

# ANNUAL SCIENTIFIC REPORT

2022



A black and white photograph capturing a moment in an operating room. In the foreground, a surgeon's hands are visible, wearing blue surgical gloves and holding a long, thin medical instrument. Behind them, three other healthcare professionals are focused on their work. One surgeon is positioned directly above the hands, while two others stand to the right. All individuals are dressed in full surgical attire, including caps, masks, and gowns. The background shows the sterile environment of the operating room with its overhead lights and equipment.

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The cover has been designed by  
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the Talent Creatiu i Empresa program

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Sant Joan de Déu, Parc Sanitari Sant  
Joan de Déu & Institut de Neurociències ·  
Universitat de Barcelona

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# 1

## FOREWORD

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Governance and structures

Global numbers 2022

Global numbers 2015-2022

Institutional awards

# Governance and structures

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# Governance and structures

## Research commission

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Dr. Cecilia Jiménez-Mallebrera

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Dr. Guerau Fernández Isern

### Secretary

Ms. Júlia Ribot Ballabriga



# Global numbers 2022

## 411 PROJECTS



- 53** Competitive autonomic projects
- 195** Competitive national projects
- 46** Competitive international projects
- 3** Non competitive autonomic projects
- 102** Non competitive national projects
- 12** Non competitive international projects

## 719 RESEARCH STAFF



- 60** Group leader
- 292** Staff researcher
- 76** Postdoc - associate researcher
- 97** PhD student
- 194** Assistant researcher & Technician



## 14.5 M BUDGET\*



- 67%** Private
- 33%** Public

\* Millions of euros



## 355 CLINICAL TRIALS & OBSERVATIONAL STUDIES



- 28** Phase I
- 71** Phase II
- 140** Phase III
- 9** Phase IV
- 21** Other clinical trial
- 86** Observational studies

## 617 PUBLICATIONS



Articles & reviews published in scholarly journals indexed in Clarivate's JCR 2021

- 4685** Impact Factor
- 58%** Publications in first quartile
- 7.6** Mean impact factor
- 48%** Articles with main contributing authors

# Global numbers 2015-2022

**411**  
**PROJECTS**



**719**  
**RESEARCH STAFF**



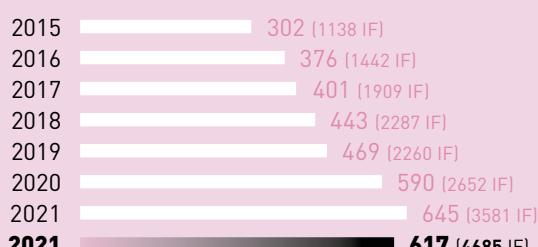
**14.5 M**  
**BUDGET\*\***



**355**  
**CLINICAL TRIALS  
& OBSERVATIONAL STUDIES**



**617**  
**PUBLICATIONS**



\* Only the projects managed by the Fundació de Recerca Sant Joan de Déu have been accounted for.  
\*\* Millions of euros

# Institutional awards

## HRS4R award

### HRS4R Human Resources Strategy for Researchers

<https://www.irsjd.org/en/research/hrs4r-hr-excellence-research/>

Fundació de Recerca Sant Joan de Déu is awarded with the logo "HR Excellence in research" since 2018. Thus, it has acquired a commitment to implement the European Charter for Researchers and the Code of Conduct for the Recruitment of Researchers (Charter & Code) in their policies and practices.

## CERCA

The Institut de Recerca Sant Joan de Déu (IRSJD) has been recognized by the board of trustees of the Institution for Research Centers in Catalonia (CERCA) as a CERCA centre in 2020

This award involves the development of a frontier research aimed at the scientific and economic impact and the improvement of the social and individual wellbeing.

IRSJD is a research center in biomedicine formed by groups from two university hospitals, Sant Joan de Déu Hospital and the Health Park Sant Joan de Déu, and three research institutes, the Institute of Biomedicine (IBUB) and the Institute of Neurosciences (UBNeuro), both from the UB, and thUPC's Research Centre in Biomedical Engineering.

The CERCA institute focuses its actions on the contribution to the international presence of research centres from Catalonia and to promote the cooperation and scientific exchange with the top centres and universities worldwide. Moreover, it promotes the adoption of joint policies in research management, scientific development and knowledge transfer. Also, it works in order to increase and ease knowledge transfer in the business sector and general public, promoting the involvement of these agents in the field of research.



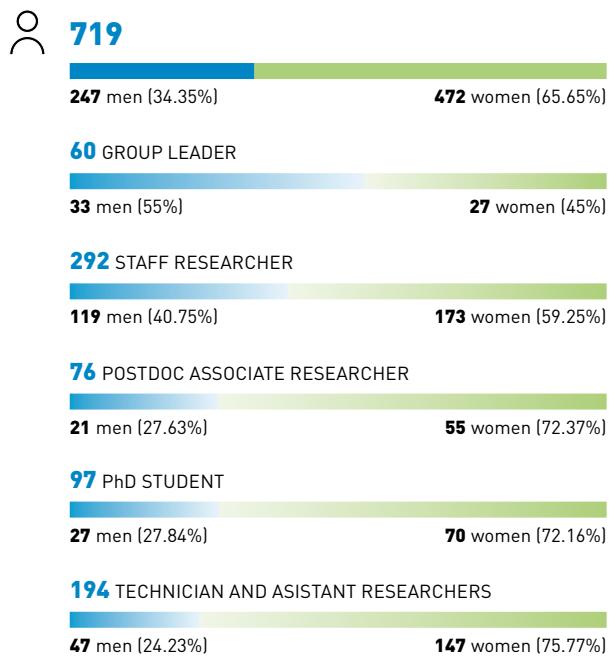
# 2 STAFF

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Research staff



# Research staff





# 3

## PROJECTS AND CLINICAL TRIALS

New projects

Active projects

New clinical trials and observational studies

Active clinical trials and observational studies



# New projects

# Active projects

**141**

120 competitive	21 non competitive
<b>20 INTERNATIONAL</b>	
17 competitive	3 non competitive
<b>88 NATIONAL</b>	
70 competitive	18 non competitive
<b>33 AUTONOMIC</b>	
33 competitive	0 non competitive

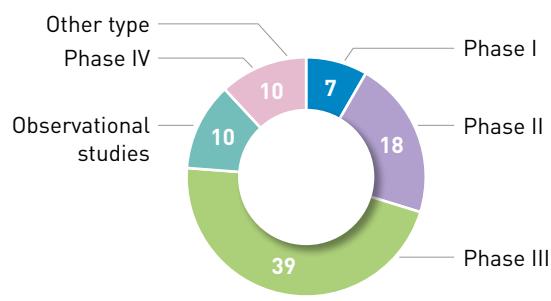
**411**

294 competitive	117 non competitive
<b>58 INTERNATIONAL</b>	
46 competitive	12 non competitive
<b>297 NATIONAL</b>	
195 competitive	102 non competitive
<b>56 AUTONOMIC</b>	
53 competitive	3 non competitive



# New clinical trials and observational studies

**Clinical Trials by Type**

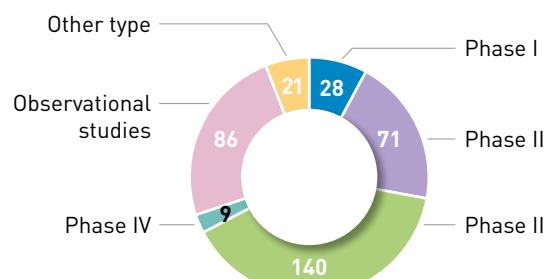


<b>64</b> CLINICAL TRIALS	<b>10</b> OBSERVATIONAL STUDIES	<b>10</b> OTHERS BY RESEARCH AREA
---------------------------------	---------------------------------------	---

- 26 NEUROLOGY**  
(23 clinical trials & 1 observational study & 2 others)
- 6 ENDOCRINOLOGY**  
(6 clinical trials)
- 5 ONCOLOGY**  
(4 clinical trials & 1 observational study)
- 6 ALLERGY AND CLINICAL IMMUNOLOGY**  
(6 clinical trials)
- 5 INFECTIOUS DISEASES**  
(5 clinical trials)
- 4 DERMATOLOGY**  
(4 clinical trials)
- 4 CLINICAL HAEMATOLOGY**  
(1 clinical trial & 3 observational studies)
- 4 GASTROENTEROLOGY, HEPATOLOGY AND NUTRITION**  
(3 clinical trials & 1 others)
- 4 PNEUMOLOGY**  
(4 clinical trials)
- 4 RHEUMATOLOGY**  
(3 clinical trials & 1 observational study)
- 2 INTERNAL MEDICINE**  
(2 Observational studies)
- 2 EMERGENCY ROOM**  
(2 others)
- 2 NEPHROLOGY**  
(1 clinical trial & 1 observational study)
- 2 PSYCHIATRY AND PSYCHOLOGY**  
(1 clinical trial & 1 other)
- 2 GYNAECOLOGY AND OBSTETRICS**  
(2 clinical trials)
- 1 ORTHOPAEDICS AND TRAUMATOLOGY**  
(1 others)
- 1 NURSERY**  
(1 others)
- 1 NEUROSURGERY**  
(1 others)
- 1 OPHTHALMOLOGY**  
(1 clinical trial)
- 1 OTOLARYNGOLOGY**  
(1 Others)
- 1 PHARMACY**  
(1 observational study)

# Active clinical trials and observational studies

**Clinical Trials by Type**



<b>230</b> CLINICAL TRIALS	<b>86</b> OBSERVATIONAL STUDIES	<b>21</b> OTHERS BY RESEARCH AREA
----------------------------------	---------------------------------------	---

- 76 NEUROLOGY**  
(61 clinical trials & 12 observational studies & 3 others)
- 59 ONCOLOGY**  
(49 clinical trials & 9 observational studies & 1 others)
- 29 RHEUMATOLOGY**  
(21 clinical trials & 7 observational studies & 1 others)
- 22 DERMATOLOGY**  
(19 clinical trials & 3 observational studies)
- 21 INFECTIOUS DISEASES**  
(19 clinical trials & 1 observational studies & 1 other)
- 20 CLINICAL HAEMATOLOGY**  
(9 clinical trials & 11 observational studies)
- 17 ALLERGY AND CLINICAL IMMUNOLOGY**  
(11 clinical trials & 6 observational studies)
- 17 ENDOCRINOLOGY**  
(10 clinical trials & 7 observational studies)
- 16 GASTROENTEROLOGY, HEPATOLOGY AND NUTRITION**  
(9 clinical trials & 5 observational studies & 2 others)
- 12 INTERNAL MEDICINE**  
(7 clinical trials & 5 observational studies)
- 10 GYNAECOLOGY AND OBSTETRICS**  
(5 clinical trials & 5 observational studies)
- 10 PNEUMOLOGY**  
(8 clinical trials & 2 observational studies)
- 8 NEPHROLOGY**  
(4 clinical trials & 4 observational studies)
- 8 PSYCHIATRY AND PSYCHOLOGY**  
(6 clinical trials & 2 others)
- 5 CARDIOLOGY**  
(3 clinical trials & 1 observational study & 1 others)
- 5 INTENSIVE CARE UNIT**  
(5 clinical trials)
- 4 NEONATOLOGY**  
(3 clinical trials & 1 other)
- 4 OPHTHALMOLOGY**  
(3 clinical trials & 1 observational study)
- 3 ANAESTHESIA**  
(2 observational studies & 1 other)
- 3 ORTHOPAEDICS AND TRAUMATOLOGY**  
(1 clinical trial & 1 others)
- 3 DIGESTIVE MEDICINE**  
(1 clinical trial & 2 observational studies)
- 2 EMERGENCY ROOM**  
(1 others)
- 1 REHABILITATION AND PHYSICAL MEDICINE**  
(1 clinical trial)

# 4

## SCIENTIFIC PRODUCTION AND RESULTS

Scientific publications  
Highest impact factor journals  
Patents, utility models and spin-offs  
Doctoral Theses

# Scientific publications

	Output (1)	Total IF	Mean IF	%Q1	%D1
Article	598	4553	7.6	58%	22%
Editorial material	12	147	12.3	58%	33%
Letter	28	479	17.1	54%	32%
Review	19	132	6.9	74%	26%
Others (2)	70	649	9.3	69%	19%
<b>Total</b>	<b>727</b>	<b>5961</b>	<b>8.2</b>	<b>59%</b>	<b>22%</b>

(1) Publications with Impact Factor. JCR 2021

(2) Other: case report, correction, meeting abstract, proceeding paper)



# Highest impact factor journals

## ORIGINALS ARTICLES IN FIRST DECIL

Number of original articles in first decil journals: 169

Journal	Total articles	IF
LANCET	1	202,731
N ENGL J MED	1	176,079
LANCET RESPIR MED	1	102,642
NAT. MED.	1	87,241
LANCET MICROBE	1	86,208
WORLD PSYCHIATRY	1	79,683
NATURE	2	69,504
LANCET NEUROL	2	59,935
NAT GENET	1	41,307
CIRCULATION	1	39,918
LANCET GLOB HEALTH	2	38,927
J INFECT	1	38,637
CANCER DISCOV	1	38,272
LANCET RHEUMATOL.	1	35,482
GASTROENTEROLOGY	1	33,883
EUR RESPIR J	1	33,795
JAMA ONCOL.	1	31,777
CELL METAB	1	31,373
LANCET HAEMATOL	1	30,153
J HEPATOL	2	30,083
JAMA NEUROL	2	29,907
ANN RHEUM DIS	1	27,973
J. AM. COLL. CARDIOL.	2	27,203
BLOOD	2	25,476
EUROSURVEILLANCE	1	21,286
CLIN INFECT DIS	5	20,999
J MED VIROL	5	20,693
J CLIN INVEST	2	19,456
BR J SPORTS MED	1	18,473

Journal	Total articles	IF
NAT COMMUN	2	17,694
ECLINICALMEDICINE	2	17,033
EMERG INFECT DIS	3	16,126
J THROMB HAEMOST	1	16,036
NPJ DIGIT MED	1	15,357
GENOME MED	1	15,266
BRAIN	2	15,255
SCI ADV	1	14,957
ALLERGY	3	14,71
AM J BIOETH	1	14,676
J ALLERGY CLIN IMMUNOL	1	14,29
OPHTHALMOLOGY	2	14,277
EMBO MOL MED	1	14,26
QJM	1	14,04
MOL PSYCHIATRY	4	13,437
JAMA NETW OPEN	2	13,353
CANCER RES	2	13,312
CLIN MICROBIOL INFECT	1	13,31
AM J HEMATOL	1	13,265
MOL THER	1	12,91
LEUKEMIA	2	12,883
NEUROLOGY	2	11,8
AGEING RES REV	1	11,788
J CONTROL RELEASE	1	11,467
NEUROL NEUROIMMUNOL NEUROINFLAMM	1	11,36
ANN NEUROL	1	11,274
PSYCHIATRY RES.	4	11,225
ADV HEALTHC MATER	1	11,092

# Highest impact factor journals

Journal	Total articles	IF
AGING CELL	1	11,005
OBES REV	1	10,867
REDOX BIOL	1	10,787
INT J BIOL SCI	1	10,75
CARBOHYDR POLYM	1	10,723
AM J OBSTET GYNECOL	5	10,693
PSYCHOL MED	3	10,592
THROMB RES	2	10,407
PHARMACOL RES	1	10,334
STROKE	1	10,17
J PATHOL	1	9,883
PEDIATRICS	1	9,703
MOV DISORD	1	9,698
INT J EPIDEMIOL	1	9,685
HEALTH PSYCHOL REV	1	9,638
ALIMENT PHARMACOL THER	1	9,524
REV ENDOCR METAB DISORD	2	9,306
SENS ACTUATOR B-CHEM	3	9,221
NEUROSCI BIOBEHAV REV	3	9,052
GENET MED	1	8,864
ONCOGENE	1	8,756
AM J OBSTET GYNECOL MFM	1	8,679
ULTRASOUND OBSTET GYNECOL	6	8,678
EXPERT SYST APPL	1	8,665
EVID BASED MENT HEALTH	2	8,541
ENVIRON RES	2	8,431
JAMA OPHTHALMOL	1	8,253
DEPRESS ANXIETY	2	8,128
ANAL CHEM	1	8,008
J BIOMED INFORM	1	8
ANTIOXIDANTS (BASEL)	5	7,675

Journal	Total articles	IF
ACTA PHYSIOL	1	7,523
BIOMED PHARMACOTHER	1	7,419
J MED INTERNET RES	1	7,076
ARCH DIS CHILD FETAL NEONATAL ED	3	6,643
FRONT PUBLIC HEALTH	3	6,461
J PEDIATR	1	6,314
NONLINEAR DYN	1	5,741
PEDIATR ALLERGY IMMUNOL.	2	5,464
EUR CHILD ADOLESC PSYCHIATRY	1	5,349
INDIAN J PEDIATR	1	5,319
EUR J HEALTH ECON	1	5,271
INT J MENT HEALTH NURS	1	5,1
J NURS MANAG	1	4,68
J CLIN NURS	2	4,423
BRAIN STRUCT FUNCT	1	3,748
J INTELLECT DISABIL RES	1	3,646
MATHEMATICS	1	2,592

# Highest impact factor journals

**Originals articles D1 with IRSJD main contribution: 63**

Original article	Journal	IF	Program
Agüera T, Carreras N, Agut T. Cerebral Ultrasonography of Cytotoxic Edema in a Newborn With a Mitochondrial Disorder. <i>JAMA Neurol.</i> 2022;79(7):722-723. IF:29,907. (Q1).	JAMA NEUROL	29,907	Neuroscience
Jordán I, Fernández de Sevilla M, Fumadó V, Bassat Q, Bonet E, Fortuny C et al. Transmission of Severe Acute Respiratory Syndrome Coronavirus 2 Infection Among Children in Summer Schools Applying Stringent Control Measures in Barcelona, Spain. <i>Clin Infect Dis.</i> 2022;74(1):66-73. IF:20,999. (Q1).	CLIN INFECT DIS	20,999	Neuroscience
Jordán I, Fernández de Sevilla M, Fumadó V, Bassat Q, Bonet E, Fortuny C et al. Transmission of Severe Acute Respiratory Syndrome Coronavirus 2 Infection Among Children in Summer Schools Applying Stringent Control Measures in Barcelona, Spain. <i>Clin Infect Dis.</i> 2022;74(1):66-73. IF:20,999. (Q1).	CLIN INFECT DIS	20,999	Adaptive Response, Metabolism and Immunobiology
Jordán I, Fernández de Sevilla M, Fumadó V, Bassat Q, Bonet E, Fortuny C et al. Transmission of Severe Acute Respiratory Syndrome Coronavirus 2 Infection Among Children in Summer Schools Applying Stringent Control Measures in Barcelona, Spain. <i>Clin Infect Dis.</i> 2022;74(1):66-73. IF:20,999. (Q1).	CLIN INFECT DIS	20,999	Adaptive Response, Metabolism and Immunobiology
Jordán I, Fernández de Sevilla M, Fumadó V, Bassat Q, Bonet E, Fortuny C et al. Transmission of Severe Acute Respiratory Syndrome Coronavirus 2 Infection Among Children in Summer Schools Applying Stringent Control Measures in Barcelona, Spain. <i>Clin Infect Dis.</i> 2022;74(1):66-73. IF:20,999. (Q1).	CLIN INFECT DIS	20,999	Health Technologies and Innovation
Ciruela P, Soldevila N, García JJ, González S, Álvaro C, Redin A et al. Effect of COVID-19 Pandemic on Invasive Pneumococcal Disease in Children, Catalonia, Spain. <i>Emerg Infect Dis.</i> 2022;28(11):2321-2325. IF:16,126. (Q1).	EMERG INFECT DIS	16,126	Adaptive Response, Metabolism and Immunobiology
Ruiz A, Gassiot S, Edurne G, Zubicaray J, José Luis Dapena D, Rives S et al. Venous thromboembolism in pediatric patients with acute lymphoblastic leukemia under chemotherapy treatment. Risk factors and usefulness of thromboprophylaxis. Results of LAL-SEHOP-PETHEMA-2013. <i>J. Thromb. Haemost.</i> 2022;20(6):1390-1399. IF:16,036. (Q1).	J. THROMB. HAEMOST.	16,036	Paediatric Cancer
Rodríguez Del Río P, Alvarez A, Blumchen K, Caimmi D, Caubet JC, Konstantinopoulos AP et al. Food immunotherapy practice: Nation differences across Europe, the FIND project. <i>Allergy.</i> 2022;77(3):920-932. IF:14,710. (Q1).	ALLERGY	14,71	Adaptive Response, Metabolism and Immunobiology
Esquerda M, Lorenzolzquierdo D, Torralba F. Certainties and Uncertainties in Genetic Information: Good Ethics Starts with Good Data. <i>Am J Bioeth.</i> 2022;22(2):48-50. IF:14,676. (Q1).	AM J BIOETH	14,676	Adaptive Response, Metabolism and Immunobiology

# Highest impact factor journals

Original article	Journal	IF	Program
Antón E, Dalla E, Orlandi JG, Castro G, Gualda EJ, Young A et al. Deficiency of the <i>ywhaz</i> gene, involved in neurodevelopmental disorders, alters brain activity and behaviour in zebrafish. <i>Mol Psychiatry</i> . 2022;27(9):3739-3748. IF:13,437. (Q1).	MOL PSYCHIATRY	13,437	Precision Medicine in Rare Diseases
Fernandez N, Cabana J, Corominas L, Cormand B. Molecular genetics of cocaine use disorders in humans. <i>Mol Psychiatry</i> . 2022;27(1):624-639. IF:13,437. (Q1).	MOL PSYCHIATRY	13,437	Precision Medicine in Rare Diseases
Kostev K, Abeler S, Koyanagi A, Haro JM, Smith L, Jacob L. Sales of Iodine-Containing Drugs in Europe Following the Beginning of the War Between Russia and Ukraine. <i>JAMA Netw Open</i> . 2022;5(10) IF:13,353. (Q1).	JAMA NETW OPEN	13,353	Mental Health and Society
Ortiz V, Alonso A, Español M, Martínez N, Anna F, Magnano L et al. Results of ARI-0001 CART19 cell therapy in patients with relapsed/refractory CD19-positive acute lymphoblastic leukemia with isolated extramedullary disease. <i>Am J Hematol</i> . 2022;97(6):731-739. IF:13,265. (Q1).	AM J HEMATOL	13,265	Adaptive Response, Metabolism and Immunobiology
Ortiz V, Alonso A, Español M, Martínez N, Anna F, Magnano L et al. Results of ARI-0001 CART19 cell therapy in patients with relapsed/refractory CD19-positive acute lymphoblastic leukemia with isolated extramedullary disease. <i>Am J Hematol</i> . 2022;97(6):731-739. IF:13,265. (Q1).	AM J HEMATOL	13,265	Paediatric Cancer
Pascual G, Castillo H, Unceta N, Aschero MR, Resa C, Gómez A et al. SPARC-mediated long-term retention of nab-paclitaxel in pediatric sarcomas. <i>J Control Release</i> . 2022;342:81-92. IF:11,467. (Q1).	J CONTROL RELEASE	11,467	Paediatric Cancer
Pascual G, Castillo H, Unceta N, Aschero MR, Resa C, Gómez A et al. SPARC-mediated long-term retention of nab-paclitaxel in pediatric sarcomas. <i>J Control Release</i> . 2022;342:81-92. IF:11,467. (Q1).	J CONTROL RELEASE	11,467	Paediatric Cancer
Pascual G, Castillo H, Unceta N, Aschero MR, Resa C, Gómez A et al. SPARC-mediated long-term retention of nab-paclitaxel in pediatric sarcomas. <i>J Control Release</i> . 2022;342:81-92. IF:11,467. (Q1).	J CONTROL RELEASE	11,467	Paediatric Cancer
Kuseyri O, Julià NA, Olivella M, Guder P, Zafeiriou DI, Horvath G et al. Integrative Approach to Predict Severity in Nonketotic Hyperglycinemia. <i>Ann Neurol</i> . 2022;92(2):292-303. IF:11,274. (Q1).	ANN NEUROL	11,274	Neuroscience
Isabel D, Moreno B, García M, Barrigón ML, Fermín H, López R et al. Is Metacognitive Training effective for improving neurocognitive function in patients with a recent onset of psychosis?. <i>Psychiatry Res</i> . 2022;318:114941-114941. IF:11,225. (Q1).	PSYCHIATRY RES.	11,225	Scientific base and approach to mental disorders
Diago M, Vila R, Serra C, Butjosa A, del Cacho N, Marina S et al. Emotional abuse and perceived stress: The most relevant factors in suicide behavior in first-episode psychosis patients. <i>Psychiatry Res</i> . 2022;315:114699-114699. IF:11,225. (Q1).	PSYCHIATRY RES.	11,225	Scientific base and approach to mental disorders

# Highest impact factor journals

Original article	Journal	IF	Program
Diago M, Vila R, Serra C, Butjosa A, del Cacho N, Marina S et al. Emotional abuse and perceived stress: The most relevant factors in suicide behavior in first-episode psychosis patients. <i>Psychiatry Res.</i> 2022;315:114699-114699. IF:11,225. (Q1).	PSYCHIATRY RES.	11,225	Neuroscience
Narita Z, Banawa R, Zhou S, DeVylder J, Koyanagi A, Hans OH. Loneliness and psychotic experiences among US university students: Findings from the Healthy Minds Study 2020. <i>Psychiatry Res.</i> 2022;308:114362-114362. IF:11,225. (Q1).	PSYCHIATRY RES.	11,225	Mental Health and Society
Oliver LL, Martin H, Mandakhbayar N, Jo YW, Cavalcanti EA, Kim HW et al. Mimicking Bone Extracellular Matrix: From BMP-2-Derived Sequences to Osteogenic-Multifunctional Coatings. <i>Adv. Healthc. Mater.</i> 2022;11(20):IF:11,092. (Q1).	ADV. HEALTHC. MATER.	11,092	Health Technologies and Innovation
Cuadrado M, Liu J, Paco S, Aschero MR, Burgeño V, Sirab N et al. Identification of immunosuppressive factors in retinoblastoma cell secretomes and aqueous humor from patients. <i>J Pathol.</i> 2022;257(3):327-339. IF:9,883. (Q1).	J PATHOL	9,883	Associated Clinical Research Groups
Cuadrado M, Liu J, Paco S, Aschero MR, Burgeño V, Sirab N et al. Identification of immunosuppressive factors in retinoblastoma cell secretomes and aqueous humor from patients. <i>J Pathol.</i> 2022;257(3):327-339. IF:9,883. (Q1).	J PATHOL	9,883	Precision Medicine in Rare Diseases
Cuadrado M, Liu J, Paco S, Aschero MR, Burgeño V, Sirab N et al. Identification of immunosuppressive factors in retinoblastoma cell secretomes and aqueous humor from patients. <i>J Pathol.</i> 2022;257(3):327-339. IF:9,883. (Q1).	J PATHOL	9,883	Paediatric Cancer
Cuadrado M, Liu J, Paco S, Aschero MR, Burgeño V, Sirab N et al. Identification of immunosuppressive factors in retinoblastoma cell secretomes and aqueous humor from patients. <i>J Pathol.</i> 2022;257(3):327-339. IF:9,883. (Q1).	J PATHOL	9,883	Paediatric Cancer
Cuadrado M, Liu J, Paco S, Aschero MR, Burgeño V, Sirab N et al. Identification of immunosuppressive factors in retinoblastoma cell secretomes and aqueous humor from patients. <i>J Pathol.</i> 2022;257(3):327-339. IF:9,883. (Q1).	J PATHOL	9,883	Paediatric Cancer
Carbonell C, Rubio M, Marqués S, Peñarrubia MT, María Montserrat Gil G, García V et al. Pediatric Medication Noninitiation in Spain. <i>Pediatrics.</i> 2022;149(1):IF:9,703. (Q1).	PEDIATRICS	9,703	Mental Health and Society
Lara E, María M, Olaya B, Caballero FF, Morillo D, Moneta MV et al. Cohort Profile: The Spanish Longitudinal Study on Ageing and Health (Edad Con Salud). <i>INT J EPIDEMIOL.</i> 2022;51(4):IF:9,685. (Q1).	INT J EPIDEMIOL	9,685	Mental Health and Society
García I, Michaud A, Jurado MA, Dagher A, Morys F. Mechanisms linking obesity and its metabolic comorbidities with cerebral grey and white matter changes. <i>Rev Endocr Metab Disord.</i> 2022;23(4):833-843. IF:9,306. (Q1).	REV ENDOCR METAB DISORD	9,306	Neuroscience

# Highest impact factor journals

Original article	Journal	IF	Program
Gavaldà A, Villarroya J, Cereijo R, Giralt M, Villarroya F. The endocrine role of brown adipose tissue: An update on actors and actions. <i>Rev Endocr Metab Disord.</i> 2022;23(1):31-41. IF:9,306. (Q1).	REV ENDOCR METAB DISORD	9,306	Adaptive Response, Metabolism and Immunobiology
Solorzano A, Eichmann J, Fernandez L, Ziems B, Jimenez JM, Marco S et al. Early fire detection based on gas sensor arrays: Multivariate calibration and validation. <i>Sens. Actuator B-Chem.</i> 2022;352:IF:9,221. (Q1).	SENS. ACTUATOR B-CHEM.	9,221	Health Technologies and Innovation
Boutet ML, Eixarch E, Ahumada P, Nakaki A, Crovetto F, Cívico MS et al. Fetal neurosonography and infant neurobehavior following conception by assisted reproductive technology with fresh or frozen embryo transfer. <i>Ultrasound Obstet Gynecol.</i> 2022;60(5):646-656. IF:8,678. (Q1).	ULTRASOUND OBSTET GYNECOL	8,678	Adaptive Response, Metabolism and Immunobiology
Basso A, Youssef L, Nakaki A, Paules C, Miranda J, Casu G et al. Fetal neurosonography at 31-35 weeks reveals altered cortical development in pre-eclampsia with and without small-for-gestational-age fetus. <i>Ultrasound Obstet Gynecol.</i> 2022;59(6):737-746. IF:8,678. (Q1).	ULTRASOUND OBSTET GYNECOL	8,678	Adaptive Response, Metabolism and Immunobiology
Pérez M, Gómez O, Gibert M, Masoller N, Marimon E, Lip D et al. Corpus callosum size by neurosonography in fetuses with congenital heart defect and relationship with expected pattern of brain oxygen supply. <i>Ultrasound Obstet Gynecol.</i> 2022;59(2):220-225. IF:8,678. (Q1).	ULTRASOUND OBSTET GYNECOL	8,678	Adaptive Response, Metabolism and Immunobiology
Manzini E, Garrido J, Fonollosa J, Perera A. Mapping layperson medical terminology into the Human Phenotype Ontology using neural machine translation models. <i>Expert Syst. Appl.</i> 2022;204:IF:8,665. (Q1).	EXPERT SYST. APPL.	8,665	Health Technologies and Innovation
Cahuana MJ, Mazarico E, Cahuana AJ, Pascal R, Alonso L, Targa J et al. Maternal short-term exposure to NO <sub>2</sub> during pregnancy and its relationship with Doppler markers of placental function. <i>Environ Res.</i> 2022;214[Pt 1]:113813-113813. IF:8,431. (Q1).	ENVIRON RES	8,431	Adaptive Response, Metabolism and Immunobiology
Smith L, Shin JI, Butler L, Barnett Y, Hans OH, Jacob L et al. Physical multimorbidity and depression: A mediation analysis of influential factors among 34,129 adults aged ≥50 years from low- and middle-income countries. <i>Depress. Anxiety.</i> 2022;39(5):376-386. IF:8,128. (Q1).	DEPRESS. ANXIETY	8,128	Mental Health and Society
Gabarrell A, Moneta MV, Ayuso JL, Maria M, Lara E, Haro JM et al. The effect of loneliness and social support on the course of major depressive disorder among adults aged 50 years and older: A longitudinal study. <i>Depress. Anxiety.</i> 2022;39(2):147-155. IF:8,128. (Q1).	DEPRESS. ANXIETY	8,128	Mental Health and Society
Manzini E, Vlacho B, Franch J, Escudero J, Génova A, Reixach E et al. Longitudinal deep learning clustering of Type 2 Diabetes Mellitus trajectories using routinely collected health records. <i>J. Biomed. Inform.</i> 2022;135:104218-104218. IF:8,000. (Q1).	J. BIOMED. INFORM.	8	Health Technologies and Innovation

# Highest impact factor journals

Original article	Journal	IF	Program
Paredes AJ, Oliva C, Montero R, Alcaide P, Ruijter GJG, García J et al. Technical Aspects of Coenzyme Q(10) Analysis: Validation of a New HPLC-ED Method. <i>Antioxidants (Basel)</i> . 2022;11(3):IF:7,675. (Q1).	ANTIOXIDANTS (BASEL)	7,675	Precision Medicine in Rare Diseases
Cañizo D, Hadley SM, Marta O, López M, Valls A, Viñals ML et al. Oxidative Stress and Indicators of Brain Damage Following Pediatric Heart Surgery. <i>Antioxidants (Basel)</i> . 2022;11(3):489IF:7,675. (Q1).	ANTIOXIDANTS (BASEL)	7,675	Adaptive Response, Metabolism and Immunobiology
Escobar MC, Pérez M, Arráez M, Cascant MM, Albiach A, Kuligowski J et al. Brain Oxygen Perfusion and Oxidative Stress Biomarkers in Fetuses with Congenital Heart Disease-A Retrospective, Case-Control Pilot Study. <i>Antioxidants (Basel)</i> . 2022;11(2):IF:7,675. (Q1).	ANTIOXIDANTS (BASEL)	7,675	Adaptive Response, Metabolism and Immunobiology
Sebastiani G, Navarro E, Almeida L, Serra M, Paltrinieri AL, García Ó et al. Effects of Antioxidant Intake on Fetal Development and Maternal/Neonatal Health during Pregnancy. <i>Antioxidants (Basel)</i> . 2022;11(4):IF:7,675. (Q1).	ANTIOXIDANTS (BASEL)	7,675	Adaptive Response, Metabolism and Immunobiology
Escobar MC, Pérez M, Arráez M, Cascant MM, Albiach A, Kuligowski J et al. Brain Oxygen Perfusion and Oxidative Stress Biomarkers in Fetuses with Congenital Heart Disease-A Retrospective, Case-Control Pilot Study. <i>Antioxidants (Basel)</i> . 2022;11(2):IF:7,675. (Q1).	ANTIOXIDANTS (BASEL)	7,675	Adaptive Response, Metabolism and Immunobiology
Cañizo D, Hadley SM, Marta O, López M, Valls A, Viñals ML et al. Oxidative Stress and Indicators of Brain Damage Following Pediatric Heart Surgery. <i>Antioxidants (Basel)</i> . 2022;11(3):489IF:7,675. (Q1).	ANTIOXIDANTS (BASEL)	7,675	Adaptive Response, Metabolism and Immunobiology
Escobar MC, Pérez M, Arráez M, Cascant MM, Albiach A, Kuligowski J et al. Brain Oxygen Perfusion and Oxidative Stress Biomarkers in Fetuses with Congenital Heart Disease-A Retrospective, Case-Control Pilot Study. <i>Antioxidants (Basel)</i> . 2022;11(2):IF:7,675. (Q1).	ANTIOXIDANTS (BASEL)	7,675	Adaptive Response, Metabolism and Immunobiology
Cañizo D, Hadley SM, Marta O, López M, Valls A, Viñals ML et al. Oxidative Stress and Indicators of Brain Damage Following Pediatric Heart Surgery. <i>Antioxidants (Basel)</i> . 2022;11(3):489IF:7,675. (Q1).	ANTIOXIDANTS (BASEL)	7,675	Adaptive Response, Metabolism and Immunobiology
Nolla C, Casabella S, Jimenez V, Vallmitjana A, Tarifa C, Herraiz A et al. beta 2-adrenergic stimulation potentiates spontaneous calcium release by increasing signal mass and co-activation of ryanodine receptor clusters. <i>Acta Physiol.</i> 2022;234(4):IF:7,523. (Q1).	ACTA PHYSIOL.	7,523	Health Technologies and Innovation
Nolla C, Casabella S, Jimenez V, Vallmitjana A, Tarifa C, Herraiz A et al. beta 2-adrenergic stimulation potentiates spontaneous calcium release by increasing signal mass and co-activation of ryanodine receptor clusters. <i>Acta Physiol.</i> 2022;234(4):IF:7,523. (Q1).	ACTA PHYSIOL.	7,523	Health Technologies and Innovation
Nuñez C, Stephan C, Arca G, Agut T, Arnaez J, Cordeiro M et al. Neonatal arterial stroke location is associated with outcome at 2 years: a voxel-based lesion-symptom mapping study. <i>Arch Dis Child Fetal Neonatal Ed.</i> 2022;107(1):45-50. IF:6,643. (Q1).	ARCH DIS CHILD FETAL NEONATAL ED	6,643	Neuroscience
Nuñez C, Stephan C, Arca G, Agut T, Arnaez J, Cordeiro M et al. Neonatal arterial stroke location is associated with outcome at 2 years: a voxel-based lesion-symptom mapping study. <i>Arch Dis Child Fetal Neonatal Ed.</i> 2022;107(1):45-50. IF:6,643. (Q1).	ARCH DIS CHILD FETAL NEONATAL ED	6,643	Scientific base and approach to mental disorders

# Highest impact factor journals

Original article	Journal	IF	Program
Rico LA, Morillo D, Rodríguez Á, Vorstenbosch E, Weser AJ, Fincias L et al. Worldwide mapping of initiatives that integrate population cohorts. <i>Front Public Health</i> . 2022;10:964086-964086. IF:6,461. (Q1).	FRONT PUBLIC HEALTH	6,461	Mental Health and Society
Corral C, Sánchez A, María Montserrat Gil G, Peñarrubia MT, Aznar I, Serrano A et al. Improving Initial Medication Adherence to cardiovascular disease and diabetes treatments in primary care: Pilot trial of a complex intervention. <i>Front Public Health</i> . 2022;10:1038138-1038138. IF:6,461. (Q1).	FRONT PUBLIC HEALTH	6,461	Mental Health and Society
Barjau A, Batlle JA. Predicting impact scenarios of a rimless wheel: a geometrical approach. <i>Nonlinear Dyn.</i> 2022;110(4):3209-3227. IF:5,741. (Q1).	NONLINEAR DYN.	5,741	Health Technologies and Innovation
Rodríguez M, Álvarez X, Tor J, Pardo M, de la Serna E, Sugranyes G et al. Major depressive disorder and attenuated negative symptoms in a child and adolescent sample with psychosis risk syndrome: the CAPRIS study. <i>Eur Child Adolesc Psychiatry</i> . 2022;31(9):1431-1440. IF:5,349. (Q1).	EUR CHILD ADOLESC PSYCHIATRY	5,349	Neuroscience
Alsina L, Montoro JB, Moral PM, Neth O, Pica MO, Sánchez S et al. Cost-minimization analysis of immunoglobulin treatment of primary immunodeficiency diseases in Spain. <i>Eur J Health Econ.</i> 2022;23(3):551-558. IF:5,271. (Q1).	EUR J HEALTH ECON	5,271	Adaptive Response, Metabolism and Immunobiology
Migliorelli C, Medina IF, Bachiller A, Tost A, Alonso JF, López A et al. Cognitive stimulation has potential for brain activation in individuals with Rett syndrome. <i>J Intellect Disabil Res.</i> 2022;66(3):213-224. IF:3,646. (Q1).	J INTELLECT DISABIL RES	3,646	Neuroscience
Migliorelli C, Medina IF, Bachiller A, Tost A, Alonso JF, López A et al. Cognitive stimulation has potential for brain activation in individuals with Rett syndrome. <i>J Intellect Disabil Res.</i> 2022;66(3):213-224. IF:3,646. (Q1).	J INTELLECT DISABIL RES	3,646	Precision Medicine in Rare Diseases
Migliorelli C, Medina IF, Bachiller A, Tost A, Alonso JF, López A et al. Cognitive stimulation has potential for brain activation in individuals with Rett syndrome. <i>J Intellect Disabil Res.</i> 2022;66(3):213-224. IF:3,646. (Q1).	J INTELLECT DISABIL RES	3,646	Adaptive Response, Metabolism and Immunobiology
Migliorelli C, Medina IF, Bachiller A, Tost A, Alonso JF, López A et al. Cognitive stimulation has potential for brain activation in individuals with Rett syndrome. <i>J Intellect Disabil Res.</i> 2022;66(3):213-224. IF:3,646. (Q1).	J INTELLECT DISABIL RES	3,646	Health Technologies and Innovation
Marimon X, Traserra S, Jimenez M, Ospina A, Benítez R. Detection of Abnormal Cardiac Response Patterns in Cardiac Tissue Using Deep Learning. <i>Mathematics</i> . 2022;10(15)IF:2,592. (Q1).	MATHEMATICS	2,592	Health Technologies and Innovation

# Patents, utility models and spin-offs

## New patents, license agreements and utility models

### Perfiles de biomarcadores genéticos para la aptitud para el deporte de resistencia

Country: Germany  
Number: DE602018037391T2  
Inventors: Alexandre Perera Lluna & Maria de Los Angeles Maqueda Gonzalez  
Proprietors: Universitat Politècnica de Catalunya

### System to assist walking

Country: Australia  
Number: AU2017344951B2  
Inventors: Josep Maria Font Llagunes & Daniel Clos Costa  
Proprietors: Universitat Politècnica de Catalunya

## Portfolio of spin-offs

### Virmedex Virtual Experiences S.L.

2022 · <https://virmedex.com/>  
Co-founders: Dani Tost, Núria Bonet & Ariel von Barnekow  
Virmedex provides the next generation of training platforms for high performance risk-aware professionals. With more than 20 years in the field of serious games and gamification for health and education, they have partnered with hospitals, research centers, institutions and companies. They have designed gamification strategies and serious games for rehabilitation, screening, professional training and other applications.

### Vincer.AI

2022 · <https://www.vincer.ai/>  
Co-founders: Pol Solà-Santos, Alexandre Perera Lluna & Isidro Pascual.  
Vincer. AI assist technological companies to create trusted AI technologies and therefore help them provide credible AI models and output in a trusted business environment. Their aim is to build trustworthy AI through auditing, rating, and tracing AI solutions in order to help organizations to improve their decision-making in AI and provide traceability and transparency to their clients.

### Ephion Health S.L.

2021 · <https://ephion.health/>  
Co-founders: Eurecat, Hospital Sant Joan de Déu and The Collider  
Ephion Health is the platform to develop and deploy clinical measures for patient mobility. They assist the complete parametrization of mobility for several diseases and conditions, to create a positive impact on the patient's diagnosis, treatment and quality of life. Ephion's technology allows to save time and resources in clinical care and clinical trials.

### Gate2Brain

2020 · <https://gate2brain.com/>  
Co-founders: Universidad de Barcelona (UB), Hospital Sant Joan de Déu (HSJD)  
Gate2Brain is a biotech company focused on the development of therapeutics that efficiently cross biological barriers such as the blood-brain barrier. We pursue our goals using a radically innovative peptide-based patented technology platform.

### OMADRE

2019 · <https://www.omadre.com/>  
Dr. Jaume Pérez Payarols (IRSJD)  
The company Omadre was created to provide hospitals with the tools required for the efficient management of mother's milk, increasing the safety of existing processes in this way.

### ABLE Human Motion

2018 ·  
<https://www.ablehumanmotion.com/>  
Co-founders: Dr. Alfons Carnicero, Dr. Alex García & Dr. Josep Maria Font (IRSJD)  
ABLE Human Motion is a medical device spin-off from Universitat Politècnica de Catalunya (UPC) that is developing and will commercialize robotic exoskeletons to enhance mobility and independence of people with lower-limb paralysis. We have created ABLE, the first lightweight, easy-to-use and affordable exoskeleton that allows walking again after a spinal cord injury. It helps palliating health complications while boosting self-confidence and independence.

# Patents, utility models and spin-offs

## DBGEN Ocular Genomics

2018 · <https://dbgen.com/>

Co-founders: Dr. Roser González-Duarte and Dr. Gemma Marfany (IRSJD)

DBGen is a company focused on the genetic diagnosis of hereditary eye disorders, providing the highest level of quality, diagnostic effectiveness and close contact with patients, ophthalmologists, and enterprises in need of its services. At DBGen, we offer high-quality, reliable and accessible genetic diagnosis, allowing the clinical diagnosis to be assured and guidance to be provided to the patient with respect to emerging therapies for genetic disorders of the eyes. Genetic diagnosis is indispensable to identify family members possessing a genetic defect, permitting the most suitable advice to be given in each case.

## CEBIOTEX

2012 · <http://www.cebiotex.com/>

Dr. Joan Bertran Llavina (CEO) & Dr. Angel Montero (IRSJD)  
Cebiotex has developed and characterised at preclinical level, in vitro and in vivo, nonwoven nanofiber membranes for the local release of active ingredients in the local treatment of the surgical site after tumour excision surgery. The purpose of the local release of drugs is to achieve high concentrations of active ingredients in the area where the release device is placed, avoiding systemic toxicity and increasing the effectiveness of the drug against possible tumoral remnants. The activity of Cebiotex is based on the interaction of researchers from two Catalan institutions: the Polytechnic University of Catalonia (UPC) and Sant Joan de Déu-Barcelona Children's Hospital (HSJD).

## BCN Innova

2008 · <http://www.bcninnova.com/>

Dr. Joan Prat (IRSJD)

BCN Innova, a spin-off company of Sant Joan de Déu-Barcelona Children's Hospital, has developed a device (GazeLab) that substantially improves the diagnosis and treatment of strabismus, reducing the number of second operations that are now performed to achieve the expected results. GazeLab offers two main benefits: 1. It allows objective exploration on a revolutionary level of precision, with much better results than the subjective-measurement model that has prevailed until now. 2. It provides a 50% reduction in the number of second operations, obviating those which are surgically unnecessary.



# Doctoral Theses

Thesis author	Title	University	Thesis directors	Month of lecture	Research group
Judit Rabassa Blanco	<b>Seqüèles dentals en adolescents sotmesos a tractament oncològic durant la primera infància</b>	Universitat de Barcelona	Lluís Brunet Llobert & Jaume Miranda Rius	December	Hospital dentistry and periodontal medicine
Marta Ferrer Quintero	<b>Towards personalized medicine in psychosis: the roles of social cognition and metacognition</b>	Universitat de Barcelona	Elena Huerta and Juana Gómez	December	Etiopathogenesis and treatment of severe mental disorders (meritt)
Sara Pérez-Jaume	<b>On the diagnostic ability assessment of medical tests with right-censored data</b>	Universitat de Barcelona	Josep Lluis Carrasco Jordan	December	Paediatric cancer
Abraham José Paredes Fuentes	<b>Estudio de biomarcadores en los defectos de la fosforilación oxidativa mitocondrial y en la deficiencias de coenzima CoQ10</b>	Universitat de Barcelona	Rafael Artuch	December	Metabolic and mitochondrial medicine
Lluís Oliver-Cervelló	<b>Novel multifunctional biomimetic peptides for bone tissue engineering</b>	Universitat Politècnica de Catalunya	Carles Mas-Moruno	November	Medical technologies: Biomaterials and Tissue Engineering
Regina Vila Badia	<b>Experiències traumàtiques en la infància i conductes suïcides en pacients amb un primer episodi psícotic</b>	Universitat de Barcelona	Judith Usall i Rodié	November	Etiopathogenesis and treatment of severe mental disorders (meritt)
Javier Alvarez Subiela	<b>Predictors de la conducta suïcida en població adolescent</b>	Universitat Autònoma de Barcelona	Diego J Palao, Carmina Castellano Tejedor, Fran Villar Cabezas	November	Research in child and adolescent mental health
Lluís Oliver Cervelló	<b>Novel multifunctional biomimetic peptides for bone tissue engineering</b>	Universitat Politècnica de Catalunya	Carles Mas-Moruno	November	Medical technologies: biomaterials and tissue engineering
Izarbe Aísa-Marín	<b>Exploring the role of NR2E3 in photoreceptor cell fate determination</b>	Universitat de Barcelona	Gemma Marfany Nadal	October	Human Molecular Genetics II

# Doctoral Theses

Thesis author	Title	University	Thesis directors	Month of lecture	Research group
Maria Cuadrado Vilanova	<b>Identification of immunosuppressive factors and patterns of cell-free DNA in retinoblastoma cells secretomes and aqueous humor from patients</b>	Universitat de Barcelona	Angel Montero Carcaboso	October	Therapy development in pediatric oncology
Jonathan Olival Tremaria	<b>Application of next generation sequencing to study the genetics of idiopathic neonatal arterial ischemic stroke</b>	Universitat de Barcelona	Francesc Palau Martínez & Alfredo García-Alix Pérez	October	Neurogenetics and molecular medicine
Mònica Girona	<b>Infeccions relacionades amb l'assistència sanitària en el pacient pediàtric: factors de risc i evolució i estudi de resistències</b>	Universitat de Barcelona	Iolanda Jordan	September	Immune and respiratory dysfunction in the critical child
Cristina Jou	<b>Biomarcadores histopatológicos en las enfermedades mitocondriales en la edad pediátrica</b>	Universitat de Barcelona	Cecilia Jiménez-Mallebrera & Rafael Artuch	September	Applied research in neuromuscular diseases
Elenea Juárez Escoto	<b>Fisiopatología de modelos murinos de A-sinucleína y enfermedad de Parkinson</b>	Universitat de Barcelona	Francesc Palau Martínez & Janet Hoenicka	July	Neurogenetics and molecular medicine
Miguel Mateu	<b>Cold Plasma-Derived Oxidative Stress for Osteosarcoma Therapy</b>	Universitat Politècnica de Catalunya	Cristina Canal & Juan Tornín	June	Medical technologies: Biomaterials and Tissue Engineering
Maria Andreu Pascual		Universitat de Barcelona	Prof. Boris Birmaher (University of Pittsburgh, USA)	June	Research in child and adolescent mental health
Rosa Bosch Munso	<b>Els trastorns del neurodesenvolupament com predictors del baix rendiment acadèmic</b>	Universitat de Barcelona	Miguel Casas Brugué i Dr. Josep Antoni Ramos-Quiroga (UAB)	June	Research in child and adolescent mental health
Desirée Henares Bonilla	<b>Microbiota nasofaríngea en niños con enfermedad neumocócica invasiva, infección respiratoria banal y niños sanos: caracterización, factores moduladores y valor diagnóstico diferencial</b>	Universitat de Barcelona	Caren Muñoz-Almagro	June	Infectious diseases and microbiome

# Doctoral Theses

Thesis author	Title	University	Thesis directors	Month of lecture	Research group
Anna Solé Ribalta	Disseny i validació d'una eina pel reconeixement i estratificació precoç de la Sèpsia i Xoc Sèptic en Pediatría	Universitat de Barcelona	lolanda Jordan	June	Immune and respiratory dysfunction in the critical child
Miguel Mateu Sanz	Cold plasma-derived oxidative stress for osteosarcoma therapy	Universitat Politècnica de Catalunya	Cristina Canal Barnils & Juan Tornin Caviaelles	June	Medical technologies: biomaterials and tissue engineering
Javier Aparicio Calvo	Aplicació de noves tècniques de neuroimatge a l'avaluació pre-quirúrgica de l'epilèpsia: la combinació de la RM per tensor de difusió (DTI) i la PET-FDG, i la sostracció de la SPECT ictal amb PET-FDG interictal co-registrada amb la RM (PSICOM)	Universitat de Barcelona	Jaume Campistol & FJ Setoain	June	Epilepsy and movement disorders in pediatrics
Mar Bonan	Nanotechnology-based Approaches for Bone Tissue Engineering	Universitat Politècnica de Catalunya	Maria-Pau Ginebra & Montserrat Español	March	Medical technologies: Biomaterials and Tissue Engineering
Jesica Fernández Cano	Modelo sistemático del proceso proyectual para una ingeniería que diseña mediante materiales avanzados	Universitat Politècnica de Catalunya	Jose M Manero Planella & Francisco Javier Peña Andres	March	Medical technologies: biomaterials and tissue engineering
Helena Castillo Écija	Development and application of xenograft models to predict the prognosis and study the treatment-driven evolution of pediatric sarcomas	Universitat de Barcelona	Angel Montero Carcaboso	March	Therapy development in pediatric oncology
Mar Bonany	Nanotechnology-based approaches for bone tissue engineering	Universitat Politècnica de Catalunya	Maria Pu Ginebra & Montserrat Español	March	Medical technologies: biomaterials and tissue engineering
Eva Mª Luján Luján	Valoración del grado de funcionamiento y de salud mental positiva en pacientes diagnosticados de esquizofrenia, atendidos en la Comunidad	Universitat de Barcelona	Mª Dolores Miguel & Edith Pomarol	March	Allergic diseases in childhood and adolescence

# Doctoral Theses

Thesis author	Title	University	Thesis directors	Month of lecture	Research group
Jorge Núñez Camarena	<b>Estudio comparativo de la marcha postoperatoria en artroplastia total de cadera. Abordaje anterior directo vs. Abordaje posterolateral</b>	Universitat Autònoma de Barcelona	Cesar García Fontecha & A. Hernández	March	Orthopaedic surgery and traumatology
Alba Castells-Sánchez	<b>Active aging. Healthy aging? A molecular, brain volume and behavioural approach</b>	Universitat de Barcelona	Maria Mataró	January	Neuropsychology
Cristina Gómez Pérez	<b>Clinically relevant gait parameters in children with bilateral spastic cerebral palsy</b>	Universitat de Vic – Universitat Central de Catalunya	Josep M. Font-Llagunes, Joan Carles Martori (UVIC) & Joan Vidal (Institut Guttmann)	January	Medical Technologies: Biomechanics

# 5

## CORE FACILITIES

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# Core facilities

The Institut de Recerca Sant Joan de Déu makes available to its researchers a set of fundamental services and infrastructures for the performance of their projects. In order to facilitate high-level research, the IRJSO offers its professionals technical, scientific and support services as well as the most suitable infrastructures required to carry out their work:

## Scientific core facilities

- Biological Sample Processing and Molecular Analysis
- Confocal Microscopy and Cellular Imaging Unit
- Clinical Trials Unit
- Library
- Methodology and statistics
- Neuroimaging
- Neurological tissues Biobank for research
- Neuropsychological evaluations
- Paediatric Biobank for research
- Patient Engagement in Research Area
- Sequencing Unit
- Animal housing facility
- Unit of Design, Analysis and Interpretation of Omic data (DAI0mics)

## Management support services

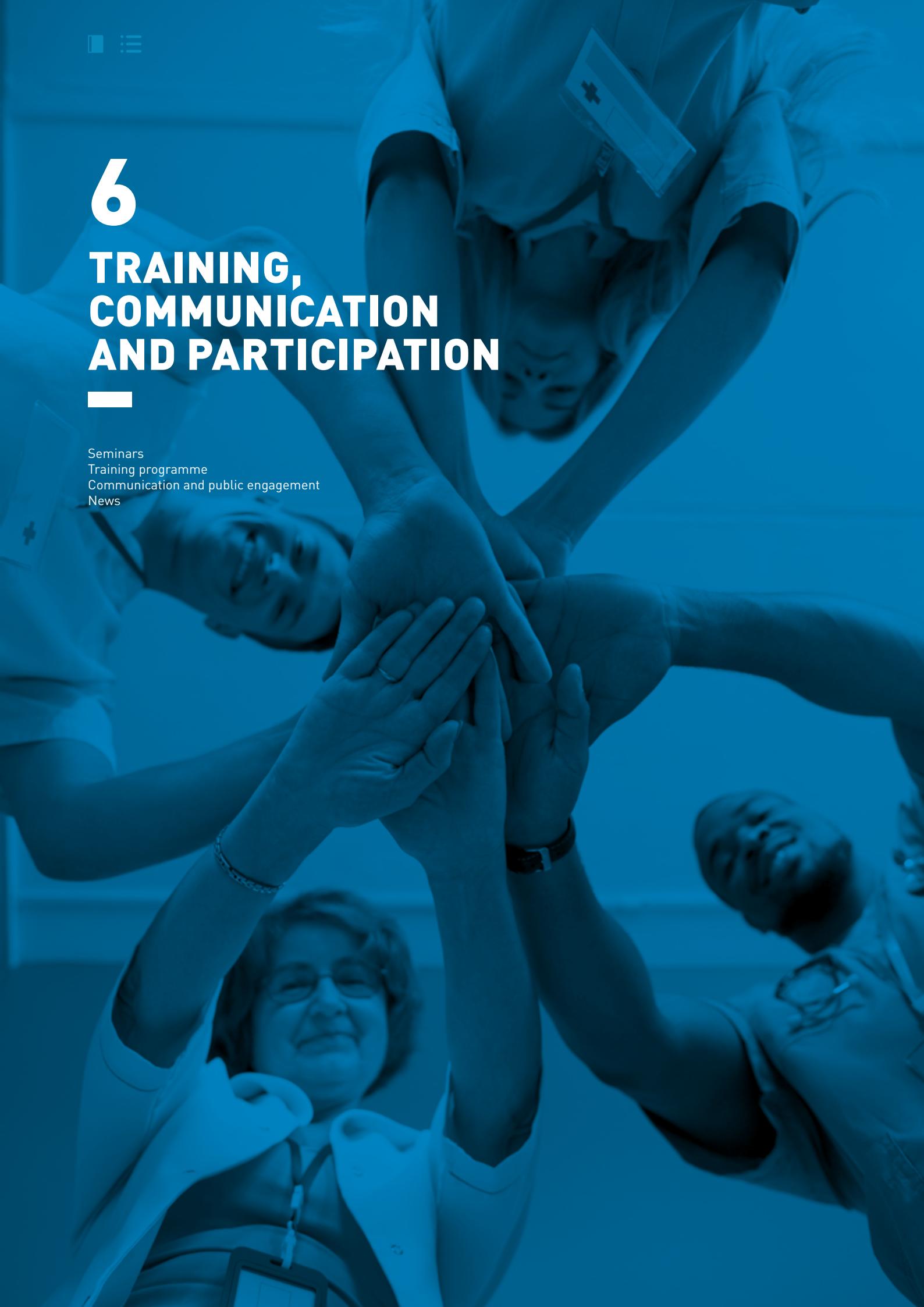
- Administration and corporate finance
- Human Area
- Clinical Research Ethics Committee
- Research management and Promotion
- Management of knowledge and communication



# 6

## TRAINING, COMMUNICATION AND PARTICIPATION

Seminars  
Training programme  
Communication and public engagement  
News



# Seminars

**19**  
SEMINARS

## Relation of research seminars and IRSJD day of debates

25 JANUARY

**Ciencia y COVID-19. Una perspectiva multidisciplinar**  
Dra. Clara Prats, BIOCOP-UPC

1 FEBRUARY

**Compassion practice quality:  
exploring new frontiers of self-criticism treatment**  
Dr Jaime Navarrete Hidalgo, IRSJD

15 FEBRUARY

**Obesidad infantil: una aproximación epigenómica**  
Dra. Marcela Parra-Vargas, IRSJD

23 FEBRUARY

**Model de síndrome alcohòlic fetal en ratolí segons dos patrons de consum humà i el paper de la EGCG en la seva prevenció**  
Dra. Laura Almeida, IRSJD

28 FEBRUARY

**Nano Rare Disease Day 2022**  
Organise: Nanomed Spain with IBEC & IRSJD

1 MARCH

**CRISPR-rescue of intracellular pathological phenotypes in congenital muscular disorders triggered by Collagen VI mutation**  
Dr. Enrico Castroflorio, ICFO

15 MARCH

**Emotional dysregulation, psychiatric disorders and trauma adolescents**  
Laia Vilalta, IRSJD

11 APRIL

**Novedades en el procesamiento de imagen y aplicación de la IA en endoscopia digestiva pediátrica**  
Dr. Víctor Vila, SJD Barcelona Children's Hospital

19 APRIL

**Systematic exploration of the contribution of dopamine and serotonin genes to psychiatric disorders**  
Dr. Bru Cormand, IRSJD

26 APRIL

**Vesícules extracel·lulars de la microbiota intestinal com a moduladors de la homeostasi intestinal i salut humana**  
Dra. Laura Baldomà, IRSJD

10 MAY

**Microbiota nasofaríngea en niños con enfermedad neumocócica invasiva, infección respiratoria banal y sanos: caracterización, factores moduladores y valor diagnóstico diferencial**  
Desireé Henares, IRSJD

24 MAY

**The Use of Deep Learning for Patient Stratification**  
Enrico Manzini, IRSJD

7 JUNE

**Estudio de pacientes dentro del espectro Rett mediante una aproximación multiómica: descifrando la huella molecular de la red funcional**  
Dra. Judith Armstrong, IRSJD

21 JUNE

**CERKL, a retinal dystrophy gene, regulates mitochondrial function and dynamics in both mammalian neuroretina and retinal pigment epithelium**  
Dra. Serena Mirra, IRSJD

5 JULY

**Reptes i oportunitats de la robòtica en cirurgia**  
Dra. Alicia Casals, IRSJD

13 SEPTEMBER

**Identification of gene signatures and potential therapeutic targets for acquired chemotherapy resistance in neuroblastoma**  
Dra. Marta García, IRSJD

27 SEPTEMBER

**Antimicrobial calcium phosphates**  
Dra. Montserrat Español, IRSJD

11 OCTOBER

**Towards personalizing psychological treatment in psychosis: the role of social cognition and metacognition**  
Marta Ferrer, IRSJD

25 OCTOBER

**Development of gene therapies for neuromuscular diseases**  
Dra. Ana Maria BujBello, INSERM

29 NOVEMBER

**IV Jornada de Debats IRSJD 2022**  
Salud Mental

# Training programme

**13**  
**COURSES IN 2022**

**246**  
**ATTENDEES IN 2022**

To promote the development of the different competence dimensions, four types of training modalities are established:

- Knowledge and techniques
- Soft Skills
- Organization and Governance
- Institutional training (Values)

## Courses

### February

Trabajo colaborativo a distancia y teletrabajo en Office 365 (Teams)

### February

Revisions sistemàtiques i meta-anàlisis (3<sup>a</sup> Ed)

### March

Introduction to scientific publishing & How to read a paper (1<sup>a</sup>Ed)

### March

Excel: Nivel intermedio (2<sup>a</sup> Ed)

### April

Habilidades Búsqueda Bibliográfica en PubMed

### April

Estadística: Nivell bàsic i nivell avançat

### May

Mindfulness (1<sup>a</sup>Ed)

### May

Introducción al Mindfulness para personal investigador (1<sup>a</sup>Ed)

### June

Metodología per projectes de recerca: Nivell bàsic i nivell avançat

### Septiembre

Taller de comunicación interpersonal (Bridge) (1<sup>a</sup>Ed)

### October

Getting the Right Person for your Team (1<sup>a</sup>Ed)

### October

Sharpen your reasoning skills: logic and critical thinking for scientists (1<sup>a</sup>Ed)

### November

Participación del Paciente en Recerca (2<sup>a</sup>Ed)

# Communication and public engagement

## Activities

**18**  
ACTIVITIES ORGANIZED

**170**  
RESEARCHERS

**1.000**  
AUDIENCE REACHED

## Internet and Social media

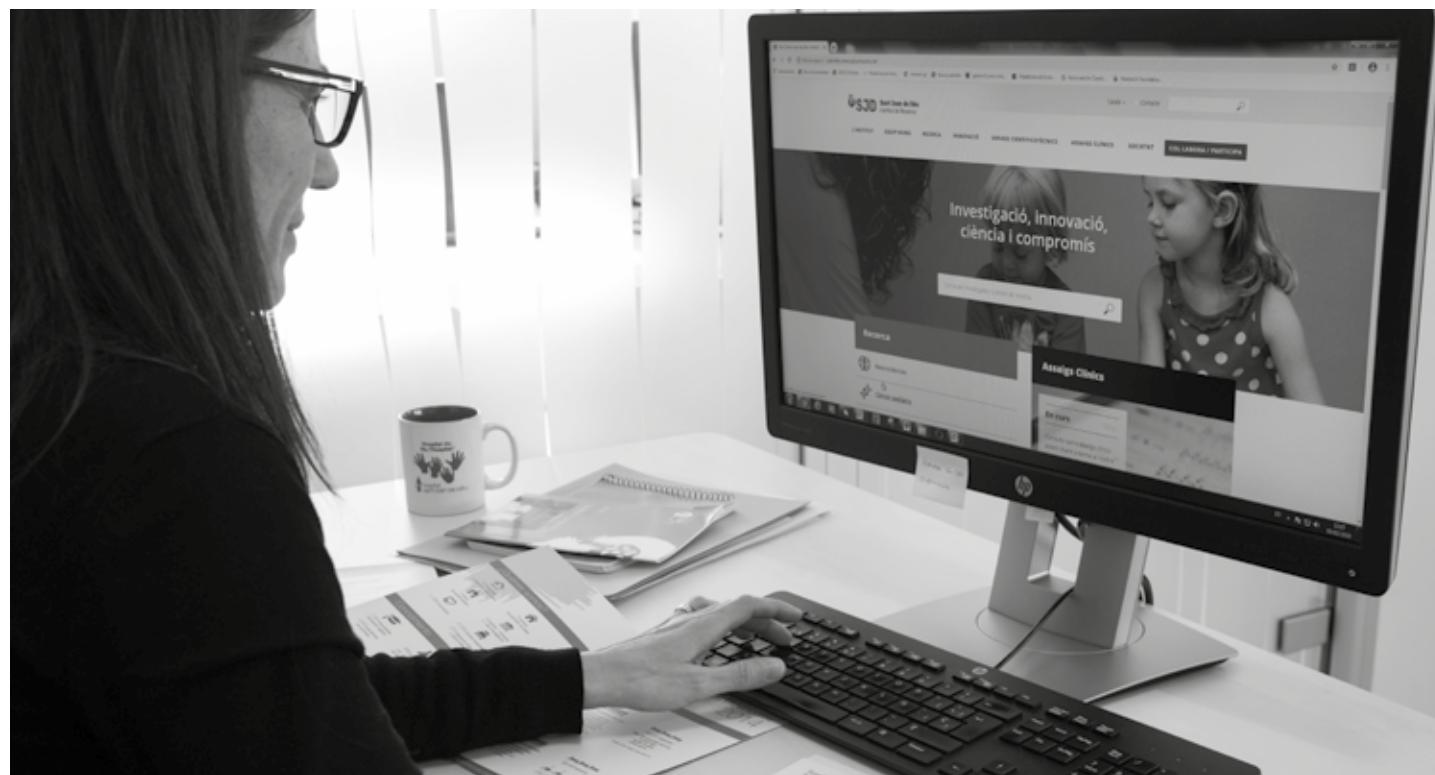
**LINKEDIN**  
**17.902** FOLLOWERS

**TWITTER**  
**1.597** FOLLOWERS

**WEBSITE**  
**47.275** USERS  
**209.231** NUMBER OF VISITS TO PAGES

Participation in:

- International Day of women and girls in science
- Europeans Researcher's Night
- Setmana de la Ciència
- Festa de la Ciència



# News

## **The overexpression of the CERKL gene, which is responsible for retinitis pigmentosa, protects the mitochondria in the retina from oxidative stress**

A new study led by Drs. Gemma Marfany and Serena Mirra has revealed that the overexpression of the CERKL gene, which is responsible for retinitis pigmentosa, protects the mitochondria in the retina from oxidative stress. This finding points to potential therapeutic targets for treating patients with mutations in this gene. Retinitis pigmentosa encompasses a group of genetically diverse diseases that result in the death of photoreceptors, leading to night blindness and tunnel vision. The study has demonstrated the expression of CERKL in the retinal pigment epithelium (RPE) and its partial colocalization with mitochondria. These findings could pave the way for therapies that improve the survival of photoreceptors and slow down retinal neurodegeneration.

## **Nanomedicine improves the efficacy of a drug against Ewing sarcoma**



A research team led by Dr. Ángel Montero Carcaboso has discovered that albumin nanoparticles can improve the bio-pharmaceutical

properties of paclitaxel, a cancer drug, and prolong its effectiveness in treating pediatric sarcomas like Ewing sarcoma. The study shows that tumors secreting a protein called SPARC have a better uptake of albumin nanoparticles. By interacting with SPARC, the nanoparticles enhance the concentration and prolonged presence of the drug within the tumor cells, leading to increased sensitivity to treatment. This finding offers a potential new approach for improving sarcoma treatment and opens up avenues for predicting the efficacy of the nanopharmaceutical in pediatric sarcomas.

## **61% of individuals with first-episode psychosis have experienced childhood trauma**



A study led by Dr. Judith Usall reveals that 61% of people experiencing their first episode of psychosis have a history of childhood trauma, including emotional, physical, and sexual abuse, as well as neglect. The research highlights the significant impact of childhood trauma on the development of severe mental disorders, particularly first-episode psychosis symptoms such as delusions and hallucinations. The study emphasizes the need for increased awareness and effective therapeutic interventions to address the influence of childhood trauma

on mental health, particularly in the context of early psychosis diagnosis and treatment.

## **Mitochondria-lysosome contacts: a new cell phenotype in neurogenetic diseases**



The Neurogenetics and Molecular Medicine research group at the Institut de Recerca Sant Joan de Déu (IRSJD) and the Neuromuscular Pathology Unit at SJD Barcelona Children's Hospital have conducted a study on the pathophysiology of mitochondrial dynamics and mitochondrial-lysosome contacts in patients with neurogenetic diseases. The research reveals alterations in the morphology of the mitochondrial network and lysosomes, as well as the identification of a new protein pair involved in mitochondrial-lysosome contacts. These findings shed light on the understanding of mitochondrial pathophysiology and provide potential biomarkers and therapeutic targets for neurogenetic diseases. Translational research using patient fibroblasts plays a crucial role in advancing clinical knowledge and treatment options for these conditions.

## **One in Ten Prescribed Pharmacological Treatments for Children Are Not Initiated**

A recent observational study conducted by researchers from the

# News

PRISMA research group at the Institut de Recerca Sant Joan de Déu (IRSJD) and the CIBER of Epidemiology and Public Health (CIBERESP) has found that 9% of pharmacological treatments prescribed to pediatric patients are not initiated. The study, published in Pediatrics, evaluated medication patterns in patients under 18 years old who received at least one new prescription across 28 drug subgroups. Factors influencing non-initiation varied depending on the type of medication, age group, and socioeconomic factors. The study highlights the need to address inequities in access to pharmacological treatments in pediatric populations.

## **Supplementation with Dopamine and Serotonin Precursors Could Have Neuroprotective Effects in Early-Onset Epilepsy Children**

A research team from the Sant Joan de Déu Research Institute has demonstrated that supplementation with neurotransmitters such as dopamine and serotonin could protect the brains of children under 3 years old with early-onset epilepsy. The study, published in the scientific journal Developmental Medicine & Child Neurology, focused on the analysis of cerebrospinal fluid in patients with epilepsy of early origin. Abnormal neurotransmitter levels were found in 33% of patients, especially in younger children. The study suggests that supplementation with neurotransmitter precursors may lead to a reduction in seizure frequency, severity, and improved neurological development in these patients. These findings have potential implications for clinical practice, recommending neurotransmitter level determination and supplementation in relevant cases.

## **Key Epigenetic Markers Identified in Vulnerability to Develop Food Addiction**

Researchers, including Drs. Bru Cormand and Noèlia Fernàndez from the Sant Joan de Déu Research Institute, have identified common epigenetic mechanisms in rodents and humans related to food addiction. The study, led by researchers from Pompeu Fabra University and the Biomedical Research Institute of Girona Dr. Josep Trueta, revealed the involvement of specific epigenetic markers, including microRNAs, in cortical brain regions associated with food addiction. The study's findings offer insights into the development of food addiction and may contribute to the identification of biomarkers for early diagnosis and future therapeutic interventions. The research highlights the role of epigenetics in understanding vulnerability to food addiction.

## **The hidden cells behind B-cell acute lymphoblastic leukemia relapse spotlighted for the first time**

Dr. Pablo Menéndez, along with Drs. Susana Rives, Montserrat Torrebadell, and Nerea Vega from the Sant Joan de Déu Research Institute, led a study revealing a previously unnoticed population of pre-leukemic cells that may be responsible for relapses in B-cell Acute Lymphoblastic Leukemia (B-ALL). The research, published in the hematological journal Blood, identified a subset of CD19-negative (CD19-/CD22+) cells that coexist with CD19-positive leukemic cells. These CD19-negative cells, which are not targeted by CD19-directed immunotherapies, possess similar mutations to the primary leukemia and could contribute to relapses. The findings suggest the potential use of

dual therapies targeting both CD19 and CD22 to reduce relapse rates and extend remission in B-ALL patients.

## **CRISPR/Cas9 Gene Editing Offers a New Therapeutic Strategy for Congenital Muscular Dystrophies with Collagen VI Deficiency**



A study published in the International Journal of Molecular Sciences introduces a novel therapeutic approach for patients with congenital muscular dystrophies caused by collagen VI deficiency. The study, led by Drs. Cecilia Jimenez-Mallabriga and Aristides López-Márquez from the Sant Joan de Déu Research Institute (IRSJD), employed CRISPR/Cas9 gene editing to target specific mutations in the COL6A1 gene, enhancing the collagen VI fiber network in fibroblasts. By silencing dominant mutations, the technique aims to improve the assembly of collagen VI, offering potential treatment prospects for these currently untreatable muscle disorders. The research involved collaborations with the Ramón y Cajal Institute for Health Research, the University of Barcelona Biomedical Research Institute, and the Biomedical Research Network on Rare Diseases (CIBERER).

# News

## Dr. Cristina Canal Develops Innovative Treatment for Bone Cancer



Dr. Cristina Canal, a researcher from the Sant Joan de Déu Research Institute (IRSJD) and the CREB-UPC, has developed a new therapy for bone cancer that avoids the side effects of conventional treatments, such as chemotherapy, while promoting bone regeneration after surgical tumor resection. The project, called TRANSFORMER (Transforming bone cancer therapy with composite biomaterials encapsulating plasma-generated RONS), combines biomaterials for bone regeneration with an innovative treatment using gas plasma-treated hydrogels that induce the death of cancer cells. This research aims to improve survival rates for osteosarcoma patients, especially those with metastatic tumors. The Proof of Concept grant from the European Research Council (ERC) will support the advancement of this technology towards clinical development and commercial viability.

## Prevalence of Suicidal Thoughts and Behavior Among Spanish Adults During Confinement Was 4.5%

A study published in Current Psychology, led by researchers from the Sant Joan de Déu Research Institute and the Sant Joan de

Déu Health Park, reveals that 4.5% of the Spanish population exhibited suicidal behaviors during the confinement period. Within this group, 60% reported passive behaviors such as wishing to be dead, while 40% indicated suicide attempts or planning. Previous studies have reported similar figures when assessing suicidal behaviors in the Spanish adult population over a lifetime. Factors such as social disconnection and emotional disorders like depression or anxiety contribute to the increased likelihood of suicidal thoughts and behavior. The study highlights the importance of implementing preventive interventions that address both the individual and community dimensions of social relationships, providing psychological assistance to those experiencing social disconnection and improving their opportunities for satisfying social connections.

## Genomics: A Tool for the Development of Vaccines Against Bacterial Infections

A groundbreaking genomic surveillance study published in Lancet Microbe has provided valuable insights into the ongoing battle between *Streptococcus pneumoniae*, a bacterium responsible for pneumonia and meningitis, and vaccines designed to protect against dominant strains. The study revealed that a strain called GPSC10 posed a significant threat due to its ability to transform its structure, evading vaccines, and its high virulence, coupled with resistance to common antibiotics. The research underscores the importance of genomic surveillance in vaccine design and highlights the challenges posed by highly adaptable pneumococcal strains. *Streptococcus pneumoniae* is a major cause of infant mortality worldwide, and effective strategies to combat the bacterium are crucial.

## First clinical guideline on Schaaf-Yang syndrome presented for professionals and families

The first clinical guideline aimed at healthcare professionals and families of children affected by Schaaf-Yang syndrome (SYS) has been developed to improve understanding of this ultra-rare disease caused by mutations in the MAGEL2 gene. The research, published in the Journal of Medical Genetics, provides insights into the effects of truncated MAGEL2 protein on cell physiology. The guideline, based on a project led by researchers from various institutions, addresses the diverse symptoms and clinical features of SYS, including neurodevelopmental delay, intellectual disability, sleep disorders, facial dysmorphia, and joint contractures. The collaboration with families and associations is emphasized as crucial in rare disease research.

## Environmental exposure to nitrogen oxides linked to severity of rhinovirus infection



A study led by the Sant Joan de Déu Research Institute (IRSJD) and the Sant Joan de Déu Barcelona Hospital has found that higher exposure to atmospheric nitrogen oxides during childhood is associated with more severe rhinovirus infections and an increased number of admissions

# News

to the Pediatric Intensive Care Unit (PICU). The research, published in the journal *Pediatric Pulmonology*, analyzed data from nine years and identified a correlation between elevated levels of nitrogen oxides and ICU admissions three days after the exposure. Understanding the impact of environmental factors on respiratory virus infections like rhinovirus can contribute to better mitigation strategies.

## **Dr. Alexandra Avgustinova awarded prestigious scientific grant from the European Research Council (ERC)**



The ERC Starting Grant, totaling approximately 1.5 million euros, will fund the development of the project "Oncogenic competence during development - When, Where and Why?" led by Dr. Alexandra Avgustinova of the Sant Joan de Déu Research Institute (IRSJD). The research aims to understand the origins of malignant rhabdoid tumors, a rare and aggressive form of cancer primarily diagnosed in children under 2 years old. By employing a multidisciplinary approach and utilizing patient cell models, cell cultures, bioinformatics, genomics, transcriptomics, and gene editing, the project seeks to unravel the factors that determine the oncogenic potential of cells carrying the SMARCB1 gene mutation. The findings could

lead to more targeted and less toxic treatment strategies for pediatric cancer patients.

## **Genetic alteration responsible for COVID-19-induced multisystem inflammatory syndrome in children discovered**



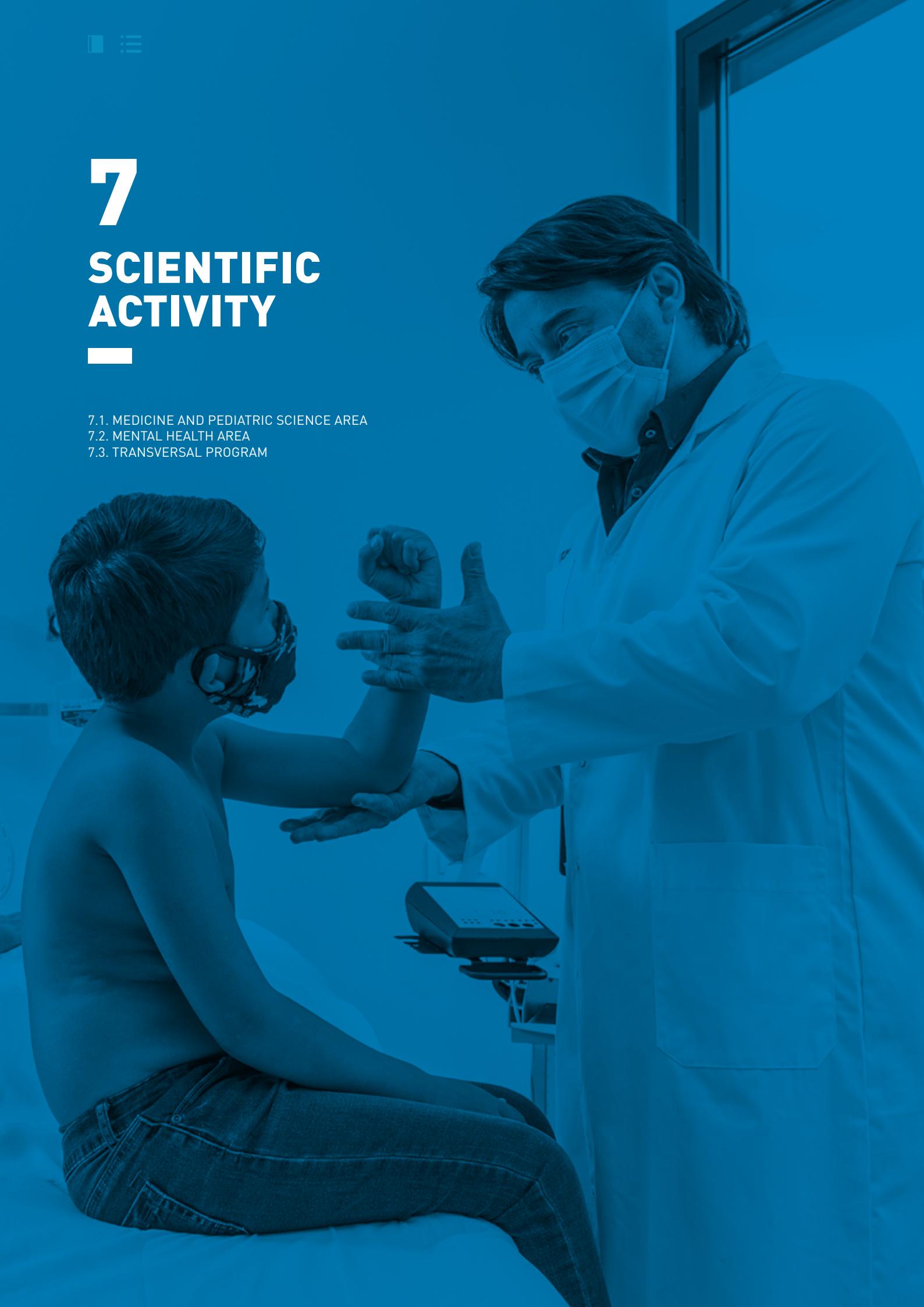
An international study, involving researchers from the Sant Joan de Déu Research Institute, has identified genetic mutations in three genes (OAS1, OAS2, and RNASEL) in five pediatric patients with multisystem inflammatory syndrome in children (MIS-C) caused by SARS-CoV-2, the virus responsible for COVID-19. These mutations, published in the journal *Science*, provide insight into why some patients, particularly children, develop this syndrome weeks after being infected with the virus. The findings contribute to understanding the genetic and immunological factors that impact the severity of the disease and pave the way for personalized treatments based on genetic and molecular defects.

# 7

## SCIENTIFIC ACTIVITY

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- 7.1. MEDICINE AND PEDIATRIC SCIENCE AREA
- 7.2. MENTAL HEALTH AREA
- 7.3. TRANSVERSAL PROGRAM



# 7.1

## MEDICINE AND PEDIATRIC SCIENCE AREA

### 7.1.1 **Neuroscience**

Applied research in neuromuscular diseases

Pediatric neurometabolism: neural communication mechanisms  
and personalized therapies

Neonatal brain

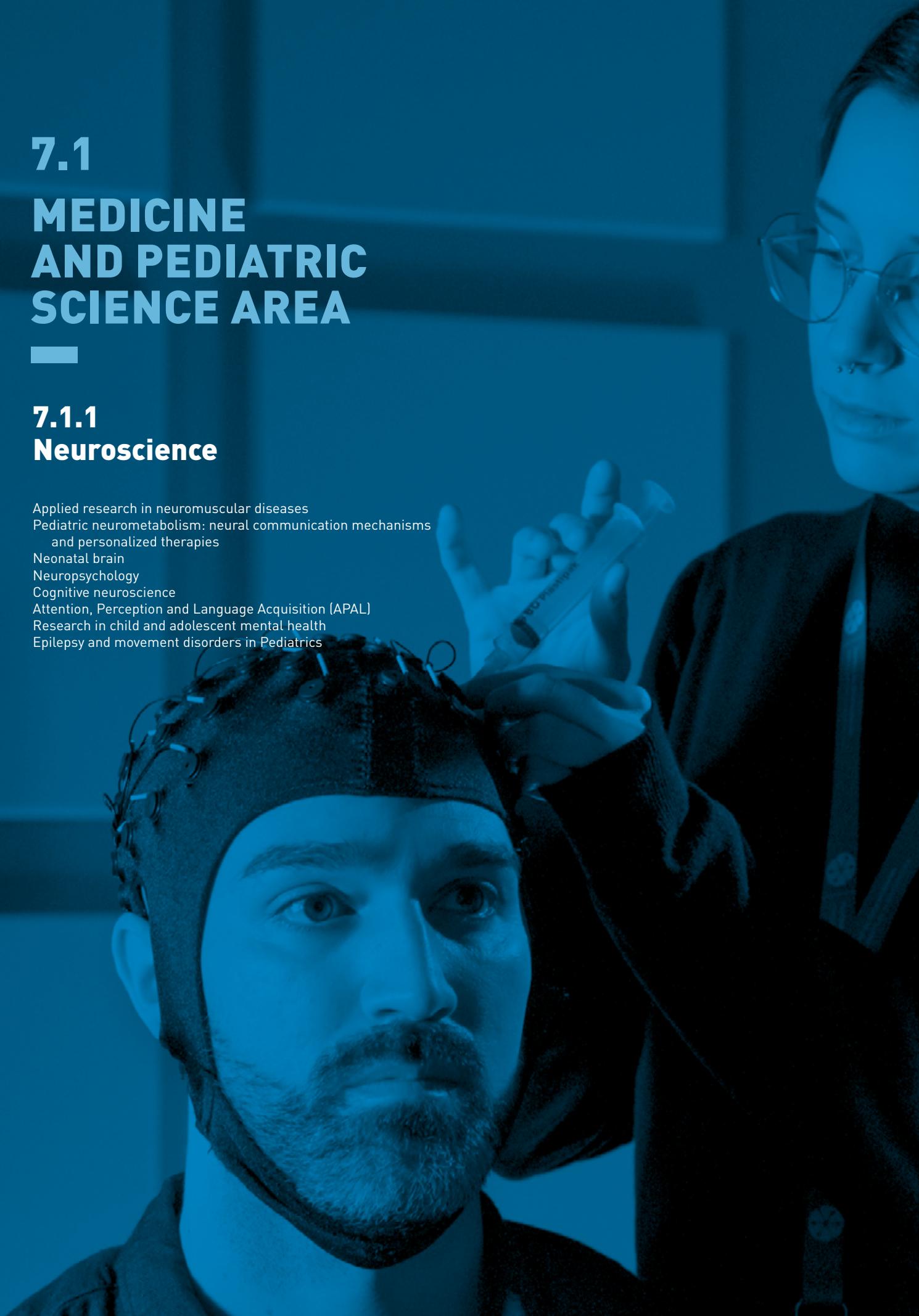
Neuropsychology

Cognitive neuroscience

Attention, Perception and Language Acquisition (APAL)

Research in child and adolescent mental health

Epilepsy and movement disorders in Pediatrics



## Applied research in neuromuscular diseases

- 17** COMMERCIAL CLINICAL TRIALS
- 1** ACADEMIC CLINICAL TRIALS
- 18** PROJECTS
- 25** PUBLICATIONS
- 218.78** IMPACT FACTOR
- 52%** PUBLICATIONS IN 1RTS QUARTIL
- 12%** PUBLICATIONS IN 1RTS DECIL
- 60%** LEADERSHIP

### Cecilia Jiménez Malebrera, PhD

In 2022 we started a collaboration with the Institute of Material Sciences in Barcelona after obtaining competitive funding from the Orphan Disease Centre to investigate a new family of lipid nanoparticles as vehicles for the targeted delivery of nucleic acids to treat collagen VI-related dystrophies. This is a significant step in the translational therapeutic pathway that we started 4 years ago using CRISPR/Cas9 technology and which was described in Dr. López-Márquez publication also this year / doi: 10.3390/ijms23084410.). Dr. C. Jimenez-Mallebrera spent 3 months at the Institute de Myologie in Paris which has allowed us to establish strategic alliances with Inserm and Genethon. In the second half of the year, Dr. D. Natera de Benito was awarded a Miguel-Servet Fellowship from the ISCIII and Dr. C. Jou obtained her PhD. Work has continued on the Arm Tracker funded by the Marató Foundation as well as on the RD Solve project which has allowed us to establish a genetic diagnosis in 14 out of 45 patients without diagnosis.

The number of Clinical Trials and observational studies in which we participate has increased due to the new potential treatments available mainly for Duchenne Muscular Dystrophy and Spinal Muscular Atrophy including the administration

of a gene therapy in DMD boys for the first time as part of a Clinical Trial in which we participate as the sole centre in Spain. We are also participating in a Clinical trial assessing the combined effect of Gene Therapy and Antisense Oligonucleotides for the treatment of SMA. We have participated in various national and international meetings. To highlight the Elsevier prize awarded to Dr. Natera de Benito at the World Muscle Society Congress in Halifax (Canada) for best poster presentation and the best poster prize awarded to Dr. Julita Medina communication at the EACD congress in Barcelona.

A paper that we published in 2021 was awarded the prize for the best paper by the Spanish Society of Laboratory Medicine- José Luis Castaño Foundation (doi: 10.1093/clinchem/hvab091. PMID: 34352085).

### MEMBERS

#### STAFF RESEARCHER

Julita Medina, Irene Beatriz Zschaeck Luzardo, Andrés Nascimento Osorio, Carlos Ignacio Ortez Gonzalez

#### POSTDOC ASSOCIATED RESEARCHERS

Arístides Lopez Marquez, Selena Trifunov, Daniel Natera de Benito

#### PHD STUDENTS

Cristina Jou Muñoz

#### ASSISTANT RESEARCHER

Cristina Puig Ram, Sonia Segovia Simon, Jessica Maria Exposito Escudero, Laura Carrera García, María del Carmen Badosa Gallego

#### TECHNICIAN

Ruth Lavilla Hidalgo, Anna Codina Bergadà

### SELECTED PROJECTS

- ArmTracker: A state-of-the-art wearable system to assess upper limb motor function in real life conditions for patients with Duchenne muscular dystrophy and spinal muscular atrophy. [561/C/2020]. Fundació La Marató de TV3. 2021-2024. IP: Daniel Natera de Benito.
- DYSTRO-SMARTY: A novel nanotechnology platform for delivery of nucleic acids to treat Collagen VI-related Congenital Muscular Dystrophy. Fundación Noelia. Niños Contra la Distrofia Muscular Congénita por Déficit de Colágeno VI. IP: Cecilia Jiménez Mallebrera.
- Cohort of Spanish Patients with Nonsense Mutation Duchenne Muscular Dystrophy (nmdMD). PTC Therapeutics International Limited.

2022-2024. IP: Carlos Ignacio Ortez Gonzalez.

### SELECTED CLINICAL TRIALS & OBSERVATIONAL STUDIES

- Estudio de fase III multinacional, aleatorizado, doble ciego, controlado con placebo, de liberación génica sistémica para evaluar la seguridad y la eficacia de SRP-9001 en sujetos con distrofia muscular de Duchenne. [SRP-9001-301]. Merchán; Sarepta Therapeutics, Inc. 2022. IP: Andrés Nascimento Osorio.
- Estudio de Fase 4 de Nusinersen (BIIB058) en Pacientes con Atrofia Muscular Espinal que Recibieron Onasemnogene Abeparvovec. [232SM404]. Biogen IDEC Research LTD; IQVIA RDS Spain SL; Quispe. 2021. IP: Andrés Nascimento Osorio.
- Estudio de fase 3, multicéntrico, aleatorizado, doble ciego, controlado con placebo para evaluar la seguridad y eficacia de PF-06939926 en el tratamiento de la distrofia muscular de Duchenne. [C3391003]. Altimira; PFIZER INC; Pharmaceutical Product Development Spain, S.L. 2020. IP: Andrés Nascimento Osorio.

### SELECTED PUBLICATIONS

- Castroflorio E, Pérez AJ, López A, Badosa MC, Loza P, Roldan M et al. The Capillary Morphogenesis Gene 2 Triggers the Intracellular Hallmarks of Collagen VI-Related Muscular Dystrophy. *Int J Mol Sci.* 2022;23(14):7651. IF:6,208. (Q1).
- Jou C, Nascimento A, Codina A, Montoya J, López E, Emperador S et al. Pathological Features in Paediatric Patients with TK2 Deficiency. *Int J Mol Sci.* 2022;23(19):11002. IF:6,208. (Q1).
- López A, Morín M, Fernández S, Badosa MC, Hernández A, Natera D et al. CRISPR/Cas9-Mediated Allele-Specific Disruption of a Dominant COL6A1 Pathogenic Variant Improves Collagen VI Network in Patient Fibroblasts. *Int J Mol Sci.* 2022;23(8): 4410. IF:6,208. (Q1).
- Natera D, Jurgens JA, Yeung A, Zaharieva IT, Manzur A, DiTroia SP et al. Recessive variants in COL25A1 gene as novel cause of arthrogryposis multiplex congenita with ocular congenital cranial dysinnervation disorder. *Hum Mutat.* 2022;43(4):487-498. IF:4,700. (Q2).

# Pediatric neurometabolism: neural communication mechanisms and personalized therapies

-  **13** COMMERCIAL CLINICAL TRIALS
-  **0** ACADEMIC CLINICAL TRIALS
-  **14** PROJECTS
-  **27** PUBLICATIONS
-  **157.22** IMPACT FACTOR
-  **52%** PUBLICATIONS IN 1RTS QUARTIL
-  **30%** PUBLICATIONS IN 1RTS DECI
-  **44%** LEADERSHIP

## M<sup>a</sup> Angels García Cazorla, MD, PhD

During 2022, our team has been awarded a number of competitive and private projects. These have contributed to strengthening our line of research in neurometabolic diseases and the modulation of brain metabolism as a therapeutic strategy in rare neurogenetic diseases. On the one hand, it has allowed us to start developing a "synapse-on-a-chip" model to test in vitro treatments for various diseases that affect neuronal communication. On the other hand, we have obtained projects that allow us to develop treatments proposed by our team for Rett syndrome and infantile Parkinsonism.

In addition to this, we have continued our activity in personalised therapies and have developed a "Neuroprotect" formula which is in the process of being patented, which we have applied in a clinical trial in Rett syndrome and other genetic neurodevelopmental diseases.

Numerous clinical trials have been carried out by our team. These include the use of metabolic treatments in Grinopathies, X-ADL (X-linked adrenoleukodystrophy), Friedreich's Ataxia and fatty acid beta-oxidation

disorders. In addition, an enzyme treatment that crosses the blood-brain barrier has been initiated in Hunter syndrome.

Finally, numerous scientific articles have been published, including the development of a clinical algorithm in a metabolic neurotransmitter disease (non-ketotic hyperglycinemia), the description of neurotransmitter depletion in epileptic encephalopathies and its possible implication in the development of therapies, and the description and new approach to cellular trafficking diseases.

## MEMBERS

### STAFF RESEARCHER

Roser Colomé Roura, Silvia Maria Meavilla Olivas, Alejandra Darling, Jordi Muchart López, M<sup>a</sup>del Mar O'Callaghan Gordo, Federico Ramos

### POSTDOC ASSOCIATED RESEARCHERS

Alfonso Luis De Oyarzabal Sanz

### PHD STUDENTS

Juliana Ribeiro Constante, Uliana Musokhranova Matveeva, Natalia Alexandra Julià Palacios

### ASSISTANT RESEARCHER

Júlia Romagosa I Pérez, Jennifer Anticona Huaynate, Leticia Diana Pias Peleteiro, Ariadna Borras Martínez, Rosanna Marí Vico, Silvia Constancia Liendo Vallejo, Mariya Sigatullina Bondarenko

### TECHNICIAN

Anna Serradell Salís, Cristina Grau Páez

## SELECTED PROJECTS

- Development of leriglitazone for Rett Syndrome and related disorders. (CPP2021-008554). Ministerio De Ciencia E Innovacion. 2022-2025. IP: Alfonso Luis De Oyarzabal Sanz.
- Sinapsis en un chip: medicina personalizada en trastornos genéticos del neurodesarrollo que afectan a la comunicación sináptica. (PI21/00073). Instituto de Salud Carlos III (ISCIII). 2022-2024. IP: M<sup>a</sup> Angels García Cazorla.
- Developing patient-tailored therapeutic candidates for rare genetic forms of pediatric parkinsonism. (202012-30). Fundació La Marató de TV3. 2021-2024. IP: M<sup>a</sup> Angels García Cazorla.

## SELECTED CLINICAL TRIALS & OBSERVATIONAL STUDIES

- Estudio multicéntrico, aleatorizado y doble ciego para determinar

el efecto de la triheptanoína en comparación con triglicéridos de cadena media (TCM) par sobre los episodios clínicos importantes (ECI) en pacientes pediátricos con trastornos de la oxidación de ácidos grasos de cadena larga (LC-FAOD). (UX007-CL302). Rodriguez; Ultragenyx Pharmaceutical, Inc. 2022IP: M<sup>a</sup> Angels García Cazorla.

- ESTUDIO EN FASE III DE JR-141 EN PACIENTES CON MUCOPOLISACARIDOSIS TIPO II (SÍNDROME DE HUNTER) A Phase III study of JR-141 in Mucopolysaccharidosis type II (Hunter Syndrome) patients. (JR-141-GS31). Bermejo; JCR Pharmaceuticals Co., Ltd.; Parexel International S.L. 2022IP: M<sup>a</sup> del Mar O'Callaghan Gordo.
- AN EXPLORATORY, OPEN-LABEL, MULTICENTER STUDY IN MALE PEDIATRIC PATIENTS WITH CEREBRAL X-LINKED ADRENOLEUKODYSTROPHY (CALD) TO ASSESS THE EFFECTS OF MIN 102 TREATMENT ON THE PROGRESSION OF CEREBRAL LESIONS. (MT-2-02). CTI Clinical Trial & consulting services Spain S.L; Hernández; Minoryx Therapeutics S.L. 2019IP: M<sup>a</sup> Angels García Cazorla.
- A DOUBLE-BLIND, PLACEBO-CONTROLLED STUDY ON THE EFFECTS OF MIN-102 ON BIOCHEMICAL, IMAGING, NEUROPHYSIOLOGICAL, AND CLINICAL MARKERS IN PATIENTS WITH FRIEDREICH'S ATAXIA. (MT-2-03). Minoryx Therapeutics BE SA. 2019IP: Alejandra Darling.

## SELECTED PUBLICATIONS

- Kuseyri O, Julià-Palacios N, Olivella M, Guder P, Zafeiriou DI, Horvath G et al. Integrative Approach to Predict Severity in Nonketotic Hyperglycinemia. Ann Neurol. 2022;92(2):292-303. IF:11,274. (Q1).
- Julià-Palacios NA, Molina C, Sigatullina M, Cortés E, Aparicio J, Cuadras D et al. Monoamine neurotransmitters in early epileptic encephalopathies: New insights into pathophysiology and therapy. Dev Med Child Neurol. 2022;64(7):915-923. IF:4,864. (Q1).
- Garcia A, De Oyarzabal AL, Saudubray JM, Martinelly D, Dionisi C. Genetic disorders of cellular trafficking. Trends Genet. 2022;38(7):724-751. IF:11,821. (Q1).

## Neonatal brain

- 1** COMMERCIAL CLINICAL TRIALS
- 0** ACADEMIC CLINICAL TRIALS
- 2** PROJECTS
- 7** PUBLICATIONS
- 61.90** IMPACT FACTOR
- 71%** PUBLICATIONS IN 1RTS QUARTIL
- 29%** PUBLICATIONS IN 1RTS DECIL
- 71%** LEADERSHIP

**Thais Agut Quijano, MD, PhD & Ana Alarcon Allen, MD, PhD**

The Neonatal Brain Group has upheld its commitment to generating knowledge about brain disorders affecting children's health and capacities in the foetal/neonatal period.

The group's main active lines of research continue to be hypoxic-ischaemic encephalopathy, stroke, extrauterine maturation of the preterm brain, foetal and neonatal infections affecting the developing brain, and care practices for newborns with neurological pathology or risk and their families.

Likewise, the group continues to collaborate with other specialists and researchers in areas relating to neonatal neurology, as well as with groups of the Institut de Recerca Sant Joan de Déu and of other institutions and universities. Our projects have fed on multiple approaches (clinical practice of doctors or nurses, basic science, biomedical engineering, epidemiology), all of which favour the translational character of our research activities.

Through our participation in large-scale multi-centre clinical trials (such as the SafeBoosC-III trial on near-infrared spectroscopy in premature newborns), we seek to

incorporate the results of research into clinical practice. Our innovation and assessment activities notably include our collaboration on a project for the development of ultrasound-based non-invasive technology for the diagnosis of neonatal meningitis. We also participate in AI4NICU Action, a pan-European multidisciplinary (clinical and technical) network focused on implementing artificial intelligence-based neuromonitoring tools in neonatal units in order to support the clinician's decision-making process.

### MEMBERS

#### STAFF RESEARCHER

Sílvia Planas Román, Nuria Herranz Rubia, María del Mar Velilla Aparicio

#### POSTDOC ASSOCIATED RESEARCHERS

Nuria Carreras Blesa

#### TECHNICIAN

Nerea Liñan Rosa

### SELECTED PROJECTS

- Estudio CRIB (Cribado de citomegalovirus congénito en recién nacidos sanos e ingresados - Barcelona). (PI19/00095). Instituto de Salud Carlos III (ISCIII). 2020-2023. IP: Antoni Noguera Julián, Ana Alarcon Allen.
- Elaboración de un atlas morfométrico normalizado de la maduración cerebral del recién nacido prematuro y valoración del impacto de la patología cerebral en el proceso madurativo. (PI18/00110). Instituto de Salud Carlos III (ISCIII). 2019-2023. IP: Thais Agut Quijano.
- CA20124 – AI4NICU Action: “Maximising impact of multidisciplinary research en early diagnosis of neonatal brain injury”. COSTE (European Cooperation in Science and Technology), Horizon 2020. (INFANT Research Centre). 2021-2025. Ana Alarcón.

### SELECTED CLINICAL TRIALS & OBSERVATIONAL STUDIES

- UNITED Meningitis - Ultrasound Non-Invasive Technology for Early Diagnosis of Meningitis Newborn Solutions. (Hospital Sant Joan de Déu). 2020-2023. Estudio clínico. Ana Alarcón, Thais Agut, Nuria Carreras.
- Pre-Wallerian degeneration according to topographic

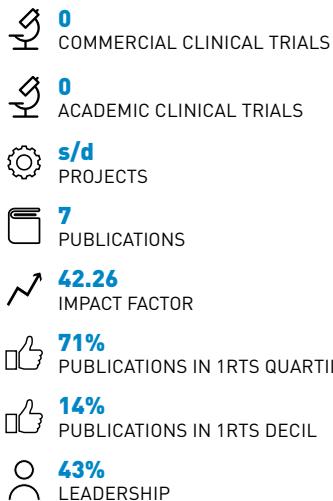
distribution of Neonatal Arterial Ischemic Stroke. Estudio observacional. IP: Thais Agut

- “Eficacia y tolerabilidad de la suplementación con nutraceuticos en pacientes prematuros que presentan trastornos del neurodesarrollo”, PIC-158-21. Ensayo clínico. IP Angels García-Cazorla. Thais Agut, colaboradora.
- Neuroimaging in infants with congenital cytomegalovirus and its correlation with outcome: emphasis on white matter disease. 2021-2023. Estudio observacional. IP: Ana Alarcón.
- Automatic segmentation of sylvian fissure in brain ultrasound images of preterm infants. Investigadoras: Thais Agut, Nuria Carreras.

### SELECTED PUBLICATIONS

- Agüera T, Carreras N, Agut T. Cerebral Ultrasonography of Cytotoxic Edema in a Newborn With a Mitochondrial Disorder. JAMA Neurol. 2022;79(7):722-723. IF:29,907. (Q1).
- Arnaez J, Herranz N, García-Alix A, ESP-EHI Group. Perinatal hypoxic-ischaemic encephalopathy: a national survey of end-of-life decisions and palliative care. BMJ Support Palliat Care. 2022;12(e6):e771-4. IF:4,633. (Q1).
- Nuñez C, Stephan C, Arca G, Agut T, Arnaez J, Cordeiro M et al. Neonatal arterial stroke location is associated with outcome at 2 years: a voxel-based lesion-symptom mapping study. Arch Dis Child Fetal Neonatal Ed. 2022;107(1):45-50. IF:6,643. (Q1).
- Schmidt G, Pillay K, Adams E, Alarcon A, Andritsou F, Cobo MM et al. The impact of premature extrauterine exposure on infants' stimulus-evoked brain activity across multiple sensory systems. Neuroimage Clin. 2022;33:102914-102914. IF:4,891. (Q2).

# Neuropsychology



## Roser Pueyo Benito, PhD

During 2022, the group made further advancements in the study of neuropsychological functioning in individuals with risk factors such as obesity, as well as in the neuropsychological consequences of congenital, perinatal, and/or acquired injuries (including cerebral palsy and stroke) and their rehabilitation. Projects funded through national competitive calls also continued, and the ICREA-Acadèmia program continued to support one of the team's researchers. In addition, a Marie Skłodowska-Curie Actions Postdoctoral Fellowship for the evaluation of autism spectrum disorder in children with cerebral palsy was submitted and successfully funded in 2022. The selected projects and publications demonstrate that the adopted neuropsychological approach enables the development of neuropsychological assessment guidelines and an exploration of the underlying mechanisms of these diseases, including the analysis of neuroimaging, genetics, microbiota, and more. Furthermore, the group is actively studying the effectiveness of interventions designed to improve cognitive and emotional function, such as physical exercise, mindfulness, and cognitive training. Notably, the group's prominent publications also highlight its ability to respond to emerging challenges, such as understanding

the neuropsychological sequelae of post-COVID situations.

## MEMBERS

### STAFF RESEARCHER

Isabel García García, María Mataró Serrat, Xavier Caldú Ferrús, María Angeles Jurado Luque

### POSTDOC ASSOCIATED RESEARCHERS

Cristina Sánchez Castañeda

### PHD STUDENTS

Anna Prunell Castañe, Adrià Bermudo Gallaguet, Montse Blasco Sierra, María García Galant, Sandra Luis Ruiz, Xavier Prats Soteras, Noemí Lamonja Vicente, Júlia Ballester Plané, Olga Laporta Hoyos, Jonatan Ottino González

### ASSISTANT RESEARCHER

Gemma Monté Rubio

## SELECTED PROJECTS

- Inflamación, estrés y microbiota como mediadores entre la obesidad y el funcionamiento cerebral durante la adolescència. PSI2017-86536-C2-1-R. Ministerio de Economía y Competitividad. Convocatòria d'ajuts del Programa Estatal d'Investigació, Desenvolupament i Innovació Orientada als reptes de la Societat. Modalitat Projectes I+D+I. PI: María Angeles Jurado Luque. 2018-2022
- Ajut per incentivar i consolidar la recerca d'excel·lència ja existent a les universitats públiques de Catalunya. Programa ICREA Academia 2018. Fundació Institució Catalana de Recerca i Estudis Avançats (ICREA). Convocatòria del Programa Icrea Acadèmia. PI: María Mataró Serrat. 2019-2023.
- Guias basadas en la evidencia y consensuadas para la evaluación neuropsicológica de las personas con parálisis cerebral grave y parálisis cerebral discinética. PID2020-117163RB-I00. Ministerio de Economía y Competitividad. Convocatòria d'ajuts del Programa Estatal d'Investigació, Desenvolupament i Innovació Orientada als reptes de la Societat. Modalitat Reptes. PI: María del Roser Pueyo Benito. 2021 - 2025

## SELECTED PUBLICATIONS

- Ariza M, Cano N, Segura B, Adan A, Bargalló N, Caldú X et al. Neuropsychological impairment in

post-COVID condition individuals with and without cognitive complaints. *Front Aging Neurosci.* 2022;14:1029842-1029842. IF:4,800. (Q2).

- Bermudo A, Ariza M, Dacosta R, Agudelo D, Camins N, Boldó M et al. Effects and mechanisms of mindfulness training and physical exercise on cognition, emotional wellbeing, and brain outcomes in chronic stroke patients: Study protocol of the MindFit project randomized controlled trial. *Front Aging Neurosci.* 2022;14:936077-936077. IF:4,800. (Q2).
- Forné S, López A, Mateu R, Adan A, Caldú X, Rifà X et al. Improving Reading Skills Using a Computerized Phonological Training Program in Early Readers with Reading Difficulties. *Int. J. Environ. Res. Public Health.* 2022;19(18) IF:4,614. (Q1).
- García I, Michaud A, Jurado MA, Dagher A, Morys F. Mechanisms linking obesity and its metabolic comorbidities with cerebral grey and white matter changes. *Rev. Endocr. Metab. Disord.* 2022;23(4):833-843. IF:8,200. (Q1).

## Cognitive neuroscience

- 0** COMMERCIAL CLINICAL TRIALS
- 0** ACADEMIC CLINICAL TRIALS
- s/d** PROJECTS
- 6** PUBLICATIONS
- 24.49** IMPACT FACTOR
- 71%** PUBLICATIONS IN 1RTS QUARTIL
- 14%** PUBLICATIONS IN 1RTS DECIL
- 43%** LEADERSHIP

### Carles Escera Micó, PhD

During 2022, two major milestones were achieved by our research group. First, in collaboration with the Foetal Environment and Obstetric Complications research group, led by Dr Gómez-Roig, we were able to identify a neurophysiological consequence of being born small for gestational age. Specifically, we showed, in a sample of 53 newborns with fetal growth restriction (FGR) compared to 48 newborns with adequate weight for gestational age, an abnormal neural encoding of speech sounds at birth in the FGR group (Ribas-Prats et al., 2022). These results were obtained with a variant of the auditory evoked potential typically used in the universal neonatal hearing screening, termed frequency-following response (FFR). We published during this period a review of the recording procedures of the neonatal FFR, along with the results achieved so far by our group in this research approach (Gorina-Careta et al., 2022). The second milestone during this period was securing the funding from three competitive calls (LLAVOR-AGAUR; Prueba de Concepto – MICINN; and F2I-Fundació Bosch i Gimpera/Universitat de Barcelona) to start our innovation strategy aiming at developing a predictive tool of neurodevelopmental derail during early development (target age: 2 years), based on the neuronal

response to a speech sound at birth and its interpretation by an artificial intelligence algorithm. Our goal here is to establish a universal “cognitive” screening at birth based on neurophysiological responses.

### MEMBERS

#### STAFF RESEARCHER

Jordi Costa Faidella, Imma Clemente Lapena, María José Corral López, María Isabel Núñez Peña, Iria San Miguel Insúa, Hans Supèr, Marc Via Garcia

#### PHD STUDENTS

Sonia Arenillas Alcón, Marta Puertollano Rodríguez, Teresa Ribas Prats, Alejandro Lerer

### SELECTED PROJECTS

- Predicting neurodevelopment delay in newborns born low weight with artificial intelligence applied to speech sound-related EEG. PDC2022-133044-I00. Spanish Ministry of Science and Innovation. Proof of Concept call. PI: Carles Escera. 2022-2024.

### SELECTED PUBLICATIONS

- Bermudo A, Ariza M, Dacosta R, Agudelo D, Camins N, Boldó M et al. Effects and mechanisms of mindfulness training and physical exercise on cognition, emotional wellbeing, and brain outcomes in chronic stroke patients: Study protocol of the MindFit project randomized controlled trial. *Front Aging Neurosci.* 2022;14:936077-936077. IF:4,800. (Q2).
- Gorina N, Ribas T, Arenillas S, Puertollano M, Gómez MD, Escera C. Neonatal Frequency-Following Responses: A Methodological Framework for Clinical Applications. *Semin Hear.* 2022;43(3):162-176.
- Heurteloup C, Merchie A, Roux S, Bonnet F, Escera C, Gomot M. Neural repetition suppression to vocal and non-vocal sounds. *Cortex.* 2022;148:1-13. IF:3,600. (Q1).
- Ribas T, Arenillas S, Lip DL, Costa J, Mazarico E, Gómez MD et al. Deficient neural encoding of speech sounds in term neonates born after fetal growth restriction. *Dev Sci.* 2022;25(3)IF:3,700. (Q1).

## Attention, Perception and Language Acquisition (APAL)

- 0** COMMERCIAL CLINICAL TRIALS
- 0** ACADEMIC CLINICAL TRIALS
- s/d** PROJECTS
- 0** PUBLICATIONS
- 0** IMPACT FACTOR
- 0%** PUBLICATIONS IN 1RTS QUARTIL
- 0%** PUBLICATIONS IN 1RTS DECIL
- 0%** LEADERSHIP

### Laura Bosch Galceran, PhD

Research activity from our group has been developed in three directions. First, the study of language and cognitive outcomes in infants born deaf and explored two years post-implantation has been completed (communicative, lexical and morpho-syntactic development; fast mapping and word learning skills). An age-matched sample of hearing infants has also been gathered and tested on the same skills. Results revealed important levels of variability within the implanted population. Data analyses have been undertaken taking into account the role of biological pre-implantation markers, as well as socio-educational aspects and early communicative skills. This is a collaborative research involving the ORL unit (Dra. M.A. Claveria and Dr. O. Haag), Neuro-radiology unit (M. Rebollo) and Psychology unit (D. Polo). A second research line from our group (APAL Lab) has been focused on audiovisual speech processing in the first year of life, involving infants from monolingual and bilingual environments. The aim is to gain knowledge on the basic mechanisms in native language recognition and the development of infants' early capacity

to associate face and language from familiar and non-familiar speakers. Finally, a new collaborative research project has been started with the UnimTEA Unit (Multidisciplinary Unit of Autism Spectrum Disorder) with Dr. A. Aranbarri and Sra. Díez, in the program TEA CARE-Mas Casadevall. This research aims at analyzing possible change biomarkers related to the social motivation hypothesis, by using experimental measures with an eye-tracking methodology. This methodology has been validated in the context of the early detection of population at risk for TEA. A first pilot study has been developed with a sample of children with a TEA diagnosis.

### MEMBERS

#### STAFF RESEARCHER

Ferran Pons Gimeno, Marta Ramon Casas

#### POSTDOC ASSOCIATED RESEARCHERS

Joan Birulés Muntané, Clément François

#### ASSISTANT RESEARCHER

Jessica Sánchez-Galán Frauca

### SELECTED PROJECTS

- Indexical representation and lexical learning in audiovisual contexts: A challenge or an advantage for typically developing infants and clinical populations?. PID2021-128159NB-I00. Ministerio de Ciencia, Innovación y Universidades. Convocatòria d'ajuts del Programa Estatal de Foment de la Investigació Científica i Tècnica d'Excel·lència. Subprograma Estatal de generació del coneixement. Modalitat Projectes d'R+D de Generació del Coneixement. co-PI: Ferran Pons Gimeno i Laura Bosch Galceran. 2022-2024.
- Resonancia magnética funcional e imagen de tensor de difusión en la valoración prequirúrgica del paciente pediátrico con hipoacusia prelingual neurosensorial profunda bilateral de detección en el cribado neonatal como potenciales biomarcadores de desarrollo de lenguaje oral tras la colocación de un implante coclear. Sociedad Española de Neuroradiología. SERN 2017. PCP00215. PI: Mónica Rebollo, MD. 2017-2022.

### SELECTED PUBLICATIONS

- Birulés J, Martínez A, Lewkowicz DJ, de Diego R, Pons F. Violation of non-adjacent rule dependencies elicits greater attention to a talker's mouth in 15-month-old infants. *Infancy*. 2022;27(5):963-971. IF:2,600. (Q2).

## Research in child and adolescent mental health

- 2** COMMERCIAL CLINICAL TRIALS
- 1** ACADEMIC CLINICAL TRIALS
- 11** PROJECTS
- 19** PUBLICATIONS
- 100,48** IMPACT FACTOR
- 58%** PUBLICATIONS IN 1RTS QUARTIL
- 11%** PUBLICATIONS IN 1RTS DECIL
- 47%** LEADERSHIP

### Montserrat Dolz Abadia, MD, PhD

Our research group is aimed at the investigation of psychiatric disorders in pediatric populations, with special emphasis on neurodevelopment disorders and early-onset disorders. Consolidated lines of research are on childhood-onset psychosis (PI: Dr Dolz), attention deficit hyperactivity disorder (PI: Dr Alda), autism spectrum disorder (PI: Dr Mairena, Dr Aranbarri), child witnesses of domestic violence (PI: Dr Álvarez, Dr Lacasa), neurodevelopmental disorders prevalence and school failure (Dr Casas, Dra Bosch). Emerging lines of research are in eating disorders (clinical PI: Dr Serrano; neuroimaging in eating disorders, PI: Dra Vial), self-injurious behaviour (PI: Dr Sintes), Early detection and prevention in mental health problems (PI: Dra Camprodón) and parenting interventions (Dra Villalta).

In recent years, our group has been sustained by awarded finantiation by public and private institutions, both national and European. Part of our group is within the CIBERSAM G11, and we have stable collaborations with other CIBERSAM groups (G04 and G01). We are also within the recognised SGR research group (SGR 01380; PI Judith Usall).

Several projects have been awarded with competitive grants, with IRSJD researchers as main PIs ("Early Treatment for children with mental health problems and Genetic Abnormalities through aParenting intervention (The GAP): a pragmatic randomized controlled trial" (PI: Dra Villalta, Marató TV3), "Precursors del trastorno por atracón en adolescentes con obesidad: marcadores MRI temprano de disfunción de circuitos cerebrales de refuerzo e inhibición (Estudio PREC-BED)" (Dra Via, FIS, ISCI), "RESCCUE4COVID: REcover Spaces for traumatized Children in CommUnity sEttings post COVID-19" (Dra Álvarez, European Union: CERV-2021-DAPHNE), Determinants primerencs ambientals, psicosocials i genètiques dels problemes neuroconductuals i emocionals (PNCEs) en els primers 4 anys de vida: l'estudi ENGENE, Dra. Gascon, Co-. IP: Dra. Camprodón, Marató TV3. IRSJD researchers were included as collaborators in other competitive grants (Marató TV3 and Fundación Ramon Areces). Finally, Dr Vidal-Ribas has joined the research group with a Ramón y Cajal post-doctoral research scholarship (ISCI).

### MEMBERS

#### STAFF RESEARCHER

Alazne Gorrotxategi Aizpitarte, Aritz Aranbarri Paredes, Xavier Estrada Prat, Francisco Villar Cabeza, Laia Mollà Cusí, Esther Via Virgili, Neus Elias Carbonell, José Ángel Alda Díez, Mar Alvarez Segura, Ester Camprodón Rosanas, María Diez Juan, Fernando Lacasa Saludes, Ma Ángeles Mairena García de la Torre, Josep Lluís Matalí Costa, Daniel Muñoz Samons, Jordi Navarra Ordoño, Marta Pardo Gallego, Isabel Rueda Barcena, Bernardo Sánchez Fernández, Eduardo Serrano Troncoso, Anna Sintes Estevez

#### POSTDOC ASSOCIATED RESEARCHERS

Pablo Vidal-Ribas Belil, Inmaculada Insa Pineda, Anna Butjosa Molines

#### PHD STUDENTS

Laia Villalta Maciá, Ania Perez Racana, Anna Huguet Miguel, Claudia Caprile Elola - Olaso, Laia Portoles , Laura Puigcerver Navarro, Jordina Tor Fabra

#### ASSISTANT RESEARCHER

Carolina Morales Guerrero, Paula Resina Curado, Berta Sanfelix Salvà, Anna Martínez García, Sara Parrilla Carrasco, Ingrid Tortadès Arumí,

Marina Fabrega Ribera, Carlota Saumell Andreu, Andrea González Contijoch

#### COLLABORATOR RESEARCHER

Carme Tello

### SELECTED PROJECTS

- Ayudas Ramón y Cajal\_Beneficiari: Pablo Vidal-Ribas (RYC2021-033369-I). Ministerio De Ciencia E Innovación. 2022-2027.
- Estudio multicéntrico longitudinal sobre marcadores de predicción de la transición a psicosis en niños y adolescentes con riesgo de psicosis: el papel del estrés intrauterino. (PI21/00090). Instituto de Salud Carlos III (ISCI). 2022-2024. IP: Montserrat Dolz Abadia.
- Impacte de l'estressor ambiental COVID19 sobre les emocions i la conducta en població infantो-juvenil: Macro-Estudí longitudinal EmCoVID19 i EmCoVID19-unitats clíniques. (BML 2021\_HSJD). ZZZZ\_O.H.S.J.D.-Curia Provincial. 2021-2023. IP: Esther Via Virgili.
- Disrupció de los ritmos circadianos en población infantil con Trastornos del Neurodesarrollo (TDAH y TEA) y su impacto sobre la salud y calidad de vida. (PI19/00113). Instituto de Salud Carlos III (ISCI). 2020-2024. IP: José Ángel Alda Díez.
- First Steps: estudio piloto aleatorizado para la implementación del programa de parentalidad Incredible Years® en preescolares con autismo y retraso del lenguaje en España. Fundación Alicia Koplowitz. 2019-2022. IP: Laia Villalta Maciá.
- Afegir el de MIND Escolees (espera info de J de Dios)
- Programa MIMeS\_Detección, prevención y tratamiento precoz de Salud Mental en pacientes con "Malalties Minoritàries Cognitivo-Conductuals de base Genètica. (Programa MIMeS). OSSJD - Obra Social Sant Joan de Déu. 2018-2022. IP: Mercedes Serrano Gimaré, Laia Villalta Maciá.

### SELECTED CLINICAL TRIALS & OBSERVATIONAL STUDIES

- A Double-blind, Randomized, Psychoactive Placebo-controlled, Study to Evaluate the Efficacy and Safety of 3 Fixed Doses (28 mg, 56 mg and 84 mg) of Intranasal Esketamine in Addition to

Comprehensive Standard of Care for the Rapid Reduction of the Symptoms of Major Depressive Disorder, Including Suicidal Ideation, in Pediatric Subjects Assessed to be at Imminent Risk for Suicide; Janssen - Cilag, S.A. 2021. Local. IP: Montserrat Dolz Abadia. Sub-IPs: X Estrada, M Vila, M Andreu. Recruiters: A Torres, X Alvarez.

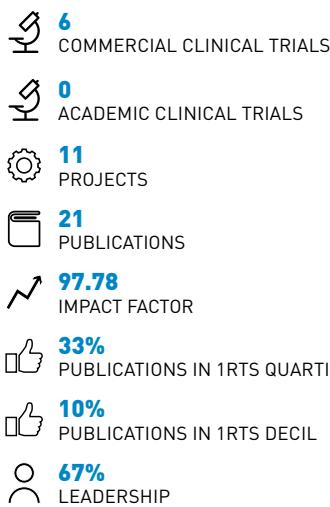
- Placebo-controlled trial in ultra-high risk for psychosis with omega-3 fatty acids in Europe (PURPOSE; EudraCT\_2015-003503-39). Stanley Medical Research Institute. University Medical Center Utrecht, 2016 - 2023. IP centro promotor: René Khan. IP local: Montserrat Dolz Abadia. Sub-IP Jordina Tor
- Assaig clinic RHC-PEP
- A Double-blind, Randomized, Psychoactive Placebo-controlled, Study to Evaluate the Efficacy and Safety of 3 Fixed Doses (28 mg, 56 mg and 84 mg) of Intranasal Esketamine in Addition to Comprehensive Standard of Care for the Rapid Reduction of the Symptoms of Major Depressive Disorder, Including Suicidal Ideation, in Pediatric Subjects Assessed to be at Imminent Risk for Suicide. Janssen - Cilag, S.A. 2018-2022. Local. IP: Montserrat Dolz Abadia. Su-IPs: M Vila, E Via.

#### SELECTED PUBLICATIONS

- Alvarez-Subiela X, Castellano-Tejedor C, Verge-Muñoz M, Esnaola-Letemendia K, Palao-Vidal D, Villar-Cabeza F. Predictors of Suicide Re-Attempt in a Spanish Adolescent Population after 12 Months' Follow-Up. *Int J Environ Res Public Health.* 2022 Jun;19(13):7566. IF:4,614(Q1)
- Bosch R, Payerols M, Rivas C, Sixto L, Bricollé L, Español-Martín G, et al. Neurodevelopmental disorders among Spanish school-age children: prevalence and sociodemographic correlates. *Psychol Med.* 2022 Oct;52(14):3062–72. IF:10,592(Q1)
- Rodríguez-Pascual M, Álvarez-Subiela X, Tor J, Pardo M, de la Serna E, Sugranyes G, et al. Major depressive disorder and attenuated negative symptoms in a child and adolescent sample with psychosis risk syndrome: the CAPRIS study. *Eur Child Adolesc Psychiatry.* 2022 Sep;31(9):1431–40. IF:5,349(Q1).



# Epilepsy and movement disorders in Pediatrics



Alexandros Arzimanoglou, MD, PhD

## MEMBERS

### STAFF RESEARCHER

Javier Aparicio, María del Carmen Fons Estupiña, Anna López Sala, Juan Darío Ortigoza Escobar

### ASSISTANT RESEARCHER

Itziar Alonso Colmenero, Santiago Antonio Candela Cantó, Verónica Delgadillo Chilavert, Andrea Palacio Navarro, Alia Ramírez Camacho

## SELECTED PROJECTS

- El análisis de la conectividad cerebral como herramienta para evaluar los trastornos neuroconductuales y de regulación emocional (EpileTRE.net). (PI21/01268). Instituto de Salud Carlos III (ISCIII). 2022-2024. IP: Alexandros Arzimanoglou.
- TREAT-AHC. Identification of compounds for the treatment of Alternating Hemiplegia of Childhood by drug repositioning. Molecular and pre-clinical evaluation and clinical outcome measure validation. (283/C/2020). Fundació La Marató de TV3. 2021-2024. IP: María del Carmen Fons Estupiña.
- Aplicación de una nueva técnica de neuroimagen en la evaluación pre-quirúrgica de la epilepsia: la

sustracción de la SPECT ictal con la 18F-FDG-PET (PISCOM). Sociedad Española de Epilepsia. 2021-2022. IP: Javier Aparicio.

Spike-and-Wave During Sleep. (NBI-827104-CSWS2010). ACCELSIORS; Neurocrine Biosciencies, inc. 2022. IP: Alia Ramírez Camacho.

## SELECTED CLINICAL TRIALS & OBSERVATIONAL STUDIES

- Estudio de extensión abierto a largo plazo para evaluar la seguridad y tolerabilidad de NBI-827104 en sujetos pediátricos con encefalopatía epiléptica con puntas-ondas continuas durante el sueño. Long-Term, Open-Label Extension Study to Evaluate the Safety and Tolerability of NBI-827104 in Pediatric Subjects with Epileptic Encephalopathy with Continuous Spike-and-Wave During Sleep. (NBI-827104-CSWS2025). Neurocrine Biosciencies, inc.; Vaquero. 2022. IP: Alia Ramírez Camacho.
- Estudio de prolongación sin enmascaramiento con XEN496 en niños que padecen encefalopatía epiléptica y del desarrollo por KCNQ2. An Open-Label Extension Study of XEN496 in Pediatric Subjects with KCNQ2 Developmental and Epileptic Encephalopathy. (XPF-009-302). Icon Clinical Research España, S.L.; Xenon Pharmaceuticals Inc. 2022. IP: María del Carmen Fons Estupiña.
- Estudio en fase III de XEN496 complementario en pacientes pediátricos con encefalopatía epiléptica y del desarrollo asociada al gen KCNQ2 A Phase 3 Study of Adjunctive XEN496 in Pediatric Subjects with KCNQ2 Developmental and Epileptic Encephalopathy. (XPF-009-301). Icon Clinical Research España, S.L.; Luque; Xenon Pharmaceuticals Inc. 2022. IP: María del Carmen Fons Estupiña.
- Estudio multicéntrico de fase 2, aleatorizado, doble ciego, controlado con placebo, de grupos paralelos para evaluar la eficacia, seguridad, tolerabilidad y farmacocinética de NBI-827104 en sujetos pediátricos con encefalopatía epiléptica con pico y onda continuos durante el sueño. Phase 2 Multicenter, Randomized, Double-Blind, Placebo-Controlled, Parallel-Group Study to Assess the Efficacy, Safety, Tolerability, and Pharmacokinetics of NBI-827104 in Pediatric Subjects with Epileptic Encephalopathy with Continuous

## SELECTED PUBLICATIONS

- Casas D, Oliva C, Vaz MA, Codina A, Agut T, García-Alix A et al. Cerebrospinal Fluid Ion Analysis in Neonatal Seizures. *Pediatr Neurol*. 2022;128:16-19. IF:4,210. (Q1).
- Julià NA, Molina C, Sigatullina M, Cortés E, Aparicio J, Cuadras D et al. Monoamine neurotransmitters in early epileptic encephalopathies: New insights into pathophysiology and therapy. *Dev Med Child Neurol*. 2022;64(7):915-923. IF:4,864. (Q1).
- Nevado J, García S, Palomares M, Vallespín E, Guillén E, Rosell J et al. Variability in Phelan-McDermid Syndrome in a Cohort of 210 Individuals. *Front Genet*. 2022;13:652454-652454. IF:4,772. (Q1).
- Ponce J, Benítez L, Baños N, Goncé A, Benassar M, Muñoz M et al. Latency to delivery in physical examination-indicated cerclage in twins is similar to that in singleton pregnancies. *Int J Gynaecol Obstet*. 2022;159(1):188-194. IF:4,447. (Q1).

# 7.1 MEDICINE AND PEDIATRIC SCIENCE AREA

## 7.1.2 **Adaptive response, metabolism and immunobiology**

Metabolic endocrinology

Metabolic diseases of paediatric origin

Molecular biology and gene regulation of adipose tissue and its pathologies

Neonatology research group

Pharmacological targets in inflammation and metabolic diseases

Study of diseases due to immune dysfunction in pediatrics (GEMDIP)

Infectious diseases and microbiome

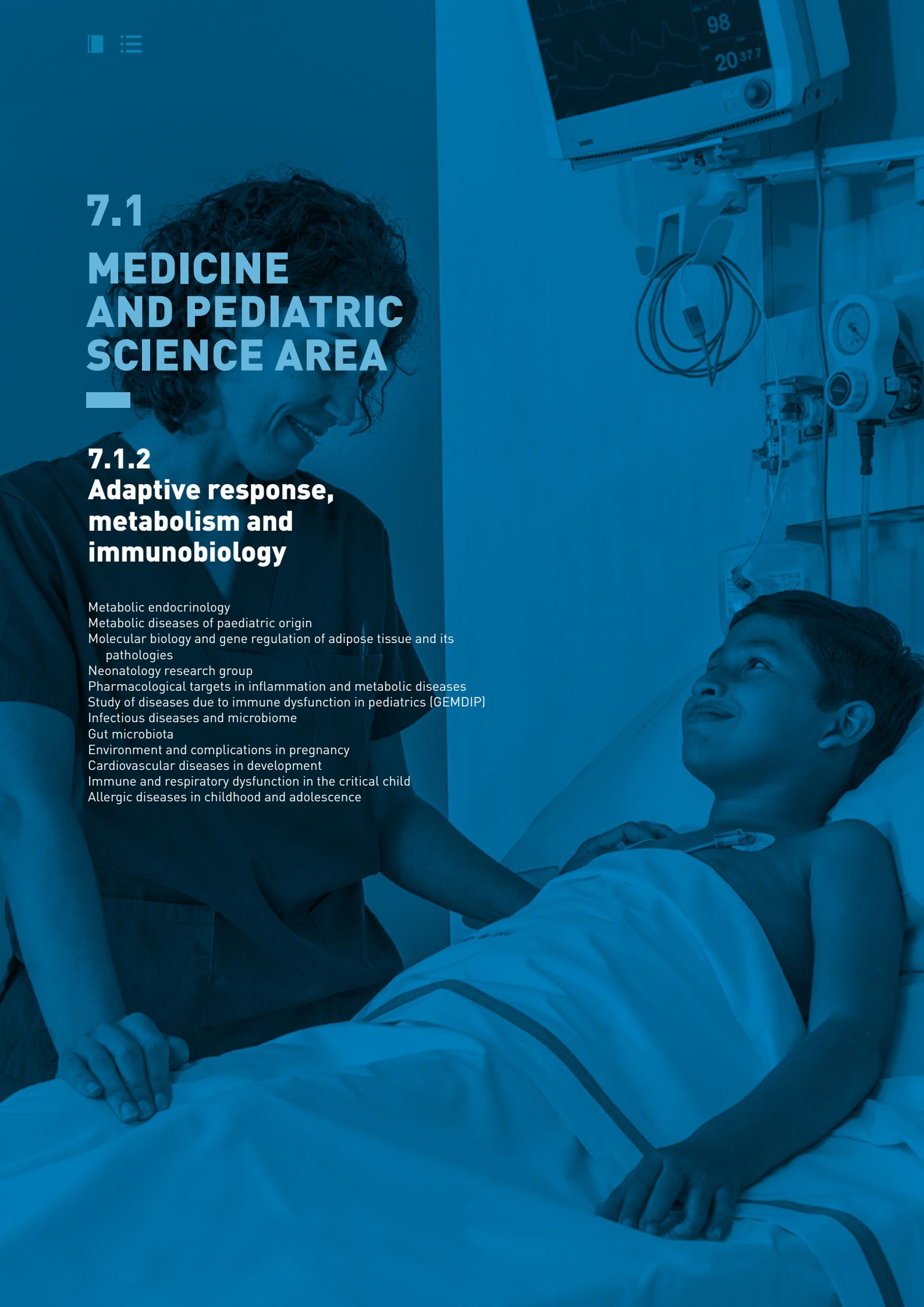
Gut microbiota

Environment and complications in pregnancy

Cardiovascular diseases in development

Immune and respiratory dysfunction in the critical child

Allergic diseases in childhood and adolescence



# Metabolic endocrinology

- 0** COMMERCIAL CLINICAL TRIALS
- 3** ACADEMIC CLINICAL TRIALS
- 5** PROJECTS
- 10** PUBLICATIONS
- 50.78** IMPACT FACTOR
- 70%** PUBLICATIONS IN 1RTS QUARTIL
- 0%** PUBLICATIONS IN 1RTS DECIL
- 80%** LEADERSHIP

**Lourdes Ibáñez Toda, MD, PhD**

## MEMBERS

### STAFF RESEARCHER

Silvia Marin Del Barrio, Silvia Xargay Torrent, Paula Casano Sancho, Marta Díaz Silva

### PHD STUDENTS

Cristina García Beltran

### COLLABORATOR RESEARCHER

Judith Bassols Casadevall, Abel López Bermejo

## SELECTED PROJECTS

- Efectos del tratamiento con spiomet a mitad de dosis en niñas con pubertad adelantada y aceleración de la maduración ósea: estudio multicéntrico, aleatorizado y controlado con placebo. (ICI21/00005). Institut d'Investigació Biomèdica de Girona Dr. Josep Trueta. 2022-2025. IP: Lourdes Ibáñez Toda.
- SPIOMET4HEALTH\_PCOS in adolescent girls and young women: Toward a treatment guided by pathophysiology. (899671). European Commission. 2021-2026. IP: Lourdes Ibáñez Toda.
- Subvencions per a la contractació de personal investigador en formació (PIF-Salut). Beneficiari: Cristina García Beltran. (SLT017/20/000228). Departament de Salut - Generalitat de Catalunya. 2021-2024. IP: Lourdes Ibáñez Toda.

## SELECTED CLINICAL TRIALS & OBSERVATIONAL STUDIES

- Efectos del tratamiento con spiomet a mitad de dosis en niñas con pubertad adelantada y aceleración de la maduración ósea: estudio multicéntrico, aleatorizado y controlado con placebo. (MINI-SPIOMET). Institut d'Investigació Biomèdica de Girona Dr. Josep Trueta; Serrate. 2022. IP: Lourdes Ibáñez Toda.
- Ensayo clínico de fase II, aleatorizado, multicéntrico y multinacional para evaluar la eficacia, la tolerabilidad y la seguridad de una combinación de dosis fijas de espironolactona, pioglitazona y metformina (SPIOMET) para adolescentes y mujeres adultas jóvenes (AYA) con síndrome de ovario poliquístico (SOP) A Phase II, randomised, multi-centric, multi-national clinical trial to evaluate the efficacy, tolerability, and safety of a fixed dose combination of Spironolactone, Pioglitazone & Metformin (SPIOMET) for adolescent girls and young adult women (AYAs) with polycystic ovary syndrome (PCOS) / Ensayo clínico de fase II, aleatorizado, multicéntrico y multinacional para evaluar la eficacia, la tolerabilidad y la seguridad de una combinación de dosis fijas de espironolactona, pioglitazona y metformina (SPIOMET) para adolescentes y mujeres adultas jóvenes con síndrome de ovario poliquístico (SOP). (SPIOMET4HEALTH).
- Carranza; Fundació Privada per a la Recerca i la Docència Sant Joan de Déu - FSJD; Garcia; Optimapharm Contract Research Organisation, S.L.U. 2022. IP: Lourdes Ibáñez Toda.

and CXCL14 Levels in the First Year of Life: Sex Differences and Association With Adiposity. *J Clin Endocrinol Metab.* 2022;107(3):1148-1158. IF:6,134. (Q1).

- Gonzalez C, Manuel B, Díaz M, García C, Lerin C, Barbas C et al. Birth Weight and Early Postnatal Outcomes: Association with the Cord Blood Lipidome. *Nutrients.* 2022;14(18)IF:6,706. (Q1).

## SELECTED PUBLICATIONS

- Casano P, Ana Carolina Izurieta P. Endocrine Late Effects in Childhood Cancer Survivors. *Cancers (Basel).* 2022;14(11): 2630. IF:6,575. (Q1).
- García C, Carreras G, Bassols J, Malpique R, Plou C, de Zegher F et al. microRNAs in newborns with low birth weight: relation to birth size and body composition. *Pediatr Res.* 2022;92(3):829-837. IF:3,953. (Q1).
- García C, Cereijo R, Plou C, Gavaldà A, Malpique R, Villarroyna J et al. Posterior Cervical Brown Fat

# Metabolic diseases of paediatric origin

	<b>5</b>	COMMERCIAL CLINICAL TRIALS
	<b>0</b>	ACADEMIC CLINICAL TRIALS
	<b>11</b>	PROJECTS
	<b>10</b>	PUBLICATIONS
	<b>70.84</b>	IMPACT FACTOR
	<b>80%</b>	PUBLICATIONS IN 1RTS QUARTIL
	<b>20%</b>	PUBLICATIONS IN 1RTS DECIL
	<b>50%</b>	LEADERSHIP

**Josep Jiménez Chillarón, PhD & Carles Lerin Martínez, PhD**

Our main scientific goal is to understand the molecular mechanisms by which nutrition during early neonatal/infant development determines the risk of obesity and obesity-associated co-morbidities during adulthood. For this purpose, we combine human clinical cohorts, a murine model of childhood obesity, and *in vitro* cell cultures. Secondly, grounded on the previous goal, we aim to design novel knowledge-based prevention/intervention strategies intended to tackle childhood obesity and its late onset co-morbidities.

## Aim 1. Defining molecular mechanisms of disease

Over the last year, we underscored that the circadian rhythm played a critical role in mediating the development of hepatic steatosis and insulin resistance in a mouse model of childhood obesity (MINECO, PID2021-126441NB-100). We reported that these effects may be transmitted to the following generation offspring, likely through epigenetic mechanisms (Templenton Foundation, ID-62167). Furthermore, in the context of the recent COVID-19 epidemic, we have performed a comprehensive characterization of the effects of maternal SARS-

CoV-2 infection and vaccination on breast milk composition and the potential consequences for the infants (MilkCorona Study, Fundació la Marató de TV3, 202106-32).

## Aim 2. Designing evidence-based novel prevention and intervention strategies

Interventions at early stages of development offer the possibility of exerting long-term health effects. In particular, we are conducting two approaches: In the first one, we are modifying infant diet exclusively during lactation with the aim of reducing the risk of childhood obesity (MINECO, PDC2021-121816-100). We are carrying out the study in human subjects at risk of childhood obesity. Secondly, we are developing two nutritional interventions in children and adolescents with obesity. Specifically, we are now implementing a Time-Restricted Feeding protocol onto children and adolescents with obesity to ameliorate metabolic disturbances through modulating, in part, the circadian rhythm [ClinicalTrials.gov, ID NCT05174871]. Furthermore, we are also treating children with Prader-Willi syndrome with probiotics (ISCIII, PI20/00301).

## MEMBERS

### STAFF RESEARCHER

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### POSTDOC ASSOCIATED RESEARCHERS

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### COLLABORATOR RESEARCHER

Ivonne Palacios Marin

### ASSISTANT RESEARCHER

Ana Lopez Bataller, Lorena González López

## SELECTED PROJECTS

- Impact of maternal SARS-CoV-2 infection and vaccination on breast milk antibodies, milk bioactive compounds and maternal-child health (MilkCORONA). (202106-32). Fundació La Marató de TV3. 2021-2023. IP: Carles Lerin Martinez.
- Non-genomic Inheritance of environmentally-induced traits:

Deciphering epigenetically-driven genetic variation in mammals. (ID 62167). John Templenton Foundation. 2021-2024. IP: José Carlos Jiménez Chillarón.

- El microbioma intestinal como diana terapéutica en el síndrome de Prader-Willi: efectos metabólicos y en el comportamiento de los pacientes. (PI20/00301). Instituto de Salud Carlos III (ISCIII). 2021-2023. IP: Marta Ramon Krauel.
- Cronobiología durante el desarrollo neonatal: Implicaciones sobre la obesidad infantil y enfermedades metabólicas asociadas. (PID2021-126441NB-100). Ministerio de Economía Y Competitividad (MINECO). 2022-2024. IP: José Carlos Jiménez Chillarón.

## SELECTED CLINICAL TRIALS & OBSERVATIONAL STUDIES

- A time-restricted feeding intervention in children and adolescents with obesity: The TRansForm study (ClinicalTrials.gov ID NCT05174871). Fundació Privada per a la Recerca i la Docència Sant Joan de Déu - FSJD. 2022. IP: Carles Lerin.
- El microbioma intestinal como diana terapéutica en el síndrome de Prader-Willi: efectos metabólicos y en el comportamiento de los pacientes. (PI20/00301). Instituto de Salud Carlos III (ISCIII). IP: Marta Ramon Krauel.
- Estudio en fase III, aleatorizado a doble ciego y controlado con placebo: 5 subestudios independientes de setmelanotide en pacientes con defectos en los genes POMC, PCSK1, LEPR, SRC1, SH2B1 o PCSK1 N221D en la vía del receptor 4 de melanocortina. A Phase 3, Randomized, Double-Blind, Placebo-Controlled, Four-Arm Parallel-Group Study of Setmelanotide in Patients with Specific Gene Defects in the Melanocortin-4 Receptor Pathway. (RM-493-035). Medina; RHYTHM Pharmaceuticals, Inc. 2022. IP: Marta Ramon Krauel.
- Estudio en fase II, doble ciego y controlado con placebo de setmelanotide en pacientes con defectos genéticos específicos en la vía del receptor de la melanocortina 4, en dos etapas (preinclusión abierta seguida de retirada aleatorizada). A 2-Stage

(Open-Label Run-in followed by Randomized Withdrawal), Double-Blind, Placebo-Controlled, Phase 2 Study of Setmelanotide in Patients with Specific Gene Defects in the Melanocortin-4 Receptor Pathway. (RM-493-034). IQVIA RDS Spain SL; Medina; RHYTHM Pharmaceuticals, Inc. 2022. IP: Marta Ramon Krauel.

#### SELECTED PUBLICATIONS

- Molina PX, Murillo S, Meis L, Sans O, Amat M, Llobet M et al. A time-restricted feeding intervention in children and adolescents with obesity: The TRansForm study protocol. *Front Nutr.* 2022;9:1026694-1026694. IF:6,590. (Q1).
- Selma M, Bäuerl C, Mena D, Aguilar L, Pérez FJ, Parra A et al. Anti-SARS-CoV-2 IgA and IgG in human milk after vaccination is dependent on vaccine type and previous SARS-CoV-2 exposure: a longitudinal study. *Genome Med.* 2022;14(1):42-42. IF:15,266. (Q1).
- Bäuerl C, Randazzo W, Sánchez G, Selma M, García E, Martínez L et al. SARS-CoV-2 RNA and antibody detection in breast milk from a prospective multicentre study in Spain. *Arch Dis Child Fetal Neonatal Ed.* 2022;107(2):216-221. IF:6,643. (Q1).



# Molecular biology and gene regulation of adipose tissue and its pathologies

- 0** COMMERCIAL CLINICAL TRIALS
- 0** ACADEMIC CLINICAL TRIALS
- s/d** PROJECTS
- 14** PUBLICATIONS
- 118.73** IMPACT FACTOR
- 71%** PUBLICATIONS IN 1RTS QUARTIL
- 21%** PUBLICATIONS IN 1RTS DECIL
- 57%** LEADERSHIP

**Francesc Villarroya Gombau, PhD**

## MEMBERS

### STAFF RESEARCHER

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### TECHNICIAN

Mercedes Morales Rueda, Gemma Moreno Besora, Albert Però Garcia

## SELECTED PUBLICATIONS

- Domingo P, Quesada T, Villarroya J, Cairo M, Gutierrez MDM, Mateo MG et al. Differential effects of dolutegravir, bictegravir and raltegravir in adipokines and inflammation markers on human adipocytes. *Life Sci.* 2022;308:120948-120948. IF:6,780. (Q1).
- García C, Cereijo R, Plou C, Gavaldà A, Malpique R, Villarroya J et al.

Posterior Cervical Brown Fat and CXCL14 Levels in the First Year of Life: Sex Differences and Association With Adiposity. *J Clin Endocrinol Metab.* 2022;107(3):1148-1158. IF:6,134. (Q1).

- Gavaldà A, Villarroya J, Cereijo R, Giralt M, Villarroya F. The endocrine role of brown adipose tissue: An update on actors and actions. *Rev Endocr Metab Disord.* 2022;23(1):31-41. IF:9,306. (Q1).
- Ruperez C, Blasco A, Kular D, Cairo M, Ferrer G, Villarroya J et al. Autophagy is Involved in Cardiac Remodeling in Response to Environmental Temperature Change. *Front Physiol.* 2022;13:864427-864427. IF:4,755. (Q1).

## Neonatology research group

	<b>0</b>	COMMERCIAL CLINICAL TRIALS
	<b>1</b>	ACADEMIC CLINICAL TRIALS
	<b>9</b>	PROJECTS
	<b>17</b>	PUBLICATIONS
	<b>77.153</b>	IMPACT FACTOR
	<b>53%</b>	PUBLICATIONS IN 1RTS QUARTIL
	<b>24%</b>	PUBLICATIONS IN 1RTS DECIL
	<b>53%</b>	LEADERSHIP

### Martin Iriondo Sanz, MD, PhD

Our research group studies how neonatal disease and early exposures could determine immediate later effects that will manifest in childhood or adulthood, both from a metabolic and neurodevelopmental point of view.

Since 2021, a competitive project is being developed that includes a prospective multicenter study of validation of predictive models of mortality in premature infants (mortality calculator in premature infants - [https://mbosio85.github.io/SEN\\_calc/](https://mbosio85.github.io/SEN_calc/)).

During 2022 we have been following up a cohort of very preterm patients at between 7-9 years of life thanks to a competitive grant obtained in 2020: "Consequences in childhood of perinatal growth failure in very preterm newborns: clinical and metabolomic study [FIS PI20/00294]". This project has been the basis to apply for a new grant to analyze the impact of the introduction of probiotics in our Neonatal unit in the care of very preterm newborns.

During the year 2022, the first translational studies have been initiated to evaluate the new continuous neuromonitoring device (jointlab-sjd. icfo.eu), with the aim of determining the origin of brain lesions in critically ill neonatal patients. This work has been carried out together with the cardiology department and the Institute of Photonic Sciences (ICFO) within the framework of a European Horizon 2020 grant (<https://tinybrains.eu/>). Also in this line, many projects studying the

origin of the cerebral alterations in patients with congenital heart disease have been published, some of them analyzing some aspects of the fetal life and others including the study of the perioperative period and follow up.

The neonatology nursing team has also collaborated in many projects: "*Mindfulness to improve stress in mothers and fathers during kangaroo care in a neonatal intensive care unit*", "*implementation of an intervention to increase milk production in mothers of premature newborns admitted to a NICU*" and "*Emotional impact of parents on the admission of newborns to the Neonatal Intensive Care Unit*", in collaboration with the University of Barcelona.

### MEMBERS

#### STAFF RESEARCHER

Marta del Olmo Fernández, Cristina Carrasco Carrasco, Isabel Iglesias Platas, Ana Morillo Palomo

#### POSTDOC ASSOCIATED RESEARCHERS

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Carla Balcells Espinera, Jordi Clotet Caba, Ruth Del Rio Florentino, Beatriz Del Rey Hurtado De Mendoza, Montserrat Izquierdo Renau, Alba Rivas Piorno

#### ASSISTANT RESEARCHER

Ines Fernanda Medina Rivera

#### COLLABORATOR RESEARCHER

Maria Dolores Salvia Roiges, Mattia Bosio, Marta Thió Lluch, Cristina Borràs Novell, Ana Herranz Barbero

### SELECTED PROJECTS

- Validación prospectiva mediante un estudio multicéntrico estatal (REDSAMID) de modelos predictivos de mortalidad neonatal en prematuros. [PI20/00295]. Instituto de Salud Carlos III (ISCIII). 2021-2023. IP: Martin Iriondo Sanz.
- Consecuencias en la infancia del fallo perinatal de crecimiento en Recién Nacidos Muy Prematuros: estudio clínico y metabolómico. [PI20/00294]. Instituto de Salud Carlos III (ISCIII). 2021-2023. IP: Isabel Iglesias Platas.
- TinyBrains\_Bio-photonic imaging of the infant brain, the missing link between the cellular brain damage and the neurovascular unit during acute illness. [101017113]. European Commission; Fundació Privada per a la Recerca i la Docència Sant Joan de Déu - FSJD. 2021-2024. IP: Marta Camprubi Camprubi, Joan Sánchez de Toledo.

de Toledo.

- Estrés oxidativo y mielinización en pacientes con cardiopatía congénita: nuevos horizontes terapéuticos. [PI19/00118]. Instituto de Salud Carlos III (ISCIII). 2020-2023. IP: Marta Camprubi Camprubi.
- Electricity and light; together as a novel tool for understanding the neonatal brain metabolism during acute illness. Torrons Vicens SL. 2020-2023. IP: Marta Camprubi Camprubi, Joan Sánchez de Toledo.

### SELECTED CLINICAL TRIALS & OBSERVATIONAL STUDIES

- Efecto del Alopurinol en pacientes con encefalopatía hipóxico-isquémica tratados con hipotermia. ALBINO ensayo clínico multicéntrico, aleatorizado, ciego, controlado con placebo. [H2020-PHC-18-2015-667224]. Instituto de Investigación Sanitaria Hospital La Fe. 2017. IP: Marta Camprubi Camprubi.
- Evaluation of Real Time Continuous Glucose Monitoring in the very preterm infant. (REACT RCT). University of Cambridge. 2017. IP: Isabel Iglesias Platas.
- Recién nacidos con edad gestacional extremadamente baja asignados al azar, a ciegas, para ser resucitados resuscitado con un 21% frente al 60% de oxígeno: influencia sobre la mortalidad y las enfermedades crónicas en el período neonatal. [1645-CI-057]. Instituto de Investigación Sanitaria Hospital La Fe. 2015. IP: Martin Iriondo Sanz.

### SELECTED PUBLICATIONS

- Cañizo D, Hadley SM, Marta O, López M, Valls A, Viñals ML et al. Oxidative Stress and Indicators of Brain Damage Following Pediatric Heart Surgery. *Antioxidants* (Basel). 2022;11(3). IF:7,675. (Q1).
- Escobar MC, Pérez M, Arráez M, Cascant MM, Albiach A, Kuligowski J et al. Brain Oxygen Perfusion and Oxidative Stress Biomarkers in Fetuses with Congenital Heart Disease-A Retrospective, Case-Control Pilot Study. *Antioxidants* (Basel). 2022;11(2): 229. IF:7,675. (Q1).
- Avila A, Zozaya C, Pértiga S, Sanchez M, Iriondo M, Elorza MD et al. Temporal trends in respiratory care and bronchopulmonary dysplasia in very preterm infants over a 10-year period in Spain. *Arch Dis Child Fetal Neonatal Ed.* 2022;107(2):143-149. IF:6,643. (Q1).

# Pharmacological targets in inflammation and metabolic diseases



## Manuel Vázquez Carrera, PhD

Our group has reported that activating transcription factor 4 (ATF4) is a new factor involved in the downregulation of the peroxisome proliferator-activated receptor  $\gamma$  (PPAR $\gamma$ ) coactivator 1 $\alpha$  (PGC-1 $\alpha$ ) under endoplasmic reticulum stress conditions in skeletal muscle, suggesting that its inhibition might be a therapeutic target for insulin resistant states. Likewise, we have described that loss of the nuclear receptor PPAR $\beta/\delta$ -/- affects neuronal and synaptic structure, contributing to memory dysfunction, suggesting that this receptor might be a possible new target for the treatment of memory impairment. Finally, we have delineated mechanistic insights into the beneficial effects of recently explored therapeutic approaches that target growth differentiation factor 15 (GDF15) for the treatment of type 2 diabetes mellitus.

## MEMBERS

### STAFF RESEARCHER

F. Xavier Palomer Tarridas

### POSTDOC ASSOCIATED RESEARCHERS

Emma Barroso Fernández, Marta Montori Grau

## SELECTED PROJECTS

- Targeting the PPAR $\beta/\delta$ -GDF15 pathway for the treatment of insulin resistance and type 2 diabetes mellitus (PPAR $\beta/\delta$ -GDF15). PID2021-122116OB-I00. Ministerio de Ciencia e Innovación (MICINN). Generación del Conocimiento 2021. PI: Manuel Vázquez Carrera and Xavier Palomer. 2022-2025
- Unveiling the role of the growth differentiation factor 15 (GDF15) in non-alcoholic fatty liver disease (NAFLD). CIBERDEM 2021/PIM04. CIBERDEM. CIBERDEM Intramurales. PI: Emma Barroso/Ricardo Rodríguez/María García Altares. 2021-2022.

## SELECTED PUBLICATIONS

- Aguilar D, Barroso E, Palomer FX, Wahli W, Vazquez M. Knocking on GDF15's door for the treatment of type 2 diabetes mellitus. Trends Endocrinol Metab. 2022;33(11):741-754. IF:10,586. (Q1).
- Espinosa T, Busquets O, Cano A, Sánchez E, Verdaguér E, Parcerisas A et al. Peroxisomal Proliferator-Activated Receptor  $\beta/\delta$  Deficiency Induces Cognitive Alterations. Front Pharmacol. 2022;13:902047-902047. IF:5,988. (Q1).
- Montori M, Aguilar D, Zarei M, Pizarro J, Palomer FX, Vazquez M. Endoplasmic reticulum stress downregulates PGC-1 alpha in skeletal muscle through ATF4 and an mTOR-mediated reduction of CRTC2. Cell Commun Signal. 2022;20(1):53-53. IF:7,525. (Q2).

# Study of diseases due to immune dysfunction in pediatrics (GEMDIP)

- 19** COMMERCIAL CLINICAL TRIALS
- 2** ACADEMIC CLINICAL TRIALS
- 19** PROJECTS
- 32** PUBLICATIONS
- 265.126** IMPACT FACTOR
- 66%** PUBLICATIONS IN 1RTS QUARTIL
- 22%** PUBLICATIONS IN 1RTS DECIL
- 34%** LEADERSHIP

**Laia Alsina Manrique de Lara, MD, PhD & Jordi Antón López, MD, PhD**

Highlighting the year 2022, the official recognition as a research group consolidated by the AGAUR, and the progressive alignment of the group's research towards diseases due to immune dysregulation. This has successfully translated into top notch publications by our own group and collaborations with other leading national and international teams. Also of note is obtaining competitive grants to develop basic and clinical translational research to understand the pathophysiology and identify new therapeutic targets for these disorders. All this aligned with our prolific participation in clinical trials that will provide new treatments for patients with rare immunological diseases.

## MEMBERS

### STAFF RESEARCHER

Alessandra Magnani , Violeta Bittermann , Juan Manuel Mosquera Angarita, Ana M<sup>a</sup> Plaza Martín, Andrea Zarcarias Crovato, Rosa Bou i Torrent, Àngela Deyà Martínez, Estibaliz Iglesias Jimenez, Judith Sánchez Manubens

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### COLLABORATOR RESEARCHER

Jordi Yagüe Ribes, Manel Juan Otero, Juan Ignacio Arostegui Gorospe

## SELECTED PROJECTS

- Caracterización de las alteraciones inmunológicas, específicamente en células Treg, implicadas en la desregulación inmune en pacientes pediátricos con errores innatos de inmunidad, hacia terapias dirigidas. (PI21/00211). Instituto de Salud Carlos III (ISCIII). 2022-2024. IP: Laia Alsina Manrique de Lara.
- Characterization of the nasopharyngeal microbiota in Kawasaki disease. (PI20/00517). Instituto de Salud Carlos III (ISCIII). 2021-2023. IP: Jordi Antón López.
- PIVIT\_Construcció d'una sala blanca per al seu ús en teràpies avançades en pacients pediàtrics, dirigit principalment a la producció de vectors virals per a teràpia gènica. (IU16-016824). Agaur - Agència de Gestió d'Ajuts Universitaris i de Recerca; Departament de Salut - Generalitat de Catalunya. 2020-2023. IP: Laia Alsina Manrique de Lara, Manel Juan Otero.
- ImmunAID\_Immunome project consortium for AutoInflammatory Disorders. (779295). European Commission. 2018-2023. IP: Jordi Antón López.
- Los nuevos criterios de PRINTO para la clasificación de la artritis idiopática juvenil. Un proyecto basado en la evidencia científica para la revisión de los criterios de la Liga Internacional contra el Reumatismo (ILAR). (ILAR). Accountancy Departament Instituto Giannina Gaslini. 2019. IP: Jordi Antón López.

## SELECTED CLINICAL TRIALS & OBSERVATIONAL STUDIES

- Multi-centre, randomised, open-label, blinded endpoint assessed, trial of corticosteroids plus intravenous immunoglobulin (IVIG) and aspirin, versus IVIG and aspirin for prevention of coronary artery aneurysms in Kawasaki disease. (KD-CAAP [C4C]). University College London. 2022. IP: Jordi Antón López.
- Estudio abierto, multicéntrico, de dos cohortes y un solo grupo para evaluar la eficacia, la seguridad y la tolerabilidad, la farmacocinética y la farmacodinámica de emapalumab en niños y adultos con síndrome de activación de macrófagos (SAM) en la enfermedad de Still (incluida la artritis idiopática juvenil sistémica y la enfermedad de Still del adulto) o con SAM en el lupus eritematoso sistémico. A two-cohort, open-label, single arm, multicenter study to evaluate efficacy, safety and tolerability, pharmacokinetics (PK) and pharmacodynamics (PD), of emapalumab in children and adults with macrophage activation syndrome (MAS) in Still's disease (including systemic juvenile idiopathic arthritis and adult onset Still's disease) or with MAS in systemic lupus erythematosus. (NI-0501-14). Pharmaceutical Research Associates España SAU; Swedish Orphan Biovitrum AG. 2021. IP: Jordi Antón López.
- A Phase 3, Randomized, Double-Blind, Placebo-Controlled, Multicenter Study of Mavorixafor in Patients with WHIM Syndrome with Open-Label Extension. (X4P-001-103). González; Syneos Health UK Limited; X4 Pharmaceuticals Inc. 2020. IP: Laia Alsina Manrique de Lara.

farmacodinámica de emapalumab en niños y adultos con síndrome de activación de macrófagos (SAM) en la enfermedad de Still (incluida la artritis idiopática juvenil sistémica y la enfermedad de Still del adulto) o con SAM en el lupus eritematoso sistémico. A two-cohort, open-label, single arm, multicenter study to evaluate efficacy, safety and tolerability, pharmacokinetics (PK) and pharmacodynamics (PD), of emapalumab in children and adults with macrophage activation syndrome (MAS) in Still's disease (including systemic juvenile idiopathic arthritis and adult onset Still's disease) or with MAS in systemic lupus erythematosus. (NI-0501-14). Pharmaceutical Research Associates España SAU; Swedish Orphan Biovitrum AG. 2021. IP: Jordi Antón López.

• A Phase 3, Randomized, Double-Blind, Placebo-Controlled, Multicenter Study of Mavorixafor in Patients with WHIM Syndrome with Open-Label Extension. (X4P-001-103). González; Syneos Health UK Limited; X4 Pharmaceuticals Inc. 2020. IP: Laia Alsina Manrique de Lara.

## SELECTED PUBLICATIONS

- Deyà A, Rivière JG, Roxo P, Ramakers J, Bloomfield M, Guisado P et al. Impact of JAK Inhibitors in Pediatric Patients with STAT1 Gain of Function (GOF) Mutations-10 Children and Review of the Literature. *J Clin Immunol*. 2022;42(5):1071-1082. IF:8,542. (Q1).
- Grasa CD, Fernández E, Sánchez J, Carazo B, Aracil J, Anton J et al. Kawasaki disease in children younger than 6 months of age: characteristics of a Spanish cohort. *Eur J Pediatr*. 2022;181(2):589-598. IF:3,860. (Q1).
- Margarit A, Deyà A, Canizales JT, Vlagea A, Ana G, Marsal J et al. Case report: Challenges in immune reconstitution following hematopoietic stem cell transplantation for CTLA-4 insufficiency-like primary immune regulatory disorders. *Front. Immunol*. 2022;13:1070068-1070068. IF:8,786. (Q1).
- Pfeifer J, Thurner B, Kessel C, Fadle N, Kheiruddin P, Regitz E et al. Autoantibodies against interleukin-1 receptor antagonist in multisystem inflammatory syndrome in children: a multicentre, retrospective, cohort study. *Lancet Rheumatol*. 2022;4(5):329-337. IF:35,482. (Q1).

## Infectious diseases and microbiome

- 14** COMMERCIAL CLINICAL TRIALS
- 4** ACADEMIC CLINICAL TRIALS
- 19** PROJECTS
- 44** PUBLICATIONS
- 432.788** IMPACT FACTOR
- 52%** PUBLICATIONS IN 1RTS QUARTIL
- 23%** PUBLICATIONS IN 1RTS DECIL
- 45%** LEADERSHIP

**Carmen Muñoz Almagro, MD, PhD**

### MEMBERS

#### STAFF RESEARCHER

Clàudia Fortuny Guasch, Maria Goretti López Ramos, Antoni Noguera Julián, Pedro Brotons de los Reyes, Victoria Fumadó, Juan José García García, Cristian Launes Montaña, Silvia Simó Nebot

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#### TECHNICIAN

Carles Cisneros Camps, Amareesh Pérez Argüello, Jesica Saucedo Verdes

#### COLLABORATOR RESEARCHER

Emilia Sánchez Ruiz

### SELECTED PROJECTS

- Tecnologías NGS para la Vigilancia Epidemiológica Molecular de *S. pneumoniae* y para determinar su invasividad. Quo vadis y Quo invades

pneumococcus?. (PI19/00104). Instituto de Salud Carlos III (ISCI). 2020-2023. IP: Carmen Muñoz Almagro.

- Estudio CRIB [Cribado de citomegalovirus congénito en recién nacidos sanos e ingresados - Barcelona]. (PI19/00095). Instituto de Salud Carlos III (ISCI). 2020-2023. IP: Antoni Noguera Julián, Ana Alarcon Allen.
- Vaccine Effectiveness, Burden and Impact Studies (VEBIS) of COVID-19 and Influenza. European Centre for Disease Prevention and Control (ECDC). 2021-2023. IP: Carmen Muñoz Almagro.
- Contract ECD.11486- "Developing an infrastructure and performing vaccine effectiveness studies for COVID-19 vaccine in the EU/EEA". LOT3 (HCW). European Centre for Disease Prevention and Control (ECDC). 2021-2022. IP: Carmen Muñoz Almagro.

### SELECTED CLINICAL TRIALS & OBSERVATIONAL STUDIES

- Estudio de fase III, abierto, de un solo grupo y con dosis repetidas, para evaluar la seguridad y tolerabilidad, la farmacocinética y la actividad antiviral de maribavir para el tratamiento de la infección por el citomegalovirus (CMV) en niños y adolescentes que han recibido un trasplante de células madre hematopoyéticas (HSCT) o un trasplante de órgano sólido (TOS). A Phase 3, open-label, single-arm, repeated-dose study to evaluate the safety and tolerability, pharmacokinetics, and antiviral activity of maribavir for the treatment of cytomegalovirus (CMV) infection in children and adolescents who have received a hematopoietic stem cell transplant (HSCT) or a solid organ transplant (SOT). (TAK-620-2004). Takeda Farmacéutica España S.A. 2022. IP: Clàudia Fortuny Guasch.
- Ensayo en fase III, aleatorizado, doble ciego para evaluar la seguridad de una vacuna conjugada antineumococica 20-valente en lactantes sanos a phase 3, randomized, partial double-blind trial to evaluate the safety and immunogenicity of 20-valent pneumococcal conjugate vaccine in healthy children  $\geq 12$  to  $< 24$  months of age with two prior infant doses of preventar 13. (B7471027). Gonzalez; ICON Clinical Research, Ltd; Koncalova; PFIZER INC. 2022. IP: Antoni Noguera Julián.
- Estudio de fase 3, multicéntrico, alatorizado, con enmascaramiento parcial y controlado con palivizumab para evaluar la seguridad, la eficacia y la farmacocinética de MK-1654 en lactantes y niños con riesgo elevado de enfermedad grave por el VRS. Phase 3, Multicenter, Randomized, Partially Blinded, Palivizumab-Controlled Study to Evaluate the Safety, Efficacy, and Pharmacokinetics of MK-1654 in Infants and Children at Increased Risk for Severe RSV Disease. (MK-1654-007). Jiménez; Merck, Sharp & Dohme de España. S.A; Ventosa. 2021. IP: Clàudia Fortuny Guasch.
- Estudio fase III, aleatorizado, doble ciego, controlado con placebo y multicéntrico para demostrar la eficacia de una dosis única de la vacuna materna no adyuvada frente al Virus Respiratorio Sincitrial (VRS), administrada por vía IM a gestantes de 18 a 49 años de edad, para la prevención de la Enfermedad vías respiratorias Bajas (EVRB) asociada al VRS en sus hijos hasta los 6 meses de edad. (212171 (RSV MAT-009)). Glaxosmithkline, S.A. 2021. IP: Victoria Fumadó.
- Estudio de la infección por virus influenza en niños hospitalizados en España en dos estaciones gripe consecutivas. Análisis de costos médicos directos. (HOSPIGRIP-2016). Asociación Instituto Biodonostia. 2017. IP: Juan José García García

### SELECTED PUBLICATIONS

- Armero G, Penela D, Belmonte J, Gómez D, Larrauri A, Henares D et al. Concentrations of nitrogen compounds are related to severe rhinovirus infection in infants. A time-series analysis from the reference area of a pediatric university hospital in Barcelona. Pediatr Pulmonol. 2022;57(9):2180-2188. IF:4,090. (Q1).
- Jordán I, Fernández de Sevilla M, Fumadó V, Bassat Q, Bonet E, Fortuny C et al. Transmission of Severe Acute Respiratory Syndrome Coronavirus 2 Infection Among Children in Summer Schools Applying Stringent Control

Measures in Barcelona, Spain.  
Clin Infect Dis. 2022;74(1):66-73.  
IF:20,999. (Q1).

- Soler A, Gamell A, Pérez T, Soriano A, Begoña G, Tórtola T et al. Performance of QuantiFERON-TB Gold Plus assays in children and adolescents at risk of tuberculosis: a cross-sectional multicentre study. Thorax. 2022;77(12):1193-1201. IF:9,102. (Q1).
- Lo SW, Mellor K, Cohen R, Redin A, Belman S, Kumar N et al. Emergence of a multidrug-resistant and virulent *Streptococcus pneumoniae* lineage mediates serotype replacement after PCV13: an international whole-genome sequencing study. Lancet Microbe. 2022;3(10):735-743. IF:86,208. (Q1).



## Gut microbiota

- 0** COMMERCIAL CLINICAL TRIALS
- 0** ACADEMIC CLINICAL TRIALS
- 1** PROJECTS
- 4** PUBLICATIONS
- 20.722** IMPACT FACTOR
- 75%** PUBLICATIONS IN 1RTS QUARTIL
- 0%** PUBLICATIONS IN 1RTS DECIL
- 25%** LEADERSHIP

### Laura Baldomà Llavinés, PhD

There is growing evidence that gut microbiota derived extracellular vesicles (EVs) are key players in interkingdom communication as mediators of the effects of gut resident bacteria on host immune and defense responses. Our studies on *E. coli* EVs have broadened the current understanding of how probiotic and beneficial gut microbes positively affect the intestinal epithelial barrier and immune modulation. In 2022, the main contribution to this topic was centered on the immunomodulatory properties of microbiota EVs. To deepen into the mechanisms underlying the ability of microbiota EVs to activate dendritic cells (DCs) and subsequent T cell responses in a strain-specific manner, we analyzed the influence of EVs on DC-secreted cytokines and exosomes. In the absence of direct cellular contacts, DC-secreted factors triggered secretion of effector cytokines by T cells. The main differences between strains influenced Th1 and Treg cytokines. Moreover, exosomes released by EV-activated DCs were enriched in surface proteins involved in antigen presentation and T cell activation but differ in the content of immune-related miRNA depending on the origin of EVs. Importantly, differences in the miRNA profile were consistent with the induced immune responses. Currently, therapies targeted at modulating DC function

are receiving great interest for certain diseases, such IBD or cancer. Our results suggest the potential of microbiota EVs as a novel strategy to treat these disorders. We are currently exploring the modulation by microbiota EVs of genes and miRNAs dysregulated in IBD such as miR-7-5p. This miRNA downregulates TFF3, a peptide with a crucial role in maintaining and repairing the intestinal mucosa. In goblet cells, EVs from *E. coli* strains differentially regulate TFF3 and miR-7-5p. Preliminary results show that EVs from the probiotic EcN increase TFF3 expression through TLR2 activation and miR-7-5p down-regulation.

### MEMBERS

#### STAFF RESEARCHER

Josefa Badia Palacin

### SELECTED PROJECTS

- Evaluación de nuevas actividades de vesículas secretadas por probióticos y microbiota intestinal y su potencial aplicación como postbióticos en salud humana. PID2019-107327RB-I00. Ministerio de Ciencia, Innovación y Universidades. Retos 2020. PI: Laura Baldomà, co-IP: Josefa Badia. 2020-2023.

### SELECTED PUBLICATIONS

- Diaz N, Badia J, Baldoma L. Modulation of Dendritic Cells by Microbiota Extracellular Vesicles Influences the Cytokine Profile and Exosome Cargo. *Nutrients*. 2022;14(2):344-344. IF:5,900. (Q1).
- Limon D, Gil P, Rodriguez L, Alvarado HL, Diaz N, Mallardrich M et al. Supramolecular Hydrogels Consisting of Nanofibers Increase the Bioavailability of Curcuminoids in Inflammatory Skin Diseases. *ACS Appl. Nano Mater.* 2022;5(10):13829-13839. IF:5,900. (Q2).
- Sarango P, Espinoza LC, Jorge G, Alvarado H, Rodríguez MJ, Baldoma L et al. Effect of Penetration Enhancers and Safety on the Transdermal Delivery of Apremilast in Skin. *Pharmaceutics*. 2022;14(5):1011. IF:5,400. (Q1).

## Environment and complications in pregnancy

- 0** COMMERCIAL CLINICAL TRIALS
- 4** ACADEMIC CLINICAL TRIALS
- 8** PROJECTS
- 23** PUBLICATIONS
- 160.875** IMPACT FACTOR
- 78%** PUBLICATIONS IN 1RTS QUARTIL
- 48%** PUBLICATIONS IN 1RTS DECIL
- 65%** LEADERSHIP

**Maria Dolores Gómez Roig, MD, PhD**

### MEMBERS

#### STAFF RESEARCHER

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### SELECTED PROJECTS

- Primary care interventions to prevent maternal and child chronic diseases of perinatal and developmental origin. (RD21/0012/0003). Instituto de Salud Carlos III (ISCIII). 2022-2024. IP: Maria Dolores Gómez Roig.
- Impacto de un programa de intervención prenatal estructurado, de disminución de consumo de alcohol, en los resultados perinatales y de neurodesarrollo. (PI20/00490). Instituto de Salud Carlos III (ISCIII). 2021-2023. IP:

- Maria Dolores Gómez Roig.
- Respuesta de seguimiento de frecuencia (RSF) como biomarcador de desarrollo neurocognitivo en recién nacidos con exposición prenatal al alcohol. Hospital Sant Joan de Déu - Esplugues HSJD. 2020-2023. IP: Maria Dolores Gómez Roig.
- ATHLETE\_Advancing Tools for Human Early Lifecourse Exposome research and Translation. (874583). European Commission. 2020-2025. IP: Maria Dolores Gómez Roig.

### SELECTED CLINICAL TRIALS & OBSERVATIONAL STUDIES

- Efficacy of low dose acetylsalicylic acid in preventing adverse maternal and perinatal outcomes in SARS-CoV-2 infected pregnant women. [SARA]. Fundació Privada ISGLOBAL; Serrano; Stöger. 2022. IP: Edurne Mazarico Gallego.
- Optimización del manejo antenatal de mujeres con amenaza de parto pretérmino mediante modelos de predicción: estudio clínico randomizado multicéntrico abierto (OPTIM-PTL). (OPTIM-PTL). Fundació de Recerca Clínic Barcelona-Institut d'Investigacions Biomèdiques August Pi i Sunyer. 2022. IP: Silvia - Irena Ferrero Martínez.
- Hydroxychloroquine efficacy in preventing SARS-CoV-2 infection and CoVid-19 disease severity during pregnancy. (CoVid-Preg). Fundació Privada ISGLOBAL. 2020. IP: Edurne Mazarico Gallego.
- Tratamiento del crecimiento intrauterino restringido precoz con heparina de bajo peso molecular: Ensayo Clínico Randomizado (TRACIP). (FSJD-TRACIP-2017). Fundació Privada per a la Recerca i la Docència Sant Joan de Déu - FSJD. 2017. IP: Edurne Mazarico Gallego.

### SELECTED PUBLICATIONS

- Basso A, Youssef L, Nakaki A, Paules C, Miranda J, Casu G et al. Fetal neurosonography at 31-35 weeks reveals altered cortical development in pre-eclampsia with and without small-for-gestational-age fetus. Ultrasound Obstet Gynecol. 2022;59(6):737-746. IF:8,678. (Q1).
- Boutet ML, Eixarch E, Ahumada

- P, Nakaki A, Crovetto F, Cívico MS et al. Fetal neurosonography and infant neurobehavior following conception by assisted reproductive technology with fresh or frozen embryo transfer. Ultrasound Obstet Gynecol. 2022;60(5):646-656. IF:8,678. (Q1).
- Meler E, Martinez RJ, Caradeux J, Mazarico E, Gil C, Boada D et al. Severe smallness as predictor of adverse perinatal outcome in suspected late small-for-gestational-age fetuses: systematic review and meta-analysis. Ultrasound Obstet Gynecol. 2022;60(3):328-337. IF:8,678. (Q1).
- Rubio R, Aguilar R, Bustamante M, Muñoz E, Vázquez M, Santano R et al. Maternal and neonatal immune response to SARS-CoV-2, IgG transplacental transfer and cytokine profile. Front. Immunol. 2022;13:999136-999136. IF:8,786. (Q1).

# Cardiovascular diseases in development

- 3** COMMERCIAL CLINICAL TRIALS
- 0** ACADEMIC CLINICAL TRIALS
- 15** PROJECTS
- 27** PUBLICATIONS
- 230.699** IMPACT FACTOR
- 59%** PUBLICATIONS IN 1RTS QUARTIL
- 19%** PUBLICATIONS IN 1RTS DECIL
- 52%** LEADERSHIP

**Joan Sánchez de Toledo, MD, PhD & Georgia Sarquella Brugada, MD, PhD**

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### TECHNICIAN

Adrian Ortega Sanabria

## SELECTED PROJECTS

- Identificación de predictores de arritmias cardíacas y muerte súbita en pacientes pediátricos afectos de enfermedades neuromusculares. (PI21/00094). Instituto de Salud Carlos III (ISCIII). 2022-2024. IP: Georgia Sarquella Brugada.
- Fundación Daniel Bravo Dr. Joan Sánchez de Toledo. Fundació Privada Daniel Bravo Andreu. 2022-2022. IP: Joan Sánchez de Toledo.
- Utilidad de la neuromonitorización y biomarcadores de daño cerebral como predictores del neurodesarrollo en pacientes sometidos a cirugía cardiaca los

primeros 6 meses de vida. Sociedad Española de Neonatología. 2021-2023. IP: Débora Cañizo Vázquez.

- Antioxidantes y neuroprotección en un modelo animal pediátrico de circulación extracorporea y parada circulatoria: nuevos horizontes terapéuticos. Hospital Sant Joan de Déu - Esplugues HSJD. 2021-2024. IP: Joan Sánchez de Toledo.

## SELECTED CLINICAL TRIALS & OBSERVATIONAL STUDIES

- Estudio multicéntrico para evaluar la seguridad y tolerabilidad a largo plazo de un tratamiento en abierto con sacubitrilo/valsartán en pacientes pediátricos con insuficiencia cardíaca debida a disfunción sistólica de ventrículo izquierdo sistémico, que han completado el estudio CLCZ696B2319. (CLCZ696B2319E1 - Ext.). Novartis Farmacéutica, S.A.; Pozas. 2020. IP: Joan Sánchez de Toledo.
- Estudio multicéntrico, abierto, para evaluar la seguridad, tolerabilidad, farmacocinética y farmacodinámica de LCZ696, seguido de un estudio de 52 semanas, aleatorizado, doble ciego, paralelo controlado con fármaco activo para evaluar la eficacia y seguridad de LCZ696 comparado con enalapril en pacientes pediátricos con edades desde 1 mes hasta < 18 años, con insuficiencia cardíaca debida a disfunción sistólica del ventrículo izquierdo sistémico. (CLCZ696B2319). Novartis Farmacéutica, S.A. 2019. IP: Joan Sánchez de Toledo.
- estudio abierto, fase IV, multicéntrico, para evaluar la eficacia diagnóstica de Nuubo nECGband antes del Holter Implantable. (Syn201401). Smart Solutions Tecnologies S.L. 2015. IP: Georgia Sarquella Brugada.

## SELECTED PUBLICATIONS

- Cañizo D, Hadley SM, Marta O, López M, Valls A, Viñals ML et al. Oxidative Stress and Indicators of Brain Damage Following Pediatric Heart Surgery. Antioxidants (Basel). 2022;11(3):489. IF:7,675. (Q1).
- Deyà A, Rivière JG, Roxo P, Ramakers J, Bloomfield M, Guisado P et al. Impact of JAK

Inhibitors in Pediatric Patients with STAT1 Gain of Function (GOF) Mutations-10 Children and Review of the Literature. J Clin Immunol. 2022;42(5):1071-1082. IF:8,542. (Q1).

- Escobar MC, Pérez M, Arráez M, Cascant MM, Albiach A, Kuligowski J et al. Brain Oxygen Perfusion and Oxidative Stress Biomarkers in Fetuses with Congenital Heart Disease-A Retrospective, Case-Control Pilot Study. Antioxidants (Basel). 2022;11(2): 299. IF:7,675. (Q1).
- Margarit A, Deyà A, Canizales JT, Vlagea A, Ana G, Marsal J et al. Case report: Challenges in immune reconstitution following hematopoietic stem cell transplantation for CTLA-4 insufficiency-like primary immune regulatory disorders. Front. Immunol. 2022;13:1070068-1070068. IF:8,786. (Q1).

## Immune and respiratory dysfunction in the critical child

	<b>5</b>	COMMERCIAL CLINICAL TRIALS
	<b>0</b>	ACADEMIC CLINICAL TRIALS
	<b>4</b>	PROJECTS
	<b>39</b>	PUBLICATIONS
	<b>264.948</b>	IMPACT FACTOR
	<b>59%</b>	PUBLICATIONS IN 1RTS QUARTIL
	<b>13%</b>	PUBLICATIONS IN 1RTS DECIL
	<b>72%</b>	LEADERSHIP

**Francisco José Cambra Lasaosa, MD, PhD**

At the time of this writing, we are participating in four clinical trials.

Three of these trials involve antiviral and antibacterial treatments in the field of infectious diseases. One of them seeks to evaluate an antiviral drug in the treatment of RSV bronchiolitis. Another is evaluating a drug for flu treatment in hospitalised children up to the age of six months. The third trial is analysing an antibiotic drug in the treatment of hospital-acquired pneumonia.

The fourth trial in which we are taking part evaluates the efficiency of an inhaled anaesthetic in the handling of mechanically ventilated patients.

Likewise, we are working on the analysis of the data of the financed project focused on the study of the bacterial nasopharyngeal microbiota in the development of mechanical ventilation-associated pneumonia in our intensive care unit.

We are continuing the study, on an animal model, of the neuroprotective effect of dexmedetomidine as a protector in extracorporeal circulation and circulatory arrest. The study has

been completed on rabbits and is currently being carried out on pigs.

Over 30 publications have been made in relation to our group's research lines, in journals with a high impact factor. Four of these publications are listed in the respective section.

During this period, two members of our team have obtained doctorate degrees *cum laude*.

### MEMBERS

#### STAFF RESEARCHER

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#### COLLABORATOR RESEARCHER

Montserrat Esquerda Areste

TBD

Sergi Huerta Calpe, Monica Girona Alarcon, Ricardo Suárez Ramírez

### SELECTED PROJECTS

- Implicación de la microbiota bacteriana de la nasofaringe en el desarrollo de neumonía asociada a ventilación mecánica en la unidad de cuidados intensivos pediátricos. Estudio MiNoRep. (PI19/00106). Instituto de Salud Carlos III (ISCIII). 2020-2023. IP: Iolanda Jordán García.
- Valoración del efecto neuoprotector de la dexmedetomidina en el modelo animal pediatrico de circulación extracorporea y parada circulatoria. Sociedad y Fundación Española de Cuidados Intensivos Pediátricos (SECIP). 2018-2022. IP: Sergio Benito Fernández.
- ARDS in CHILDREN and ECMO initiation strategies impact on NEURO-DEVELOPMENT - ASCEND. (SUBK00015894). National Institute of Health (IEUUU). 2020-2023. IP: Susana Segura Matute.

### SELECTED CLINICAL TRIALS & OBSERVATIONAL STUDIES

- Estudio aleatorizado con control activo para comparar la eficacia y

la seguridad de isoflurano inhalado administrado por el AnaConDa-S (dispositivo de conservación de anestésico) y el midazolam intravenoso para la sedación en pacientes pediátricos con ventilación mecánica de entre 3 y 17 años (menos de 18 años). A Randomised Active-controlled Study to Compare Efficacy and Safety of Inhaled Isoflurane Delivered by the AnaConDa-S (Anaesthetic Conserving Device) to Intravenous Midazolam for Sedation in Mechanically Ventilated Paediatric Patients 3 to 17 (Less than 18) Years Old. (SED002). LINK Medical Research; Sedana Medical AB. 2021. IP: Monica Girona Alarcon.

- Estudio de fase IIa sin enmascaramiento en lactantes con infección de las vías Respiratorias bajas por el Virus Sincitial Respiratorio, seguido de una parte con doble enmascaramiento y controlada con placebo para evaluar la seguridad, la tolerabilidad, la farmacocinética y el efecto antivírico de RV521 (REVIRAL1). A Phase I/IIa Open-label Study in Infants with REspiratory Syncytial VIRus Lower RespirAtory Tract Infection, Followed by a Doubt-blind, Placebo-controlled Part, to Evaluate the Safety, Tolerability, Pharmacokinetics and Antiviral Effect of RV521. (REVC003). Chavez; ReViral Ltd; Syneos Health Clinical Spain S.L.U. 2021. IP: Iolanda Jordán García.
- Estudio abierto, de un solo brazo para evaluar la farmacocinética, seguridad y tolerabilidad, y para explorar los resultados clínicos del tratamiento con dosis única o múltiple de zanamivir IV en neonatos e infantes menores de 6 meses con infección por gripe confirmada complicada. An open label, single arm study to evaluate the pharmacokinetics, safety and tolerability of intravenous (IV) zanamivir in hospitalised neonates and infants under 6 months of age with confirmed or suspected influenza infection. (200925). Glaxosmithkline, S.A. 2020. IP: Mónica Balaguer Gargallo.
- A Phase 1, open-label, non-comparative, multicenter clinical trial to evaluate the safety, tolerability, and pharmacokinetics of ceftolozane/tazobactam (MK-7625A) in pediatric participants

with nosocomial pneumonia./  
Estudio clínico de fase 1, abierto,  
no comparativo y multicéntrico  
para evaluar la seguridad, la  
tolerabilidad y la farmacocinética  
de ceftolozano/tazobactam (MK-  
7625A) en participantes pediátricos  
con neumonía hospitalaria. (MK-  
7625A-036). Fernández; Merck  
Sharp & Dohme LLC; Merck, Sharp  
& Dohme de España. S.A. 2019. IP:  
Iolanda Jordán García.

#### SELECTED PUBLICATIONS

- Fauroux B, Abel F, Amaddeo A, Bignamini E, Chan E, Corel L et al. ERS statement on paediatric long-term noninvasive respiratory support. *Eur Respir J.* 2022; 59(6): 2101404. IF:33,795. (Q1).
- Girona M, Argüello G, Esteve A, Bobillo S, Paolo X, Bonet E et al. Low levels of CIITA and high levels of SOCS1 predict COVID-19 disease severity in children and adults. *iScience.* 2022;25(1):103595-103595. IF:6,107. (Q1).
- Guitart C, Rodríguez J, Bobillo S, Carrasco JL, Inarejos E, Cambra FJ et al. An algorithm combining procalcitonin and lung ultrasound improves the diagnosis of bacterial pneumonia in critically ill children: The PROLUSP study, a randomized clinical trial. *Pediatr Pulmonol.* 2022;57(3):711-723. IF: 4,090. (Q1).
- Jordán I, Fernández de Sevilla M, Fumadó V, Bassat Q, Bonet E, Fortuny C et al. Transmission of Severe Acute Respiratory Syndrome Coronavirus 2 Infection Among Children in Summer Schools Applying Stringent Control Measures in Barcelona, Spain. *Clin Infect Dis.* 2022;74(1):66-73. IF: 20,999. (Q1).
- Lorenzo-Izquierdo D, Esquerda M, Palau F, Cambra FJ, Grup Investigació en Bioètica. Ethics and Genomic Editing Using the Crispr-Cas9 Technique: Challenges and Conflicts. *NanoEthics.* 2022; IF:1,333. (Q2).



## Allergic diseases in childhood and adolescence

- 10** COMMERCIAL CLINICAL TRIALS
- 0** ACADEMIC CLINICAL TRIALS
- 5** PROJECTS
- 9** PUBLICATIONS
- 82.91** IMPACT FACTOR
- 67%** PUBLICATIONS IN 1RTS QUARTIL
- 56%** PUBLICATIONS IN 1RTS DECIL
- 22%** LEADERSHIP

### Montserrat Alvaro Lozano, MD, PhD

During 2022 some of our scientific projects have been taking shape as we have been working on the recruitment of patients for some of them and statistical evaluation for others.

We have also been working on some other studies that are going to be (if accepted by Universitat de Barcelona) doctoral Thesis for two of our components (D. Olga Domínguez and Dr Adrianna Machinena).

Meanwhile we have been recruiting patients for clinical international studies (Phase II and phase III). These are going to be a great advance in the treatment of allergic children, as they involve respiratory, cutaneous an food allergy.

### MEMBERS

#### STAFF RESEARCHER

Rosa Jiménez Feijoo, Jaime Lozano Blasco, Adrianna Machinena Spera, María del Mar Folqué Giménez, Mónica Piquer Gibert

#### PHD STUDENTS

Olga Dominguez Sánchez

### SELECTED PROJECTS

- Estudio de hipersensibilidad a anti-inflamatorios no esteroideos (AINES) en población pediátrica. Sociedad

Española de Inmunología Clínica, Alergia y Asma Pediátrica. 2022-2023. IP: Adrianna Machinena Spera.

- Estudio de la alergenidad de diferentes proteïnes en la alergia alimentaria a pescado en población pediátrica. Sociedad Española de Inmunología Clínica, Alergia y Asma Pediátrica. 2021-2023. IP: Olga Dominguez Sánchez.
- Genetics of Food Allergy Barcelona Montserrat Alvaro. Max Delbrück Centrum für Molekulare medizin Berlin-Buch. 2021-2023. IP: Montserrat Alvaro Lozano.
- A safety/hypoallergenicity study for a Cow's Milk Substitute Hypo-Allergenic Rice-based infant formula with added Donkey Milk - The CHARM Study. (CHARM-01-2020). Dr. Schär SPA. 2020. IP: Montserrat Alvaro Lozano.

### SELECTED CLINICAL TRIALS & OBSERVATIONAL STUDIES

- Ensayo clínico de eficacia y seguridad, prospectivo, aleatorizado, doble ciego, de doble simulación, controlado con placebo, multicéntrico, con inmunoterapia en pacientes con asma de leve a moderada controlada y rinitis/ rinoconjuntivitis, alérgicos a Dermatophagooides pteronyssinus y/o Dermatophagooides farinae. (MM09-SIT-040). BioClever 2005, SLU; Escoruela; Inmunotek, .SL. 2022. IP: Adrianna Machinena Spera.
- Protocolo maestro de tres estudios aleatorizados, controlados con placebo, doble ciegos, multicéntricos, de grupos paralelos de dupilumab en pacientes con urticaria crónica espontánea (UCE) que siguen siendo sintomáticos a pesar del uso de tratamiento antihistamínico H1 en pacientes sin tratamiento previo con omalizumab y en pacientes intolerantes o con respuesta incompleta a omalizumab. (EFC16461-CUPID). Farmacia; Laboratori HSJD; Ranz; Sanofi-Aventis S.A; Servicio de Cardiología; Unitat de Recerca FSJD. 2022. IP: Montserrat Alvaro Lozano.
- Estudio de extensión, abierto, multicéntrico y de un solo grupo para evaluar la seguridad a largo plazo de GSK3511294 (depemokimab) en participantes adultos y adolescentes con asma grave con fenotipo eosinofílico de los estudios 206713 o 213744 (AGILE). A multi-centre,

single-arm, open-label extension study to evaluate the long-term safety of GSK3511294 (Depemokimab) in adult and adolescent participants with severe asthma with an eosinophilic phenotype from studies 206713 or 213744. (212895). GlaxoSmithKline Research and Development Ltd.; IQVIA RDS Spain SL. 2022. IP: Jaime Lozano Blasco.

- Estudio multicéntrico, aleatorizado, doble ciego y controlado con placebo de ligelizumab (QGE031) en el tratamiento de la urticaria crónica inducible (CINDU) en adolescentes y adultos inadecuadamente controlados con antihistamínicos H1. (CQGE031E12301). Novartis Farmacéutica, S.A. 2022. IP: Montserrat Alvaro Lozano.
- Estudio multicéntrico, aleatorizado, doble ciego, controlado con placebo de 52 semanas de duración para evaluar la eficacia clínica y la seguridad de ligelizumab (QGE031) en la disminución de la sensibilidad a los cacahuetes en pacientes con alergia al cacahuete. (CQGE031G12301). Balcell; Novartis Farmacéutica, S.A. 2022. IP: Montserrat Alvaro Lozano.

### SELECTED PUBLICATIONS

- Jutel M, Torres MJ, Palomares O, Akdis CA, Eiwegger T, Untermann E et al. COVID-19 vaccination in patients receiving allergen immunotherapy (AIT) or biologicals-EAACI recommendations. Allergy. 2022;77(8):2313-2336. IF:14,710. (Q1).
- Muraro A, de Silva D, Halken S, Worm M, Khaleva E, Arasi S et al. Managing food allergy: GA(2) LEN guideline 2022. World Allergy Organ J. 2022;15(9):100687-100687. IF:5,516. (Q2).
- Rodríguez Del Río P, Alvarez A, Blumchen K, Caimmi D, Caubet JC, Konstantinopoulos AP et al. Food immunotherapy practice: Nation differences across Europe, the FIND project. Allergy. 2022;77(3):920-932. IF:14,710. (Q1).
- Zuberbier T, Dörr T, Aberer W, Alvaro M, Angier E, Arasi S et al. Proposal of 0.5 mg of protein/100 g of processed food as threshold for voluntary declaration of food allergen traces in processed food-A first step in an initiative to better inform patients and avoid fatal allergic reactions: A GA<sup>2</sup>LEN position paper. Allergy. 2022;77(6):1736-1750. IF:14,710. (Q1).

# 7.1 MEDICINE AND PEDIATRIC SCIENCE AREA

## 7.1.3 Paediatric cancer

Translational genomics

Sarcomas and histiocytosis

Therapy development in pediatric oncology

Paediatric leukaemia and haematopoiesis

Molecular pharmacology and experimental therapies

Paediatric cancer epigenetics



## Translational genomics

- 2** COMMERCIAL CLINICAL TRIALS
- 0** ACADEMIC CLINICAL TRIALS
- 9** PROJECTS
- 9** PUBLICATIONS
- 100.716** IMPACT FACTOR
- 67%** PUBLICATIONS IN 1RTS QUARTIL
- 44%** PUBLICATIONS IN 1RTS DECIL
- 44%** LEADERSHIP

**Cecilia Emilia Lavarino, PhD**

### MEMBERS

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Alba Parra Checa

#### TECHNICIAN

Cristina Darbra Magrí, Sonia Paco Mercader

### SELECTED PROJECTS

- SEHOP-PENCIL study - Personalised mEdiciNe for Cancer in chILdren in Spain. (PMP21/00075). Instituto de Salud Carlos III (ISCIII). 2022-2025. IP: Cinzia Emilia Lavarino.
- Desarrollo de clasificadores epigenéticos para el Cáncer Pediátrico (ECPC). (PI20/00519). Instituto de Salud Carlos III (ISCIII). 2021-2023. IP: Cinzia Emilia Lavarino.
- Platform of Epigenetic Classifiers for Childhood CAncer (PECA). (201921-30). Fundació La Marató de TV3. 2020-2023. IP: Cinzia Emilia Lavarino.
- Método para la clasificación en subgrupos moleculares de pacientes con Meduloblastoma. (PI17/00353). Instituto de Salud Carlos III (ISCIII). 2018-2022. IP: Cinzia Emilia Lavarino.

### SELECTED CLINICAL TRIALS & OBSERVATIONAL STUDIES

- Estudio posautorización de seguridad de pacientes pediátricos que inician el tratamiento con selumetinib: Estudio de cohortes prospectivo de varios países. Post-Authorisation Safety Study of Paediatric Patients Initiating Selumetinib: A Multiple-Country Prospective Cohort Study. (D1346R00004). Astrazeneca Farmaceutica Spain, S.A.; Labcorp Drug Development Inc. 2022. IP: Héctor Salvador Hernández.
- Ensayo de Fase I/II, Abierto y de un solo brazo para Evaluar la Farmacocinética, Seguridad/ Tolerabilidad y Eficacia de la Formulación Granulada de Selumetinib en niños de  $\geq 1$  a  $< 7$  Años con Neurofibromas Plexiformes (NP) Sintomáticos e Inoperables relacionados con Neurofibromatosis Tipo 1 (NF1) (SPRINKLE). (D1346C00004). Astrazeneca Farmaceutica Spain, S.A.; Diagnóstico para la imagen (DXI); Farmacia; Hospital de dia PCCB; Hospitalización HSJD; Laboratori HSJD; Medina; Servicio de Cardiología; Servicio Oftalmología; Unitat de Recerca FSJD. 2021. IP: Héctor Salvador Hernández.
- Ensayo fase I, de un solo brazo, secuencial, para evaluar el efecto de los alimentos sobre la seguridad y farmacocinética de selumetinib después de dosis múltiples en niños adolescentes con neurofibromas plexiformes (NP) asociados a neurofibromatosis tipo 1 (NF1). A Phase I, Single-Arm, Sequential Study to Evaluate the Effect of Food on the Gastrointestinal Toxicity and Pharmacokinetics of Selumetinib after Multiple Doses in Adolescent Children with Neurofibromatosis Type 1 (NF1) Related Plexiform Neurofibromas (PN). (D1346C00015). Astrazeneca Farmaceutica Spain, S.A.; Diagnóstico para la imagen (DXI); Farmacia; Hospitalización HSJD; Laboratori HSJD; Servicio de Cardiología; Servicio Oftalmología; Unitat de Recerca FSJD; Vázquez. 2021. IP: Héctor Salvador Hernández.

### SELECTED PUBLICATIONS

- Carvalho DM, Richardson PJ, Olaciregui N, Stankunaite R, Lavarino C, Molinari V et al. Repurposing Vandetanib plus

- Everolimus for the Treatment of ACVR1-Mutant Diffuse Intrinsic Pontine Glioma. *Cancer Discov.* 2022;12(2):416-431. IF:38,272. (Q1).
- Cuadrado M, Liu J, Paco S, Aschero MR, Burgeño V, Sirab N et al. Identification of immunosuppressive factors in retinoblastoma cell secretomes and aqueous humor from patients. *J Pathol.* 2022;257(3):327-339. IF:9,883. (Q1).
- Gonçalves E, Garcia M, Rodriguez CJ, Gomez S, Ecker RC, Suñol M et al. AC-265347 Inhibits Neuroblastoma Tumor Growth by Induction of Differentiation without Causing Hypocalcemia. *Int J Mol Sci.* 2022;23(8): 4323. IF:6,208. (Q1).
- Pascual G, Castillo H, Unceta N, Aschero MR, Resa C, Gómez A et al. SPARC-mediated long-term retention of nab-paclitaxel in pediatric sarcomas. *J Control Release.* 2022;342:81-92. IF:11,467. (Q1).

## Sarcomas and histiocytosis

- 14** COMMERCIAL CLINICAL TRIALS
- 8** ACADEMIC CLINICAL TRIALS
- 18** PROJECTS
- 15** PUBLICATIONS
- 166.075** IMPACT FACTOR
- 73%** PUBLICATIONS IN 1RTS QUARTIL
- 33%** PUBLICATIONS IN 1RTS DECIL
- 53%** LEADERSHIP

### Jaume Mora Graupera, MD, PhD

2022 culminated 20 years of group development with the opening in June of the Pediatric Cancer Center Barcelona (PCCB), a unique center in Europe where science and clinical assistance meets for pediatric cancer patients. This is a fully philanthropic sponsored center where families and donors understood, long time ago, that only through research we change the life history of patients affected with developmental cancer. In October of 2022 we hosted the annual congress of the International Society of Pediatric Oncology (SIOP) in Barcelona with record assistance and success at all levels which was accredited to the local organizing committee led by the PCCB researchers.

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Óscar Muñoz Aznar, Noelia Salvador Marcos

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Maria Inmaculada Hernandez Muñoz

### SELECTED PROJECTS

- Contratos predoctorales de formación en investigación en salud\_Beneficiario: Pablo Taboas. [FI21/00047]. Instituto de Salud Carlos III [ISCI]. 2022-2025. IP: Jaume Mora Graupera.
- Influencia del contexto epigenético en la edad como factor pronóstico en Ewing Sarcoma. [PI20/00179]. Instituto de Salud Carlos III [ISCI]. 2021-2023. IP: Jaume Mora Graupera.
- Virtual patient derived xenografts for tumor treatment. [HR20-00411]. Fundació Bancària "La Caixa". 2020-2023. IP: Jaume Mora Graupera.
- Unveiling the Ewing sarcoma cell of origin: a multistep transformation process that involves prenatal genetic and postnatal epigenetic mechanisms. [201928-30]. Fundació La Marató de TV3. 2020-2023. IP: Jaume Mora Graupera.
- Molecular analysis of the metastatic process in developmental tumors. [201925-31]. Fundació La Marató de TV3. 2020-2023. IP: Silvia Mateo Lozano.

### SELECTED CLINICAL TRIALS & OBSERVATIONAL STUDIES

- Estudio de fase 2, aleatorizado, sin enmascaramiento, en el que se evalúa abemaciclib en combinación con irinotecán y temozolomida en participantes con sarcoma de Ewing recidivante o resistente al tratamiento. A Randomized, Open-Label, Phase 2 Study Evaluating Abemaciclib in Combination with Irinotecan and Temozolomide in Participants with Relapsed or Refractory Ewing's Sarcoma. [J1S-MC-JP04]. Alfano; Anatomía patológica; Diagnóstico para la imagen [DXI]; Farmacia; Hospital de dia PCCB; Laboratori HSJD; Lilly S.A; Servicio de Cardiología; SIMM Molecular S.L.; Unitat de Recerca FSJD. 2022. IP: Jaume Mora Graupera.

- Estudio (Pegasus Lymphoma 205) de fase 2 no aleatorizado, abierto, multicéntrico y de múltiples cohortes que evalúa el beneficio clínico de SAR444245 (THOR-707) con o sin otros tratamientos antineoplásicos para el tratamiento de adultos y adolescentes con linfoma de células B en recaída o refractario. A Phase 2 non-randomized, open-label, multi-cohort, multi-center study assessing the clinical benefit of SAR444245 (THOR-707) with or without other anticancer therapies for the treatment of adults and adolescents with relapsed or refractory B-cell lymphoma. [ACT16941]. Sanofi-Aventis S.A. 2022. IP: Verónica Celis Passini.
- Ensayo Clínico plataforma en fase II de inmuno-oncología de precisión con indicación tumor agnóstica para mutaciones somáticas específicas [TAPISTRY]. Tumor-agnostic precision immuno-oncology and somatic targeting rational for you (Tapistry) phase II platform trial. [B041932]. Anatomía patológica; Diagnóstico para la imagen [DXI]; Farmacia; Hospital de dia PCCB; Hospitalización HSJD; Laboratori HSJD; Roche Farma, S.A; Servicio de Cardiología; Servicio Dermatología; Servicio Genética i Molecular; Servicio Oftalmología; SIMM Molecular S.L.; Unitat de Recerca FSJD. 2021. IP: Jaume Mora Graupera.

### SELECTED PUBLICATIONS

- Castañeda A, Gorostegui M, Miralles SL, Chamizo A, Patiño SC, Flores MA et al. How we approach the treatment of patients with high-risk neuroblastoma with naxitamab: experience from the Hospital Sant Joan de Déu in Barcelona, Spain. ESMO Open. 2022;7(2):100462-100462. IF:6,883. (Q1).
- Mora J. Autologous Stem-Cell Transplantation for High-Risk Neuroblastoma: Historical and Critical Review. Cancers [Basel]. 2022;14(11)IF:6,575. (Q1).
- Molnar C, Reina J, Herrero A, Heinen JP, Méndiz V, Bonnal S et al. Human EWS-FLI protein recapitulates in Drosophila the neomorphic functions that induce Ewing sarcoma tumorigenesis. PNAS Nexus. 2022;1(4):222.

# Therapy development in pediatric oncology

- 0** COMMERCIAL CLINICAL TRIALS
- 3** ACADEMIC CLINICAL TRIALS
- 19** PROJECTS
- 20** PUBLICATIONS
- 369.9** IMPACT FACTOR
- 80%** PUBLICATIONS IN 1RTS QUARTIL
- 45%** PUBLICATIONS IN 1RTS DECIL
- 20%** LEADERSHIP

**Angel Montero Carcaboso, PhD**

## MEMBERS

### POSTDOC ASSOCIATED RESEARCHERS

Maria del Rosario Aschero

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Ana Jiménez Cabaco, Mònica Vilà Ubach

### COLLABORATOR RESEARCHERS

Guillermo Chantada , Ofelia Cruz Martínez, Andrés Morales La Madrid

## SELECTED PROJECTS

- Immune microenvironment of retinoblastoma. (PI21/00216). Instituto de Salud Carlos III (ISCIII). 2022-2024. IP: Angel Montero Carcaboso.
- Desenvolupament preclínic del fàrmac G2B-001 i aplicació en el tractament de tumors sòlids pediàtrics. (2021 DI 008). Agaur - Agència de Gestió d'Ajuts Universitaris i de Recerca. 2021-2024. IP: Angel Montero Carcaboso.
- Inhibition of the PI3K/Akt pathway in pediatric high grade and diffuse

intrnsic pontine gliomas (pHGG/ DIPG). [201927-30]. Fundació La Marató de TV3. 2020-2023. IP: Angel Montero Carcaboso.

Sci. 2022;23(8): 4323. IF:6,208. (Q1).

- Pascual G, Castillo H, Unceta N, Aschero MR, Resa C, Gómez A et al. SPARC-mediated long-term retention of nab-paclitaxel in pediatric sarcomas. J Control Release. 2022;342:81-92. IF:11,467. (Q1).

## SELECTED CLINICAL TRIALS & OBSERVATIONAL STUDIES

- SIOP Ependimoma II - Programa clínico internacional para el diagnóstico y tratamiento de niños, adolescentes y adultos jóvenes con ependimoma. [ET-13-002]. Cetir Centre Diagnòstic Pedralbes S.A; Sociedad Española de Hematología y Oncología Pediátricas. 2017. IP: Ofelia Cruz Martínez.
- Phase Ib Clinical Trial on the Safety of Immunotherapy With Autologous Dendritic Cells Primed With Lysate Allogeneic Tumor Lines in Patients With Diffuse Intrinsic Pontine Glioma (DIPG). [FSJD-DIPG-DC]. Fundació Privada per a la Recerca i la Docència Sant Joan de Déu - FSJD. 2016. IP: Andrés Morales La Madrid.
- Estudio prospectivo internacional en niños mayores de 3 a 5 años con meduloblastoma de riesgo estándar clínico de perfil biológico de bajo riesgo. [SIOP - PNET 5 MB - LR]. Anatomía patológica; Diagnóstico para la imagen (DXI); Laboratori HSJD; Servicio de Cardiología; SERVICIO DE CIRUGIA; Servicio Otorrinolaringología; Sociedad Española de Hematología y Oncología Pediátricas; Unitat de Recerca FSJD. 2016. IP: Ofelia Cruz Martínez.

## SELECTED PUBLICATIONS

- Cuadrado M, Liu J, Paco S, Aschero MR, Burgeño V, Sirab N et al. Identification of immunosuppressive factors in retinoblastoma cell secretomes and aqueous humor from patients. J Pathol. 2022;257(3):327-339. IF:9,883. (Q1).
- Gállego Pérez J, Garcia M, Labiano S, Patiño A, Dobbs J, Gonzalez M et al. Oncolytic DNX-2401 Virus for Pediatric Diffuse Intrinsic Pontine Glioma. N. Engl. J. Med. 2022;386(26):2471-2481. IF:176,079. (Q1).
- Gonçalves E, Garcia M, Rodriguez CJ, Gomez S, Ecker RC, Suñol M et al. AC-265347 Inhibits Neuroblastoma Tumor Growth by Induction of Differentiation without Causing Hypocalcemia. Int J Mol

## Paediatric leukaemia and haematopoiesis

- 22** COMMERCIAL CLINICAL TRIALS
- 6** ACADEMIC CLINICAL TRIALS
- 11** PROJECTS
- 23** PUBLICATIONS
- 222.462** IMPACT FACTOR
- 65%** PUBLICATIONS IN 1RTS QUARTIL
- 35%** PUBLICATIONS IN 1RTS DECIL
- 43%** LEADERSHIP

**Mireia Camós Guijosa, MD, PhD**

Listed below are some of the advances made by the Paediatric Leukaemia and Other Haematologic Diseases Group in 2022:

- In the GEMMA study for the complete characterisation of childhood myelodysplastic syndromes and hereditary acute myeloid leukaemias, a project funded by the TV3 Telethon Foundation, Dr Català has succeeded in establishing the first register in Spain of patients with syndromes of predisposition to haematological neoplasias, and we are advancing in the biological characterisation of these syndromes.
- Thanks to the project financed by the One-in-a-hundred-thousand Foundation, we have succeeded in implementing the complex methodology required to launch the ALLTogether international clinical trial, which will be benefiting in the near future all the children with acute lymphoblastic leukaemia in Spain.
- The CAR-T Unit team continues to lead several clinical trials on CAR-T cell therapy in acute leukaemia. Likewise, Drs Rives, Torrebadell

and Vega are leading clinical and biological projects financed with competitive funds, aimed to improve the prognosis of patients undergoing this type of treatments.

- Drs Camós, Torrebadell and Vega are leading projects addressed to improving the diagnosis, follow-up and prognosis of high-risk leukaemias. These projects have shown significant results in 2022, some of which have recently been published in international journals.
- The international CLOSER project, funded by the European Commission and led by Dr Camós, continued to advance on standardisation and training in diagnostic techniques and on the fostering of the active participation of patients, families and support organizations in the handling of children with leukaemia in Europe and Latin America ([www.closerleukemia.org](http://www.closerleukemia.org)).

### MEMBERS

#### STAFF RESEARCHER

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Mercé Richarte Franqués, Alba Crespo Carrasco, Andrea Ruiz Sanjuan

### SELECTED PROJECTS

- Nuevas técnicas para la caracterización molecular completa y la mejora del seguimiento de la enfermedad mínima residual en leucemia aguda pediátrica. (PI21/00213). Instituto de Salud Carlos III (ISCIII). 2022-2024. IP: Mireia Camós Guijosa, Nerea Vega García.
- Análisis de los factores pronósticos de fracaso del tratamiento tras

terapia con células T con receptores antigenicos químéricos (CAR-T) en pacientes pediátricos con leucemia linfoblástica aguda de línea B. (PI21/00218). Fundació Privada per a la Recerca i la Docència Sant Joan de Déu - FSJD; Instituto de Salud Carlos III (ISCIII). 2022-2024. IP: Susana Rives Solà, Montserrat Torrebadell Burriel.

- Implementación de la estrategia de tratamiento ALLTogether para leucemia linfoblástica aguda pediátrica en España. Fundación UnoentreCienMil. 2021-2022. IP: Susana Rives Solà, Mireia Camós Guijosa.

### SELECTED CLINICAL TRIALS & OBSERVATIONAL STUDIES

- Estudio multicéntrico fase 2 en pacientes pediátricos (0-18 años) con leucemia linfoblástica aguda B CD19+ en recidiva o refractaria. (ICI21/00033). Instituto de Salud Carlos III (ISCIII). 2022-2025. IP: Susana Rives Solà.
- A Phase Ib study of Vyxeos® (liposomal daunorubicin and cytarabine) in combination with Clofarabine in children with relapsed/refractory AML. (ITCC-092). Diagnóstico para la imagen (DXI); Farmacia; Laboratori HSJD; Prinses Máxima Centrum voor Kinderoncologie B.V; Servicio de Cardiología; Unitat de Recerca FSJD. 2022. IP: Albert Català Temprano.
- Un estudio de fase III, abierto, de un solo grupo, multicéntrico de Ravulizumab junto al mejor tratamiento sintomático en participantes pediátricos (de 1 mes a < 18 years of age) with Thrombotic Microangiopathy (TMA) after Hematopoietic Stem Cell transplantation (HSCT). (ALXN1210-TMA-314). Alexion Pharmaceuticals Inc.; Diagnóstico para la imagen (DXI); Farmacia; Hospital de dia PCCB; IQVIA RDS Spain SL; Laboratori HSJD; Mollica; Servicio de Cardiología; Servicio de Trasplante TPH; Unitat de Recerca FSJD. 2021. IP: Julia Marsal Ricomà.
- Estudio de fase 2, multicéntrico, abierto para evaluar la dosis adecuada y la seguridad de crizanlizumab, con o sin hidroxiurea/hydroxicarbamida, en grupos secuenciales de

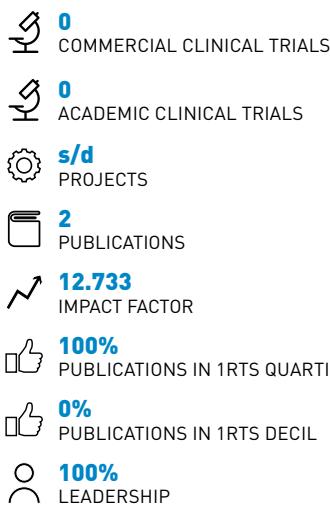
edad descendente de pacientes pediátricos con Enfermedad de Células Falciformes con crisis vaso-occlusivas. [CSEG101B2201]. Miranda; Novartis Farmacéutica, S.A. 2019. IP: Anna Ruiz Llobet.

#### SELECTED PUBLICATIONS

- Bueno C, Barrena S, Bataller A, Ortiz V, Elliott N, O'Byrne S, et al. CD34+CD19-CD22+ B-cell progenitors may underlie phenotypic escape in patients treated with CD19-directed therapies. *Blood*. 2022;140(1):38-44. IF:25,476. (Q1).
- Laetsch TW, Maude SL, Rives S, Hiramatsu H, Bittencourt H, Bader P et al. Three-Year Update of Tisagenlecleucel in Pediatric and Young Adult Patients With Relapsed/Refractory Acute Lymphoblastic Leukemia in the ELIANA Trial. *J Clin Oncol*. 2022;IF:50,717. (Q1).
- Ortiz-Maldonado V, Alonso-Saladrígues A, Español-Rego M, Martínez-Cibrián N, Faura, A, Magnano L, et al. Results of ARI-0001 CART19 cell therapy in patients with relapsed/refractory CD19-positive acute lymphoblastic leukemia with isolated extramedullary disease. *Am J Hematol*. 2022;97(6):731-739. IF:13,265. (Q1).
- Ruiz-Llobet A, Gassiot S, Sarrate E, Zubicaray J, Dapena JL, Rives S, et al. Venous thromboembolism in pediatric patients with acute lymphoblastic leukemia under chemotherapy treatment. Risk factors and usefulness of thromboprophylaxis. Results of LAL-SEHOP-PETHEMA-2013. *J. Thromb Haemost*. 2022;20(6):1390-1399. IF:16,036. (Q1).
- Vicente-Garcés C, Esperanza-Cebollada E, Montesdeoca S, Torrebadell M, Rives S, Dapena JL, et al. Technical Validation and Clinical Utility of an NGS Targeted Panel to Improve Molecular Characterization of Pediatric Acute Leukemia. *Front Mol Biosci*. 2022;9:854098-854098. IF:6,113. (Q1).



# Molecular pharmacology and experimental therapies



## Marçal Pastor-Anglada, PhD

The bioavailability of certain anti-tumour drugs whose mechanism of action is interference with nucleotide metabolism depends on transporter proteins located in the cell membrane. The expression of these transporters is altered in both solid and liquid tumours and it is not uncommon for a specific subtype responsible for the uptake of these drugs, such as the hCNT1 protein, to present negligible levels in tumours, which in turn may contribute to tumour metabolic reprogramming, and is also a cause of chemoresistance. The main contribution of our group last year was the demonstration that the loss of hCNT1 expression in a wide range of tumours has an epigenetic origin, associated with the deregulation of the expression pattern of miRNAs in tumours. The oncomiRs miR-106a and miR-17 negatively and directly regulate the nucleoside-derived drug transporter hCNT1. Silencing these miRNAs restores the expression and part of the function of this drug transporter, which contributes to the improvement of the cellular response to these anti-tumour agents. We believe that this is a new epigenetic mechanism that may condition the response to chemotherapy.

## MEMBERS

### STAFF RESEARCHER

Sandra Perez-Torras

### POSTDOC ASSOCIATED RESEARCHERS

Liska Caviedes Cárdenas, Jose Antonio Agüero Fernández, Aida Mata Ventosa, Nerea Urtasun Plans

### PHD STUDENTS

Andrea Valentina Prieto Carruyo

- Pastor M, Mata A, Perez S. Inborn Errors of Nucleoside Transporter (NT)-Encoding Genes (SLC28 and SLC29). *Int. J. Mol. Sci.* 2022;23(15):8770IF:5,600. (Q1).

## SELECTED PROJECTS

- Doctorats industrials 2019.  
Empresa: Laboratorios Reig Jofré, S.A. Agència de Gestió d'Ajuts Universitaris i de Recerca (AGAUR). Ajuts a universitats i a empreses per al desenvolupament d'un projecte de doctorat industrial, en qualsevol àmbit de coneixement i qualsevol sector empresarial. Doctorats industrials 2019. 2019DI005. PI: Sandra Pérez Torras. 2019-2022.
- Proteínas transportadoras de nucleósidos (NT), metabolismo de nucleótidos y cáncer. Ministerio de Ciencia, Innovación y Universidades. Convocatòria d'ajuts del Programa Estatal d'Investigació, Desenvolupament i Innovació Orientada als reptes de la Societat. Modalitat Reptes. RTI2018-094655-B-I00. PI: Marçal Pastor Anglada. 2019-2022.
- Descubrimiento y desarrollo de nuevos tratamientos Inmuno-Oncológicos [RTC2019-006865-1]. Ministerio de Economía y Competitividad. Convocatòria Reptes-Col·laboració del Programa Estatal de Recerca, Desenvolupament i Innovació Orientada als Reptes de la Societat. RTC2019-006865-1. PI: Marçal Pastor Anglada. 2020-2022.
- Desoxirribonucleòsids com a teràpia per als trastorns de la replicació de l'ADN mitocondrial: investigació dels mecanismes terapèutics i ampliació del tractament a les mutacions en POLG i altres gens. Fundació La Marató de TV3. Ajuts econòmics a projectes de recerca de la Fundació Marató de TV3. 97/C/2020. PI: Marçal Pastor Anglada. 2021-2024.

## SELECTED PUBLICATIONS

- Brunet M, Pastor M. Insights into the Pharmacogenetics of Tacrolimus Pharmacokinetics and Pharmacodynamics. *Pharmaceutics*. 2022;14(9):1755IF:5,400. (Q1).

# Paediatric cancer epigenetics



## Alexandra Avgustinova, PhD

The overarching goal of our research group is to identify specific epigenomic vulnerabilities of developmental tumours, and to exploit them in the pursuit of personalised therapies, thus improving patient welfare and prognosis. We believe that one size does not fit all, and we need to treat each individual, not the disease. During 2022 we have progressed along our established lines of research, but also the research topics we work on to include studying the metabolic function of histone methylation, and developing novel models for Ewing Sarcoma. In addition, our team was awarded an ERC Starting Grant entitled "Oncogenic Competence – When, Where and Why?" (ERC), which constitutes funding for 5 years that will allow us to initiate work with transgenic mouse models, and incorporate further members into our research group during 2023. Additionally, during 2022 we have incorporated new members into the group and expanded our national and international collaborator network: together we will bring about change sooner.

## MEMBERS

### POSTDOC ASSOCIATED RESEARCHERS

Carlos Javier Rodriguez Hernandez

### PHD STUDENTS

Valentin-Cosmin Pletea, Àlex Cebrià Xart

## SELECTED PROJECTS

- Ayudas para contratos predoctorales para la formación de doctores 2021. [PRE2021-098532]. Ministerio de Economía Y Competitividad (MINECO). 2022-2026. IP: Alexandra Avgustinova.
- GLEBioassay\_Nano-Monitoring of Cancer Immunotherapy Efficiency: The Lateral Electrophoretic Bioassay platform. [AC21\_2/00045]. Instituto de Salud Carlos III (ISCIII). 2022-2024. IP: Carlos Javier Rodriguez Hernandez.
- El epigenoma malvado de los tumores rabdoídes - definir para tratar. [PID2020-118241RA-I00]. Ministerio de Economía Y Competitividad (MINECO). 2021-2024. IP: Alexandra Avgustinova.
- Ayudas para contratos Ramón y Cajal (RYC) 2019. Beneficiario: Alexandra Avgustinova. [RYC2019-027738-I]. Ministerio de Economía Y Competitividad (MINECO). 2020-2025. IP: Alexandra Avgustinova.

## SELECTED PUBLICATIONS

- Jenkins L, Jungwirth U, Avgustinova A, Iravani M, Mills AP, Haider S et al. Cancer-Associated Fibroblasts Suppress CD8[+]T-cell Infiltration and Confer Resistance to Immune-Checkpoint Blockade. *Cancer Res.* 2022;82(16):2904-2917. IF:13,312. [Q1].
- Gonçalves E, Garcia M, Rodriguez CJ, Gomez S, Ecker RC, Suñol M et al. AC-265347 Inhibits Neuroblastoma Tumor Growth by Induction of Differentiation without Causing Hypocalcemia. *Int J Mol Sci.* 2022;23(8)IF:6,208. [Q1].

# 7.1 MEDICINE AND PEDIATRIC SCIENCE AREA

## 7.1.4 Precision medicine in rare diseases

Metabolic and mitochondrial medicine  
Neurogenetics and molecular medicine  
Human molecular genetics I  
Human molecular genetics II  
Genomics for the diagnosis of rare diseases  
Diagnostic and therapeutic imaging



## Metabolic and mitochondrial medicine

- 6** COMMERCIAL CLINICAL TRIALS
- 0** ACADEMIC CLINICAL TRIALS
- 20** PROJECTS
- 17** PUBLICATIONS
- 88.085** IMPACT FACTOR
- 59%** PUBLICATIONS IN 1RTS QUARTIL
- 12** PUBLICATIONS IN 1RTS DECIL
- 41%** LEADERSHIP

### Rafael Artuch Iriberry, MD, PhD

During the year 2022, we have maintained a good scientific activity, both at the level of publications and scientific projects funded under a competitive way. We are also developing a growing activity in observational studies and clinical trials. Our targeted metabolomics laboratory has established itself as a reference center for hereditary metabolic diseases, both nationally and internationally, reaching 20,000 samples analyzed this year for diagnostic and treatment follow-up purposes. We are also developing a growing activity in metabolomic phenotyping of cell and animal models for different basic research groups. From the point of view of innovation, we have published the validation of a biosensor for ammonium monitoring in Point-of-Care format.

Finally, it should be noted that the PI of the group has been appointed director of the executive committee of the ERNDIM laboratory quality control program ([www.ERNDIM.org](http://www.ERNDIM.org)), a program that brings together more than 400 laboratories from around the world that work in the field of hereditary metabolic diseases.

## MEMBERS

### STAFF RESEARCHER

Roser Urreizti Frexedas, Jaume Campistol Plana, Mercedes Serrano Gimaré, Camila García Volpe, Rosa Gassió Subirachs, Aida Ormazabal Herrero, Cristina Sierra March

### POSTDOC ASSOCIATED RESEARCHERS

Irene Pazos Capell, Mercedes Casado Rio, Raquel Montero Sánchez

### PHD STUDENTS

Mónica Centeno Pla

### ASSISTANT RESEARCHER

Helena Rodriguez González, Eric Kramer Rosado, Florencia Epifani, Gregorio Alexander Nolasco Tovar

### TECHNICIAN

Lidia Celina Sánchez Silvestre, Adriana Balen Valeiro, Yahel Calzadilla Borras, Aroa Fernández de Miguel, Juan Moreno García, Tania Pardal Chofle

## SELECTED PROJECTS

- Stroke-like episodes en CDG: propuesta multicéntrica internacional para descubrir el verdadero fenotipo, mejorar el diagnóstico y tratamiento y entender los mecanismos subyacentes. [PI21/00068]. Fundació Privada per a la Recerca i la Docència Sant Joan de Déu - FSJD; Instituto de Salud Carlos III (ISCIII). 2022-2024. IP: Mercedes Serrano Gimaré.
- PHENYLKETONURIA: from Childhood to Adults through Brain Functional Connectomics, Cardiovascular Changes, Metabolomic and Microbiota Characteristics. [18/C/2020]. Fundació La Marató de TV3. 2021-2024. IP: Aida Ormazabal Herrero.
- Secuenciación de exoma para el diagnóstico de pacientes con enfermedades mitocondriales: investigación de aspectos fisiopatológicos y terapéuticos de las deficiencias de coenzima Q10. [PI20/00340]. Instituto de Salud Carlos III (ISCIII). 2021-2023. IP: Rafael Artuch Iriberry.
- Epilepsy and Mood Regulation Disorders: a Prospective and Longitudinal Study in Children With Newly Diagnosed Epilepsy. EPILETRE. [EPILETRE]. Fundació Bancària "La Caixa"; Fundació La Caixa. 2016-2022. IP: Jaume Campistol Plana, Alexandros Arzimanoglou.

## SELECTED CLINICAL TRIALS & OBSERVATIONAL STUDIES

- Un estudio de fase 2, aleatorizado, abierto, de 12 semanas de duración, para evaluar la farmacodinámica, seguridad, tolerabilidad y farmacocinética de múltiples dosis de GLM101 administradas por vía intravenosa a participantes adultos con PMM2-CDG. A Phase 2, Randomized, Open-Label, 12-Week Study to Assess the Pharmacodynamics, Safety, Tolerability, and Pharmacokinetics of Multiple Doses of GLM101 Administered Intravenously to Adult Participants with PMM2-CDG. (GLM101-002). And Glycomine, Inc; Diagnóstico para la imagen (DXI); Farmacia; García; Hospital de dia HSJD; Laboratori HSJD; Precision for medicine (HU) KFT; Servicio de Cardiología; Servicio Neurofisiología; Servicio Oftalmología; Unitat de Recerca FSJD. 2022. IP: Mercedes Serrano Gimaré.
- Estudio abierto, multicéntrico para evaluar la seguridad, tolerabilidad, farmacocinética y farmacodinamia de R07248824 en participantes con Síndrome de Angelman. An open-label, multicenter study to investigate the safety, tolerability, pharmacokinetics and pharmacodynamics of R07248824 in participants with angelman syndrome. (BP41674). Ortega; Roche Farma, S.A. 2020. IP: Mercedes Serrano Gimaré.
- Estudio de fase IV, abierto y de una sola cohorte, de los resultados neurocognitivos (NC) a largo plazo en niños de 4 a 5 años con fenilcetonuria tratados con dihidrocloruro de saproterina (Kuvan®) durante 7 años. (EMR 700773-002). Merck KGaA; Parexel International S.L. 2014. IP: Jaume Campistol Plana.

## SELECTED PUBLICATIONS

- Jou C, Nascimento A, Codina A, Montoya J, López E, Emperador S et al. Pathological Features in Paediatric Patients with TK2 Deficiency. Int J Mol Sci. 2022;23(19):11002. IF:6,208. (Q1).
- Sánchez O, Yubero D, Leal F, Couce ML, González Gutiérrez L, López E et al. The clinical and biochemical hallmarks generally associated with

GLUT1DS may be caused by defects in genes other than SLC2A1. Clin Genet. 2022;102(1):40-55. IF:4,296. (Q2).

- Calvo A, Rebollo B, Ormazabal A, Artuch R, Rosell X, Alonso J et al. Biomedical point-of-care microanalyzer for potentiometric determination of ammonium ion in plasma and whole blood. Anal Chim Acta. 2022;1205:339782-339782. IF:6,911. (Q1).
- Paredes AJ, Oliva C, Montero R, Alcaide P, Ruijter GJG, García J et al. Technical Aspects of Coenzyme Q(10) Analysis: Validation of a New HPLC-ED Method. Antioxidants (Basel). 2022;11(3)IF:7,675. (Q1).



## Neurogenetics and molecular medicine

- 0** COMMERCIAL CLINICAL TRIALS
- 0** ACADEMIC CLINICAL TRIALS
- 14** PROJECTS
- 22** PUBLICATIONS
- 124** IMPACT FACTOR
- 59%** PUBLICATIONS IN 1RTS QUARTIL
- 5%** PUBLICATIONS IN 1RTS DECIL
- 50%** LEADERSHIP

**Janet Hoenicka, PhD  
& Francesc Palau Martínez, MD, PhD**

In the scientific field, the research group maintains lines of research in neurogenetic diseases, highlighting new findings and publications on (i) mitochondrial dynamics and mitochondrial-lysosome membrane contacts in the cellular pathophysiology of neurological diseases, (ii) the relationship between the phenotype and genotype of neurodevelopmental disorders and movement disorders, (iii) the development of molecular models functional validation of genetic variants through the Diagnostic and Translational Therapy (PDTT) program. Within this program, during 2022 we have carried out the exceptional treatment of a child affected by Menkes disease with Elesclomol-Copper, with results that are very encouraging to develop a future clinical trial.

International activities include participation in the Global Parkinson's Genetics Program (GP2), the conferences given by doctors Janet Hoenicka and Francesc Palau at the Mellowes Center on Genomic Sciences and Precision Medicine of the Medical College of Wisconsin, at the 1st Congress of Clinical Genetics of Honduras and at the EuroDysmorpho

- 32nd European Meeting on Dysmorphology, and scientific editorial management as editor-in-chief of the Orphanet Journal of Rare Diseases.

Our group also participates in the IMPaCT project in its pillars on genomics and data science, and in research networks such as CIBERER and clinical networks such as the ERN Ithaca on Intellectual Disability and other Neurodevelopmental Disorders.

### MEMBERS

#### STAFF RESEARCHER

Joan Maynou Fernández, Mónica Roldan Molina, Guerau Fernandez Isern

#### POSTDOC ASSOCIATED RESEARCHERS

Mar Borregán Prats, Esther Cuatrecasas Capdevila, Cristina Hernando Davalillo, Laura Martí Sanchez, Jordi Pijuan Marquilles, Lara Cantarero Abad, Loreto Martorell Sampol

#### PHD STUDENTS

Berta Estévez Arias

#### ASSISTANT RESEARCHER

Marcos Frías Nestares, Natalia Caelles Gramunt, Natalia Lourdes Serrano, Claire Melody Nguyen, Gemma Tort Vázquez, Yaiza Díaz Osorio, Adrián Alcalá San Martín, Jodie Kane, Alba Pascual Rodríguez, Mercè Bolasell Gargas, Antonio F Martínez Monseny

#### TECHNICIAN

Daniel Castillo Mestres, Jordi Genovès Escarré

### SELECTED PROJECTS

- Genes y fenotipos de síndromes parkinsonianos: estudio clínico y experimental. (PI19/00126). Instituto de Salud Carlos III (ISCIII). 2020-2023. IP: Janet Hoenicka.
- Dinámica de la membrana mitocondrial y sus sitios de contacto entre orgánulos en la fisiopatología celular y del nervio de la enfermedad neurogenética, PID2020-114655RB-100 (AEI, MICIN) 2021-2024. IP: Francesc Palau
- Ayudas Juan de la Cierva-Formación. Beneficiario: Claire Nguyen. (FJC2021-047447-I). Ministerio de Ciencia e Innovación. 2022-2024. IP: Janet Hoenicka.
- The Copper (less) program: Treating and Futures Perspectives for Menkes. Menkes International Association. 2022-2023. IP:

Francesc Palau, Janet Hoenicka.

• Infraestructura de Medicina de Precisión asociada a la Ciencia y Tecnología (IMPaCT). Ciencia de datos. (IMP/00019). Instituto de Salud Carlos III (ISCIII). 2021-2023. IP: Joan Maynou Fernández.

• El cerebro clínico del síndrome Coffin-Siris: Estudio integral de un trastorno del neurodesarrollo. Hospital Sant Joan de Déu - Esplugues HSJD. 2020-2023. IP: Francesc Palau.

### SELECTED PUBLICATIONS

- Fernandez G, Yubero D, Palau F, Armstrong J. Molecular Modelling Hurdle in the Next-Generation Sequencing Era. *Int J Mol Sci.* 2022;23(13):7176. IF:6,208. (Q1)
- Vela-Desojo L, Urso D, Osuna-López M, Hoenicka J. Non-Motor Symptoms in PLA2G6-Associated Dystonia-Parkinsonism: A Case Report and Literature Review. *J Clin Med.* 2022; 11(6):1590. IF: 4,96 (Q1)
- Pijuan J, Cantarero L, Natera-de Benito D, Altimir A, Altisent-Huguet A, Díaz-Osorio Y, Carrera-García L, Expósito-Escudero J, Ortez C, Nascimento A, Hoenicka J, Palau F. Mitochondrial Dynamics and Mitochondria-Lysosome Contacts in Neurogenetic Diseases. *Front Neurosci.* 2022 Jan 31;16:784880. IF: 5,15 (Q2)
- Muñoz DC, Mollá B, Sáenz JJ, Insuasty E, de la Iglesia M, Pook MA et al. Frataxin Deficit Leads to Reduced Dynamics of Growth Cones in Dorsal Root Ganglia Neurons of Friedreich's Ataxia YG8sR Model: A Multilinear Algebra Approach. *Front Mol Neurosci.* 2022;15:912780-912780. IF:6,261. (Q1).

## Human molecular genetics I

- 0** COMMERCIAL CLINICAL TRIALS
- 0** ACADEMIC CLINICAL TRIALS
- s/d** PROJECTS
- 18** PUBLICATIONS
- 177.356** IMPACT FACTOR
- 61%** PUBLICATIONS IN 1RTS QUARTIL
- 39%** PUBLICATIONS IN 1RTS DECIL
- 44%** LEADERSHIP

**Daniel Raúl Grinberg Vaisman, PhD**

In 2022 we have undertaken molecular pathology studies of Schaaf-Yang syndrome (SYS) and TRAF7 syndrome in patients' fibroblasts. We discovered that the truncated MAGEL2 proteins of SYS accumulate in the nucleus, which constitutes a useful cellular biomarker. To better understand the neurological phenotype of SYS we have derived iPSCs from patients' fibroblasts and are currently differentiating neurons, astrocytes and brain organoids, which we will characterize by -omics approaches. To advance into therapeutic avenues, we are assaying nutraceutical compounds on TRAF7 patients' fibroblasts and assaying their effects with previously identified RNA biomarkers.

Taking over on exome results of a family segregating a novel skeletal dysplasia, we have undertaken functional assays of promising variants in TBX18 and SEMA4D. We have also explored the contribution of Osteogenesis Imperfecta (OI genes) to the genetic architecture of postmenopausal osteoporosis.

In our research on genetics of stroke recovery, we have initiated in vitro analyses involving a blood brain barrier cell model to assay the rare variants identified in a novel gene with protective effects.

We have studied genetic and epigenetic risk factors underlying various complex psychiatric and neurological disorders. On one hand, we used several animal models to study the contribution of specific genes to autism (YWHAZ, mouse) or to multiple psychiatric conditions (RBFOX1, zebrafish). Also, mouse models have been used to identify microRNA alterations involved in addiction to food or to cocaine. We have also studied the shared and differentiating genomic architecture of autism and ADHD. We have studied, at genome-wide scale, genetic risk variants that alter the methylation of multiple genes in psychiatric disorders. Finally, we made progress in our project studying the Williams-Beuren and 7q11.23 microduplication syndromes. We generated iPSC-derived neurons and performed scRNAseq studies that we are now analyzing.

### MEMBERS

#### STAFF RESEARCHER

Susana Balcells Comas, Bru Cormand Rifà

#### POSTDOC ASSOCIATED RESEARCHERS

Raquel Rabionet Janssen, Noelia Fernández Castillo, Roser Corominas

#### TECHNICIAN

Mónica Cozar Morillo

### SELECTED PROJECTS

- Pleiotropía y comorbilidad en el trastorno por déficit de atención con hiperactividad (PID2021-127776OB-I00). Ministerio de Ciencia e Innovación (MICINN). Pls: Bru Cormand Rifa i Noelia Fernandez Castillo. 2022-2025.
- ISEDAP7: Integrative studies of early neurodevelopment alterations in patients with 7q11.23 rearrangements (PID2021-1282150B-I00). Ministerio de Ciencia e Innovación. PI: Roser Corominas Castiñeira. 2022-2025.
- Estudios genéticos y funcionales hacia aproximaciones terapéuticas para varias enfermedades óseas y síndromes de discapacidad intelectual (PID2019-107188RB-C21). Ministerio de Ciencia, Innovación y Universidades. Convocatòria d'ajuts del Programa Estatal d'Investigació, Desenvolupament i Innovació Orientada als reptes de la Societat.

Modalitat Reptes. PI: Susanna Balcells Comas. 2020-2023.

### SELECTED PUBLICATIONS

- Antón E, Dalla E, Orlandi JG, Castro G, Gualda EJ, Young A et al. Deficiency of the ywhaz gene, involved in neurodevelopmental disorders, alters brain activity and behaviour in zebrafish. *Mol Psychiatry*. 2022;27(9):3739-3748. IF:11,000. (Q1).
- Fernandez N, Cabana J, Corominas L, Cormand B. Molecular genetics of cocaine use disorders in humans. *Mol Psychiatry*. 2022;27(1):624-639. IF:11,000. (Q1).
- Martinez N, Ovejero D, Garcia N, Brueque CD, Mellibovsky L, Nogues X et al. Genetic Analysis in a Familial Case With High Bone Mineral Density Suggests Additive Effects at Two Loci. *JBMR Plus*. 2022;6(4)
- Ovejero D, Garcia N, Martinez N, Rabionet R, Balcells S, Grinberg DR et al. Clinical description and genetic analysis of a novel familial skeletal dysplasia characterized by high bone mass and lucent bone lesions. *Bone*. 2022;161:116450-116450. IF:4,100. (Q2).

## Human molecular genetics II

- 0 COMMERCIAL CLINICAL TRIALS
- 0 ACADEMIC CLINICAL TRIALS
- s/d PROJECTS
- 6 PUBLICATIONS
- 35.517 IMPACT FACTOR
- 67% PUBLICATIONS IN 1RTS QUARTIL
- 0% PUBLICATIONS IN 1RTS DECI
- 83% LEADERSHIP

### Gemma Marfany Nadal, PhD

Our group has a main line of research focused on the generation and characterization of animal and cell models to study the function of the inherited retinal dystrophy genes and search for a possible precision therapy.

In order to study the molecular bases of hereditary blindness, we generated different mouse models by genetic editing techniques (CRISPR-Cas9) on two genes that cause retinitis pigmentosa, cone-rod dystrophy, and S-cone syndrome: *CERKL* and *NR2E3*, respectively. These last years, we have performed more in-depth molecular studies to elucidate why mutations in these genes cause hereditary blindness. For *NR2E3*, we are currently performing single cell transcriptomics to characterize the photoreceptor populations of the mutant strains compared to wild-type (collaboration with Dr. Vaquerizas at Imperial College London). Regarding *CERKL*, we have collaborated with other IBUB and IRSJD groups (Dr. Villaroya and Dr. Artuch) to show that its depletion causes alterations in mitochondrial morphology, dynamics, and metabolism, in retinal neurons, photoreceptors, retinal ganglion cells (RGC) as well as in cells of the retinal pigment epithelium (RPE) and hippocampus, opening new perspectives for a

potential treatment by regulation of mitochondrial metabolism. A senior postdoc from our group has carried out short research stays in Naples (collaboration with Drs. Ursini and Trapani) to explore potential gene therapy treatments by subretinal injection of adeno-associated viruses (AAVs). Finally, we are also investigating several genes that may contribute to the formation and stability of the sensory cilium, so they could be candidates for causing ciliopathies, one of the symptoms of which is retinal neurodegeneration, specifically *ATXN3*, *RPGK* and *USP48*.

In the future, we intend to delve into new avenues of research, such as the generation of human retinal organoids from hiPSCs. The year 2022 has been very intense in terms of work and strengthening very fruitful national and international collaborations.

### MEMBERS

#### POSTDOC ASSOCIATED RESEARCHERS

Serena Mirra

### SELECTED PROJECTS

- Explorando las vías de señalización neurosensoriales en enfermedades hereditarias de retina como posible aproximación a la terapia. Ministerio de Ciencia, Innovación y Universidades. Convocatoria d'ajuts del Programa Estatal d'Investigació, Desenvolupament i Innovació Orientada als reptes de la Societat. Modalitat Reptes. PID2019-108578RB-I00. PI: Gemma Marfany Nadal, co-IP: Serena Mirra. 2020-2023.
- Enfermedades Raras. Ministerio de Sanidad y Consumo. Convocatoria d'ajuts destinats a finançar estructures estables d'investigació cooperativa, a l'àrea de biomedicina i ciències de la salut, en el marc del Programa Ingenio 2010, Programa Consolider, acciones CIBER. CB06/07/0041. PI: Gemma Marfany Nadal. 2006-2022.
- Terapia génica para la Retinitis Pigmentosa causada por mutaciones en *CERKL*: prueba preclínica mediante virus adenoasociados (AAVs) en el modelo de ratón CerkIKD/K.
- Financiador: Ministerio de Sanidad y Consumo. Acciones intramulares CIBER. ACCI21-04. PI: Serena Mirra. 2021-2023.

### SELECTED PUBLICATIONS

- García R, Marfany G, Mirra S. *CERKL*, a Retinal Dystrophy Gene, Regulates Mitochondrial Transport and Dynamics in Hippocampal Neurons. *Int. J. Mol. Sci.* 2022;23(19):115931F:5,600. (Q1).
- Sánchez L, Férriz A, Carrillo M, Rabanal L, García FR, Marfany G. The Deubiquitinating Enzyme USP48 Interacts with the Retinal Degeneration-Associated Proteins UNC119a and ARL3. *Int. J. Mol. Sci.* 2022;23(20)F:5,600. (Q1).
- Toulis V, Casaroli R, Camós A, Figueiras M, Sánchez B, Muñoz E et al. Altered retinal structure and function in Spinocerebellar ataxia type 3. *Neurobiol Dis.* 2022;170:105774-105774. IF:6,100. (Q1).

## Genomics for the diagnosis of rare diseases

- 0** COMMERCIAL CLINICAL TRIALS
- 0** ACADEMIC CLINICAL TRIALS
- 11** PROJECTS
- 9** PUBLICATIONS
- 52.749** IMPACT FACTOR
- 56%** PUBLICATIONS IN 1RTS QUARTIL
- 22%** PUBLICATIONS IN 1RTS DECIL
- 56%** LEADERSHIP

**Judith Armstrong Morón, PhD**

### MEMBERS

#### STAFF RESEARCHER

Núria Brandi Tarrrau, Délia Yubero Siles

#### PHD STUDENTS

Ainhoa Pascual Alonso, Clara Xiol Viñas

#### TECHNICIAN

Carlota Ros Bonaventura, Paola Pacheco Fernández

### SELECTED PROJECTS

- Catalan Interhospital Network of Genetic Variants to Improve Genetic Diagnosis in Rare Diseases. (590/C/2020). Fundació La Marató de TV3. 2021-2024. IP: Délia Yubero Siles.
- Estudio de pacientes dentro del Espectro Rett mediante un enfoque multiómico integrativo: delucidar la huella molecular de la vía funcional. (PI20/00389). Instituto de Salud Carlos III (ISCIII). 2021-2023. IP: Judith Silvia Armstrong Morón.
- Convocatòria d'ajuts per a la contractació de personal investigador novell (FI-2020). Beneficiari: Ainhoa Pascual (co-financiado por el FSE). (2020 FI\_B\_00888). Agaur - Agència de Gestió d'Ajuts Universitaris i de Recerca. 2020-2023. IP: Judith Silvia Armstrong Morón.

- FPU\_Clara Xiol. (FPU18/02152). Ministerio Educación, Cultura y Deporte (MECD). 2019-2023. IP: Judith Silvia Armstrong Morón.
- I Edición de los premios AELMHU. (AELMHU). Asociación Española de Laboratorios de Medicamentos Huérfanos y Ultrahuérfanos – AELMHU. 2017-2023. IP: Judith Silvia Armstrong Morón.

### SELECTED PUBLICATIONS

- Fernandez G, Yubero D, Palau F, Armstrong J. Molecular Modelling Hurdle in the Next-Generation Sequencing Era. *Int J Mol Sci.* 2022;23(13):IF:6,208. (Q1).
- Schlüter A, Rodríguez A, Verdura E, Vélez V, Ruiz M, Fourcade S et al. Diagnosis of Genetic White Matter Disorders by Singleton Whole-Exome and Genome Sequencing Using Interactome-Driven Prioritization. *Neurology.* 2022;98(9):912-923. IF:11,800. (Q1).
- Migliorelli C, Medina IF, Bachiller A, Tost A, Alonso JF, López A et al. Cognitive stimulation has potential for brain activation in individuals with Rett syndrome. *J Intellect Disabil Res.* 2022;66(3):213-224. IF:3,646. (Q1).
- Siqueira E, Obiols A, Jorge OC, Oliveira C, Soler M, Ramesh D et al. Analysis of the circRNA and T-UCR populations identifies convergent pathways in mouse and human models of Rett syndrome. *Mol Ther Nucleic Acids.* 2022;27:621-644. IF:10,183. (Q1).

# Diagnostic and therapeutic imaging

- 0** COMMERCIAL CLINICAL TRIALS
- 0** ACADEMIC CLINICAL TRIALS
- 8** PROJECTS
- 16** PUBLICATIONS
- 53.593** IMPACT FACTOR
- 19%** PUBLICATIONS IN 1RTS QUARTIL
- 0%** PUBLICATIONS IN 1RTS DECIL
- 69%** LEADERSHIP

**Josep Munuera del Cerro, MD, PhD**

## MEMBERS

### STAFF RESEARCHER

Ignasi Barber Martínez de la Torre, Marta Gómez Chiari, Emilio Inarejos Clemente, Mónica Rebollo Polo

### ASSISTANT RESEARCHER

Sandra Bernaus Tomé, Martí Engli Rueda, Christian Mata Miquel

## SELECTED PROJECTS

- Sistema de ayuda a la decisión clínica para la evaluación y predicción de enfermedad vascular cerebral pediátrica: Radiómica a partir de RM vascular e Inteligencia Artificial (Vasculómica). (PI20/00296). Instituto de Salud Carlos III (ISCIII). 2021-2023. IP: Jose Luis Munuera del Cerro.
- Plataforma ISCIII de apoyo a la I+D+ en Biomedicina y Ciencias de la Salud. Biomodelos-Impresión 3D. (PT20/00090). Instituto de Salud Carlos III (ISCIII). 2021-2023. IP: Jose Luis Munuera del Cerro.
- Node d'imatge computacional pediàtrica (PEDIATRIC\_ICON). (PR15-019756). Departament de Salut - Generalitat de Catalunya. 2020-2023. IP: Jose Luis Munuera del Cerro.
- EXPERT3D - Transversal programme in medical image

postprocess, training professionals: 3D printing to research - 20494. EIT Health e.V. 2020-2022. IP: Jose Luis Munuera del Cerro.

## SELECTED PUBLICATIONS

- Guitart C, Rodríguez J, Bobillo S, Carrasco JL, Inarejos E, Cambra FJ et al. An algorithm combining procalcitonin and lung ultrasound improves the diagnosis of bacterial pneumonia in critically ill children: The PROLUSP study, a randomized clinical trial. *Pediatr Pulmonol*. 2022;57(3):711-723. IF:4,090. (Q1).
- Inarejos E, Navarro OM, Navallas M, Enrique G, Torner F, Suñol M et al. Multiparametric MRI evaluation of bone sarcomas in children. *Insights Imaging*. 2022;13(1):33. IF:5,036. (Q1).
- Quihui P, Ochoa B, Gonzalez M, Rodriguez G, Mata C. Comparison of Automatic Prostate Zones Segmentation Models in MRI Images Using U-net-like Architectures. *Lect Notes Comput Sc*. 2022;13612:282-296. IF:0,402. (Q4).
- Raymond C, Fuentes P, García MÁ, Ramiro N, Soler J, Torres ML et al. Regularized Functional Connectivity in Schizophrenia. *Front Hum Neurosci*. 2022;16:878028. IF:3,473. (Q2).

# 7.1 MEDICINE AND PEDIATRIC SCIENCE AREA

## 7.1.5 Applied clinical research

Influence of the environment on child and adolescent welfare

Hospital dentistry and periodontal medicine

Paediatric ophthalmology

Research in nursing

Orthopaedic surgery and traumatology



## Influence of the environment on child and adolescent welfare

- 0** COMMERCIAL CLINICAL TRIALS
- 0** ACADEMIC CLINICAL TRIALS
- 1** PROJECTS
- 10** PUBLICATIONS
- 38.628** IMPACT FACTOR
- 10%** PUBLICATIONS IN 1RTS QUARTIL
- 0%** PUBLICATIONS IN 1RTS DECILO
- 70%** LEADERSHIP

### Carles Luaces Cubells, MD, PhD

Collaboration was begun on three new research projects in 2022.

We have also continued to emphasise the promotion of good treatment of children and the prevention of mistreatment, together with the study of the consequences of sexual violence in childhood. Likewise, we have collaborated on various courses and sessions of the Spanish Society for Paediatric Emergencies and on the implantation of the Barnahus project.

A group has also been set up for collaboration with the Patient Experience Area to detect suitable actions for the improvement of the care provided in the Emergency Service.

### MEMBERS

#### STAFF RESEARCHER

Gemma Claret Teruel, Ana Isabel Curcoy Barcenilla, Lidia Martínez Sánchez, Victoria Trenchs Sainz de la Maza

#### POSTDOC ASSOCIATED RESEARCHERS

Andrea Aldemira Liz, Vanessa Arias Constantí, Eva Gargallo Burriel, Cristina Parra Cotanda

#### PHD STUDENTS

David Muñoz Santanach, Sergi Navarro Vilarrubi, Marta Simó Nebot

### SELECTED PROJECTS

- ACCELERATE\_AI Accelerator – A Smart Hospital Care Pathway Engine. (101016902). European Commission. 2021-2024. IP: Arnaud Valls Esteve, Sergi Navarro Vilarrubi, Joan Sánchez de Toledo.
- Evaluación del uso de FebriDx en el Entorno Pediátrico y en el Manejo de Niños con Neumonía Adquirida en la Comunidad. (FebriDx). 2022. IP: Carles Luaces Cubells.
- STEPS\_Step by step standing up a Barnahus in Catalonia: A simulation-based program to avoid secondary victimization in the assessment and treatment of child sexual abuse. European Commission. 2020-2022. IP: Marta Simó Nebot.
- RSV in Hospital Emergency Departments in Iberia [RHEDI] Study. (RHEDI). 2022. IP: Carles Luaces Cubells.

### SELECTED PUBLICATIONS

- Asenjo S, López A, Muñoz D, Trenchs V, Luaces C, Parra C. Impact of a Disaster Drill on Waiting Times in a Pediatric Emergency Department. Disaster Med Public Health Prep. 2022;16(5):1814-1816. IF:5,556. (Q1).
- Burillo G, Ibrahim D, Galicia M, Supervía A, Martínez L, Ortega J et al. Clinical manifestations and serious adverse effects after cannabis use: role of age according to sex and coingestion of alcohol. Emergencias. 2022;34(4):275-281. IF:5,345. (Q1).
- Burillo G, Ibrahim D, Martínez L, Galicia M, Supervía A, Puiguriguer J et al. Differences in clinical signs and severity of intoxication due to street drugs in adolescents and young adults treated in emergency departments. Emergencias. 2022;34(5):352-360. IF:5,345. (Q1).
- Hernández S, Trenchs V, Soler A, Marina B, Ciutad M, Luaces C. Outpatient and oral management is suitable for infants 60-90 days old with urinary tract infections at low risk of bacteremia. Eur J Pediatr. 2022;181(2):671-677. IF:3,860. (Q1).

## Hospital dentistry and periodontal medicine

-  **0** COMMERCIAL CLINICAL TRIALS
-  **0** ACADEMIC CLINICAL TRIALS
-  **0** PROJECTS
-  **3** PUBLICATIONS
-  **6.5** IMPACT FACTOR
-  **33%** PUBLICATIONS IN 1RTS QUARTIL
-  **0%** PUBLICATIONS IN 1RTS DECIL
-  **100%** LEADERSHIP

Alejandro Rivera Baró, MD, PhD

### MEMBERS

#### STAFF RESEARCHER

Mireia Aznar Gómez, Lluis Brunet Llobet, Jaume Miranda Rius, Jordi Moncunill Mira, Maria Lluïsa Tobella Camps

#### SELECTED PUBLICATIONS

- Rabassa J, Brunet LL, Marcote P, Balsells S, Correa MG, Miranda J. Prevalence of, and risk factors for, dental sequelae in adolescents who underwent cancer therapy during childhood. *Oral Dis.* 2022;IF:3,800. (Q1).
- Brunet LL, Auria B, Gonzalez Y, Cahuana P, Mashala El, Miranda J. The use of silver diamine fluoride in a children's hospital: Critical analysis and action protocol. *Clin Exp Dent Res.* 2022;8(5):1175-1184. FI: 1,8.
- Lapiitzkaya A, Cahuana P, Brunet LL, Miranda J. Oral irritation fibroma associated with the pathological migration of a primary tooth. *BMJ Case Rep.* 2022;15(3):e249369. IF: 0,9.



## Paediatric ophthalmology

- 1** COMMERCIAL CLINICAL TRIALS
- 1** ACADEMIC CLINICAL TRIALS
- 1** PROJECTS
- 9** PUBLICATIONS
- 82.429** IMPACT FACTOR
- 78%** PUBLICATIONS IN 1RTS QUARTIL
- 44%** PUBLICATIONS IN 1RTS DECIL
- 11%** LEADERSHIP

**Joan Prat Bartomeu, MD, PhD**

### MEMBERS

#### STAFF RESEARCHER

Elisa Carreras Bertran, Ester Casas Gimeno, Jaume Català Mora, Jesús Díaz Cascajosa, Ana Isabel Díaz Cortés, Carlos Fresno Cañada, Carmen García de Vicuña Muñoz, Enrique Jimenez Gañan, Ana Llorca Cardeñosa, Marta Morales Ballús, Núria Planas Domènech, Mª Josep Planas Tapias, Enric Puigventós, Gemma Romeu Cerrillo, Alicia Serra Castanera, Dowon, Simone Suh, Begonya Yeste Mayoral

### SELECTED PROJECTS

- Desenvolupament d'un equip de realitat virtual per l'avaluació i seguiment de la visió funcional en pacients amb distrofies hereditàries de retina abans i després del tractament amb teràpies intraoculars avançades; (2021 LLAV 00019) Agència de Gestió d'Ajuts Universitaris i de Recerca;. IP: Jaume Català

### SELECTED CLINICAL TRIALS & OBSERVATIONAL STUDIES

- A Phase 3, multicenter, randomized, evaluator-blinded clinical trial to assess the safety and efficacy of Clobetasol propionate ophthalmic nanoemulsion, 0.05% compared

to Prednisolone acetate, 1% in the treatment of inflammation after cataract surgery in pediatric population 0 to 3 years of age [CLOSE-3]. [CLOBOF3-17IA03]. del Pino; Farmacia; Laboratorios Salvat, S.A.; Unitat de Recerca FSJD. 2022. IP: Marta Morales Ballús.

- A randomized, 2-arm, open-label, phase 3 study to evaluate the efficacy, safety, and tolerability of intravitreal afibercept versus laser photocoagulation in patients with retinopathy of prematurity (ROP). (BAY 86-5321/20090). Bayer AG. 2019. IP: Alicia Serra Castanera.
- Ensayo clínico fase I, unicéntrico, abierto y de escalado de dosis, para evaluar la seguridad y la actividad del adenovirus oncolítico VCN-01 en pacientes con retinoblastoma refractario. (FSJD-RTB-2015). Fundació Privada per a la Recerca i la Docència Sant Joan de Déu - FSJD; VCN Biosciences S.L. 2017. IP: Jaume Català Mora.
- Estudio postautorización, multicéntrico, multinacional, longitudinal y observacional para el registro de la seguridad en pacientes tratados con voretigén neparvovec. (CLTW888A12401/ NOV-LUX-2019-01). Novartis Farmacéutica, S.A. 2020. IP: Jaume Català Mora.

### SELECTED PUBLICATIONS

- Cuadrado M, Liu J, Paco S, Aschero MR, Burgeño V, Sirab N et al. Identification of immunosuppressive factors in retinoblastoma cell secretomes and aqueous humor from patients. *J Pathol.* 2022;257(3):327-339. IF:9,883. (Q1).
- Tomar AS, Finger PT, Gallie B, Kivelä TT, Mallipatna A, Zhang C et al. High-risk Pathologic Features Based on Presenting Findings in Advanced Intraocular Retinoblastoma A Multicenter, International Data-Sharing American Joint Committee on Cancer Study. *Ophthalmology.* 2022;129(8):923-932. IF:14,277. (Q1).
- Tomar AS, Finger PT, Gallie B, Kivelä TT, Mallipatna A, Zhang C et al. Metastatic Death Based on Presenting Features and Treatment for Advanced Intraocular Retinoblastoma A Multicenter Registry-Based Study. *Ophthalmology.* 2022;129(8):933-945. IF:14,277. (Q1).

- Wissinger B, Baumann B, Buena E, Ravesz Z, Cideciyan AV, Stingl K et al. The landscape of submicroscopic structural variants at the OPN1LW/OPN1MW gene cluster on Xq28 underlying blue cone monochromacy. *Proc. Natl. Acad. Sci. U. S. A.* 2022;119(27):211,553,811. IF:12,779. (Q1).

## Research in nursing

- 0** COMMERCIAL CLINICAL TRIALS
- 0** ACADEMIC CLINICAL TRIALS
- 3** PROJECTS
- 4** PUBLICATIONS
- 13.786** IMPACT FACTOR
- 50%** PUBLICATIONS IN 1RTS QUARTIL
- 0%** PUBLICATIONS IN 1RTS DECIL
- 75%** LEADERSHIP

From the responsibility of the Group Clínic Associats: Nursing Research, aspects of revaluation of the directors of support to research are promoted in relation to:

- The activity of continuous training in the search to strengthen the nursing discipline.
- The impulses for the creation of research teams from each of the areas of patient/family care for the improvement of the quality of the services that lend themselves to the nursing discipline.
- Development of a fourth line of research for the contribution to the improvement of the health level of the patient/family.

Initiatives promoted by the research support nurse:

- Creation of the course: "Research Methodology. Evidence-Based Clinical Practice". Continuous training. Accreditation of the Consell Català de la Formació Continuada de les Professions Sanitàries (CCFCPS). Hours awarded: 12.00. Credits: 1.5. Quality Factor: 1.10. HSJD Training Classroom. Address: Montserrat Gutiérrez Juárez.
- Research Commission. The expert nurses of the commission for the detection and accompaniment of the research projects of the nurses of our institution are established by new directors. They are part of the group of teachers of the course:

"Research Methodology. Evidence-Based Clinical Practice."

- Fourth line of research Patient/family experience. She is self-assured and is presented to the HSJD Nursing Department and to the Cap de l'Àrea d'Experiència del Paciente.
- #SJDScientific Friday. Consolidation of the project and restructuring of work dynamics to obtain an annual agenda in advance. Coordination: Jordi Mitjà Costa.
- ECHO Nursing at the Working Group Meeting, May 10-11, 2022, Children's Health Ireland. Participation (in person) with a member.
- "CAT Nurse Research". Participation of the Grup d'infermeres referents in creative research to establish synergies between emerging groups of nursing research and consolidated groups.
- Final Residency Work (TFR): Accompaniment of the research nurse in the memory of the TFR to present to the CR of the IRSJD and to the CEIm.

### MEMBERS

#### STAFF RESEARCHER

Maribel Ferrer Orona, Concepción Agustín Mata, Silvia Ciprés Roig, Encarna Gómez Gamboa, Jordi Mitjà, Gemma Pérez Acevedo

#### POSTDOC ASSOCIATED RESEARCHERS

Mª Dolors Navarro Rubio

#### PHD STUDENTS

Marta Fajardo Samper

#### ASSISTANT RESEARCHER

Pau Miquel Diego, Ikram Allaoui, Montserrat Gutierrez Juarez, Ares Sentenach Carbó

#### COLLABORATOR RESEARCHER

Carmen Jerez Molina

### SELECTED PUBLICATIONS

- Gómez E, Olga P, San M, Saz MA, Negre A, Puig M. The Perceptions of Children and Adolescents with Cancer Regarding Nurses' Communication Behaviors during Needle Procedures. *Int J Environ Res Public Health*. 2022;19(15)IF:4,614. (Q1).
- Pérez G, Bosch A, Torra JE. Larval Therapy for Treatment of Chronic Wounds Colonized by Multi-resistant Pathogens in a Pediatric Patient A Case Study. *J Wound Ostomy Cont Nurs*. 2022;49(4):373-378. IF:1,970. (Q3).
- Gutierrez L, Garcia S, Leon A, Gomez AJ, Perez B, Rodriguez S et al. Competency Gradients in Advanced Practice Nurses, Specialist Nurses, and Registered Nurses: A Multicentre Cross-Sectional Study. *Int J Environ Res Public Health*. 2022;19(14)IF:4,614. (Q1).
- Gómez E, Rodrigo O, Saz MA, Gómez A, Negre A, Llobet MP. Spiritual pain as part of the hospitalization experience of children and adolescents with acute lymphoblastic leukemia: A phenomenological study. *Eur J Oncol Nurs*. 2022;58:102141-102141. IF:2,588. (Q2).

### SELECTED PROJECTS

- Subvencions per a la intensificació de professionals d'infermeria, llevaduria, fisioteràpia i nutrició, òptica i optometria, podologia i protètica dental. Beneficiari: Gemma Pérez Acevedo. (SLT017/20/000194). Departament de Salut - Generalitat de Catalunya. 2021-2022. IP: Gemma Pérez Acevedo.

# Orthopaedic surgery and traumatology



0

COMMERCIAL CLINICAL TRIALS



0

ACADEMIC CLINICAL TRIALS



5

PROJECTS



2

PUBLICATIONS



5.88

IMPACT FACTOR



50%

PUBLICATIONS IN 1RTS QUARTIL



0%

PUBLICATIONS IN 1RTS DECIL



100%

LEADERSHIP

**César García Fontecha, MD, PhD**

## MEMBERS

### STAFF RESEARCHER

Antoni Compte Verdaguer, Laura Corominas Francés, Lydia De Sena De Cabo, Dimosthenis Kiimetoglou, David Moreno Romo, Laura Montserrat Pérez López, Melisa Stitzman Wengrowicz, Ferran Torner Rubies, Alejandro Peiró García, Maria Pilar Rovira Martí

### PHD STUDENTS

Rocio Garcia Garcia

### ASSISTANT RESEARCHER

Gerard Marturià Navarro

## SELECTED PROJECTS

- Estudio longitudinal para evaluar las mediciones de crecimiento y los biomarcadores digitales en participantes pediátricos con acondroplasia. A study to evaluate longitudinal growth velocity and digital biomarkers in pediatric participants with achondroplasia. [OBS16647]. Sanofi-Aventis S.A. 2022. IP: César Galo García Fontecha.
- Computational model of guided growth in immature skeleton for custom-made correction of deformities. [PI20/00293]. Instituto de Salud Carlos III (ISCIII). 2021-2023. IP: César Galo García

Fontecha.

- Randomized clinical trial to compare day-time and night-time brace. [PI20/00962]. Instituto de Salud Carlos III (ISCIII). 2021-2023. IP: Alejandro Peiró García.
- Exploring the relationship between Legg-Calvé-Perthes disease (LCPD) and Attention Deficit Hyperactivity Disorder (ADHD). European Paediatric Orthopaedic Society (EPOS). 2022-2023. IP: César Galo García Fontecha.

## SELECTED PUBLICATIONS

- Cabrera M, Gutierrez D, García CG, Garcia R, Ortega A, Pérez LM. Poland syndrome: a new classification system based on a retrospective analysis of 74 cases. *J Hand Surg Eur Vol.* 2022;47(10):1027-1031. IF:0,844. (Q3).
- Torner F, Nuñez JH, Inarejos E, Garraus M, Suñol M, Deyà A et al. Total calcaneal allograft reconstruction of an Ewing's sarcoma in a child: Outcome and review of the literature. *Cancer Rep (Hoboken)*. 2022;5(9):e1626.
- Inarejos E, Navarro OM, Navallas M, Ladera E, Torner F, Suñol M et al. Multiparametric MRI evaluation of bone sarcomas in children. *Insights Imaging*. 2022;13(1):33. IF:5,036. (Q1).

## 7.2 MENTAL HEALTH AREA

### 7.2.1 Mental health and society

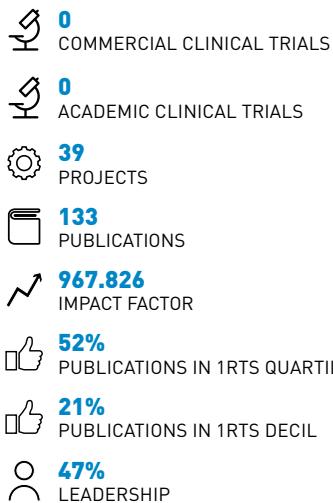
Epidemiology of mental disorders and aging

Health technology assessment in primary care and mental health  
(PRISMA)

Psychological research group on fibromyalgia and chronic pain  
(AGORA group)



# Epidemiology of mental disorders and aging



**Josep Maria Haro Abad, MD, PhD & Ai Koyanagi, MD, PhD**

## MEMBERS

### STAFF RESEARCHER

Louis Etienne Charles Jacob , Mireia Félez Nóbrega, Paula Cristobal Narvaez, Sara Siddi , Stefanos Tyrovolas

### POSTDOC ASSOCIATED RESEARCHERS

Rodrigo Antunes Lima, Joan Domènec Abella, Beatriz Olaya Guzmán

### PHD STUDENTS

Elena Condominas Casanovas, Carlota de Miquel Bleier, Anna Monistrol Mula

### ASSISTANT RESEARCHER

Mireia Esplugues Tormo, Clara Szymanski Cortés, Helena Sainz Elias, Andrea Estefanía Miranda Mendizabal, Leticia Gonzalez Spinoglio, Emilio Tomas Lavilla, Fajar Matloob Butt, Rachid El Hafi Elmokhtari , Marta Franch Roca, Lydia Hanaa Faris , Aina Gabarrell Pascuet, Desireé Gutiérrez Marin, Iago Giné Vázquez, Ivet Bayes Marin, Blanca Mellor Marsa, Maria Victoria Moneta , Ziggi Santini , Elisabeth CW Vorstenbosch

### MANAGEMENT

Silvena Pashova Velikova, Iñigo Chivite Araiz, Judit Lopez Luque, Iris Mauricio Arribas, Carla Obradors Tarragó

## SELECTED PROJECTS

- MIO: Migration Impact On health. (SR21-00779). Fundació Bancària " La Caixa". 2021-2023. IP: Josep Maria Haro Abad.
- EDIT-B\_Edit-B: The RNA blood test for bipolar disorder. (220628). EIT Health e.V. 2022-2024. IP: Josep Maria Haro Abad.
- Xarxa de R+D+I en Tecnologies de la Salut (XARTEC). (2018 XARDI 00016). Agaur - Agència de Gestió d'Ajuts Universitaris i de Recerca. 2018-2022. IP: Josep Maria Haro Abad.
- CRETSAM\_Centre de Recerca i Transferència en Salut Mental. (IU16-016625). Agaur - Agència de Gestió d'Ajuts Universitaris i de Recerca; Departament de Salut - Generalitat de Catalunya. 2020-2023. IP: Josep Maria Haro Abad.

## SELECTED PUBLICATIONS

- Kostev K, Abeler S, Koyanagi A, Haro JM, Smith L, Jacob L. Sales of Iodine-Containing Drugs in Europe Following the Beginning of the War Between Russia and Ukraine. *JAMA Netw open*. 2022 Oct;5(10):e2240032. IF:13,353(Q1)
- Lara E, Miret M, Olaya B, Caballero FF, Morillo D, Moneta MV, et al. Cohort Profile: The Spanish Longitudinal Study on Ageing and Health [Edad Con Salud]. *Int J Epidemiol*. 2022 Aug;51(4):e189–99. IF:9,685(Q1)
- Narita Z, Banawa R, Zhou S, DeVylder J, Koyanagi A, Oh H. Loneliness and psychotic experiences among US university students: Findings from the Healthy Minds Study 2020. *Psychiatry Res*. 2022 Feb;308:114362. IF:11,225(Q1)
- Tyrovolas S, Tyrovolas D, Giné-Vázquez I, Koyanagi A, Bernabe-Ortiz A, Rodriguez-Artalejo F, et al. Global, regional, and national burden of aortic aneurysm, 1990-2017: a systematic analysis of the Global Burden of Disease Study 2017. *Eur J Prev Cardiol*. 2022 May;29(8):1220–32. IF:8,526(Q1)

# Health technology assessment in primary care and mental health (PRISMA)

	<b>0</b>	COMMERCIAL CLINICAL TRIALS
	<b>1</b>	ACADEMIC CLINICAL TRIALS
	<b>23</b>	PROJECTS
	<b>20</b>	PUBLICATIONS
	<b>104.752</b>	IMPACT FACTOR
	<b>50%</b>	PUBLICATIONS IN 1RTS QUARTIL
	<b>20%</b>	PUBLICATIONS IN 1RTS DECIIL
	<b>60%</b>	LEADERSHIP

**Maria Rubio Valera, PhD & Antoni Serrano Blanco, MD, PhD**

Throughout this year 2022, PRISMA has carried out the IMA intervention in 12 Primary Care Centers in Catalonia and protocols of the intervention have been published, similarly, PRISMA has developed the cost of pre-implementation analysis of the Equity intervention (two ERC projects) in Colombia, Chile and Ecuador. Additionally, PRISMA has received funding for assessing the prevalence and associated factors of benzodiazepines users from Instituto de Salud Carlos III (ISCIII) as well as for assessing an intervention to improve mental health resilience in adolescents with key stakeholders in mental health care. Several members of PRISMA has been included in the RICAPPS network from ISCIII. Alexandra Dima has received a Miguel Servet contract from ISCIII. PRISMA research group has reinforced their collaboration with the public institutions as Diputació de Barcelona or the Pacte Nacional de Salut Mental from Generalitat de Catalunya, positioning as a key actor in mental health policy planning. PRISMA research members have published a book about Mental Health

Primary Support Programme which details the network coordination between Primary Care and Mental Health professionals. Several PRISMA members received a prize from the Barcelna Medical College for the article non-initiation of prescribed medication from a Spanish health professionals' perspective: A qualitative exploration based on Grounded Theory considered the best articlue published in 2022 in the region of Baix Llobregat. PRISMA, with the support of CIBER in Epidemiology and Public Health from ISCIII has done great advances in the development of E-CONOS, a database which records the unit cost of health services of all Spanish regions.

## MEMBERS

### STAFF RESEARCHER

Claudia Palma Vásquez, Alexandra Lelia Dima , Víctor Chavarría Romero, Luisa Baladon Higuera, Vanessa Barneda , María Iglesias Gonzalez, Cristina López Ortiz, Enric Vicens Pons

### POSTDOC ASSOCIATED RESEARCHERS

Mercè Soler Font, María Montserrat Gil Girbau, Carlos Peña Salazar, Ignacio Aznar Lou

### PHD STUDENTS

Alba Sanchez Viñas, Cristina Carbonell Duacastella

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Marcos Antonio Catalan Vega

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Salvatore Fabrizio Contaldo, María del Carmen Gallardo González, Silvia Ramírez Perdomo, Montserrat Iracheta Todó, Ana Fernández Sánchez, María Antonia March Pujol, Juan Manuel Mendive Arbeloa, María Teresa Peñarrubia María, Ramón Sabes Figuera, Luís Salvador Carulla

## SELECTED PROJECTS

- Red de Investigación en Cronicidad, Atención Primaria y Promoción de la Salud (RICAPPS). (RD21/0016/0018). Instituto de Salud Carlos III (ISCIII). 2022-2024. IP: María Montserrat Gil Girbau.
- IMA-cRCT\_Effectiveness and cost-effectiveness of the Initial Medication Adherence intervention: cluster randomized controlled trial and economic model. (948973).

European Commission. 2021-2025. IP: Maria Rubio Valera.

- CIBER\_Epidemiology and Public Health. (CB16/02/00322). Instituto de Salud Carlos III (ISCIII). 2017-2030. IP: Antoni Serrano Blanco.
- Necessitats actuals i reptes de la Taula del Pacte Nacional de Salut Mental: Evaluació qualitativa. Fundació Universitària Balmes de Vic. 2022-2023. IP: Antoni Serrano Blanco.

## SELECTED PUBLICATIONS

- Carbonell-Duacastella C, Rubio-Valera M, Marqués-Ercilla S, Peñarrubia-María MT, Gil-Girbau M, García-Cardenas V, et al. Pediatric Medication Noninitiation in Spain. *Pediatrics*. 2022 Jan;149(1):e2020034371. IF:9,703(Q1)
- Dima AL, Allemann SS, Dunbar-Jacob J, Hughes DA, Vrijens B, Wilson IB. Methodological considerations on estimating medication adherence from self-report, electronic monitoring and electronic healthcare databases using the TEOS framework. *Br J Clin Pharmacol*. 2022 May. IF:9,473(Q1)
- Parody-Rua E, Bustamante A, Montaner J, Rubio-Valera M, Serrano D, Pérez-Sánchez S, et al. Modeling the potential efficiency of a blood biomarker-based tool to guide pre-hospital thrombolytic therapy in stroke patients. *Eur J Heal Econ HEPAC Heal Econ Prev care*. 2023 Jun;24(4):621-32. IF:5,271(Q1)
- Peñarrubia-María MT, Gil-Girbau M, Gallardo-González MC, Aznar-Lou I, Serrano-Blanco A, Mendive Arbeloa JM, et al. Non-initiation of prescribed medication from a Spanish health professionals' perspective: A qualitative exploration based on Grounded Theory. *Health Soc Care Community*. 2022 Jan;30(1):e213-21. IF:2,395(Q2)

## Psychological research group on fibromyalgia and chronic pain (AGORA group)

	<b>0</b>	COMMERCIAL CLINICAL TRIALS
	<b>1</b>	ACADEMIC CLINICAL TRIALS
	<b>9</b>	PROJECTS
	<b>14</b>	PUBLICATIONS
	<b>71.948</b>	IMPACT FACTOR
	<b>71%</b>	PUBLICATIONS IN 1RTS QUARTIL
	<b>14%</b>	PUBLICATIONS IN 1RTS DECIL
	<b>71%</b>	LEADERSHIP

**Juan Vicente Luaciano Devis, PhD**

In 2022, the most important developments with respect to the group have been its incorporation into the Centre for Network Biomedical Research – Epidemiology and Public Health (CIBERESP; CB22/02/00052), attached to ISCIII, and the recognition of the group's consolidated status by AGAUR (SGR 721). In addition to its accreditation, the group has received financing as a consolidated SGR group.

Likewise, it should be noted that Dr Jesús Montero Marín, a researcher with the University of Oxford's Psychiatry Department, obtained a Miguel Servet contract from ISCIII (CP21/00080) to join our group as a senior researcher.

Over the course of the year, the group has mainly focused its efforts on three clinical trials. The first one deals with the efficacy and cost-utility of two third-generation psychological therapies for patients with chronic lumbar pain and comorbid depression (IMPACT Project). The second trial has the purpose of evaluating the efficacy and cost-utility of a new

pharmacological therapy (low-dose naltrexone) for the handling of pain in fibromyalgia (INNOVA Project) in comparison to a placebo, while the third study is on the efficacy and cost-utility of a multicomponent treatment programme for the handling of fibromyalgia, FIBROWALK, analysing its benefits for physical and mental health in two formats: online and outdoor (On&Out Project).

### MEMBERS

#### STAFF RESEARCHER

Jesús Montero Marín, Juan Pablo Sanabria Mazo, Lluís Torruella Barraquer, Eugenia Cardeñosa Valera, Antoni Rozadilla Sacanell

#### POSTDOC ASSOCIATED RESEARCHERS

Adrián Pérez Aranda

#### PHD STUDENTS

Ariadna Colomer Carbonell

#### ASSISTANT RESEARCHER

Jaime Navarrete Hidalgo

#### COLLABORATOR RESEARCHER

Albert Feliu Soler, Xavier Borràs Hernández, María Teresa Navarro Gil

### SELECTED PROJECTS

- Effectiveness, cost-utility and neurobiological effects of low-dose naltrexone in patients with fibromyalgia (INNOVA project). (ICI20/00080). Instituto de Salud Carlos III (ISCIII). 2021-2024. IP: Juan Vicente Luciano Devis. CO-. IP: Antoni Rozadilla Sacanell
- Coste-utilidad y efectos fisiológicos de la Terapia de Aceptación y Compromiso y la Activación Conductual en pacientes con dolor crónico y depresión mayor comórbida (Proyecto IMPACT). (PI19/00112). Instituto de Salud Carlos III (ISCIII). 2020-2022. IP: Juan Vicente Luciano Devis.
- Coste-utilidad y efectos inmuno-inflamatorios de la terapia multicomponente FIBROWALK en formato online o al aire libre en fibromialgia: un ensayo controlado aleatorizado (PID2020-117667RA-I00). Proyectos I+D+I 2020 - Modalidades «Retos Investigación» y «Generación de Conocimiento». 2021-2023. IP: Dr. Albert Feliu Soler
- Serveis de Consultoria Grup AGORA. Swing Therapeutics, Inc. 2020-2022. IP: Juan Vicente Luciano Devis.

### SELECTED CLINICAL TRIALS & OBSERVATIONAL STUDIES

- Ensayo clínico farmacológico controlado con placebo, aleatorizado, doble ciego y 12 meses de tratamiento para evaluar la efectividad, coste-utilidad y efectos neurobiológicos de la Naltrexona a dosis baja en pacientes con fibromialgia (Proyecto INNOVA). (FSJD-INNOVA-2020). Adkroma Health Research, S.L; Fundació Privada per a la Recerca i la Docència Sant Joan de Déu - FSJD. 2022. IP: Juan Vicente Luciano Devis; CO-. IP: Antoni Rozadilla Sacanell.

### SELECTED PUBLICATIONS

- López-del-Hoyo Y, Collado-Navarro C, Pérez-Aranda A, García-Campayo J, López-Montoya A, Feliu-Soler A, et al. Assessing mindfulness and self-compassion facets as mediators of change in patients with depressive, anxious and adjustment disorders: Secondary data analysis of a randomized controlled trial. *J Context Behav Sci.* 2022;24:171-178. IF:5,138(Q1)
- Medina S, O'Daly OG, Howard MA, Feliu-Soler A, Luciano J V. Differential Brain Perfusion Changes Following Two Mind-Body Interventions for Fibromyalgia Patients: an Arterial Spin Labelling fMRI Study. *Mindfulness (N Y).* 2022;13(2):449-61. IF:3,801(Q2)
- Sanabria-Mazo JP, Colomer-Carbonell A, Carmona-Cervelló M, Feliu-Soler A, Borràs X, Grasa M, et al. Immune-inflammatory and hypothalamic-pituitary-adrenal axis biomarkers are altered in patients with non-specific low back pain: A systematic review. *Front Immunol.* 2022;945513. IF:8,786(Q1)
- Serrat M, Albajes K, Navarrete J, Almirall M, Lluch Girbés E, Neblett R, et al. Effectiveness of two video-based multicomponent treatments for fibromyalgia: The added value of cognitive restructuring and mindfulness in a three-arm randomised controlled trial. *Behav Res Ther.* 2022 Nov;158:104188. IF:5,321(Q1).

## 7.2 MENTAL HEALTH AREA

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### 7.2.2 Scientific bases and approach to mental disorders

Etiopathogenesis and treatment of severe mental disorders (MERITT)  
Cerebral imprints of gender, development and pathology

# Etiopathogenesis and treatment of severe mental disorders (MERITT)

	<b>0</b>	COMMERCIAL CLINICAL TRIALS
	<b>0</b>	ACADEMIC CLINICAL TRIALS
	<b>34</b>	PROJECTS
	<b>32</b>	PUBLICATIONS
	<b>161.788</b>	IMPACT FACTOR
	<b>31%</b>	PUBLICATIONS IN 1RTS QUARTIL
	<b>99%</b>	PUBLICATIONS IN 1RTS DECILO
	<b>69%</b>	LEADERSHIP

**Susana Ochoa Güerre, MD, PhD & Judith Usall i Rodié, MD, PhD**

The MERITT group has participated actively in 2022 in various financed research projects, including some multi-centre projects led by researchers of our group.

Three new research projects have been granted in the call made by the Instituto de Salud Carlos III. Their objectives are, respectively, the study of traumatic experiences, the determination of the efficacy of a family metacognitive intervention, and the analysis of the inflammatory effect of COVID on cognition. Likewise, a European project on personalised medicine (ERA-PERMED) has been obtained. It will begin in 2023 and its purpose is to evaluate a personalised metacognitive intervention based on machine learning. In addition, an ERASMUS project on correctional psychiatry has also been granted.

As far as publications are concerned, our impact factor has grown over those of previous years, registering a figure of 189.77 for this period. We made a total of 38 indexed publications, with 12 in Q1 and 21 in Q2. Many of them have been led by members of the group in collaborations with other national and international centres.

We also obtained a predoctoral grant from the Ministry and two grants allowing predoctoral members of the team to make stays at research centres abroad.

## MEMBERS

### STAFF RESEARCHER

Andrea Fontán Abeijón, Noemí del Prado Sánchez, José Manuel Fernández Rodríguez, Jaume Martínez Mas, Alvaro Muro Alvarez, Aitor-Eneko Olivé Albiztut, Jesús Pérez Pazos, María Ribas Siñol, Vicente Tort Herrando, María Luisa Barrigón Estevez, María José Escandell Morcillo, Belen Ramos JoseMaria, Antonio Manuel Alba Muñoz, Hilari Andres Mora, Belen Arranz Martí, Adela Berrozpe Blasco, Emma Casas Anguera, Marta Coromina Sadurní, Jorge Cuevas Esteban, Ruth Cunill Clotet, Gemma Escartín Martin, Gemma Escuder Romeva, Alexandrina Foix Sanjuan, Mar García Franco, Marcia Irazábal Giménez, Francisco Martínez Zambrano, Eva Miquel Fernandez, María Trinidad Peláez Martínez, Elena Rubio Abadal, Luis San Molina, Sonia Vilamala Anton

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### ASSISTANT RESEARCHER

Francisco Vidal Domenech, Gemma Riquelme Alacid, Alicia Colomer Salvans, Jordi Galiano Landeira, Marta Domínguez del Campo, Clara Serra Arumí, Marta Ferrer Quintero, Anna Butjosa Molines, Gabriel Ledesma Iparraguirre, Regina Vila Badia

### COLLABORATOR RESEARCHER

Joan Costa Escolà, Maria Elena Huerta Ramos, Raquel López Carrilero, Alfonso Monje Hernández

## SELECTED PROJECTS

- Impact of Traumatic Childhood Experiences (TCE) on cerebrocortical parameters and cognitive functioning in patients with a First-Episode Psychosis (FEP). (PI21/00067). Instituto de Salud Carlos III (ISCIII). 2022-2024. IP: Judith Usall i Rodié.
- Study of neuropsychological sequelae in patients affected by

SARS-CoV-2 infection: relationship with inflammatory and blood brain barrier disruption biomarkers. (PI21/00059). Instituto de Salud Carlos III (ISCIII). 2022-2024. IP: Belen Ramos JoseMaria.

- Pilot study to evaluate the effectiveness of online Family Metacognitive Training (MCTf) in mothers with psychosis and their adolescent children. (PI21/00012). Instituto de Salud Carlos III (ISCIII). 2022-2024. IP: Susana Ochoa Güerre.
- Creació i implementació d'una plataforma digital per a la detecció, comunicació i intervenció en la població adolescent i adults joves que pateixen o tenen risc de patir transtorns psicòtics. (SA51-006943). Departament de Salut - Generalitat de Catalunya. 2019-2023. IP: Marta Coromina Sadurní. IP: Belen Arranz Martí.

## SELECTED PUBLICATIONS

- Diago M, Vila-Badia R, Serra-Arumí C, Butjosa A, Del Cacho N, Esteban Sanjusto M, et al. Emotional abuse and perceived stress: The most relevant factors in suicide behavior in first-episode psychosis patients. Psychiatry Res. 2022 Sep;315:114699. IF:11,225(Q1)
- Ferrer-Quintero M, Fernández D, López-Carrilero R, Birulés I, Barajas A, Lorente-Rovira E, et al. Males and females with first episode psychosis present distinct profiles of social cognition and metacognition. Eur Arch Psychiatry Clin Neurosci. 2022 Oct;272(7):1169-81. IF:5,760(Q1)
- Ruiz-Delgado I, Moreno-Küstner B, García-Medina M, Barrigón ML, Gonzalez-Higueras F, López-Carrilero R, et al. Is Metacognitive Training effective for improving neurocognitive function in patients with a recent onset of psychosis? Psychiatry Res. 2022 Dec;318:114941. IF:11,225(Q1)
- Vila-Badia R, Serra-Arumí C, Butjosa A, Del Cacho N, Abella M, Colomer-Salvans A, et al. Risk and protective factors for the appearance of first-episode psychosis: The role of childhood trauma and coping strategies. Vol. 78, Asian J Psychiatr. 2022;78:103284. IF:13,890(Q1)

# Cerebral imprints of gender, development and pathology

- 0** COMMERCIAL CLINICAL TRIALS
- 3** ACADEMIC CLINICAL TRIALS
- 2** PROJECTS
- 8** PUBLICATIONS
- 33.357** IMPACT FACTOR
- 25%** PUBLICATIONS IN 1RTS QUARTIL
- 13%** PUBLICATIONS IN 1RTS DECIL
- 63%** LEADERSHIP

## Christian Stephan-Otto Attolini, PhD

During 2022, our group continued work on 3 main lines of research: Our large neuroimaging dataset project, in collaboration with Dr Maria Portella (Institut de Recerca Sant Pau) produced a recently accepted paper. Based on the study of 8000 children, we reported how longer-lasting breastfeeding results in better neurocognitive and cerebral tissue status of at 9-11 years old.

Our research on rare diseases with neurological involvement, in collaboration with Dr Angels García Cazorla (IRSJD), generated a first exploration on brain structure abnormalities related to neurotransmitter disorders. We showed the potential of assessing the brain volume of key gray matter structures to aid in the diagnosis and prognosis of these diseases.

From our collaboration with Dr Alfredo García-Alix (Fundación NeNe) and Dr Thais Agut (IRSJD) we reported how the precise location of neonatal arterial ischemic stroke lesions has an effect on cognitive and motor function at 2 years of age.

## MEMBERS

### STAFF RESEARCHER

Antonio Callen Soto, Maria Rosa Cambra Martí

### ASSISTANT RESEARCHER

Christian Núñez Leánez

### COLLABORATOR RESEARCHER

Gildas Yanin Guillaume Brebion

a voxel-based lesion-symptom mapping study. Arch Dis Child Fetal Neonatal Ed. 2022 Jan;107(1):45-50. IF:6,643(Q1).

## SELECTED PROJECTS

- Identificació dels principals trets cerebrals associats a les etapes més primerenques de diversos trastorns psiquiàtrics i neurològics. (05\_2020). Parc Sanitari de Sant Joan de Déu -Serveis de Salut Mental. 2021-2023. IP: Maria Rosa Cambra Martí.
- COGNIVITRA: COGNItive Vitality TRAining at home. (AC18/00015). Instituto de Salud Carlos III (ISCIII). 2019-2023. IP: Antonio Callen Soto.

## SELECTED CLINICAL TRIALS & OBSERVATIONAL STUDIES

- ACSF2004 - RGS@Home. (RGS@ Home). Fundació Privada per a la Recerca i la Docència Sant Joan de Déu - FSJD. 2020. IP: Antonio Callen Soto.
- European multicenter retrospective-prospective cohort study to observe Safinamide safety profile and pattern of use in clinical practice during the first post-commercialization phase SYNAPSES. (Z7219N02 / ZAM-SAF-2016-01). Zambon S.A. 2017. IP: Antonio Callen Soto.
- Estudio Prospectivo No Intervencionista De Pacientes Con Enfermedad De Alzheimer Leve A Moderada Y Sus Cuidadores En Cuatro Países Europeos. (MER-ALZ-2016-01). Merck, Sharp & Dohme de España. S.A. 2016. IP: Antonio Callen Soto.

## SELECTED PUBLICATIONS

- Alfonsi C, Stephan-Otto C, Cortès-Saladelafont E, Palacios NJ, Podzamczer-Valls I, Cruz NG, et al. Volumetric study of brain MRI in a cohort of patients with neurotransmitter disorders. *Neuroradiology*. 2022 Nov;64(11):2179-90. IF:2,995(Q3)
- Núñez C, Stephan-Otto C, Arca G, Agut T, Arnaez J, Cordeiro M, et al. Neonatal arterial stroke location is associated with outcome at 2 years:

# 7.3

## TRANSVERSAL PROGRAM

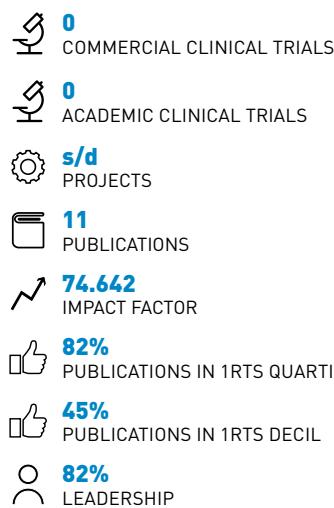
### 7.3.1

#### Health technologies and innovation

Medical technologies: bioinformatics and biomedical signals  
Medical technologies: computer graphics: serious games and gamification  
Medical technologies: biomaterials and tissue engineering  
Medical technologies: analysis for rehabilitation  
Medical technologies: instrumentation and eHealth  
Medical technologies: robotics and vision  
Medical technologies: biomechanics  
Patients engagement in research  
Innovation in health technologies



# Medical technologies: bioinformatics and biomedical signals



**Alexandre Perera Lluna, PhD**

## MEMBERS

### STAFF RESEARCHER

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### ASSISTANT RESEARCHER

Pol Gil Figuerola, Joshua Llano Viles, Pol Sola de los Santos

### TECHNICIAN

Joana Palés Huix, Enrico Manzini, Maria Maqueda

## SELECTED PUBLICATIONS

- Manzini E, Garrido J, Fonollosa J, Perera A. Mapping layperson medical terminology into the Human Phenotype Ontology using neural machine translation models. *Expert Syst. Appl.* 2022;204: 117446. IF:8,665. (Q1).
- Manzini E, Vlacho B, Franch J, Escudero J, Génova A, Reixach E

et al. Longitudinal deep learning clustering of Type 2 Diabetes Mellitus trajectories using routinely collected health records. *J Biomed Inform.* 2022;135:104218-104218. IF:8,000. (Q1).

- Muritala BA, Hernández AB, Sánchez MV, Perera A. #CoronavirusCruise: Impact and implications of the COVID-19 outbreaks on the perception of cruise tourism. *Tour. Manag. Perspect.* 2022;41:100948-100948. IF:7,608. (Q1).
- Solorzano A, Eichmann J, Fernandez L, Ziems B, Jimenez JM, Marco S et al. Early fire detection based on gas sensor arrays: Multivariate calibration and validation. *Sens. Actuator B-Chem.* 2022;352I: 130961. F:9,221. (Q1).

## SELECTED PROJECTS

- Métodos y aplicaciones de aprendizaje profundo y superficial de fenotipos para análisis y predicción de datos biomédicos. PROYECTOS DE GENERACIÓN DE CONOCIMIENTO. Agencia Estatal De Investigacion. PI: Alexandre Perera. 2022-2025.
- CAIXAIMPULSE VALIDATE CI21-00186. AJUTS A PROJECTES I ACTIVITATS DE VALORIZACIÓ DE LA RECERCA. Fundació La Caixa. PI: Alexandre Perera. 2021-2023.
- Xarxa R+D+I en Tecnologies de la Salut (XarTEC SALUT). 2018 XARDI 00016 / IU68-013944. XARXES R+D+I. Agencia De Gestio D'ajuts Universitaris I De Recerca (agaur). PI: Alex Perera. 2020-2022.

# Medical technologies: computer graphics: serious games and gamification

- 0** COMMERCIAL CLINICAL TRIALS
- 0** ACADEMIC CLINICAL TRIALS
- s/d** PROJECTS
- 3** PUBLICATIONS
- 14.872** IMPACT FACTOR
- 67%** PUBLICATIONS IN 1RTS QUARTIL
- 67%** PUBLICATIONS IN 1RTS DECIL
- 67%** LEADERSHIP

## Raül Benítez Iglesias, PhD

In 2022, we conducted numerous studies that employed computer vision, artificial intelligence, and pattern recognition techniques across various domains. These applications spanned areas such as biomedical materials, translational cardiology, and the detection of physiological anomalies. Our efforts resulted in the publication of nine articles in highly-ranked JCR journals. Additionally, we initiated a pioneering research direction focused on developing AI explainability methods, aimed at gaining deeper insights into how deep learning models identify specific image classes based on particular image regions and details. Furthermore, we established a highly active collaboration in the field of human-centric artificial intelligence with researchers specializing in computer vision at Amrita University in India.

## MEMBERS

### STAFF RESEARCHER

Dolors Ayala Vallespí, Maria Amor Duch Guillen, Jose Luis Eguía Gómez, Mercè Ginjaume Egido, Robert Joan Arinyó, Núria Pla García, Josep

Sempau Roma, Lluís Solano Albages, Daniela Tost Pardell, Marc Vigo Anglada

### PHD STUDENTS

Núria Bonet Codina, Ariel von Barnekow

## SELECTED PROJECTS

- Biologia Computacional i Sistemes Complexos. 2021 SGR 00582. AJUTS PER DONAR SUPORT A LES ACTIVITATS DELS GRUPS DE RECERCA DE CATALUNYA. Agència De Gestió D'ajuts Universitaris I De Recerca (agaur). PI: Clara Prats. 2022-2024
- Análisis computacional del impacto de riesgos genéticos y clínicos en la señalización molecular y disfunciones electrofisiológicas en fibrilación auricular. PID2020-116927RB-C22. RETOS DE INVESTIGACIÓN: PROYECTOS DE I+D+. Agencia Estatal De Investigacion. PI: Raul Benitez. 2021-2024.
- Xarxa R+D+I en Tecnologies de la Salut (XarTEC SALUT). 2018 XARDI 00016 / IU68-013944. XARXES R+D+I. Agència De Gestió D'ajuts Universitaris I De Recerca (agaur). PI: Alex Perera. 2020-2022.

## SELECTED PUBLICATIONS

- Marimon X, Traserra S, Jimenez M, Ospina A, Benítez R., Detection of Abnormal Cardiac Response Patterns in Cardiac Tissue Using Deep Learning. Mathematics. 2022;10(15) IF:2,592. (Q1).
- Nolla C, Casabella S, Jimenez V, Vallmitjana A, Tarifa C, Herráiz A et al.,  $\beta$ 2-adrenergic stimulation potentiates spontaneous calcium release by increasing signal mass and co-activation of ryanodine receptor clusters. Acta Physiol (Oxf). 2022;234(4)IF:7,523. (Q1).
- C Mata, J Munuera, A Lalande, G Ochoa-Ruiz, R Benitez, MedicalSeg: A Medical GUI Application for Image Segmentation Management, Algorithms 15 (6), 200. IF: 2,69 (Q2)
- A Herráiz-Martínez, C Tarifa, V Jiménez-Sábado, A Llach, H Godoy-Marín, et al., Influence of sex on intracellular calcium homoeostasis in patients with atrial fibrillation, Cardiovascular research 118 (4), 1033-1045. IF: 10,8 (Q1).

# Medical technologies: biomaterials and tissue engineering

- 0** COMMERCIAL CLINICAL TRIALS
- 0** ACADEMIC CLINICAL TRIALS
- s/d** PROJECTS
- 20** PUBLICATIONS
- 116.512** IMPACT FACTOR
- 75%** PUBLICATIONS IN 1RTS QUARTIL
- 15%** PUBLICATIONS IN 1RTS DECIL
- 50%** LEADERSHIP

## Cristina Canal Barnils, PhD

The Biomaterials, Biomechanics and Tissue Engineering Research Group brings together a multidisciplinary team of researchers with complementary backgrounds, such as engineers, chemists, physicists and biologists, which ensures the capacity to tackle the scientific challenges in the biomedical field from a global and integrated perspective and in 2022 has secured, again, its appointment as SGR research group by Generalitat de Catalunya.

During 2022, members of the BBT have published a total of 29 papers in JCR indexed peer-reviewed journals and have presented their work in over 47 Plenary, Keynote, Invited, Oral and Poster presentations in international conferences and workshops.

Dr. Maria-Pau Ginebra was granted with an ERC Advanced Grant (BAMMBI) by the European Research Council (ERC), while Dr. Cristina Canal was got a Proof of Concept project (TRANSFORMER), also funded by ERC, and Dr. Marta Pegueroles was granted with a grant by the MISTI Global Seed Funds of the MIT (USA). During 2022, the BBT was able to secure a total of 13 projects granted

by Agencia Estatal de Investigación, from several calls. In addition to this, the group has intensely carried out the design, management, organization and execution of R+D+I projects for national and international companies, and the implementation of studies and specific services of analysis and/or experimental characterization, with a focus on biomedical applications.

## MEMBERS

### STAFF RESEARCHER

Andrea Malandrino, Albert Espina Noguera, José Manuel García Torres, Montserrat Español Pons, Francesc Xavier Gil Mur, Jordi Guillem Martí, Jose María Manero Planella, Carles Mas Moruno, Marta Pegueroles Neyra, Daniel Rodríguez Rius

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### PHD STUDENTS

David Piñera Avellaneda, Laura del Mazo Barbara, Victor Chausse Calbet, Claudia García Mintegui, Xavier Solé Martí, Mar Bonany Mariñosa, Judit Buxadera Palomero, Miguel Mateu Sanz, Joanna Magdalena Konka, Lluís Oliver Cervelló, Yago Raymond Llorens, Elisa Rupérez de Gracia

### TECHNICIAN

Meritxell Sánchez Hervás, Noelia Aparicio Bádenas, Meritxell Molmeneu Trias, Miquel Punset Fusté

## SELECTED PUBLICATIONS

- García JM, Colombi S, Macor LP, Aleman C. Multitasking smart hydrogels based on the combination of alginate and poly(3,4-ethylenedioxythiophene) properties: A review. *Int. J. Biol. Macromol.* 2022;219:312-332. IF:8,200. (Q1).
- Oliver LL, Martin H, Mandakhbayar N, Jo YW, Cavalcanti EA, Kim HW et al. Mimicking Bone Extracellular Matrix: From BMP-2-Derived Sequences to Osteogenic-Multifunctional Coatings. *Adv Healthc Mater.* 2022;11(20)IF:10,000. (Q1).
- Przekora A, Penolazzi L, Kalisz G, Kazimierczak P, Canal C, Wojcik M et al. Osteoclast-mediated acidic hydrolysis of thermally gelled curdlan component of the bone scaffolds: Is it possible?. *Carbohydr Polym.* 2022;295:119914-119914. IF:11,200. (Q1).
- Raymond Y, Lehmann C, Thorel E, Benítez R, Riveiro A, Pou J et al. 3D printing with star-shaped strands: A new approach to enhance *in vivo* bone regeneration. *Biomater Adv.* 2022;137:212807-212807.

## SELECTED PROJECTS

- BAMMBI: Bio-inspired AntiMicrobial Bone Bloceramics: Deciphering contact-based biocidal mechanisms. Funding Agency: European Research Council. AdG Grant agreement ID: 101055053. PI: Prof. Maria-Pau Ginebra. 2022-2027
- TRANSFORMER: Transforming bone cancer therapy with composite biomaterials encapsulating plasma-generated RONS. Funding Agency: European Research Council. PoC – ERC. - Grant agreement: 101082096 – TRANSFORMER. PI: Dr. Cristina Canal. 2022-2024
- PLASMABOOST: Hidrogeles biomiméticos para liberar estrés oxidativo de plasmas fríos estimulan la regeneración de tejidos. Funding Agency: Instituto de Salud Carlos III (ISCIII). IHRC22/00003. PI: Dra. Cristina Canal. 2023-2024.

# Medical technologies: analysis for rehabilitation

- 0** COMMERCIAL CLINICAL TRIALS
- 0** ACADEMIC CLINICAL TRIALS
- s/d** PROJECTS
- 5** PUBLICATIONS
- 21.842** IMPACT FACTOR
- 40%** PUBLICATIONS IN 1RTS QUARTIL
- 20%** PUBLICATIONS IN 1RTS DECIL
- 40%** LEADERSHIP

**Miquel Ángel Mañas Villanueva,  
PhD**

## MEMBERS

### STAFF RESEARCHER

Joan Francesc Alonso Lopez, Sergio Romero Lafuente

## SELECTED PROJECTS

- MYOARM: tecnología inteligente para la rehabilitación motora. PDC2021-120818-I00. AYUDAS A PROYECTOS DE I+D+I PARA LA REALIZACIÓN DE «PRUEBAS DE CONCEPTO». Agencia Estatal De Investigación. PI: Miquel Ángel Mañas Villanueva. 2021-2024.
- Tecnologías innovadoras para monitorizar y personalizar la rehabilitación interdisciplinar de pacientes de cuidado intensivo. PID2020-117751RB-I00. RETOS DE INVESTIGACIÓN: PROYECTOS DE I+D+I. Agencia Estatal De Investigacion. PI: Miquel Ángel Mañas Villanueva. 2021-2024.
- MV-OPTIMIZER: A DIGITAL HEALTH SYSTEM FOR SAFE-PATIENT AND COST-EFFECTIVE MANAGEMENT OF MECHANICAL VENTILATION. 2021PROD00085. AJUTS DESTINATS A DONAR SUPORT A L'OBTENCIÓ DE PROTOTIPUS, I LA VALORIZACIÓ I TRANSFERÈNCIA

DELS RESULTATS DE LA INVESTIGACIÓ DELS EQUIPS DE RECERCA DE CATALUNYA. Agència De Gestió D'ajuts Universitaris I De Recerca (agaur). PI: Miquel Ángel Mañas Villanueva. 2022-2024.

## SELECTED PUBLICATIONS

- Borras M, Romero S, Alonso JF, Bachiller A, Serna LY, Migliorelli C et al. Influence of the number of trials on evoked motor cortical activity in EEG recordings. *J Neural Eng.* 2022;19(4): 046050. IF:5,043. (Q2).
- Ghaderi P, Nosouhi M, Jordanic M, Marateb HR, Mañas MA, Farina D. Kernel Density Estimation of Electromyographic Signals and Ensemble Learning for Highly Accurate Classification of a Large Set of Hand/Wrist Motions. *Front Neurosci.* 2022;16:796711. IF:5,152. (Q2).
- Migliorelli C, Medina IF, Bachiller A, Tost A, Alonso JF, López A et al. Cognitive stimulation has potential for brain activation in individuals with Rett syndrome. *J Intellect Disabil Res.* 2022;66(3):213-224. IF:3,646. (Q1).
- Ona G, Sampedro F, Romero S, Valle M, Camacho V, Migliorelli C et al. The Kappa Opioid Receptor and the Sleep of Reason: Cortico-Subcortical Imbalance Following Salvinorin-A. *Int J Neuropsychopharmacol.* 2022;25(1):54-63. IF:5,678. (Q1).

## Medical technologies: instrumentation and eHealth

- 0** COMMERCIAL CLINICAL TRIALS
- 0** ACADEMIC CLINICAL TRIALS
- s/d** PROJECTS
- 7** PUBLICATIONS
- 36.218** IMPACT FACTOR
- 43%** PUBLICATIONS IN 1RTS QUARTIL
- 14%** PUBLICATIONS IN 1RTS DECIL
- 43%** LEADERSHIP

### Xavier Rosell Ferrer, PhD

The "Medical technologies: instrumentation and eHealth" group has continued its collaboration with several hospitals on the following topics: with Hospital Sant Joan de Deu and the Autonomous University of Barcelona for the development of a point-of-care system for ammonium assessment, with the Cardiology service of the Hospital de Sant Pau and Santa Cruz on the diagnosis of cardiac arrhythmias, and in the same hospital with the pulmonology service for the evaluation of a minimally invasive method for the distinction of tumour tissues in the lung. A new three-year research project has been obtained in the latter area.

### MEMBERS

#### STAFF RESEARCHER

Ramon Bragós Badia, Mireya Fernández Chimeno, Miguel Angel García Gonzalez, Miguel J García Hernández, Lexa Nescolarde Selva, Juan Ramos Castro, Pere Joan Riu Costa

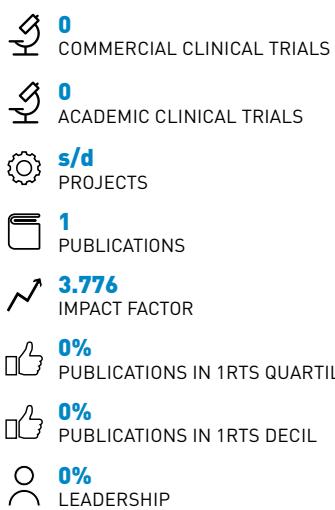
### SELECTED PROJECTS

- Biopsia Pulmonar Electrónica Guiada por Broncoscopia de Navegación Electromagnética. PID2021-1286020B-C21. Agencia Estatal De Investigacion. PLAN ESTATAL DE INVESTIGACIÓN CIENTÍFICA Y TÉCNICA Y DE INNOVACIÓN 2017-2020. PI: Nescolarde Selva, Lexa Digna and Bragos Bardia, Ramon. 2022-2025.
- European Training Network on Electromagnetic Risks in Medical Technology. H2020-955816-Eternity. EU. 2021. PI: Fernandez Chimeno, Mireya. 2021-2025.

### SELECTED PUBLICATIONS

- Amoros G, Casabella S, Company G, Arzamendi D, Macias Y, Jorge E et al. A closed-chest model of selective atrial myocardial infarction for the study of induced electrophysiological and structural derangements. *Eur. Heart J.* 2022;43(s2):2907IF:39,300. (Q1).
- Calvo A, Rebollo B, Ormazabal A, Artuch R, Rosell X, Alonso J et al. Biomedical point-of-care microanalyzer for potentiometric determination of ammonium ion in plasma and whole blood. *Anal. Chim. Acta.* 2022;1205:339782-339782. IF:6,200. (Q1).
- Company G, Nescolarde L, Pajares V, Torrego A, Riu P, Rosell X et al. Effect of Calibration for Tissue Differentiation Between Healthy and Neoplasm Lung Using Minimally Invasive Electrical Impedance Spectroscopy. *IEEE Access.* 2022;10:103150-103163. IF:3,900. (Q2).
- Company G, Nescolarde L, Pajares V, Torrego A, Riu P, Rosell X et al. Minimally Invasive Lung Tissue Differentiation Using Electrical Impedance Spectroscopy: A Comparison of the 3-and 4-Electrode Methods. *IEEE Access.* 2022;10:7354-7367. IF:3,900. (Q2).

## Medical technologies: robotics and vision



### Alicia Casals Gelpí, PhD

Research has had a clear continuity in its different fields in the area of robotics: Surgery and Rehabilitation and assistance. The scientific development relying on the advances on assisted teleoperation and in the automatization of certain processes to provide a level of autonomy to the user, either the patient, the surgeon or the therapist or assistant clinician. A special emphasis in this period has been put on the user interface as a way to improve the human -robot interaction and providing means to facilitate the control of a robot from a human both, by providing information (mainly visual) or providing some autonomous or assistive actions that release the user from tedious or too stressing tasks. This implies research on perception systems, mainly based on vision, force, or distance. In the surgical areas, both laparoscopy and colonoscopy have been the main fields of work, in assistance provide a means to easily control a complete robotized wheelchair by providing multifunctionality, mobility and actuations, with an intelligent interface that interpret the user's intention.

The motivation of research translation has also been a key point of the

activity, both assisting companies in the health sector in the development of new products and in making new steps towards the creation of a company in devices, rollators, for assisting user's mobility.

Intraluminal and Endovascular Autonomous Navigation: a Systematic Review. *IEEE Transact Robot.* 2022;IF:7,800. (Q1).

### MEMBERS

#### STAFF RESEARCHER

Joan Aranda López, Josep Fernández Ruzafa, Manel Frigola Bourlon, Enric Xavier Martín Rull, Antonio Benito Martínez Velasco

#### POSTDOC ASSOCIATED RESEARCHERS

Albert Hernansanz Prats

#### PHD STUDENTS

Narcís Sayols Baixeras

#### TECHNICIAN

Oriol Escoté Lluís

#### COLLABORATOR RESEARCHER

Antonio Benedico Blanes, Laureano Tinoco, Carlos Morata

### SELECTED PROJECTS

- ATLAS: Autonomous Intraluminal Surgey. H-2020 INT Marie Curie 813782. EU. H2020-MSCA-ITN-2018. PI: Alícia Casals. 2019-2023.
- Plataforma Híbrida Ortesis-Silla para hacer compatible la Movilidad, Funcionalidad Y Aceptabilidad de aplicación en entornos domésticos. PID2020-116091RB-I00. MINECO-Retos 2019. PI: Alícia Casals. 2021-2024.

### SELECTED PUBLICATIONS

- Farahi M, Casals A, Sarrafzadeh O, Zamani Y, Ahmadi H, Behbood N et al. Beat-to-beat fetal heart rate analysis using portable medical device and wavelet transformation technique. *Heliyon.* 2022;8(12):e12655. IF:4,000. (Q2).
- Finocchiaro M, Arezzo A, Menciassi A, Casals A, Hernansanz A, Ciuti G. Human machine interfaces for robot-assisted colonoscopy: a clinical survey. *EasyChair preprint.* 2022. 2p.
- Pore A, Finocchiaro M, Dall D, Hernansanz A, Ciuti G, Arezzo A et al. Colonoscopy Navigation using End-to-End Deep Visuomotor Control: A User Study. *IEEE International Conference on Intelligent Robots and Systems.* 2022:9582-9588.
- Pore A, Hernansanz A, Casals A. Motion Planning for Robotassisted

## Medical technologies: biomechanics

- 0** COMMERCIAL CLINICAL TRIALS
- 0** ACADEMIC CLINICAL TRIALS
- s/d** PROJECTS
- 7** PUBLICATIONS
- 30.694** IMPACT FACTOR
- 29%** PUBLICATIONS IN 1RTS QUARTIL
- 14%** PUBLICATIONS IN 1RTS DECIL
- 57%** LEADERSHIP

### Josep Maria Font Llagunes, PhD

In 2022, the BIOMECH group received funding for two new competitive research projects financed by the Spanish Research Agency [AEI]. One project aims to do basic research related to neuromusculoskeletal modeling and simulation of motor function recovery after spinal cord injury (Generation of Knowledge call), and the other is related to the development of smart robotic exoskeletons to assist people with disabilities in everyday life activities (Public-private partnership call together with ABLE Human Motion). This year the group also continued the collaboration with Sant Joan de Déu Barcelona Children's Hospital under the project ArmTracker (La Marató de TV3). Eight articles were published in JCR indexed journals in this period, 10 papers were presented in scientific conferences, and a doctoral thesis was presented. Additionally, the group's PI chaired the IFToMM Technical Committee for Multibody Dynamics. The group has been also an active member of the innovation networks Xartec Salut and i4KIDS, led by CREB (Research Centre for Biomedical Engineering) and Sant Joan de Déu Barcelona Children's Hospital, respectively. Lastly, Dr. Josep Maria Font was keynote speaker at the 18th ASME International Conference

on Multibody Systems, Nonlinear Dynamics, and Control (MSNDC), St. Louis, USA; and Rosa Pàmies won the best paper award at the Meeting of the National Chapter of the European Society of Biomechanics (ESB).

### MEMBERS

#### STAFF RESEARCHER

Oriol Alís González, Ana Barjau Condomines, Daniel Clos Costa, Rosa Pàmies Vilà

#### POSTDOC ASSOCIATED RESEARCHERS

Massimo Cenciarini, Albert Peiret Giménez, Joan Lobo Prat

#### PHD STUDENTS

Jesús de Miguel Fernández, Michael Ray Epke, Antonio Rodríguez Fernández, Míriam Febrer Nafría

### SELECTED PROJECTS

- Neuromusculoskeletal modeling and simulation of motor function recovery after robot-assisted walking rehabilitation (NIMBLE). Spanish Ministry of Science and Innovation (MICINN), AEI. PID2021-1236570B-C33. PI: Josep Maria Font Llagunes. 2022-2025.
- ABLE DailyExo: The next generation of smart robotic exoskeletons to assist people with disabilities in everyday life activities. Spanish Ministry of Science and Innovation (MICINN), AEI. CPP2021-008429. PI: Josep Maria Font Llagunes. 2022-2025.
- ArmTracker: A state-of-the-art wearable system to assess upper limb motor function in real-life conditions for patients with Duchenne muscular dystrophy and spinal muscular atrophy. La Marató de TV3 Foundation, call 2019. 202023-32. PI: Josep Maria Font Llagunes. 2021-2024.

### SELECTED PUBLICATIONS

- Rodríguez A, Lobo J, Tarragó R, Chaverri D, Iglesias X, Guiarao L et al. Comparing walking with knee-ankle-foot orthoses and a knee-powered exoskeleton after spinal cord injury: a randomized, crossover clinical trial. *Sci Rep.* 2022;12(1):19150-19150. IF:4,996. (Q2).
- Vega MM, Li G, Shourijeh MS, Ao D, Weinschenk RC, Patten C et al. Computational evaluation of psoas muscle influence on

walking function following internal hemipelvectomy with reconstruction. *Front Bioeng Biotechnol.* 2022;10:855870-855870. IF:6,064. (Q1).

- de Miguel J, Pescatore C, Mesa A, Rikhof C, Prinsen E, Font JM et al. Immediate Biomechanical Effects of Providing Adaptive Assistance With an Ankle Exoskeleton in Individuals After Stroke. *IEEE Robot. Autom. Lett.* 2022;7(3):7574-7580. IF:4,321. (Q2).
- Cárdenas AM, Uribe J, Font JM, Hernández AM, Plata JA. The effect of prosthetic alignment on the stump temperature and ground reaction forces during gait in transfemoral amputees. *Gait Posture.* 2022;95:76-83. IF:2,746. (Q2).
- Febrer M, Fregly BJ, Font JM. Evaluation of Optimal Control Approaches for Predicting Active Knee-Ankle-Foot-Orthosis Motion for Individuals With Spinal Cord Injury. *Front Neurorobot.* 2022;15:748148-748148. IF:3,493. (Q2).

# Patients engagement in research

- 0** COMMERCIAL CLINICAL TRIALS
- 0** ACADEMIC CLINICAL TRIALS
- 9** PROJECTS
- 7** PUBLICATIONS
- 48.16** IMPACT FACTOR
- 29%** PUBLICATIONS IN 1RTS QUARTIL
- 14%** PUBLICATIONS IN 1RTS DECIL
- 57%** LEADERSHIP

**Joana Claverol Torres, MSc & Begonya Nafria Escalera, MSc**

## MEMBERS

### STAFF RESEARCHER

Estibaliz Urarte Rodríguez, Júlia Giner Gràcia, Elisa Mora de Checa, Victor Royo Calavera, Claudia Alsina Steinemann

## SELECTED PUBLICATIONS

- Alonso S, Català M, López D, Álvarez E, Jordán I, García JJ et al. Individual prevention and containment measures in schools in Catalonia, Spain, and community transmission of SARS-CoV-2 after school re-opening. *PLoS One.* 2022;17(2): e0263741. IF:3,752. (Q2).
- Jordán I, Fernández de Sevilla M, Fumadó V, Bassat Q, Bonet E, Fortuny C et al. Transmission of Severe Acute Respiratory Syndrome Coronavirus 2 Infection Among Children in Summer Schools Applying Stringent Control Measures in Barcelona, Spain. *Clin Infect Dis.* 2022;74(1):66-73. IF:20,999. (Q1).
- Mele M, Launes C, Fernández de Sevilla M, Hernandez M, Pons G, Bassat Q et al. Low transmission of SARS-CoV-2 derived from children in family clusters: An observational study of family households in the Barcelona Metropolitan Area, Spain. *PLoS One.* 2022;17(11): e0277754. IF:3,752. (Q2).
- Rocafort M, Henares D, Brotons P, Launes C, Fernández de Sevilla M, Fumadó V et al. Impact of COVID-19 Lockdown on the Nasopharyngeal Microbiota of Children and Adults Self-Confining at Home. *Viruses.* 2022;14(7): 1521. IF:5,818. (Q2).

## SELECTED PROJECTS

- EU Network of Youth Cancer Survivors. (101056918). European Commission. 2022-2024. IP: Begoña Nafria Escalera.
- Science4Pandemics: Cizitens engagement digital platform for collective intelligence in pandemics. (220365). EIT Health e.V; Fundació Privada per a la Recerca i la Docència Sant Joan de Déu - FSJD. 2022-2024. IP: Begoña Nafria Escalera.
- EJP RD: European Joint Programme on Rare Diseases. (825575). European Commission. 2019-2023. IP: Joana Claverol Torres.
- c4c\_connect4children (COllaborative Network for European Clinical Trials For Children). (777389). European Commission. 2018-2024. IP: Joana Claverol Torres.

# Innovation in health technologies

- 0** COMMERCIAL CLINICAL TRIALS
- 0** ACADEMIC CLINICAL TRIALS
- 19** PROJECTS
- 2** PUBLICATIONS
- 6.99** IMPACT FACTOR
- 0%** PUBLICATIONS IN 1RTS QUARTIL
- 0%** PUBLICATIONS IN 1RTS DECIL
- 100%** LEADERSHIP

Salut - Generalitat de Catalunya.  
2019-2022. IP: Arnau Valls Esteve.

## SELECTED PUBLICATIONS

- COVIDSurg C, GlobalSurg C. SARS-CoV-2 infection and venous thromboembolism after surgery: an international prospective cohort study. *Anaesthesia*. 2022;77(1):28-39. IF:12,893. (Q1).
- Garcia P, Isabel A, Aurensanz E, Valls A, Miguel FA, Ormazabal D et al. Machine Learning-Based Systems for the Anticipation of Adverse Events After Pediatric Cardiac Surgery. *Front Pediatr.* 2022;10:930913. IF:3,569. (Q2).
- Pereira HR, Barzegar M, Hamadelseed O, Valls A, Munuera JL. 3D surgical planning of pediatric tumors: a review. *Int J Comput Assist Radiol Surg.* 2022;17(4):805-816. IF:3,421. (Q2).

**Jaume Pérez Payarols, MD, PhD**

## MEMBERS

### STAFF RESEARCHER

Carmen De la Gala Otero, José M. Quintillá Martínez, Josep Rubio Palau, Arian Tarbal Roquer, Arnau Valls Esteve

### ASSISTANT RESEARCHER

Trinidad Arenas Costa, Júlia Meca Santamaría, Gerard Marturià Navarro, Sofia Ferreira Quartino, Alba Clavero Ros, Nuria Esperanza Adell Gómez, Pilar Puig i Sàrries, Xavier Puñet Ortiz, Ariadna Ballesteros Herruzo

## SELECTED PROJECTS

- FUNNY FRIENDS. Fundació Barcelona Mobile World Capital Foundation. 2021-2022. IP: Arnau Valls Esteve.
- AICCELERATE\_AI Accelerator – A Smart Hospital Care Pathway Engine. (101016902). European Commission. 2021-2024. IP: Arnau Valls Esteve, Sergi Navarro Vilarrubi, Joan Sánchez de Toledo.
- Hub d'Innovació Pediàtrica. (2018 XARDI 00011). Agaur - Agència de Gestió d'Ajuts Universitaris i de Recerca. 2020-2022. IP: Jaume Pérez Payarols.
- P2 - FUSE3D: Tecnologies per a deposició de material semifos. (IU16-011722). Departament de



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