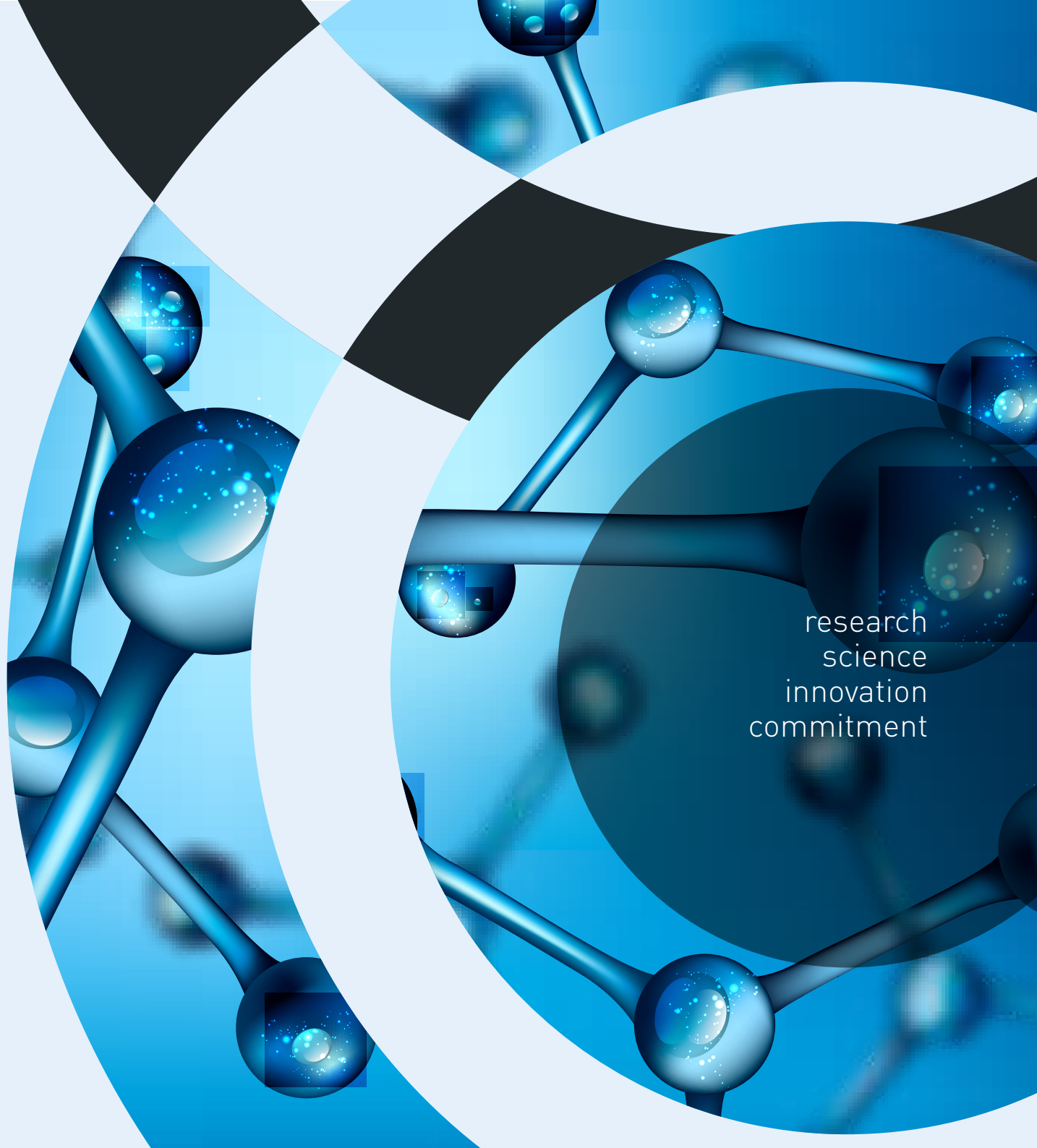
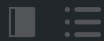


ANNUAL SCIENTIFIC
REPORT
2018

 **SJD**
Sant Joan de Déu
Institut de Recerca

research
science
innovation
commitment





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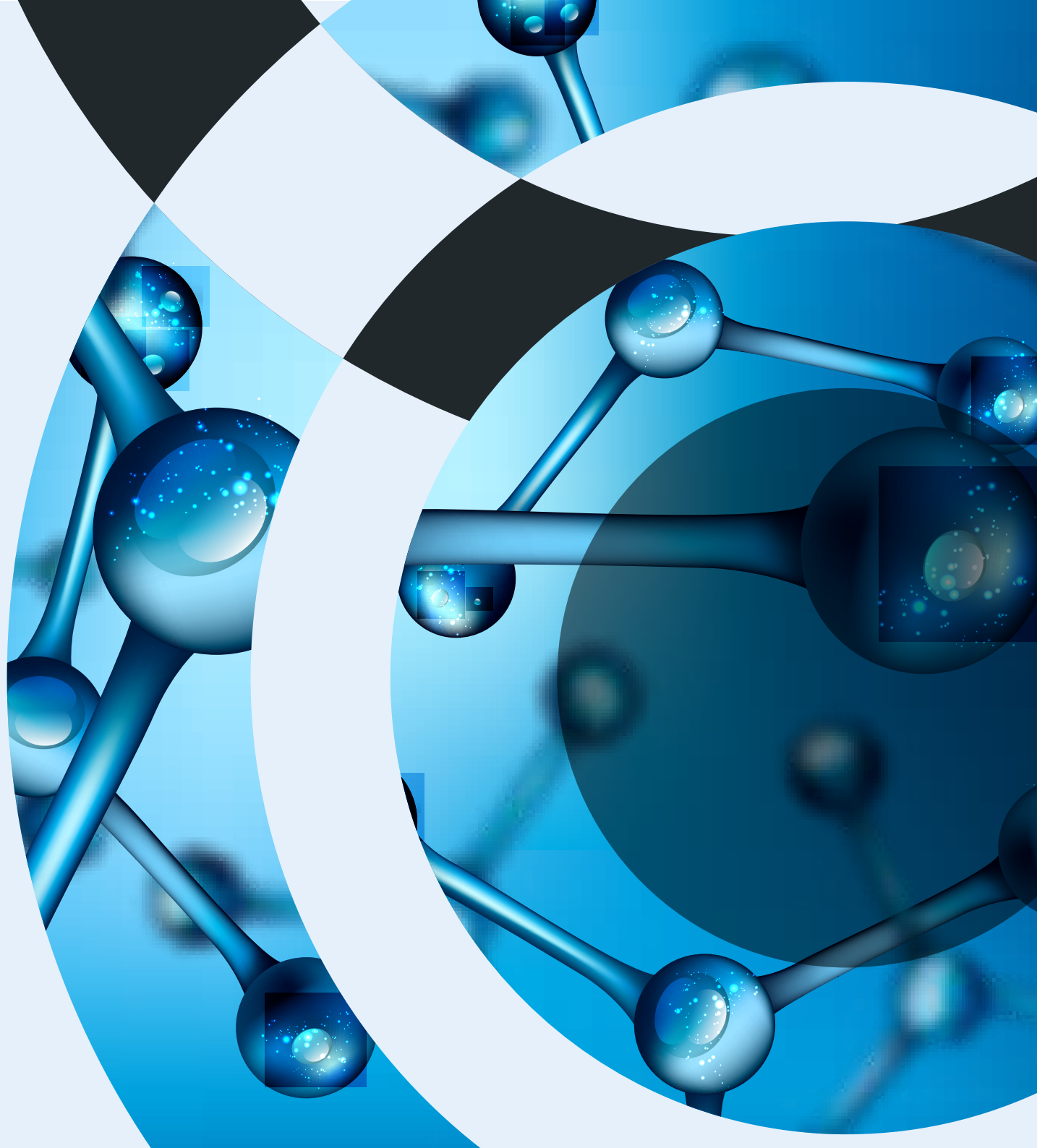
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1 FOREWORD

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- 1.3. Services and infrastructures



1.1. Introduction

It is a pleasure to show you the annual scientific report of the Institut de Recerca Sant Joan de Déu (IRSJD), which shows once again the great work done by all IRSJD's people. From here, our acknowledgment and gratitude to everyone who, in one way or another, makes possible the continuous improvement of our institution, the scientific results and the transfer of knowledge for the improvement of patients.

During this year, from the Direction of the Institute has been worked on the Strategic Plan for the next four years. Scientific excellence, the consolidation of the human team and the infrastructures, and the promotion and support for the establishment of scientific collaborations and international alliances are the main objectives of this new Plan. In parallel, there has been a long process of reflection, together with the people leading the research groups of the Institute, on the interconnections between the groups of the different centers that make up the IRSJD. This has allowed redefining the cooperative scientific project model of the Institute.

The scientific framework we have had since 2015 has been very useful for showing the nuclear research lines of the Hospital and the transversally of the research groups of the other IRSJD's Centers. The new scientific structure aims to reflect and enhance the common research objectives and lines, and facilitate collaboration between the medical and hospital medical activity and the basic and technological research groups. It is also necessary to make reference to the different agents that are fundamental for quality research, with translation and responsible capacity, such as healthcare professionals, experts in innovation and patient representatives. As of 2019, this new structure will be fully developed.

During 2018, the IRSJD has increased the research projects, clinical trials and scientific publications; in such a way that the indicators obtained allow us to continue with sustained growth. The upstream line in the collection of financing funds and scientific production since the creation of the IRSJD places us in a good position among the research centers in our environment. Just to make some mention in relation to scientific production, we would like to highlight, among others, some publications led by researchers from the different research programs, such as Paediatric Cancer in Clinical Cancer Research (C. Lavarino) and Journal of Controlled Release (A. Montero-Carcaboso); in the field of Neuroscience with publications in Developmental Medicine & Child Neurology by A. García-Cazorla and published by the group of M. Pastor-Anglada at Comprehensive Physiology; and the publications conducted by L. Alsina in Pediatric Allergy and Immunology and L. Ibáñez in Trends in Endocrinology and Metabolism in the Adaptive Response area; Importantly, there have been international collaborations that have been reflected in works published in JAMA Psychiatry, with the participation of A. Koyanagi of the area of Mental Health in the Neurosciences program.

Finally, from the Directorate we would like to encourage you to continue working and collaborating with all of us, and with other national and international centers. We know that we have a great human team to achieve our priority objective: a job of excellence to achieve the best solutions for our patients.

1.2. Governing and advisory bodies

Board of trustees

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Deputy Director, Institut de Recerca Sant Joan de Déu
Director, Research Foundation Sant Joan de Déu
Director for Innovation, Hospital Sant Joan de Déu
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Universitat Politècnica de Catalunya*
Director, Institut de Biomedicina - Universitat de Barcelona (IBUB)
Institut de Neurociències - Universitat de Barcelona (UBNeuro)
Director for Research, Innovation and Teaching, Parc Sanitari Sant Joan de Déu
Scientific manager, Institut de Recerca Sant Joan de Déu

Scientific Advisory Council

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Montserrat Vendrell i Rius

Scientific director. European infrastructure for translational medicine (EATRIS)
Director. Centro Nacional de Investigaciones Oncológicas (CNIO)
Director. Parc de Recerca Biomèdica de Barcelona & Fundació Pasqual Maragall
London's Global University. Division of Psychiatry
Scientific director. Duchenne Parent Project, Spain
Executive vice-president. St. Jude Children's Research Hospital
Paediatric endocrinologist. Institut des Sciences du Cerveau, de la Cognition et du Comportement de Toulouse (ISC3T)
Partner. Alta Life Sciences

Research commission

Rafael Artuch Iriberry, *president*
Asteria Albert Cazalla
Pere Caminal Magrans
Carmen de Torres Gómez-Pallete
Carles Escera i Micó
Claudia Fortuny Guasch
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Àngel Montero Carcaboso
Carmen Muñoz Almagro
Susana Ochoa Güerre
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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

- Support of the 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

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
- Management of knowledge and communication



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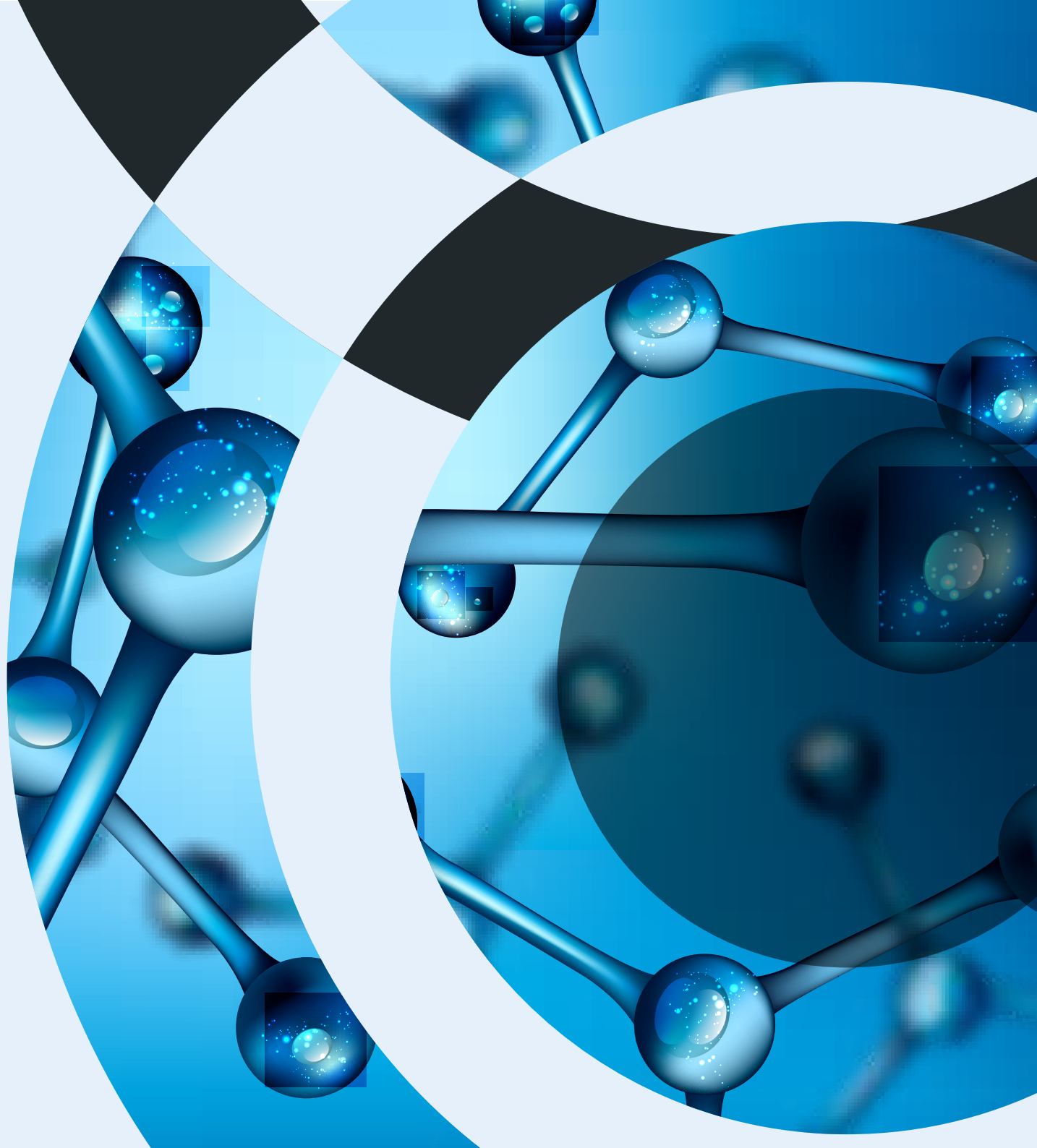
GLOBAL NUMBERS

- 2.1. Global numbers 2018
- 2.2. Global numbers 2015-2018
- 2.3. Research and innovation outputs
 - Scientific publications
 - Patents and utility models
 - Spin-off
- 2.4. Training
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 - Scientific seminars

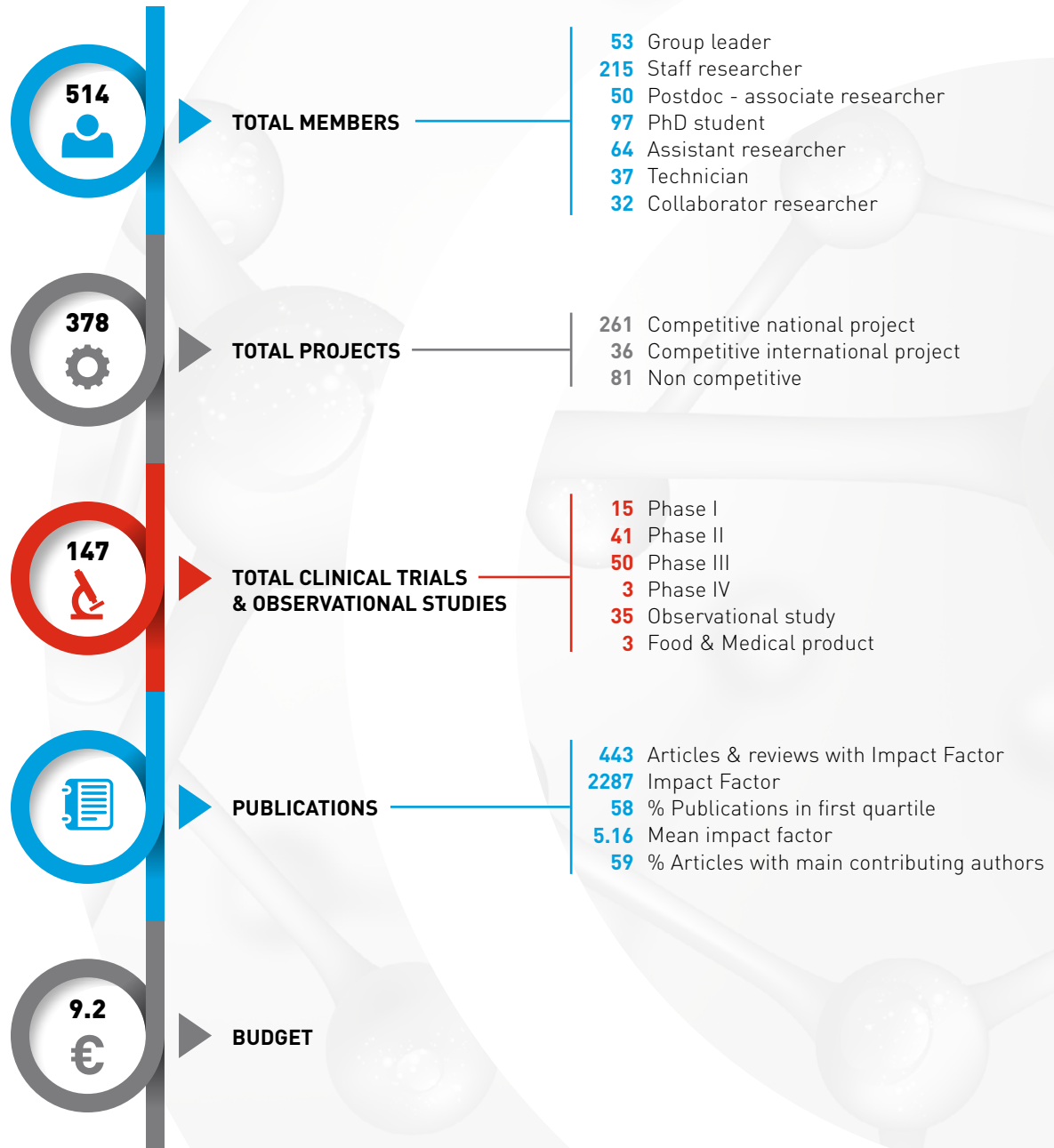


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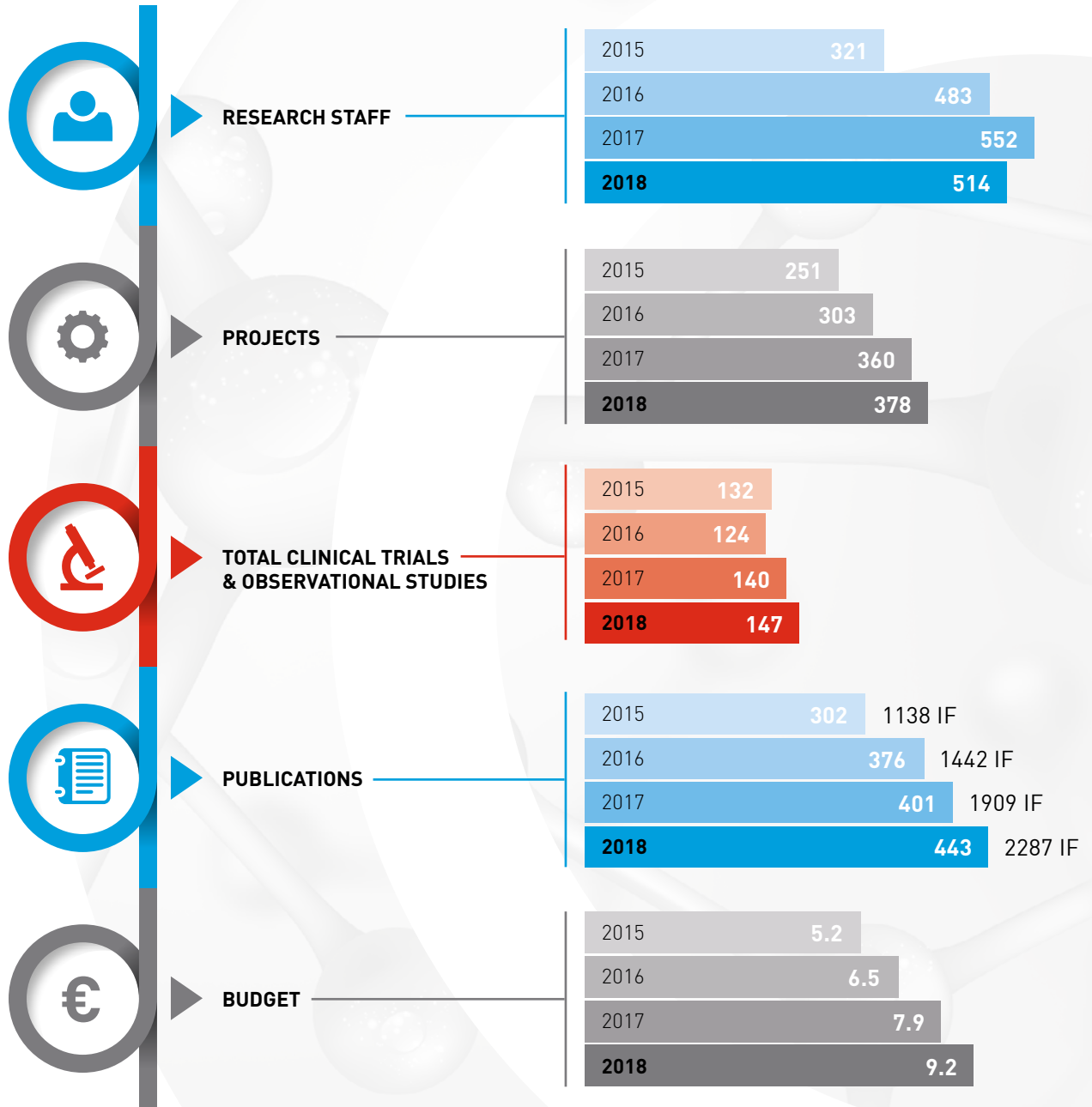
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2.1. Global numbers 2018

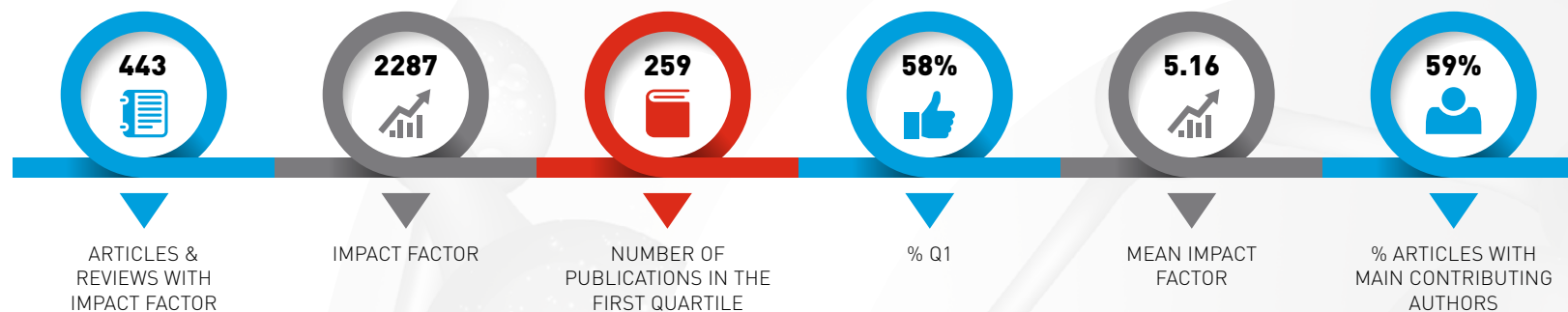


2.2. Global numbers 2015-2018



2.3. Research and Innovation Outputs

SCIENTIFIC PUBLICATIONS



%Q1: % of publications published in journals ranked in the first quartile (top 25%)
Mean IF: Mean impact factor of all the scientific publications

Highest impact factor journals

The list reflects the 99 original articles and reviews published in the most relevant scientific journals (1st decil)

Journal	IF	Total articles
THE NEW ENGLAND JOURNAL OF MEDICINE	79,258	2
JOURNAL OF THE AMERICAN MEDICAL ASSOCIATION	47,661	1
NATURE	41,577	1
SCIENCE	41,058	3
NATURE MEDICINE	32,621	1
LANCET NEUROLOGY	27,138	1
NATURE GENETICS	27,125	1
CANCER CELL	22,844	1
NATURE REVIEWS NEUROLOGY	19,819	1
JOURNAL OF THE AMERICAN COLLEGE OF CARDIOLOGY	16,834	2
JAMA PSYCHIATRY	16,642	1

Journal	IF	Total articles
ACTA NEUROPATHOLOGICA	15,872	5
INTENSIVE CARE MEDICINE	15,008	1
JOURNAL OF THE AMERICAN CHEMICAL SOCIETY	14,357	1
JOURNAL OF ALLERGY AND CLINICAL IMMUNOLOGY	13,258	1
ANNALS OF THE RHEUMATIC DISEASES	12,350	5
TRENDS IN PHARMACOLOGICAL SCIENCES	12,108	1
BIOLOGICAL PSYCHIATRY	11,982	1
PLOS MEDICINE	11,675	2
MOLECULAR PSYCHIATRY	11,640	1
JOURNAL OF EXPERIMENTAL MEDICINE	10,790	1
TRENDS IN ENDOCRINOLOGY & METABOLISM	10,769	2

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FOREWORD2.3.
Research and
Innovation Outputs

Journal	IF	Total articles
EMBO MOLECULAR MEDICINE	10,293	1
CLINICAL CANCER RESEARCH	10,199	2
LEUKEMIA	10,023	1
MICROBIOME	9,133	1
BMC MEDICINE	9,088	2
GENOME MEDICINE	8,898	1
BIOMATERIALS	8,806	1
AUTOIMMUNITY REVIEWS	8,745	1
JOURNAL OF THE AMERICAN SOCIETY OF NEPHROLOGY	8,655	1
MOVEMENT DISORDERS	8,324	1
BIOSENSORS AND BIOELECTRONICS	8,173	1
JAMA DERMATOLOGY	8,107	1
JOURNAL OF CONTROLLED RELEASE	7,877	2
ARTHRITIS & RHEUMATOLOGY	7,871	2
EUROPEAN JOURNAL OF NUCLEAR MEDICINE AND MOLECULAR IMAGING	7,704	1
AGING CELL	7,627	1
ENVIRONMENT INTERNATIONAL	7,297	1
BMJ QUALITY & SAFETY	7,226	1
JOURNAL OF INTERNAL MEDICINE	6,754	1
JOURNAL OF NANOMEDICINE AND NANOTECHNOLOGY	6,500	1
CRITICAL REVIEWS IN CLINICAL LABORATORY SCIENCES	6,481	1
JOURNAL OF INVESTIGATIVE DERMATOLOGY	6,448	1
ACTA BIOMATERIALIA	6,383	3
JOURNAL OF BONE AND MINERAL RESEARCH	6,314	1
MOLECULAR METABOLISM	6,291	1
BRITISH JOURNAL OF DERMATOLOGY	6,129	1
ADDICTION	5,953	1
COMPREHENSIVE PHYSIOLOGY	5,797	1

Journal	IF	Total articles
EPIDEMIOLOGY AND PSYCHIATRIC SCIENCES	5,684	1
ULTRASOUND IN OBSTETRICS & GYNECOLOGY	5,654	3
FORENSIC SCIENCE INTERNATIONAL: GENETICS	5,637	1
ADVANCED HEALTHCARE MATERIALS	5,609	2
INTERNATIONAL JOURNAL OF BEHAVIORAL NUTRITION AND PHYSICAL ACTIVITY	5,548	1
CLINICAL NUTRITION	5,496	2
BIOINFORMATICS	5,481	1
PSYCHOLOGICAL MEDICINE	5,475	5
NEUROIMAGE	5,426	1
JOURNAL OF THE AMERICAN MEDICAL DIRECTORS ASSOCIATION	5,325	2
DEPRESSION AND ANXIETY	5,043	1
THE JOURNALS OF GERONTOLOGY	4,902	2
BRAIN STRUCTURE AND FUNCTION	4,231	1
JOURNAL OF THE AMERICAN GERIATRICS SOCIETY	4,155	1
PEDIATRIC ALLERGY AND IMMUNOLOGY	4,137	1
EUROPEAN CHILD & ADOLESCENT PSYCHIATRY	3,553	1
DEVELOPMENTAL MEDICINE & CHILD NEUROLOGY	3,289	2
JOURNAL OF ADVANCED NURSING	2,267	1

PATENTS AND UTILITY MODELS

Nonwoven membrane as a drug delivery System

Date of grant	25/07/2018
Country of protection	AL, AT, BE, BG, CH, CY, CZ, DE, DK, EE, ES, FI, FR, GB, GR, HR, HU, IE, IS, IT, LI, LT, LU, LV, MC, MK, MT, NL, NO, PL, PT, RO, RS, SE, SI, SK, SM, TR
Grant number	EP3108881A1
Applicants	Universitat Politècnica de Catalunya i Hospital Sant Joan de Déu
Inventors	Jose Antonio Tornero Garcia, Angel Montero Carcaboso, Joan Bertran Llavina

Immunotherapy for Casr-Expressing Cancer (E.G. Neuroblastoma)

Date of grant	18/01/2018
Country of protection	Estats Units
Grant number	US20180207111A1
Applicants	Hospital Sant Joan de Déu i Fundació Cellex
Inventors	Carmen de torres Gómez-Pallete

Método de clasificación en subgrupos moleculares de pacientes con meduloblastoma

Date of grant	22/11/2018
Country of protection	AE, AG, AL, AM, AO, AT, AU, AZ, BA, BB, BG, BH, BN, BR, BW, BY, BZ, CA, CH, CL, CN, CO, CR, CU, CZ, DE, DJ, DK, DM, DO, DZ, EC, EE, EG, ES, FI, GB, GD, GE, GH, GM, GT, HN, HR, HU, ID, IL, IN, IR, IS, JO, JP, KE, KG, KH, KN, KP, KR, KW, KZ, LA, LC, LK, LR, LS, LU, LY, MA, MD, ME, MG, MK, MN, MW, MX, MY, MZ, NA, NG, NI, NO, NZ, OM, PA, PE, PG, PH, PL, PT, QA, RO, RS, RU, RW, SA, SC, SD, SE, SG, SK, SL, SM, ST, SV, SY, TH, TJ, TM, TN, TR, TT, TZ, UA, UG, US, UZ, VC, VN, ZA, ZM, ZW
Grant number	PCT/ES2018/070347
Applicants	Hospital Sant Joan de Déu
Inventors	Cinzia Emilia Lavarino, Soledad Gomez Gonzalez

Betaine for the prevention of obesity

Date of grant	15/11/2018
Country of protection	AE, AG, AL, AM, AO, AT, AU, AZ, BA, BB, BG, BH, BN, BR, BW, BY, BZ, CA, CH, CL, CN, CO, CR, CU, CZ, DE, DJ, DK, DM, DO, DZ, EC, EE, EG, ES, FI, GB, GD, GE, GH, GM, GT, HN, HR, HU, ID, IL, IN, IR, IS, JO, JP, KE, KG, KH, KN, KP, KR, KW, KZ, LA, LC, LK, LR, LS, LU, LY, MA, MD, ME, MG, MK, MN, MW, MX, MY, MZ, NA, NG, NI, NO, NZ, OM, PA, PE, PG, PH, PL, PT, QA, RO, RS, RU, RW, SA, SC, SD, SE, SG, SK, SL, SM, ST, SV, SY, TH, TJ, TM, TN, TR, TT, TZ, UA, UG, US, UZ, VC, VN, ZA, ZM, ZW
Grant number	WO2018/206763A1
Applicants	Hospital Sant Joan de Déu i Ajinomoto
Inventors	Carlos Lerín Martínez, Jose Carlos Jiménez Chillaron, Marta Ramón Krauel

SPIN-OFF

DBGEN Ocular Genomics (co-funders Roser Gonzàlez-Duarte and Gemma Marfany)

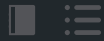
2.4. Training

DOCTORAL THESES

A list provided below the 33 doctoral theses presented during 2018 in original language.

Thesis Author	Title	Thesis directors	Date
Javier Rodríguez Fanjul	Neuroprotección mediante terapia combinada (hipotermia+alopurinol) en un modelo animal de daño cerebral hipóxico-isquémico	Alfredo Garcia-Alix	January
Eva Maria Guix Comellas	Influència del seguiment infermer en l'adherència al tractament amb fàrmacs antituberculosos en la població pediàtrica	Antoni Noguera Julian Enriqueta Force Sanmartín	January
Sally Sabra	Maternal heavy metals and toxins exposure and their influences on fetal growth restriction clinical subtypes	Eduard Gratacos Maria Dolores Gómez-Roig	January
María José Fábrega Fernández	Modulation of intestinal immune response by outer membrane vesicles from probiotic and commensal Escherichia coli strains	Laura Baldomà Llavínés Rosa Giménez Claudio	January
Alejandra Darling	Malalties Neurodegeneratives Associades a Dipòsit Cerebral de Ferro: caracterització clínica i genética	Belén Pérez Dueñas	February
Estrella Rubio Solsona	Estudio funcional de la proteína ANKK1 en el músculo esquelético: implicaciones en distrofias musculares	Janet Hoenicka y Francesc Palau	March
Tannia Valeria Carpio Arias	Aspectos cronobiológicos, emocionales y nutricionales en pacientes con Trastorno de Déficit de Atención e Hiperactividad	Jose Angel Alda	March
Gaia Botteri	Novel potential determinants in endoplasmic reticulum stress, inflammation and insulin resistance: Apo CIII and sAPPbeta	Manel Vazquez Carrera	March
Gemma Escuder Romeva	Necessitats de la població atesa a les unitats psiquiàtriques penitenciàries de Catalunya	Susana Ochoa Güerre	April
Miguel M ^a Alsina Casanova	Disfunción multiorgánica en el recién nacido con encefalopatía hipóxico-isquémica en la era de la hipotermia terapéutica	Alfredo Garcia-Alix Pérez	May
Ana Esteve Solé	Estudio de los defectos primarios y secundarios de la vía IL12- Interferón gamma-TNFalfa con susceptibilidad a la infección por microorganismos intracelulares.	Laia Alsina Manrique de Lara Manel Juan Otero	May
Albert Viel Oliva	Determinants estructurals i implicacions funcionals de l'interactoma proteic d'hCNT1	Marçal Pastor-Anglada	June
Hamsa Jameel Banjer	Role of the human Concentrative Nucleoside Transporter 1 (hCNT1) in oncogenesis	Marçal Pastor-Anglada	June
Gabriel Porcel Fernandez	Estudio del matrisoma y secretoma de fibroblastos humanos de piel, en el contexto de las distrofias musculares congenitas	Cecilia Jiménez Mallebrera	June
Guillem Pascual Pastó	Farmacocinètica i eficàcia preclínica de teràpies innovadores pel retinoblastoma refractari.	Angel Montero Carcaboso	June

Thesis Author	Title	Thesis directors	Date
Montserrat Torredadell Burriel	Análisis del valor pronóstico del perfil de expresión génica en la leucemia mieloblástica aguda de riesgo citogenético intermedio según la opción terapéutica post-remisión	Jordi Esteve Reyner Mireia Camós Guijosa	June
Anna Mensa-Vilaró	Implicació del mosaïcisme genètic en les immunodeficiències primàries	Juan Ignacio Aróstegui Jordi Antón	July
Daniel Natera	Application of CRISPR/Cas9 technology in collagen VI dominant mutations on human dermal fibroblasts	Caterina Marqués Gornés	July
Caterina Marques	Aplicación de CRISPR/Cas9 para editar mutaciones dominantes en genes de colágeno VI en fibroblastos	Cecilia Jiménez Mallebrera	July
Julia Rebeca Sevilla Llewellyn-Jones	Clinical personality traits in high risk and recent onset of psychosis patients	Berta Moreno Kustner Susana Ochoa Güerre	July
Mohammad Zarei	Unveiling the role of PPARbeta/delta on non-alcoholic fatty liver disease	Manel Vazquez Carrera	September
Nerea Vega García	Estudi del perfil d'expressió de les histones deacetilasa (HDAC) en pacients pediàtrics amb leucèmia aguda	Mireia Camós Guijosa	October
Júlia Ballester-Plané	Beyond the motor impairment in dyskinetic cerebral palsy: neuropsychological and connectome-based approach	Roser Pueyo Benito	October
Liska Caviedes Cárdenas	Proteína multifuncional ADE2 como nexo entre vías de recuperación y síntesis de novo de nucleósidos	Marçal Pastor-Anglada	October
Marta Ramon Krauel	Nutrició perinatal i programació de risc metabòlic a llarg termini: estudi de possibles mecanismes i intervencions en models d'experimentació animal.	Josep Jimenez-Chillarón Carles Lerín	October
Judit Cabana Dominguez	Genètica i dependència de cocaïna: Estudis d'associació, transcriptòmica i models animals	Bru Cormand Noèlia Fernández	October
Olga Laporta-Hoyos	Neuroimaging and executive function in dyskinetic cerebral palsy	Roser Pueyo Benito	October
Lorena Medina Pulido	Nuevas funciones de los transportadores concentrativos de nucleósidos CNT1 y CNT2.	Marçal Pastor-Anglada Sandra Pérez Torras	November
Sandra Moro Villa	Nuevas implicaciones de los transportadores concentrativos de nucleósidos asociadas a la biología tumoral	Marçal Pastor-Anglada Sandra Pérez Torras	November
Rosa Torres Ferraz	Pla de desenvolupament d'una nova combinació de fàrmacs pel tractament de la Síndrome de l'Ovari Poliquístic: del mercat al mercat	Lourdes Ibáñez Clara Campàs	Desember
Marta Chamorro Fernández	Evaluación de los patrones de sueño en niños con TDAH de nuevo diagnóstico comparado con controles sanos	Jose Angel Alda	Desember
Laura Domènech-Salgado	A comprehensive multiomics approach towards understanding obsessive-compulsive disorder	Raquel Rabionet Eulàlia Martí Xavier Estivill	Desember
Olga Casulleras Gras	Regulació del transportador de nucleòsids d'alta afinitat hCNT3 per proteïnes d'interacció	Marçal Pastor-Anglada Sandra Pérez Torras	Desember



SCIENTIFIC SEMINARS

- January, 9. Flu vaccination among healthcare personnel and vaccination recommendations for the adult population. **Ángela Domínguez García, PhD.**
- January, 23. Initial medication non-adherence: what lies behind this attitude?. **Montserrat Gil Girbau.**
- February, 1. 2nd Scientific Conference of the Institut de Recerca Sant Joan de Déu Barcelona (IRSJD).
- February, 6. Electrophysiological brain markers of cognitive function in neonatology and neuropaediatrics. **Carles Escera, PhD.**
- February, 20. Neonatal immune system in health and illness. **Laia Alsina, MD, PhD & Ana Esteve-Solé MSc.**
- March, 6. New insulin resistance mechanisms in skeletal muscle. **Gaia Botteri.**
- March, 20. Molecular mechanisms underlying development of the nervous system in health and disease. **Elisa Martí, PhD.**
- April, 10. NGS approaches in the study of complex disorders: a study of association of rare variants in obsessive-compulsive disorder. **Raquel Rabionet Jansse, PhD.**
- April, 24. Tyrosine hydroxylase deficiency: studies in patients and in a cellular model. **Alba Tristan Noguero.**
- May, 7. Training of executive function in childhood obesity: food choice, quality of life and brain connectivity. **Ma. Àngels Jurado, PhD.**
- May, 22. ANKK1 and the molecular bases of addiction: from vulnerable brain structure to induced changes. **Janet Hoenicka, PhD.**
- June, 5. Impact of a mindfulness intervention (MBSR) on clinical severity and inflammatory biomarkers in patients with fibromyalgia. **Laura Andrés Rodríguez.**
- June, 7. Early detection of infant's at risk for Cerebral Palsy: Early neuroimaging and clinical biomarkers. **Roslyn N. Boyd, PhD.**
- June, 16. Roche Scientific Seminar: Multidisciplinary approach of clinical microbiology. **Pedro Brotons, Carmen Muñoz-Almagro, PhD & MD.**
- June, 19. New generation sequencing for diagnosis of hereditary metabolic diseases. **Dèlia Yubero Siles, PhD.**
- June, 28. Calibration-based routines for imputing categorical variables in complex data. **Recal Yucel, PhD.**
- July, 17. Advances in regenerative medicine. **Gorka Oriv, PhD.**
- October, 9. Changes in GABAergic synapse in Rett syndrome. **Alfonso Luis de Oyarzabal Sanz, PhD.**
- October, 12th edition of the Course on Economic Assessment of Healthcare and Social Programmes and Services.
- October, 23. Gate2Brain peptides: from discovery to applications. **Meritxell Teixidó, PhD.**
- October, 24. 1st Discussion Conference IRSJD 2018 – Mental Health.
- November, 6. Mitochondrial DNA deletions: Quantitative evaluation of single and multiple deletions generation and expansion in single cells. **Selena Trifunov, PhD.**
- November, 20. EUDAIMON STUDY. Effectiveness of the MBSR programme in the treatment of fibromyalgia: a controlled randomised clinical trial. **Adrià Pérez Aranda.**
- November, 23 & 26. International mini-symposium: Models and therapies for Rare Diseases. **Cecília Jiménez-Mallebrera, PhD & Alba Tristán.**
- December, 4. Effectiveness of Metacognitive Training (MCT) in persons with recent-onset psychosis. **Susana Ochoa, PhD & MD.**
- December, 13. Workshop H2020: "How to Write a Competitive Proposal for Horizon 2020". **Sean McCarthy.**
- December, 18. Translational Data Science in Pediatric Oncology. **Soledad Gómez González, PhD.**



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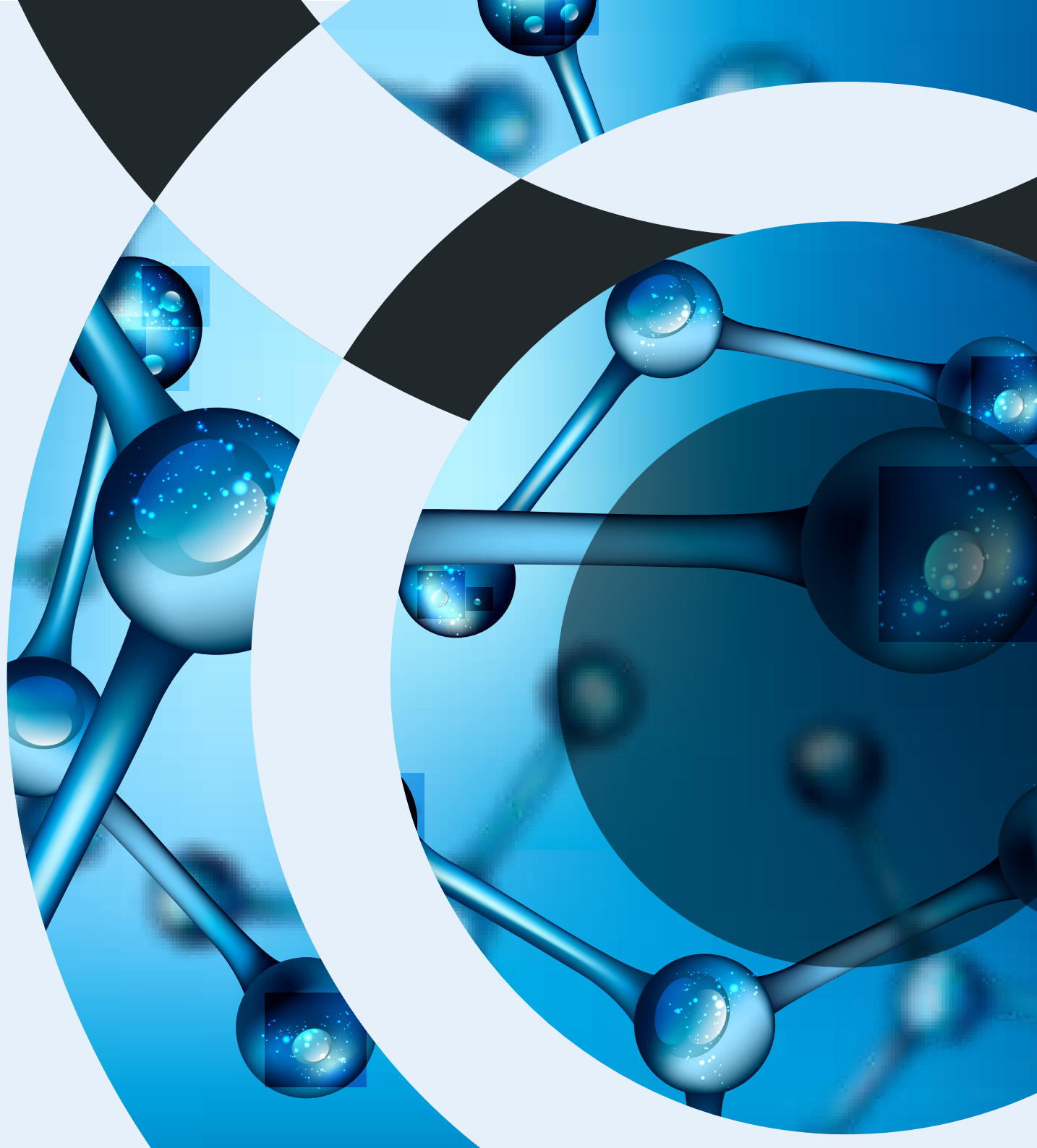
VERTICAL AREAS

- 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



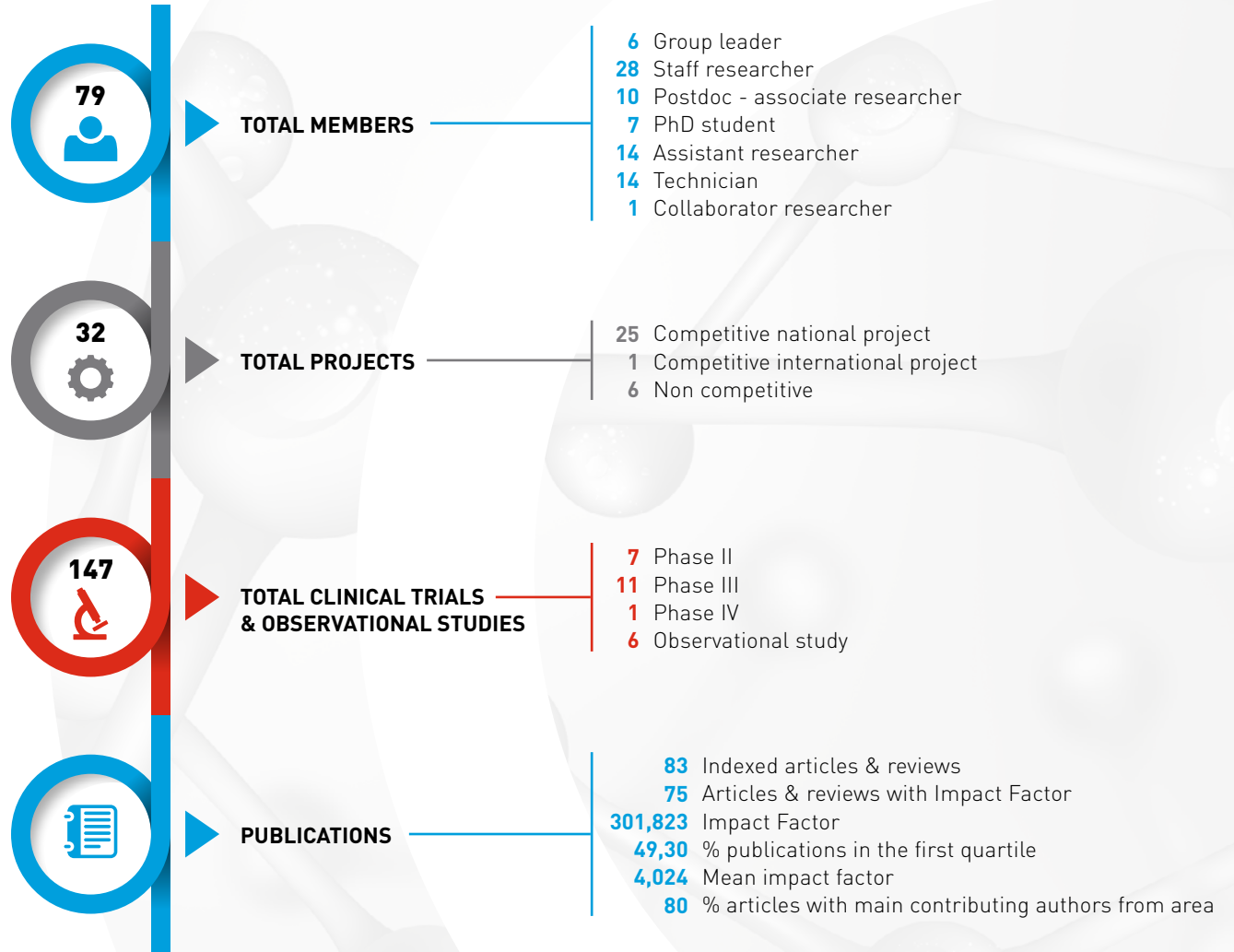
SJD

Sant Joan de Déu
Institut de Recerca



3.1. Paediatric neuroscience

Coordinators: Jaume Campistol Plana, MD, PhD and Rafael Artuch Iriberry, MD, PhD



PKU AND OTHER AMINO ACID GENETIC DISEASES

Jaume Campistol Plana, MD, PhD

Members

Staff researcher

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

Assistant researcher

Irene Galceran Nou

3

VERTICAL AREAS

3.1.

Pediatric
neuroscience

Coordinators:

Jaume Campistol Plana
Rafael Artuch Iriberrí



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. 15/11/2016-14/11/2018.

Selected clinical trials & observational studies

- 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. Codi EUDRACT: 2009-015844-41. Promotor: 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). Codi EUDRACT: 2013-002702-30. Promotor: Alexion Pharma Spain, S.L. PI: María Julieta González. 28/06/2016.

Selected publications

- Chakrapani A, Valayannopoulos V, Segarra NG, Del Toro M, Donati MA, Garcia-Cazorla A, et al. Effect of carglumic acid with or without ammonia scavengers on hyperammonaemia in acute decompensation episodes of organic acidurias. Orphanet J Rare Dis 2018. 13(1): 97-97. FI: 3,607(Q2)
- González MJ, Rebollo M, Ripollés P, Gassió-Subirachs R, Ormazabal-Herrero A, Sierra-March C, et al. White matter microstructural damage in early treated phenylketonuric patients. Orphanet J Rare Dis 2018. 13(1): 188-188. FI: 3,607(Q2)
- Tonduti D, Panteghini C, Pichiecchio A, Decio A, Carecchio M, Reale C, et al. Encephalopathies with intracranial calcification in children: clinical and genetic characterization. Orphanet J Rare Dis 2018. 13(1): 135-135. FI: 3,607(Q2)

Networks

- CIBERER
- TREAT-NMD

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

Rafael Artuch Iriberry, MD, PhD

This year's highlights

Our group increased its critical mass with the inclusion of two new post-doctoral researchers from another group in the neurosciences area who are expert clinical researchers on movement-related and neurodevelopmental disorders. We consequently increased our financing in competitive research projects and in the generation of scientific knowledge via publications. In the field of internationalisation, it may be noted that two members of our group were designated in 2018 as members of the scientific advisory board of the international laboratory quality control programme called ERNDIM, which is specialised in the analysis of biomarkers for the study of hereditary metabolic disorders.

Members

Staff researcher

Aida Ormazabal Herrero, Cristina Sierra March, Délia Yubero Siles, Mercedes Serrano Gimaré

Postdoc - associate researcher

Mercedes Casado Rio, Raquel Montero Sánchez, Juan Darío Ortigoza Escobar

PhD student

Abraham José Paredes Fuentes

Technician

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



PERSONAL RESEARCHER



PROJECTS



CLINICAL TRIALS & OBSERVATIONAL STUDIES



PUBLICATIONS WITH IF



FIRST QUARTILE PUBLICATIONS

Selected projects

- Molecular diagnosis of mitochondrial oxidative phosphorylation defects: pathogenesis of CoQ10 deficiencies. FIS 2017. Instituto de Salud Carlos III (ISCIII). PI: Rafa Artuch Iriberry. 01/01/2018-31/12/2020
- FIS 2017_Cerebellar atrophy in childhood: development of clinical-radiological tools for precision phenotyping, integral diagnostic algorithm, and identification of new genes. FIS 2017. Instituto de Salud Carlos III (ISCIII). PI: Mercedes Serrano Gimaré. 01/01/2018-31/12/2020
- 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. 01/01/2015-30/06/2018

Selected clinical trials & observational studies

- FSJD-PMM2-2016. Phase II clinical trial to assess the effectiveness and safety of acetazolamide in the treatment of cerebellar syndrome in patients with PMM2-CDG deficiency. Codi EUDRACT: 2017-000810-44. PI: Mercedes Serrano Gimaré. 02/06/2017-

Selected publications

- Batllori M, Molero M, Ormazabal A, Montero R, Sierra C, Ribes A, et al. Cerebrospinal fluid monoamines, pterins, and folate in patients with mitochondrial diseases: systematic review and hospital experience. *J Inher Metab Dis* 2018. 41(6): 1147-1158. FI: 4,092(Q1)
- Casado M, Sierra C, Batllori M, Artuch R and Ormazabal A. A targeted metabolomic procedure for amino acid analysis in different biological specimens by ultra-high-performance liquid chromatography-tandem mass spectrometry. *Metabolomics* 2018. 14(6): 76-76. FI: 3,511(Q2)
- Izquierdo-Serra M, Martínez-Monseny T, Lopez L, Carrillo-García J, Edo A, Ortigoza-Escobar JD, et al. Stroke-Like Episodes and Cerebellar Syndrome in Phosphomannomutase Deficiency (PMM2-CDG): Evidence for Hypoglycosylation-Driven Channelopathy. *Int J Mol Sci* 2018. 19(2): 619. FI: 3,687(Q2)
- Vilches C, Boiadjieva-Knopfel E, Bodoy S, Camargo S, Lopez de Heredia M, Prat-Reixach E, et al. Cooperation of Antiporter LAT2/CD98hc with Uniporter TAT1 for Renal Reabsorption of Neutral Amino Acids. *J Am Soc Nephrol* 2018. 29(6): 1624-1635. FI: 8,655(Q1)

Networks

- CIBERER: 2007
- ERN (metabERN). European reference networks: 2017
- EUROGLYCANomics: Grupo europeo CDG

APPLIED RESEARCH ON NEUROMUSCULAR DISORDERS

Cecilia Jiménez Mallebrera, MD, PhD

This year's highlights

We began 2019 with the award of two new grants for two projects that start off two research lines. One line, dealing with spinal muscular atrophy, is financed by the association Galicia AME for two years and its aim is to identify early biomarkers of response to treatment with antisense oligonucleotides in collaboration with Hospital de Sant Pau. The goal of the other research line is to investigate the efficiency of CRISPR/Cas genome-editing to correct dominant mutations in collagen VI genes and it is financed by CIBERER and by the association Noelia. These two grants have allowed us to contract pre-doctoral and post-doctoral personnel to carry out these projects.

Members

Staff researcher

Andrés Nascimiento Osorio, Irene Beatriz Zschaecck Luzardo, Julita Medina.

Postdoc - associate researcher

Daniel Natera de Benito, Selena Trifunov, Jaume Colomer

PhD student

Carlos Ignacio Ortez Gonzalez, Cristina Jou Muñoz, Sandra Pérez Ramos

Assistant researcher

Maria del Carmen Badosa Gallego, Laura Carrera García, Jessica Exposito Escudero

Collaborated researcher

Susana Graciela Kalko Witruk

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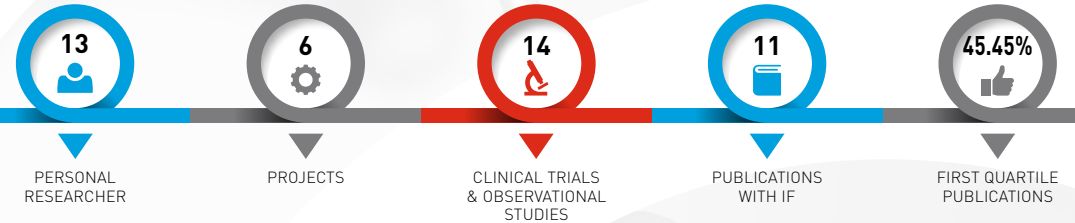
VERTICAL AREAS

3.1.

Pediatric neuroscience

Coordinators:

Jaume Campistol Plana
Rafael Artuch Iriberrí



Selected projects

- Application of proteomics and systems biology to the study of matrisome and miRNAs in congenital muscular dystrophies. FIS 2016. Instituto de Salud Carlos III (ISCIII). PI: Cecilia Jimenez Mallebrera. 01/01/2017-31/12/2019
- New therapeutic approach for Ullrich and Bethlem myopathies based on editing of dominant collagen VI mutations in patient fibroblasts. Obra Social Sant Joan de Déu. PI: Cecilia Jimenez Mallebrera. 08/10/2018-07/10/2021
- 780262, SHARE4RARE: Social media platform dedicated to rare diseases, using collective intelligence for the generation of awareness and advanced knowledge on this large group of diseases European Commission. H2020. PI: Begoña Nafria Escalera. (Fundació Privada per a la Recerca i la Docència Sant Joan de Déu - FSJD). 01/01/2018-31/12/2020. 562.500 €. Collaborator: Cecilia Jimenez-Mallebrera
- Delivery of RNA therapy: DART COST ACTION, European Commission. PI: Virginia Arechavala-Gomez. (Hospital de Cruces). 01/01/2018-31/12/2022. Collaborator: Cecilia Jimenez-Mallebrera, Sandra Perez

Selected clinical trials & observational studies

- A multicenter, randomised, double-blind, placebo-controlled, phase 3 safety and efficacy 48-week trial. Codi EUDRACT: 2017-004554-42. Promotor: Universitätskinderhospital beider Basel (UKBB). PI: Andres Nascimiento. 02/02/2018-
- Multicentre randomised parallel-group double-blind multiple-dose placebo-controlled study to assess the effectiveness and safety of MNK-1411 in male patients

aged 4 to 8 years with Duchenne muscular dystrophy. Codi EUDRACT: 2017-004139-35. Promotor: Mallinckrodt ARD Inc. PI: Andres Nascimiento. 29/06/2018

- Retrospective study of the combination of pyrimidine nucleos(t)ides in patients with thymidine kinase 2 (TK2) deficiency. PI: Andres Nascimiento. 02/11/2018-

Selected publications

- Batllori-Tragant M, Molero M, Ormazabal-Herrero A, Montero-Sanchez R, Sierra-March C, Ribes A, et al. Cerebrospinal fluid monoamines, pterins, and folate in patients with mitochondrial diseases: systematic review and hospital experience. *J Inherit Metab Dis* 2018. 41(6): 1147-1158. FI: 4,092(Q1)
- Darling A, Aguilera-Albesa S, Tello CA, Serrano M, Tomás M, Camino-León R, et al. PLA2G6-associated neurodegeneration: New insights into brain abnormalities and disease progression. *Parkinsonism Relat Disord* 2018. FI: 4,721(Q1)
- Garone C, Taylor RW, Nascimiento-Osorio A, Poulton J, Fratrer C, Dominguez-Gonzalez C, et al. Retrospective natural history of thymidine kinase 2 deficiency. *J Med Genet* 2018. 55(8): 515-521. FI: 5,751(Q1)
- Thompson K, Mai N, Oláhová M, Scialó F, Formosa LE, Stroud DA, et al. OXA1L mutations cause mitochondrial encephalopathy and a combined oxidative phosphorylation defect. *EMBO Mol Med* 2018. 10(11). FI: 10,293(Q1)

Networks

- CIBERER
- TREAT-NMD
- EURO-NMD: European reference network for neuromuscular diseases

NEUROMETABOLIC DISORDERS AND SYNAPTIC STUDIES IN RARE NEUROGENETIC CONDITIONS

M^a Àngels García Cazorla, MD, PhD

Members

Staff researcher

M^a del Mar O'Callaghan Gordo, Jordi Muchart López, Judith Armstrong Morón, Federico Ramos, Anna López Sala

Postdoc - associate researcher

Alfonso Luis de Oyarzabal Sanz, Alejandra Darling

PhD student

Alba Tristian Noguero

Assistant researcher

Ines Fernanda Medina Ribera, Uliana Musokhranova, Natalia Alexandra Julia Palacios

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VERTICAL AREAS

3.1.

Paediatric neuroscience

Coordinators:

Jaume Campistol Plana
Rafael Artuch Iriberrí



Selected projects

- U-IMD_Unified European Registry for Inherited Metabolic Disorders. Third-Programme HEALTH 2017. European Commission. PI: M^a Àngels García Cazorla. 01/02/2018-31/01/2021
- Search for new therapeutic indications in the field of neurodegenerative and mitochondrial diseases for the PPAR-gamma agonist MIN-102 compound. Proyectos Retos Colaboración 2018. Ministerio de Economía Y Competitividad (MINECO). PI: M^a Àngels García Cazorla. 01/01/2018-31/01/2019
- Study of the frequency of attention difficulties in paediatric patients with epilepsy and the effect of pharmacological treatment with methylphenidate. Laboratorios Rubió. PI: M^a Àngels García Cazorla. 31/07/2017-30/07/2019

Selected clinical trials & observational studies

- Open extension study to assess the long-term safety and effectiveness of UX007 in subjects with glucose transporter type-1 deficiency syndrome (Glut1DS). Codi EUDRACT: 2015-000389-69. Promotor: Ultragenyx Pharmaceutical, Inc. PI: M^a Àngels García Cazorla. 05/06/2017-
- Randomised double-blind placebo-controlled study to investigate the effectiveness and safety of cannabidiol (GWP42003-P, CBD) as a complementary therapy in patients with tuberous sclerosis complex suffering poorly controlled epileptic seizures. Codi EUDRACT: 2015-002154-12. Promotor: GW Research. PI: Federico Ramos. 20/02/2017-
- Effect of lamotrigine on cognitive deficits associated with neurofibromatosis type 1: a

random placebo-controlled phase-II study conducted at various centres (NF1-EXCEL). Codi EUDRACT: 2013-003405-26. Promotor: Erasmus MC. PI: Federico Ramos. 03/07/2017-

Selected publications

- Armangue-Salvador T, Spatola M, Vlagea A, Mattozzi S, Cárceles-Cordon M, Martinez-Heras E, et al. Frequency, symptoms, risk factors, and outcomes of autoimmune encephalitis after herpes simplex encephalitis: a prospective observational study and retrospective analysis. LANCET NEUROL 2018. 17(9): 760-772. FI: 27,138(Q1)
- Soto D, Olivella M, Grau C, Armstrong-Moron J, Alcon C, Gasull X, et al. RETRACTED: Rett-like Severe Encephalopathy Caused by a De Novo GRIN2B Mutation Is Attenuated by D-serine Dietary Supplement (Retracted article. See vol. 83, pg. 715, 2018). Biol Psychiatry 2018. 83(2): 160-172. FI: 11,982(Q1)
- Bosch de Basea M, Moriana D, Figuerola J, Barber I, Muchart-Lopez J, Lee C and Cardis E. Subtle excess in lifetime cancer risk related to CT scanning in Spanish young people. Environ Int 2018. 120: 1-10. FI: 7,297(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

NEUROGENETICS AND MOLECULAR MEDICINE

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

This year's highlights

The Neurogenetics and Molecular Medicine (NeuroGene) research group carries out its scientific and translational work in the field of experimental biomedicine for neurogenetic and neuropsychiatric disorders, and in the clinical sphere of the Paediatric Institute for Rare Diseases (IPER). The scientific milestones reached in 2018 include, among others: (1) Description of ANKK1 as a myogenesis biomarker and its relation to Duchenne muscular dystrophy (Rubio-Solsona et al. *PLoS One* 2018). (2) Relation between variants of the D3 dopamine receptor and addiction in early-onset Parkinson's disease (Castro-Martínez et al. *Parkinsonism Relat Disord* 2018). The training and dissemination activities included, among others, the coordination of the monographic issue "Science, medicine and reality of rare diseases" published by the journal *ARBOR*, and the work of F. Palau as editor-in-chief of the publication *Orphanet Journal of Rare Diseases*.

Members

RESEARCH LABORATORY

Postdoc - associate researcher

Laura Domínguez Berzosa, Lara Cantarero Abad

Assistant researcher

Montserrat Roura Llerda, Mireia Osuna López

PhD student

Azahara Civera Tregon, Elena Juárez Escoto, Abel Sola Bacaicoa

Technician

Dolors López López, Johanna Troya Balseca

CLINICAL RESEARCH

Staff researcher

Antonio Martínez Monseny, Mar Borregan Prats

Assistant researcher

Mercè Bolasell Girgas, Cesar Arjona Fernandez

CLINICAL LABORATORY - GENOMICS AND HUMAN GENETICS

Staff researcher

Loreto Martorell Sampol, Dèlia Yubero, Ma. Del Mar Pérez Iribarne,

Isabel Plensa Nebot, Cristina Hernando Devalillo

Assistant researcher

Núria Brandí, Silvia Vidal Falcó, Ainhoa Pascual Alonso

Technician

Jordi Genovès Escarré, Daniel Castillo Mestres, Paola Pacheco Fernández,

Patricia Peral, Sara Manresa, Teresa Zabala, Lluïsa Ramírez, Patricia Fernández

DANIEL BRAVO CENTER

Bioinformatics Unit

Guerau Fernández, Jan Maynou

Confocal Microscopy Unit

Mònica Roldan Molina

3

VERTICAL AREAS

3.1.

Paediatric neuroscience

Coordinators:

Jaume Campistol Plana
Rafael Artuch Iriberrí



PERSONAL
RESEARCHER



PROJECTS



PUBLICATIONS
WITH IF



FIRST QUARTILE
PUBLICATIONS

Selected projects

- GenomicScientia_Centre for Genomic Sciences in Medicine. Special institutional projects for construction, acquisition, conditioning and/or substantial expansion of buildings for R&D infrastructures. 2015. Departament de Salut - Generalitat de Catalunya. Projectes singulars institucionals de construcció, adquisició, habilitació i/o ampliació substancial d'edificacions per infraestructures d'R+D. PI: Francesc Palau Martínez. 01/01/2016-31/12/2018
- Role of ANKK1 in the genetics and pathophysiology of Parkinson's disease. Instituto de Salud Carlos III (ISCIII). FIS. PI15/01013. PI: Janet Hoenicka. 18/04/2016-31/12/2018
- 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. 01/01/2016-31/12/2019

Selected publications

- Casas-Alba D, Martínez-Monseny T, Pino-Ramírez RM, Alsina-Manrique de Lara L, Castejón Ponce E, et al. Hyaline fibromatosis syndrome: Clinical update and phenotype-

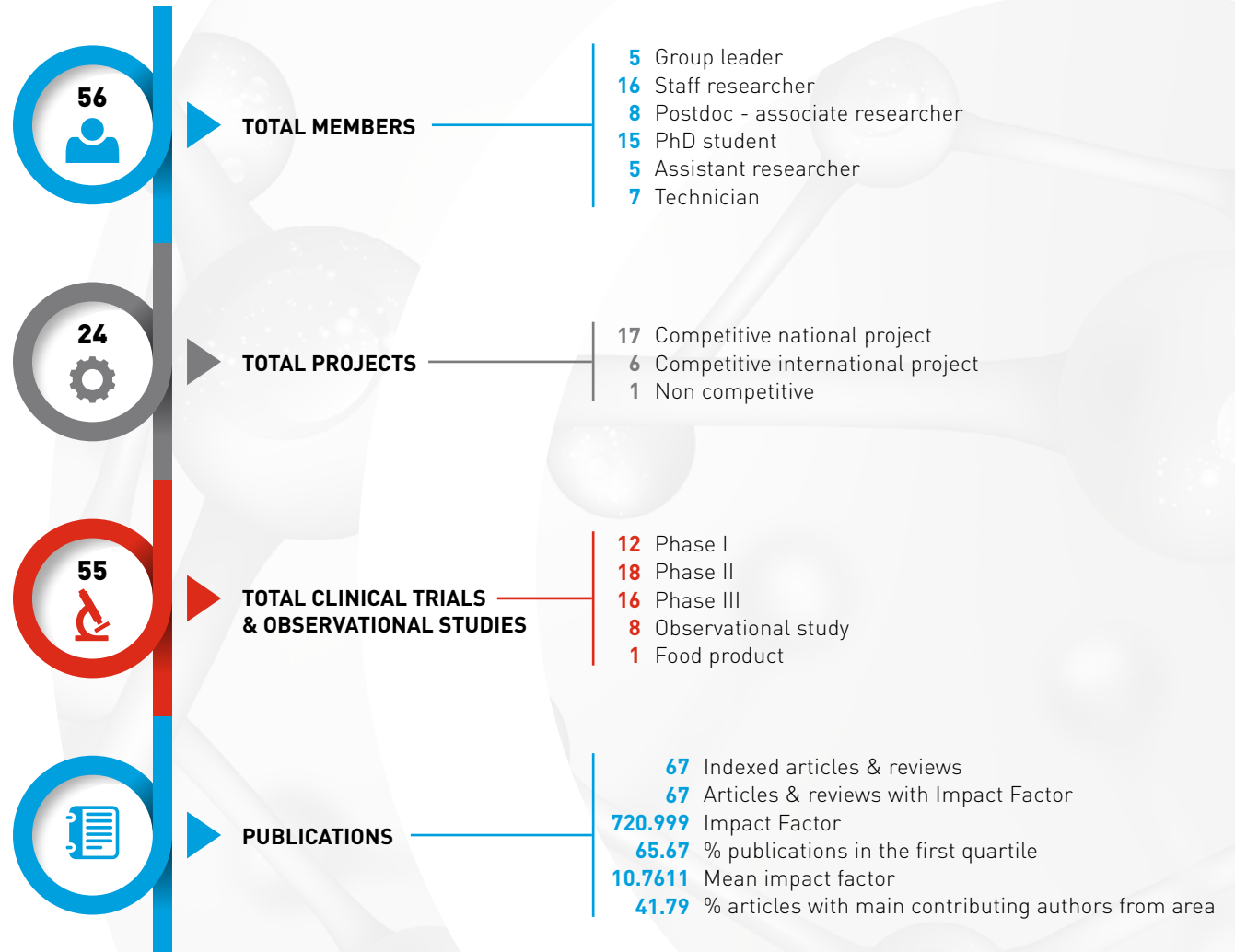
- genotype correlations. *Hum Mutat* 2018. 39(12): 1752-1763. FI: 5,359(Q1)
- Castro-Martínez X, García-Ruiz PJ, Martínez-García C, Martínez-Castrillo JC, Vela L, Mata M, et al. Behavioral addictions in early-onset Parkinson disease are associated with DRD3 variants. *Parkinsonism Relat Disord* 2018. 49: 100-103. FI: 4,721(Q1)
- Martínez-Monseny T, Mercè Bolasell Girgas, Arjona-Fernandez C, Martorell-Sampol L, Yubero-Siles D, Armstrong-Moron J, et al. Mutation of PACS1: the milder end of the spectrum. *Clin Dysmorphol* 2018. 27(4): 148-150. FI: 0,427(Q4)
- Rubio-Solsona E, Martí S, Vílchez JJ, Palau-Martínez F and Hoenicka J. ANKK1 is found in myogenic precursors and muscle fibers subtypes with glycolytic metabolism. *PLoS One* 2018. 13(5): e0197254. FI: 2,766(Q1)

Networks

- CIBERER, 2006
- CIBERSAM, 2007
- RD-ACTION - Data and Policies for Rare Diseases
- European Reference Network for Rare Neuromuscular Diseases (ERN-NMD)
- Orphanet-Spain

3.2. Molecular and cellular biology of developmental tumour

Coordinator: Jaume Mora Graupera, MD, PhD



PRECLINICAL PHARMACOLOGY AND DRUG RELEASE IN SOLID PAEDIATRICS TUMOURS

Angel Montero Carcaboso, PhD

3

VERTICAL AREAS

3.2.

Molecular and cellular
biology of developmental
tumour

Coordinator:
Jaume Mora Graupera

This year's highlights

Notable achievements in 2018 included the following: (i) Award of the Schweisguth Prize of the International Society for Paediatric Oncology (SIOP) for our research on a new treatment with oncolytic adenoviruses in retinoblastoma. This prize was awarded to Dr. Guillem Pascual Pastó and it was presented orally at the 50th SIOP Congress (Kyoto, Japan, 16-19 November 2018). (ii) Award of a competitive project of ISCiii to continue our work on retinoblastoma (starting in January 2019). (iii) Award of a Miguel Servet Type 2 contract of ISCiii (starting in January 2019). Our proposal scored the highest in the 2018 call.

Members

Staff researcher

Guillermo Chantada, Ofelia Cruz Martínez, Andrés Morales La Madrid

Postdoc - associate researcher

Gaia Botteri, Sonia Paco Mercader

PhD students

Helena Castillo Ecija, Maria Cuadrado Vilanova, Nagore Gene Olaciregui, Guillem Pascual Pastó

Technician

Mònica Vilà Ubach



PERSONAL
RESEARCHER



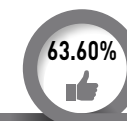
PROJECTS



CLINICAL TRIALS
& OBSERVATIONAL
STUDIES



PUBLICATIONS
WITH IF



FIRST QUARTILE
PUBLICATIONS

Selected projects

- Cure4RB. Ministry of Economy and Competitiveness (MINECO) - Cooperation challenges 2015. RTC-2015-4319-1. PI: Angel Montero Carcaboso. 01/07/2015-31/12/2018. The most significant scientific results are published in the journal Science Translational Medicine (January 2019). The clinical trial FSJD-RTB-2015 was continued in 2018 and patients continued to be recruited as well.
- Immune therapy of diffuse intrinsic pontine glioma (DIPG). Instituto de Salud Carlos III (ISCIII) - FIS. PI15/01161. PI: Angel Montero Carcaboso. 01/01/2016-31/12/2018. Part of this project's most significant scientific results were published in the journal Frontiers in Oncology (April 2018). The clinical trial FSJD-DIPG-DC completed the inclusion of patients.

Selected clinical trials & observational studies

- 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). Codi EUDRACT: 2015-003362-84. Promotor: Fundació Sant Joan de Déu. PI: Andrés Morales La Madrid. 06/06/2016 - We took part in the conception, preclinical justification and analysis of samples.
- 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. Codi EUDRACT: 2016-001060-11. Promotor: Fundació Sant Joan de Déu. PI: Jaume Catalá Mora. 03/08/2017-. We participated in the conception, preclinical justification, methodological design and analysis of samples, and shared the intellectual property.
- 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. Codi EUDRACT: 2016-002464-14. Promotor: Grupo Español de Investigación en Sarcoma Grupo GEIS. PI: Jaume Mora Graupera. 18/05/2017-. We took part in the preclinical justification and coordinated the translational study of the clinical trial.

Selected publications

- Bukchin A, Pascual-Pastó G, Cuadrado-Vilanova M, Castillo-Ecija H, Monterrubio C, Olaciregui NG, et al. Glucosylated nanomicelles target glucose-avid pediatric patient-derived sarcomas. J Control Release 2018. 276: 59-71. FI: 7,877(Q1)
- In this study we proved that the release of drugs to paediatric sarcomas (rhabdomyosarcomas and Ewing's sarcomas) can be increased, taking advantage of these tumours' glucose avidity. If we encapsulate the drugs with glucose-coated polymers, we obtain some micelles (a type of nanoparticles) that are captured by tumours better and more quickly than non-encapsulated drugs. The publication Journal of Controlled Release is one of the most highly reputed in the field of pharmacy and pharmacology.
- Manzanares-Quintela A, Restrepo-Perdomo CA, Botteri G, Castillo-Ecija H, Pascual-Pastó G, Cano F, et al. Tissue Compatibility of SN-38-Loaded Anticancer Nanofiber Matrices. Adv Healthc Mater 2018. 7(15): 1800255. FI: 5,609(Q1)
- This study is the first preclinical characterisation of the tissular compatibility of a new drug release system based on polymer nanofibre matrixes containing a powerful anticancer drug. We proved that nanofibre matrixes, placed on various healthy tissues including liver, blood vessels and nerves, produce only a mild local inflammatory reaction with no changes in the biochemical profiles relating to haematological, hepatic or renal activity. The journal Advanced Healthcare Materials is one of the most highly reputed publications in the field of biomedical engineering.
- Meel, M. H., M. C. de Gooijer, M. Guillen Navarro, P. Waranecki, M. Breur, L. C. M. Buil, L. E. Wedekind, J. W. R. Twisk, J. Koster, R. Hashizume, E. H. Raabe, A. Montero Carcaboso, M. Bugiani, O. van Tellinggen, D. G. van Vuurden, G. J. L. Kaspers and E. Hulleman (2018). "MELK Inhibition in Diffuse Intrinsic Pontine Glioma." Clin Cancer Res 24(22): 5645-5657.

Networks

- Innovative Therapies for Children with Cancer

LEUKEMIA AND NON-MALIGNANT BLOOD DISORDERS

Mireia Camós Guijosa, MD, PhD

This year's highlights

The Hereditary Erythropathology Unit of Hospital Sant Joan de Déu - Hospital de la Santa Creu i Sant Pau has been accredited as a CSUR (National Health System Reference Centre), which will help to strengthen our research activity in this field.

At the international meeting of the NOPHO group held in Gothenburg, Dr Montserrat Torredadell presented the nation-wide results of the clinical trial NOPHO-DBH-AML 2012 for acute myeloid leukaemia.

At our hospital, Dr Susana Rives leads various clinical trials of cell therapy with CART19, which provide a therapeutic alternative for patients with high-risk acute lymphoblastic leukaemia.

Members

Staff researcher

Isabel Badell Serra, Rubén Berruenco Moreno, Albert Català Guijosa, Izaskun Elorza Alvarez, Susanna Gassiot Riu, Julia Marsal Ricoma, Susana Rives Solà, Anna Ruiz Llobert, Montserrat Torredadell Burriel

Postdoc - associate researcher

Nerea Vega García

PhD students

Anna Alonso Saladrigues, Elena Esperanza Cebollada, Montserrat Messegué Meda, Sandra Martín Pont, Maria Trabazo del Castillo, Susanna Gassiot

Assistant researcher

Clara Vicente García

Technician

Maria Camino Estella Aguado, Lidia Ruiz Bocanegra, Warda Suleman



Selected projects

- Towards the personalisation of deep vein thrombosis prophylaxis in paediatric patients with acute lymphoblastic leukaemia: use of the thrombin generation test. FIS 2017. Carlos III Health Institute (ISCIII). FIS 2017. Instituto de Salud Carlos III (ISCIII). PI: Ruben Berruenco Moreno. 01/01/2018-31/12/2020
- Pilot study of infusion of genetically modified autologous T-lymphocytes to express anti-CD19 in patients with leukaemia or lymphoma CD19+ resistant or refractory to treatment. PICI 2014. Instituto de Salud Carlos III (ISCIII). PI: Susana Rives Solà. 01/01/2015-31/12/2018

Selected clinical trials & observational studies

- A Multicenter, Randomized, Phase III Registration Trial of Transplantation of NiCord®, Ex Vivo Expanded, Umbilical Cord Blood-derived, Stem and Progenitor Cells, versus Unmanipulated Umbilical Cord Blood for Patients with Hematological Malignancies. Codi EUDRACT: 2016-000704-28. Promotor: Gamida-Cell LTD. Cellt Therapy Technologies. PI: Isabel Badell Serra / Izaskun Elorza Álvarez. 04/10/2018-
- Multinational phase-IV study to assess the "real life" treatment pattern in previously treated patients with haemophilia A receiving KOVALTRY (octocog alpha) for routine prophylaxis. TAURUS. Promotor: Bayer Hispania, S.L. U. PI: Ruben Berruenco Moreno. 07/03/2018-

- Open multicentre phase-II trial to assess the effectiveness and safety of Daratumumab in paediatric and young-adult patients between ≥ 1 and ≤ 30 years with relapse or treatment-refractory acute lymphoblastic leukaemia or precursor B-cell or T-cell lymphoblastic lymphoma. Codi EUDRACT: 2017-003377-34. Promotor: Janssen-Cilag International NV. PI: Susana Rives Solà. 20/06/2018-

Selected publications

- Maude SL, Laetsch TW, Buechner J, Rives-Sola S, Boyer M, Bittencourt H, et al. Tisagenlecleucel in Children and Young Adults with B-Cell Lymphoblastic Leukemia. *N Engl J Med* 2018. 378(5): 439-448. FI: 79,258(Q1)
- Vega-García N, Malatesta R, Estella-Aguado MC, Pérez-Jaume S, Esperanza-Cebollada E, Torredadell-Burriel M, et al. Paediatric patients with acute leukaemia and KMT2A (MLL) rearrangement show a distinctive expression pattern of histone deacetylases. *Br J Haematol* 2018. 182(4): 542-553. FI: 5,128(Q1)
- Pérez-Rodríguez A, Battle J, Corrales I, Borràs N, Rodríguez-Trillo Á, Lourés E et al. Role of multimeric analysis of von Willebrand factor (VWF) in von Willebrand disease (VWD) diagnosis: Lessons from the PCM-EVW-ES Spanish project. *PLoS One* 2018. 13(6): e0197876. FI: 2,766(Q1)

Networks

- Innovative Therapies for Children with Cancer
- Grup clinic vinculat CIBER malalties rares (CIBERER), Instituto de Salud Carlos III. 2015-

3

VERTICAL AREAS

3.2.

Molecular and cellular biology of developmental tumour

Coordinator:

Jaume Mora Graupera

NEUROBLASTOMA

Carmen De Torres Gómez-Pallete, MD, PhD

Members

Postdoc - associate researcher

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

PhD student

Elia Carolina Gonçalves Alves.

Technician

Noelia Salvador Marcos

3

VERTICAL AREAS

3.2.

Molecular and cellular
biology of developmental
tumour

Coordinator:

Jaume Mora Graupera



PERSONAL
RESEARCHER



PROJECTS



PUBLICATIONS
WITH IF



FIRST QUARTILE
PUBLICATIONS

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
- 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
- Combined therapies for neuroblastoma based on the calcium receptor-sensor. FIS 2017. Instituto de Salud Carlos III (ISCIII). Proyectos de investigación. PI: Carmen De Torres Gómez-Pallete. 01/01/2018-31/12/2020

Selected publications

- de la Escosura-Muñiz A, Espinoza-Castañeda M, Chamorro-García A, Rodríguez-Hernandez CJ, de Torres C and Merkoçi A. In situ monitoring of PTHLH secretion in neuroblastoma cells cultured onto nanoporous membranes. BIOSENS BIOELECTRON 2018. 107: 62-68. FI: 8,173(Q1)

- Gomez-Gonzalez S, Garrido- García A, García-Gerique L, Lemos I, Suñol M, de Torres C, et al. A Novel Method for Rapid Molecular Subgrouping of Medulloblastoma. Clin Cancer Res 2018. 24(6): 1355-1363. FI: 10,199(Q1)
- Tamborero D, Rubio-Perez C, Deu-Pons J, Schroeder MP, Vivancos A, Rovira A, et al. Cancer Genome Interpreter annotates the biological and clinical relevance of tumor alterations. Genome Med 2018. 10(1): 25-25. FI: 8,898(Q1)
- Vinci M, Burford A, Molinari V, Kessler K, Popov S, Clarke M, et al. Functional diversity and cooperativity between subclonal populations of pediatric glioblastoma and diffuse intrinsic pontine glioma cells. Nat Med 2018. 24(8): 1204-1215. FI: 32,621(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.

SARCOMA AND HISTIOCYTOSIS

Jaume Mora Graupera, MD, PhD

This year's highlights

2018 was marked by the completion of the study period on Ewing's sarcoma financed by the cooperative group grant of AECC (5 years). It has been a key driving factor for the group in the study of the epigenetics of Ewing's sarcoma and it should culminate in 2019 with the most ambitious publication in 5 years, which is the outcome of the work carried out during these 5 years. The continuity of the study in this field is ensured by the acquisition of the FIS project now in progress, the new interactions with outstanding groups in this field, such as the CRG group led by Luciano di Croce, and the cooperation with Angel Raya's group of the Reparative Medicine Institute of Catalonia. 2018 was marked by the group's international positioning with the largest number of immunotherapy clinical trials to date through the Ymabs company for the treatment of neuroblastoma patients. These studies draw patients from around the world and this has allowed us to acquire a uniquely broad experience with an immense number of patients. This number continues to grow, with all that this entails in terms of acquiring new knowledge and generating treatment protocols specific to our group.

Members

Staff researcher

Verónica Celis Passini, Moira Garraus Oneca, María Inmaculada Hernandez Muñoz

Postdoc - associate researcher

Sara Sánchez Molina

Assistant researcher

Estela Prada Varela, Alejandro Manzanares Quintela, Sara Pérez Jaume.

PhD student

Elisabet Figuerola Bou, Maria Sanchez Jimenez

Technician

Maria Jesus Nagel Moyano

3

VERTICAL AREAS

3.2.

Molecular and cellular biology of developmental tumour

Coordinator:

Jaume Mora Graupera



PERSONAL RESEARCHER



PROJECTS



CLINICAL TRIALS & OBSERVATIONAL STUDIES



PUBLICATIONS WITH IF



FIRST QUARTILE PUBLICATIONS

Selected projects

- Search for and validation and clinical translation of new therapeutic targets based on integrative genomics and proteomics studies on Ewing's sarcoma. Grupos estables. Modalidad B 2013. Asociación Española Contra el Cáncer. Grupos Estables Mod. B. PI: Jaume Mora Graupera. 01/10/2013-31/12/2018
- Characterisation and relevance of the epigenetic environment in the development of Ewing's sarcoma. FIS 2016. Instituto de Salud Carlos III (ISCIII). Proyectos de investigación en salud. PI: Jaume Mora Graupera. 01/01/2017-31/12/2019

Selected clinical trials

- FSJD-RTB-2015. Single-centre, open Phase I clinical trial with escalating doses to assess the safety and activity of VCN-01 oncolytic adenovirus in patients with refractory retinoblastoma. Código EUDRACT: 2016-001060-11. Promotor: Fundació Privada per a la Recerca i la Docència Sant Joan de Déu - FSJD FSJD. PI: Jaume Català Mora. 06/09/2017-
- 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. Código EUDRACT: 2016-001060-11. Promotor: Grupo Español de Investigación en Sarcoma Grupo GEIS. PI: Jaume Mora Graupera. 18/05/2017-
- Y-MABS 201. Pivotal phase 2 trial of antibody naxitamab (hu3F8) and

granulocyte-macrophage colony stimulating factor (GM-CSF) in high-risk neuroblastoma patients with primary refractory disease or with incomplete response to salvage treatment in bone and/or bone marrow. Codi EUDRACT: 2017-001829-40. Promotor: Y-mAbs Therapeutics A/S. PI: Jaume Mora Graupera. 05/03/2018

Selected publications

- Ding YY, Panzer J, Maris JM, Castañeda-Heredia A, Marta Gómez Chiari and Mora J. Transverse myelitis as an unexpected complication following treatment with dinutuximab in pediatric patients with high-risk neuroblastoma: A case series. *Pediatr Blood Cancer* 2018. 65(1): e26732. FI: 2,646(Q1)
- Filbin MG, Tirosh I, Hovestadt V, Shaw ML, Escalante LE, Mathewson ND, et al. Developmental and oncogenic programs in H3K27M gliomas dissected by single-cell RNA-seq. *Science* 2018. 360(6386): 331-335. FI: 41,058(Q1)
- Gomez-Gonzalez S, Garrido- García A, García-Gerique L, Lemos I, Suñol M, de Torres C, et al. A Novel Method for Rapid Molecular Subgrouping of Medulloblastoma. *Clin Cancer Res* 2018. 24(6): 1355-1363. FI: 10,199(Q1)
- Mora J, Pérez-Jaume S and Cruz-Martínez O. Treatment of childhood astrocytomas with irinotecan and cisplatin. *Clin Transl Oncol* 2018. 20(4): 500-507. FI: 2,392(Q3)

Networks

- Innovative Therapies for Children with Cancer

TRANSLATIONAL GENOMICS

Cinzia Emilia Lavarino, MD, PhD

This year's highlights

- Publication of the method of classification in molecular subgroups of patients with medulloblastoma - S. Gómez et al. *Clinical Cancer Research* 2018.
- Prize for the best scientific publication IRSJD 2018.
- International patent (Registration no. OEPM - PCT/ES2018/070347). Title: Method for classification of medulloblastoma in molecular subgroups.
- Implementation of the method for molecular classification of medulloblastoma in the clinical routine of our centre.
- Implementation of the Omics Design, Analysis and Interpretation (DAI-Omics) Unit in the catalogue of services of the Sant Joan de Déu Barcelona Research Institute.

Members

Staff researcher

Héctor Salvador Hernández

Postdoc - associate researcher

Soledad Gomez Gonzalez

Assistant researcher

Alba Parra Checa

PhD student

Laura Garcia Gerique, Alicia Garrido Garcia

Technician

Isadora Lemos

3

VERTICAL AREAS

3.2.

Molecular and cellular biology of developmental tumour

Coordinator:

Jaume Mora Graupera



Selected projects

- Method for classification in molecular subgroups of patients with medulloblastoma. FIS 2017. Instituto de Salud Carlos III (ISCIII). Proyectos de investigación. PI: Cinzia Emilia Lavarino. 01/01/2018-31/12/2020
- 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. 01/01/2015-31/12/2018.

Selected clinical trials & observational studies

- SPAIN-CCSS. Creation of a Spanish cohort of childhood cancer survivors: SPAIN-CCSS cohort. Promotor: Fundació Privada ISGLOBAL. PI: Héctor Salvador Hernández. 20/01/2014-

Selected publications

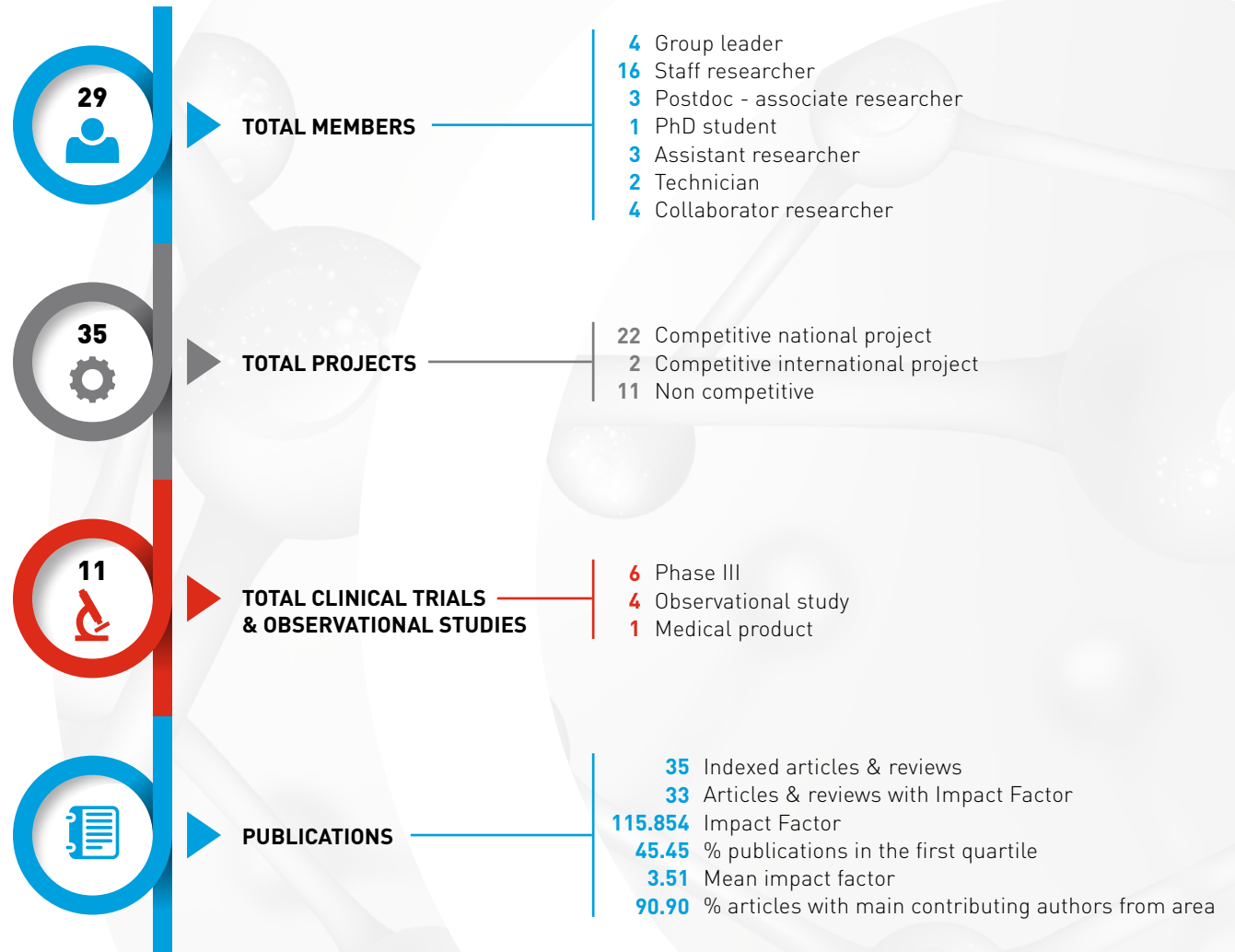
- Castellanos E, Plana A, Carrato C, Carrio M, Rosas I, Amilibia E, et al. Early Genetic Diagnosis of Neurofibromatosis Type 2 From Skin Plaque Plexiform Schwannomas in Childhood. *JAMA Dermatol* 2018. 154(3): 341-346. FI: 8,107(Q1)
- Filbin MG, Tirosh I, Hovestadt V, Shaw ML, Escalante LE, Mathewson ND, et al. Developmental and oncogenic programs in H3K27M gliomas dissected by single-cell RNA-seq. *Science* 2018. 360(6386): 331-335. FI: 41,058(Q1)
- Gomez-Gonzalez S, Garrido-García A, García-Gerique L, Lemos I, Suñol M, de Torres C, et al. A Novel Method for Rapid Molecular Subgrouping of Medulloblastoma. *Clin Cancer Res* 2018. 24(6): 1355-1363. FI: 10,199(Q1)
- Soria Gondek A, Julià V, Jou-Munoz C, Salvador-Hernandez H, Rovira-Zurriaga C and Tarrado X. Adolescent Hydrocele Carrying a Surprise: A Case of Papillary Cystadenoma of the Epididymis. *Urology* 2018. 112: 172-175. FI: 2,300(Q2)

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

Coordinator:
Lourdes Ibáñez Toda

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

Coordinator: Lourdes Ibáñez Toda, MD, PhD



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

Coordinator:
Lourdes Ibáñez Toda

METABOLIC ENDOCRINOLOGY INCLUDING OVARIAN HYPERANDROGENISM, INSULIN RESISTANCE, OBESITY, METABOLIC SYNDROME AND DIABETES

Lourdes Ibáñez Toda, MD, PhD

This year's highlights

In 2018 the metabolic endocrinology group has focused its research on the longitudinal prospective study of epigenetic, clinical and biochemical aspects of subjects born small for gestational age (SGA), and of adolescents with ovarian hyperandrogenism or polycystic ovary syndrome (PCOS). The longitudinal cohorts of SGA children and of adolescents with PCOS have been followed up, and new patients have been recruited, allowing us to determine the relation between liver volume and fat, and visceral adiposity in prepubertal SGA children (Malpique et al. Int J Obes 2018), and the effects of a new combination of insulin action sensitizers and an antiandrogen (SPIOMET) on Fetuin-A levels in adolescents with PCOS (Díaz-Silva M et al. Int J Endocrinol 2018).

Members

Staff researcher

Paula Casano Sancho, Marta Díaz Silva

Postdoc students

Rita Maria Martins de Sousa Maia Malpique, Georgia Sebastiani

Collaborator researcher

Judith Bassols Casadevall, Francis de Zegher, Abel López Bermejo

Assistant researcher

Cristina García Beltran, Esther Lizagarra Mollinedo, Cristina Plou Pérez



Selected projects

- Pioglitazone+spironolactone+metformin versus oral contraception in adolescents with ovarian hyperandrogenism and hyperinsulinism. Instituto de Salud Carlos III (ISCIII). FIS 2015. PI: Lourdes Ibáñez Toda. 01/01/2016-31/12/2019
- MiRNAs and metabolic disorders of foetal origin: pathophysiological mechanisms and risk markers. Hospital Sant Joan de Déu - Esplugues HSJD. BEQUES RECERCA 2016. PI: Lourdes Ibáñez Toda. 07/03/2017-06/03/2020

Selected clinical trials & observational studies

- FSJD-PIOSPIMET-2015. Random open prospective clinical trial with two groups of patients to assess the effectiveness and safety of the combination of ethinyl estradiol and levonorgestrel versus the combination of pioglitazone, spironolactone and metformin at low doses in adolescents with ovarian hyperandrogenism and hyperinsulinism: effects on ovulatory function, chronic inflammation parameters, markers of treatment evolution and effectiveness, and of development of type 2 diabetes. Codi EUDRACT: 2015-005092-24. Fundació Privada per a la Recerca i la Docència Sant Joan de Déu - FSJD FSJD. PI: Lourdes Ibáñez Toda. 11/02/2016-

- WI191603. Treatment with recombinant growth hormone (rGH) in short patients born small-for-gestational-age (SGA) followed at Hospital Sant Joan de Déu: Analysis of early predictors of good response useful in clinical practice. Fundació Privada per a la Recerca i la Docència Sant Joan de Déu - FSJD FSJD. PI: Lourdes Ibáñez Toda. 14/11/2014-

Selected publications

- Malpique R, Bassols-Casadevall J, López-Bermejo A, Díaz-Silva M, Villarroya F, Pavia J, et al. Liver volume and hepatic adiposity in childhood: relations to body growth and visceral fat. Int J Obes (Lond) 2018. 42(1): 65-71. FI: 5,151(Q1)
- de Zegher F, López-Bermejo A and Ibáñez-Toda L. Central Obesity, Faster Maturation, and 'PCOS' in Girls. Trends Endocrinol Metab 2018. 29(12): 816-819. FI: 10,769(Q1)
- Díaz-Silva M, García-Beltran C, López-Bermejo A, de Zegher F and Ibáñez-Toda L. GLP-1 and IGF-I levels are elevated in late infancy in low birth weight infants, independently of GLP-1 receptor polymorphisms and neonatal nutrition. Int J Obes (Lond) 2018. 42(4): 915-918. FI: 5,151(Q1)

Networks

- CIBERDEM

3 VERTICAL AREAS

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

Coordinator:
Lourdes Ibáñez Toda

EARLY ORIGINS OF METABOLIC DISEASE

José Carlos Jiménez Chillarón, PhD &
Carles Lerin Martínez, PhD

This year's highlights

Three new projects subsidised by MINECO and Barcelona Magic Line were started, in addition to the publication of 5 articles in scientific journals. Likewise, one patent was granted and an application was submitted for a second patent.

Members

Staff researcher

Ruben Díaz Naderi, Marta Ramon Krauel

Postdoc students

Francesc Ribas Aulinas

PhD student

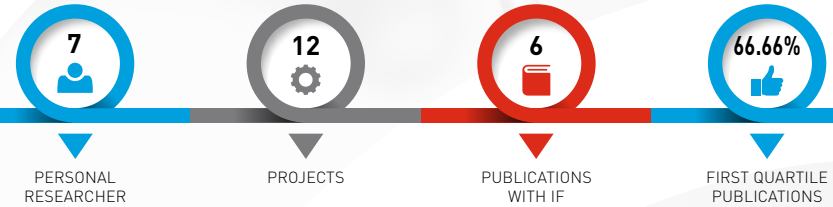
Montse Amat Bou

Collaborator researcher

Ivonne Palacios, Marin

Technician

Judith Cebrià Romeo



Selected projects

- BetAkk: Intestinal microbiota in the first stages of life and long-term risk of obesity and metabolic disorders. Retos Investigación: Proyectos I+D+i. Ministerio de Economía Y Competitividad (MINECO). PI: Carles Lerin Martínez. 13/04/2018-31/12/2020.
- TransEpiR: Non-genomic inheritance of obesity through small non-coding RNAs. Retos Investigación: Proyectos I+D+i. Ministerio de Economía Y Competitividad (MINECO). PI: Josep Jimenez Chillarón. 13/04/2018-31/12/2020.
- Encapsulated Synthetic Cellular Circuits to Restore Glycemic Control in Type 1 Diabetes. Convocatòria de projectes sobre diabetes i obesitat. Fundació La Marató de TV3. PI: Ruben Díaz Naderi. 22/05/2017-21/05/2020.

Selected publications

- Leal-Witt MJ, Llobet-Garcès M, Samino S, Castellano P, Cuadras-Palleja D, Jimenez-Chillarón JC, et al. Lifestyle Intervention Decreases Urine Trimethylamine N-Oxide Levels in Prepubertal Children with Obesity. *Obesity (Silver Spring)* 2018. 26(10): 1603-1610. FI: 4,042(Q1)
- Leal-Witt MJ, Ramon-Krauel M, Samino S, Llobet-Garcès M, Cuadras-Palleja D, Jimenez-Chillarón JC, et al. Untargeted metabolomics identifies a plasma sphingolipid-related signature associated with lifestyle intervention in prepubertal children with obesity. *Int J Obes (Lond)* 2018. 42(1): 72-78. FI: 5,151(Q1)
- Montori M, Pedreira-Casahuga R, Boyer-Díaz Z, Lassot I, García-Martínez C, Orozco A, et al. GNIP1 E3 ubiquitin ligase is a novel player in regulating glycogen metabolism in skeletal muscle. *Metabolism* 2018. 83: 177-187. FI: 5,963(Q1)
- Pellitero S, Piquer-Garcia I, Ferrer-Curriu G, Puig R, Martínez Franco E, Moreno P, et al. Opposite changes in meteorin-like and oncostatin m levels are associated with metabolic improvements after bariatric surgery. *Int J Obes (Lond)* 2018. 42(4): 919-922. FI: 5,151(Q1)
- Ruperez CH, Lerin C, Ferrer G, Cairo M, Mas-Stachurska A, Sitges M, et al. Autophagic control of cardiac steatosis through FGF21 in obesity-associated cardiomyopathy. *Int J Cardiol* 2018. 260: 163-170. FI: 4,034(Q2)

NEONATAL PHYSIOPATHOLOGY

Alfredo García-Alix Pérez, MD, PhD

Members

Staff researcher

Thais Agut Quijano, Miguel M^a Alsina Casanova, Marta Camprubi Camprubi, Ruth del rio Florentino, Isabel Iglesias Platas, Martin Iriondo Sanz, Ana Martin Ancel, Julia Hernando Moreno, Ana Morillo Palomo, Africa Pertierra Cortada, Ana Riverola de Veciabam, Javier Rodríguez Fanjul

3

VERTICAL AREAS

3.3.

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

Coordinator:
Lourdes Ibáñez Toda



Selected projects

- Rare Coding Genetic Variants Associated with neonatal arterial ischemic stroke. Fundació La Marató de TV3. PI: Alfredo García-Alix. 13/03/2018-11/03/2021.
- Human milk: Nutrients and bioactive factors. Relation to intestinal microbiota development, growth and health of preterm newborns. FIS 2017. Instituto de Salud Carlos III (ISCIII). PI: Isabel Iglesias Platas. 01/01/2018-31/12/2020.

Selected clinical trials & observational studies

- Effect of alopurinol on patients with hypoxic-ischaemic encephalopathy treated with hypothermia. ALBINO multicentre randomised blind placebo-controlled clinical trial. Codi EUDRACT: 2016-000222-19. Promotor: Instituto de investigación Sanitaria Hospital La Fe. PI: Marta Camprubi Camprubi. 05/05/2017-
- Evaluation of Real Time Continuous Glucose Monitoring in the very preterm infant. Codi. University of Cambridge. PI: Isabel Iglesias Platas. 04/05/2017

Selected publications

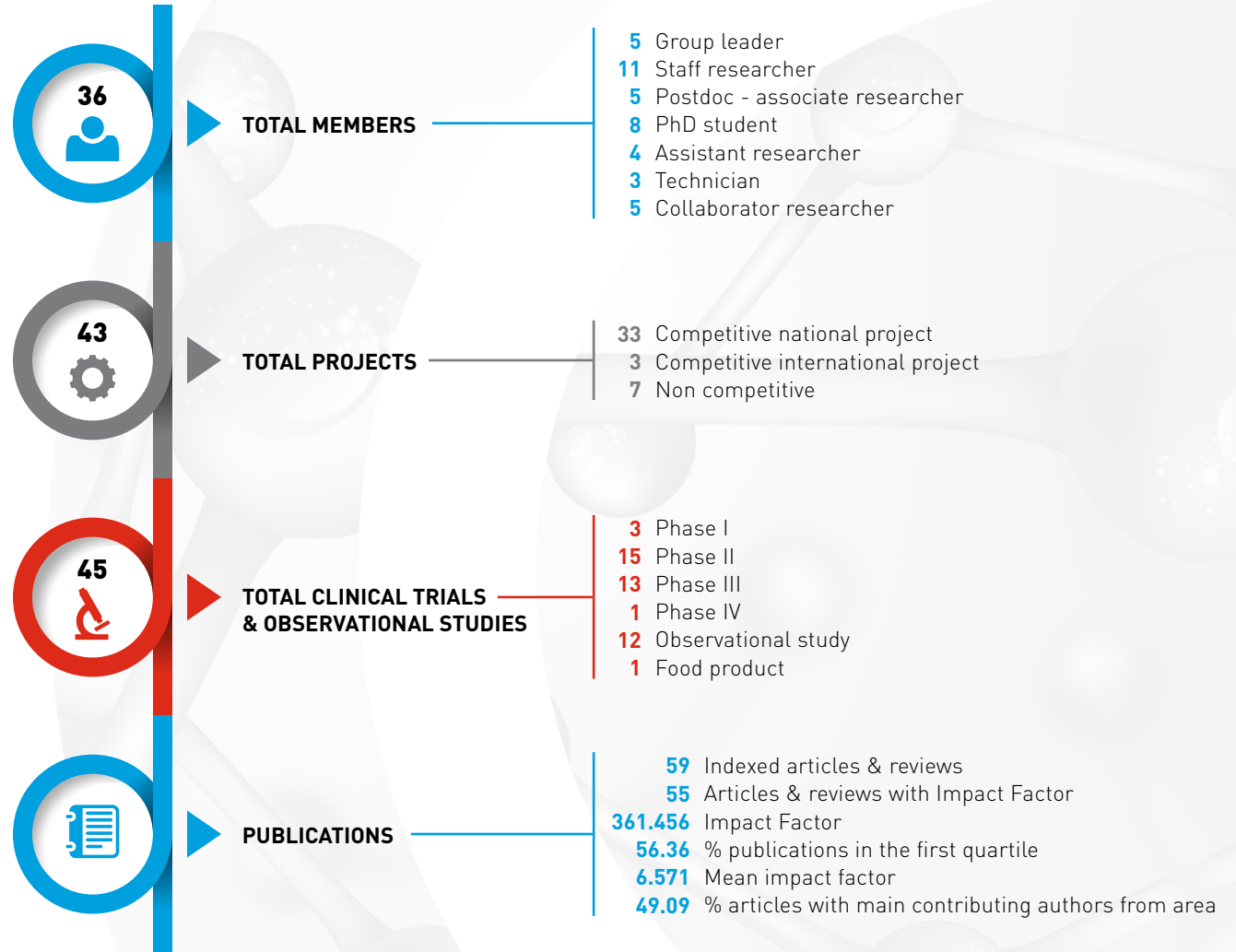
- Arnaez J, Arca-Diaz G, Martín-Ancel A and García-Alix A. Coagulation factor V G1691A, factor II G20210A and methylenetetrahydrofolate reductase C677T gene mutations do not play a major role in symptomatic neonatal arterial ischaemic stroke. *Br J Haematol* 2018. 180(2): 290-292. FI: 5,128(Q1)
- Casas-Alba D, Martínez-Monseny T, Pino-Ramírez RM, Alsina-Manrique de Lara L, Castejón Ponce E, Navarro-Vilarrubi S, et al. Hyaline fibromatosis syndrome: Clinical update and phenotype-genotype correlations. *Hum Mutat* 2018. 39(12): 1752-1763. FI: 5,359(Q1)
- Hernández Mora JR, Sánchez-Delgado M, Petazzi P, Moran S, Esteller M, Iglesias-Platas I and Monk D. Profiling of oxBS-450K 5-hydroxymethylcytosine in human placenta and brain reveals enrichment at imprinted loci. *Epigenetics* 2018. 13(2): 182-191. FI: 4,918(Q1)

Networks

- BCNatal

3.4. Infectious diseases and systemic inflammatory response in paediatrics

Coordinator: Carmen Muñoz-Almagro, MD, PhD



VACCINE-PREVENTABLE DISEASES. DIAGNOSTIC INNOVATION TRANSVERSAL SUBLINE

Carmen Muñoz Almagro, MD, PhD

3

VERTICAL AREAS

3.4.

Infectious diseases and systemic inflammatory response in paediatrics

Coordinator:
Carmen Muñoz-Almagro

This year's highlights

In 2018 the group continued to consolidate its research activity with 5 FIS projects on characterisation of nasopharyngeal microbiota in invasive pneumococcal disease, characterisation of nasopharyngeal microbiota in rhinovirus infection, molecular characterisation of B. pertussis, impact of adding PCV13 vaccine to the systematic calendar, and assessment of a new diagnostic algorithm for severe pneumonia in PICU. Likewise, the group has continued to cooperate with research networks of excellence of both national (CIBERESP) and European (PERTINENT, SPIDNET) scope, as well as with industry, conducting 4 validation or verification studies on new rapid diagnosis devices for infections and 5 pharmacological clinical trials. These activities have been disseminated through 12 scientific articles published in impact journals (5 of them in the first quartile).

Members

Staff researcher

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

Post-doc researchers

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

PhD students

Sara Bobillo Perez, Didac Casas Alba, Desiree Henares Bonilla, Míriam Triviño Rodríguez

Assistant researcher

Selene Garcia Garcia, Susana Hernández Bou

Technician

Carles Cisneros Camps, Amaresh Pérez Argüello, Jesica Saucedo Verdes



PERSONAL
RESEARCHER



PROJECTS



CLINICAL TRIALS
& OBSERVATIONAL
STUDIES



PUBLICATIONS
WITH IF



FIRST QUARTILE
PUBLICATIONS

Selected projects

- Assessment of the Safety and Clinical Performance Characteristics of the DiagCORE Gastrointestinal Panel Assay. STAT-Dx Life S.L. PI: Carmen Muñoz Almagro. 05/02/2018-16/03/2018.
- Human rhinovirus (HRV) in children under age 5 years: from the common cold to the intensive care unit - the role of microbiota. Proyectos de investigación. Instituto de Salud Carlos III (ISCIII). PI: Cristina Luanes Montaña. 01/01/2018-31/12/2020.
- Evolution of pediatric invasive pneumococcal disease after the introduction of the tridecavalent pneumococcal conjugate vaccine in the systematic calendar. Proyectos de investigación. Instituto de Salud Carlos III (ISCIII). PI: Juan José García García. 01/01/2018-31/12/2020.

Selected clinical trials & observational studies

- Study of influenza virus infection in children hospitalised in Spain in two consecutive flu seasons. Direct medical cost analysis. Promotor: Asociación Instituto Bionostia. PI: Juan José García García. 10/05/2017-
- Multicentre open randomised phase-II/III study to determine the safety and effectiveness of solithromycin in adolescents (12 to 17 years of age, both inclusive) and children (> 2 months to < 12 years of age) with suspected or confirmed community-acquired bacterial pneumonia. Codi EUDRACT: 2014-004039-37. Promotor: Cempra Pharmaceuticals, Inc. PI: Juan José García García. 12/09/2016-

- Phase-III multicentre open randomised comparator-controlled study to assess the safety and effectiveness of dalbavancin versus an active comparator in paediatric patients with acute bacterial infections of the skin and soft tissues. Codi EUDRACT: 2014-005281-30. PI: Juan José García García. 02/12/2016-

Selected publications

- Casas-Alba D, Martinez-Monseny T, Pino-Ramirez RM, Alsina-Manrique de Lara L, Castejón Ponce E, Navarro-Vilarrubi S, et al. Hyaline fibromatosis syndrome: Clinical update and phenotype-genotype correlations. Hum Mutat 2018. 39(12): 1752-1763. FI: 5,359(Q1)
- Balsells E, Dagan R, Yildirim I, Gounder PP, Steens A, Munoz-Almagro C, et al. The relative invasive disease potential of Streptococcus pneumoniae among children after PCV introduction: A systematic review and meta-analysis. J Infect 2018. 77(5): 368-378. FI: 4,603(Q1)
- Ciruela Navas P, Izquierdo C, Broner S, Munoz-Almagro C, Hernández-Bou S, Ardanuy C, et al. The changing epidemiology of invasive pneumococcal disease after PCV13 vaccination in a country with intermediate vaccination coverage. Vaccine 2018. 36(50): 7744-7752. FI: 3,285(Q2)

Networks

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

INFECCIONES DE TRANSMISIÓN VERTICAL E INFECCIONES EN PACIENTES VULNERABLES

Clàudia Fortuny Guasch, PhD, MD

3

VERTICAL AREAS

3.4.

Infectious diseases and systemic inflammatory response in paediatrics

Coordinator:
Carmen Muñoz-Almagro

This year's highlights

In 2018, we began the study financed by the FIS grant in relation to the mitochondrial toxicity of the drugs used in the treatment of congenital CMV infection. The recruitment was slower than expected but by the end of the year we had included 8 patients. Despite the slow and laborious recruitment process, we believe that we now have a significant number of included patients and this should allow us to obtain some very interesting results. Additionally, we continued with the CCTs with antivirals for treatment of HCV and HIV infection. We also continued to cooperate with other groups on the study of patients with HIV infection, in relation to its treatment, effects, natural history, co-infections, transfer to adult units, etc.

Members

Staff researcher

Vicky Fumadó, Silvia Simó Nebot

Postdoc students

Anna Gamell

PhD students

Ferran Bossacoma

Collaborator researcher

Emilia Sanchez



PERSONAL RESEARCHER



PROJECTS



CLINICAL TRIALS & OBSERVATIONAL STUDIES



PUBLICATIONS WITH IF



FIRST QUARTILE PUBLICATIONS

Selected projects

- Study of mitochondrial toxicity in newborns and infants with congenital cytomegalovirus infection treated with ganciclovir or valganciclovir. FIS 2017. Instituto de Salud Carlos III (ISCIII). PI: Clàudia Fortuny Guasch. 01/01/2018-31/12/2020
- Longitudinal study of mitochondrial DNA mutations in children exposed to HIV (infected and non-infected) and to antiretrovirals. FIS 2013. Instituto de Salud Carlos III (ISCIII). PI: Clàudia Fortuny Guasch. 01/01/2014-30/06/2018

Selected clinical trials & observational studies

- 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. Codi EUDRACT: 2016-004102-34. Promotor: AbbVie Deutschland GmbH & Co. KG "AbbiVie". PI: Clàudia Fortuny Guasch. 03/11/2017-
- Study of the safety, tolerability and pharmacokinetics of two formulations of posaconazole (POS) given intravenously (IV) and in oral suspension powder in immunodepressed paediatric patients with neutropenia. Codi EUDRACT: 2014-002807-10. Promotor: Merck, Sharp & Dohme de España. S.A. PI: Clàudia Fortuny Guasch. 07/07/2015-
- 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 to C. difficile toxin B) in children and adolescents from 1 to under 18 years of age receiving antibacterial treatment for C. difficile infection (MODIFY III). Codi EUDRACT: 2017-000070-11. Promotor: Merck, Sharp & Dohme de España. S.A. PI: Clàudia Fortuny Guasch. 09/10/2017-

Selected publications

- Collaborative Initiative for Paediatric HIV Education and Research (CIPHER) Glob, Slogrove AL, Schomaker M, Davies MA, Williams P, Balkan S, et al. The epidemiology of adolescents living with perinatally acquired HIV: A cross-region global cohort analysis. PLoS Med 2018. 15(3): e1002514. FI: 11,675(Q1)
- Richarz NA, Fustà-Novell X, Fatsini-Blanch V, Fortuny-Guasch C, González-Enseñat MA and Vicente-Villa MA. Lichen striatus following scarlet fever in a 3-year-old female patient. Int J Dermatol 2018. 57(9): 1118-1119. FI: 1,541(Q3)
- Soriano-Arandes A, Noguera-Julían A, López-Lacort M, Soler-Palacín P, Mur A, Méndez M, et al. Pregnancy as an opportunity to diagnose human-immunodeficiency virus immigrant women in Catalonia. Enferm Infecc Microbiol Clin 2018. 36(1): 9-15. FI: 1,707(Q3)
- Velasco-Arnaiz E, Pérez E, Henares-Bonilla D, Fernández-López A, Valls-Lafon A, Brotons-de los Reyes P, et al. Use of procalcitonin in the diagnosis of tuberculosis in infants and preschool children. Eur J Pediatr 2018. 177(9): 1377-1381. FI: 2,242(Q2)
- Velasco-Arnaiz E, Soriano-Arandes A, Latorre-Rueda I, Altet N, Domínguez J, Fortuny-Guasch C, et al. Performance of Tuberculin Skin Tests and Interferon-gamma Release Assays in Children Younger Than 5 Years. Pediatr Infect Dis J 2018. 37(12): 1235-1241. FI: 2,305(Q2)

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

TUBERCULOSIS

Antoni Noguera Julián, PhD, MD

This year's highlights

Recruitment for the studies of isoniazid pharmacokinetics in newborns and small children continued in 2018. This recruitment is a slow and laborious process but we hope that we now have a significant number of included patients, which should allow us to obtain some very interesting results.

On the other hand, the studies are continuing on the diagnosis of tuberculosis infection in at-risk paediatric populations and on the role that may be played in this connection by the new tests for detection of *Mycobacterium tuberculosis*-specific gamma interferon. Lastly, the first doctoral thesis arising from the research line, on adherence to tuberculosis treatment in children and adolescents (Dr Eva Maria Guix Comellas), was successfully defended in 2018.

Members

PhD students

Maria Goretti López Ramos, Eneritz Velasco, Joan Vinent Genestar

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VERTICAL AREAS

3.4.

Infectious diseases and systemic inflammatory response in paediatrics

Coordinator:
Carmen Muñoz-Almagro



Selected projects

- Validation of the single daily dose of 10 mg isoniazid/kg of weight in newborns under 3 months of age. Instituto de Salud Carlos III (ISCIII). FIS 2013. PI: Antoni Noguera Julián. 01/01/2014-30/06/2018
- Phase IIA open clinical trial on absorption of a suspension of 10 mg isoniazid/ml for treatment of tuberculosis infection in patients under 6 years of age. Instituto de Salud Carlos III (ISCIII). PICI 2014. PI: Antoni Noguera Julián. 01/01/2015-30/06/2019
- Intensification of the research activity of IRSJD-HSJD. Hospital Sant Joan de Déu - Esplugues HSJD. AJUT INTENSIFICACIÓ 2016. PI: Antoni Noguera Julián. 05/03/2018-31/12/2018

Selected clinical trials & observational studies

- FSJD-ISON-2016. Phase IIA open clinical trial on absorption of a suspension of 10 mg isoniazid/ml for treatment of tuberculosis infection in patients under 6 years of age. Codi EUDRACT: 2016-002000-31. Fundació Privada per a la Recerca i la Docència Sant Joan de Déu - FSJD FSJD. PI: Antoni Noguera Julián. 10/10/2016-
- FUN-ISO-2014-01 Validation of the single daily dose of 10 mg isoniazid/kg of weight in newborns under 3 months of age. Fundació Privada per a la Recerca i la Docència Sant Joan de Déu - FSJD FSJD. PI: Antoni Noguera Julián. 16/04/1014-

Selected publications

- Guix-Comellas EM, Rozas L, Velasco-Arnaiz E, Ferrés-Canals A, Estrada-Masllorens JM, Force-Sanmartín E and Noguera-Julián A. Impact of nursing interventions on adherence to treatment with antituberculosis drugs in children and young people: A nonrandomized controlled trial. *J Adv Nurs* 2018. 74(8): 1819-1830. FI: 2,267(Q1)
- Judd A, Chappell E, Turkova A, Le Coeur S, Noguera-Julián A, Goetghebuer T, et al. Long-term trends in mortality and AIDS-defining events after combination ART initiation among children and adolescents with perinatal HIV infection in 17 middle- and high-income countries in Europe and Thailand: A cohort study. *PLoS Med* 2018. 15(1): e1002491. FI: 11,675(Q1)
- Velasco-Arnaiz E, Pérez E, Henares-Bonilla D, Fernández-López A, Valls-Lafon A, Brotons-de los Reyes P, et al. Use of procalcitonin in the diagnosis of tuberculosis in infants and preschool children. *Eur J Pediatr* 2018. 177(9): 1377-1381. FI: 2,242(Q2)
- Velasco-Arnaiz E, Soriano-Arandes A, Latorre-Rueda I, Altet N, Domínguez J, Fortuny-Guasch C, et al. Performance of Tuberculin Skin Tests and Interferon-gamma Release Assays in Children Younger Than 5 Years. *Pediatr Infect Dis J* 2018. 37(12): 1235-1241. FI: 2,305(Q2)

Networks

- 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 &
Laia Alsina Manrique, MD, PhD

This year's highlights

Our group is the leader in Spain in the study of immunodeficiencies secondary to the use of anti-TNFα monoclonal drugs during pregnancy, and in the study of primary immunodeficiencies associated with intracellular infections (such as mycobacteria and leishmania). Likewise, a research line has been started in the growing field of primary immunodeficiencies with predominant component of immune deregulation (inflammation, lymphoproliferative disorders, autoimmunity), with particular interest in the study of cells with a regulatory function (regulatory T and B lymphocytes) and in the use of sirolimus/rapamycin in their treatment. Our group has taken part in key clinical trials on the handling of autoinflammatory disorders, juvenile idiopathic arthritis and systemic lupus erythematosus, and on the analysis of safety profiles with biological drugs. We are also a national reference centre in the study of such disorders as juvenile dermatomyositis, juvenile idiopathic arthritis-associated uveitis, and Kawasaki disease.

Members

PhD students

Rosa Bou i Torrent, Àngela Deyà Martínez, Estibaliz Iglesias Jiménez, Mónica Piquer Gibert, Ana Pilar Garcia Garcia, Judith Sánchez Manubens

Post-doc researcher

Ana Esteve Solé

Assistant researcher

Violeta Bittermann, Clara Isabel Giménez Roca

Collaborator researcher

Alexandru Vlăgea, Anna Mensa Vilaró, Juan Ignacio Aróstegui Gorospe, Manel Juan Otero

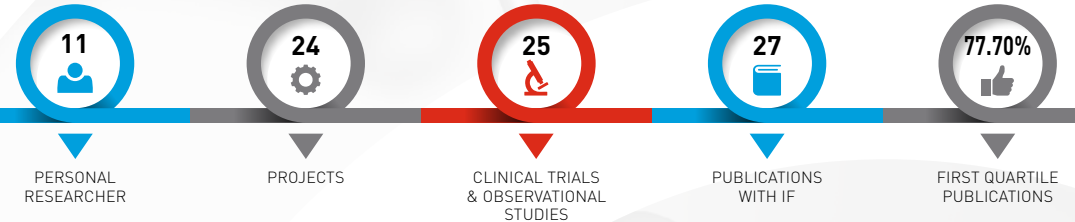
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VERTICAL AREAS

3.4.

Infectious diseases and systemic inflammatory response in paediatrics

Coordinator:
Carmen Muñoz-Almagro



Selected projects

- WINDBIOME_Aerial micro-biome diversity and their role in Kawasaki disease. Fundació Privada Daniel Bravo Andreu. Projectes Daniel Bravo 2015. PI: Jordi Antón López. 15/12/2015-14/12/2018
- ImmunAID_Immune project consortium for Autoinflammatory Disorders. European Commission. H2020-SC1-PM-03-2017. PI: Jordi Antón López. 01/04/2018-31/03/2023.
- Modulating factors of primary immunodeficiency with susceptibility to severe intracellular pathogen infections in paediatric age. Instituto de Salud Carlos III (ISCIII). FIS 2015. PI: Laia Alsina Manrique de Lara. 01/01/2016-31/12/2019
- MDRTBNet_Host-pathogen interactions in MultidrugResistant Tuberculosis Study of the Molecular Epidemiology, Host Immune Responses and GenomeBased Prediction and Early Identification of MDR-TB in High Tuberculosis Burden Settings. Instituto de Salud Carlos III (ISCIII). Acciones Complementarias 2016. 01/01/2017-31/12/2019

Selected clinical trials & observational studies

- GTI1503. Phase III. Multi-centre open trial with a single group to assess the effectiveness, pharmacokinetics, safety and tolerability of IGSC, 20% solution, in subjects with primary immunodeficiency. Instituto Grifols, S.A. Codi EUDRACT: 2015-003290-15. PI: Laia Alsina Manrique de Lara. 05/05/2016-
- NI-0501-06. Open, multi-centre pilot study with a single group to assess the safety, tolerability, pharmacokinetics and effectiveness of intravenous administration of NI-0501, an anti-interferon gamma (anti-IFNγ) monoclonal antibody, in patients with systemic-onset juvenile idiopathic arthritis (SoJIA) who develop

macrophage activation syndrome/secondary HLH (MAS/HLHs). Novimmune SA. Codi EUDRACT: 2016-004223-23. PI: Jordi Antón López. 06/11/2017-

Selected publications

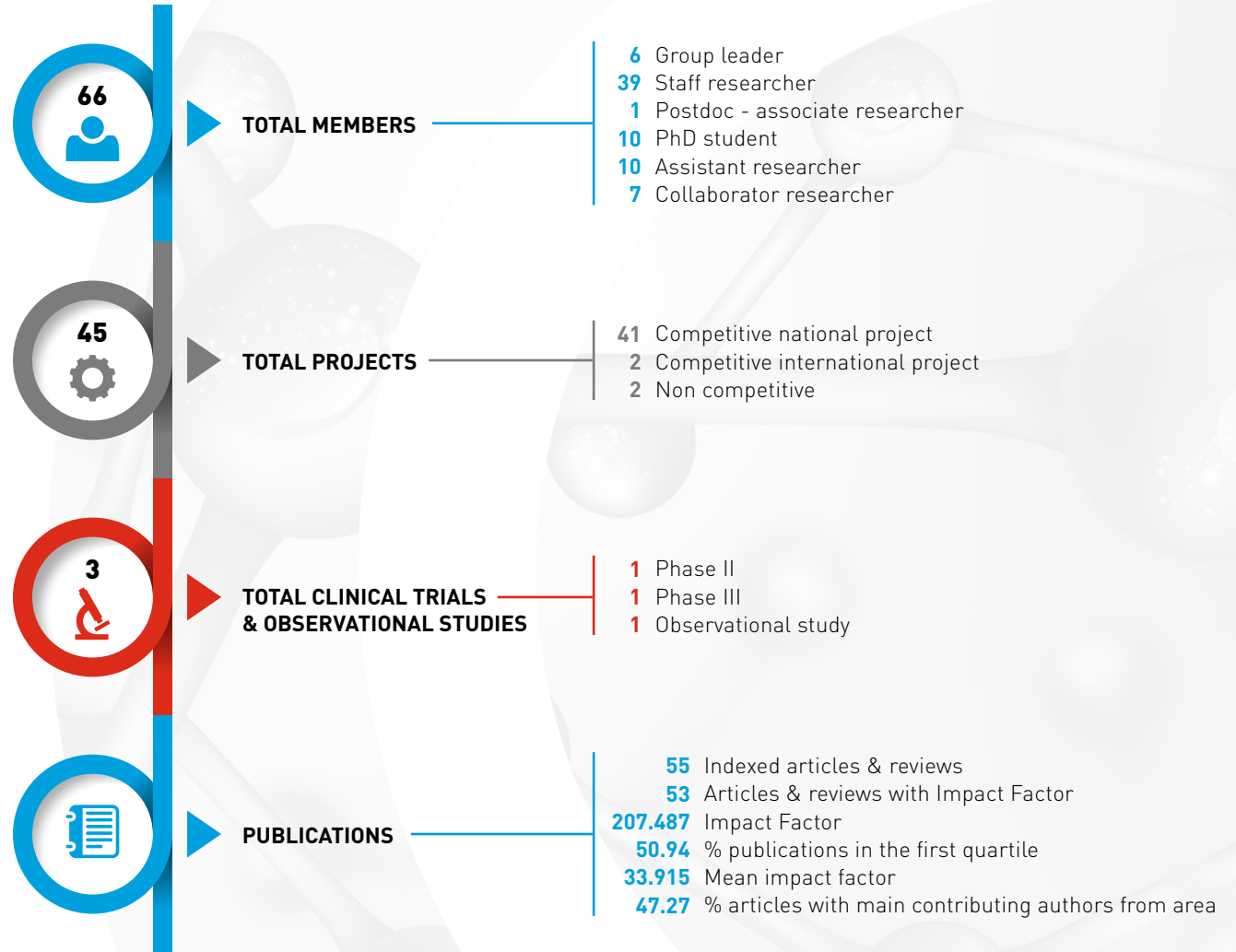
- De Benedetti F, Gattorno M, Anton-Lopez J, Ben-Chetrit E, Frenkel J, Hoffman HM, et al. Canakinumab for the Treatment of Autoinflammatory Recurrent Fever Syndromes. *N Engl J Med* 2018. 378(20): 1908-1919. FI: 79,258(Q1)
- de Inocencio J, Anton-Lopez J, Calvo Penades I, Mesa Del Castillo Bermejo P, Alcobendas R, Boteanu AL, et al. The Castilian Spanish version of the Juvenile Arthritis Multidimensional Assessment Report (JAMAR). *Rheumatol Int* 2018. 38(Suppl 1): 91-98. FI: 1,952(Q4)
- Deyà-Martinez A, Esteve-Sole A, Vélez-Tirado N, Celis-Passini V, Costa Escolà J, Cols M, et al. Sirolimus as an alternative treatment in patients with granulomatous-lymphocytic lung disease and humoral immunodeficiency with impaired regulatory T cells. *Pediatr Allergy Immunol* 2018. 29(4): 425-432. FI: 4,137(D1)
- Esteve-Sole A, Sologuren I, Martínez-Saavedra MT, Deyà-Martinez A, Oleaga-Quintas C, Martínez-Barricarte R, et al. Laboratory evaluation of the IFN-gamma circuit for the molecular diagnosis of Mendelian susceptibility to mycobacterial disease. *Crit Rev Clin Lab Sci* 2018. 55(3): 184-204. FI: 6,481(D1)

Networks

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

3.5. Mental Health

Coordinators: Susana Ochoa Güerre, MD, PhD and Judith Usall Rodié, MD, PhD



CLINICAL, COGNITIVE AND PSYCHOSOCIAL BASES OF SEVERE MENTAL DISORDERS

Susana Ochoa Güerre, PhD, MD

This year's highlights

In the follow-up study of patients with a first psychotic episode, we have continued to work in the FIS line on traumatic and protective factors granted in 2016. In this year we obtained a PFIS grant linked to the project. A FIS grant has been obtained to assess the effect of attachment on metacognition and social cognition in patients with schizophrenia, patients with first psychotic episodes and healthy controls.

Work was continued on the validation of instruments for patients with psychosis. In the field of prison mental health, this year the first thesis was presented at the PSSJD on this subject.

A series of papers was published in connection with the POMONA study in the sphere of intellectual disability and mental health.

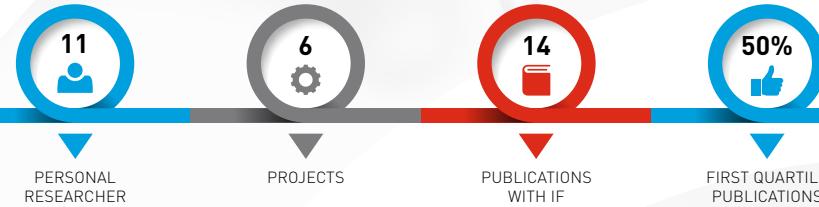
Members

Staff researcher

Hilari Andres Mora, Emma Casas Anguera, Jorge Cuevas Esteban, Gemma Escuder Romeva, Alexandrina Foix Sanjuan, Marcia Irazabal Jiménez, Maria Trinidad Peláez Martínez, Regina Vila-Badía

Assistant researcher

Anna Butjosa Molines, Núria del Cacho Ortega



Selected projects

- Social cognition and cognitive biases in patients with first psychotic episodes in comparison to chronic schizophrenia and healthy controls. Projectes Recerca Orientats Salut Mental. Departament de Salut - Generalitat de Catalunya. PI: Susana Ochoa Güerre. 17/02/2018-31/12/2020
- Traumatic experiences and protective factors in patients with a first psychotic episode and healthy controls. Ajuts recerca PSSJD. Parc Sanitari de Sant Joan de Déu - Serveis de Salut Mental. PI: Anna Butjosa Molines. 01/02/2018-31/01/2019
- Effectiveness of Individualised Metacognitive Training (MCT+) in patients with recent-onset psychotic disorder. Instituto de Salud Carlos III (ISCIII). PI: Susana Ochoa Güerre. 01/01/2015-31/12/2018

Selected publications

- Amoretti S, Cabrera Llorca B, Torrent C, Mezquida G, Lobo A, González-Pinto A, et al. Cognitive reserve as an outcome predictor: first-episode affective versus non-affective psychosis. Acta Psychiatr Scand 2018. 138(5): 441-455. FI: 4,984(Q1)

- Farreny A, Usall J, Cuevas-Esteban J, Ochoa S and Brebion G. Amendment of traditional assessment measures for the negative symptoms of schizophrenia. Eur Psychiatry 2018. 49: 50-55. FI: 4,129(Q1)
- Paipa N, Stephan-Otto C, Cuevas-Esteban J, Núñez Navarro A, Usall J and Brebion G. Second-to-fourth digit length ratio is associated with negative and affective symptoms in schizophrenia patients. Schizophr Res 2018. 199: 297-303. FI: 3,958(Q1)
- Sevilla-Llewellyn-Jones J, Cano-Domínguez P, de-Luis-Matilla A, Espina-Eizaguirre A, Moreno-Küstner B and Ochoa S. Personality traits in recent-onset-of-psychosis patients compared to a control sample by gender. Schizophr Res 2018. 195: 86-92. FI: 3,958(Q1)
- González LE, López-Carrilero R, Barrigón ML, Grasa E, Barajas A, Pousa Tomas E, et al. Neuropsychological functioning and jumping to conclusions in recent onset psychosis patients. Schizophr Res 2018. 195: 366-371. FI: 3,958(Q1)

Networks

- CIBERSAM
- Consolidated research group in AGAUR

3 VERTICAL AREAS

3.5. Mental Health

Coordinators:
Susana Ochoa Güerre
Judith Usall Rodié

MENTAL HEALTH INTERVENTIONS

Judith Usall Rodié, MD, PhD

This year's highlights

A new paper was published on the EU m-Resist project.

The FIS Individual Metacognitive Training project on patients with a first psychotic episode was closed in 2018 and a paper on the results is being prepared for publication. We have continued to recruit patients in the SCORES study of psychological intervention to improve emotional recognition in patients with postmenopausal schizophrenia.

A new paper has been published on the results of biomarkers in the clinical trial of treatment with raloxifene in patients with schizophrenia.

Members

Staff researchers

Belen Arraz Martí, Adela Berrozpe Blasco, Marta Coromina Sadurní, Ruth Cunill Clotet, Gemma Escartin Martín, Mar García Franco, Gabriel Ledesma Iparraguirre, Eva Miquel Fernandez, Elena Rubio Abadal, Luis San Molina, Sonia Vilamala Anton

PhD students

Marta Ferrer Quintero, Silvia Marcó Garcia

Assistant researcher

Raquel López Carrilero, Elia Vila Andreu

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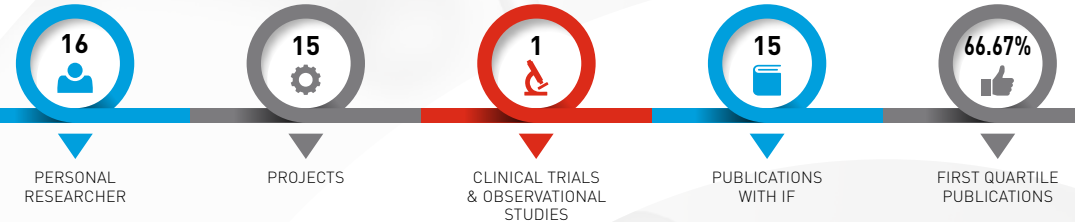
VERTICAL AREAS

3.5.

Mental Health

Coordinators:

Susana Ochoa Güerre
Judith Usall Rodié



Selected projects

- Traumatic experiences and protective factors in patients with a first psychotic episode and healthy controls. Ajutsrececera PSSJD. Parc Sanitari de Sant Joan de Déu - Serveis de Salut Mental. PI: Anna Butjosa Molines. 01/02/2018-31/01/2019
- Intestinal permeability as a cause of low-degree chronic inflammation in patients with schizophrenia: relation to diet, metabolic syndrome and severity of the illness. Proyectos de investigación. Instituto de Salud Carlos III (ISCIII). PI: Belen Arranz Martí. 01/01/2018-31/12/2020
- Legal incapacitation in patients with severe mental disturbance: description of the clinical and neuropsychological profile, and development of a predictive model. OSSJD - Obra Social Sant Joan de Déu. PI: Elena Huerta Ramos. 01/01/2018-31/12/2018
- m-RESIST_Mobile Therapeutic Attention for Patients with Treatment Resistant Schizophrenia. H2020-PHC-2014-single-stage. EuropeanCommission. PI: Judith Usall Rodie. 01/01/2015-31/01/2018

Selected clinical trials & observational studies

- Multicentre observational study of the cost-effectiveness of long-acting antipsychotics versus oral antipsychotics in patients with schizophrenia. Promotor: Cluster de Salut Mental Catalunya. PI: Luis San Molina. 23/11/2016-

Selected publications

- Ballesteros A, Sánchez-Torres AM, López-Ilundain JM, Cabrera B, Lobo A, González-Pinto AM, et al. Is cognitive impairment associated with antipsychotic dose and anticholinergic equivalent loads in first-episode psychosis?. Psychol Med 2018. 48(13): 2247-2256. FI: 5,475(Q1)
- Soria V, González-Rodríguez A, Huerta-Ramos ME, Usall J, Cobo J, Bioque M, et al. Targeting hypothalamic-pituitary-adrenal axis hormones and sex steroids for improving cognition in major mood disorders and schizophrenia: a systematic review and narrative synthesis. Psychoneuroendocrinology 2018. 93: 8-19. FI: 4,731(Q1)
- Paipa N, Stephan-Otto C, Cuevas-Esteban J, Núñez Navarro A, Usall J and Brebion G. Second-to-fourth digit length ratio is associated with negative and affective symptoms in schizophrenia patients. Schizophr Res 2018. 199: 297-303. FI: 3,958(Q1)
- González LE, López-Carrilero R, Barrigón ML, Grasa E, Barajas A, Pousa Tomas E, et al. Neuropsychological functioning and jumping to conclusions in recent onset psychosis patients. Schizophr Res 2018. 195: 366-371. FI: 3,958(Q1)

Networks

- CIBERSAM

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

Gildas Brebion, PhD

3

VERTICAL AREAS

3.5.

Mental Health

Coordinators:

Susana Ochoa Güerre
Judith Usall Rodié

This year's highlights

We have continued with the analyses of the data derived from earlier projects and we have written the respective articles. We have also continued to recruit patients with schizophrenia and depression for our study of auditory verbal hallucination, financed by ISCIII (file PI14/00047). Likewise, inasmuch as we have a sample of 48 healthy participants, we have written a preliminary article on the relation between the incapacity to monitor inner speech and the propensity for non-clinical auditory verbal hallucination. We have also analysed the data corresponding to the effects of depression and anxiety on verbal memory in the healthy sample and we have begun to report the results in an article.

Members

Staff researcher

Christian Núñez Leánez,
Christian Stephan-Otto Attolini



PERSONAL
RESEARCHER



PROJECTS



PUBLICATIONS
WITH IF



FIRST QUARTILE
PUBLICATIONS

Selected projects

- SGR 2017_Cerebral imprints of gender, development and pathology. Agaur - Agència de Gestió d'Ajuts Universitaris i de Recerca. SGR 2017-2019. PI: Christian Stephan-Otto Attolini. 01/01/2017-31/12/2020
- Neurofunctional bases of anomalous inner speech in auditory verbal hallucinations. Instituto de Salud Carlos III (ISCIII). FIS 2014. PI: Gildas Brebion. 01/01/2015-30/06/2019
- Miguel Servet tipo II - Beneficiario: Gildas Brébion. Instituto de Salud Carlos III (ISCIII). Miguel Servet II 2015. PI: Gildas Brebion. 15/01/2016-14/01/2019

Selected publications

- Nuñez C, Theofanopoulou C, Senior C, Cambra-Martí MR, Usall J, Stephan-Otto C and Brebion G. A large-scale study on the effects of sex on gray matter asymmetry. *BRAIN STRUCT FUNCT* 2018. 223(1): 183-193. FI: 4,231(Q1)
- Farreny A, Usall J, Cuevas-Esteban J, Ochoa S and Brebion G. Amendment of traditional assessment measures for the negative symptoms of schizophrenia. *Eur Psychiatry* 2018. 49: 50-55. FI: 4,129(Q1)
- Paipa N, Stephan-Otto C, Cuevas-Esteban J, Araceli Núñez Navarro, Usall J and Brebion G. Second-to-fourth digit length ratio is associated with negative and affective symptoms in schizophrenia patients. *Schizophr Res* 2018. 199: 297-303. FI: 3,958(Q1)
- Brebion G, Stephan-Otto C, Ochoa S, María de Lourdes Nieto García, Contel M and Usall J. Verbal fluency in male and female schizophrenia patients: Different patterns of association with processing speed, working memory span, and clinical symptoms. *Neuropsychology* 2018. 32(1): 65-76. FI: 2,699(Q2)
- Siddi S, Ochoa S, Farreny A, Brebion G, Larøi F, Cuevas-Esteban J, et al. Measurement invariance of the Spanish Launay-Slade Hallucinations Scale-Extended version between putatively healthy controls and people diagnosed with a mental disorder. *Int J Methods Psychiatr Res* 2018. 27(4): e1741. FI: 2,500(Q2)

CHILD AND YOUTH MENTAL HEALTH RESEARCH

Montserrat Dolz Abadia, MD, PhD

This year's highlights

The "Mental disturbances in childhood and adolescence" group (CIBERSAM) has become consolidated in recent years, both in terms of research collaborations established on various levels, and in terms of project financing. On the one hand, the group cooperates in a stable manner with other groups of the CIBERSAM mental health biomedical research network, such as group G04 of the Hospital Clínic i Provincial de Barcelona (psychosis project). Additionally, a national networked research cooperation project led by the Autonomous University of Madrid has been started (autism project). It should be pointed out that, over the course of the year, we have consolidated our cooperation with international centres (MIND Institute, ISI-Global, MNC Melbourne). Likewise, public projects (FIS, MINECO projects) and European projects (European Commission) as well as private projects have been financed. Notable achievements in the past year have included the grant of two projects with participation of competitive funds: PERIS, Dr Sintes, with the collaboration of the GRETA group (Group for Study and Treatment of Self-Injury) in the study of self-harming behaviour; and Dr Alda in the study of ADHD and ASD and microbiota. Also to be noted was the award of a mobility grant to Dr Dolz by the Carlos III Health Institute. The group (PI: Dr Camprodon) has recently become a collaborating centre in a European Horizon 2020 project (ERC, reference: No 785994) which is conducted at ISGlobal Barcelona (PI: Dr Sunyer), studying the effects of prenatal exposure to urban air pollution and pre- and postnatal cerebral development. We also participated in two clinical trials: one is a European test (reference: 2015-003503-39) addressed to assessing the effectiveness of omega-3 treatment in patients with risk of developing psychosis, and the other is a trial of worldwide scope, with private financing and using drugs, that deals with depression with suicidal ideation (ESKETINSUI2002). Likewise, the research group has been involved in R&D projects in collaboration with the universities URL, UOC and UB (PI: Dr Serrano; PI: Dr Matalí). Lastly, at national level, we have received a number of prizes for different research studies (for example, the AEPNyA award).

Members

Staff researcher

José Ángel Alda Díez, Mar Álvarez Segura, Aritz Arranbarri Paredes, Ester Camprodon Rosanas, Maria Diez Juan, Neus Elias Carbonell, Fernando Lacasa Saludas, Maria Ángeles Mairena García de la Torre, Josep Lluís Matalí Costa, Daniel Muñoz Samons, Marta Pardo Gallego, Bernardo Sánchez Fernández, Eduardo Serrano Troncoso, Anna Sintes Estevez, Esther Vía Virgili, Francisco Villar Cabeza

PhD students

Claudia Caprile Elola-Olaso, Anna Huguet Miguel, Jordina Tor Fabra

Assistant researchers

Carlota Alcover Van de Walle, Marina Fabrega Ribera, Sara Parrilla Carrasco, Carlota Saumell Andreu, Ingrid Tortadès Arumí, Ania Perez Racana

Collaborator researchers

Jordi Navarra Ordoño, Laura Puigcerver Navarro

3

VERTICAL AREAS

3.5.

Mental Health

Coordinators:

Susana Ochoa Güerre
Judith Usall Rodié



PERSONAL
RESEARCHER



PROJECTS



CLINICAL TRIALS
& OBSERVATIONAL
STUDIES



PUBLICATIONS
WITH IF



FIRST QUARTILE
PUBLICATIONS

Selected projects

- Mindfulness mediante realidad virtual en niños recientemente diagnosticado de TDAH versus programa de mindfulness estándar (estudio aleatorizado). Premio AEPNyA de Investigación. Asociación Española de Psiquiatría del Niño y el Adolescente. PI: Anna Huguet Miguel. 17/06/2017-16/06/2019
- Non-suicidal self-injury in Catalonia: prevalence, characteristics and psychopathological implications in adolescents and young adults. Projectes Recerca Orientats Salut Mental. Departament de Salut - Generalitat de Catalunya. PI: Anna Sintes Estevez. 17/02/2018-17/12/2020
- Multicentre longitudinal study on neurobiological markers of stress in children and adolescents with psychosis- and transition to psychosis-risk syndrome. FIS. Instituto de Salud Carlos III (ISCIII). PI: Montserrat Dolz Abadia. 01/01/2016-30/06/2020

Selected clinical trials & observational studies

- A Double-blind, Randomized, Psychoactive Placebo-controlled, Study to Evaluate the Efficacy and Safety of 3 Fixed Doses (28 mg, 56 mg and 84 mg) of Intranasal Esketamine in Addition to Comprehensive Standard of Care for the Rapid Reduction of the Symptoms of Major Depressive Disorder, Including Suicidal Ideation, in Pediatric Subjects Assessed to be at Imminent Risk for Suicide. Codi EUDRACT: 2016-004422-42. Promotor: Janssen - Cilag, S.A. PI: Montserrat Dolz Abadia. 27/11/2018-
- Placebo-controlled trial in subjects at high risk for psychosis compared 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

- Insa I, Alda JA, Chamorro M, Espadas M and Huguet A. Difference in Psychic Distress Lived by Parents With ADHD Children and Parents With Healthy Children: Focus on Gender Differences. J Atten Disord 2018. 1087054718790010. FI: 3,668(Q1)
- Tor J, Dolz M, Sintes A, Muñoz Samons D, Pardo M, de la Serna E, et al. Clinical high risk for psychosis in children and adolescents: a systematic review. Eur Child Adolesc Psychiatry 2018. 27(6): 683-700. FI: 3,553(Q1)NETWORKS
- Baeza I, Serna E, Calvo-Escalona R, Merchán-Naranjo J, Rodríguez-Latorre P, Martínez-Cantarero MC, et al. One-Year Prospective Study of Liver Function Tests in Children and Adolescents on Second-Generation Antipsychotics: Is There a Link with Metabolic Syndrome?. J Child Adolesc Psychopharmacol 2018. 28(7): 463-473. FI: 2,901(Q1)
- Quintero J, Ramos-Quiroga JA, Sebastián JS, Montañés F, Fernández-Jaén A, Martínez-Raga J, et al. Health care and societal costs of the management of children and adolescents with attention-deficit/hyperactivity disorder in Spain: a descriptive analysis. BMC Psychiatry 2018. 18(1): 40-40. FI: 2,419(Q2)
- Matalí JL, Simons J, Pardo M, Lleras M, Pérez A and Andiñón O. Spanish version validation of the Marijuana Motives Measure in a drug-consuming adolescent sample. Adicciones 2018. 30(4): 282-291. FI: 2,021(Q3)

Networks

- CIBERSAM

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

Juan Vicente Luciano Devis, PhD

This year's highlights

Last year the AGORA group obtained a predoctoral FI grant from AGAUR awarded to Laura Andrés Rodríguez (with Dr Luciano as thesis director). Dr Feliu, together with Dr Borràs, directed the master's degree dissertation of Natalia Angarita Osorio, presented at UAB. One of the most important aspects was the start-up of the ULTREYA project (2018-2020), which seeks to assess the short- and long-term effects of the pilgrimage to Santiago de Compostela on mental health and quality of life, and to determine which personal characteristics predict a bigger change in the collected health variables. This is a prospective observational study in which participants answer an online battery of questionnaires (on anxiety, depression, psychological well-being, etc.) on beginning the pilgrimage, on ending it and 3 months after ending it. This project includes the collaboration of researchers of PSSJD (Dr Luciano and Dr Feliu), Hospital de la Santa Creu i Sant Pau (Dr Soler), University of Zaragoza (Dr García Campayo) and Sao Paulo Federal University (Dr Demarzo). Project website: <http://estudiocamino.org/>

Members

Staff researcher

Antoni Rozadilla Sacanell, Lluís Torruella Barraquer

Post-doc researchers

Albert Feliu Soler

PhD students

Laura Andrés Rodríguez, Natalia Angarita Osorio, Adrián Pérez Aranda.

Collaborator researcher

Jaume guado Carné, Xavier Borràs Hernández, Eugenia Cardeñoso Valera



PERSONAL
RESEARCHER



PROJECTS



PUBLICATIONS
WITH IF



FIRST QUARTILE
PUBLICATIONS

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 2014. PI: Juan Vicente Luciano Devis. 01/01/2015-30/06/2019
- Neurobiological effects of a mindfulness-based intervention (MBSR) vs. a psycho-educational intervention (FibroQol) in patients with fibromyalgia (EUDAIMON project). Instituto de Salud Carlos III (ISCIII). FIS 2015. PI: Juan Vicente Luciano Devis. 01/01/2016-31/12/2019
- SGR 2017_Grupo de Investigación Psicológica en Fibromialgia y Dolor Crónico (Grupo ÁGORA). Agaur - Agència de Gestió d'Ajuts Universitaris i de Recerca. SGR 2017-2019. PI: Juan Vicente Luciano Devis. 01/01/2017-31/12/2020

Selected publications

- Feliu-Soler A, Cebolla A, McCracken LM, Knapp M, D'Amico F, López-Montoyo A, García-Campayo J, Soler J, Baños RM, Pérez-Aranda A, Andrés-Rodríguez L, Rubio-Valera M, & Luciano JV. Economic

- impact of 'third-wave' cognitive and behavioral therapies: A systematic review and quality assessment of economic evaluations in randomized controlled trials. *Behav Ther.* 2018; 49: 124-147. FI: 3,228(Q1)
- Cuesta-Vargas A, Neblett R, Chiarotto A, Kregel J, Nijs J, van Wilgen CP, Pitance L, Knezevic A, Gatchel RJ, Mayer TG, Viti C, Roldán-Jiménez C, Testa M, Caumo W, Jeremic-Knezevic M, & Luciano JV. Dimensionality and reliability of the Central Sensitization Inventory (CSI) in a pooled multi-country sample. *J Pain* 2018; 19: 317-329. FI: 4,859 (Q1)
- Cebolla A, Galiana L, Campos-Bacas D, Oliver A, Soler J, Demarzo M, Baños RM, Feliu-Soler A, García-Campayo J. How does mindfulness work? Exploring a theoretical model using samples of meditators and non-meditators. *Mindfulness.* 2018; 9: 860-870. FI: 3,024 (Q1)

Networks

- The following members of the AGORA group participate in the RedIAPP (ISCIII; RD16/0007/0012): Laura Andrés Rodríguez, Albert Feliu Soler, Juan Vicente Luciano Devis, Adrián Pérez Aranda

MOLECULAR PSYCHIATRY

Belen Ramos Josemaria, PhD

This year's highlights

- A FIS Health Research multi-centre project has been obtained to study gene expression regulators as biomarkers in psychoses related to cognitive functioning and neuroimage changes. Our group is the project coordinator.
- We have continued recruitment in the project associated with the Miguel Servet contract for the study of biomarkers in recent-onset psychosis and chronic schizophrenia.
- A study of large-scale quantitative proteomics in post-mortem brain tissue has been conducted in the project associated with the Miguel Servet contract for the study of biomarkers in recent-onset psychosis and chronic schizophrenia. Collaboration with Dr Villen's group at the University of Washington (Seattle, USA)
- A new paper has been published on the SP1 and SP4 transcription factors in relation to the improvement of symptoms in the clinical trial of treatment with raloxifene in patients with postmenopausal schizophrenia.
- A new paper has been published on 22 genes involved in inflammatory processes and immune response in post-mortem prefrontal dorsolateral cortex in geriatric patients with chronic schizophrenia in relation to microglia.
- A new doctoral student from the Neurosciences Programme of the Autonomous University of Barcelona (UAB) has been added to the group for the development of the project on detection of lipid drops in cells of the white series in psychosis as possible biomarkers. Collaboration with Dr Montserrat Torreadall (HSJD) and Dr Enrique Claro (UAB).

Members

PhD students

America Jennifer Vera Montecinos, Elia Vila Andreu, Francisco Vidal Domènech, Ania Eugui Anta

Collaborator researcher

Joan Costa Escolà, Alfonso Monje Hernández

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VERTICAL AREAS

3.5.

Mental Health

Coordinators:

Susana Ochoa Güerre
Judith Usall Rodié



PERSONAL
RESEARCHER



PROJECTS



PUBLICATIONS
WITH IF



FIRST QUARTILE
PUBLICATIONS

Selected projects

- Effectiveness of Individualized Metacognitive Training (MCT+) in patients with recent-onset psychotic disorder. FIS. Instituto de Salud Carlos III (ISCIII). PI: Susana Ochoa Güerre. Co-PI: Belén Josemaría Ramos. 01/01/2015-31/12/2018
- Molecular alterations linked to symptoms and cognitive functioning in chronic schizophrenia and brief psychosis: Biomarkers for predicting cognitive decline and symptom progression. Beneficiario: Dra. Belen Ramos. Miguel Servet I. Instituto de Salud Carlos III (ISCIII). PI: Belen Ramos Josemaria. 01/01/2017-31/12/2021
- Molecular alterations linked to symptoms and cognitive functioning in chronic schizophrenia and brief psychosis: Biomarkers for predicting cognitive decline and symptom progression. Miguel Servet I (Proyecto). Instituto de Salud Carlos III (ISCIII). PI: Belen Ramos Josemaria. 01/01/2017-31/12/2019

Selected publications

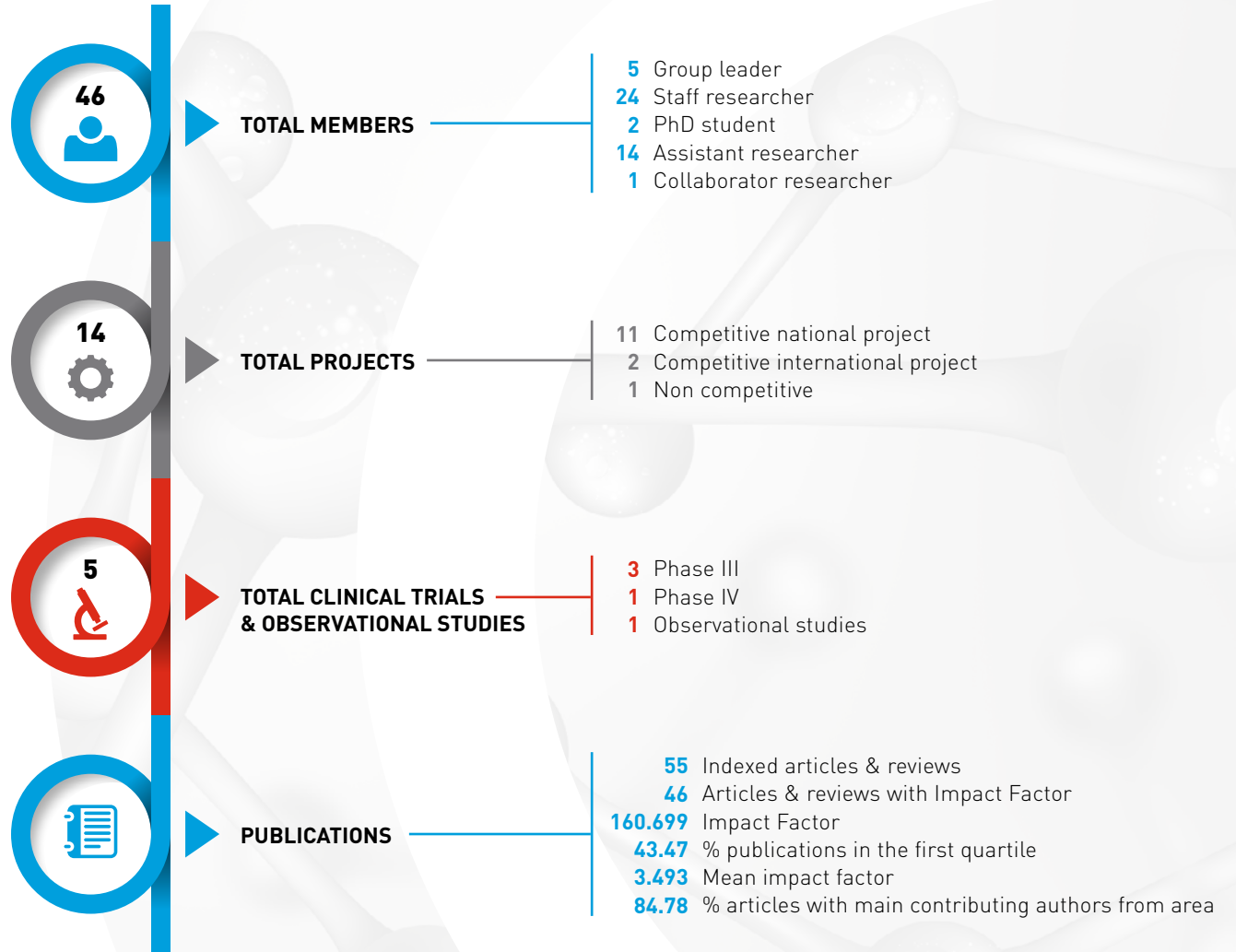
- Deyà-Martinez A, Esteve-Sole A, Vélez-Tirado N, Celis-Passini V, Costa Escolà J, Cols M, Jou-Munoz C, Vlasea A, Plaza-Martín AM, Juan M and Alsina-Manrique de Lara L. Sirolimus as an alternative treatment in patients with granulomatous-lymphocytic lung disease and humoral immunodeficiency with impaired regulatory T cells. *Pediatr Allergy Immunol* 2018. 29(4): 425-432. FI: 4,137(Q1)
- Lores L, Monje A, Bergada M, Arellano E, Rodriguez-Larrea J and Miravittles M. Prevalence of smoking in a psychiatric hospital and its relationship with respiratory symptoms and the prevalence of COPD. *Int J Chron Obstruct Pulmon Dis* 2018. 13: 2797-2804. FI: 2,917(Q2)

Networks

- (CIBERSAM) Grupo: CB/07/09/0028. Fecha de inicio: 01/06/2008
- 2017 SGR 964, Grup de Recerca en trastorns mentals greus Generalitat de Catalunya.
- AGAUR programa for recognized research groups. Judith Usall i Rodié. (FUNDACIO PER A LA RECERCA I LA DOCENCIA SANT JOAN DE DEU (FSJD)). 01/01/2018-31/12/2020. 35.200 €. Miembro de equipo.

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

Coordinator: Maria Dolores Gómez-Roig, MD, PhD



PAEDIATRIC INTENSIVE CARE UNIT RESEARCH PROJECT GROUP

Francisco José Cambra Lasasoa, MD, PhD

Members

Staff researchers

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

Assistant researcher

Carme Alejandre Galobardes, Sergio Benito Fernández, Patricia Corniero Alonso, Aida Felipe Villalobos, Elena Fresán Ruiz, Lluïsa Hernandez Platero, Nuria Millán García del Real, Susana Segura Matute, Marta Serradell Orea, David Vila Pérez

Collaborator researcher

Montserrat Esquerda Areste

3

VERTICAL AREAS

3.6.

Foetal / Paediatric Diseases and environment, metabolic and genetic factors

Coordinator:

María Dolores Gómez-Roig



Selected projects

- Analysis of a diagnostic algorithm for severe pneumonia in critically ill paediatric patients, using pulmonary ultrasound and procalcitonin, as a plan to improve healthcare quality. FIS 2016. Instituto de Salud Carlos III (ISCIII). Proyectos de investigación en salud. PI: Iolanda Jordán García. 01/01/2017-31/12/2019
- Predoctoral contracts for health research training. Beneficiari: Sara Bobillo. PFIS 2017. Instituto de Salud Carlos III (ISCIII). PFIS. PI: Iolanda Jordán García. 31/01/2018-29/01/2022
- Assessment of the neuroprotective effect of dexmedetomidine on a paediatric animal model of extracorporeal circulation and circulatory failure. BECA FRANCISCO RUZA 2018. Sociedad y Fundación Española de Cuidados Intensivos Pediátricos (SECIP). BECA FRANCISCO RUZA. PI: Sergio Benito Fernández. 01/04/2018-31/03/2020

Selected clinical trials & observational studies

- LJ501-CRH04. Randomised placebo-controlled double-blind multicentre study on the effectiveness and safety of LJPC-501 in paediatric patients aged > 2 to 17 years with catecholamine-resistant hypotension associated with distributive shock. Codi EUDRACT: 2017-005153-37. PI: Iolanda Jordán García. 06/11/2018-
- 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

- Bosch-Alcaraz A, Falcó Pegueroles A and Jordán-García I. A literature review of comfort in the paediatric critical care patient. J Clin Nurs 2018. 27(13-14): 2546-2557. FI: 1,635(Q2)
- Mayordomo-Colunga J, Pons-Odena M, Medina A, Rey C, Milesi C, Kallio M, et al. Non-invasive ventilation practices in children across Europe. Pediatr Pulmonol 2018. 53(8): 1107-1114. FI: 3,157(Q1)
- Oulego-Erroz I, González-Cortes R, García-Soler P, Balaguer-Gargallo M, Frías-Pérez M, Mayordomo-Colunga J, et al. Ultrasound-guided or landmark techniques for central venous catheter placement in critically ill children. Intensive Care Med 2018. 44(1): 61-72. FI: 15,008(Q1)
- Solé A, Jordán-García I, Bobillo-Perez S, Moreno-Hernando J, Balaguer-Gargallo M, Hernandez-Platero L, et al. Venoarterial extracorporeal membrane oxygenation support for neonatal and pediatric refractory septic shock: more than 15 years of learning. Eur J Pediatr 2018. 177(8): 1191-1200. FI: 2,242(Q2)

Networks

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

INFLUENCE OF THE ENVIRONMENT ON CHILD AND ADOLESCENT WELFARE

Carles Luaces Cubells, MD, PhD

This year's highlights

- *Contribution to the drafting of the Protocol of action in response to childhood/adolescence abuse in the health field (published by the Secretariat for Public Health. Health Department. Barcelona, March 2019). http://salutpublica.gencat.cat/content/minisite/aspcat/promocio_salut/infancia-i-adolescencia/05maltractamentinfantojuvenil/protocol-actuacio-maltractaments-infancia.pdf, forming part of the Working Group for the Revision of the Protocol for Childhood/Adolescence Abuse, and of the Subcommittee on the Healthcare and Judicial Communication Report.*
- *Drafting and updating of the hospital's new plan for catastrophes and multiple-victim incidents.*

Members

Staff researchers

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

Assistant researcher

David Muñoz Santanach



Selected publications

- Casas-Alba D, Martínez-Monseny T, Pino-Ramírez RM, Alsina-Manrique de Lara L, Castejón Ponce E, Navarro-Vilarrubi S, Pérez-Dueñas B, Serrano M, Palau-Martínez F and García-Alix A. Hyaline fibromatosis syndrome: Clinical update and phenotype-genotype correlations. *Hum Mutat* 2018. 39(12): 1752-1763. FI: 5,359(Q1)
- Cristina Parra Cotanda, Mele M, Alonso I, Trenchs-Sainz de la Maza V and Luaces-Cubells C. Parent experience in the resuscitation room: how do they feel?. *Eur J Pediatr* 2018. 177(12): 1859-1862. FI: 2,242(Q2)
- Pinedo-Painous I, Garrido-Romero R, Valls-Lafon A, Muñoz-Santanach D and Martínez-Sánchez L. Cannabis poisoning under the age of 3 years. *Emergencias* 2018. 30(6): 408-411. FI: 3,608(Q1)

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), 2012
- Red de investigación y desarrollo de la emergencia pediátrica latinoamericana (RIDEPLA)

3

VERTICAL AREAS

3.6.

Foetal / Paediatric Diseases and environment, metabolic and genetic factors

Coordinator:
María Dolores Gómez-Roig

FOETAL ENVIRONMENT AND OBSTETRICAL COMPLICATION

Maria Dolores Gómez Roig, MD, PhD

This year's highlights

Our goal is to characterise the effect of exposure during pregnancy to all these substances in order to expand our knowledge about them and to be able to predict pregnancy complications relating to environmental factors such as growth restriction.

In 2018:

- (1) *We published an article evidencing the serious problem of alcohol consumption during pregnancy. During the course of this study, the presence of EtG (an alcohol metabolite) was detected in the hair of a large number of pregnant women. For this reason, in 2018 we began an intervention study to reduce alcohol consumption in pregnant women.*
- (2) *We also began a study called BiSC (Barcelona Life Study Cohort), the purpose of which is to understand the effect of air pollution on the placental function, foetal growth, and cardiovascular and cerebral development of the foetus. This is a cooperative project of ISGlobal (Barcelona Institute for Global Health), BCNatal (Hospital Sant Joan de Déu and Hospital Clínic i Provincial de Barcelona) and the Hospital de la Santa Creu i Sant Pau. At present the group is in the process of recruiting 600 volunteers who live in Barcelona and who are in their first three months of pregnancy.*

Our group seeks to study the influence of the environment on obstetric complications and to decrease their effects in the prenatal and postnatal periods of newborns.

Members

Staff researchers

Sergio Cabré Gili, Silvia-Irena Ferrero Martínez, Miriam Pérez Cruz, Joan Sabrià Bach, María José Tojo Grandio, Eduarne Mazarico, Patricia Pagès

PhD students

Edda Marimon Garcia, Isabel Miró Sotelo

Assistant researcher

Laura Almeida Toledano, Erica Muñoz Sanz, Estela Rodríguez Quinteiro

Technician

Daniel Cuadras Palleja

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VERTICAL AREAS

3.6.

Foetal / Paediatric Diseases and environment, metabolic and genetic factors

Coordinator:

Maria Dolores Gómez-Roig



PERSONAL RESEARCHER



PROJECTS



CLINICAL TRIALS & OBSERVATIONAL STUDIES



PUBLICATIONS WITH IF



FIRST QUARTILE PUBLICATIONS

Selected projects

- Study on the conditions of use of Cariban® and other antiemetic drugs in the treatment of nausea and vomiting in pregnancy: longitudinal observational prospective study. Proyectos de investigación. Proyectos de investigación en salud. Instituto de Salud Carlos III (ISCIII). PI: Maria Dolores Gómez Roig. 01/01/2018-31/12/2020
- Traffic-related air pollution and birth weight: the roles of noise, placental function, green space, diet, physical activity, and socioeconomic status (FRONTIER). RFA 17-1: Assessing adverse health effects of exposure to traffic-related air pollution, noise, and their interactions with socio-economic status. Health Effects Institute (HEI). PFIS. PI: Maria Dolores Gómez Roig. 01/06/2018-31/02/2019

Selected clinical trials & observational studies

- Study on the conditions of use of Cariban® and other anti-emetics in the treatment of nausea and vomiting in pregnancy: prospective observational longitudinal study. Promotor: Laboratorios Inbisa, S.A. PI: Jose María Bogaña Ponsa. 02/06/2016-
- Treatment of early onset foetal growth restriction with low molecular weight heparin: randomised clinical trial (TRACIP). Codi EUDRACT: 2017-001905-33. Promoter: Fundació Privada per a la Recerca i la Docència Sant Joan de Déu - FSJD FSJD. PI: Eduarne Mazarico Gallego. 02/11/2017-

Selected publications

- Lin X, Torrabadella M, Amat Tardiu L, Gomez S, Azqueta C, Sánchez M, Cuadras-Palleja

- D, Martínez Lorenzo MJ, Brull JM, Gayà A, Cemborain A, Pérez Garcia C, Arroyo J, Querol S and Gómez-Roig MD. Estimated fetal weight percentile as a tool to predict collection of cord blood units with higher cellular content: implications for prenatal selection of cord blood donors. Transfusion 2018. 58(7): 1732-1738. FI: 3,423(Q2)
- Pérez-Cruz M, Crispi F, Fernández-Pérez MT, Parra-Hernandez JA, Valls-Lafon A, Gómez-Roig MD and Gratacós E. Cord Blood Biomarkers of Cardiac Dysfunction and Damage in Term Growth-Restricted Fetuses Classified by Severity Criteria. Fetal Diagn Ther 2018. 44(4): 271-276. FI: 1,813(Q3)
- Sabra S, Malmqvist E, Almeida-Toledano L, Gratacós E and Gómez-Roig MD. Differential correlations between maternal hair levels of tobacco and alcohol with fetal growth restriction clinical subtypes. Alcohol 2018. 70: 43-49. FI: 2,425(Q3)
- Gómez-Roig MD, Marchei E, Sabra S, Busardò FP, Mastrobattista L, Pichini S and Gratacós E. Maternal hair testing to disclose self-misreporting in drinking and smoking behaviour during pregnancy. Alcohol 2018. 67: 1-6. FI: 2,425(Q3)
- Mazarico-Gallego E, Llorba E, Cabero L, Sanchez O, Valls-Lafon A, Martín-Ancel A, Cardenas D and Gómez-Roig MD. Associations between neural injury markers of intrauterine growth-restricted infants and neurodevelopment at 2 years of age.. J Matern Fetal Neonatal Med 2018. 1-7. FI: 1,493(Q4)

Networks

- 2009 - Red SAMID-RETIC
- 2013 - BCNatal

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

Joan Sanchez de Toledo, MD, PhD

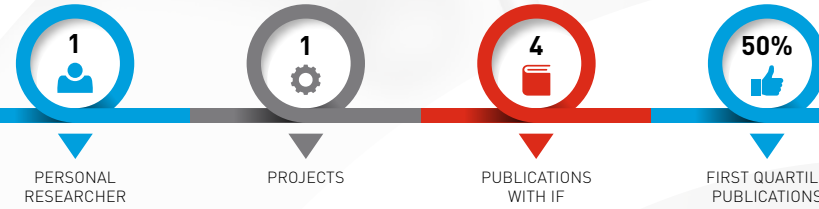
3

VERTICAL AREAS

3.6.

Foetal / Paediatric
Diseases and
environment, metabolic
and genetic factors

Coordinator:
María Dolores Gómez-Roig



Selected projects

- Usefulness of integrated neuromonitoring as a predictor of neurodevelopment in patients subjected to heart surgery in paediatric age: from the clinic to the paediatric animal model. Proyectos de investigación en salud. Instituto de Salud Carlos III (ISCIII). PI: Joan Sanchez de Toledo. 01/01/2018-31/12/2020

Selected publications

- Bautista Rodríguez C, Sanchez-de-Toledo J and Da Cruz EM. The Role of Echocardiography in Neonates and Pediatric Patients on Extracorporeal Membrane Oxygenation. *Front Pediatr* 2018. 6: 297-297. FI: 2,335(Q2)
- Del Rey Hurtado de Mendoza B, Sanchez-de-Toledo J and Rodríguez-Fanjul J. Chest ultrasound for guiding lung recruitment maneuvers. *Med Intensiva* 2018. 42(8): e20. FI: 1,755(Q4)
- Rodríguez-Fanjul J, Moreno-Hernando J and Sanchez-de-Toledo J. Lung Ultrasound for Cardiogenic Shock in VA-ECMO. *Rev Esp Cardiol* 2018. 71(5): 393-393. FI: 5,166(Q1)
- Torres-Andres F, Fink EL, Bell MJ, Sharma MS, Yablonsky EJ and Sanchez-de-Toledo J. Survival and Long-Term Functional Outcomes for Children With Cardiac Arrest Treated With Extracorporeal Cardiopulmonary Resuscitation. *Pediatr Crit Care Med* 2018. 19(5): 451-458. FI: 3,092(Q1)

PAEDIATRIC ARRHYTHMIAS, SUDDEN DEATH AND CARDIOVASCULAR GENETICS

Georgia Sarquella Brugada, MD, PhD

Members

Staff researchers

Esther Carro Fernandez, Sergio César Diaz,
Isaac Moll Adrian

3

VERTICAL AREAS

3.6.

Foetal / Paediatric
Diseases and
environment, metabolic
and genetic factors

Coordinator:

Maria Dolores Gómez-Roig



PERSONAL
RESEARCHER



PROJECTS



CLINICAL TRIALS
& OBSERVATIONAL
STUDIES



PUBLICATIONS
WITH IF



FIRST QUARTILE
PUBLICATIONS

Selected projects

- Phase-II predictors of arrhythmias and sudden death in paediatric patients with laminopathy. Fundación Andrés Marcio, niños contra la Laminopatía. PI: Georgia Sarquella Brugada. 01/02/2017-31/01/2020
- SGR 2017_ Paediatric arrhythmias, familial heart diseases, cardiovascular genetics and sudden death (SAMS). SGR 2017-2019. Agaur - Agència de Gestió d'Ajuts Universitaris i de Recerca. PI: Georgia Sarquella Brugada. 01/01/2017-31/12/2020
- GUARD-HEART: Gateway to Uncommon And Rare Diseases of the HEART. Third-Programme HEALTH- ERN-01-2016: ERN-2016 - European Reference Networks. European Commission. PI: Georgia Sarquella Brugada. 01/03/2017-28/02/2019

Selected clinical trials & observational studies

- Open phase IV multicentre study to assess the diagnostic effectiveness of Nuubo nECGband prior to Holter monitor implantation. EUDRACT: N/A. Promoter: Smart Solutions Technologies S.L. PI: Georgia Sarquella Brugada. 11/03/2015-

Selected publications

- Brugada-Terradellas J, Campuzano O, Arbelo E, Sarquella-Brugada G and Brugada R. Present Status of Brugada Syndrome JACC State-of-the-Art Review. J Am Coll Cardiol 2018. 72(9): 1046-1059. FI: 16,834(Q1)

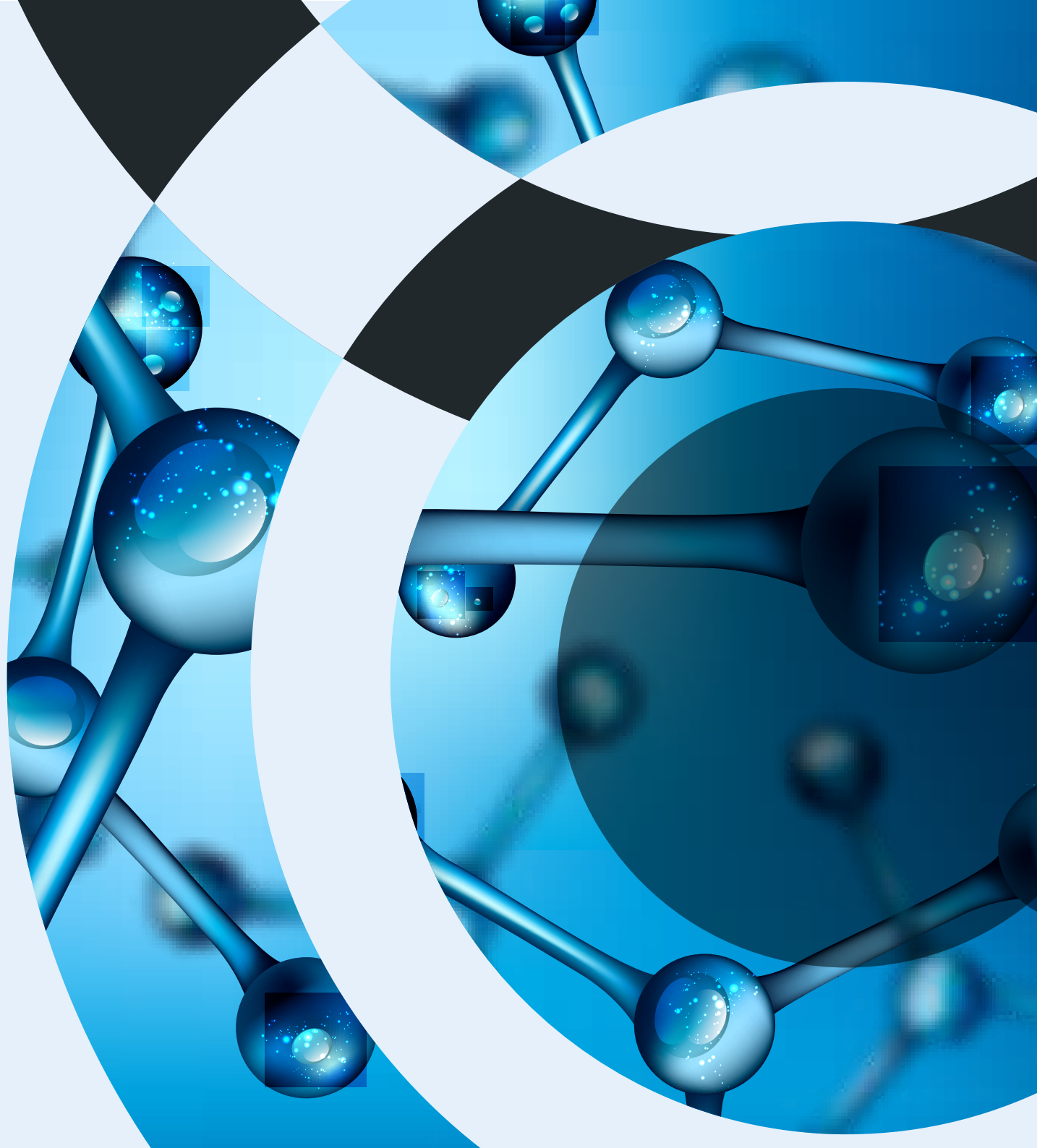
- Campuzano O, Beltramo P, Fernández A, Iglesias A, García L, Allegue C, et al. Molecular autopsy in a cohort of infants died suddenly at rest. Forensic Sci Int Genet 2018. 37: 54-63. FI: 5,637(Q1)
- Mates J, Mademont-Soler I, Del Olmo B, Ferrer-Costa C, Coll M, Pérez-Serra A, et al. Role of copy number variants in sudden cardiac death and related diseases: genetic analysis and translation into clinical practice. Eur J Hum Genet 2018. 26(7): 1014-1025. FI: 3,636(Q2)
- Michowitz Y, Milman A, Sarquella-Brugada G, Andorin A, Champagne J, Postema PG, et al. Fever-related arrhythmic events in the multicenter Survey on Arrhythmic Events in Brugada Syndrome. Heart Rhythm 2018. 15(9): 1394-1401. FI: 4,743(Q1)
- Milman A, Andorin A, Gourraud JB, Postema PG, Sacher F, Mabo P, et al. Profile of patients with Brugada syndrome presenting with their first documented arrhythmic event: Data from the Survey on Arrhythmic Events in BRUGADA Syndrome (SABRUS). Heart Rhythm 2018. 15(5): 716-724. FI: 4,743(Q1)
- Milman A, Gourraud JB, Andorin A, Postema PG, Sacher F, Mabo P, et al. Gender differences in patients with Brugada syndrome and arrhythmic events: Data from a survey on arrhythmic events in 678 patients. Heart Rhythm 2018. 15(10): 1457-1465. FI: 4,743(Q1)
- White JL, Chang AM, Cesar S and Sarquella-Brugada G. Can sudden cardiac death in the young be predicted and prevented? Lessons from autopsy for the emergency physician. Emergencias 2018. 30(3): 194-200. FI: 3,608(Q1)



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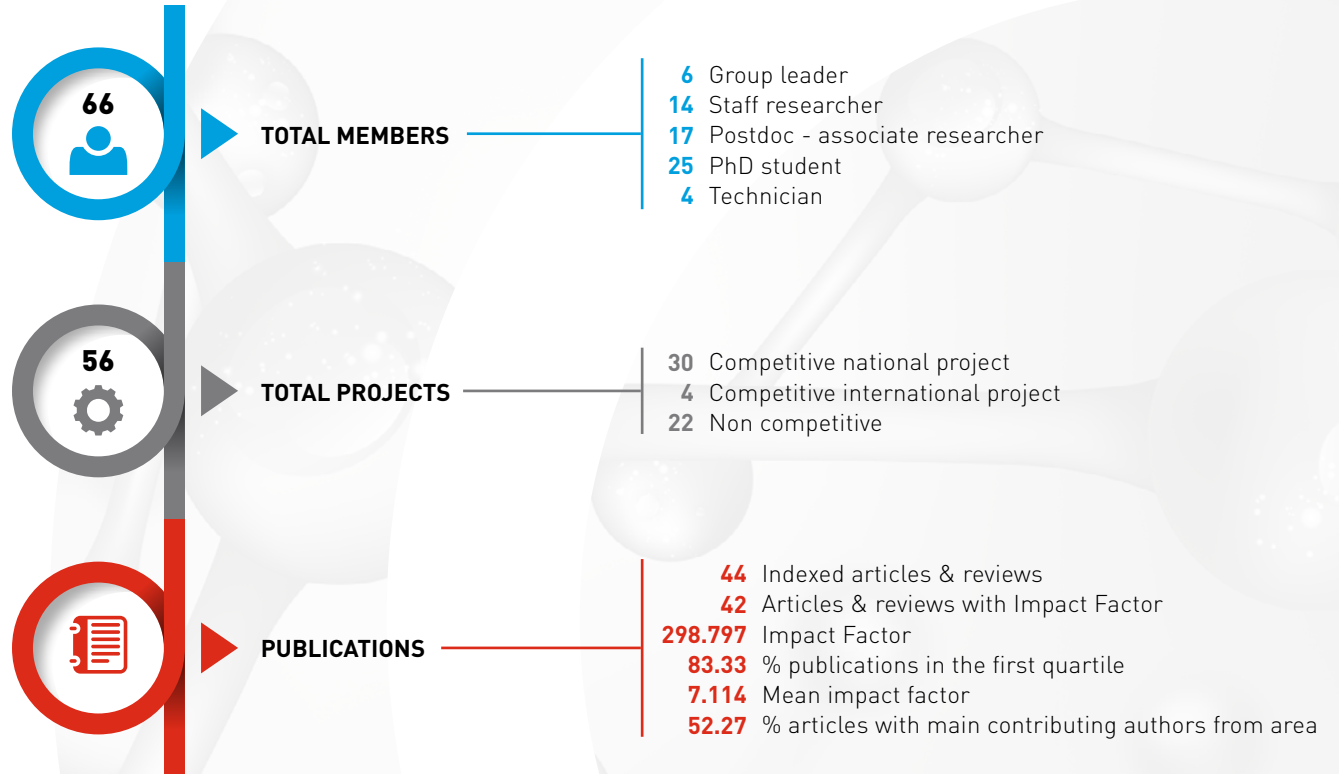
CROSS-SECTIONAL AREAS

- 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, PhD



MOLECULAR BIOLOGY AND GENE REGULATION OF ADIPOSE TISSUE AND ITS PATHOLOGIES

Francesc Villarroya Gombau, PhD

Members

Staff researcher

Marta Giralt Oms, Roser Iglesias Coll, Teresa Mampel Astals, Marion Peyrou, Anna Planavila Posta.

Postdoc - associate researcher

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

PhD students

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

Technician

Mercedes Morales Rueda, Gemma Moreno Besora, Albert Però Garcia



PERSONAL
RESEARCHER



PROJECTS



PUBLICATIONS
WITH IF



FIRST QUARTILE
PUBLICATIONS

Selected projects

- PI17/00420. Study of inter-organ molecular signalling mediators involved in the prevalence and progression of subclinical atheromatosis in patients with HIV infection. Ajuts de l'Acció Estratègica en Salut: Subprograma de projectes d'investigació en salut (PI). Ministerio de Economía y Competitividad. PI: Marta Giralt Oms. 2018-2020
- 2017SGR330. Genetics and molecular biology of mitochondrial proteins and associated pathologies. Convocatòria d'ajuts per donar suport a les activitats dels grups de recerca (SGR). Agència de Gestió d'Ajuts Universitaris i de Recerca (AGAUR). PI: Francesc Villarroya Gombau. 2018-2020
- SAF2017-85772-R. New secreted factors of brown adipose tissue ("batokines") involved in its beneficial functions for metabolism. Convocatòria d'ajuts del Programa Estatal d'Investigació, Desenvolupament i Innovació Orientada als reptes de la Societat. Modalitat Projectes I+D+i. Ministerio de Economía y Competitividad. PI: Francesc Villarroya Gombau. 2018-2020
- CB06/03/0025P. athophysiology of obesity and nutrition. Convocatòria d'ajuts destinats a finançar estructures estables d'investigació cooperativa, a l'àrea de biomedicina i ciències de la salut, en el marc del Programa Ingenio 2010, Programa Consolider, accions CIBER. Ministerio de Sanidad y Consumo. PI: Francesc Villarroya Gombau. 2018-2019
- BFU2017-90578-REDT. Plasticity of adipose tissue, applications and associated pathologies. Convocatòria d'ajuts del Programa Estatal de Foment de la Investigació Científica i Tècnica d'Excel·lència. Subprograma Estatal de generació

del coneixement. Accions de dinamització 'Xarxes d'Excel·lència'. Ministerio de Economía y Competitividad. PI: Francesc Villarroya Gombau. 2018-2020

Selected publications

- Villarroya J, Gallego-Escuredo JM, Delgado-Anglés A, Cairo M, Moure R, Gracia Mateo M, et al. Aging is associated with increased FGF21 levels but unaltered FGF21 responsiveness in adipose tissue. *Aging Cell* 2018. 17(5): e12822. FI: 7,627(Q1)
- Zarei M, Barroso E, Palomer FX, Dai J, Rada P, Quesada T, et al. Hepatic regulation of VLDL receptor by PPAR β /d and FGF21 modulates non-alcoholic fatty liver disease. *Mol Metab* 2018. 8: 117-131. FI: 6,291(Q1)
- Villarroya F, Cereijo R, Gavaldà A, Villarroya J and Giralt M. Inflammation of brown/beige adipose tissues in obesity and metabolic disease. *J Intern Med* 2018. 284(5): 492-504. FI: 6,754(Q1)
- Domingo P, Fernández Carasa I, Gallego-Escuredo JM, Torres F, Gutierrez MDM, Mateo MG, et al. Effects of docosahexanoic acid on metabolic and fat parameters in HIV-infected patients on cART: A randomized, double-blind, placebo-controlled study. *Clin Nutr* 2018. 37(4): 1340-1347. FI: 5,496(Q1)

Networks

- CIBEROBN (Fisiopatología de la obesidad y nutrición)
- ADIPOPLAST - RED temática de excelencia (Red de investigación sobre plasticidad adiposa y su impacto fisiopatológico)

HUMAN MOLECULAR GENETICS I

Daniel Raúl Grinberg Vaisman, PhD

This year's highlights

In 2018 we described new genes and mutations responsible for intellectual disability in patients originally diagnosed with Opitz C or Bohring-Opitz syndrome (FOXP1, ASXL1, PIGT, DPH1, MAGEL2 and others). We analysed the role of the WNT16, DKK1 and SOST genes and of miRNAs in relation to osteoporosis. We have also participated in a multi-centre study on the determinants of osteoporotic fractures and we conducted functional studies on the GGPS1 gene in relation to atypical femoral fractures. In the field of lysosomal disorders, we continued our studies on therapeutic approaches in neurons derived from iPS cells for Sanfilippo C syndrome and in the Niemann-Pick C murine model. In the field of neurogenetics, we identified genes involved in various neurological and psychiatric disorders (autism, migraine, aggressiveness, ADHD) and we investigated the shared inheritability between 25 brain disorders. We also proved the importance of paternally inherited regulatory genetic variants in autism. Dr R. Corominas joined this line of research this year. Her work is notable in such fields as 7q11.23 aneuploidies and autism.

Members

Staff researcher

Susana Balcells Comas, Bru Cormand Rifà, Roser Corominas Castiñera, Lluïsa Vilageliu Arqués
Postdoc - associate researcher

Judit Cabana Domínguez, Noelia Fernández Castillo, Raquel Rabionet Janssen, Bárbara Torrico Avilés, Roser Urrizti Frexadas.

PhD student

Ester Antón Galindo, Noelia Benetó Gándia, Laura Castilla Vallmanya, Núria Martínez Gil, Laura Pineda Cirera, Neus Roca Ayats, Anu Shivalikanjli

Technician

Mónica Cozar Morillo



PERSONAL
RESEARCHER



PROJECTS



PUBLICATIONS
WITH IF



FIRST QUARTILE
PUBLICATIONS

Selected projects

- 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
- Mastering skills in the training Network for attention deficit hyperactivity and autism spectrum Disorders (MiND). Unió Europea (H2020). Horizon 2020 (Research and Innovation Framework Programme: Training Networks). 643051. PI: Bru Cormand Rifà. 2015-2018

Selected publications

- Urreiziti R, Damanti S, Esteve-Matanza C, Franco-Valls H, Castilla-Vallmanya L, Tonda R, et al. A De Novo FOXP1 Truncating Mutation in a Patient Originally Diagnosed as C Syndrome. *Sci Rep* 2018. 8(1): 694-694. FI: 4,122(Q1)
- Martínez-Gil N, Roca-Ayats N, Monistrol-Mula A, García-Giralt N, Díez-Pérez A, Nogués X, et al. Common and rare variants

- of WNT16, DKK1 and SOST and their relationship with bone mineral density. *Sci Rep* 2018. 8(1): 10951-10951. FI: 4,122(Q1)
- Roca-Ayats N, Ng PY, Garcia-Giralt N, Falcó-Mascaró M, Cozar M, Abril JF, et al. Functional Characterization of a GGPPS Variant Identified in Atypical Femoral Fracture Patients and Delineation of the Role of GGPPS in Bone-Relevant Cell Types. *J Bone Miner Res* 2018. 33(12): 2091-2098. FI: 6,314(Q1)
- Cabana-Domínguez J, Arenas C, Cormand B and Fernández-Castillo N. MiR-9, miR-153 and miR-124 are down-regulated by acute exposure to cocaine in a dopaminergic cell model and may contribute to cocaine dependence. *Transl Psychiatry* 2018. 8(1): 173-173. FI: 4,691(Q1)
- Brandler WM, Antaki D, Gujral M, Kleiber ML, Whitney J, Maile MS, et al. Paternally inherited cis-regulatory structural variants are associated with autism. *Science* 2018. 360(6386): 327-331. FI: 41,058(Q1)

Networks

- CIBERER, 2006.
- GEFOS/GENOMOS (international consortium for the study of the genetic bases of osteoporosis), 2002.
- IMpACT (International Multicenter Persistent ADHD Collaboration), 2007.
- IHGC (International Headache Genetics Consortium), 2006.
- PGC (Psychiatric Genomics Consortium), 2007.
- ECNP (European College of Neuropsychopharmacology) Network: ADHD across the lifespan, 2015.
- Gene Stroke, Red Española de Genética del Ictus, 2007.

4 CROSS-SECTIONAL AREAS

4.1.
Genetics, molecular
biology and gene
therapy

Coordinator:
Marçal Pastor-Anglada

HUMAN MOLECULAR GENETICS II

Gemma Marfany Nadal, PhD

This year's highlights

The research of our group, UB CIBERER U-718, is focused on identifying new genes causing hereditary retinal dystrophy, and on characterising their function. It should be pointed out that the composition of the group changed greatly in 2018. The group's senior leader is Dr. Marfany, and one CIBERER postdoctoral researcher and three predoctoral researchers in different stages of their doctoral theses have joined us. In 2018 we formed a spin-off, DBGen Ocular Genomics, to offer genetic diagnosis to families affected by hereditary retinal disorders. In the research field, the group has developed new gene analysis tools: 1) cellular tools (primary cultures of retinal and hippocampus neurons), 2) organoids (retinal cultures, generation of optical cups from patient-derived hiPSC cells), and 3) animal cells (zebrafish, knockout mice generated by cre/loxP recombination, and mice models generated using CRISPR gene editing). In short, 2018 was a year of change, consolidation and growth.

Members

Postdoc - associate researcher

Serena Mirra

PhD student

Izarbe Aísa Marín, Elena Borrego Domènech,
Vasileios Toulis



Selected projects

- Contractes de recerca/serveis. 309247. Spark Therapeutics England, Inc. PI: Roser Gonzalez Duarte. 2017-2019
- PROJETE 201417-30-31-32. Dissecting protein trafficking in retinal neurodegeneration by super-resolution imaging on animal models and human iPSCs. LA MARATÓ 2013. Fundació de La Marató TV3. PI: Roser Gonzalez Duarte & Gemma Marfany. 2015-2018
- SAF2016-80937-R. R&D&i 2016 projects: a nationwide research, development and innovation programme addressing the challenges of society. Ministerio de Economía y Competitividad. PI: Gemma Marfany & Roser Gonzalez. 2016-2019

Selected publications

- De-Castro M, Tonda R, Marfany G, Casaroli-Marano RP and Gonzalez R. Novel mutation in the choroideremia gene and multi-Mendelian phenotypes in Spanish families. Br J Ophthalmol 2018. 102(10): 1378-1386. FI: 3,384(Q1)

Networks

- CIBERER - UNITAT 718 (Centro de Investigación Biomédica en Red- Enfermedades Raras)
- COST ACTION BM1307 - PROTEOSTASIS (Member of the Core Management Group) 2013-2018

MOLECULAR PHARMACOLOGY AND EXPERIMENTAL THERAPIES

Marçal Pastor-Anglada, PhD

Members

Staff researcher

Sandra Perez-Torras, 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, Eduard Manel Mas
Marin

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CROSS-SECTIONAL
AREAS

4.1.

Genetics, molecular
biology and gene
therapy

Coordinator:

Marçal Pastor-Anglada



PERSONAL
RESEARCHER



PROJECTS



PUBLICATIONS
WITH IF



FIRST QUARTILE
PUBLICATIONS

Selected projects

- 2017SGR416. Molecular pharmacology and experimental therapies. (MPET). Convocatòria d'ajuts per donar suport a les activitats dels grups de recerca (SGR). Agència de Gestió d'Ajuts Universitaris i de Recerca (AGAUR). PI: Marçal Pastor-Anglada. 2018-2020
- CB06/04/0063 Liver and digestive disorders. Convocatòria d'ajuts destinats a finançar estructures estables d'investigació cooperativa, a l'àrea de biomedicina i ciències de la salut, en el marc del Programa Ingenio 2010, Programa Consolider, accions CIBER. Ministerio de Sanidad y Consumo. PI: Marçal Pastor-Anglada. 2018-2019

Selected publications

- Pastor-Anglada M, Urtasun N and Perez-Torras S. Intestinal Nucleoside Transporters: Function, Expression, and Regulation. *COMPR PHYSIOL* 2018. 8(3): 1003-1017. FI: 5,797(Q1)
- Pastor-Anglada M and Perez-Torras S. Emerging Roles of Nucleoside Transporters. *FRONT PHARMACOL* 2018. 9: 606-606. FI: 3,831(Q1)
- Pastor-Anglada M and Perez-Torras S. Who Is Who in Adenosine Transport. *FRONT PHARMACOL* 2018. 9: 627-627. FI: 3,831(Q1)

Networks

- CIBER-EHD

PHARMACOLOGICAL TARGETS IN INFLAMMATION AND METABOLIC DISEASES

Manuel Vázquez Carrera, PhD

This year's highlights

In our group we made two notable findings in 2018. On the one hand, we advanced in the study of the PPAR (peroxisome proliferator-activated receptor) β/δ nuclear receptor in the regulation of non-alcoholic fatty liver development and, more specifically, of how this receptor modulates the VLDL and FGF21 (fibroblast growth factor 21) receptor levels in the development of this disorder. Additionally, we also proved how the BACE1 (β -secretase/ β -site amyloid precursor protein (APP)-cleaving enzyme 1) enzyme and the product of its activity, soluble APP β (sAPP β), take part in the occurrence of endoplasmic reticulum stress, inflammation, and insulin resistance induced by lipids in skeletal muscle.

Members

Staff researcher

F. Xavier Palomer Tarridas

Postdoc - associate researcher

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

PhD student

Mohammad Zarei



Selected projects

- New HRI activators for the treatment of type 2 Diabetes Mellitus. Fundació Caixa de Pensions "La Caixa". Caixa Impulse Programme 2018. CI18-00006. PI: Manuel Vazquez Carrera. 2018-2020
- 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

Selected publications

- Palomer FX, Pizarro J and Vazquez M. Emerging Actors in Diabetic Cardiomyopathy: Heartbreaker Biomarkers or Therapeutic Targets?. Trends Pharmacol Sci 2018. 39(5): 452-467. FI: 12,108(Q1)
- Zarei M, Barroso E, Palomer FX, Dai J, Rada P, Quesada T, et al. Hepatic regulation of VLDL receptor by PPAR β /d and FGF21 modulates non-alcoholic fatty liver disease. Mol Metab 2018. 8: 117-131. FI: 6,291(Q1)
- Palomer FX, Pizarro J, Barroso E and Vazquez M. Palmitic and Oleic Acid: The Yin and Yang of Fatty Acids in Type 2 Diabetes Mellitus. Trends Endocrinol Metab 2018. 29(3): 178-190. FI: 10,769(Q1)
- Botteri G, Salvadó L, Gumà A, Lee Hamilton D, Meakin PJ, Montagut G, et al. The BACE1 product sAPP β induces ER stress and inflammation and impairs insulin signaling. METABOLISM 2018. 85: 59-75. FI: 5,963(Q1)

Networks

- 2007 CIBERDEM

MICROBIOTA INTESTINAL

Laura Baldomà Llavines, PhD

This year's highlights

In the context of the research line focused to the study of membranes vesicles (MVs) secreted by gut microbiota and probiotics we have contributed to prove the role of these MVs in the immune priming mediated by gut microbiota. Activation of the cytosolic immune receptor NOD-1 by internalized microbiota MVs in intestinal epithelial cells leads to controlled inflammatory responses that contribute to the training of immune system. This is an essential mechanism to trigger appropriate immune responses against invading microbes to assist pathogen eradication and host survival. In addition, we have provided new insights on the role of microbiota MVs and secreted factors on the modulation of intestinal tight junctions, expanding their barrier-protective effects against pathogen-induced disruption.

Members

Staff researcher

Josefa Badia Palacin, Rosa Giménez Claudio
Postdoc - associate researcher

Rodrigo Vera García

PhD student

M^a Alexandra Cañas Pacheco, Natalia Díaz Garrido, M^a José Fábrega Fernández



PERSONAL
RESEARCHER



PROJECTS



PUBLICATIONS
WITH IF



FIRST QUARTILE
PUBLICATIONS

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

- Limon D, Fabrega MJ, Calpena AC, Badia J, Baldoma L and Pérez-García L. Multifunctional Serine Protease Inhibitor-Coated Water-Soluble Gold Nanoparticles as a Novel Targeted Approach for the Treatment of Inflammatory Skin Diseases. *Bioconjug Chem* 2018. 29(4): 1060-1072. FI: 4,485(Q1)
- Solé M, Balcells C, Crespo M, Quirante J, Badia J, Baldoma L, et al. Synthesis, characterization and biological activity of new cyclometallated platinum(IV) complexes containing a para-tolyl ligand. *Dalton Trans* 2018. 47(27): 8956-8971. FI: 4,099(Q1)
- Cañas MA, Fabrega MJ, Gimenez R, Badia J and Baldoma L. Outer Membrane Vesicles From Probiotic and Commensal Escherichia coli Activate NOD1-Mediated Immune Responses in Intestinal Epithelial Cells. *FRONT MICROBIOL* 2018. 9: 498-498. FI: 4,019(Q1)
- Reig M, Bosque R, Font-Bardía M, Calvis C, Messeguer R, Baldoma L, et al. A study of the properties, reactivity and anticancer activity of novel N-methylated-3-thiazolyl or 3-thienyl carbazoles and their Pd(II) and Pt(II) complexes. *J Inorg Biochem* 2018. 184: 134-145. FI: 3,063(Q1)

4

CROSS-SECTIONAL AREAS

4.1.

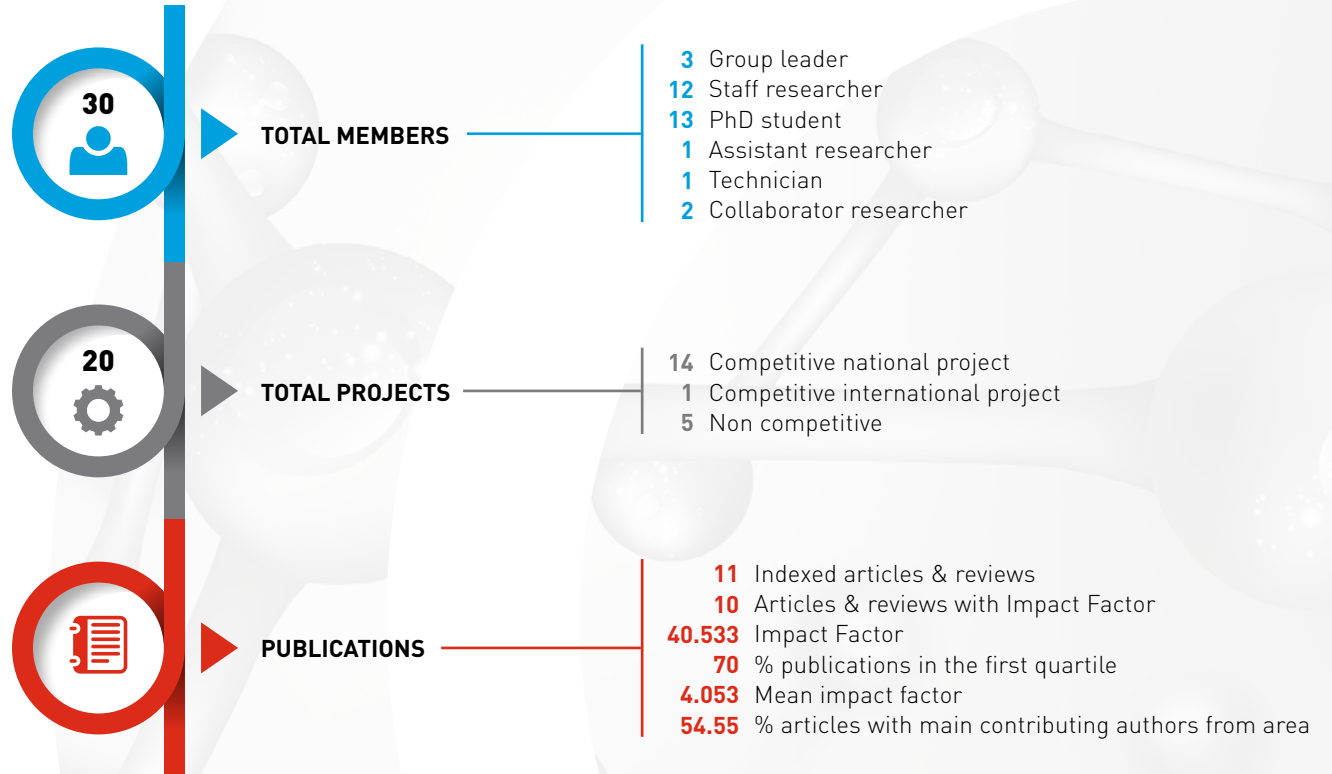
Genetics, molecular biology and gene therapy

Coordinator:

Marçal Pastor-Anglada

4.2. Neuropsychology, cognitive and development neuroscience

Coordinator: Carles Escera Micó, PhD



NEUROPSYCHOLOGY

Maria Mataró Serra, PhD

This year's highlights

The group published 3 articles in impact journals, read 2 theses with international mention, and has 7 competitive projects.

Members

Staff researcher

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

PhD student

Júlia Ballester Plané, Montse Blasco Sierra, Alba Castells Sánchez, María García Galant, Noemí Lamónja Vicente, Olga Laporta Hoyos, Sandra Luís Ruiz, Jonatan Ottino González, Xavier Prats Soteras

Collaborator researcher

Cristina Sánchez Castañeda

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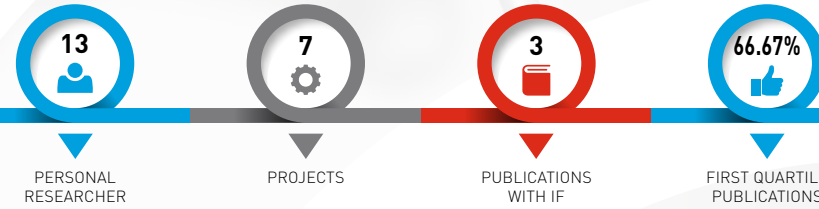
CROSS-SECTIONAL AREAS

4.2.

Neuropsychology, cognitive and development neuroscience

Coordinator:

Carles Escera Micó



Selected projects

- PSI2017-86536-C2-1-R. Inflammation, stress and microbiota as mediators between obesity and brain functioning during adolescence. Convocatòria d'ajuts del Programa Estatal d'Investigació, Desenvolupament i Innovació Orientada als reptes de la Societat. Modalitat Projectes I+D+I. Ministerio de Economía y Competitividad PI: Maria Angeles Jurado Luque. 2018-2021
- Effects of combined therapies including exercise, mindfulness and cognitive stimulation on cognition and neuroplasticity in patients with chronic ischaemic stroke. Ajuts econòmics a projectes de recerca de la Fundació Marató de TV3. PI: Maria Mataró Serrat. 2018-2020
- Virtual training raising citizens' awareness about sleep and healthy living' (SweetDreams). KIC Health 2018. Unió Europea. PI: Maria Mataró Serrat. 2018

Selected publications

- Ballester J, Laporta O, Macaya A, Poo P, Meléndez-Plumed M, Toro-Tamargo E, et al. Cognitive functioning in dyskinetic cerebral palsy: Its relation to motor function, communication and epilepsy. Eur J Paediatr Neurol 2018. 22(1): 102-112. FI: 2,362(Q2)
- Laporta-Hoyos O, Fiori S, Pannek K, Ballester-Plané J, Leiva D, Reid LB, et al. Brain lesion scores obtained using a simple semi-quantitative scale from MR imaging are associated with motor function, communication and cognition in dyskinetic cerebral palsy. Neuroimage Clin 2018 19:892-900. FI: 3,869 (Q1)
- Ottino-gonzález J, Jurado MA, García-García I, Segura B, Marqués-Iturria I, Sender-Palacios MJ, et al. Allostatic load and disordered White matter microstructure in overweight adults. Sci Rep 2018. 8:15898. FI: 4,122 (Q1)

COGNITIVE NEUROSCIENCE

Carles Escera Micó, PhD

This year's highlights

In 2018, our research group continued with its activity in the study of brain mechanisms of auditory perception, in both the healthy population and in ageing population, especially promoting the research line addressed to identifying biomarkers for neurocognitive development in newborns. The most notable activities during this period were the holding of the 8th Mismatch Negativity Conference "MMN from basic science to clinical applications", in Helsinki (Finland) in June 2018; the guest lecture held at the University of Toronto (Canada) "Neurocognitive mechanisms in audition as revealed with the frequency-following response"; and the leading of two work packs (RL2-Psychoacoustics and RL3-Neuroscience) in the project "ARTSOUNDSCAPES - The sound of special places: exploring rock art soundscapes and the sacred" financed with an advanced grant from the European Research Council (ERC; EC grant agreement number 787842).

Members

Staff researcher

Imma Clemente Lapena, Maria José Corral López, Jordi Costa Faidella, Maribel Nuñez Peña, Iria San Miguel Insúa, Hans Supèr, Marc Via Garcia

PhD student

Natàlia Gorina, Alejandro Lerer, Teresa Ribas Prats.

Technician

Sonia Arenillas



PERSONAL
RESEARCHER



PROJECTS



PUBLICATIONS
WITH IF



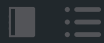
FIRST QUARTILE
PUBLICATIONS

Selected projects

- PSI2017-85600-P. Coding of self-reference stimuli versus external-reference stimuli: short- and long-term consequences. Ministerio de Economía y Competitividad. PI: Iria San Miguel Insua. 2018-2020
- Research/service contracts. Soundev. PI: Carles Escera Mico. 2016-2018
- SGR2014-1413. Grant to incentivise and consolidate existing research of excellence in the public universities of Catalonia. Programa ICREA Academia 2015. Convocatòria del Programa Icrea Acadèmia. Fundació Institució Catalana de Recerca i Estudis Avançats (ICREA). PI: Carles Escera Mico. 2016-2020
- 2015 DI 051. Development of improvement attention measures by novel eye tracking technology. Industrial Doctorates. Agència de Gestió d'Ajuts Universitaris i de Recerca (AGAUR). PI: Hans Supèr. 2016-2019

Selected publications

- Marzecova A, Schettino A, Widmann A, San-Miguel I, Kotz SA and Schroger E. Attentional gain is modulated by probabilistic feature expectations in a spatial cueing task: ERP evidence. *Sci Rep* 2018. 8(1): 54-54. FI: 4,122(Q1)
- Escera C, López-Caballero F and Gorina N. The Potential Effect of Forbrain as an Altered Auditory Feedback Device. *J Speech Lang Hear Res* 2018. 61(4): 801-810. FI: 1,906(Q1)
- do Vale S and Escera C. Dehydroepiandrosterone and Dehydroepiandrosterone-Sulfate and Emotional Processing. *Vitam Horm* 2018. 108: 413-441. FI: 3,286(Q2)
- Aghamolaei M, Jafari Z, Grimm S, Zarnowiec K, Najafi-Koopae M and Escera C. The effects of aging on early stages of the auditory deviance detection system. *Clin Neurophysiol* 2018. 129(11): 2252-2258. FI: 3,614(Q2)



DEVELOPMENTAL NEUROSCIENCE

Laura Bosch Galceran, PhD

Members

Staff researcher
Ferran Pons Gimeno, Marta Ramon Casas
Postdoc - associate researcher
Clément François
PhD student
Joan Birulés Muntané
Assistant researcher
Jessica Sánchez-Galán
Collaborator researcher
Horgina Solé Montoliu

4

CROSS-SECTIONAL AREAS

4.2.
Neuropsychology,
cognitive and
development
neuroscience

Coordinator:
Carles Escera Micó



Selected projects

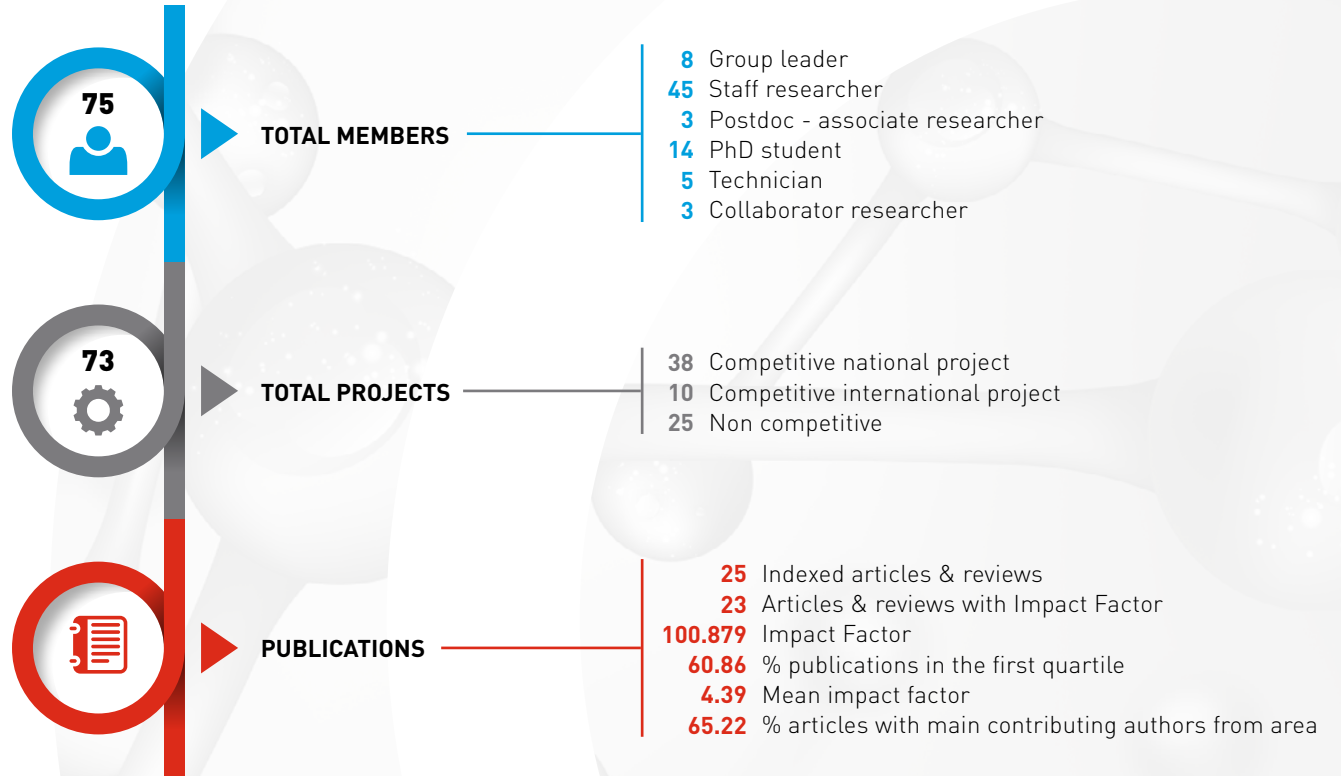
- PSI2014-55105-P. Multimodal information at start of language learning: use of audiovisual keys in neurotypical development and in populations at neurocognitive risk. Convocatòria d'ajuts del Programa Estatal de Foment de la Investigació Científica i Tècnica d'Excel·lència. Subprograma Estatal de generació del coneixement. Modalitat Projectes d'R+D. Ministerio de Economía y Competitividad. PI: Laura Bosch Galceran. 2015-2018
- Collaboration agreement for research on language acquisition in multilingual children under 18 years of age. St. Peter's School. PI: Ferran Pons Gimeno. 2017-2018
- Collaboration agreement for research on cognition and speech perception in children. Fundació Privada Betània-Patmos. PI: Ferran Pons Gimeno. 2017-2018

Selected publications

- Perissinotti A, Niñerola-Baizán A, Rubí S, Carreño M, Martí-Fuster B, Aparicio J, et al. PISCOM: a new procedure for epilepsy combining ictal SPECT and interictal PET. Eur J Nucl Med Mol Imaging 2018. 45(13): 2358-2367. FI: 7,704(Q1)
- Elmer S, Albrecht J, Valizadeh SA, François C and Rodríguez-Fornells A. Theta Coherence Asymmetry in the Dorsal Stream of Musicians Facilitates Word Learning. Sci Rep 2018. 8(1): 4565-4565. FI: 4,122(Q1)
- Vaquero L, Ramos-Escobar N, François C, Penhune V and Rodríguez-Fornells A. White-matter structural connectivity predicts short-term melody and rhythm learning in non-musicians. Neuroimage 2018. 181: 252-262. FI: 5,426(Q1)

4.3. Biomedical engineering

Coordinator: Pere Caminal Magrans, PhD



BIOINFORMATICS AND DATA ANALYSIS PLATFORM

Pere Caminal Magrans, PhD &
Alexandre Perera Lluna, PhD

Members

Staff researcher

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

4

CROSS-SECTIONAL
AREAS

4.3.

Biomedical engineering

Coordinator:

Pere Caminal Magrans



PERSONAL
RESEARCHER



PROJECTS



PUBLICATIONS
WITH IF



FIRST QUARTILE
PUBLICATIONS

Selected projects

- H2020-780262-SHARE4RARE. H2020-780262-SHARE4RARE, Social media platform Dedicated to rare diseases, using collective intelligence for the advanced generation of Awareness and knowledge on this large group of diseases. European Commission. PI: Alexandre Perera Lluna. 2018-2020
- Predictive cancer biomarkers in peripheral blood. Universal Diagnostics. PI: Alexandre Perera Lluna. 2018-2020
- Online platform for Classification of Samples of meduloblastoma. Hospital Sant Joan de Déu. PI: Alexandre Perera Lluna. 2018-2019

Selected publications

- Lupón J, Gavidia G, Eva Ferrer Vidal-Barraquer, de Antonio M, Perera A, López-Ayerbe J, et al. Dynamic Trajectories of Left Ventricular Ejection Fraction in Heart Failure. J Am Coll Cardiol 2018. 72(6): 591-601. FI: 16,834(Q1)
- Picart-Armada S, Thompson WK, Buil A and Perera A. diffuStats: an R package to compute diffusion-based scores on biological networks. Bioinformatics 2018. 34(3): 533-534. FI: 5,481(Q1)
- Ziarrusta H, Mijangos L, Picart-Armada S, Irazola M, Perera A, Usobiaga A, et al. Non-targeted metabolomics reveals alterations in liver and plasma of gilt-head bream exposed to oxybenzone. Chemosphere 2018. 211: 624-631. FI: 4,427(Q1)
- Picart-Armada S, Fernández-Albert F, Vinaixa M, Yanes O and Perera A. FELLA: an R package to enrich metabolomics data. BMC Bioinformatics 2018. 19(1): 538-538. FI: 2,213(Q1)



COMPUTER GRAPHICS

Daniela Tost Pardell, PhD

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



PERSONAL
RESEARCHER



PROJECTS

Selected projects

- BEST: Bienestar y Salud mental en el puesto de trabajo. Institut de Recerca Sant Joan de Déu. PI: Dani Tost. 2018-2019

BIOMATERIALS AND TISSUE ENGINEERING

Maria Pau Ginebra Molins, PhD

Members

Staff researcher

Albert Barba Serrahima, Cristina Canal Barnils, Montserrat Español Pons, Francesc Xavier Gil Mur, Jordi Guillem Martí, Mireia Hoyis Nogués, Jose María Manero Planella, Carles Mas Moruno, Marta Pegueroles Neyera, Daniel Rodríguez Rius, Joanna Maria Sadowska, Èlia Vidal Girona

Postdoc - associate researcher

Juan Tornín

PhD student

Mar Bonany, Inès Hamouda, Joanna Konka, Miguel Mateu, Joaquim Minguella, Luis Oliver, Santiago Raymond, Diego Torres
Technician

Meritxell Molmeneu Trias, Mònica Ortiz Hernández, Miquel Punset Fusté

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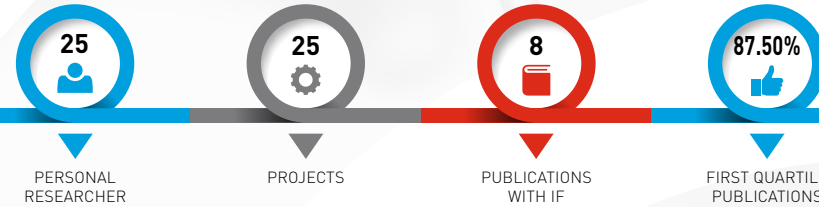
CROSS-SECTIONAL
AREAS

4.3.

Biomedical engineering

Coordinator:

Pere Caminal Magrans



Selected projects

- H2020-MSCA-IF-2016. DEBBIE: Database of Experimental Biomaterials and their Biological Effect. H2020. European Commission. PI: Maria Pau Ginebra. 2018-2020
- H2020-MSCA-IF-2018 Proposal: 838621. ENGAGE: ENGINEERING extracellular matrix-based de novo proteins with high Affinity to Growth factors for Enhancing bone regeneration. European Commission. PI: Maria Pau Ginebra, Jordi Guillem. 2018-2022
- EQC2018-004902-P. Characterisation system of the mechanical response of advanced materials to multidimensional scales. AEI. PI: Lluís Llanes, Maria Pau Ginebra. 2018-2019
- Cátedra AMES-UPC. AMES: PI: José María Manero. 2018-2019
- In-vitro biological assessment of coatings by means of in-vitro biological tests. AIN: Asociación de la Industria Navarra. PI: José María Manero. 2018-2019
- Preparation, texturing, functionalising and characterisation of polypropylene (PP) discs, and posterior surface characterisation by Raman spectroscopy of discs cultured in the presence of human fibroblasts in order to analyse the extracellular matrix which they deposit, for the VHIR General Surgery Research Group. VHIR: Fundació Hospital Universitari Vall Hebron - Institut de Recerca. PI: José María Manero. 2018-2019

Selected publications

- Sadowska JM, Wei F, Guo J, Guillem J, Ginebra MP and Xiao Y. Effect of nano-structural properties of biomimetic hydroxyapatite on osteoimmunomodulation. Biomaterials 2018. 181: 318-332. FI: 8,806(Q1).
- Raymond S, Maazouz Y, Montufar EB, Perez RA, González B, Konka J, et al. Accelerated hardening of nanotextured 3D-plotted self-setting calcium phosphate inks. Acta Biomater 2018. 75: 451-462. FI: 6,383(Q1).
- Barba A, Maazouz Y, Díez Escudero A, Rappe K, Español M, Montufar EB, et al. Osteogenesis by foamed and 3D-printed nanostructured calcium phosphate scaffolds: Effect of pore architecture. Acta Biomater 2018. 79: 135-147. FI: 6,383(Q1)
- Maria Sadowska J, Guillem J, Español M, Stähli C, Döbelin N and Ginebra MP. In vitro response of mesenchymal stem cells to biomimetic hydroxyapatite substrates: A new strategy to assess the effect of ion exchange. Acta Biomater 2018. 76: 319-332. FI: 6,383(Q1)

Networks

- 2014-2018 - IMPROMEDI: Improved Protection of Medical Devices Against Infection.
- 2015-2020 - AMICI: Anti-Microbial Coating Innovations to prevent infectious diseases.

PROCESSING AND INTERPRETATION OF BIOMEDICAL SIGNALS

Miquel A. Mañanas Villanueva, PhD

Members

Staff researcher

Joan Francesc Alonso Lopez,
Sergio Romero Lafuente

4

CROSS-SECTIONAL
AREAS

4.3.
Biomedical engineering

Coordinator:
Pere Caminal Magrans



Selected projects

- H2020-PHC-2014. Ref. 643535. Cost effective self-management of urinary incontinence addressed to women across Europe (WOMEN-UP). MINECO. PI: Miquel A. Mañanas. 2015-2018
- DPI2017-83989-R. Social network mobile app to promote patient adherence to treatment and healthy lifestyles. Fundación MAPFRE-2017. PI: Joan F. Alonso/ Joan Ramos. 2017-2018
- 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

- Ballesteros-Urpí A, Pardo-Hernández H, Ferrero-Gregori A, Torralbas-Ortega J, Puntí-Vidal J, Tàpies Olivet P, et al. Validation of the Spanish and Catalan versions of the Health of the Nation Outcome Scale for Children and Adolescents (HoNOSCA). *Psychiatry Res* 2018. 261: 554-559. FI: 2,223(Q2)
- Marateb HR, Mohebian MR, Javanmard SH, Tavallaei AA, Tajadini MH, Heidari-Beni M, et al. Prediction of dyslipidemia using gene mutations, family history of diseases and anthropometric indicators in children and adolescents: The CASPIAN-III study. *Comput Struct Biotechnol J* 2018. 16: 121-130. FI: 4,148(Q1)
- Muñoz IC, Hernández AM, Ortega YM and Mañanas MA. Respiratory muscular response to obstructive maneuvers in non-invasively ventilated healthy subjects. *Respir Physiol Neurobiol* 2018. 258: 76-81. FI: 1,792(Q3)
- Sarrafzadegan N, Hassannejad R, Marateb HR, Talaei M, Sadeghi M, Roohafza HR, et al. Correction: PARS risk charts: A 10-year study of risk assessment for cardiovascular diseases in Eastern Mediterranean Region. *PLoS One* 2018. 13(1): e0191379. FI: 2,766(Q1)
- Serna LY, Mañanas MA, Hernandez AM and Rabinovich RA. An Improved Dynamic Model for the Respiratory Response to Exercise. *Front Physiol* 2018. 9: 69-69. FI: 3,394(Q1)

INSTRUMENTATION AND BIOENGINEERING

Xavier Rosell Ferrer, PhD

Members

Staff researcher

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



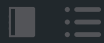
PERSONAL
RESEARCHER



PROJECTS

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
- RWECS. IAAF. PI: Xavier Rosell. 2016-2018



ROBOTICS AND VISION

Alicia Casals Gelpí, PhD

Members

Staff researcher

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

Postdoc - associate researcher

Albert Hernansanz Prats

PhD student

Narcís Sayols Baixeras

Collaborator researcher

Antonio Benedico Blanes, Carlos Morat,
Laureano Tinoco



PERSONAL
RESEARCHER



PROJECTS



PUBLICATIONS
WITH IF

Selected projects

- H2020-ICT-2017 Ref 779813. Smart Autonomous RoboticAssistant Surgeon (SARAS). H2020. PI: Alicia Casals Gelpi. 2018-2020
- H2020-ITN 2018 REF: 813782. Autonomous Intraluminal Surgery (ATLAS). H2020. PI: Alicia Casals Gelpi. 2018-2022
- Agreement Regarding Purchase of R&D. PI: Antonio B. Martinez. 2018-2019
- Development of a physical simulation platform for training in selective biopsy of sentinel lymph node. PI: Alicia Casals Gelpi, Albert hernansanz. 2018

Selected publications

- Aviles-Rivero AI, Alsaleh SM and Casals-Gelpi A. Sliding to predict: vision-based beating heart motion estimation by modeling temporal interactions. Int J Comput Assist Radiol Surg 2018. 13(3): 353-361. FI: 1,961(Q2)

Networks

- 2015 - Clúster de Salut Mental de Catalunya

MEDICAL RADIOPHYSICS

Mercè Ginjaume Egido

Members

Staff researcher
Maria Amor Duch Guillen,
Josep Sempau Roma
Postdoc - associate researcher
Sara Principi

4

CROSS-SECTIONAL
AREAS

4.3.
Biomedical engineering

Coordinator:
Pere Caminal Magrans



PERSONAL
RESEARCHER



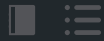
PROJECTS

Selected projects

- Personal Online Dosimetry Using computational Methods (PODIUM). H2020-662287. H2020, CONCERT EJP second call. PI: Maria Amor Duch. 2018-2019
- 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

Networks

- 2000 - EURADOS, European Radiation Dosimetry Group, www.eurados.org - Voting Member i Investigadors WG12 medical imaging
- SGR 2014, SGR 2017 Dosimetria i Radiofísica mèdica



BIOMECHANICAL ENGINEERING

Josep Maria Font Llagunes, PhD

Members

Staff researcher

Ana Barjau Condomines, Daniel Clos Costa,
Rosa Pàmies Vilà, Gil Serrancolí Masferrer

PhD student

Miria Febrer Nafria



PERSONAL
RESEARCHER



PROJECTS

Selected projects

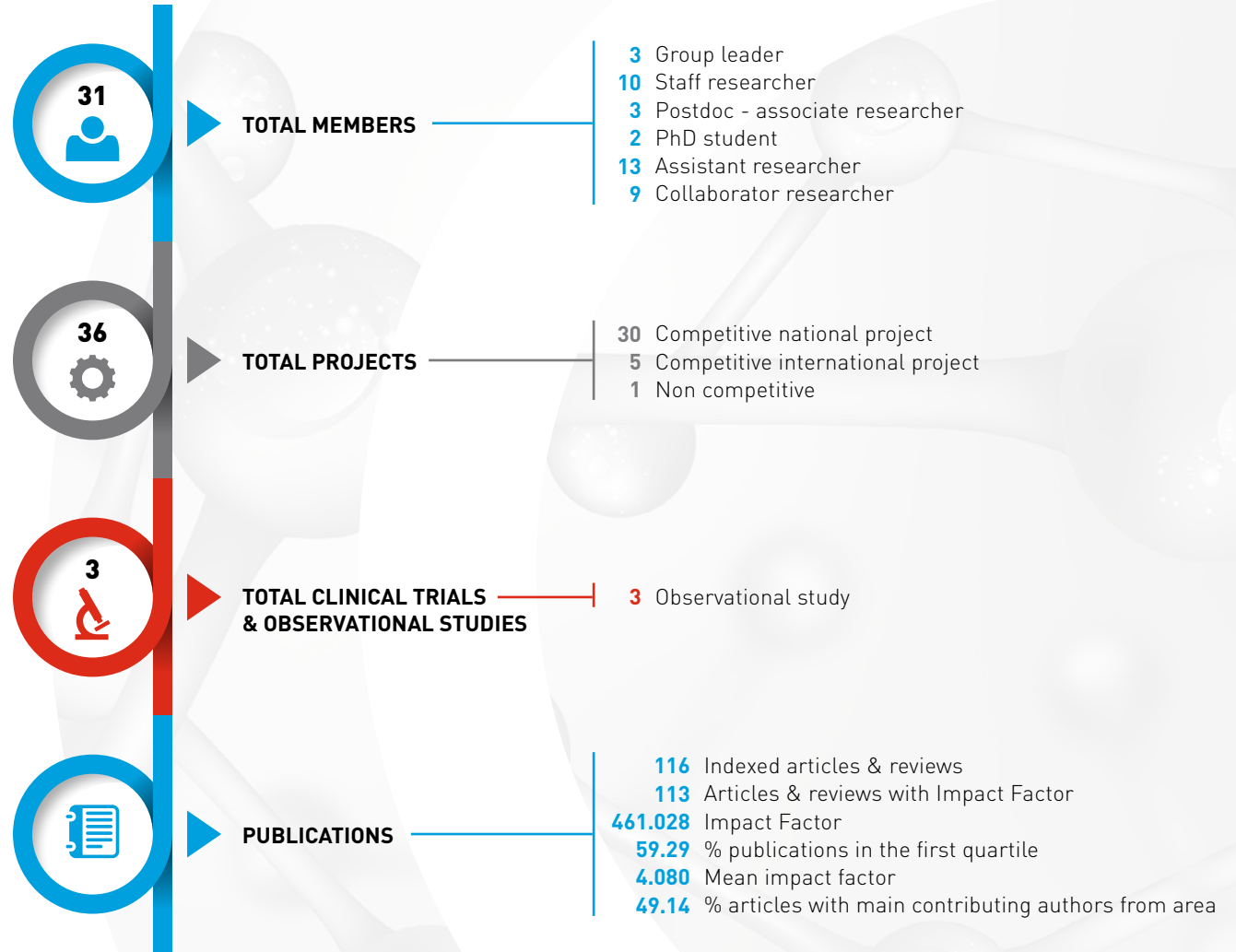
- CI17-00009. ABLE: Assistive Biorobotic Low-cost Exoskeleton. Fundació La Caixa. PI: Josep Maria Font. 2018
- Personalized robotic exoskeleton for gait rehabilitation. Fundació BBVA. PI: Josep Maria Font. 2018-2019
- DPI2017-90572-REDT. Red Nacional de Investigación en Biomecánica. MINECO. PI: Josep Maria Font. 2017-2019

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

4.4. Epidemiology

Coordinator: Josep Maria Haro Abad, MD, PhD



EPIDEMIOLOGY OF AGING: FACTORS ASSOCIATED WITH HEALTHY AGING

Josep Maria Haro Abad, MD, PhD

Members

Staff researcher

Andra de la Garza Puentes, Albert Sánchez Niubó, Sara Siddi

Assistant researcher

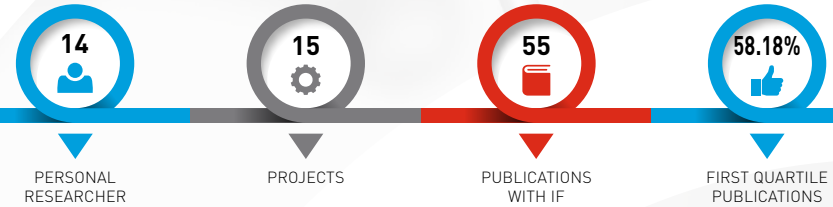
Ivet Bayes Marin, Joan Domènech Abella, Laia Egea Cortes, Iago Giné Vazquez, Federica Lombardini, Blanca Mellor Marsa, Ziggi Santini, Teresa Torres Moral, Elisabeth Catharina Wilhelmina Vorstenbosch, Carla Obradors Tarragó

4

CROSS-SECTIONAL
AREAS

4.4.
Epidemiology

Coordinator:
Josep Maria Haro Abad



Selected projects

- MULTI-MODE III. EIT HEALTH. European Commission. PI: Josep Maria Haro. 01/01/2018-31/12/2018
- Assessment of effectiveness and cost-effectiveness of a programme for prevention, detection and approach of mental disturbances in small and medium enterprises. Projectes Recerca Orientats Salut Mental. Departament de Salut - Generalitat de Catalunya. PI: Josep Maria Haro. 17/02/2018-31/12/2020
- Research on mental disturbances in the immigrant and refugee population: prevalence, associated factors and received treatment. Convocatoria FAMI. Ministerio de Trabajo, Migraciones y Seguridad Social. PI: Josep Maria Haro. 01/01/2018-31/12/2018

Selected publications

- Global Burden of Disease 2016 Injury Collaborators, Naghavi M, Marczak LB, Kutz M, Shackelford KA, Arora M, et al. Global Mortality From Firearms, 1990-2016. JAMA 2018. 320(8): 792-814. FI: 47,661(Q1)
- Shevick SL, Carla Obradors Tarragó and Shenvi RA. Mechanistic Interrogation of Co/Ni-Dual Catalyzed Hydroarylation. J Am Chem Soc 2018. 140(38): 12056-12068. FI: 14,357(Q1)
- Kessler RC, Aguilar-Gaxiola S, Alonso J, Bromet EJ, Gureje O, Karam EG, et al. The associations of earlier trauma exposures and history of mental disorders with PTSD after subsequent traumas. Mol Psychiatry 2018. 23(9): 1-8. FI: 11,640(Q1)
- Degenhardt L, Saha S, Lim CCW, Aguilar-Gaxiola S, Al-Hamzawi A, Alonso J, et al. The associations between psychotic experiences and substance use and substance use disorders: findings from the World Health Organization World Mental Health surveys. Addiction 2018. 113(5): 924-934. FI: 5,953(Q1)

Networks

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

IMPACT, RISK AND PREVENTION OF MENTAL DISORDERS

Ai Koyanagi, MD, PhD

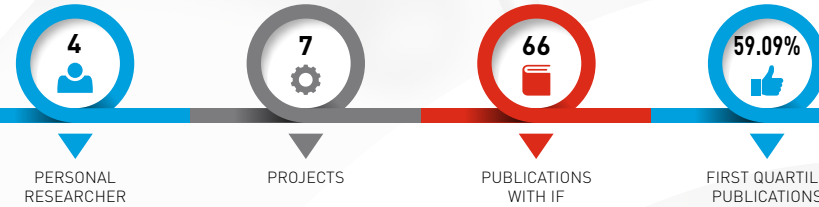
Members

Postdoc - associate researcher
Beatriz Olaya Guzmán
Assistant researcher
Daniel Fernández Martínez,
Maria Victoria Moneta

4 CROSS-SECTIONAL AREAS

4.4. Epidemiology

Coordinator:
Josep Maria Haro Abad



Selected projects

- Sara Borrell_Beneficiari: Hans Oh. Instituto de Salud Carlos III (ISCIII). PI: Ai Koyanagi. 01/01/2018-31/12/2020
- Rio Hortega_Beneficiari: Núria del Cacho. Instituto de Salud Carlos III (ISCIII). PI: Ai Koyanagi. 15/01/2018-02/07/2018
- PFP00133 - Incorporació de científics i tecnòlegs. Departament de Salut - Generalitat de Catalunya. PI: Ai Koyanagi. 17/02/2018-31/12/2020

Selected publications

- DeVylder JE, Kelleher I, Lalane M, Oh H, Link BG and Koyanagi A. Association of Urbanicity With Psychosis in Low- and Middle-Income Countries. *JAMA Psychiatry* 2018. 75(7): 678-686. FI: 16,642(Q1)
- Machado MO, Veronese N, Sanches M, Stubbs B, Koyanagi A, Thompson T, et al. The association of depression and all-cause and cause-specific mortality: an umbrella review of systematic reviews and meta-analyses. *BMC Med* 2018. 16(1): 112-112. FI: 9,088(Q1)
- DeVylder JE and Koyanagi A. Evaluating the Clinical Relevance of Psychotic Experiences in Low- and Middle-Income Countries. *Schizophr Bull* 2018. 44(6): 1167-1169. FI: 6,944(Q1)
- Vancampfort D, Stubbs B, Firth J, Van Damme T and Koyanagi A. Sedentary behavior and depressive symptoms among 67,077 adolescents aged 12-15 years from 30 low- and middle-income countries. *Int J Behav Nutr Phys Act* 2018. 15(1): 73-73. FI: 5,548(Q1)

Networks

- CIBERSAM

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

Antonio Serrano Blanco, MD, PhD

4 CROSS-SECTIONAL AREAS

4.4. Epidemiology

Coordinator:
Josep Maria Haro Abad

This year's highlights

The set of projects on initial medication non-adherence received three distinctions: the prize for the best article on health economy in Spain from the Health Economy Association; the ASISA research grant to expand the study in the paediatric population; and the CAMFIC grant to study initial non-adherence to cardiovascular treatment.

The group —together with CIBERESP's GenRisk consortium— also obtained a research grant from the Spanish Association against Cancer for the study of genetic and metabolic screening to prevent colon cancer. We continued to carry out territorial diagnoses of health services and risk factors in various areas of Barcelona province through Barcelona Provincial Council. We held the 12th edition of the Economic Assessment and Community Healthcare Policy Course, likewise participating in the courses of the School of Public Health in Mahon.

Members

Staff Researcher

Luisa Baldon Higuera, Vanessa Barneda, Laura González Suñer, Maria Iglesias Gonzalez, Cristina López Ortiz, Maria Rubio Valera, Enric Vicens Pons
[Postdoc - associate researcher](#)

Ignacio Aznar Lou, Elisabeth Parody Rua
[PhD student](#)

Gabriela Murugarra Centurión, Carles Vilaplana Carnerero

Assistant researcher

Montserrat Gil Gribau

Collaborator researcher

Ana Fernández Sánchez, Ana Maria Jove Masso, Martin Knapp, Maria Antonia March Pujol, Paul McCrone, Juan Manuel Mendive Arbeloa, Maria Teresa Peñarrubia Maria, Ramon Sabes Figuera, Luís Salvador Carulla



Selected projects

- Modelling of the complex intervention "Initial Medication Adherence" (IMA) in primary care: Non-Adherence phase-I study. Ajuts recerca PSSJD. Parc Sanitari de Sant Joan de Déu - Serveis de Salut Mental. PI: Maria Rubio Valera. 01/02/2018-31/01/2019
- Prevalence and impact of initial non-adherence to pharmacological treatment in the paediatric population: paediatric initial treatment non-adherence study. Proyectos de Investigación en Salud. Proyectos de Investigación en Salud. PI: Maria Rubio Valera. 01/12/2018-30/11/2019
- Intensificació facultatiu. Departament de Salut - Generalitat de Catalunya. PI: Antoni Serrano Blanco. 17/02/2018-31/12/2019
- Genetic and metabolomics screening towards the precision medicine in cancer prevention. Grupos Coordinados Traslacionales. Fundación Científica de la Asociación Española Contra el Cáncer (AECC). PI: Antoni Serrano Blanco. 01/08/2018-31/07/2023

Selected clinical trials & observational studies

- Estudio observacional de postautorización para evaluar la seguridad de ADASUVE® (Loxapina para inhalador Staccato) en personas agitadas en la práctica clínica habitual. Promoter: Alexa Pharmaceuticals. PI: Antoni Serrano Blanco. 16/07/2015-
- 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 Blanco. 30/05/2017-

- Predictors of persistence in patients with schizophrenia treated with once-monthly aripiprazole long-acting injection in the Spanish clinical practice: a retrospective, observational study. Promoter: Promotor Otsuka Pharmaceutical Europe Ltd. PI: Antoni Serrano Blanco. 2017-

Selected publications

- Aznar-Lou I, Pottegård A, Fernández A, Peñarrubia-María MT, Serrano-Blanco A, Sabés-Figuera R, Maria Montserrat Gil Gribau, Fajó-Pascual M, Moreno-Peral P and Rubio-Valera M. Effect of copayment policies on initial medication non-adherence according to income: a population-based study. *BMJ QUAL SAF* 2018. 27(11): 878-891. FI: 7,226(Q1)
- Iglesias Gonzalez M, Aznar-Lou I, Peñarrubia-María MT, Gil Gribau MM, Fernández-Vergel R, Alonso J, Serrano-Blanco A and Rubio-Valera M. Effectiveness of watchful waiting versus antidepressants for patients diagnosed of mild to moderate depression in primary care: A 12-month pragmatic clinical trial (INFAP study). *Eur Psychiatry* 2018. 53: 66-73. FI: 4,129(Q1)
- Aznar-Lou I, Iglesias Gonzalez MM, Gil Gribau MM, Serrano-Blanco A, Fernández A, Peñarrubia-María MT, Sabés-Figuera R, Murrugarra-Centurión AG, March-Pujol M, Bolívar-Prados M and Rubio-Valera M. Impact of initial medication non-adherence to SSRIs on medical visits and sick leaves. *J Affect Disord* 2018. 226: 282-286. FI: 3,786(Q1)

Networks

- 2017 - CIBERESP
- 2006 Red IAPP

Notes

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 2018.

Clinical trials and **observational studies** correspond to all those actives during 2018.

Publications include original articles and reviews published during 2018 (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 2017.

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

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