



research  
science  
innovation  
commitment

ANNUAL SCIENTIFIC  
**REPORT 2017**

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# 1 FOREWORD

- 1.1. Introduction
- 1.2. Governing and advisory bodies
- 1.3. Services and infrastructures

## 1.1. Introduction

Our thanks to all the researchers who have made it possible, once again this year, for our research institute to continue to raise its high standard of expertise and to enhance the quality of its knowledge, as well as the translation of this knowledge to clinical practice. The intense rigorous work carried out by the Institut de Recerca Sant Joan de Déu (IRSJD) is unquestionably the most important contribution that we can offer to our patients.

IRSJD is a young institution that was formally created in 2015 and as such it has many challenges to face. One of these is to achieve the Public Administration's recognition of the Institute's high level of research quality. In 2017, IRSJD was assessed by the prestigious CERCA institution of the Government of Catalonia and the outcome was highly encouraging, not only because of the positive result of the appraisal but also because it gave us the chance to receive a broad series of recommendations which will help us to grow and to improve.

The year 2017 has also been marked by the effective incorporation of some of the research groups of the Parc Sanitari Sant Joan de Déu within the structure of IRSJD. The Parc Sanitari Sant Joan de Déu is an institution which counts among its strategic goals the integration of mental health as a natural aspect of our society. The mental health research groups provide a high level of quality research and scientific output to our Institute. We welcome them and sincerely hope that our scientific collaboration will prove very beneficial.

Drawing up a balance of the results obtained in 2017, we find that the Institute continues to follow a trend of sustained growth, having achieved more clinical trials and more competitive projects as well as a substantial increase of scientific publications reflecting qualitative growth and an impact factor of over 450 points, which goes to show the high level of competence of our researchers. Our joint collaborative high-quality endeavours have allowed us to improve some basic infrastructures such as the new Daniel Bravo Centre for Rare Disease Diagnosis and Research, to be a reference institution in paediatric clinical trials under European financing, to have increased our participation in financed European research projects and networks, and to publish significant papers in leading scientific journals of great prestige and impact.

This work in collaboration of the IRSJD research groups, which belong to highly regarded universities and hospitals, focuses scientific and technical solutions on the clinical problems of people and especially of those who are most vulnerable, like children and adolescents, and mental patients, improving the results of the medicine which we seek to practise and of the science which we seek to develop.

## 1.2. Governing and advisory bodies

### Board of trustees

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 Director, Research Foundation Sant Joan de Déu  
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 Director of Innovation, Hospital Sant Joan de Déu  
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 Director, Institut de Biomedicina - Universitat de Barcelona (IBUB)  
 Institut de Neurociències - Universitat de Barcelona (UBNeuro)  
 Scientific manager, Institut de Recerca Sant Joan de Déu

### Scientific Advisory Council

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Instituto de Investigación Sanitaria Hospital 12 de Octubre  
 Parc Recerca Biomèdica de Barcelona. Fundació Pasqual Maragall - Universitat Pompeu Fabra  
 Genomic Regulation Centre of Barcelona.  
 London's Global University. Division of Psychiatry  
 Deputy Director, General Management of the Biomedical Research Institute of Bellvitge (IDIBELL)  
 St. Jude Children's Research Hospital  
 General Director, BIOCAT  
 Institut de Recerca de l'Hospital de la Santa Creu i Sant Pau - Universitat Autònoma de Barcelona

### Research commission

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 Josep Jiménez Chillarón  
 Ángel Montero Carcaboso  
 Carmen Muñoz Almagro  
 Susana Ochoa Güerre  
 Francesc Palau Martínez  
 Marçal Pastor Anglada  
 Antoni Serrano Blanco  
 Judith Usall Rodie  
 Júlia Ribot Ballabriga, secretary

## 1.3. Services and infrastructures

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 IRJSD offers its professionals technical, scientific and support services as well as the most suitable infrastructures required to carry out their work.

### Scientific Core facilities

Clinical Trials Unit  
Sequencing Unit  
Confocal Microscopy Unit  
Animal housing facility  
Biobank  
Methodological and statistical support  
Methodological support in neuroimaging  
Scientific and technical evaluations service  
Library

### Other scientific and technical services

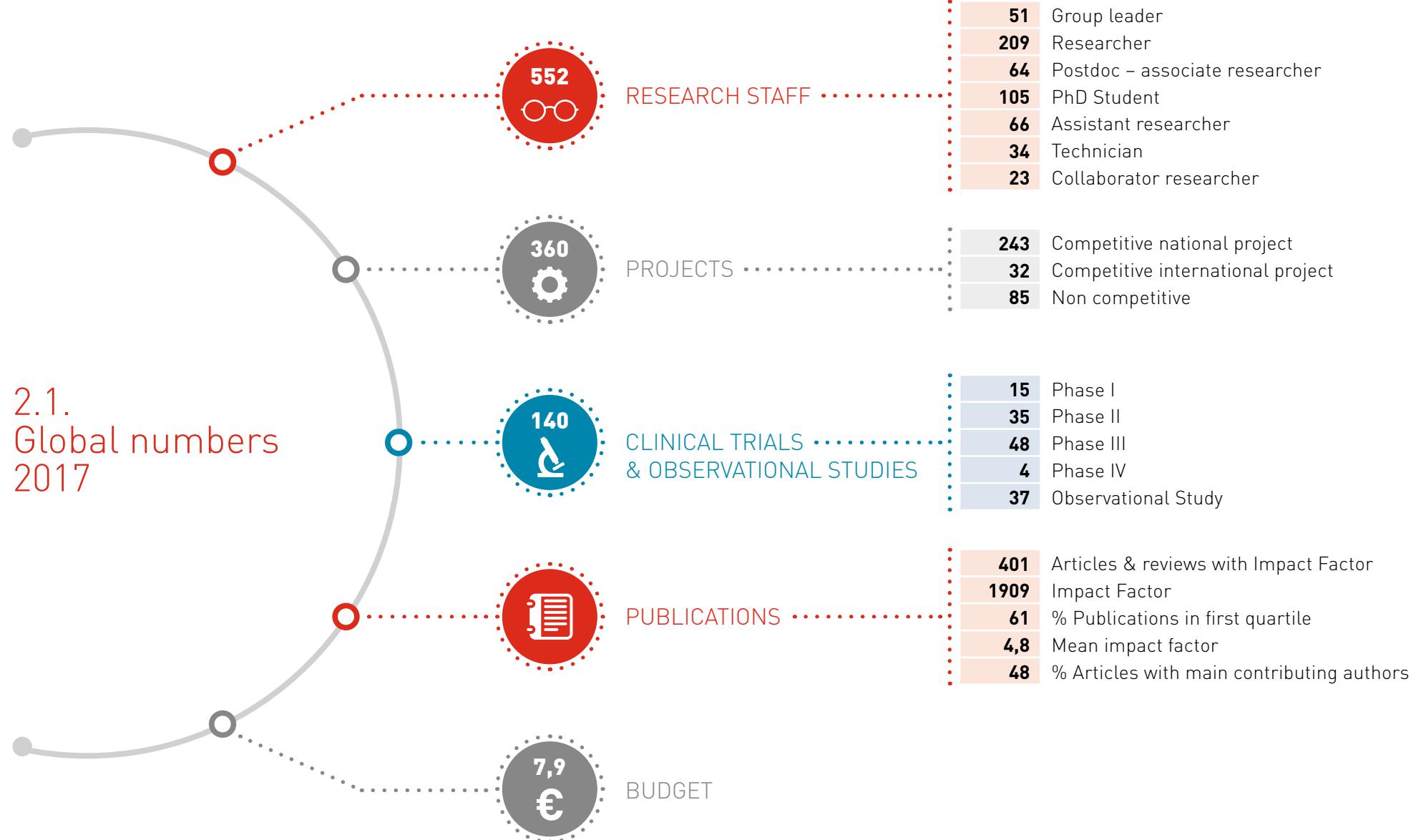
UBNeuro Services  
IBUB Services  
CREB Services

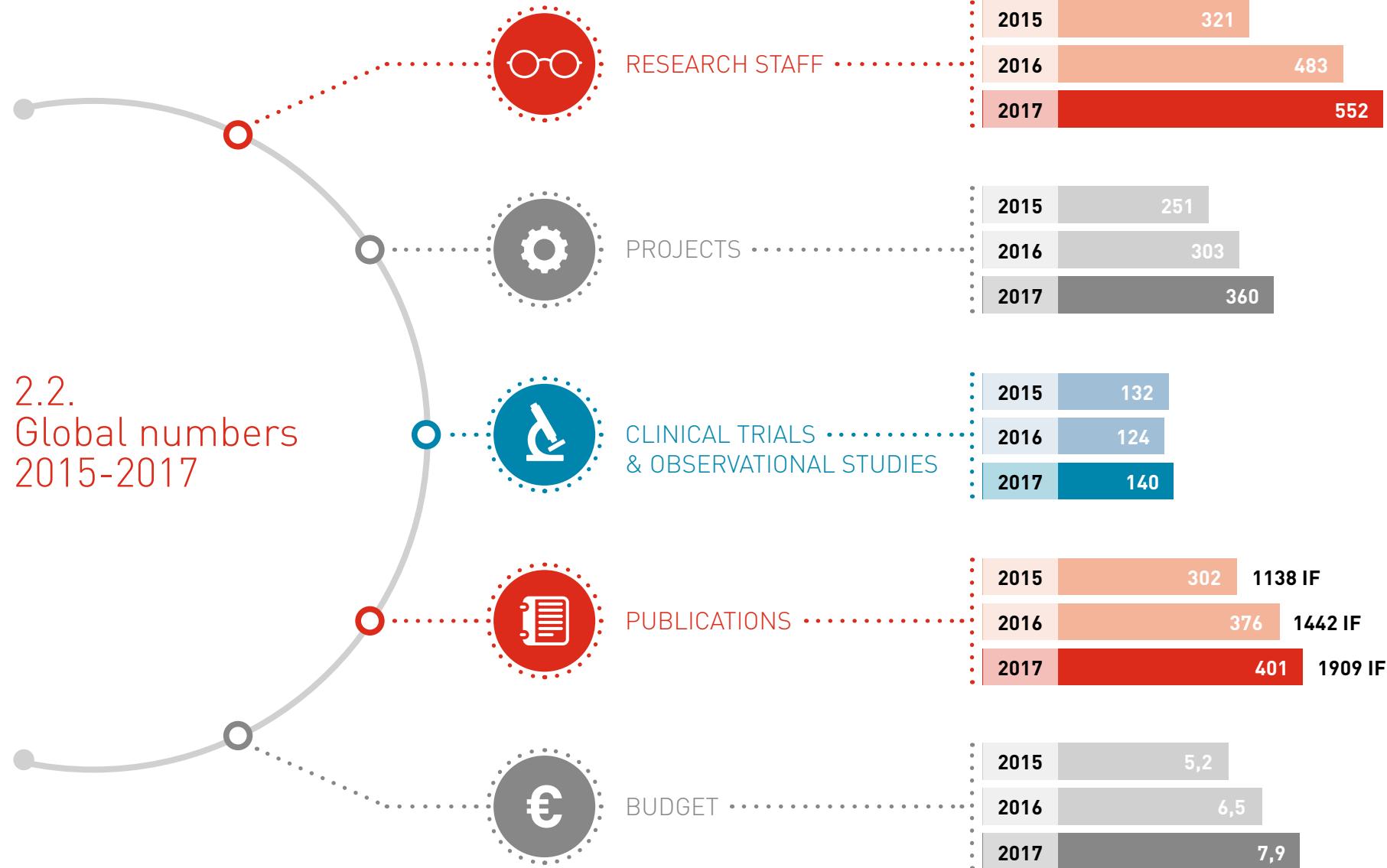
### Management support services

Administration and human resources  
Clinical Research Ethics Committee  
Research management and Promotion  
Knowledge management and communication

## **2** GLOBAL NUMBERS

- 2.1. Global numbers 2017
- 2.2. Global numbers 2015-2017
- 2.3. Research and innovation outputs
  - Scientific publications
  - Patents and utility models
- 2.4. Training
  - Doctoral theses
  - Scientific seminars





## 2.3. Research and innovation outputs

### Scientific publications



ARTICLES &  
REVIEWS WITH  
IMPACT FACTOR



IMPACT FACTOR



% ARTICLES  
IN THE FIRST  
QUARTILE



MEAN IMPACT  
FACTOR



% ARTICLES WITH  
MAIN CONTRIBUTING  
AUTHORS

% publications in the first quartile: % of publications published in journals ranked in the first quartile (top 25%)  
Mean impact factor: Mean impact factor of all the scientific publications

### Highest impact factor journals

The list reflects the 98 original articles and reviews published in the most relevant scientific journals (1st decil)

Journal	IF	Total articles
LANCET	47,831	1
NATURE	40,137	1
NATURE MEDICINE	29,886	1
NATURE GENETICS	27,959	2
CANCER CELL	27,407	3
WORLD PSYCHIATRY	26,561	1
JOURNAL OF CLINICAL ONCOLOGY	24,008	1
JOURNAL OF THE AMERICAN COLLEGE OF CARDIOLOGY	19,896	1
LANCET DIABETES & ENDOCRINOLOGY	19,742	1
CIRCULATION	19,309	1
LANCET RESPIRATORY MEDICINE	19,287	2

Journal	IF	Total articles
CELL METABOLISM	18,164	1
JAMA PSYCHIATRY	15,307	4
ANNALS OF THE RHEUMATIC DISEASES	12,811	2
NATURE COMMUNICATIONS	12,124	2
INTENSIVE CARE MEDICINE	12,015	1
JOURNAL OF EXPERIMENTAL MEDICINE	11,991	1
PLOS MEDICINE	11,862	1
ANNALS OF ONCOLOGY	11,855	1
TRENDS IN ENDOCRINOLOGY AND METABOLISM	10,893	1
EUROPEAN RESPIRATORY JOURNAL	10,569	1
JAMA PEDIATRICS	10,251	1

Journal	IF	Total articles
NAT PROTOC	10,032	1
ANNALS OF NEUROLOGY	9,890	1
CLINICAL CANCER RESEARCH	9,619	3
DIABETES	8,684	1
CLINICAL INFECTIOUS DISEASES	8,216	1
BMC MEDICINE	8,097	2
JOURNAL OF CONTROLLED RELEASE	7,786	2
INTERNATIONAL JOURNAL OF EPIDEMIOLOGY	7,738	1
HAEMATOLOGICA	7,702	1
SCHIZOPHRENIA BULLETIN	7,575	1
ACS APPLIED MATERIALS & INTERFACES	7,504	1
AGEING RES REV	7,452	1
MOVEMENT DISORDERS	7,072	1
CLINICAL EPIDEMIOLOGY	7,056	1
AMERICAN JOURNAL OF CLINICAL NUTRITION	6,926	1
ARTHRITIS & RHEUMATOLOGY	6,918	1
SPORTS MEDICINE	6,832	1
ACTA PSYCHIATRICA SCANDINAVICA	6,790	2
AGING CELL	6,714	1
CEREBRAL CORTEX	6,559	1
BRITISH JOURNAL OF PSYCHIATRY	6,347	2
INT J NEURAL SYST	6,333	1
ACTA BIOMATERIALIA	6,319	3
JOURNAL OF INFECTIOUS DISEASES	6,273	1
STROKE	6,032	1
NEUROIMAGE	5,835	1
PEDIATRICS	5,705	1
BIORESOURCE TECHNOLOGY	5,651	1

Journal	IF	Total articles
STEM CELLS	5,599	1
CLINICAL MICROBIOLOGY AND INFECTION	5,292	1
JOURNAL OF CLINICAL PSYCHIATRY	5,291	1
PSYCHOLOGICAL MEDICINE	5,230	7
AMERICAN JOURNAL OF OBSTETRICS AND GYNECOLOGY	5,226	2
DEPRESSION AND ANXIETY	4,971	1
AMERICAN JOURNAL OF EPIDEMIOLOGY	4,825	1
TISSUE ENGINEERING PART A	4,697	1
J AM GERIATR SOC	4,388	1
CORTEX	4,279	1
JOURNAL OF ADOLESCENT HEALTH	3,974	1
JOURNAL OF PEDIATRICS	3,874	2
JOURNAL OF NEUROPSYCHOLOGY	3,634	1
PEDIATRIC CRITICAL CARE MEDICINE	3,495	4
PEDIATRIC OBESITY	3,400	2
BRITISH JOURNAL OF GENERAL PRACTICE	3,041	1
BILING-LANG COGN	3,010	1
J WOMENS HEALTH	2,322	1
J APPL RES INTELLECT	2,112	1

## Patents and utility models

### Nonwoven Membrane as a drug delivery System

Date of grant	07/02/2017
Country of protection	United States
Grant number	9,561,190
Applicants	Universitat Politècnica de Catalunya & Hospital Sant Joan de Déu
Inventors	Montero Carcaboso A – Hospital Sant Joan de Déu. Others

### Treatment of hepatic steatosis related oligo-ovulation

Date of grant	04/05/2017
Grant number	WO 2017/072243
Applicants	Katholieke Universiteit Leuven & Hospital Sant Joan de Déu
Inventors	Ibáñez L – Hospital Sant Joan de Déu. Others

### Immunotherapy for CaSR-expressing cancer (e.g., neuroblastoma)

Date of grant	26/01/2017
Grant number	WO 2017/013276 A1
European patent	Nº 15756941.9
Applicants	Hospital Sant Joan de Déu, Barcelona & Fundació Cellex, Barcelona
Agent	Herrero & Asociados.
Inventors	de Torres C – Hospital Sant Joan de Déu

### System to assist walking / Sistema para asistir a caminar

Date of grant	17/10/2017
Country of protection	Spain
Grant number	P201600886. PCT extension: PCT/EP2017/076482
Owner	UPC, UDC, UEX
Inventors	Lugrís U, Cuadrado J, Font-Llagunes JM, Clos D, Alonso FJ, Romero F - CREB

### Sistema de contención y estimulación sensorial para bebés prematuros

Date of grant	04/04/2017
Grant number	ES1180334
Applicants	Fundació Privada Elisava Escola Universitària i Hospital Sant Joan de Déu
Inventors	Díaz Mecchia E, del Corral AM, González Colominas M, García Fernández M

## 2.4. Training

### Doctoral theses

A list provided below the 32 doctoral theses presented during 2017 in original language.

Thesis Author	Title	Supervisors
Angélica Ivone Avilés Rivero	Estimating and understanding motion: from diagnostic to robotic surgery	Alicia Casals
Ignacio Azanar Lou	El problema de la no iniciación del tratamiento farmacológico: evaluación con métodos cuantitativos	Maria Rubio Antoni Serrano
Carles Bautista-Rodríguez	Relación entre la enfermedad neumocócica invasiva y el déficit genético de MBL (mannose-binding lectin)	Carmen Muñoz-Almagro Carles Launes Iolanda Jordán
Anna Butjosa Molines	Stressful life events and first-episode psychosis	Susana Ochoa Güerre Juana Gómez
Judit Buxadera	Development of polymeric coatings with combined antifouling/antibacterial properties for titanium dental implants	Daniel Rodríguez Rius Xavier Gil
Montserrat Cairó Calzada	Autofagia en la activación termogénica del tejido adiposo marrón	Francesc Villarroya Joan Villarroya
María Alexandra Cañas Pacheco	Comunicación microbiota-huésped: internalización y señalización intracelular de vesículas de membrana externa de cepas probióticas y comensales de <i>E. coli</i> en células de epitelio intestinal	Laura Baldomà Josefa Badia
María Isabel Castellanos Arboleda	Functionalized CoCr surfaces with adhesive molecules to improve endothelialization	Marta Pegueroles José M. Manero
Marta de Castro Miró	Metodologies d'alt rendiments per a la identificació de noves mutacions i gens causants de distrofies de retina hereditàries	Roser González-Duarte Bru Cormand Rifà
Albert Català Temprano	Caracterització de les alteracions moleculars de FLT3 en pacients pediàtrics afectes de leucèmia aguda. Correlació amb l'activitat i expressió dels transportadors de fàrmacs derivats de nucleòsids	Mireia Camós Guijosa Marçal Pastor Anglada
Anna Díez-Escudero	Tuning the biological performance of calcium phosphates through microstructural and chemical modifications	Maria Pau Ginebra Montserrat Español
Behjat Gholami	Functional analyses of candidate genes for osteoporosis: RUNX2 and LRP5	Susana Balcells Daniel Grinberg
Soledad Gómez-González	Developmental Tumors, Epigenetics and microRNAs	Cinzia Lavarino
Marcia Irazabal Jiménez	Carga e impacto familiar de las cuidadoras y los cuidadores de personas adultas con discapacidad intelectual con o sin trastorno mental asociado	Susana Ochoa Güerre Crescencia Pastor Vicente

Thesis Author	Title	Supervisors
Kanupriya Khurana	Bone Regeneration by tuning the drug release from calcium phosphate scaffolds	Maria Pau Ginebra Cristina Canal Frank Mücklich
María Jesús Leal-Witt	Efecto de una intervención del estilo de vida en el perfil metabólico de sujetos prepuberales con obesidad	Carles Lerin
Hadiseh Mahdavi	A contribution to unobtrusive measurement methods for sleep monitoring based on magnetic induction technique	Javier Rosell Ferrer
Ricardo Moure Ortega	Alteraciones en la señalización de FGF21 asociadas a la terapia antirretroviral y la adiposidad	Francesc Villarroya Marta Giralt
Diana Carolina Muñoz Lasso	Fisiopatología de la ataxia de Friedreich: transporte y degeneración axonal	Francesc Palau Martínez Pilar González Cabo
Maria del Mar O'Callaghan	Nuevos aspectos clínicos, bioquímicos, genéticos y terapéuticos en la encefalomielopatías mitocondriales en la edad pediátrica	Mercè Pineda Rafael Artuch
Juan Darío Ortigoza Escobar	Estudio de defectos en el transporte y el metabolismo de tiamina asociados a encefalopatías recurrentes en la infancia	Belén Pérez Dueñas
Monica Piquer-Gisbert	Fenotips d'immunodeficiència comuna variable (IDCV) en pacients pediàtrics. Correlació genotip-fenotip en l'evolució clínica i el tractament dels pacients	Mª Anunciación Martín Mateos Manel Juan Otero
Sara Principi	Development of methodologies for estimating the dose to the eye lens in Interventional radiology. Operational implications of the eye lens new dose limit	Mercè Ginjaume
Alejandra Margarita Ríos Hernández	Diet and sleep in children and adolescents with Attention-Deficit Hyperactivity Disorder	María Luz Izquiero Pulido José Ángel Alda Díez
Alaleh Safari	Time dependent flow of biolubricant and suspended particles behavior within total hip replacement	Maria Pau Ginebra Nazanin Emami Michel J. Cervantes
Romain Schieber	Novel functionalized and patterned surfaces for cardiovascular applications	Marta Peguerolés Frank Mücklich
Carina Shianya Álvarez	Modulación de la barrera epitelial intestinal por vesículas de membrana y factores solubles secretados por cepas probióticas y comensales de Escherichia coli	Laura Baldomà Josefa Badia
Sara Siddi	Neurociencia de las alucinaciones: mecanismos neurocognitivos, neurofuncionales y neuroestructurales	Gildas Brebion Christian Stephan-Otto
Antonio Soriano Asrandes	Influencia de los cambios migratorios en la epidemiología de la transmisión vertical del virus de la inmunodeficiencia humana en Catalunya (período 2000-2014)	Clàudia Fortuny Antoni Noguera
Maria Teixidó Ibáñez	Constraints on early word segmentation and mapping	Laura Bosch Galceran
Albert Viel Oliva	Determinants estructurals i implicacions funcionals de l'interactoma protèic d'hCNT1	Sandra Pérez Torras Marçal Pastor Anglada
David Vila-Pérez	Evaluación de los factores pronósticos de sepsis en la edad pediátrica	Iolanda Jordán

## Scientific seminars

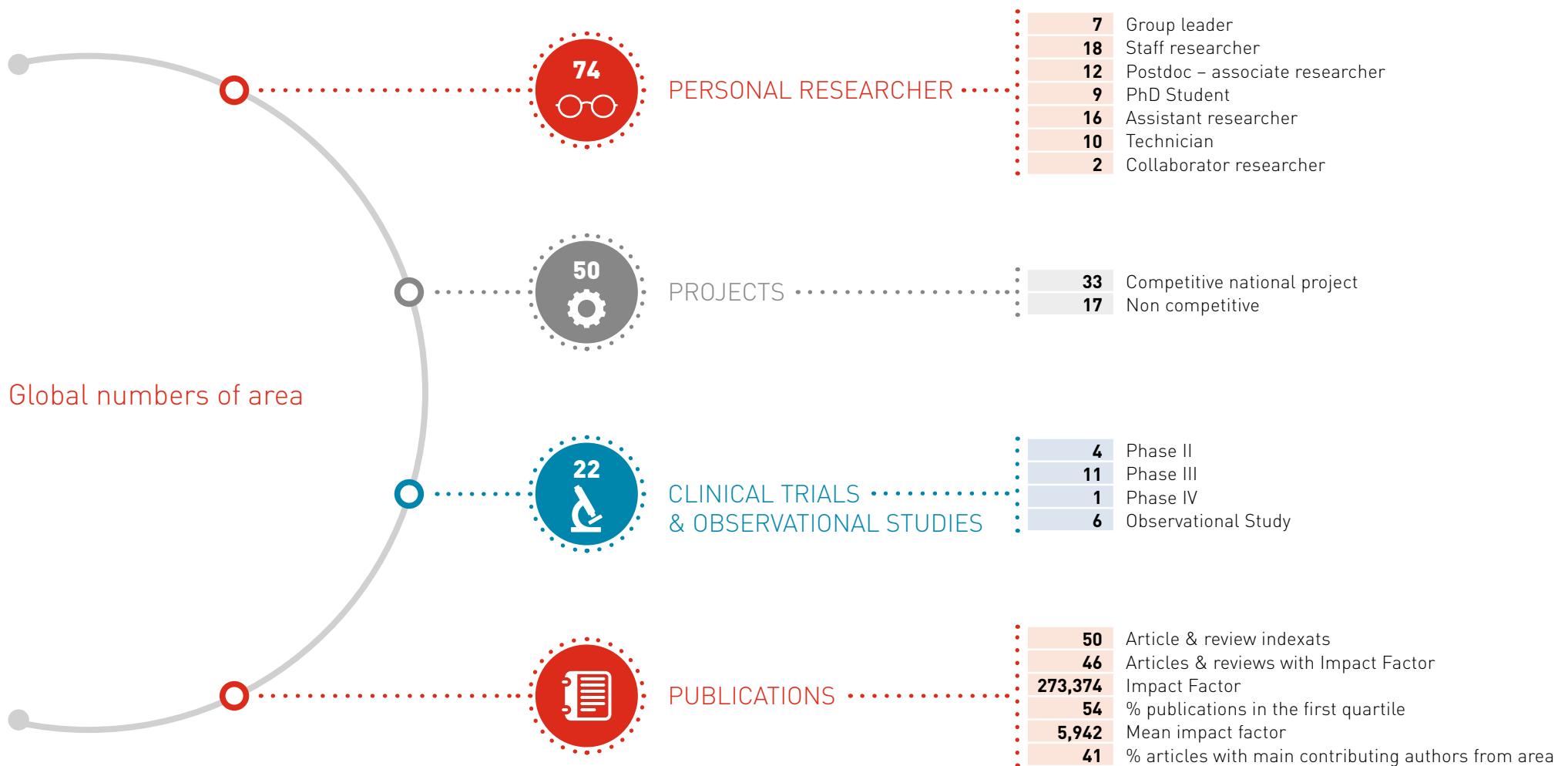
- January, 24. Neuroprotection and regeneration/repair by combined therapy (hypothermia + allopurinol) in an animal model of hypoxic-ischaemic brain injury
- February, 7. Evolution of nosocomial infection in paediatric intensive care units: ENVIN-HELICS study
- February, 21. Sudden death in children and adolescents in Catalonia. How genetics helps us and implications for the family
- March, 7. Insulin receptor and endothelial adenosine axis in gestational diabetes mellitus
- April, 4. New generation genetic sequencing of patients with radiological anomalies of the basal ganglia
- April, 24. Assessment of effectiveness of a multiple intervention addressed to improving early empirical antibioticotherapy in child sepsis: paediatric ABISS-EDUSEPSIS study
- May, 9. New generation genetic sequencing of patients with radiological anomalies of the basal ganglia
- May, 16. Best practices for project management
- May, 23. A journey from the gene to the phenotype
- June, 6. Studies of disorders using advanced tools in systems biomedicine: derivation of multi-level maps
- June, 20. Imprinting in humans: the role of imprinting in the placenta and in cancer
- October, 3. Our preclinical path to a phase I trial on advanced therapy in retinoblastoma
- October, 17. Influence of nurse supervision on adherence to treatment with antituberculosis drugs in the paediatric population
- October, 31. European Financing Opportunities in Research in the Health Field - 2018
- November, 14. ABLE: A low-cost robotic exoskeleton for gait rehabilitation
- November, 11. Neonatal ECG screening for early detection of long QT syndrome
- November, 29. RNA therapies for inherited retinal diseases: design & proof of concept studies
- December, 1. Identification and characterization of the molecular pathway controlling SAMHD1 function: implication in HIV infection and beyond
- December, 12. Design and assessment of innovative arm supports for children and adults with Duchenne muscular dystrophy

## **3 VERTICAL AREAS**

- 3.1. Paediatric neuroscience
- 3.2. Molecular and cellular biology of developmental tumour
- 3.3. Adult-age disorders originating in the foetal period or in the early years of life
- 3.4. Infectious diseases and systemic inflammatory response in paediatrics
- 3.5. Mental Health
- 3.6. Foetal / Paediatric Diseases and environment, metabolic and genetic factors

## 3.1. Paediatric neuroscience

**Coordinator:** Jaume Campistol and Rafael Artuch



## PKU AND OTHER AMINO ACID GENETIC DISEASES

Jaume Campistol Plana, MD, PhD

*The fact that our hospital institution is a reference centre for phenylketonuria in Catalonia has allowed us to study in depth the pathophysiological mechanisms involved in this disease in our patients and to test new treatments aimed to improve their metabolic control and neurologic prognosis. Our studies have led to the conclusion that some neuropsychological and neurological alterations are associated, on the one hand, with metabolic control, and on the other with antioxidant deficiencies. We have begun new treatments (with tetrahydrobiopterin) on our patients with phenylketonuria that have provided a better metabolic control and the possibility of freeing them from the special diet they were following. The monitoring of this treatment will be an important subject of study for the next three years. With respect to other disorders of the intermediate metabolism, mention should be made of our work on disorders of the metabolism of homocysteine and other amino acids, primarily addressed to characterising new disorders and new phenotypic presentations.*

### Members

#### Staff researcher

Roser Colomé Roura, Carme Fons Estupiña, Camila García Volpe, Rosa Gassió Subirachs, María Julieta González, Silvia Maria Meavilla Olivas

#### Assistant researcher

Pol Canal Noguer, Irene Galceran Nou, Leticia Diana Pias Peleteiro

### Selected projects

- Epilepsy and Mood Regulation Disorders: a Prospective and Longitudinal Study in Children With Newly Diagnosed Epilepsy. EPILETRE. Fundació La Caixa. EPILETRE. PI: Alexis Arzimanoglou / Jaume Campistol Plana. 2016 - 2018.
- SGR 2014\_Grup de recerca en neurociències i metabolisme pediàtric. Agaur - Agència de Gestió d'Ajuts Universitaris i de Recerca - SGR. 2014SGR1150. PI: Jaume Campistol Plana. 2014 - 2017.

### Selected clinical trials

- EMR 700773-002. Phase IV. Open single-cohort phase IV study of long-term neurocognitive (NC) results in children aged 4-5 years with phenylketonuria treated with sapropterin dihydrochloride (Kuvan®) for 7 years. EUDRACT: 2009-015844-41. Promoter: Merck KGaA. PI: Jaume Campistol Plana. 05/02/2014 -
- ALXN1101-MCD-202. Phase III. Open multi-centre multinational phase II/III study to assess the effectiveness and safety of ALXN1101 in newborns with type A molybdenum cofactor deficiency (MoCD). EUDRACT: 2013-002702-30. Promoter: Alexion Pharma Spain, S.L. PI: María Julieta González. 28/06/2016 -

### Selected publications

- Batllori-Tragant M, Molero M, Arrabal L, Heras JL, Fernandez-Ramos JA, Gutiérrez-Solana LG, et al. Urinary sulphatoxymelatonin as a biomarker of serotonin status in biogenic amine-deficient patients. Sci Rep 2017. 7: 14675-0. FI: 4,259(Q1).
- Mendes MI, Smith DE, Pop A, Lennertz P, Ojeda MR, Kanhai WA, et al. Clinically Distinct Phenotypes of Canavan Disease Correlate with Residual Aspartoacylase Enzyme Activity. Hum Mutat 2017. 38: 524-531. FI: 4,601(Q1).
- van Spronsen FJ, van Wegberg AM, Ahring K, Bélanger-Quintana A, Blau N, Bosch AM, et al. Key European guidelines for the diagnosis and management of patients with phenylketonuria. Lancet Diabetes Endocrinol 2017. 5: 743-756. FI: 19,742(Q1).
- Vidal-Falcó S, Brandi-Tarrau N, Pacheco P, Gerotina E, Blasco L, Trotta JR, et al. The utility of Next Generation Sequencing for molecular diagnostics in Rett syndrome. Sci Rep 2017. 7: 12288-0. FI: 4,259(Q1).

### Networks

- CIBERER
- TREAT-NMD



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PROJECTS



CLINICAL TRIALS  
& OBSERVATIONAL  
STUDIES



PUBLICATIONS  
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PUBLICATIONS

# MITOCHONDRIAL ENERGY METABOLISM AND DEVELOPMENT OF BIOMARKERS FOR THE INVESTIGATION OF INBORN ERRORS OF METABOLISM

Rafael Artuch Iriberry, MD, PhD

*We have implemented a metabolomic platform for the phenotyping of patients with genetically-based rare metabolic disorders, using animal and cellular models. This activity has been possible thanks to the acquisition of a UHPLC system coupled in tandem to a mass spectrometer, financed by the Daniel Bravo Foundation.*

## Members

### Staff researcher

Mercedes Casado Rio, Raquel Montero Sánchez, Cristina Sierra March

### Postdoc – associate researcher

Aida Ormazabal Herrero, Délia Yubero Siles

### Technician

Yahel Calzadilla Borras, Aroa Fernández de Miguel, Juan Moreno García, Tania Pardal Chofle

## Selected projects

- Mitochondrial encephalomyopathies and secondary deficiencies of Q10 coenzyme: studies on paediatric patients and on cellular and murine models.. Instituto de Salud Carlos III (ISCIII). FIS. PI14/00028. PI: Rafael Artuch Iriberry. 2015 - 2017.
- Study of trace elements [Cu, Zn, Se and Mn] in cerebrospinal fluid in paediatric patients and analysis of related biomarkers. Instituto de Salud Carlos III (ISCIII). FIS. PI14/00032. PI: Aida Ormazabal Herrero. 2015 - 2018.
- Contracts for intensification of research activity in the National Health System (SNS) 2015. Beneficiario: Dr. Rafael Artuch. Instituto de Salud Carlos III (ISCIII). Intensificación. INT16/00120. PI: Rafael Artuch Iriberry. 2017.

## Selected publications

- Batllori-Tragant M, Molero M, Arrabal L, Heras JL, Fernandez-Ramos JA, Gutiérrez-Solana LG, et al. Urinary sulphatoxymelatonin as a biomarker of serotonin status in biogenic amine-deficient patients. Sci Rep 2017. 7: 14675-0. FI: 4,259(Q1).

- Batllori-Tragant M, Molero M, Ormazabal-Herrero A, Casado M, Sierra-March C, Garcia-Cazorla A, et al. Analysis of human cerebrospinal fluid monoamines and their cofactors by HPLC. Nat Protoc 2017. 12: 2359-2375. FI: 10,032(Q1).
- Mancuso M, McFarland R, Klopstock T, Hirano M; consortium on Trial Readiness in Mitochondrial Myopathies. International Workshop: Outcome measures and clinical trial readiness in primary mitochondrial myopathies in children and adults. Consensus recommendations. Neuromuscul Disord. 2017;27(12):1126-1137. FI 2,487(Q3).
- Wassenberg T, Molero M, Jeltsch K, Hoffmann GF, Assmann B, Blau N, et al. Consensus guideline for the diagnosis and treatment of aromatic l-amino acid decarboxylase (AADC) deficiency. Orphanet J Rare Dis 2017. 12: 12-0. FI: 3,507(Q2).

## Networks

- CIBERER
- ERN (metabERN). European reference networks



PERSONAL RESEARCHER



PROJECTS



PUBLICATIONS WITH IF



FIRST QUARTILE PUBLICATIONS

## APPLIED RESEARCH ON NEUROMUSCULAR DISORDERS

Cecilia Jiménez Mallebrera, MD, PhD

We coordinate the national register (CIBERER Database of Neuromuscular Disorders) of patients with congenital myasthenias and with collagen VI deficiency. On the latter group of patients we are conducting studies of natural history (in collaboration with the HSJD Rehabilitation Service, Dr. J. Medina) and of prognostic factors (in collaboration with several international centres: Prof. C. Bonnemann, NINDS-NIH, Washington, USA; Dr. Nicholas Deconinck, Ghent University, Belgium).

In 2017 Dr. Daniel Natera obtained his PhD at the University of Barcelona for the study "Congenital myasthenic syndromes in the Spanish paediatric population: Clinical characterisation, genetics and therapeutic response", and in 2018 he obtained a Rio Ortega contract for the study of clinical and genetic aspects of congenital myopathies and congenital myasthenias (coordinated by Dr. Palau, IRSJD). In June 2017 we began a project in collaboration with the group of Profs. Daniel Grinberg and Susana Balcells (IBUB) for the purpose of editing dominant mutations in collagen VI genes by means of CRISPR/Cas9 technology and other gene editing methodologies. The project is funded by the Noelia Foundation for Collagen VI Deficiency Myopathies. In this project (in which Dr. Jimenez-Mallebrera and Mari Carmen Badosa participate) we also collaborate with the group of Dr. Francesc Palau and Janet Hoenicka (IRSJD) and with Dr. Mónica Roldán of the Confocal Microscopy Service of the Daniel Bravo Centre (IRSJD).

### Members

#### Staff researcher

Carlos Ignacio Orterz Gonzalez, Jaume Colomer

#### Postdoc – associate researcher

Daniel Natera de Benito

#### Assistant researcher

Maria del Carmen Badosa Gallego, Anna Lia Frongia, Irene Beatriz Zschaack Luzado, Anna Codina

#### PhD Student

Andres Nascimento Osorio, Cristina Jou Muñoz

#### Collaborator researcher

Pol Castellano Escuder, Susana Graciela Kalko Witruk

### Selected projects

- Application of proteomics and systems biology to the study of matrisoma and miRNAs in Congenital Muscular Dystrophies. Instituto de Salud Carlos III (ISCIII). Proyectos de investigación en salud. PI16/00579. PI: Cecilia Jiménez Mallebrera. 2017 - 2019.
- Miguel Servet tipo II - Beneficiary: Dra. Cecilia Jimenez. Instituto de Salud Carlos III (ISCIII). Miguel Servet II. CPII15/00020. PI: Cecilia Jiménez Mallebrera. 2016 - 2019.
- Myopathies associated with collagen VI deficiency: Natural history, functional assessment and identification of new diagnostic and prognostic markers. Fundación Andrés Marcio, niños contra la Laminopatia. PI: Cecilia Jiménez Mallebrera. 2015 - 2019.

### Selected clinical trials

- DSC/14/2357/48. Phase III: Randomised, double blind, placebo controlled, multicentre study to evaluate the efficacy and safety of givinostat in ambulant patients with Duchenne Muscular Dystrophy. EUDRACT: 2016-000401-36. Promoter: Italfarmaco,S.A. PI: Andrés Nascimento Osorio. 31/05/2017 -
- 4045-301. Phase III: A Double-Blind, Placebo-Controlled, Multicenter Study With an Open-Label Extension to Evaluate the Efficacy and Safety of SRP-4045 and SRP-4053 in Patients With Duchenne Muscular Dystrophy. EUDRACT: 2015-002069-52. Promoter: Sarepta Therapeutics, Inc. PI: Andrés Nascimento Osorio. 19/07/2017 -
- WN40227 [CN001016]. Phase III: A randomized, double blind, placebo-controlled, study to assess the efficacy, safety, and tolerability of R07239361 (BMS-986089) in ambulatory boys with Duchenne Muscular dystrophy. EUDRACT: 2016-001654-18. Promoter: F. Hoffmann- La Roche Ltd. PI: Andrés Nascimento Osorio. 28/09/2017 -
- with enterovirus-A71 in; Catalonia, Spain (2016): a clinical observational study in a children's; reference centre in Catalonia. Clin Microbiol Infect 2017. 23: 874-881. FI: 5,292(Q1).
- McDonald CM, Campbell C, Torricelli RE, Finkel RS, Flanigan KM, Goemans N, Heydemann P, et al. Ataluren in patients with nonsense mutation Duchenne muscular dystrophy (ACT DMD): a multicentre, randomised, double-blind, placebo-controlled, phase 3 trial. Lancet 2017. 390: 1489-1498. FI: 47,831(Q1).
- Rodríguez-García MA, Del Rio-Baquero LM, Ortez-Gonzalez CI, Jou-Munoz C, Vigo-Moranco M, Medina J, Febrer A, et al. Differences in Adipose Tissue and Lean Mass Distribution in Patients with Collagen VI Related Myopathies Are Associated with Disease Severity and Physical Ability.. Front Aging Neurosci 2017. 9: 268-0. FI: 4,504(Q1).
- Romero-Moya D, Santos-Ocaña C, Castaño J, Garrabou G, Rodríguez-Gómez JA, Ruiz-Bonilla V, et al. Genetic Rescue of Mitochondrial and Skeletal Muscle Impairment in an Induced Pluripotent Stem Cells Model of Coenzyme Q10 Deficiency. Stem Cells 2017. 35: 1687-1703. FI: 5,599(Q1).

### Selected publications

- Casas-Alba D, Fernández de Sevilla-Estrach M, Valero-Rello A, Fortuny-Guasch C, García-García JJ, Ortez-Gonzalez CI, Muchart-Lopez J, et al. Outbreak of brainstem encephalitis associated



PERSONAL  
RESEARCHER



PROJECTS



CLINICAL TRIALS & OBSERVATIONAL STUDIES



PUBLICATIONS  
WITH IF



FIRST QUARTILE  
PUBLICATIONS

## NEUROMETABOLIC DISORDERS AND SYNAPTIC STUDIES IN RARE NEUROGENETIC CONDITIONS

M<sup>a</sup> Àngels García Cazorla, MD, PhD

In 2017 we consolidated the study models available to us at the laboratory (cellular models for the study of tyrosine hydroxylase deficiency and Rett syndrome) and we developed new techniques for the study of brain function (connectivity). We have had a very active participation in the European networks, which took shape in the coordination of a disorders subgroup (neurotransmitter defects) in the metabERN network. Likewise, we have made a significant contribution to the development of clinical guides and to the description of a new cause of early Parkinsonism and of new molecular aspects of known neurometabolic disorders.

### Members

#### Postdoctoral researcher

Alfonso Luis De Oyarzabal Sanz

#### Staff researchers

Anna López Sala, M<sup>a</sup> del Mar O'Callaghan Gordo,

Federico Ramos

#### PhD Students

Elisenda Cortès-Saladelafont, Alba Tristan Noguero

#### Assistant researcher

Inés Fernanda Medina Rivera

### Selected projects

- Neurotransmission systems in paediatric encephalopathies of genetic origin: identification of new molecular and pathophysiological aspects. Instituto de Salud Carlos III (ISCIII). PI15/01082. FIS. PI: M<sup>a</sup> Àngels García Cazorla. 2016 - 2018.
- Therapeutic approaches in a cellular model of Rett syndrome. OSSJD - Obra Social Sant Joan de Déu. Mi Princesa Rett. PI: M<sup>a</sup> Àngels García Cazorla. 2014 - 2020.
- Brain mapping and advanced therapies in childhood neurological disorders. PepsiCo Iberia. PI: M<sup>a</sup> Àngels García Cazorla. 2016 - 2019.

### Selected clinical trials

- UX007G-CL201. Phase II. A double-blind placebo-controlled clinical trial of parallel groups to assess the safety and effectiveness of UX007 in subjects with glucose transporter type 1 deficiency syndrome. Codi EUDRACT: 2013-003771-35. PI: M<sup>a</sup> Àngels García Cazorla. 07/01/2016 -
- UX007G-CL202. Phase II. Open extension study to assess the long-term safety and effectiveness of UX007 in subjects with glucose transporter type 1 deficiency syndrome (Glut1 DS). EUDRACT: 2015-000389-69. Promoter: Ultragenyx Pharmaceutical, Inc. PI: M<sup>a</sup> Àngels García Cazorla. 05/06/2017 -

### Selected publications

- Batllori-Tragant M, Molero M, Ormazabal-Herrero A, Casado M, Sierra-March C, Garcia-Cazorla A, et al. Analysis of human cerebrospinal fluid monoamines and their cofactors by HPLC. Nat Protoc 2017. 12: 2359-2375. FI: 10,032(Q1).
- Bravo-Alonso I, Navarrete R, Arribas-Carreira L, Perona A, Abia D, Couce ML, et al. Nonketotic hyperglycinemia: Functional assessment of missense variants; in GLDC to understand phenotypes of the disease. Hum Mutat 2017. 38: 678-691. FI: 4,601(Q1).
- Díez H, Cortès-Saladelafont E, Ormazábal A, Marmiese AF, Armstrong J, Matalonga L, et al. Severe infantile parkinsonism because of a de novo mutation on DLP1 mitochondrial-peroxisomal protein. Mov Disord. 2017 Jul;32(7):1108-1110. FI: 7,072 (Q1).
- Vidal-Falcó S, Brandi-Tarau N, Pacheco P, Gerotina E, Blasco L, Trotta JR, et al. The utility of Next Generation Sequencing for molecular diagnostics in; Rett syndrome. Sci Rep 2017. 7: 12288-0. FI: 4,259(Q1).

### Networks

- 2007 - CIBERER
- 2017 - European Reference Network for Hereditary Metabolic Diseases (METABERN)
- 2014 - Connecting the growing brain
- 2015 - INTD - International Working Group on Neurotransmitter Related Disorders



PERSONAL RESEARCHER



PROJECTS



CLINICAL TRIALS & OBSERVATIONAL STUDIES



PUBLICATIONS WITH IF



FIRST QUARTILE PUBLICATIONS

## MOVEMENT DISORDERS OF GENETIC AND NEUROMETABOLIC ORIGIN

Belén Pérez Dueñas, MD, PhD

In 2017, our group published 11 articles with a total impact factor of 66.743; it was engaged in 13 active projects (including 2 FIS projects, 1 TV3 Marathon grant, and 1 Rio Hortega grant, among others) and the reading of one doctoral thesis (doctoral candidate Juan D. Ortigoza-Escobar). Dr. Serrano led a clinical trial (Phase II) on acetazolamide in patients with PMM2-congenital disorder of glycosylation. The unit also belongs to several national research networks (CSUR) and European ones (ERN-NMD).

### Members

#### Staff researchers

Jordi Muchart López, Mercedes Serrano Gimaré

#### Postdoc – associate researcher

Alejandra Darling, Juan Darío Ortigoza Escobar

#### PhD Students

Laura Martín Sanchez, María Isabel Vanegas Grisales

### Selected projects

- Neurodegeneration with Brain Iron Accumulation: Clinical Assessment and Genetic Characterization by means of a Spanish Multi-Centre Research Network. Fundació La Marató de TV3. Investigación enfermedades neurodegenerativas. Marató TV3 - 20143130. PI: Belén Pérez Dueñas. 2015 - 2018.
- Clinical and radiological quantitative assessment in PMM2-CDG patients through a national network of professionals. Instituto de Salud Carlos III (ISCIII). FIS. PI14/00021. PI: Mercedes Serrano Gimaré. 2015 - 2017.
- Biomarkers and genes in infantile bilateral striatal necrosis. Instituto de Salud Carlos III (ISCIII). FIS. PI15/00287. PI: Belén Pérez Dueñas. 2016 - 2018.

### Selected clinical trials

- FSJD-PMM2-2016. Phase II clinical trial to evaluate the effectiveness and safety of acetazolamide in the treatment of cerebellar syndrome in patients with PMM2-CDG deficiency. EUDRACT: 2017-000810-44. Promoter: Fundació Privada per a la Recerca i la Docència Sant Joan de Déu - FSJD FSJD. PI: Mercedes Serrano Gimaré. 2017 -

### Selected publications

- Meyer E, Carss KJ, Rankin J, Nichols JM, Grozeva D, Joseph AP, et al. Mutations in the histone methyltransferase gene KMT2B cause complex early-onset dystonia. Nat Genet 2017. 49: 223-237. FI: 27,959(Q1).

- Ortigoza-Escobar JD, Alfadhel M, Molero M, Darin N, Spiegel R, de Coo IF, et al. Thiamine deficiency in childhood with attention to genetic causes: Survival and outcome predictors. Ann Neurol 2017. 82: 317-330. FI: 9,890(Q1).
- Darling A, Tello C, Martí MJ, Garrido C, Aguilera-Albesa S, Tomás Vila M, et al. Clinical rating scale for pantothenate kinase-associated neurodegeneration: A pilot study. Mov Disord 2017. 32: 1620-1630. FI: 7,072(Q1).
- Casas-Alba D, Fernández de Sevilla-Estrach M, Valero-Rello A, Fortuny-Guasch C, García-García JJ, Ortez-Gonzalez CI, et al. Outbreak of brainstem encephalitis associated with enterovirus-A71 in Catalonia, Spain [2016]: a clinical observational study in a children's reference centre in Catalonia. Clin Microbiol Infect 2017. 23: 874-881. FI: 5,292(Q1).

### Networks

- CIBERER
- TREAT-NMD
- Unitat integrada de referència del sistema nacional de salut en "enfermedades raras que cursan con trastornos del movimiento (csur\_53)
- "Health care provider" European Reference Network on Rare Neurologic Diseases



## NEUROGENETICS AND MOLECULAR MEDICINE

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

The scientific and translational endeavours of the Neurogenetics and Molecular Medicine research group (NeuroGene) unfold in the field of experimental biomedicine on neurogenetic and neuropsychiatric disorders, specifically Charcot-Marie-Tooth neuropathy (CMT), Friedreich's ataxia (FRDA), Parkinson's disease (PD) and neurodevelopmental disorders, as well as in the clinical field of the Paediatric Institute for Rare Diseases (IPER). The scientific landmarks achieved in 2017 notably include: (1) determination of the role of ANKK1 protein in the cell cycle during neurodevelopment (España-Serrano et al., Cereb Cortex 2017); (2) determination of the differential effects on calcium homeostasis of recessive GDAP1 mutations vs. dominant mutations in CMT neuropathy (González-Sánchez et al., Sci Rep 2017); and (3) application of NGS mass sequencing in the genetic diagnosis and study of a long series of patients with Rett or Rett-type syndrome (Vidal et al., Sci Rep 2017). In December 2017, F. Palau was proposed by France's INSERM as editor-in-chief of the Orphanet Journal of Rare Diseases.

### Members

#### Staff researchers

Judith Silvia Armstrong Morón

#### Postdoc – associate researcher

Lara Cantarero Abad, Alba Aina Castells Santamaría, Laura Domínguez Berzosa, Loreto Martorell Sampol, Mª del Mar Pérez Iribarne, Isabel Plensa Nebot

#### Assistant researcher

Cesar Arjona Fernandez, Núria Brandi Tarrau, Antonio Martínez Monseny, Mireia Osuna López, Ainhoa Pascual Alonso, Judith Reina Castillón, Montserrat Roura Llerda, Silvia Vidal Falcó

#### PhD Student

Azahara Civera Tregon, Elena Juárez Escoto, Abel Sola Bacaicoa

#### Technician

Daniel Castillo Mestres, Jordi Genovès Escarré, Dolores López López, Montserrat Naudó Lahoz, Paola Pacheco Fernández, Johanna Troya Balseca

### Selected projects

- Clinical and molecular characterisation of Rett syndrome: elucidation of unsolved cases. Instituto de Salud Carlos III (ISCIII). FIS. PI15/01159. PI: Judith Armstrong Morón. 2016 -2018.
- Role of ANKK1 in the genetics and pathophysiology of Parkinson's disease. Instituto de Salud Carlos III (ISCIII). FIS. PI15/01013. PI: Janet Hoenicka. 2016 - 2018.
- The landscape of axonal biology and mitochondria-associated membranes in neurogenetic disorders. Ministerio de Economía y Competitividad (MINECO). Retos colaboración 2015. SAF2015-66625-R. PI: Francesc Palau Martínez. 2016 - 2019.

### Selected publications

- España-Serrano L, Guerra Martín-Palanco N, Montero-Pedrazuela A, Pérez-Santamarina E, Vidal R, García-Conseguera I, et al. The Addiction-Related Protein ANKK1 is Differentially Expressed During the Cell Cycle in Neural Precursors.. Cereb Cortex 2017. 27: 2809-2819. FI: 6,559(Q1).

- González-Sánchez P, Pla-Martín D, Martínez-Valero P, Rueda CB, Calpena E, Del Arco A, et al. CMT-linked loss-of-function mutations in GDAP1 impair store-operated Ca(2+) entry-stimulated respiration. Sci Rep 2017. 7: 42993-0. FI: 4,259(Q1).
- Mollá B, Muñoz-Lasso DC, Riveiro F, Bolinches-Amorós A, Pallardó FV, Fernandez-Vilata A, et al. Reversible Axonal Dystrophy by Calcium Modulation in Frataxin-Deficient Sensory Neurons of YG8R Mice. Front Mol Neurosci 2017. 10: 264-0. FI: 5,076(Q1).
- Vidal-Falcó S, Brandi-Tarrau N, Pacheco P, Gerotina E, Blasco L, Trotta JR, et al. The utility of Next Generation Sequencing for molecular diagnostics in; Rett syndrome. Sci Rep 2017. 7: 12288-0. FI: 4,259(Q1).

### Networks

- CIBERER
- CIBERSAM
- TREAT-NMD
- ACTION - Data and Policies for Rare Diseases
- European Reference Network for Rare Neuromuscular Diseases
- Orphanet-Spain



PERSONAL  
RESEARCHER

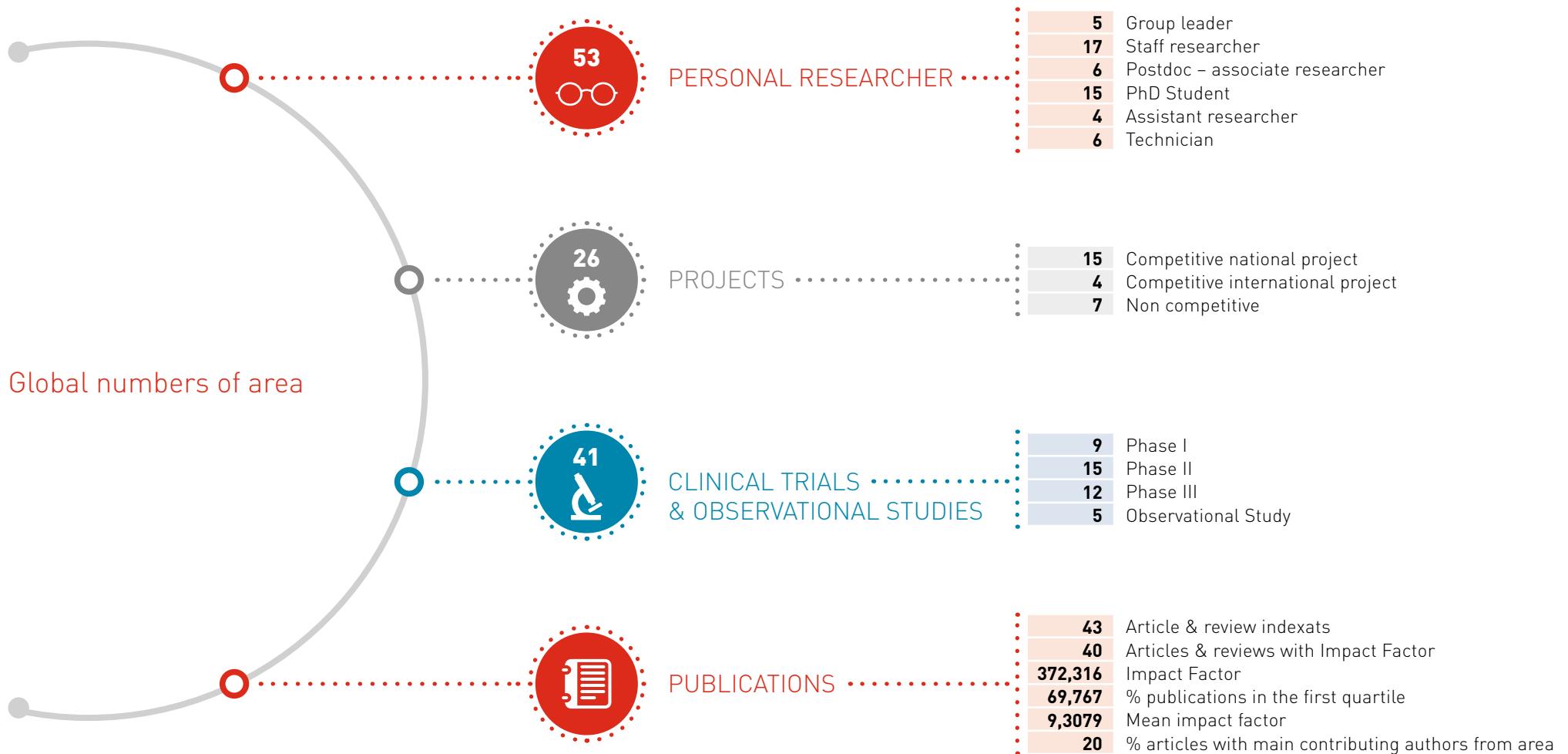
PROJECTS

PUBLICATIONS  
WITH IF

FIRST QUARTILE  
PUBLICATIONS

## 3.2. Molecular and cellular biology of developmental tumour

**Coordinator: Jaume Mora**



## PRECLINICAL PHARMACOLOGY AND DRUG RELEASE IN SOLID PAEDIATRICS TUMOURS

Angel Montero Carcaboso, PhD

A treatment of retinoblastoma with the VCN-01 oncolytic adenovirus reached the clinical phase (phase 1 trial) at Sant Joan de Déu Barcelona Children's Hospital after carrying out all the preclinical phases at our laboratory. We thus achieved a goal pursued over the last 4 years. Part of the results of another two 4-year projects were published in 2017. In the first paper we described a new strategy for studying the intratumoural distribution of drugs targeting paediatric cancer and its application in neuroblastoma. The results were published in the Journal of Controlled Release. In the second study, which was published in the same journal, we showed that the barrier between the blood and the retina works by restricting the entry of drugs into the retinoblastoma tumour and can be pharmacologically modulated to counteract this effect.

### Members

#### Staff researchers

Guillermo Chantada, Ofelia Cruz Martínez,  
Andrés Morales La Madrid

#### Postdoc – associate researcher

Sonia Paco Mercader

#### PhD Student

Nagore Gene Olaciregui, Guillem Pascual Pastó

#### Assistant researcher

Helena Castillo Ecija, María Cuadrado Vilanova,  
Alejandro Manzanares

#### Technician

Mònica Vilà Ubach

### Selected projects

- ITCC-P4\_ITCC Pediatric Preclinical POC Platform. European Commission - H2020-JTI-IMI2-2015-07-two-stage. PI: Angel Montero Carcaboso. 2017 - 2021.
- Cure4RB. Ministerio de Economía Y Competitividad (MINECO) - Retos colaboración 2015. RTC-2015-4319-1. PI: Angel Montero Carcaboso. 2015 - 2018.
- Immune therapy of diffuse intrinsic pontine glioma (DIPG). Instituto de Salud Carlos III (ISCIII) - FIS. PI15/01161. PI: Angel Montero Carcaboso. 2016 - 2018.

### Selected clinical trials

- FSJD-RTB-2015. Phase I. Single-centre, open phase I clinical trial with escalating dose to assess the safety and activity of VCN-01 oncolytic adenovirus in patients with refractory retinoblastoma. EUDRACT: 2016-001060-11. Promoter: Fundació Sant Joan de Déu. PI: Jaume Catalá Mora. 03/08/2017 -
- FSJD-DIPG-DC. Phase I. Phase Ib clinical trial on the safety of immune therapy with autologous dendritic cells pulsed with lysate of allogenic tumour cell lines in patients with diffuse intrinsic pontine glioma (DIPG). EUDRACT: 2015-003362-84. Promoter: Fundació Sant Joan de Déu. PI: Andrés Morales La Madrid. 06/06/2016 -

### Selected publications

- Mohammad F, Weissmann S, Leblanc B, Pandey DP, Höjfeldt JW, Comet I, et al. EZH2 is a potential therapeutic target for H3K27M-mutant pediatric gliomas. *Nat Med* 2017; 23: 483-492. FI: 29,886(Q1).
- Monterrubio C, Paco-Mercader S, Gene-Olaciregui N, Pascual-Pastó G, Vilà-Ubach M, Cuadrado-Vilanova M, et al. Targeted drug distribution in tumor extracellular fluid of GD2-expressing neuroblastoma patient-derived xenografts using SN-38-loaded nanoparticles conjugated to the monoclonal antibody 3F8. *J Control Release* 2017; 255: 108-119. FI: 7,786(Q1).
- Pascual-Pastó G, Gene-Olaciregui N, Opezzo JAW, Castillo-Ecija H, Cuadrado-Vilanova M, Paco-Mercader S, et al. Increased delivery of chemotherapy to the vitreous by inhibition of the blood-retinal barrier. *J Control Release* 2017; 264: 34-44. FI: 7,786(Q1).

### Networks

- Innovative Therapies for Children with Cancer



## LEUKEMIA AND NON-MALIGNANT BLOOD DISORDERS

Mireia Camós Guijosa, MD, PhD

In 2017 the group combined an intense clinical research activity with translational research. Dr. Susana Rives continued the clinical trials involving cell therapy with T CAR19 lymphocytes, one with the pharmaceutical industry and the other academic. Our centre was the first in Spain to administer this therapy and the one that recruited the largest number of patients in Europe, and in 2017 it passed the inspection of the U.S. Food and Drug Administration (FDA). The results of this trial were published in the New England Journal of Medicine. In the translational field, this year we obtained several competitive grants, including the FIS grant of Dr. Berrueco, the first in the line of haemostasis, which seeks to reduce the risk of thrombosis in patients with acute leukaemia. In 2017 Dr. Albert Català defended his thesis and is now training in Canada where he is carrying out a project on bone marrow failures.

### Members

#### Staff researchers

Isabel Badell Serra, Ruben Berrueco Moreno, Albert Català Temprano, Susanna Gassiot Riu, Julia Marsal Ricoma, Susana Rives Solà, Anna Ruiz Llobet, Montserrat Torrebadell Burriel

#### PhD Student

Anna Alonso Saladrigues, Elena Esperanza Cebollada, Montserrat Mesegue Meda, Sandra Pont Martí, María Trabazo del Castillo, Nerea Vega García

#### Technician

Maria Camino Estella Aguado, Lidia Ruiz Bocanegra

### Selected projects

- Prognostic impact of genetic mutations in patients with acute childhood lymphoblastic leukaemia treated by the SEHOP-PETHEMA 2013 protocol: a cooperative project of the biological studies committee of the leukaemias group of the Spanish Society for Paediatric Haematology and Oncology (SEHOP). Fundación Uno entre Cien Mil - III Beca Fundación Uno entre cien mil. PI: Mireia Camós Guijosa. 2016 - 2018.
- Pilot study on the infusion of autologous T lymphocytes genetically modified to express anti-CD19 in patients with chemotherapy resistant or refractory CD19+ leukaemia or lymphoma. Instituto de Salud Carlos III (ISCIII) - PICI. ICI14/00224. PI: Susana Rives Solà. 2015 - 2018.

- Epigenetic regulators in patients with high-risk paediatric leukaemia: the role of the HDAC family and microRNAs as prognostic risk biomarkers and therapeutic targets. Instituto de Salud Carlos III (ISCIII) - Proyectos de investigación en salud. PI16/00246. PI: Mireia Camós Guijosa. 2017 - 2019.

### Selected clinical trials

- CCTL019B2001X . Phase II. CTL019 expanded treatment protocol for paediatric/young adult patients with relapsing/refractory acute lymphoblastic leukaemia. EUDRACT: 2016-001991-31. Promoter: Novartis Farmacéutica, S.A. PI: Susana Rives Solà. 21/04/2017 -
- NOPHO-DBH AML 2012 . Fase III. Research study for treatment of children and adolescents with acute myeloid leukaemia 0-18 years. EUDRACT: 2012-002934-35. Promoter: Sociedad Española de Hematología y Oncología Pediátricas. PI: Susana Rives Solà. 24/08/2017 -
- CART19-BE-01. Phase I. Pilot study of the infusion of autologous T lymphocytes genetically modified to express anti-CD19 in patients with chemotherapy resistant or refractory CD19+ leukaemia or lymphoma. EUDRACT: 2016-002972-29. Promoter: Hospital Clínic Barcelona. PI: Susana Rives Solà. 04/07/2017 -

### Selected publications

- Borràs N, Batlle J, Pérez-Rodríguez A, López-Fernández MF, Rodríguez-Trillo Á, Lourés E, et al. Molecular and clinical profile of von Willebrand disease in Spain [PCM-EVW-ES]: comprehensive genetic analysis by next-generation sequencing of 480 patients. Haematologica 2017. 102: 2005-2014. FI: 7,702(Q1).
- Forero-Castro M, Robledo C, Benito R, Bodegas-Mayor I, Rapado I, Hernández-Sánchez M, et al. Mutations in TP53 and JAK2 are independent prognostic biomarkers in B-cell precursor acute lymphoblastic leukaemia. Br J Cancer 2017. 117: 256-265. FI: 6,176(Q1).
- Lipka DB, Witte T, Toth R, Yang J, Wiesenfarth M, Nollke P, et al. RAS-pathway mutation patterns define epigenetic subclasses in juvenile myelomonocytic leukemia. Nat Commun 2017. 8: 2126-0. FI: 12,124(Q1).
- Robles-Valero J, Lorenzo-Martín LF, Menacho-Márquez M, Fernández-Pisonero I, Abad A, Camós-Guijosa M, et al. A Paradoxical Tumor-Suppressor Role for the Rac1 Exchange Factor Vav1 in T Cell Acute Lymphoblastic Leukemia. Cancer Cell 2017. 32: 608-6239. FI: 27,407(Q1).

### Networks

- Innovative Therapies for Children with Cancer



PERSONAL RESEARCHER



PROJECTS



CLINICAL TRIALS & OBSERVATIONAL STUDIES



PUBLICATIONS WITH IF



FIRST QUARTILE PUBLICATIONS

## NEUROBLASTOMA

Carmen de Torres Gómez-Pallete, MD, PhD

*The group continued the study of the pathway of the calcium sensor receptor and PTHrP as potential therapeutic tools in neuroblastoma. Financing was achieved in 2017 for a European project for the study of nanosensors as detection systems for proteins excreted to the environment using the PTHrP model in neuroblastoma as a study model. The group continued to form part of the European network for the study of CasR, which includes the training of researchers shared by various laboratories.*

### Members

#### Postdoc – associate researcher

Marta García López, Silvia Mateo Lozano, Carlos Javier Rodriguez Hernandez

#### PhD Student

Eliana Carolina Gonçalves Alves

#### Technician

Noelia Salvador Marcos

### Selected projects

- CaSR\_Marie Curie International Training Network - CaSR Biomedicine\_Calcium-Sensing Receptor Therapeutics for Non-Communicable Diseases. Beneficiary: Dra. Carmen de Torres. European Commission - MSCA-ITN 2015. MSCA-ITN 2015. PI: Carmen De Torres Gómez-Pallete. 01/03/2016 - 29/02/2020.
- Assessment of the sensor receptor of calcium and parathyroid hormone-related protein as new therapeutic targets in neuroblastoma. Instituto de Salud Carlos III (ISCIII) - FIS. PI14/00040. PI: Carmen De Torres Gómez-Pallete. 01/01/2015 - 31/12/2017.
- NACANCELL\_Development of a Nanodiagnostic platform for monitoring of Cancer cell secreted proteins. Instituto de Salud Carlos III (ISCIII) - Acciones complementarias. AC16/00026. PI: Carmen De Torres Gómez-Pallete. 01/01/2017 - 31/12/2019.

### Selected publications

- Lassaletta A, Zapotocky M, Mistry M, Ramaswamy V, Honnorat M, Krishnatry R, et al. Therapeutic and Prognostic Implications of BRAF V600E in Pediatric Low-Grade Gliomas. J Clin Oncol 2017; 35: 2934-2941. FI: 24,008(Q1).

- Mackay A, Burford A, Carvalho D, Izquierdo E, Fazal-Salom J, Taylor KR, et al. Integrated Molecular Meta-Analysis of 1,000 Pediatric High-Grade and Diffuse Intrinsic Pontine Glioma. Cancer Cell 2017; 32 : 520-537.e5. FI: 27,407(Q1).
- Mohammad F, Weissmann S, Leblanc B, Pandey DP, Højfeldt JW, Comet I, et al. EZH2 is a potential therapeutic target for H3K27M-mutant pediatric gliomas. Nat Med 2017; 23: 483-492. FI: 29,886(Q1).
- Monterrubio C, Paco-Mercader S, Gene-Olaciregui N, Pascual-Pastó G, Vilà-Ubach M, Cuadrado-Vilanova M, et al. Targeted drug distribution in tumor extracellular fluid of GD2-expressing neuroblastoma patient-derived xenografts using SN-38-loaded nanoparticles conjugated to the monoclonal antibody 3F8. J Control Release 2017; 255: 108-119. FI: 7,786(Q1).

### Networks

- CaSR\_Marie Curie International Training Network - CaSR Biomedicine\_Calcium-Sensing Receptor Therapeutics for Non-Communicable Diseases.
- 2016 - CaSR\_Marie Curie International Training Network - CaSR Biomedicine\_Calcium-Sensing Receptor Therapeutics for Non-Communicable Diseases. MSCA-ITN 2015.
- 2016 - NACANCELL\_Development of a Nanodiagnostic platform for monitoring of cancer cell secreted proteins. Euronanomed 2.



PERSONAL  
RESEARCHER



PROJECTS



PUBLICATIONS  
WITH IF



FIRST QUARTILE  
PUBLICATIONS

## SARCOMA AND HISTIOCYTOSIS

Jaume Mora Graupera, MD, PhD

2017 saw the publication of the original study of the GEIS 21 trial on the first multi-institutional clinical trial on both adult and paediatric patients of the GEIS group on Ewing's sarcoma directed at our centre. This study was presented at the ASCO Annual Meeting in 2016 and it formed the basis of the GEIS group for the study and development of new Ewing's sarcoma treatments. The new clinical trial of the GEIS group on Ewing's sarcoma, GEIS 39, began in 2017, following up the findings of our laboratory on the activity of nab-paclitaxel in these tumours.

The treatment of patients with refractory and relapsing neuroblastoma began at our centre in June of 2017 using the Hu3F8 antibody for the first time outside MSKCC, thanks to the relation established with the company Ymabs, which was founded by the father of a neuroblastoma survivor patient whom we followed up at our centre. Ymabs has acquired all the rights on the antibodies developed at MSKCC for the treatment of paediatric tumours with the aim to register them and make them commercially available. The clinical trials of these new treatments will be conducted jointly at MSKCC and at our centre. Because of this plan, many patients from around the world have begun to come to our centre to receive these unique treatments.

### Members

#### Staff researcher

Verónica Celis Passini, Moira Garraus Oneca, María Inmaculada Hernandez Muñoz, Vicente Santa-María, Alicia Castañeda Heredia

#### Postdoc – associate researcher

Sara Sánchez Molina

#### Assistant researcher

Elisabet Figuerola Bou

#### PhD Student

Estela Prada Varela, Alejandro Manzanares Quintela,

Sara Pérez Jaume

#### Technician

Maria Jesus Nagel Moyano

### Selected projects

- Characterization and relevance of the epigenetic environment in the development of Ewing sarcoma. Instituto de Salud Carlos III (ISCIII). Proyectos de investigación en salud. PI16/00245. PI: Jaume Mora Graupera. 2017 - 2019.
- Fundación Leo Messi\_Convenio Rhabdomyosarcoma. Fundación Privada Leo Messi. PI: Jaume Mora Graupera. 2016 - 2019.
- SGR 2014\_Research Group on Developmental Tumors. Agaur - Agència de Gestió d'Ajuts Universitaris i de Recerca. SRG. 2014SGR1398. PI: Jaume Mora Graupera. 2014 - 2017.

### Selected clinical trials

- GEIS-39. Phase II trial of nab-paclitaxel for the treatment of desmoid tumors and multiply relapsed/refractory desmoplastic small round cell tumors and Ewing sarcoma. EUDRACT: 2016-002464-14. Promoter: Grupo Español de Investigación en Sarcoma Grupo GEIS. PI: Jaume Mora Graupera. 18/05/2017 -
- HSJD-HR-NB-Ch14.18. Phase II single arm institutional study to assess Dinutuximab combined with the cytokines granulocyte-macrophage colony stimulating factor (GM-CSF) and IL-2 in patients with high-risk neuroblastoma not eligible to other immunotherapy trials. EUDRACT: 2013-004864-69. Promoter: Fundació Privada per a la Recerca i la Docència Sant Joan de Déu - FSJD FSJD. PI: Jaume Mora Graupera. 26/05/2014 -
- TRA-130/GEIS-21. Phase II: Multicenter, prospective uncontrolled phase II clinical trial of intensive chemotherapy, surgery and radiotherapy in tumors of the Ewing sarcoma family in young children and adults. EUDRACT: 2009-016027-62. Promoter: Grupo Español de Investigación en Sarcoma Grupo GEIS. PI: Jaume Mora Graupera. 01/01/2010 -

### Selected publications

- Mackay A, Burford A, Carvalho D, Izquierdo E, Fazal-Salom J, Taylor KR, et al. Integrated Molecular

Meta-Analysis of 1,000 Pediatric High-Grade and Diffuse Intrinsic Pontine Glioma. Cancer Cell 2017. 32: 520-537. FI: 27,407(Q1).

Mohammad F, Weissmann S, Leblanc B, Pandey DP, Højfeldt JW, Comet I, et al. EZH2 is a potential therapeutic target for H3K27M-mutant pediatric gliomas. Nat Med 2017. 23: 483-492. FI: 29,886(Q1).

Monterrubio C, Paco-Mercader S, Gene-Olaciregui N, Pascual-Pastó G, Vilà-Ubach M, Cuadrado-Vilanova M, et al. Targeted drug distribution in tumor extracellular fluid of GD2-expressing neuroblastoma patient-derived xenografts using SN-38-loaded nanoparticles conjugated to the monoclonal antibody 3F8. J Control Release 2017. 255: 108-119. FI: 7,786(Q1).

Mora J, Castañeda A, Pérez-Jaume S, Lopez-Pousa A, Maradiegue E, Valverde C, et al. GEIS-21: a multicentric phase II study of intensive chemotherapy including gemcitabine and docetaxel for the treatment of Ewing sarcoma of children and adults: a report from the Spanish sarcoma group (GEIS). Br J Cancer 2017. 117: 767-774. FI: 6,176(Q1).

### Networks

- Innovative Therapies for Children with Cancer

12  
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PERSONAL RESEARCHER

6  
GE

PROJECTS

16  
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CLINICAL TRIALS & OBSERVATIONAL STUDIES

14  
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PUBLICATIONS WITH IF

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FIRST QUARTILE PUBLICATIONS

## TRANSLATIONAL GENOMICS

Cinzia Emilia Lavarino, MD, PhD

*Cinzia Lavarino's research group developed a method that allows the quick, simple and precise classification of patients with medulloblastoma, a malignant tumour of the cerebellum. This method is based on the analysis of a small panel of epigenetic markers specific to each of the four main types of medulloblastoma (wingless, sonic hedgehog, group 3 and group 4), which are characterised respectively by different clinical behaviours and genetics. This biomarker panel has been developed and tested using databases of over 1,500 samples. The value of this classification method lies in its applicability to small tumoural biopsies (frozen or preserved), as well as its speed, reproducibility of results, and low financial cost.*

### Members

#### Staff researchers

Héctor Salvador Hernández

#### Postdoc – associate researcher

Soledad Gomez Gonzalez.

#### PhD Student

Laura García Gerique, Alicia Garrido García

#### Technician

Isadora Lemos

### Selected projects

- Epigenetics in neuroblastoma pathogenesis: new molecular perspectives for the development of therapeutic strategies. Instituto de Salud Carlos III (ISCIII) - FIS. PI14/00038. PI: Cinzia Emilia Lavarino. 2015 - 2018.
- Neuroblastoma NEN. Associació de familiars i amics de pacients amb neuroblastoma "cons.". PI: Cinzia Emilia Lavarino. 2012 -

### Selected observational studies

- Observational study. Creación de una cohorte española de supervivientes de cáncer infantil: cohorte SPAIN-CCSS. Promoter: Fundació Privada ISGLOBAL. PI: Héctor Salvador Hernández. 2014 -

### Selected publications

- Achatz MI, Porter CC, Brugières L, Druker H, Frebourg T, Foulkes WD, et al. Cancer Screening Recommendations and Clinical Management of Inherited Gastrointestinal Cancer Syndromes in Childhood. Clin Cancer Res 2017;23:e107-e114. FI: 9,619(Q1).
- Evans DGR, Salvador-Hernandez H, Chang VY, Erez A, Voss SD, Druker H, et al. Cancer and Central Nervous System Tumor Surveillance in Pediatric Neurofibromatosis 2 and Related Disorders. Clin Cancer Res 2017; 23: 54-61. FI: 9,619(Q1).
- Evans DGR, Salvador-Hernandez H, Chang VY, Erez A, Voss SD, Schneider KW, et al. Cancer and Central Nervous System Tumor Surveillance in Pediatric Neurofibromatosis 1. Clin Cancer Res 2017; 23: 46-53. FI: 9,619(Q1).
- Mohammad F, Weissmann S, Leblanc B, Pandey DP, Höjfeldt JW, Comet I, et al. EZH2 is a potential therapeutic target for H3K27M-mutant pediatric gliomas. Nat Med 2017; 23: 483-492. FI: 29,886(Q1).



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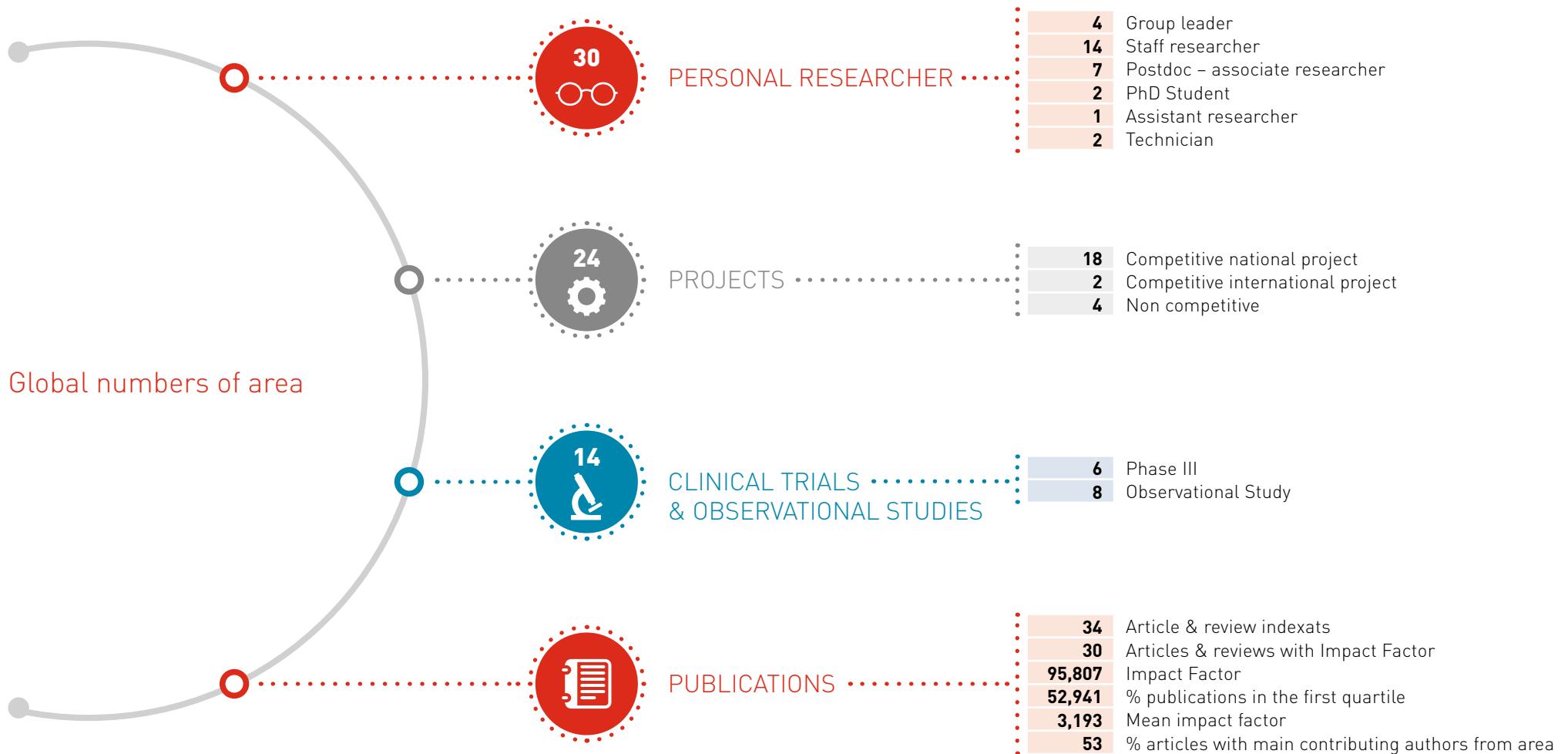
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### 3.3. Adult-age disorders originating in the foetal period or in the early years of life

**Coordinator: Lourdes Ibañez**



## METABOLIC ENDOCRINOLOGY

Lourdes Ibañez Toda, MD, PhD

*Foetal growth restriction may have irreversible consequences. The study of the various stages from intrauterine life to adolescence will allow us to advance in the prevention of this condition and in the treatment and prevention of the complications derived from it.*

### Members

#### Postdoc – associate researcher

Judith Bassols Casadevall, Paula Casano Sancho, Francis de Zegher, Marta Díaz Silva, Abel López Bermejo, Rita Maria Martins de Sousa Maia Malpique, Giorgia Sebastiani

#### Assistant researcher

Esther Lizarraga Mollinedo

#### PhD Student

Cristina García Beltran

### Selected projects

- Genetic determinants of prenatal and postnatal growth: association with early markers of cardiovascular risk and metabolic syndrome. Instituto de Salud Carlos III (ISCIII). Proyectos FIS-2011. PI11/02403. PI: Lourdes Ibañez Toda. 2012 - 2017.
- Pioglitazone+spironolactone+metformin versus oral contraception in adolescents with ovarian hyperandrogenism and hyperinsulinism. Instituto de Salud Carlos III (ISCIII). FIS. PI15/01078. PI: Lourdes Ibañez Toda. 2016 - 2018.
- Development of a new combination of insulin sensitizers and antiandrogens for the treatment of polycystic ovary syndrome in adolescent and young-adult women. Ministerio de Economía Y Competitividad (MINECO). Convocatoria Retos Colaboración 2016. RTC-2016-5671-1. PI: Lourdes Ibañez Toda. 2016 - 2019.

### Selected clinical trials

- CP-4-006. Fase III. A phase 3, open-label, randomized, multicenter, 12 months, efficacy and safety study of weekly MOD-4023 compared to daily Genotropin - therapy in pre-pubertal children with growth hormone deficiency. EUDRACT: 2016-003874-42. Promoter: OPKO Biologics Ltd. PI: Lourdes Ibañez Toda. 10/11/2017 -
- FSJD-PIOSPIMET-2015. Phase III. Prospective open randomised clinical trial with two groups of patients to assess the effectiveness and safety of the combination of ethynodiol diacetate and levonorgestrel versus the combination of pioglitazone, spironolactone and metformin in low doses in adolescent women with ovarian hyperandrogenism and hyperinsulinism: effects on the ovulatory function, chronic inflammation parameters, and markers of evolution and treatment effectiveness, and of type 2 diabetes development. EUDRACT: 2015-005092-24. Promoter: Fundació Privada per a la Recerca i la Docència Sant Joan de Déu - FSJD FSJDPI: Lourdes Ibañez Toda. 11/02/2016 -

### Selected publications

- de Zegher F, Reinehr T, de Sousa-Malpique RM, Darendeliler F, López-Bermejo A and Ibañez-Toda L. Reduced Prenatal Weight Gain and/or Augmented Postnatal Weight Gain Precedes Polycystic Ovary Syndrome in Adolescent Girls. *Obesity* (Silver Spring) 2017. 25: 1486-1489. FI: 3,873(Q1).
- Díaz-Silva M, García C, Sebastiani G, de Zegher F, López-Bermejo A and Ibañez-Toda L. Placental and Cord Blood Methylation of Genes Involved in Energy; Homeostasis: Association With Fetal Growth and Neonatal Body Composition. *Diabetes* 2017. 66: 779-784. FI: 8,684(Q1).
- Ibañez-Toda L, Del Río L, Díaz-Silva M, Sebastiani G, Pozo ÓJ, López-Bermejo A and de Zegher F. Normalizing Ovulation Rate by Preferential Reduction of Hepato-Visceral; Fat in Adolescent Girls With Polycystic Ovary Syndrome. *J Adolesc Health* 2017. 61: 446-453. FI: 3,974(Q1).
- Ibañez-Toda L, Oberfield SE, Witchell SF, Auchus RJ, Chang RJ, Codner E, et al. An International Consortium Update: Pathophysiology, Diagnosis, and Treatment of Polycystic Ovarian Syndrome in Adolescence. *Horm Res Paediatr* 2017. 88: 371-395. FI: 1,844(Q2).

### Networks

- CIBERDEM



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CLINICAL TRIALS  
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STUDIES



PUBLICATIONS  
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## EARLY ORIGINS OF LONG-TERM RISK OF OBESITY AND METABOLIC DISEASE

Josep Jiménez Chillarón, PhD

*The hypothesis of the foetal origin of adult disorders postulates that an adverse intrauterine environment may affect the growth of the foetus by producing permanent changes of gene expression in key cells and/or tissues. Such changes would persist after birth and could contribute to the development of chronic and complex disorders typical of adults, such as type 2 diabetes or obesity. Our laboratory's goal is to understand the molecular mechanisms that associate nutrition during intrauterine development with the risk of acquiring chronic disorders as an adult, with special emphasis on diabetes and obesity.*

*In order to understand the underlying molecular mechanisms, in our group we have developed an experimental model of foetal malnutrition that recapitulates the metabolic phenotype described in humans: malnourished mice in the uterus show low weight at birth and develop obesity, intolerance to glucose, and diabetes as from the 4th month of life.*

### Members

#### Staff researcher

Carles Lerin Martínez, Ruben Díaz Naderi,  
Marta Ramon Krauel

#### PhD Student

Silvia Ribó Gené

#### Technician

Judith Cebrià Romeo

### Selected projects

- Molecular determinants that link neonatal nutrition with late onset diabetes: epigenetic regulation of the circadian clock. European Foundation for the Study of Diabetes - Programme for Diabetes Research in Europe. EFSD/Lilly 2015. PI: José Carlos Jiménez Chillarón. 2016 - 2018.
- Encapsulated Synthetic Cellular Circuits to Restore Glycemic Control in Type 1 Diabetes. Fundació La Marató de TV3 - Convocatòria de projectes sobre diabetis i obesitat. 201610-30. PI: Ruben Díaz Naderi. 2017 - 2020.
- Miguel Servet II\_Beneficiario: Dr. Josep Jimenez. Instituto de Salud Carlos III (ISCIII) - Miguel Servet II. MSII16/00046. PI: José Carlos Jiménez Chillarón. 2017 - 2020.

### Selected observational studies

- International Study of Low-Size Genetics and Neuroendocrinology. Promoter: Lilly S.A. PI: Rubén Díaz. 2007 - 2017.
- "Increlex European Forum Database[mecasermin (derived from injectable rDNA)] on growth retardation: European patient registry for monitoring the long-term efficacy and safety of Increlex. Eu-IGFDD". Promoter: Ipsen Pharma, S.A. PI: Marta Ramon. 2009 -

- Recombinant growth hormone [rGH] treatment in patients with controlled GH deficiency at Hospital Sant Joan de Déu: Analysis of early markers of good response useful in clinical practice. Promoter: Fundació Privada per a la Recerca i la Docència Sant Joan de Déu - FSJD FSJD. PI: Marta Ramon. 2015 -

### Selected publications

- Duran Fernandez-Feijoo C, Carrasco-Carrasco C, Villamazo Francisco N, Cebrià Romero J, Fernández Lorenzo JR, Jimenez-Chillaron JC and Camprubí-Camprubí M. Influence of catch up growth on spatial learning and memory in a mouse model of intrauterine growth restriction. PLoS One 2017. FI: 2,806(Q1).
- Rodríguez-García MA, Del Rio-Baquero LM, Ortez-Gonzalez CI, Jou-Munoz C, Vigo-Morancho M, Medina J, et al. Differences in Adipose Tissue and Lean Mass Distribution in Patients with Collagen VI Related Myopathies Are Associated with Disease Severity and Physical Ability. Front Aging Neurosci 2017. 9: 268-0. FI: 4,504(Q1).

### Networks

- CIBERDEM



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RESEARCHER



PROJECTS



CLINICAL TRIALS  
& OBSERVATIONAL  
STUDIES



PUBLICATIONS  
WITH IF



FIRST QUARTILE  
PUBLICATIONS

## NEONATAL PHYSIOPATHOLOGY

Alfredo Garcia-Alix Pérez, MD, PhD

*Foetal growth restriction may have irreversible consequences. The study of the various stages from intrauterine life to adolescence will allow us to advance in the prevention of this condition and in the treatment and prevention of the complications derived from it.*

### Members

#### Staff researcher

Thais Agut Quijano, Miguel M<sup>a</sup> Alsina Casanova, Marta Camprubí Camprubí, Ruth Del Rio Florentino, Isabel Iglesias Platas, Martin Iriondo Sanz, Ana Martín Ancel, Julio Moreno Hernando, Ana Morillo Palomo, Africa Perttierra Cortada, Ana Riverola De Veciana, Javier Rodríguez Fanjul

#### Technician

Miriam López Abad

### Selected projects

- Rol of Beta - 2- microglobulin as a biomarker in extremely preterm infants with CNS inflammation. Sociedad Española de Neonatología - PREMIO "CARMEN PEDRAZ". PI: Marta Camprubi Camprubi. 2017 - 2018.
- Prediction of survival without neonatal morbidity and without moderate-severe disability of neurodevelopment in premature newborns of the Spanish SEN1500 network. Instituto de Salud Carlos III (ISCIII) - Proyectos de investigación en salud. PI16/00748. PI: Martin Iriondo Sanz. 2017 - 2019.
- Development of an algorithm based on multimodal neuroimaging for personalised prognosis of neonatal cerebral arterial infarction. OSSJD - Obra Social Sant Joan de Déu. BECA BOMBERS 2017. PI: Alfredo Garcia-Alix Pérez. 2017 - 2020.

### Selected clinical trials

- H2020-PHC-18-2015-667224. Effect of Allopurinol in addition to hypothermia for hypoxic-ischemic Brain Injury on Neurocognitive Outcome - a blinded randomized placebo-controlled parallel group multicenter trial for superiority [Phase III]. EUDRACT: 2016-000222-19. Promoter: Universitat Hospital Tübingen PI: Marta Camprubi Camprubi 05/05/2017 -
- REACT RCT. Producte Sanitari. Evaluation of Real Time Continuous Glucose Monitoring in the very preterm infant. PromotEr: University of Cambridge. PI: Isabel Iglesias Platas. 04/05/2017 -
- A1481316. Phase III: A multi-centre, randomized, placebo-controlled, double-blind, two-armed, parallel group study to evaluate efficacy and safety of iv sildenafil in the treatment of neonates with persistent pulmonary hypertension of the newborn (PPHN) or hypoxic respiratory failure and at risk for PPHN, with a long term follow up investigation of developmental progress 12 and 24 months after completion of study treatment. Eudract: 2012-002619-24. Promoter: Pfizer Inc. PI: Julio Moreno Hernando. 2014 -

### Selected publications

- Casanova MA, Monteagudo-Sánchez A, Rodríguez-Guerineau L, Court F, Serrano IG, Martorell-Sampol L, et al. Maternal mutations of FOXF1 cause alveolar capillary dysplasia despite not being imprinted. Hum Mutat 2017. 38: 615-620. FI: 4,601(Q1).
- François C, Teixidó M, Takerkart S, Agut-Quijano T, Bosch L and Rodriguez-Fornells A. Enhanced Neonatal Brain Responses To Sung Streams Predict Vocabulary; Outcomes By Age 18 Months. Sci Rep 2017. 7: 12451-0. FI: 4,259(Q1).
- Pérez-Cerdá C, Girós ML, Serrano M, Ecay MJ, Gort L, Pérez-Dueñas B, et al. A Population-Based Study on Congenital Disorders of Protein N- and; Combined with O-Glycosylation Experience in Clinical and Genetic; Diagnosis. J Pediatr 2017. 183: 170-0. FI: 3,874(Q1).
- Stephan-Otto C, Nuñez C, Arca-Diaz G, Agut-Quijano T and Garcia-Alix A. Three-Dimensional Map of Neonatal Arterial Ischemic Stroke Distribution; From Early Multimodal Brain Imaging. Stroke 2017. 48: 482-485. FI: 6,032(Q1).

### Networks

- BCNAtal



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RESEARCHER



PROJECTS



CLINICAL TRIALS &  
OBSERVATIONAL  
STUDIES



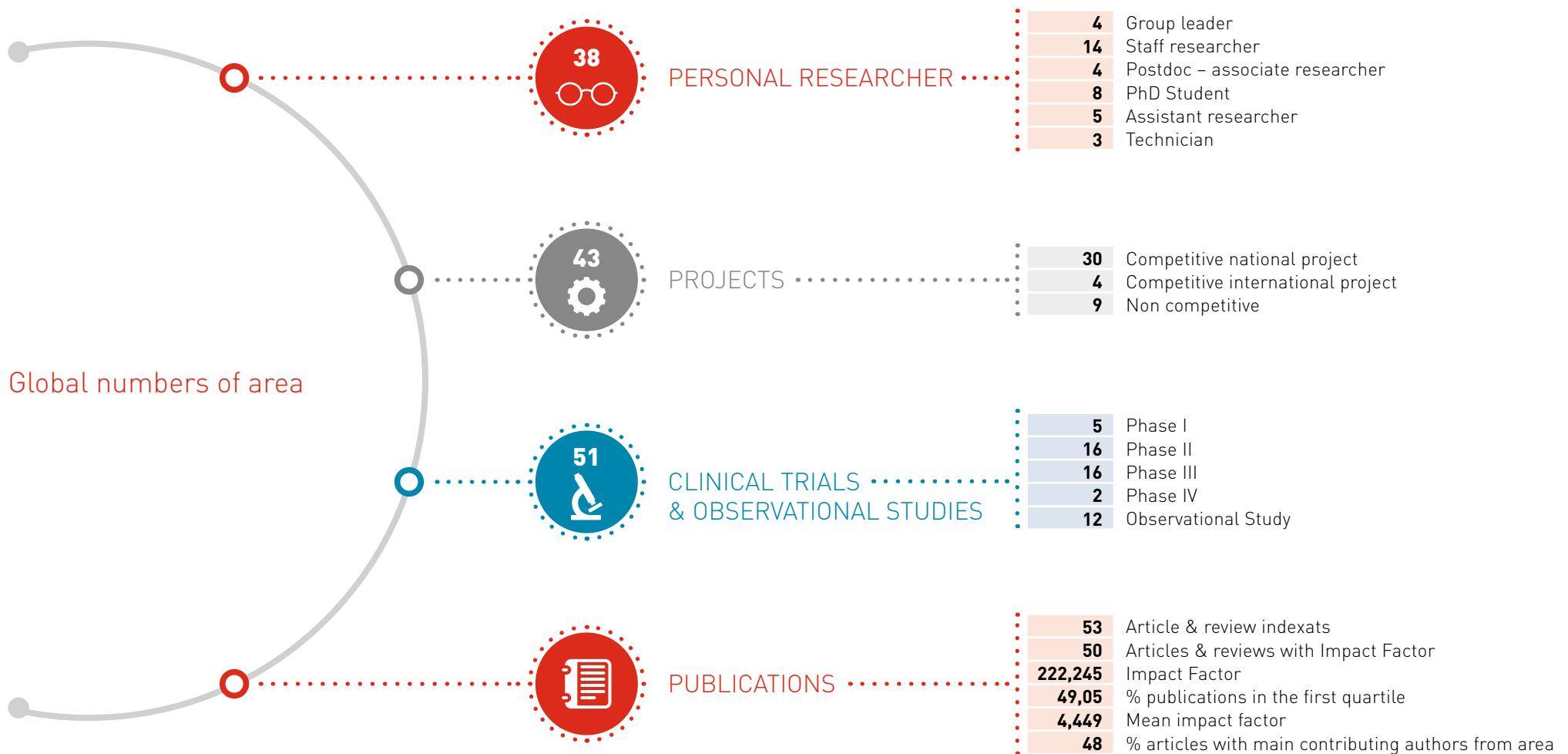
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## 3.4. Infectious diseases and systemic inflammatory response in paediatrics

**Coordinator: Carmen Muñoz-Almagro**



## VACCINE-PREVENTABLE DISEASES

Carmen Muñoz Almagro, MD, PhD

*Notable achievements during the year included the clinical and molecular characterisation of the outbreak of encephalitis caused by enterovirus A71 in Catalonia in 2016, and the implementation of the human microbiome studies in the healthy and diseased states in children.*

### Members

#### Staff researcher

Pedro Brotons de los Reyes, Juan José García García, Iolanda Jordán García, Cristian Launes Montaña

#### Postdoc – associate researcher

Cristina Esteva Afonso, Mariona Fernández de Sevilla Estrach, Natalia Timoneda Sole, Ana Valero Rello

#### Assistant researcher

Susana Hernández Bou, Ivette Rubio Martínez, Miguel García Fernández

#### PhD Student

Didac Casas Alba, Desiree Henares Bonilla, Míriam Triviño Rodríguez

#### Technician

Carles Cisneros Camps, Amaresh Pérez Argüello, Jesica Saucedo Verdes

### Selected projects

- Interactions of the nasopharyngeal microbiota and the immune response of the host: an integrated approach to the pathogenesis of invasive pneumococcal disease in paediatric patients. Hospital Sant Joan de Déu - Esplugues HSJD. Beques Recerca HSJD. BR201401. PI: Carmen Muñoz Almagro. 2016 - 2019.
- CaDMIA\_Validation of the Minimally Invasive Autopsy (MIA) tool for cause of death investigation in developing countries. Bill & Melinda Gates Foundation. Grand Challenges in Global Health. 2012. PI: Carmen Muñoz Almagro. 2012 - 2017.
- RESPOC\_Fast, cost-effective, user-friendly point of care (POC) instrument for detection of respiratory pathogens, including Streptococcus pneumoniae. European Commission. FP7-SME-2013. 606488. PI: Carmen Muñoz Almagro. 2013 - 2017.

### Selected clinical trials

- CE01-203. A Phase 2/3, Randomised, Partly blinded (blinded investigator), Multi-center, Comparative Study to Determine the Safety and activity of Solithromycin (Intravenous and oral) in paediatric patients with suspected or confirmed community acquire bacterial pneumonia (CABP). EUDRACT: 2014-004039-37. Promoter: Cempra Pharmaceuticals, Inc. PI: Juan José García García. 12/09/2016 -
- MET51. Phase 3. Immunogenicity and Safety of an Investigational Quadrivalent Meningococcal Conjugate Vaccine in Toddlers 12 to 23 Months of Age. EUDRACT: 2016-000749-30. Promoter: Sanofi-Aventis S.A. PI: Iolanda Jordán García. 15/12/2016 -

### Selected publications

- Balaguer-Gargallo M, Alejandre-Galobardes C, Vilaperez D, Esteban-Torne E, Carrasco JL, Cambra-Lasaosa FJ and Jordán-García I. Bronchiolitis Score of Sant Joan de Deu: BROSJOD Score, validation and usefulness. Pediatr Pulmonol 2017. 52: 533-539. FI: 2,758(Q1).
- Casas-Alba D, Fernández de Sevilla-Estrach M, Valero-Rello A, Fortuny-Guasch C, García-García JJ, Ortez-Gonzalez CI, et al. Outbreak of brainstem encephalitis associated with enterovirus-A71 in Catalonia, Spain [2016]: a clinical observational study in a children's reference centre in Catalonia. Clin Microbiol Infect 2017. 23: 874-881. FI: 5,292(Q1).
- Menendez C, Castillo P, Martínez MJ, Jordao D, Lovane L, Ismail MR, et al. Validity of a minimally invasive autopsy for cause of death: determination in stillborn babies and neonates in Mozambique: An observational study. PLoS Med 2017. 14 : e1002318. FI: 11,862(Q1).
- Savulescu C, Krizova P, Lepoutre A, Mereckiene J, Vestrheim DF, Ciruela Navas P, et al. Effect of high-valency pneumococcal conjugate vaccines on invasive pneumococcal disease in children in SpIDnet countries: an observational, multicentre study. Lancet Respir Med 2017. 5: 648-656. FI: 19,287(Q1).

### Networks

- 2016 - CIBERESP
- 2013 - SPIDNET
- 2015 - PERTINENT



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PROJECTS



CLINICAL TRIALS  
& OBSERVATIONAL  
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## INFECTIONS OF VERTICAL TRANSMISSION AND INFECTIONS IN VULNERABLE PATIENTS

Clàudia Fortuny Guasch, MD, PhD

*In 2017 we completed the analysis of the results of the FIS PI13/01738 project, which studied the mitochondrial toxicity secondary to exposure to human immunodeficiency virus (HIV) and to antiretroviral drugs, focused particularly on paediatric patients and on the potential impact of the depletion of mitochondrial DNA and its mutations. We are currently preparing the publications on this study.*

*We have also been awarded another FIS project (PI17/00359), addressed to studying the potential toxicity of valganciclovir/ganciclovir in patients with congenital CMV infection who receive this drug.*

### Members

#### Staff researcher

Vicky Fumadó, Silvia Simó Nebot

### Selected projects

- Longitudinal study of the mutations of mitochondrial DNA in children exposed to HIV (infected and non-infected) and to antiretrovirals. Instituto de Salud Carlos III (ISCIII). FIS. PI13/01738. PI: Clàudia Fortuny Guasch. 2014 - 2018.

### Selected clinical trials

- MK-6072-0001. Randomised double-blind placebo-controlled clinical trial to assess the safety, tolerability, pharmacokinetics and effectiveness of a single infusion of bezlotoxumab (MK-6072, human monoclonal antibody against *C. difficile* B toxin) in children and adolescents aged 1 to under 18 years receiving antibacterial treatment for *C. difficile* infection (MODIFY III). EUDRACT: 2017-000070-11. Promoter: Merck, Sharp & Dohme de España. S.A. PI: Clàudia Fortuny Guasch. 09/10/2017 -

- Codi de protocol: M16-123. Fase III. An Open-Label, Multicenter Study to Evaluate the Pharmacokinetics, Safety, and Efficacy of Glecaprevir/Pibrentasvir in Pediatric Subjects with Genotypes 1 & 6 Chronic Hepatitis C Virus (HCV) Infection. EUDRACT: 2016-004102-34. Promoter: AbbVie Deutschland GmbH & Co. KG "AbbiVie". PI: Clàudia Fortuny Guash. 03/11/2017 -

### Selected publications

- Abras A, Muñoz-Almagro C, Ballart C, Berenguer P, Llovet T, Herrero M, et al. Towards a New Strategy for Diagnosis of Congenital Trypanosoma cruzi; Infection. J Clin Microbiol 2017. 55: 1396-1407. FI: 3,712(Q2).
- Casas-Alba D, Fernández de Sevilla-Estrach M, Valero-Rello A, Fortuny-Guasch C, García-García JJ, Ortez-Gonzalez CI, et al. Outbreak of brainstem encephalitis associated with enterovirus-A71 in; Catalonia, Spain [2016]: a clinical observational study in a children's; reference centre in Catalonia. Clin Microbiol Infect 2017. 23: 874-881. FI: 5,292(Q1).

- Espiau M, Yeste D, Noguera-Julián A, Soler-Palacín P, Fortuny-Guasch C, Ferrer R, et al. Adiponectin, Leptin and Inflammatory Markers in HIV-associated Metabolic Syndrome in Children and Adolescents. Pediatr Infect Dis J 2017. 36: 31-37. FI: 2,486(Q1).
- Bossacoma-Busquets F, Noguera-Julián A, Sanchez E and Fortuny-Guasch C. Dolutegravir plus abacavir/lamivudine works in adolescents, but size; matters. J Antimicrob Chemother 2017. 72: 2958-2960. FI: 5,071(Q1).

### Networks

- CoRISPE-cat and NENEXP Cohorts for Paediatric HIV Infection in Children and Mother-to-Child Transmitted HIV Infection in Catalunya (Spain)
- PENTA-Id Network
- CIBERESP, grupo de investigación CB15/00067



PERSONAL RESEARCHER



PROJECTS



CLINICAL TRIALS & OBSERVATIONAL STUDIES



PUBLICATIONS WITH IF



FIRST QUARTILE PUBLICATIONS

## TUBERCULOSIS

Antoni Noguera Julián, MD, PhD

*Research on the adherence and toxicity associated with first-line anti-tuberculosis drugs in the paediatric age, on the pharmacokinetic characteristics of these drugs on newborns and pre-school children, on the new tuberculosis diagnostic techniques in healthy children, on children from high tuberculosis endemic areas and on immunosuppressed children, and on the clinical-epidemiological characteristics of tuberculosis in childhood in our area.*

### Members

#### PhD Student

Maria Goretti López Ramos, Eneritz Velasco, Joan Vinent Genestar

### Selected projects

- Pharmacological treatment of tuberculosis in infants under 6 months: validation of the daily single dose of isoniazid of 10mg/kg of weight. Fundación Mercedes Armengou Riera (MAR). 2014. PCP00145. PI: Antoni Noguera Julián. 2015 - 2017.
- Validation of the daily single dose of isoniazid of 10 mg/kg of weight in infants under 3 months of age. Instituto de Salud Carlos III (ISCIII). FIS. PI13/01740. PI: Antoni Noguera Julián. 2014 - 2018.
- Open phase IIA clinical trial to study the absorption of a 10 mg/ml suspension of isoniazid for treatment of tuberculosis infection in patients under 6 years of age. Instituto de Salud Carlos III (ISCIII). PICI. ICI14/00228 - FSJD-ISON-2016. PI: Antoni Noguera Julián. 2015 - 2018.

### Selected clinical trials

- TBTC S31-A5349. Rifapentine-containing treatment shortening regimens for pulmonary tuberculosis: A randomized, open-label, controlled phase 3 clinical trial. EUDRACT: 2015-003446-45. Promoter: Tuberculosis Trial Consortium. PI: Antoni Noguera Julián. 28/04/2016 -
- 7625A-035. A Phase 2, Randomized, Active Comparator-Controlled, Multicenter, Double-Blind Clinical Trial to Study the Safety and Efficacy of Ceftolozane/Tazobactam (MK-7625A) Plus Metronidazole Versus Meropenem in Pediatric Subjects with Complicated Intra- Abdominal Infection. EUDRACT: 2016-004820-41. Promoter: Merck, Sharp & Dohme de España. S.A. PI: Antoni Noguera Julián. 31/08/2017 -
- FSJD-ISON-2016. Open clinical trial phase IIa to study the isoniazid suspension absorption of 10mg/ml for the tuberculosis infection treatment in patients under the age of 6. EUDRACT: 2016-002000-31. Promoter: Fundació Privada per a la Recerca i la Docència Sant Joan de Déu - FSJD FSJD. PI: Antoni Noguera Julián. 10/10/2016 -

### Selected publications

- Espiau M, Yeste D, Noguera-Julián A, Soler-Palacín P, Fortuny-Guasch C, Ferrer R, et al. Adiponectin, Leptin and Inflammatory Markers in HIV-associated Metabolic Syndrome in Children and Adolescents. Pediatr Infect Dis J 2017. 36: 31-37. FI: 2,486(Q1).



PERSONAL  
RESEARCHER



PROJECTS



CLINICAL TRIALS  
& OBSERVATIONAL  
STUDIES



PUBLICATIONS  
WITH IF



FIRST QUARTILE  
PUBLICATIONS

### Networks

- Tuberculosis Trials Consortium (TBTC), de los CDC de Atlanta (EEUU); sub-investigador del site 31 (Barcelona)
- PENTA Steering Committee (Paediatric European Network for the Treatment of AIDS)
- CIBER en Salud Pública (CIBERESP), Grupo de Investigación CB15/0006
- pTBNet (Red Europea para Estudio de la Tuberculosis Pediátrica).

## SYSTEMIC INFLAMMATORY RESPONSE IN PAEDIATRIC AGE

Jordi Antón López, MD, PhD

This year the group's pre-consolidated status has been recognised by AGAUR. The team led by Dr. Antón has carried on with the projects on the study of autoimmune diseases (such as juvenile idiopathic arthritis, juvenile dermatomyositis and scleroderma), autoinflammatory diseases (genetic and epigenetic studies) and the aetiology of Kawasaki disease, likewise taking part in various clinical trials. As a result of this research, several papers have been communicated and published. The group led by Dr. Alsina centres its research on primary/congenital and secondary immunodeficiencies, focusing on 1) primary immunodeficiencies of the IL-12 /IFN- $\gamma$  pathway generating susceptibility to intracellular infections (mycobacteria, leishmanias, among others); 2) primary immunodeficiencies with a predominant immune dysregulation component (inflammation, lymphoproliferation, autoimmunity), with particular interest in the study of the cells possessing a regulatory role (regulatory T and B lymphocytes); 3) immunodeficiencies secondary to the use of monoclonal antibodies, especially anti-TNF alphas during pregnancy. The aim is to learn about the bases of immune dysfunction in the three pathology groups and to identify biomarkers and new therapeutic targets.

### Members

#### Staff researcher

Laia Alsina Manrique de Lara, Rosa Bou i Torrent, Ángela Deyà Martínez, Estibaliz Iglesias Jiménez, Mónica Piquer Gibert, Juan Manuel Mosquera Angarita, Judith Sánchez Manubens, Andrea Zacarias Crovato

#### Assistant researcher

Violeta Bittermann, Clara Isabel Giménez Roca

#### PhD Student

Joan Calzada Hernández, Anna Esteve Solé

### Selected projects

- WINDBIOME\_Aerial micro-biome diversity and their role in Kawasaki disease. Fundació Privada Daniel Bravo Andreu. Projectes Daniel Bravo. PI: Jordi Antón López. 2015 - 2018.
- Assessment of mechanisms responsible for illness in carrier patients of heterozygous MEV mutations. Becas FER. PI: Estibaliz Iglesias Jimenez. 2017 - 2018.
- Study of the effect of intrauterine exposure to anti-TNF-alpha drugs in the immune system of children of mothers with inflammatory rheumatic diseases. Fundación Banco Bilbao Vizcaya Argentaria. Becas Leonardo para Investigadores y Creadores. PI: Laia Alsina Manrique de Lara. 2017 - 2019.
- Modulating factors of primary immunodeficiency with susceptibility to severe infections by intracellular germs in the paediatric age. Instituto de Salud Carlos III (ISCIII). FIS. PI15/01094. PI: Laia Alsina Manrique de Lara. 2016 - 2018.

### Selected clinical trials

- GT1503. Fase III. A Multi-Centre, Open-Label, Single Arm Trial to Evaluate Efficacy, Pharmacokinetics, and Safety and Tolerability of IGSC 20% in Subjects with Primary Immunodeficiency. EUDRACT: 2015-003290-15. Promoter: Instituto Grifols, S.A. PI: Laia Alsina Manrique de Lara. 2016 -
- CACZ885N2301. Fase III. A randomized, double-blind, placebo controlled study of canakinumab in patients with Hereditary Periodic Fevers (TRAPS, HIDS, or crFMF), with subsequent randomized withdrawal/ dosing frequency reduction and open-label long term treatment epochs. EUDRACT: 2013-004291-35. Promoter: Novartis Farmacéutica, S.A. PI: Jordi Antón López. 2014 -
- WA28118. Fase I. A phase Ib, Open-Label, Multicenter study to investigate the pharmacokinetics, pharmacodynamics, and safety of tocilizumab following subcutaneous administration to patients with systemic juvenile idiopathic arthritis. EUDRACT: 2012-003490-26. Promoter: F. Hoffmann-La Roche Ltd. PI: Jordi Antón López. 2015 -

### Selected publications

- Arostegui JI, Anton-Lopez J, Calvo I, Robles A, Iglesias-Jimenez E, López-Montesinos B, et al.

Open-Label, Phase II Study to Assess the Efficacy and Safety of; Canakinumab Treatment in Active Hyperimmunoglobulinemia D With Periodic; Fever Syndrome. Arthritis Rheumatol 2017. 69: 1679-1688. FI: 6,918(Q1).

- Esteve-Sole A, Deyà-Martinez A, Teixido M, Ricart E, Gompertz M, Torradeflot M, et al. Immunological Changes in Blood of Newborns Exposed to Anti-TNF-alpha during Pregnancy. Front Immunol 2017. 8: 1123-0. FI: 6,429(Q1).
- Esteve-Solé A, Teixidó I, Deyà-Martinez A, Yagüe-Ribes J, Plaza-Martín AM, Juan M and Alsina-Manrique de Lara L. Characterization of the Highly Prevalent Regulatory CD24(hi)CD38(hi) B-Cell Population in Human Cord Blood. Front Immunol 2017. 8: 201-0. FI: 6,429(Q1).
- Navallas M, Inarejos E, Iglesias-Jimenez E, Cho Lee GY, Rodríguez N and Anton-Lopez J. MR Imaging of the Temporomandibular Joint in Juvenile Idiopathic; Arthritis: Technique and Findings(1). Radiographics 2017. 37: 595-612. FI: 3,427(Q1).

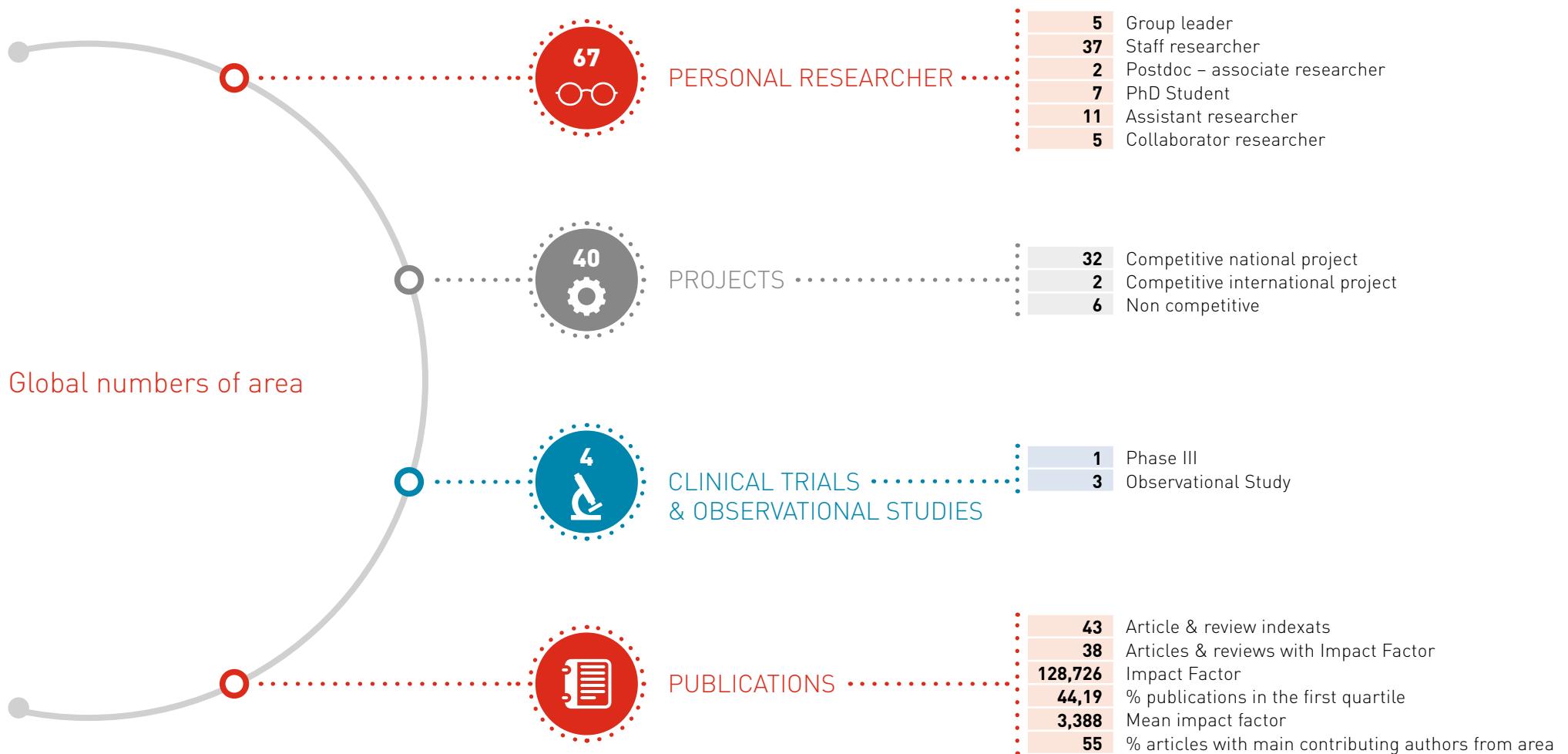
### Networks

- 2000 - Jordi Antón member of PRINTO (Pediatric Rheumatology International Trials Organization)
- 2017 - Laia Alsina member of Comité del Registro Español de Inmunodeficiencias Primarias RÉDIP



## 3.5. Mental Health

**Coordinator: Judit Usall and Susana Ochoa**



## CLINICAL, COGNITIVE AND PSYCHOSOCIAL BASES OF SEVERE MENTAL DISORDERS

Susana Ochoa Güerre, MD, PhD

*The evaluation of the clinical, psychosocial and neuropsychological characteristics of people with a severe mental disorder will allow us to identify more precisely the treatment needs in each case.*

*The main goal of this line is to evaluate the various clinical, social and neuropsychological aspects, among others, of the people who begin to manifest the first episodes of severe mental disorder. In order to carry out good evaluations in these areas, though, suitable measuring instruments must be available.*

### Members

#### Staff researcher

Antonio Manuel Alba Muñoz, Hilari Andres Mora, Emma Casas Anguera, Jorge Cuevas Esteban, Gemma Escuder Romeva, Alexandrina Foix Sanjuan, Marcia Irazabal Jiménez, Francisco Martínez Zambrano, María Trinidad Peláez Martínez

#### Assistant researcher

Anna Butjosa Molines, Núria del Cacho Ortega, Ingrid Tortadès Arumí, Regina Vila Badia

### Selected projects

- Assessment of the effectiveness of community assertive treatment addressed to patients with severe mental disturbances taking part in the Housing First programme. Beneficiaria: Inés Campo Ferreira. Parc Sanitari de Sant Joan de Déu -Serveis de Salut Mental. PI: Alexandrina Foix Sanjuan. 2015 - 2017.
- Assessment of emotion recognition in patients with schizophrenia using multimedia technology. Parc Sanitari de Sant Joan de Déu -Serveis de Salut Mental. AFR 2016. PI: 2016 - 2018.
- Effectiveness of individualised metacognitive therapy (MCT+) in patients with psychosis of brief evolution. Instituto de Salud Carlos III (ISCIII). FIS. PI14/00044. PI: Susana Ochoa Güerre. 2015 - 2018.

### Selected publications

- Nuñez C, Paipa N, Senior C, Coromina M, Siddi S, Ochoa S, Brebion G and Stephan-Otto C. Global brain asymmetry is increased in schizophrenia and related to avolition. Acta Psychiatr Scand 2017. 135: 448-459. FI: 6,790(Q1).

- Ochoa S, López-Carrilero R, Barrigón ML, Pousa Tomas E, Barajas A, Lorente-Rovira E, et al. Randomized control trial to assess the efficacy of metacognitive training compared with a psycho-educational group in people with a recent-onset psychosis. Psychol Med 2017. 47: 1573-1584. FI: 5,230(Q1).
- Stephan-Otto C, Siddi S, Carl Senior, Cuevas-Estebar J, Cambra-Martí MR, Ochoa S and Brebion G. Remembering verbally-presented items as pictures: Brain activity underlying visual mental images in schizophrenia patients with visual hallucinations. Cortex 2017. 94: 113-122. FI: 4,279(Q1).
- Butjosa Molines A, Gómez-Benito J, Myin-Germeys I, Barajas A, Baños I, Usall J, et al. Development and validation of the Questionnaire of Stressful Life Events (QSLE). J Psychiatr Res 2017. 95: 213-223. FI: 4,183(Q1).

### Networks

- CIBERSAM
- Consolidated research group in AGAUR



PERSONAL  
RESEARCHER



PROJECTS



PUBLICATIONS  
WITH IF



FIRST QUARTILE  
PUBLICATIONS

## MENTAL HEALTH INTERVENTIONS

Judith Usall Rodie, MD, PhD

*The current pharmacological and psychological treatments are not effective in all the people who suffer schizophrenia. This research group conducts clinical trials on the treatment of resistant schizophrenia or schizophrenia with predominant negative pharmacological or psychological symptoms. Four clinical trials have been carried out independently of the pharmaceutical industry, with financing from public bodies and from private foundations (Stanley Foundation). Gender- and sex-sensitive psychopharmacology is also taken into consideration in the research that is conducted.*

### Members

#### Staff researcher

Belen Arranz Martín, Adela Berrozpe Blasco, Marta Coromina Sadurní, Ruth Cunill Clotet, Gemma Escartín Martín, Mar García Franco, Eva Miquel Fernández, Elena Rubio Abadal, Luis San Molina, Sonia Vilamala Anton

#### Assistant researcher

Marta Ferrer Quintero, Gabriel Ledesma Iparraguirre, Raquel López Carrilero, Silvia Marcó García

#### Collaborator researcher

Maria Elena Huerta Ramos

### Selected projects

- Vila Saborit Award for the best article. Acadèmia de Ciències Mèdiques i de la Salut de Catalunya i de Balears. Premi Millor Article. Premi Millor Article Judith Usall. PI: Judith Usall Rodie. 2016 - 2017.
- m-RESIST\_Mobile Therapeutic Attention for Patients with Treatment Resistant Schizophrenia. European Commission. H2020-PHC-2014-single-stage. 643552. PI: Judith Usall Rodie. 2015 - 2017.
- SGR 2014\_Etiopatogenia i tractament dels trastorns mentals. Agaur - Agència de Gestió d'Ajuts Universitaris i de Recerca. SGR. SGR2014-1603. PI: Judith Usall Rodie. 2014 - 2017.

### Selected observational studies

- LSM-CLO-2016-01. Multicentre observational study of the cost-effectiveness of long-acting antipsychotics versus oral antipsychotics in patients with schizophrenia. Promoter: Cluster de Salut Mental Catalunya. PI: Luis San Molina. 2016 -

### Selected publications

- Ochoa S, López-Carrilero R, Barrigón ML, Pousa Tomas E, Barajas A, Lorente-Rovira E, et al. Randomized control trial to assess the efficacy of metacognitive training compared with a psycho-educational group in people with a recent-onset psychosis. *Psychol Med* 2017; 47: 1573-1584. FI: 5,230(Q1).
- Mas S, Gassó P, Torra M, Bioque M, Lobo A, González-Pinto A, et al. Intuitive pharmacogenetic dosing of risperidone according to CYP2D6 phenotype extrapolated from genotype in a cohort of first episode psychosis patients. *Eur Neuropsychopharmacol* 2017; 27: 647-656. FI: 4,239(Q1).
- Butjosa Molines A, Gómez-Benito J, Myin-Germeys I, Barajas A, Baños I, Usall J, et al. Development and validation of the Questionnaire of Stressful Life Events (QSLE). *J Psychiatr Res* 2017; 95: 213-223. FI: 4,183(Q1).
- Pousa Tomas E, Ochoa S, Cobo J, Nieto García ML, Usall J, Gonzalez B, et al. A deeper view of insight in schizophrenia: Insight dimensions, unawareness and misattribution of particular symptoms and its relation with psychopathological factors. *Schizophr Res* 2017; 189: 61-68. FI: 3,986(Q1).

### Networks

- CIBERSAM



## COGNITIVE AND NEUROBIOLOGICAL CORRELATES OF PSYCHOTIC AND AFFECTIVE SYMPTOMS IN SEVERE MENTAL DISORDER

Gildas Brebion, PhD

*In 2017 we published several articles on our usual research subjects, some in first decile journals.*

*We also developed a new line of research based on the quantification of brain asymmetry. Likewise, we took on a master's degree student who is exploring another line of research on body asymmetry in relation to schizophrenia symptoms.*

### Members

#### Staff researcher

Christian Stephan-Otto Attolini

#### Postdoc – associate researcher

Christian Núñez Leánez

### Selected projects

- PSSJD research grant: Unusual psychic experiences and jumping to conclusions in the clinical and general populations. Parc Sanitari de Sant Joan de Déu -Serveis de Salut Mental. PI: Sara Siddi. 2014 - 2018.
- Neurofunctional bases of anomalous inner discourse in verbal hallucinations. Instituto de Salud Carlos III (ISCIII) - FIS. PI14/00047. PI: Gildas Brebion. 2015 - 2018.
- Miguel Servet tipo II - Beneficiario: Gildas Brébion. Instituto de Salud Carlos III (ISCIII) - Miguel Servet II. MSII15/00005. PI: Gildas Brebion. 2016 - 2019.
- Differences of global brain asymmetry among people with symptoms of depression and anxiety. Institute of Neurological Specialities.- Beca Dr. Pedro Albert. Beca Dr. Pedró Albert 2016. PI: Christian Núñez Leánez. 2017.

### Selected publications

- Nuñez C, Paipa N, Senior C, Coromina M, Siddi S, Ochoa S, Brebion G and Stephan-Otto C. Global brain asymmetry is increased in schizophrenia and related to avolition. *Acta Psychiatr Scand* 2017. 135: 448-459. FI: 6,790(Q1).
- Stephan-Otto C, Nuñez C, Arca-Diaz G, Agut-Quijano T and Garcia-Alix A. Three-Dimensional Map of Neonatal Arterial Ischemic Stroke Distribution; From Early Multimodal Brain Imaging. *Stroke* 2017. 48: 482-485. FI: 6,032(Q1).
- Stephan-Otto C, Siddi S, Carl Senior, Cuevas-Esteban J, Cambra-Martí MR, Ochoa S and Brebion G. Remembering verbally-presented items as pictures: Brain activity underlying visual mental images in schizophrenia patients with visual hallucinations. *Cortex* 2017. 94: 113-122. FI: 4,279(Q1).
- Stephan-Otto C, Siddi S, Senior C, Muñoz Samons D, Ochoa S, Sánchez-Laforga AM and Brebion G. Visual Imagery and False Memory for Pictures: A Functional Magnetic Resonance Imaging Study in Healthy Participants. *PLoS One* 2017. 12: e0169551 . FI: 2,806(Q1).



## CHILD AND YOUTH MENTAL HEALTH RESEARCH

Montserrat Dolz Abadia, MD, PhD

The general goal of our research group is to conduct clinical research on various psychiatric disorders, with special emphasis on neurodevelopmental disorders. At present our efforts are focused on the study of initial psychoses in childhood [PI: Dr. Dolz], attention deficit hyperactivity disorders [ADHD; PI: Dr. Alda], and autism spectrum disorders [ASD; PI: Dr. Mairena].

In recent years the group has become consolidated, carrying out various projects financed by both public institutions [FIS projects] and private ones, and collaborations with different stable research groups of the CIBERSAM mental health biomedical research network, the G04 group from Hospital Clínic i Provincial de Barcelona and the G01 group from Hospital Gregorio Marañón in Madrid. The year was marked by the award of the European project Focus Locus centred on ADHD and gamification systems. Within the national sphere, a number of prizes were won for various research studies. Our group was awarded a research prize which allows it to continue to study ADHD and mindfulness by means of virtual reality. Likewise, we have received awards for papers and posters on intervention in social skills in children with autism, and for the identification of differential biomarkers of ADHD characterised by sluggish cognitive time.

### Members

#### Staff researcher

José Ángel Alda Díez, Mar Alvarez Segura, Ester Camprodon Rosanas, María Diez Juan, Neus Elias Carbonell, 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

#### Assistant researcher

Marina Fabrega Ribera, Andrea González Contijoch, Carlota Saumell Andreu

#### PhD Student

Claudia Caprile Elola - Olaso, Anna Huguet Miguel, Ania Pérez Racana, Laia Portoles, Jordina Tor Fabra

#### Collaborator researcher

Carme Tello

### Selected projects

- FocusLocus\_AdHD management gaming system for educational achievement and social inclusion. European Commission. H2020-ICT-24-2016. PI: Jordi Navarra. 2017 - 2019.
- Characterisation of metabolic profile (bacterial and non-bacterial) in children and adolescents with ADHD and assessment of its value as a diagnostic marker. Fundación Alicia Koplowitz. Ayudas a proyectos de Investigación. Fundación Koplowitz JAldá 2017. PI: José Ángel Alda Díez. 2017 - 2019.
- Longitudinal multicentre study on neurobiological stress markers in children and adolescents with psychosis risk syndrome (PRS) and in transition to psychosis. Instituto de Salud Carlos III (ISCIII). FIS. PI15/00509. PI: Montserrat Dolz Abadia. 2016 - 2018.

### Selected clinical trials & observational studies

- PURPOSE / ABR54654. Phase III. Placebo-controlled trial in subjects at ultra-high risk for psychosis with omega-3 fatty acids in Europe. Codi EUDRACT: 2015-003503-39. Promotor: University Medical Center Utrecht. PI: Montserrat Dolz Abadia. 05/12/2016 -

### Selected publications

- Ríos-Hernández A, Alda JA, Farran-Codina A, Ferreira Garcia E and Izquierdo-Pulido M. The Mediterranean Diet and ADHD in Children and Adolescents. Pediatrics 2017. 139: e20162027. FI: 5,705(Q1).
- Butjosa Molines A, Gómez-Benito J, Myin-Germeys I, Barajas A, Baños I, Usall J, et al. Development and validation of the Questionnaire of Stressful Life Events (QSLE). J Psychiatr Res 2017. 95: 213-223. FI: 4,183(Q1).
- Caprile C, Campistol-Plana J, Puigcerver L, Gutiérrez-Mata AP, Alonso-Colmenero I, Colomé-Roura R and Navarra J. Subtle visuomotor deficits and reduced benefit from practice in early treated phenylketonuria. J Clin Exp Neuropsychol 2017. 39: 931-940. FI: 1,839(Q2).
- Varela-Besteiro O, Serrano E, Rodríguez-Vicente V, Curet Santiesteban M, Conangla-Roselló G, Cecilia Costa R, Carulla Roig M, Matalí JL and Dolz M. Suicidal ideation and self-injurious behavior in adolescents with eating disorders. Actas Esp Psiquiatr 2017. 45: 157-166. FI: 1,339(Q3).

### Networks

- CIBERSAM



25  
PERSONAL  
RESEARCHER



13  
PROJECTS



1  
CLINICAL TRIALS  
& OBSERVATIONAL  
STUDIES



13  
PUBLICATIONS  
WITH IF



25%  
FIRST QUARTILE  
PUBLICATIONS

## PSYCHOLOGICAL RESEARCH GROUP ON FIBROMYALGIA AND CHRONIC PAIN (AGORA GROUP) grupo SGR 667

Juan Vicente Luciano Devis, PhD

The AGORA research Group, coordinated by Juan V. Luciano PhD, has been formally accredited as an Emerging Group (2017 SGR 667) by the AGAUR (Agency for Management of University and Research Grants; Government of Catalonia) and it forms part of the Spanish themed network RETICS-RedIAPP (Institute of Health Carlos III). This multidisciplinary group (5 psychologists, 1 general practitioner, 1 rheumatologist, 1 anesthesiologist, and 1 statistician) is interested in chronic pain, more specifically, in what psychological treatments work, for whom, and why. The group has experience in research and teaching of clinical psychology, psychometrics, health economics, and more specifically in the economic evaluation of different pharmacological and non-pharmacological treatments and services designed to help people with chronic pain. Along 2017, the group has published 10 manuscripts in indexed scientific journals and has obtained 3 competitive contracts (2 postdocs + 1 predoc).

Current research topics of the group:

- Cost-effectiveness of psychological treatments for fibromyalgia and chronic pain.
- Process and mechanism oriented research in chronic pain.
- Neurobiological underpinnings of chronic pain.
- Psychometric analysis of patient-reported outcome measures.

### Members

#### Staff researcher

Antoni Rozadilla Sacanell, Lluis Torruella Barraquer

#### Postdoc – associate researcher

Albert Feliu Soler

#### PhD Student

Laura Andrés Rodríguez, Adrián Pérez Aranda

#### Collaborator researcher

Jaume Aguado Carné, Xavier Borràs Hernández, Eugenia Cardeñosa Valera

### Selected projects

- Miguel Servet Tipo I (Proyecto Asociado) Cost-effectiveness of a Mindfulness-Based Stress Reduction (MBSR) treatment for fibromyalgia syndrome: A 12-month randomised, controlled trial. Instituto de Salud Carlos III (ISCIII) - Miguel Servet tipo I. CP14/00087. PI: Juan Vicente Luciano Devis. 2015 - 2018.
- Neurobiological effects of mindfulness-based stress reduction (MBSR) vs. psycho-educational intervention (FibroQoL) in patients with fibromyalgia (EUDAIMON Project). Instituto de Salud Carlos III (ISCIII) - FIS. PI15/00383. PI: Juan Vicente Luciano Devis. 2016 - 2018.

- Contracts Sara Borrell. Beneficiary: Albert Feliu. Instituto de Salud Carlos III (ISCIII) - Sara Borrell. CD16/00147. PI: Juan Vicente Luciano Devis. 2017 - 2020.

### Selected observational studies

- 10297. Patients with axial spondyloarthritis: Multinational recording of clinical features, including radiological progression, and disease burden over 5 years in real-life settings. Estudio PROOF. Promoter: Abbvie Spain, S.L.U. PI: Antoni Rozadilla Sacanell. 2014 -
- IM101348ST (ASCORE). ESTUDIO ASCORE: long-term experience with abatacept s.c. in routine medical practice. EUDRACT: BMS-ABA-2014-01. Promoter: Abbvie Spain, S.L.U. PI: Antoni Rozadilla Sacanell. 2015 -

### Selected publications

- Luciano JV, D'Amico F, Feliu-Soler A, McCracken LM, Aguado J, Peñarrubia-María MT, et al. Cost-Utility of Group Acceptance and Commitment Therapy for Fibromyalgia Versus Recommended Drugs: An Economic Analysis Alongside a 6-Month

Randomized Controlled Trial Conducted in Spain (EFFIGACT Study). J Pain 2017. 18: 868-880. FI: 4,519 (Q1).

- Feliu-Soler A, Pascual JC, Elices M, Martín-Blanco A, Carmona C, Cebolla A, Simón V and Soler J. Fostering Self-Compassion and Loving-Kindness in Patients With Borderline Personality Disorder: A Randomized Pilot Study. Clin Psychol Psychother 2017. 24: 278-286. FI: 1,933 (Q2).
- Feliu-Soler A, Reche-Camba E, Borràs X, Pérez Aranda A, Andrés Rodríguez L, Peñarrubia-María MT, et al. Psychometric Properties of the Cognitive Emotion Regulation Questionnaire (CERQ) in Patients with Fibromyalgia Syndrome. Front Psychol 2017. 8: 2075. FI: 2,323 (Q2).
- Federici S, Bracalenti M, Meloni F and Luciano JV. World Health Organization disability assessment schedule 2.0: An international systematic review. Disabil Rehabil 2017. 39: 2347-2380. FI: 1,804 (Q1).

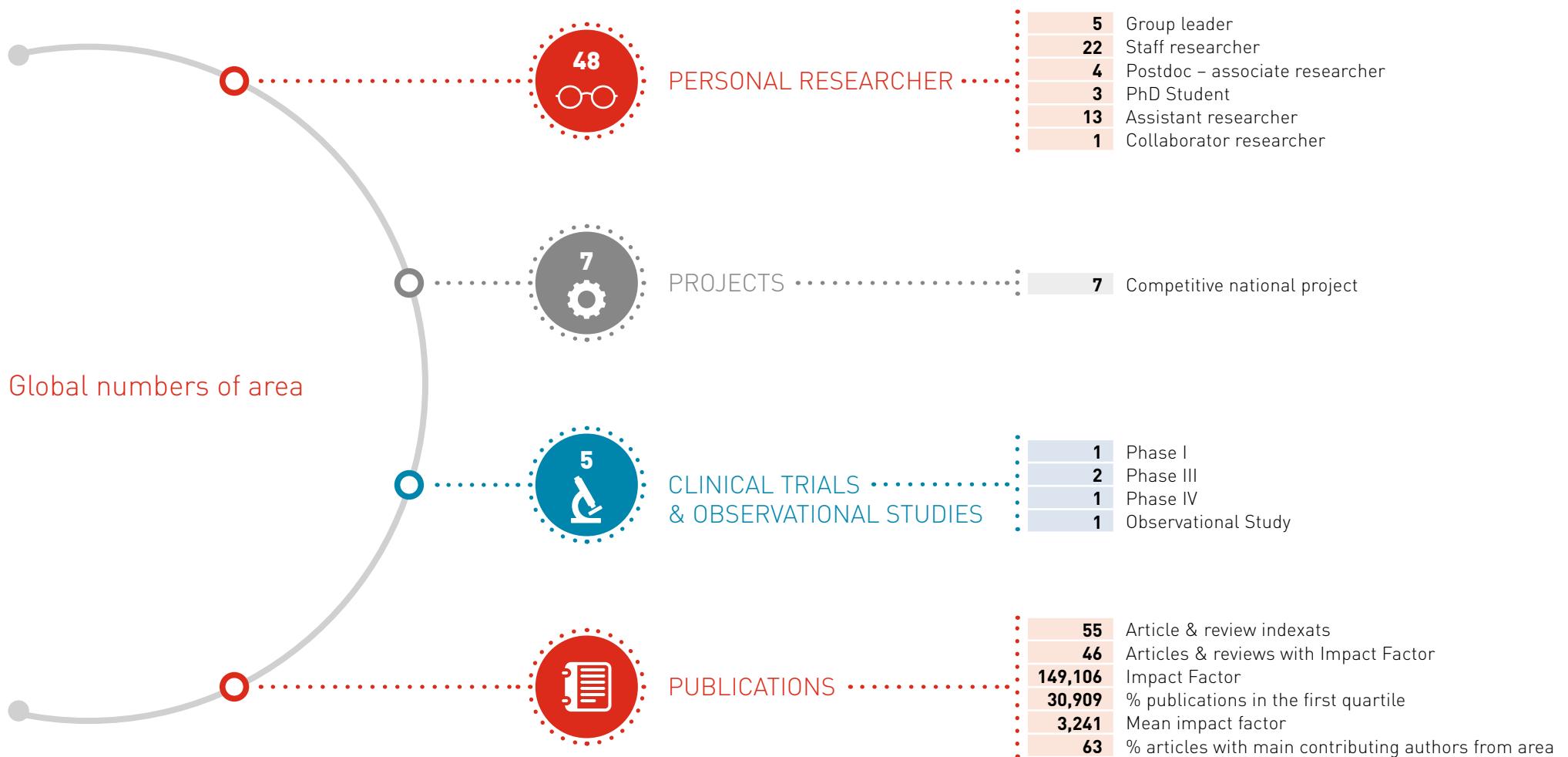
### Networks

- Coordinator participates in the redIAPP (ISCIII; RD16/0007/0012)



## 3.6. Foetal / Paediatric Diseases and environment, metabolic and genetic factors

**Coordinator: Maria Dolores Gómez-Roig**



## PAEDIATRIC INTENSIVE CARE UNIT RESEARCH PROJECT GROUP

Francisco José Cambra Lasaosa, MD, PhD

*Completion of the PFIS0116 project: Towards a body area network for measurement of stress levels. Drafting of final report. The results are currently in the process of publication.*

*PFIS0116 project. Analysis of a diagnostic algorithm for severe pneumonia in critical paediatric patients by means of lung ultrasound scanning and procalcitonin as a plan for improving healthcare quality.*

*It is unfolding appropriately within the envisaged timeframe.*

### Members

#### Staff researcher

Monica Balaguer Gargallo, Elisabeth Esteban Torne, Iolanda Jordán García, Martí Pons Òdena

#### Assistant researcher

Carme Alejandre Galobardes, Georgina Armero Campos, Sergio Benito Fernández, Sara Bobillo Pérez, Patricia Corniero Alonso, Aida Felipe Villalobos, Elena Fresán Ruiz, Lluisa Hernandez Platero, Nuria Millán García del Real, Luciana Rodríguez Guerineau, Susana Segura Matute, David Vila Pérez

#### Collaborator researcher

Montserrat Esquerda Areste

### Selected projects

- Towards a body area network for measurement of stress levels. Instituto de Salud Carlos III (ISCIII) - FIS-2012. PI12/00533. PI: Francisco José Cambra Lasaosa. 2013 - 2017.
- Analysis of a diagnostic algorithm for severe pneumonia in critical paediatric patients by means of lung ultrasound scanning and procalcitonin as a plan for the improvement of healthcare quality. Instituto de Salud Carlos III (ISCIII) - Proyectos de investigación en salud. PI16/01040. PI: Iolanda Jordán García. 2017 - 2019.

- Contrato predoctoral de Formación en Investigación. PFIS. Ministerio de Economía y Competitividad. Expediente n FI17/00253. Convocatoria 2017 de la Acción Estratégica en Salud 2013 - 2016. PI: Sara Bobillo Pérez.

### Selected clinical trials

- Codi de protocol: MET51. Phase III. Immunogenicity and safety of a quadrivalent anti-meningococcal conjugate vaccine in research on infants aged 12 to 23 months. EUDRACT: 2016-000749-30. Promoter: Sanofi-Aventis S.A. IP: Iolanda Jordán García. 15/12/2016 -

### Selected publications

- Casanova MA, Monteagudo-Sánchez A, Rodríguez-Guerineau L, Court F, Serrano IG, Martorell-Sampol L, et al. Maternal mutations of FOXF1 cause alveolar capillary dysplasia despite not being imprinted. Hum Mutat 2017; 38: 615-620. FI: 4,601(Q1).
- Casas-Alba D, Fernández de Sevilla-Estrach M, Valero-Rello A, Fortuny-Guasch C, García-García JJ, Ortez-Gonzalez CI, et al. Outbreak of brainstem encephalitis associated with enterovirus-A71 in Catalonia, Spain [2016]: a clinical observational study in a children's reference centre in Catalonia. Clin Microbiol Infect 2017; 23: 874-881. FI: 5,292(Q1).

- Domínguez A, Soldevila N, Caylà JA, García-Cenoz M, Ferrús G, Sala-Farré MR, et al. Assessment of clinical symptoms in household contacts of confirmed pertussis cases. J Infect 2017; 75: 426-432. FI: 4,201(Q1).
- Kneyber MCJ, de Luca D, Calderini E, Jarreau PH, Javouhey E, Lopez-Herce J, et al. Recommendations for mechanical ventilation of critically ill children from the Paediatric Mechanical Ventilation Consensus Conference (PEMVECC). Intensive Care Med 2017; 43: 1764-1780. FI: 12,015(Q1).

### Networks

- Multicenter study: prophylaxis against thrombosys practice (PROTRACT).
- Groupe français respiratoire GFRUP
- PALICC (Pediatric Acute Lung Injury Consensus Conference)
- Respiratory group of ESPNIC society



FIRST QUARTILE PUBLICATIONS

## INFLUENCE OF THE ENVIRONMENT ON CHILD AND ADOLESCENT WELFARE

Carles Luaces Cubells, MD, PhD

*Group focused on research in the following spheres:*

- Study of the epidemiologic and clinical factors of childhood accidents.
- Differential diagnosis of child mistreatment.
- Early detection and secondary prevention of social-health issues in adolescence: drugs, sexuality and violence.
- Impact of external agents (radiation, pollution, etc.) on child health.
- Improvement of paediatric patient care in medical visits: presence of parents, analgesia, differentiated care by age (children/adolescents), etc.

### Members

#### Staff researcher

Andrea Aldemira Liz, Gemma Claret Teruel,  
Ana Isabel Curcoy Barcenilla, Eva Gargallo Burriel,  
Lidia Martínez Sánchez, Sergi Navarro Vilarrubi,  
Marta Simó Nebot, Victoria Trenchs Sainz de la Maza

#### Assistant researcher

David Muñoz Santanach

### Selected clinical trials

- D3820C00016. A Phase I, Open-Label, Multicentre Study to Assess the Pharmacokinetics and Safety of Naloxegol in Paediatric Patients Ages ? 6 Months to < 18 Years Receiving Treatment with Opioids. EUDRACT: 2013-003935-32. Promoter: Astrazeneca Farmaceutica Spain, S.A. PI: Sergi Navarro Vilarrubi. 07/07/2014 -

### Selected publications

- Altal K, Ruiz-Artacho P, Trenchs-Sainz de la Maza V, Martínez Ortiz de Zárate M, Navarro C, Fernández C, et al. Hospital emergency room diagnosis of acute appendicitis in patients aged; 2 to 20 years: the INFURG-SEMES score from the emergency infections; study of the Spanish Society of Emergency Medicine. *Emergencias* 2017. 29: 231-236. FI: 3,028(Q1).
- Casas-Alba D, Fernández de Sevilla-Estrach M, Valero-Rello A, Fortuny-Guasch C, García-García JJ, Ortez-Gonzalez CI, et al. Outbreak of brainstem encephalitis associated with enterovirus-A71 in; Catalonia, Spain (2016): a clinical observational study in a children's; reference centre in Catalonia. *Clin Microbiol Infect* 2017. 23: 874-881. FI: 5,292(Q1).

- Hernández-Bou S, Trenchs-Sainz de la Maza V, Vanegas MI, Valls-Lafon A and Luaces-Cubells C. Evaluation of the bedside Quikread go® CRP test in the management of febrile infants at the emergency department *Eur J Clin Microbiol Infect Dis* 2017. 36: 1205-1211. FI: 2,727(Q2).
- Parra C, Vidiella N, Marín-Valencia I, Trenchs-Sainz de la Maza V and Luaces-Cubells C. Patient experience in the pediatric emergency department: do parents and; children feel the same?. *Eur J Pediatr* 2017. 176: 1263-1267. FI: 1,921(Q2).

### Networks

- Research in pediatric emergency medicine (REPEM) network (The European Paediatric Emergency Medicine Research Network. Pediatric Section of the European Society for Emergency Medicine)
- Red De Investigación De La Sociedad Española De Urgencias De Pediatría / Spanish Pediatric Emergency Research Group (RISeuP/SPERG)



## FOETAL ENVIRONMENT AND OBSTETRICAL COMPLICATION

Maria Dolores Gómez Roig, MD, PhD

With the creation of BCNatal (2013), we have achieved the integration of healthcare in our speciality with research and teaching. We continue to conduct research within the Maternal-Children's Health and Development Network (SAMID Network) financed by ISCIII. We are in charge of the line of research "Effects of environment on foetal growth" in Fetal I+D, Foetal Medicine Foundation Barcelona, and the research group "Foetal environment and obstetrical complications" of the Sant Joan de Déu Research Institute. Our research path and objectives are focused on the foetal environment, placental pathology and foetal growth restriction. We are member experts of the Work Group on Reproductive and Environmental Health of FIGO (International Federation of Gynaecology and Obstetrics, 2016-2019). Likewise, we cooperate with IsGlobal (Barcelona Institute for Global Health). It may be noted that we have now obtained the acceptance of the HEI Research Committee (Health Effects Institute, USA) for issue RFA 17-1 (November 2, 2017): "Traffic-related air pollution and birth weight: the roles of noise, placental function, green space, diet, physical activity, and socioeconomic status (FRONTIER)".

### Members

#### Staff researcher

Isidora Andujar Tore, Jose María Boguña Ponsa, Sergio Cabré Gili, Carolina Esteve Matanza, Jaume Ignasi Miñano Masip, Elisabet Palacios, María José Tojo Grandio

#### Postdoc – associate researcher

Silvia – Irena Ferrero Martínez, Edurne Mazarico Gallego, Miriam Pérez Cruz, Joan Sabrià Bach

#### PhD Student

Laura Almeida Toledo, Edda Marimon Garcia, Isabel Miró Sotelo

### Selected projects

- BR201402. Assessment of prenatal exposure to abuse substances and environmental toxics in foetuses with intrauterine growth restriction and its influence on placental pathology. Hospital Sant Joan de Déu - Esplugues HSJD. Beques de recerca HSJD. BR201402. PI: Maria Dolores Gómez Roig. 2015 - 2018.

- PFIS0187. Treatment of early IGR with low molecular weight heparin (TRACIP): randomised clinical trial. Instituto de Salud Carlos III (ISCIII). Proyectos de investigación en salud. PI: Edurne Mazarico Gallego. 2017 - 2019.
- PFIS0189. Maternal-Child Health and Development Network. Instituto de Salud Carlos III (ISCIII). RETICS. RD16/0022/0014. PI: Maria Dolores Gómez Roig. 2017 - 2021.

### Selected clinical trials & observational studies

- FSJD-TRACIP-2017. Phase III. Treatment of early intrauterine growth restriction with low-molecular-weight heparin: randomised clinical trial (TRACIP). EUDRACT: 2017-001905-33. Promoter: Fundació Privada per a la Recerca i la Docència Sant Joan de Déu - FSJD FSJD. PI: Edurne Mazarico Gallego. 2017 -
- Observational study. Study on the conditions of use of Cariban ® and other antiemetics in the treatment of nausea and vomiting in pregnancy: PROSPECTIVE STUDY OBSERVATIONAL LONGITUDINAL. Promoter: Laboratorios Inibsa, S.A. PI: Jose María Boguña Ponsa. 2016 -

### Selected publications

- Mazarico-Gallego E, Llurba E, Cumplido R, Valls-Lafon A, Melchor JC, Iglesias M, Cabero L, Gratacós E and Roig MG. Neural injury markers in intrauterine growth restriction and their relation to perinatal outcomes. Pediatr Res 2017. 82: 452-457. FI: 2,882(Q1).
- Sabra S, Gratacós E and Gómez-Roig MD. Smoking-Induced Changes in the Maternal Immune, Endocrine, and Metabolic Pathways and Their Impact on Fetal Growth: A Topical Review. Fetal Diagn Ther 2017. 41: 241-250. FI: 2,699(Q2).
- Sabra S, Malmqvist E, Saborit A, Gratacós E and Gómez-Roig MD. Heavy metals exposure levels and their correlation with different clinical forms of fetal growth restriction. PLoS One 2017. FI: 2,806(Q1).
- Sabria-Bach J, Guirado L, Miró I, Gómez-Roig MD and Borrell A. Crown-rump length audit plots with the use of operator-specific PAPP-A and beta-hCG median MoM. Prenat Diagn 2017. 37: 229-234. FI: 2,523(Q2).

### Networks

- 2009 - Red SAMID-RETIC
- 2013 - BCNatal



## IMPACT OF CONGENITAL HEART DISEASE ON DEVELOPMENT: FROM FOETUS TO ADULT

Joan Sanchez de Toledo, MD, PhD

### Selected publications

- Gil-Juanmiquel L, Gratacós M, Castilla-Fernández Y, Piquerias J, Baust T, Raguer N, et al. Bedside Ultrasound for the Diagnosis of Abnormal Diaphragmatic Motion in Children After Heart Surgery. *Pediatr Crit Care Med* 2017; 18: 159-164. FI: 3,495(Q1).
- Pérez-Casares A, Cesar S, Brunet-Garcia L and Joan Sanchez de Toledo. Echocardiographic Evaluation of Pericardial Effusion and Cardiac Tamponade. *Front Pediatr* 2017; 5: 79-0. FI: 2,172(Q2).
- Trakas E, Domnina Y, Panigrahy A, Baust T, Callahan PM, Morell VO, et al. Serum Neuronal Biomarkers in Neonates With Congenital Heart Disease Undergoing Cardiac Surgery. *Pediatr Neurol* 2017; 72: 56-61. FI: 2,018(Q2).



PERSONAL  
RESEARCHER



PUBLICATIONS  
WITH IF



FIRST QUARTILE  
PUBLICATIONS

## PAEDIATRIC ARRHYTHMIAS, SUDDEN DEATH AND CARDIOVASCULAR GENETICS

Georgia Sarquella Brugada, MD, PhD

### Members

#### Staff researcher

Esther Carro Fernandez, Sergio César Díaz, Isaac Moll Adrian



PERSONAL  
RESEARCHER



CLINICAL TRIALS  
& OBSERVATIONAL  
STUDIES



PUBLICATIONS  
WITH IF



FIRST QUARTILE  
PUBLICATIONS

### Selected publications

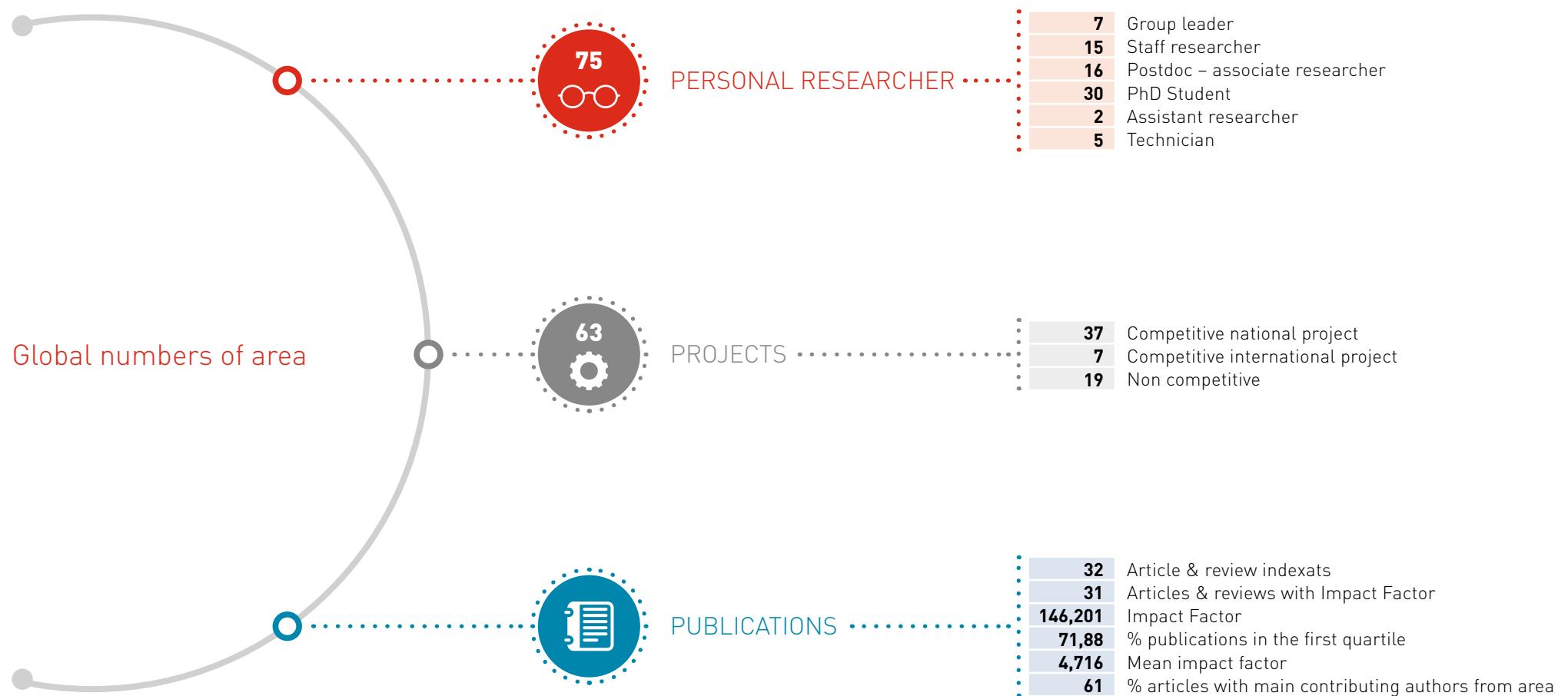
- Campuzano O, Sanchez-Molero O, Fernández A, Mademont-Soler I, Coll M, Perez-Serra A, et al. Sudden Arrhythmic Death During Exercise: A Post-Mortem Genetic Analysis.. *Sports Med* 2017; 47: 2101-2115. FI: 6,832(Q1).
- Hernandez-Ojeda J, Arbelo E, Borras R, Berne P, Tolosana JM, Gomez-Juanatey A, et al. Patients With Brugada Syndrome and Implanted Cardioverter-Defibrillators: Long-Term Follow-Up.. *J Am Coll Cardiol* 2017; 70: 1991-2002. FI: 19,896(Q1).
- Milman A, Andorin A, Gourraud JB, Sacher F, Mabo P, Kim SH, et al. Age of First Arrhythmic Event in Brugada Syndrome: Data From the SABRUS (Survey on Arrhythmic Events in Brugada Syndrome) in 678 Patients.. *Circ Arrhythm Electrophysiol* 2017;10 : e005222 . FI: 5,410(Q1).
- Pappone C, Brugada-Terradellas J, Vicedomini G, Ciccone G, Manguso F, Saviano M, et al. Electrical Substrate Elimination in 135 Consecutive Patients With Brugada Syndrome.. *Circ Arrhythm Electrophysiol* 2017;10 : e005053 . FI: 5,410(Q1)

## **4** CROSS-SECTIONAL AREAS

- 4.1. Genetics, molecular biology and gene therapy
- 4.2. Neuropsychology, cognitive and development neuroscience
- 4.3. Biomedical engineering
- 4.4. Epidemiology

## 4.1. Genetics, molecular biology and gene therapy

**Coordinator:** Marçal Pastor-Anglada



## MOLECULAR BIOLOGY AND GENE REGULATION OF ADIPOSE TISSUE AND ITS PATHOLOGIES

Francesc Villarroya Gombau, PhD

*In 2017 we kept the greater part of our research focused on the identification and characterisation of new molecules capable of acting as regulators in the pathophysiology of metabolism and adiposity. We have developed new approaches based on transcriptomics and bioinformatic prediction in both animal models and samples of cohorts of human patients and volunteers, including study cohorts at Hospital Sant Joan de Déu. We have expanded our international cooperation and new national and international cooperative projects have been obtained.*

### Members

#### Group leader

Marta Giralt Oms, Anna Planavila Porta

#### Staff researcher

Roser Iglesias Coll, Teresa Mampel Astals, Marion Peyrou, Anna Planavila Porta

#### Postdoc – associate researcher

Montserrat Cairó Calzada, Rubén Cairó Calzada, Aleix Gavaldà Navarro, Ricardo Moure Ortega, Joan Villarroya Terrade

#### PhD Student

Laura Campderrós Traver, Gemma Ferrer Curriu, Samantha Morón Ros, Tania Quesada López, Celia Helena Rupérez Gonzalo

#### Technician

Mercedes Morales Rueda, Gemma Moreno Besora, Albert Però Garcia

### Selected projects

- Role of the novel cardiomyokines FGF21 and Metrni on cardiac damage induced by alcoholism and arterial hypertension. Fundació La Marató de TV3 - Ajuts econòmics a projectes de recerca de la Fundació Marató de TV3. 20153330. PI: Ana Planavila Porta. 2016 - 2018.
- Identification and characterisation of new factors with systemic and local action responsible for the healthy metabolic effects of activation of brown adipose tissue. 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. SAF2014-55725-R. PI: Francesc Villarroya Gombau. 2015 - 2017.
- Molecular and cellular bases of changes in the FGF23/alpha-Klotho system in connection with bone, adipose and metabolic dysfunctions in HIV patients. Ministerio de Economía y Competitividad - Ajuts de l'Acció Estratégica en Salut: Subprograma de projectes d'investigació en salut (PI). PI14/00063. PI: Marta Giraltoms. 2015 - 2017.

### Selected publications

- Martínez-Sánchez N, Seoane-Collazo P, Contreras C, Varela L, Villarroya J, Rial-Pensado E, et al. Hypothalamic AMPK-ER Stress-JNK1 Axis Mediates the Central Actions of Thyroid Hormones on Energy Balance. *Cell Metab* 2017; 26: 212-229 . FI: 18,164(Q1).
- Villarroya F, Gavalda A, Peyrou M, Villarroya J and Giralt M. The Lives and Times of Brown Adipokines. *Trends Endocrinol Metab* 2017; 28: 855-867. FI: 10,893(Q1).
- Sánchez-Infantes D, Cereijo R, Peyrou M, Piquer-Garcia I, Stephens JM and Villarroya F. Oncostatin m impairs brown adipose tissue thermogenic function and the browning of subcutaneous white adipose tissue. *Obesity (Silver Spring)* 2017; 25: 85-93. FI: 3,873(Q1).
- Villarroya F, Cereijo R, Villarroya J, Giralt M. Brown adipose tissue as a secretory organ. *Nat Rev Endocrinol*. 2017 Jan;13(1):26-35. doi: 10.1038/nrendo.2016.136. Epub 2016 Sep 12. Review.

### Networks

- CIBEROBN
- 2016 - 2017 - ADIPOPLAST (BFU2015-70454-REDT)



## HUMAN MOLECULAR GENETICS I

Daniel Raúl Grinberg Vaisman, PhD

The research of this group includes the study of complex genetic diseases. Studies are currently being made of association with various pathologies using gene markers and candidate loci in order to identify genetic risk factors and to determine the molecular basis of their contribution to pathology.

We investigate: Lysosomal Diseases, Costello Syndrome, Retinitis pigmentosa, Homocystinuria, Multiple Osteochondromatosis, Osteoporosis, Migraines, Autism, Attention Deficit Hyperactivity Disorder, Opitz C Syndrome.

### Members

#### Staff researcher

Susana Balcells Comas, Bru Cormand Rifà,  
Lluïsa Vilageliu Arqués

#### Postdoc – associate researcher

Noelia Fernández Castillo, Raquel Rabionet Janssen,  
Bárbara Torrico Avilés, Roser Urreizti Frexedas

#### PhD Student

Noelia Benetó Gandia, Judit Cabana Domínguez,  
Núria Martínez Gil, Laura Pineda Cirera,  
Neus Roca Ayats, Anu Shivalikanjli

#### Technician

Mónica Cozar Morillo

### Selected projects

- Comorbid Conditions of Attention-Deficit Hyperactivity Disorder (CoCA). Unió Europea (H2020 - Horizon 2020 (Program: Personalising Health and Care). 667302. PI: Bru Cormand Rifà. 2016 - 2020.
- Synergies in the genetic study and the search for treatments for Opitz C syndrome, atypical femoral fracture, osteoporosis and lysosomal disorders. Ministerio de Economía y Competitividad - Convocatòria d'ajuts del State Programme of Research, Development and Innovation Addressed to the Challenges of Society. Modalitat Projectes I+D+I. SAF2016-75948-R. PI: Susana Balcells Comas. 2016 - 2019.
- Genetics and epigenetics of psychiatric disorders: Autism Spectrum Disorders, Attention-Deficit/Hyperactivity Disorder and Substance Use Disorders. 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. SAF2015-68341-R. PI: Bru Cormand Rifà. 2016 - 2018.

### Selected publications

- Urreizti R, Cueto-Gonzalez, Ana Maria, Franco-Valls, Héctor, Mort-Farre, Silvia, Roca N, Ponomarenko, Julia, et al. De Novo Nonsense Mutation in MAGEL2 in a Patient Initially Diagnosed as Opitz-C: Similarities Between Schaaf-Yang and Opitz-C Syndromes. Sci Rep 2017. 7: 44138-0. FI: 4,259(Q1).



### Networks

- CIBERER
- GEFOS/GENOMOS (international consortium for the study of the genetic bases of osteoporosis)
- European Network and Registry for Homocystinuria and Methylation Defects
- IMpACT (International Multicenter Persistent ADHD Collaboration)
- IHGC (International Headache Genetics Consortium)
- PGC (Psychiatric Genomics Consortium)
- ECNP (European College of Neuropsychopharmacology) Network: ADHD across the lifespan

## HUMAN MOLECULAR GENETICS II

Roser González Duarte, PhD

*This group concerns monogenic disorders associated to different degrees of genetic heterogeneity, from those of lysosomal origin to retinitis pigmentosa. Our first aim was to identify new candidate genes, disease-causing mutations and to analyse the structural and functional features of the mutant alleles. As a continuation of our work, we aim to perform functional studies at the cellular and tissular level, the construction of animal models and new strategies for diagnosis and therapy.*

### Members

#### Staff researcher

Gemma Marfany Nadal

#### PhD Student

Elena Borrego Domènech, Marta de Castro Miró,  
Vasileios Toulis

### Selected projects

- Animal models for retinal dystrophies: strategies of genomic edition, phenotyping and nanotherapy. Ministerio de Economía y Competitividad. Proyectos de I+D+I 2016, programa estatal de investigación, desarrollo e innovación orientada a los retos de la Sociedad. SAF2016-80937-R. PI: Gemma Marfany CoPI: Roser Gonzalez Duarte. 2016 - 2019.
- Research contracts/services. Spark Therapeutics, Inc. 309196. PI: Roser Gonzalez Duarte. 2017 - 2018.
- Dissecting protein trafficking in retinal neurodegeneration by super-resolution imaging on animal models and human iPSCs. Fundació La Marató de TV3. Ajuts econòmics a projectes de recerca de la Fundació Marató de TV3. 20141730. PI: Roser Gonzalez Duarte. 2016 - 2018.

### Selected publications

- Albuixech-Crespo B, Herrera-Úbeda C, Marfany G, Irimia M and Garcia-Fernández J. Origin and evolution of the chordate central nervous system: insights from amphioxus genoarchitecture. Int J Dev Biol 2017. 61: 655-664. FI: 1,981(Q3).

- De Lecuona, I.; Casado, M.; Marfany, G.; López-Baroni, M.J.; Escarrabill, M. Gene editing in humans: Towards a global and inclusive debate for responsible research. Yale Journal of Biology and Medicine. 90 - 4, pp. 673 - 681 (USA): 2017. ISSN 0044-0086
- López-Baroni, M.J.; Marfany, G.; De Lecuona, I.; Corcoy, M.; Boada, M.; Royes, A.; Santaló, J.; Casado, M. La edición genómica aplicada a seres humanos: aspectos éticos, jurídicos y sociales. Revista de Derecho y Genoma Humano. 46, pp. 317 - 340 (2017). ISSN 1134-7198
- Marfany, G. Genoma i exoma clínic (WGS i WES): retorn dels resultats genètics sol·licitats, quan, com i per què?. Treballs de la Societat Catalana de Biologia. 67, pp. 33 - 37. (España): Societat Catalana de Biologia, 2017. ISSN 0212-3037

### Networks

- CIBERER- UNITAT 718
- 2013 - 2018 - COST ACTION BM1307 - PROTEOSTASIS (Member of the Core Management Group)



PERSONAL  
RESEARCHER



PROJECTS



PUBLICATIONS  
WITH IF

## MOLECULAR PHARMACOLOGY AND EXPERIMENTAL THERAPIES

Marçal Pastor-Aneglada, PhD

*Our research group is studying diverse aspects of the cellular and molecular biology of membrane transporters, essentially in epithelial cells and in cells of the immune system. Two of the transporter gene families currently under study are CNT and ENT transporters responsible for the uptake of natural nucleosides and a wide range of drugs used in the treatment of cardiovascular diseases and antitumor therapies and antivirals.*

### Members

#### Staff researcher

Sandra Perez-Torras

#### Postdoc – associate researcher

Nerea Urtasun Plans

#### PhD Student

Hamsa Banjer, Clara Boces Pascual,  
Olga Casulleras-Gras, Liska Caviedes Cárdenas,  
Aida Mata Ventosa, Sandra Moro-Villa,  
Catalina Perelló Reus, Albert Viel Oliva

### Selected projects

- Functional dissection of the properties of human concentrative nucleoside transporter (hCNT) proteins. 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. SAF2014-52067-R. PI: Marçal Pastor-Aneglada. 2015 - 2017.
- Identification of the nucleotide cellular sensor and its interactome. Ramon Areces Foundation. PI: Marçal Pastor-Aneglada. 2017 - 2018.

### Selected publications

- Urtasun N, Boces-Pascual C, Boix L, Bruix J, Pastor-Aneglada M and Perez-Torras S. Role of drug-dependent transporter modulation on the chemosensitivity of cholangiocarcinoma. *Oncotarget* 2017. 8: 90185-90196. FI: 5,168(Q1).
- Arimany-Nardi C, Claudio-Montero A, Viel-Oliva A, Schmidtko P, Estarellas C, Barril X, Bidon-Chanal A and Pastor-Aneglada M. Identification and Characterization of a Secondary Sodium-Binding Site and the Main Selectivity Determinants in the Human Concentrative Nucleoside Transporter 3. *Mol Pharm* 2017. 14: 1980-1987. FI: 4,440(Q1).
- Minuesa G, Albert C, Pastor-Aneglada M, Martinez-Picado J and Koepsell H. Response to "Tenofovir Disoproxil Fumarate Is Not an Inhibitor of Human Organic Cation Transporter 1". *J Pharmacol Exp Ther* 2017. 360: 343-345. FI: 3,867(Q1).

### Networks

- CIBER-EHD



PERSONAL  
RESEARCHER



PROJECTS



PUBLICATIONS  
WITH IF



FIRST QUARTILE  
PUBLICATIONS

## PHARMACOLOGICAL TARGETS IN INFLAMMATION AND METABOLIC DISEASES

Manuel Vázquez-Carrera, PhD

In 2017 our group has reported that very low-density lipoproteins (VLDL) and apolipoprotein CIII, which are elevated in diabetic states, induce endoplasmic reticulum stress and inflammation and attenuate insulin signalling via Toll-like receptor 2 in mouse skeletal muscle cells. In addition, we have demonstrated that upregulation of small heterodimer partner (SHP) upon high-fat feeding leads to lipid accumulation, insulin resistance and inflammation in cardiomyocytes.

### Members

#### Staff researcher

F. Xavier Palmer Tarridas, Maria Silvia Román Azcona

#### Postdoc – associate researcher

Emma Barroso Fernández, Marta Montori Grau,  
Lucía Peña Moreno, Javier Pizarro Delgado

#### PhD Student

Gaia Botteri, Mohammad Zarei

### Selected projects

- Inflammation, insulin resistance and diabetic cardiomyopathy: assessment of PPARbeta/delta agonists and oleic acid. 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. SAF2015-64146-R. PI: Manuel Vazquez Carrera. 2016 - 2018.
- Diabetic cardiomyopathy: searching for a therapeutic target. Fundació La Marató de TV3 - Ajuts econòmics a projectes de recerca de la Fundació Marató de TV3. 20154230. PI: Manuel Vazquez Carrera. 2016 - 2018.
- HRI activators for treatment of type 2 diabetes mellitus and other associated disorders. PoC\_2016\_002. Fondo impulso innovación (F2I). Fundación Bosch i Gimpera. Universidad de Barcelona. - convocatòria Prova de Concepce, Fons per a l'Impuls de la Innovació-Bosch i Gimpera. 309109. PI: Manuel Vazquez Carrera. 2017 - 2018.

### Selected publications

- Botteri G, Montori M, Gumà A, Pizarro J, Cedó L, Escolà-Gil JC, et al. VLDL and apolipoprotein CIII induce ER stress and inflammation and attenuate insulin signalling via Toll-like receptor 2 in mouse skeletal muscle cells. Diabetologia 2017; 60: 2262-2273. FI: 6,080(Q1).
- Rodríguez-Calvo R, Tajes M and Vazquez M. The NR4A subfamily of nuclear receptors: potential new therapeutic targets for the treatment of inflammatory diseases. Expert Opin Ther Targets 2017; 21: 291-304. FI: 4,873(Q1).
- Rodríguez-Calvo R, Chanda D, Oligschlaeger Y, Miglianico M, Coumans WA, Barroso E, et al. Small heterodimer partner (SHP) contributes to insulin resistance in cardiomyocytes. Biochim Biophys Acta 2017; 1862: 541-551. FI: 5,547(Q1).

### Networks

- 2007 - CIBERDEM



PERSONAL  
RESEARCHER



PROJECTS



PUBLICATIONS  
WITH IF



FIRST QUARTILE  
PUBLICATIONS

## MICROBIOTA INTESTINAL

Laura Baldomà Llavinés, PhD

*Our research group has as objective the study of several phenomena related to the communication or dialogue between the microbiota and the intestinal epithelium. The study is framed within a very current theme that tries to elucidate the complex interactions between the host and the bacterium, as well as its association with pathophysiological phenotypes. The relevant implication of the microbiota in health and disease suggests that the modulation of its composition by different means may constitute a new therapeutic approach.*

### Members

#### Staff researcher

Josefa Badia Palacin, Rosa Giménez Claudio

#### Postdoc – associate researcher

Laura Aguilera Gil

#### PhD Student

Carina Shianya Alvarez Villagómez,

Mª Alexandra Cañas Pacheco,

Mª José Fábrega Fernández

### Selected projects

- Microbiota-derived outer membrane vesicles and Gram-negative probiotics as modulating agents of the intestinal barrier and immune response. 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. AGL2016-79113-R. PI: Laura Baldoma Llavines. 2016 - 2019.
- Cooperation agreement for the performance of the research project "Study for the regulation of the NHD gene in Escherichia coli". Universidad Nacional de Tucumán - Sense. PI: Laura Baldoma Llavines. 2009 - 2018.

### Selected publications

- Fabrega MJ, Rodríguez-Nogales A, Garrido-Mesa J, Algieri F, Badia J, Gimenez R, et al. Intestinal Anti-inflammatory Effects of Outer Membrane Vesicles from *Escherichia coli* Nissle 1917 in DSS-Experimental Colitis in Mice. *Front Microbiol* 2017. 8: 1274-0. FI: 4,076(Q1).
- Bauer E, Domingo X, Balcells C, Polat IH, Crespo M, Quirante J, et al. Synthesis, characterization and biological activity of new cyclometallated platinum(IV) iodido complexes. *Dalton Trans* 2017. 46: 14973-14987. FI: 4,029(Q1).
- Albert J, Granell J, Duran A, Lozano J, Luque A, Mate A, et al. Endo and exo cyclopalladated (E)-N-[[1,1'-biphenyl]-2-yl]-1-mesitylmethanimines: Anticancer, antibacterial and antioxidant activities. *J Organomet Chem* 2017. 839: 116-125. FI: 2,184(Q2)



PERSONAL  
RESEARCHER



PROJECTS



PUBLICATIONS  
WITH IF



FIRST QUARTILE  
PUBLICATIONS

## STEM CELL AND NEURAL PLASTICITY

Antonella Consiglio, PhD

*Our laboratory is interested in adult neuroplasticity and regeneration in the brain. We are interested in understanding the function and the potential of these newly born cells in physiology and disease. The main goal pursued in our laboratory is to identify part of the multiple signals that are responsible for endogenous precursor division, differentiation, circuit integration and survival in different regions of the brain, and to validate the possibility to manipulate neural precursors toward therapeutic neuronal repopulation for the cure of different degenerative CNS pathologies.*

### Members

#### Postdoc – associate researcher

Neus Bayo Puxan

#### Assistant researcher

Armida Faella, Irene Fernández Carasa

#### PhD Student

Carles Calatayud Aristoy, Giulia Carola, Angelique Di Domenico

### Selected projects

- Molecular, cellular and functional characterisation of dopaminergic neurons derived from iPS cells of Parkinson's disease patients. Ministerio de Economía y Competitividad - 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. BFU2016-80870-P. PI: Antonella Consiglio. 2016 - 2019.
- Elucidating early pathogenic mechanisms of neurodegeneration in Parkinson's disease through a humanized dynamic in vitro model (PD-HUMMODEL). Unió Europea - 7è Programa Marc (2007-2013). 311736. 2013 - 2018.

### Selected publications

- Calatayud Aristoy C, Carola G, Consiglio A and Raya A. Modeling the genetic complexity of Parkinson's disease by targeted genome edition in iPS cells. *Curr Opin Genet Dev* 2017. 46: 123-131. FI: 5,825(Q1).
- Mora C, Serzanti M, Consiglio A, Memo M and Dell'Era P. Clinical potentials of human pluripotent stem cells. *Cell Biol Toxicol* 2017. 33: 351-360. FI: 2,333(Q2).



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PROJECTS



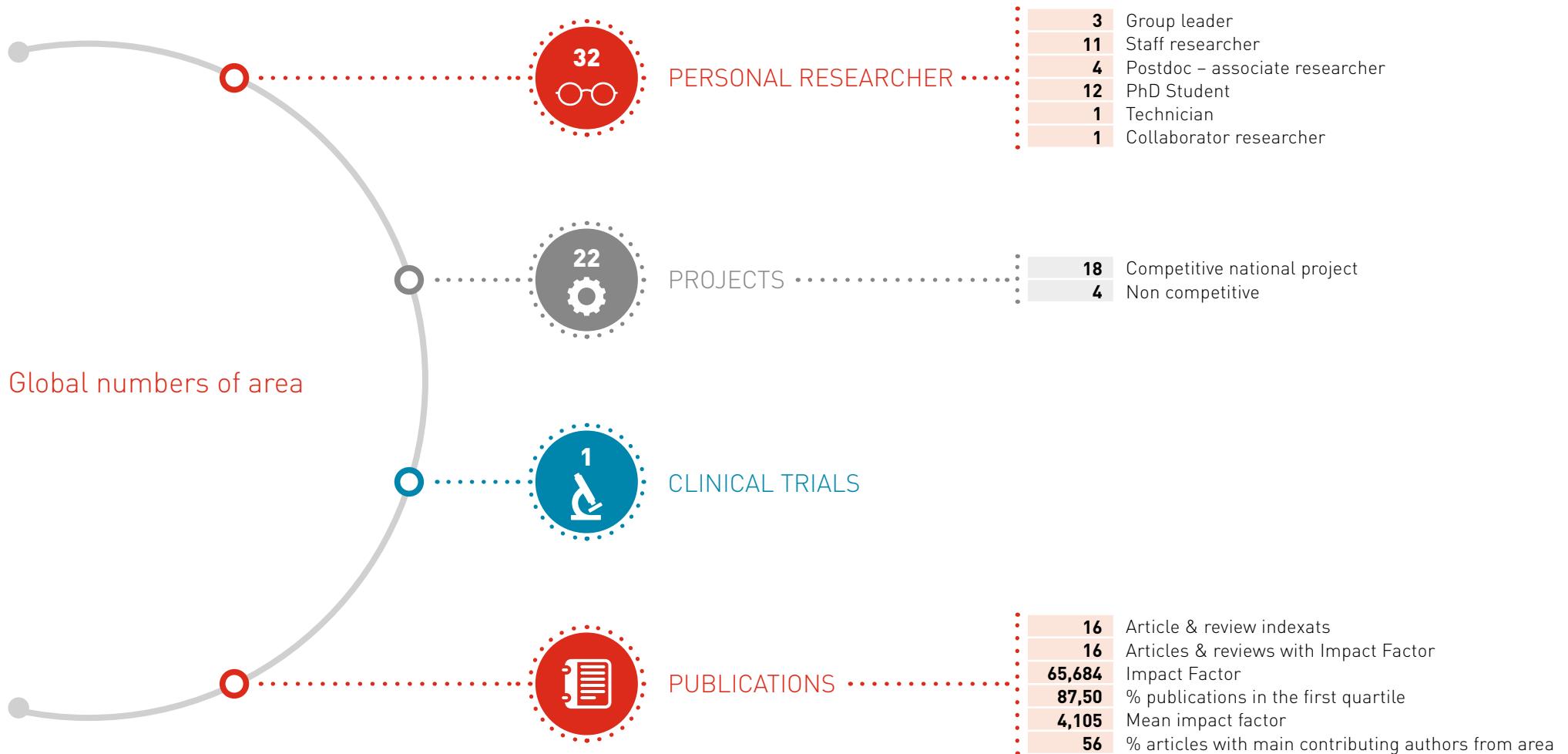
PUBLICATIONS WITH IF



FIRST QUARTILE PUBLICATIONS

## 4.2. Neuropsychology, cognitive and development neuroscience

**Coordinator:** Carles Escera



## NEUROPSYCHOLOGY

Maria Mataró Serra, PhD

*Neuropsychology implies a clinical application of this knowledge, because it studies how brain injuries or disorders affect cognitive functions and behaviour. Research groups working in this area analyse changes in cognition associated with aging and degenerative illness, as well as the mechanisms of brain reorganization after a traumatic brain injury. We aim to investigate how cognitive functions can be improved in patients with mild cognitive impairment using non-invasive brain stimulation techniques. Also, we study the effects of cerebrovascular diseases and the neurobiological mechanisms of neuroprotective interventions, like exercise and cognitive training. Other research lines from this area focus on the differences in central nervous system function between normal-weight and obese people, as well as the coexistence of substance use disorders and severe mental disorders. In addition, we analyse several learning disorders, for instance dyslexia or nonverbal learning disability. Also, we work with patients suffering from cerebral palsy and we study the major impacts in cognitive functions. In these studies we use neuroimaging techniques, genetic studies and cognitive tests.*

### Members

#### Staff researcher

Maria Angeles Jurado Luque, Roser Pueyo Benito, Xavier Caldú Ferrús

#### Postdoc – associate researcher

Cristina Sánchez Castañeda

#### PhD Student

Júlia Ballester Plané, Olga Laporta Hoyos, Jonatan Ottino González, Noemí Lamonja Vicente, Alba Castells Sánchez, Xavier Prats Soteras

### Selected projects

- Study of omics integration of neurobiological effects of physical exercise and cognitive stimulation. 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. PSI2016-77475-R. PI: Maria Mataró Serrat. 2016 - 2019.

- Executive function training in childhood obesity: food choice, quality of life and brain connectivity (TOUCH). Fundació La Marató de TV3. Ajuts econòmics a projectes de recerca de la Fundació Marató de TV3. 122/U/2016. PI: Maria Angeles Jurado Luque. 2017 - 2020.
- Executive training in cerebral palsy: participation, quality of life and brain connectivity. 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. PSI2016-75979-R. PI: Roser Pueyo Benito. 2016 - 2020.

### Selected clinical trials

- Effects of Physical and Cognitive Training in Neuroplasticity, Health and Cognition. Movement Project. Ministerio de Economía y Competitividad. IP: Maria Mataró. 2015-2018. ClinicalTrials.gov Identifier: NCT03123900.

### Selected publications

- Ballester J, Schmidt R, Laporta-Hoyos O, Junqué C, Vázquez É, Delgado I, et al. Whole-brain structural connectivity in dyskinetic cerebral palsy and its association with motor and cognitive function. *Hum Brain Mapp* 2017. 38: 4594-4612. FI: 4,530(Q1).
- Laporta-Hoyos O, Pannek K, Ballester J, Reid LB, Vázquez É, Delgado I, Zubiaurre-Elorza L, et al. White matter integrity in dyskinetic cerebral palsy: Relationship with intelligence quotient and executive function. *Neuroimage Clin* 2017. 15: 789-800. FI: 4,348(Q1).
- Ottino-González J, Jurado MA, García-García I, Segura B, Marqués-Iturria I, Sender-Palacios MJ, et al. Allostatic Load Is Linked to Cortical Thickness Changes Depending on Body-Weight Status. *Front Hum Neurosci* 2017. 11: 639-0. FI: 3,209(Q1).
- Laporta O, Ballester J, Poo P, Macaya A, Meléndez-Plumed M, Vázquez E, et al. Proxy-reported quality of life in adolescents and adults with dyskinetic cerebral palsy is associated with executive functions and cortical thickness. *Qual Life Res* 2017. 26: 1209-1222. FI: 2,344(Q1)



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RESEARCHER



PROJECTS



CLINICAL TRIALS  
& OBSERVATIONAL  
STUDIES



PUBLICATIONS  
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FIRST QUARTILE  
PUBLICATIONS

## COGNITIVE NEUROSCIENCE

Carles Escera Micó, PhD

*Major achievements of the cognitive neuroscience research group during 2017 were in understanding the neural underpinning of basic cognitive brain functions. In particular, in collaboration with University of Salamanca (Dr. M.S. Malmierca) we demonstrated that prediction errors in auditory perception emerge in a hierarchically organized pattern along the auditory system (Parras et al., 2017, *Nature Comm.*), and that conscious and attentive organization of the auditory scene relies on entrainment of neural oscillations to the tagging of auditory event patterns (Costa-Faidella et al., 2017, *Neuroimage*). We have started a new collaborative project with the Women's Area of the Hospital (Dr. Gómez-Roig) to implement the so-called Frequency-Following Response (FFR) as a candidate biomarker for neurocognitive development in fetal growth restrictions. Our group has been recognized and financed as Grup de Recerca Consolidat by the Generalitat de Catalunya (SGR2017-974).*

### Members

#### Staff researcher

Inma Clemente Lapena, María José Corral López, Maribel Nuñez Peña, Iria San Miguel Insúa, Hans Supèr, Marc Via García

#### Postdoc – associate researcher

Raffaele Cacciaglia, Miriam Cornellà Griful

#### PhD Student

Natàlia Gorina Careta, Alejandro Lerer, Teresa Ribas Prats

### Selected projects

- Exploration of the sensory patterns involved in predictive processing, using sensory-motor associations. Ministerio de Economía y Competitividad - Convocatoria 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. PSI2014-52573-P PI: Iria San Miguel Insúa. 2015 - 2017.
- Subcortical contribution to auditory cognition. Ministerio de Economía y Competitividad - Convocatoria 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. PSI2015-63664-P PI: Carles Escera Micó. 2016 - 2018.
- Grant to promote and consolidate existing research excellence at public universities in Catalonia. Programa ICREA Academia 2015. Fundació Institució Catalana de Recerca i Estudis Avançats (ICREA) - Convocatoria del Programa Icrea Acadèmia. SGR2014-1413. PI: Carles Escera Micó. 2016 - 2020.

### Selected publications

- Parras GG, Nieto-Diego J, Carballo GV, Valdés-Baizabal C, Escera C and Malmierca MS. Neurons along the auditory pathway exhibit a hierarchical organization of prediction error. *Nat Commun* 2017. 8: 2148-0. FI: 12,124(Q1).
- Costa-Faidella J, Sussman ES and Escera C. Selective entrainment of brain oscillations drives auditory perceptual organization. *Neuroimage* 2017. 159: 195-206. FI: 5,835(Q1).
- López-Caballero F and Escera C. Binaural Beat: A Failure to Enhance EEG Power and Emotional Arousal. *FRONT HUM NEUROSCI* 2017. 11: 557-0. FI: 3,209(Q1).
- Garcia-Garcia M, Via M, Zarnowiec K, SanMiguel I, Escera C and Clemente I. COMT and DRD2/ANKK-1 gene-gene interaction account for resetting of gamma neural oscillations to auditory stimulus-driven attention. *PLoS One* 2017. 12: e0172362. FI: 2,806(Q1)



83.3%  
FIRST QUARTILE PUBLICATIONS

## DEVELOPMENTAL NEUROSCIENCE

Laura Bosch Galceran, PhD

The group has carried on with the research activities focused on speech perception and language learning: a) the role of audiovisual information in language discrimination in the first year of life; b) the capacity to segment words and to associate them with a referent (6-9 months) in full-term populations and premature populations of different gestational age; c) the response to vowel/consonant changes at three moments in the first year of life (newborns, 6 months and 12 months) with EEG/ERP measurements, comparing full-term and premature infants followed up longitudinally (studies with Dr. T. Agut of neonatology), and d) assessment of language level achieved at 4 years and 7-8 years in different risk populations followed up from the first year of life (patients with neonatal cerebral infarction and children born with low weight, research studies in collaboration with Dr. A. Garcia-Alix and Dr. L. Ibáñez, respectively).

### Members

#### Staff researcher

Ferran Pons Gimeno, Marta Ramon Casas

#### Postdoc – associate researcher

Clément François

#### PhD Student

Joan Birulés Muntané, Maria Teixidó Ibáñez,  
Anna Martínez Álvarez

#### Assistant researcher

Jessica Sánchez-Galán

#### Collaborator researcher

Jorgina Solé Montoliu

### Selected projects

- Multimodal information at the start of language learning: use of audiovisual keys in normotypical development and in neurocognitive risk populations. Ministerio de Economía y Competitividad. 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. PSI2014-55105-P. PI: Laura Bosch Galceran. 2015 - 2018.
- Dynamic for segmenting continuous speech in full-term and premature newborns. Ministerio de Economía y Competitividad. 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. PSI2015-69132-P. PI: Clément François. 2016 - 2017.

### Selected publications

- Vilà-Balló A, François C, Cucurell D, Miró J, Falip M, Juncadella M and Rodríguez-Fornells A. Auditory Target and Novelty Processing in Patients with Unilateral Hippocampal Sclerosis: A Current-Source Density Study. *Sci Rep* 2017. 7: 1612-0. FI: 4,259(Q1).
- François C, Teixidó M, Takerkart S, Agut-Quijano T, Bosch L and Rodriguez-Fornells A. Enhanced Neonatal Brain Responses To Sung Streams Predict Vocabulary Outcomes By Age 18 Months. *Sci Rep* 2017. 7: 12451-0. FI: 4,259(Q1).
- Ramon M, Fennell CT and Bosch L. Minimal-pair word learning by bilingual toddlers: the Catalan /e/-/ɛ/ contrast revisited. *Biling-Lang Cogn* 2017. 20: 649-656. FI: 3,010(Q1).
- Martinez-Alvarez A, Pons F and de Diego-Balaguer R. Endogenous temporal attention in the absence of stimulus-driven cues emerges in the second year of life. *PLoS One* 2017. 12: e0184698 . FI: 2,806(Q1).
- Fernandez Prieto I, Spence C, Pons F and Navarra J. Does Language Influence the Vertical Representation of Auditory Pitch and Loudness?. *Iperception* 2017. 8: 2041669517716183. FI: 1,051(Q4).



PERSONAL  
RESEARCHER



PROJECTS



PUBLICATIONS  
WITH IF



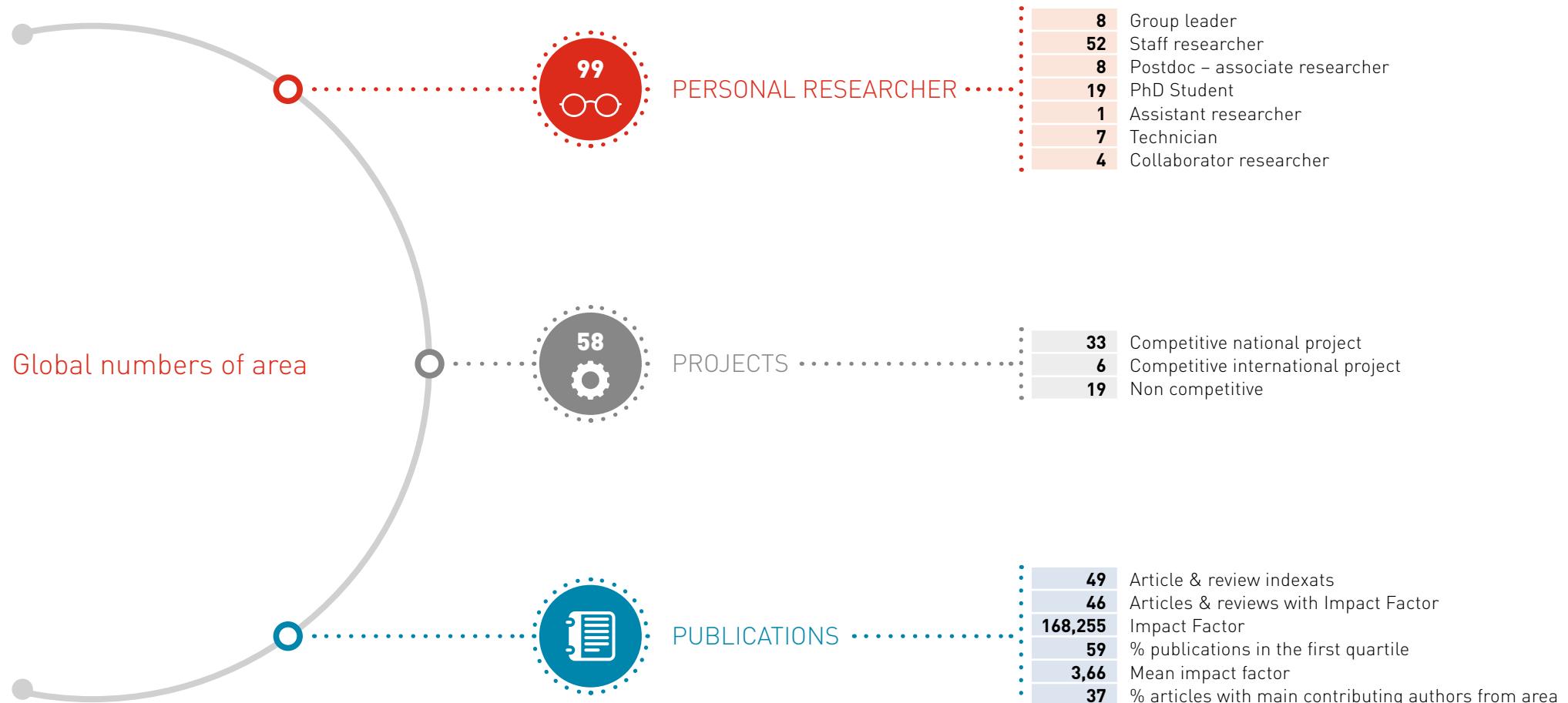
FIRST QUARTILE  
PUBLICATIONS

### Networks

- sGR-1413, 2015-2017, IP: Dr. Antoni Rodríguez-Fornells

## 4.3. Biomedical engineering

**Coordinator:** Pere Caminal



## BIOINFORMATICS AND DATA ANALYSIS PLATFORM

Pere Caminal Magrans, PhD

*It is considered essential to propose a platform for the analysis of data in the broadest sense, including bioinformatics and biosignals. The goal is to establish a bioinformatics platform for the IR-SJD with a twofold aim: to provide the set of research groups with support on massive data analysis and to carry out an active programme of research, providing added value to the platform.*

### Members

#### Staff researcher

Alexandre Perera Lluna, Raul Benítez Iglesias, Jesús Escrivá Muñoz, Giovana Elizabeth Gavidia Bovadilla, Pedro Gomis Román, Alexander Vallmitjana Lees, Montserrat Vallverdú Ferrer

#### PhD Student

Xavier Marimon Serra, Sergi Picart Armada

#### Technician

Maria Maqueda

### Selected projects

- Development of a system for monitoring the responses to nociceptive stimuli during anesthesia based on neurophysiological signals. Ministerio de Economía, Industria y Competitividad - AGAUR. 2014 DI 013. PI: Alexandre Perera Lluna. 2017 - 2017.
- ANVERSO study: continuous automatic analysis of electrocardiographic variables in relation to sudden death risk in Brugada syndrome patients. Ministerio de Economía, Industria y Competitividad - Instituto de Salud Carlos III. 2015 DI 008. PI: Alexandre Perera Lluna. 2017 -2017.
- Development of new algorithms for depth of anesthesia and pain assessment improvement integrating cerebral blood flow measurements. Agència de Gestió d'Ajuts Universitaris i de Recerca - 2016. 2015 DI 033. PI: Montserrat Vallverdú. 2016 - 2019.

### Selected publications

- Gavidia-Bovadilla G, Kanaan-Izquierdo S, Mataró-Serrat M, Perera A and Alzheimer's Disease Neuroimaging Initiative. Early Prediction of Alzheimer's Disease Using Null Longitudinal Model-Based Classifiers. PLoS One 2017.12 : e0168011. FI: 2,806(Q1).
- Maqueda M, Roca E, Brotons D, Soria JM and Perera A. Affected pathways and transcriptional regulators in gene expression response to an ultramarathon trail: Global and independent activity approaches. PLoS One 2017. 12 : e0180322.FI: 2,806(Q1).
- Martin-Fernandez L, Gavidia G, Corrales I, Brunel H, Ramírez L, López S, Souto JC, Vidal F and Soria JM. Next generation sequencing to dissect the genetic architecture of KNG1 and F11 loci using factor XI levels as an intermediate phenotype of thrombosis. PLoS One 2017. 12 : e0176301. FI: 2,806(Q1).
- Picart-Armada S, Fernández-Albert F, Vinaixa M, Rodríguez-García MA, Aivio S, Stracker TH, Yanes O and Perera A. Null diffusion-based enrichment for metabolomics data. PLoS One 2017. 12 : e0189012. FI: 2,806(Q1).



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PROJECTS



PUBLICATIONS  
WITH IF



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PUBLICATIONS

## COMPUTER GRAPHICS

Daniela Tost Pardell, PhD

The activity of the group has focused on the development of three sets of serious games and a gamified app. On one hand, a new educational video-game of the series "Personatges en Joc" has been designed. A pilot study on the efficacy of education through play for children with ADT was carried on with Dr. Alda from HSJD based on the use of this game. On the second hand, a set of video-game for the education of transversal skills at the workplace for persons with intellectual disability was initiated, as the natural continuation of the former project IntegraGame. Finally, a project on the training the perfusion technique was initiated, funded by the Spanish Association of perfusionists. In the project ADALT, funded by Eurecat, the team developed a gamified app to query the emotional state and level of pain of patients with hemiparesis of the upper limb.

### Members

#### Staff researcher

Dolors Ayala Vallespí, Jose Luis Eguía Gómez,  
Robert Joan Arinyó, Núria Pla García,  
Lluís Solano Albages, Marc Vigo Anglada

#### PhD Student

Núria Bonet Codina, Ariel von Barnekow

### Selected projects

- Figures in Play: a set of educational games on the history of Catalonia. Generalitat de Catalunya. 2017. PI: Daniela Tost Pardell. 2017.
- Activities of Daily Augmented Living Toolkit (ADALT). EURECAT. PI: Daniela Tost Pardell. 2016 - 2017.
- Smile@Work: serious games for cross-skills training in the occupational sphere for persons with intellectual disability, financed by the 1st Call for Grants to Research Projects Applied to Development of Accessible Technologies. (Indra y Fundación Universia) PI: Daniela Tost Pardell. 2017 - 2018.
- The Virtual Perfusionist: a serious game for training in the use of the heart-lung pump. Societat Espanyola de Perfusionistes. 2017 - 2018.

### Selected publications

- Alloni A, Sinforiani E, Zucchella C, Sandrini G, Bernini S, Cattani B, Tost Pardell D, Quaglini S and Pistarini C. Computer-based cognitive rehabilitation: the CoRe system. *Disabil Rehabil* 2017. 39: 407-417. FI: 1,804(Q1).
- Bottioli S, Tassorelli C, Lamonica M, Zucchella C, Cavallini E, Bernini S, Sinforiani E, Pazzi S, Cristiani P, Vecchi T, Tost Pardell D and Sandrini G. Smart Aging Platform for Evaluating Cognitive Functions in Aging: A Comparison with the MoCA in a Normal Population. *FRONT AGING NEUROSCI* 2017. 9: 379-0. FI: 4,504(Q1).
- Von Barnekow A, Bonet N and Tost Pardell D. Can 3D Gamified Simulations Be Valid Vocational Training Tools for Persons with Intellectual Disability? An Experiment Based on a Real-life Situation. *Methods Inf Med* 2017. 56: 162-170. FI: 1,772(Q3).



PERSONAL RESEARCHER



PROJECTS



PUBLICATIONS WITH IF



FIRST QUARTILE PUBLICATIONS

## BIO MATERIALS AND TISSUE ENGINEERING

Maria Pau Ginebra Molins, PhD

The Tissue Engineering and Biomaterials (TEB) group published 25 articles in high-impact index journals and two book chapters in 2017.

The TEB members obtained an ERC project, an MSCA, an H2020 NMBP project, an ERA-NET, a MINECO project for young researchers and designation as an SGR group financed by the Government of Catalonia.

In the course of the year, the group members presented 17 papers and 10 guest conferences at international congresses.

Likewise, 6 doctoral theses directed by TEB members were read in 2017.

### Members

#### Staff researcher

Cristina Canal Barnils, Jose María Manero Planella, Carles Mas Moruno, Judith Buxadera Palomero, Montserrat Español Pons, Anna Díez Escudero, Roberta Fraioli, Kanupriya Khurana, Marta Pegueroles Neyra, Daniel Rodríguez Rius, Elisa Rupérez de Gracia, Zhitong Zhao

#### Postdoc – associate researcher

Jordi Guillem Martí, Cédric Labay, Giuseppe Scionti, Anna Khlyustova, Juan Tornín, Aranzazu Villasante

#### Assistant researcher

Xavier Gil Mur

#### PhD Student

Albert Barba Serrahima, Mireia Hoyos Nogués, Joanna Maria Sadowska, Èlia Vidal Girona, Romain Hugues Marie Schieber, Mar Bonany, Inès Hamouda, Joanna Konka, Joaquim Minguela, Santiago Raymond, Diego Torres, Miguel Mateu, Lluís Oliver

#### Technician

Marc Avilés Palau, Meritxell Molmeneu Trias, Mònica Ortiz Hernández, Miquel Punset Fusté, Sergi Torrent Camarero

### Selected projects

- Càtedra Klockner. Soadco-Klockner - 2006-2018. PI: José María Manero Planella. 2006 - 2018.
- Atmospheric Pressure plAsma meets biomaterials for bone Cancer HEaling (APACHE). European Commission - ERC Starting Grants. ERC-2016-STG 714793. PI: Cristina Canal Barnils/ Maria Pau Ginebra. 2017 - 2022.
- Print4Life: "Novel bioprinting strategies for bone regeneration cáncer therapies". Ministerio de Economía, Industria y Competitividad - MINECO. MAT2015-65601- R. PI: Maria Pau Ginebra Molins i Cristina Canal Barnils. 2016 - 2019.

### Selected publications

- Barba A, Díez Escudero A, Maazouz Y, Rappe K, Español M, Montufar EB, Bonany M, Sadowska JM, Guillem J, Öhman-Mägi C, Persson C, Manzanares MC, Franch J and Ginebra MP. Osteoinduction by Foamed and 3D-Printed Calcium Phosphate Scaffolds: Effect of Nanostructure and Pore Architecture. ACS Appl Mater Interfaces 2017. 9: 41722-41736. FI: 7,504(Q1).
- Hoyos-Nogués M, Velasco F, Ginebra MP, Manero JM, Gil FJ, Mas-Moruno C. Regenerating Bone via Multifunctional Coatings: The Blending of Cell Integration and Bacterial Inhibition Properties on the Surface of Biomaterials. ACS Appl Mater Interfaces. 2017 Jul 5;9(26):21618-21630. FI: 7,504 (Q1).



FIRST QUARTILE  
PUBLICATIONS

## PROCESSING AND INTERPRETATION OF BIOMEDICAL SIGNALS

Miquel A. Mañanas Villanueva, PhD

*Analysis and interpretation of signals of cerebral origin obtained by means of non-invasive techniques such as electroencephalography (EEG) and magnetoencephalography (MEG). Analysis of signals of muscular origin as a tool for motor diagnosis and rehabilitation. Modelling and simulation of the respiratory system in order to predict the ventilatory response to different respiratory stimuli and in various conditions which may be the result of pulmonary disorders.*

### Members

#### Staff researcher

Joan Francesc Alonso Lopez, Sergio Romero Lafuente

### Selected projects

- Social network mobile app to promote patient adherence to treatment and healthy lifestyles. Fundación MAPFRE - 2017. PI: Joan F. Alonso/ Joan Ramos. 2017 - 2018.
- Thematic research network on neurotechnologies for care and rehabilitation. MINECO - 2015. DPI2015-69098-REDT. NEUROTEC. PI: José María Azorín Poveda. 2017 - 2017.
- Multimodal analysis for assessment and rehabilitation of disabling neurological disorders. MINECO - 2017. DPI2017-83989-R. PI: Miguel Ángel Mañanas/ Sergio Romero. 2017 - 2018.

### Selected publications

- Jordanic M, Rojas M, Mañanas MA, Alonso Lopez JF and Marateb HR. A Novel Spatial Feature for the Identification of Motor Tasks Using High-Density Electromyography. Sensors (Basel) 2017. 17:7. FI: 2,677(Q1).
- Karimimehr S, Marateb HR, Muceli S, Mansourian M, Mañanas MA and Farina D. A Real-Time Method for Decoding the Neural Drive to Muscles Using Single-Channel Intra-Muscular EMG Recordings. Int J Neural Syst 2017. 27: 1750025-0. FI: 6,333(Q1).
- Ochoa JF, Alonso JF, Duque JE, Tobón CA, Mañanas MA, Lopera F and Hernández AM. Successful Object Encoding Induces Increased Directed Connectivity in Presymptomatic Early-Onset Alzheimer's Disease. J Alzheimers Dis 2017. 55: 1195-1205. FI: 3,731(Q2).
- Sarrafzadegan N, Hassannejad R, Marateb HR, Talaei M, Sadeghi M, Roohafza HR, et al. PARS risk charts: A 10-year study of risk assessment for cardiovascular diseases in Eastern Mediterranean Region. PLoS One 2017. 12 : e0189389 . FI: 2,806(Q1).



PERSONAL  
RESEARCHER



PROJECTS



PUBLICATIONS  
WITH IF



FIRST QUARTILE  
PUBLICATIONS

## INSTRUMENTATION AND BIOENGINEERING

Xavier Rosell Ferrer, PhD

*Design and characterisation of medical and biotechnological instrumentation, of ultrasonic transducers, and of control and monitoring equipment; redesign of equipment and facilities. Guidance on the effect of non-ionising radiations and on compliance with EMC and electrical safety standards and directives.*

### Members

#### Staff researcher

Ramon Bragós Badia, Mireya Fernández Chimeno, Miguel Angel García González, Miguel J García Hernandez, Tomás García Sánchez, Lexa Nescolarde Selva, Juan Ramos Castro, Pere Joan Riu Costa

### Selected projects

- A novel myocardial impedance mapping system for ablation of post infarction ventricular arrhythmias in humans. Fundació La Marató de TV3. MARATO 20150830. PI: Xavier Rosell. 2016 - 2018.
- Translational development of a new system for mapping electric impedance of the myocardium for the catheter ablation of ventricular arrhythmias in myocardial infarction patients. Instituto de Salud Carlos III. 2015. DTS15/00099. PI: Xavier Rosell. 2016 - 2018.
- Development of methods for measuring the effort/recovery level in physical exercise based on cardiovascular activity, temperature and respiration. MINECO. 2015. DEP2015-68538-C2-2-R. PI: Xavier Rosell. 2016 - 2018.
- Activities of Daily Augmented Living Toolkit (ADALT). EURECAT. PI: Xavier Rosell. 2016 - 2017
- RWECS. IAAF. PI: Xavier Rosell. 2016 - 2018.

### Selected publications

- Lecina M, Sánchez Fernández B, Solà C, Prat J, Roldán M, Hernández M, et al. Structural changes of *Arthrospira* sp. after low energy sonication treatment for microalgae harvesting: Elucidating key parameters to detect the rupture of gas vesicles. Bioresour Technol 2017. 223: 98-104. FI: 5,651(Q1).
- Mahdavi H, Rosell-Ferrer J. In-bed vital signs monitoring system based on unobtrusive magnetic induction method with a concentric planar gradiometer. Physiol Meas. 2017 Jun;38(6):1226-1241. FI: 2,058 (Q3).



PERSONAL  
RESEARCHER



PROJECTS



PUBLICATIONS  
WITH IF



FIRST QUARTILE  
PUBLICATIONS

## ROBOTICS AND VISION

Alicia Casals Gelpí, PhD

*Guidance on process automation, adaptation and robotisation of facilities, and on different types of facilities, development of vision systems for inspection and guiding of robots, development of image processing systems, design of mobile robots (intelligent walkers, prams, etc.), and study and design of technical aids for persons with physical and sensory disabilities, as well as guidance on and development of advanced control systems.*

### Members

#### Staff researcher

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

#### Postdoc – Associate researcher

Albert Hernansanz Prats

#### PhD Student

Narcís Sayols Baixeras, Manuel Vinagre Ruiz

#### Collaborator researcher

Antonio Benedico Blanes, Carlos Morat, Laureano Tinoco, Alejandro Veiga López

#### Technician

Lluís Oriol Escoté

### Selected projects

- Distributed strategies of person-robot control and cooperation in healthcare environments. Ministerio de Economía, Industria y Competitividad. MINECO. DPI2015-70415-C2-1-R. PI: Alicia Casals. 2016 - 2018.
- New high-precision technologies in foetal medicine and surgery: Robotics. Fundació Cellex i Fundació La Caixa. PI: Alicia Casals. 2015 - 2018.
- Diabetes as an accelerator of cognitive impairment and Alzheimer's disease: approach and adherence EU-RIS3CAT. Generalitat de Catalunya. EU-RIS3CAT. PI: Antonio B Martínez. 2016 - 2019.

### Selected publications

- Aviles Rivero AI, Alsaleh SM, Hahn JK and Casals A. Towards Retrieving Force Feedback in Robotic-Assisted Surgery: A Supervised Neuro-Recurrent-Vision Approach.. IEEE Trans Haptics 2017. 10: 431-443. FI: 2,000(Q2).
- Aviles, A., Widlak, T., Casals A, Nillesen, M. and Ammari, H.. Robust cardiac motion estimation using ultrafast ultrasound data: a low-rank topology-preserving approach. Phys Med Biol 2017. 62: 4831-4851. FI: 2,742(Q2).
- Ballesteros, J., Urdiales, C., Martínez Velasco A and Ramos-Jimenez, G. A Biomimetical Dynamic Window Approach to Navigation for Collaborative Control. IEEE T HUM-MACH SYST 2017. 47: 1123-1133. FI: 2,493(Q2).



PERSONAL  
RESEARCHER



PROJECTS



PUBLICATIONS  
WITH IF

### Networks

- 2016 - 2017 - Hypernet: Network of excellence on neuroprosthetics and neurorobbing for functional compensation and rehabilitation of movement disorders
- 2015 - Clúster de Salut Mental de Catalunya

## MEDICAL RADIOPHYSICS

Mercè Ginjaume Egido, PhD

1. Development of radiotherapy treatment (RT) planning systems based on the use of Monte Carlo (MC) simulation, in particular the Penelope programme. The principal novelties notably include the introduction of proton applications, the study of small fields and the study of the skin dose in IMRT and SBRT.
2. Within the framework of the H2020 MEDIRAD project, a Monte Carlo programme based on the use of GPU (MC-GPU) cards is being optimised to obtain a real-time dose control system capable of minimising the probability of radiogenic skin lesions resulting from fluoroscopy-guided procedures.
3. Based on the foregoing programme, a new project has been started that allows estimation of the effective dose in organs of interest, particularly the lens of the eye and the brain, of healthcare staff participating in radiology and interventional cardiology.

### Members

#### Staff researcher

Maria Amor Duch Guillen, Youri Alexandrovich Koubychne Merkulov, Xavier Ortega Aramburu, Josep Sempau Roma

#### Postdoc – associate researcher

Sara Principi

### Selected projects

- Implications of Medical Low Dose Radiation Exposure. (MEDIRAD). H2020 - 2017. H2020-755523. PI: Mercè Ginjaume. 2017 - 2021.
- Online personal dosimetry using computational methods. MINECO - 2017. PCIN-2017-044. PI: M.Amor Duch. 2017 - 2019.

### Selected publications

- Puxeu Vaqué J, Duch Guillen MA, Nailon WH, Lizuain Arroyo MC and Ginjaume Egido M. Field correction factors for a PTW-31016 Pinpoint ionization chamber for both flattened and unflattened beams. Study of the main sources of uncertainties. Med Phys 2017. 44: 1930-1938. FI: 2,617(Q2).
- Bueno M, Duch M, Jurado-Bruggeman D, Agramunt S, Muñoz-Montplet C. Experimental verification of acuros XB in the presence of lung-equivalent heterogeneities. Radiation Measurem 2017.106: 357-360. FI: 1,442 (Q1).
- Cantone, MC, Ginjaume Egido M, Miljanic, S, Martin, CJ, Akahane, K, Mpete, L, Michelin, SC, Flannery, CM, Dauer, LT and Balter, S. Report of task group on the impact of the eye lens dose limits. J Radiol Prot 2017. 37: 527-550. FI: 1,657(Q1).

### Networks

- 2000 - EURADOS, European Radiation Dosimetry Group, [www.eurados.org](http://www.eurados.org) - Voting Member i Investigadors WG12 medical imaging
- SGR 2014, SGR 2017 Dosimetria i Radiofísica mèdica



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PROJECTS



PUBLICATIONS  
WITH IF



FIRST QUARTILE  
PUBLICATIONS

## BIOMECHANICAL ENGINEERING

Josep Maria Font Llagunes, PhD

*Kinematic and dynamic measurements of human movement in the biomechanics laboratory, dynamic analysis of human movement, mechanical design of care and rehabilitation devices.*

### Members

#### Staff researcher

Ana Barjau Condomines, Daniel Clos Costa, Rosa Pàmies Vilà, Míriam Febrer Nafría, Gil Serrancolí Masferrer, Alfons Carnicer Carmona

#### Selected projects

- Biomechanical modelling and simulation of the symptomatology of Duchenne Muscular Dystrophy (DMD)\*. Marsini Bionics - 2017. PI: Josep Maria Font. 2017.
- Low-cost motor-FES hybrid orthosis for the gait of spinal cord injured subjects and simulation methods to support design and adaptation. MINECO - 2016. (DPI2015-65959-C3-2-R). PI: Josep Maria Font. 2016 - 2018.
- Activities of Daily Augmented Living Toolkit (ADALT). EURECAT. PI: Josep Maria Font. 2016- 2017.
- ABLE: Assistive Biorobotic Low-cost Exoskeleton. Fundació La Caixa, CaixaImpulse - 2017. CI17-00009. PI: Josep Maria Font. 2017 - 2018.

#### Selected publications

- Firoozabadi AE, Ebrahimi S and Font Llagunes JM. A comparative study of elastic motions in trajectory tracking of flexible RPR planar manipulators moving with high speed. Robotica 2017. 35: 1523-1540. FI: 1,554(Q3).
- Font Llagunes JM, Muñoz JJ and Ambrósio JAC. Preface: Thematic Issue on Multibody Dynamics 2015. Multibody Syst Dyn 2017. 39: 1-2. FI: 2,286(Q1).
- Pàmies R, Pätkau O, Dòria-Cerezo A and Font Llagunes JM. Influence of the controller design on the accuracy of a forward dynamic simulation of human gait. Mech Mach Theory 2017. 107: 123-138. FI: 2,577(Q1).

#### Networks

- JM Font- Member of the Technical Committee for Multibody Dynamics, International Federation for the Promotion of Mechanism and Machine Science, IFToMM.
- JM Font- Vice-President of the Spanish Chapter of the European Society of Biomechanics, ESB.
- JM Font- PI of the Spanish Network of Biomechanics, Ministry of Economy and Competitiveness, Spain.
- JM Font- Member of the European Society of Biomechanics, ESB.



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RESEARCHER



PROJECTS



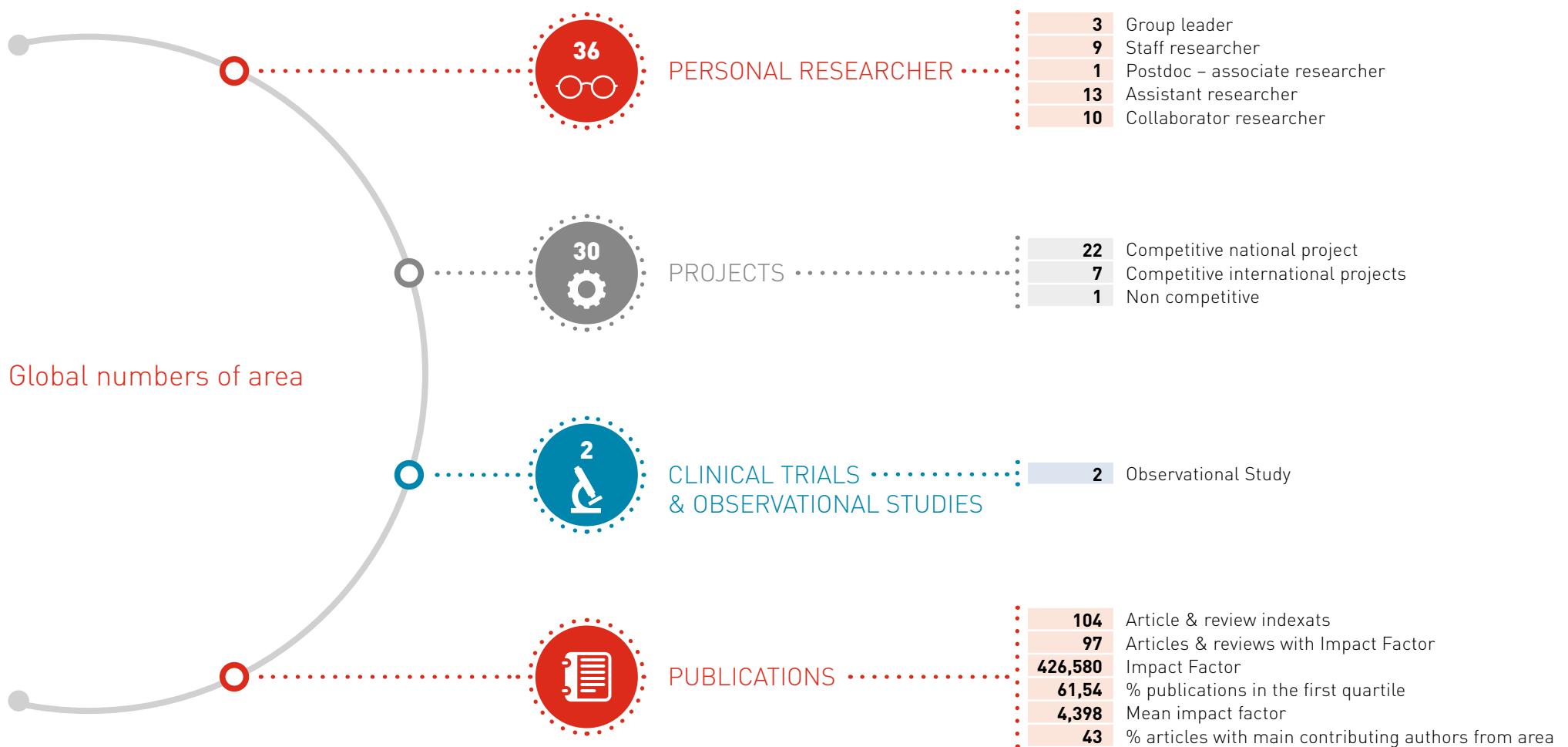
PUBLICATIONS  
WITH IF



FIRST QUARTILE  
PUBLICATIONS

## 4.4. Epidemiology

**Coordinator: Josep Maria Haro**



## EPIDEMIOLOGY OF AGING: FACTORS ASSOCIATED WITH HEALTHY AGING

Josep Maria Haro Abad, MD, PhD

*The interest of this research line lies in determining the impact (prevalence and disability) of mental disorders in the general population and the health care received by the persons who suffer such disorders. Epidemiologic studies are necessary for the planning and management of resources and to evaluate the existence and impact of factors associated with the possibility of suffering a mental disorder. The various projects of this line are focused on the adult population, the child and adolescent population, and elderly people.*

### Members

#### Staff researcher

Albert Sánchez Niubó, Stefanos Tyrovolas

#### Assistant researcher

Joan Domènech Abella, Laia Egea Cortes, Blanca Mellor Marsa, Ziggi Santini, Sara Siddi, Elisabeth Catharina Vorstenbosch

### Selected projects

- MULTI-MODE. European Commission. EIT-HEALTH/DIGITAL combined proposal. 17524. PI: Josep Maria Haro Abad. 2017 - 2017.
- Mental health paths, physical health and functioning: assessment of a Spanish adult population-based cohort. Instituto de Salud Carlos III (ISCIII). Proyectos de investigación en salud. PI16/01073. PI: Josep Maria Haro Abad. 2017 - 2019
- Estimation Global frailty Burden. Instituto de Salud Carlos III (ISCIII). M-AES. MV16/00035. PI: Stefanos Tyrovolas. 2017 - 2018.

### Selected publications

- GBD 2015 Healthcare Access and Quality Collaborators. Electronic address: [cjlm@u](mailto:cjlm@u) and GBD 2015 Healthcare Access and Quality Collaborators. Healthcare Access and Quality Index based on mortality from causes amenable to personal health care in 195 countries and territories, 1990-2015: a novel analysis from the Global Burden of Disease Study 2015. Lancet 2017. 390: 231-266. FI: 47,831(Q1).

- Degenhardt L, Glantz M, Evans-Lacko S, Sadikova E, Sampson N, Thornicroft G, et al. Estimating treatment coverage for people with substance use disorders: an analysis of data from the World Mental Health Surveys. World Psychiatry 2017. 16: 299-307. FI: 26,561(Q1).
- Bromet EJ, Nock MK, Saha S, Lim CCW, Aguilar-Gaxiola S, Al-Hamzawi A, et al. Association Between Psychotic Experiences and Subsequent Suicidal Thoughts and Behaviors: A Cross-National Analysis From the World Health Organization World Mental Health Surveys. JAMA Psychiatry 2017. 74: 1136-1144. FI: 15,307(Q1).
- Liu H, Petukhova MV, Sampson NA, Aguilar-Gaxiola S, Alonso J, Andrade LH, et al. Association of DSM-IV Posttraumatic Stress Disorder With Traumatic Experience Type and History in the World Health Organization World Mental Health Surveys. JAMA Psychiatry 2017. 74: 270-281. FI: 15,307(Q1)

### Networks

- CIBERSAM
- European Innovation Partnership on Active and Healthy Ageing (the Partnership)



10  
PERSONAL  
RESEARCHER



13  
PROJECTS



56  
PUBLICATIONS  
WITH IF



61.7%  
FIRST QUARTILE  
PUBLICATIONS

## IMPACT, RISK AND PREVENTION OF MENTAL DISORDERS

Ai Koyanagi, MD, PhD

*Mental disorders are the group of illnesses with the greatest impact on the population's health in the most economically-developed countries, as has been stated by the World Health Organisation and in the Global Burden of Disease study. Between 20% and 30% of all people suffer a mental disorder at some point in their life. In many cases, this disorder is highly incapacitating. Our interdisciplinary group, which includes psychiatrists, psychologists, statisticians, sociologists and basic researchers, studies the factors that provide protection against or increase the risk of mental disorders, and treatments for the people who suffer them, for the purpose of improving preventive and therapeutic strategies. In recent years, aware of the importance of mental health in the achievement of a physically, mentally and socially healthy ageing process, we are devoting special attention to the study of health histories in order to determine the factors which favour active healthy ageing and at exactly which moment in time it is necessary to intervene in order to improve the course of each individual's health. We seek to translate all this knowledge into healthcare innovations which can be easily incorporated into our healthcare and social support systems.*

### Members

#### Assistant researcher

Ivet Bayes Marin, Daniel Fernández Martínez,  
Maria Victoria Moneta

#### Collaborator researcher

Beatriz Olaya Guzmán

### Selected projects

- Mortality associated with mental disturbances and impact of physical comorbidity: Analysis based on big data and a representative longitudinal sample of the aged. Instituto de Salud Carlos III (ISCIII). Proyectos de investigación en salud. PI16/00321. PI: Beatriz Olaya Guzmán. 2017 - 2019.
- Severe mental disturbances and physical illness: analysis of health-related life paths and impact on functioning, quality of life, disability, mortality and the use of services, based on big longitudinal data bases. Beneficiary: Roberto Nuevo. Departament de Salut - Generalitat de Catalunya. Incorporació de científics i tecnòlegs. SLT002/16/00469. PI: Ai Koyanagi. 2017 - 2019.
- Pseudo-psychotic experiences and health: Results of epidemiological studies. Instituto de Salud Carlos III (ISCIII). FIS. PI15/00862. PI: Ai Koyanagi. 2016 - 2018.

### Selected publications

- Global Burden of Disease Child and Adolescent Health Collaboration, Kassebaum N, Kyu HH, Zoekler L, Olsen HE, Thomas K, et al. Child and Adolescent Health From 1990 to 2015: Findings From the Global Burden of Diseases, Injuries, and Risk Factors 2015 Study. JAMA Pediatr 2017. 171: 573-592. FI: 10,251(Q1).

- Koyanagi A, Vancampfort D, Carvalho AF, DeVylder JE, Haro JM, Pizzol D, et al. Depression comorbid with tuberculosis and its impact on health status: cross-sectional analysis of community-based data from 48 low- and middle-income countries. BMC Med 2017. 15: 209-0. FI: 7,901(Q1).
- Stubbs B, Vancampfort D, Veronese N, Kahl KG, Mitchell AJ, Lin PY, et al. Depression and physical health multimorbidity: primary data and country-wide meta-analysis of population data from 190 593 people across 43 low- and middle-income countries. Psychol Med 2017. 47: 2107-2117. FI: 5,230(Q1).
- Koyanagi A, Oh H, Stubbs B, Haro JM and DeVylder JE. Epidemiology of depression with psychotic experiences and its association with chronic physical conditions in 47 low- and middle-income countries. Psychol Med 2017. 47: 531-542. FI: 5,230(Q1).

### Networks

- CIBERSAM



## HEALTHCARE TECHNOLOGIES AND RESULTS IN PRIMARY CARE AND MENTAL HEALTH (PRISMA)

Antonio Serrano Blanco, MD, PhD

*Researchers of the PRISMA group estimated the costs of failure to begin medication and the impact of co-payment on such behaviour. A review was made of the evidence on the effectiveness of antidepressants as compared to active monitoring in primary care. Likewise, they typified agitation episodes, their interventions and their costs in a mental health hospital. The group also organised the 11th Course on Socio-Health Policy and Financial Assessment at the Loyola University Andalucía in collaboration with the King's College of London and the London School of Economics and Political Science.*

### Members

#### Staff researcher

Maria Rubio Valera, Luisa Baladon Higuera, Vanessa Barneda, Laura González Suñer, María Iglesias González, Cristina López Ortiz, Enric Vicens Pons

#### Postdoc – associate researcher

Ignacio Aznar Lou

#### Assistant researcher

Maria Montserrat Gil Girbau, Ana Gabriela Murrugarra Centurion, Carles Vilaplana Carnerero

#### Collaborator researcher

Ana Fernández Sánchez, Ana María Jove Massó, Martín Knapp, María Antonia March Pujol, Paul McCrone, Juan Manuel Mendive Arbeloa, María Teresa Peñarrubia, Ramón Sabes Figuera, Lluís Salvador Carulla

### Selected projects

- The problem of not starting pharmacological treatment: assessment by mixed methods. Instituto de Salud Carlos III (ISCIII). FIS. PI14/00052. PI: María Rubio Valera. 01/01/2015 - 31/12/2018.
- Financial assessment of a multi-risk complex intervention to promote healthy behaviours in personas aged 45-75 years treated in primary care: Econ-EIRA3 study. Instituto de Salud Carlos III (ISCIII). FIS. PI15/00519. PI: Antonio Serrano Blasco. 01/01/2016 - 31/12/2018.

- PRE-test-STROKE\_Personalizing REperfusion Therapeutic Strategies in Stroke. Instituto de Salud Carlos III (ISCIII). Medicina Personalizada. PMP15/00022. PI: Antoni Serrano Blasco. 01/01/2016 - 31/12/2019.

### Selected observational studies

- AMDC 204-401 (ALE-LOX-2014-01). Post-authorisation observational study to evaluate the safety of ADASUVE® (Loxapine for Staccato inhaler) in people agitated in routine clinical practice. PI: Antoni Serrano. 2015 - 2017.
- ARI-MGD-2016-01. Aripiprazole once-monthly versus daily oral atypical antipsychotic treatment in patients with recent-onset schizophrenia: an observational, non-interventional study. Promoter: Otsuka Pharmaceutical, S.A.. PI: Antoni Serrano. 2017.

### Selected publications

- Aznar-Lou I, Fernández A, Gil Gribau MM, Fajó-Pascual M, Moreno-Peral P, Peñarrubia-Teresa MT, et al. Initial medication non-adherence: prevalence and predictive factors in a cohort of 1.6 million primary care patients. Br J Clin Pharmacol 2017; 83: 1328-1340. FI: 3,493(Q1).

- Aznar-Lou I, Fernández A, Gil Gribau MM, Sabés-Figuera R, Fajó-Pascual M, Peñarrubia-María MT, et al. Impact of initial medication non-adherence on use of healthcare services and sick leave. Br J Gen Pract 2017; 67: 614-622. FI: 2,760(Q1).
- Serrano-Blanco A, Rubio-Valera M, Aznar-Lou I, Baladon L, Gibert K, Gracia Canales A, et al. In-patient costs of agitation and containment in a mental health catchment area. BMC Psychiatry 2017; 17: 212-0. FI: 2,613(Q2).
- Iglesias Gonzalez M, Aznar-Lou I, Gil Gribau MM, Moreno-Peral P, Peñarrubia-María MT, Rubio-Valera M and Serrano-Blanco A. Comparing watchful waiting with antidepressants for the management of subclinical depression symptoms to mild-moderate depression in primary care: a systematic review. Fam Pract 2017; 34: 639-648. FI: 1,804(Q2).

### Networks

- 2017 - CIBERESP
- Red IAPP



FIRST QUARTILE  
PUBLICATIONS



The **team members** of areas and groups presents all information of the group including external collaborators. The global numbers of the institution does not include external collaborators.

The number of **projects** shows the total number of projects managed and active during 2017.

**Clinical trials** and **observational studies** correspond to all those actives during 2017.

**Publications** include original articles and reviews published during 2017 (with assigned pagination) and authored by members of research groups during their membership. The impact factor and quartile assigned corresponds to the Journal Citation Reports of the Web of Science of 2016.

The **Budget** is the total resources managed.