

1

Introduction

2

Executive
summary

3

Research
areas
and groups

4

Associated
clinicians



3.5.1 INGEMM - Instituto de Genética Médica y Molecular (Institute of Medical and Molecular Genetics)



Publications: 26 | Q1: 17

COMPOSITION

Pablo Daniel Lapunzina Badía.

Facultativo Especialista de Área en Genética. Hospital Universitario La Paz

- **Pedro Arias Lajara.** *Técnico de Laboratorio. Hospital Universitario La Paz*
- **Ángel Campos Barros.** *Investigador Senior (Contrato Miguel Servet- I2). Jefe de Laboratorio. Hospital Universitario La Paz*
- **Luis Fernández García-Moya.** *Biólogo. Hospital Universitario La Paz*
- **María Victoria Fernández Montaña.** *Técnico de Laboratorio. Hospital Universitario La Paz*
- **Fe Amalia García Santiago.** *Bióloga. Hospital Universitario La Paz*
- **Isabel Gómez Nieto.** *Técnico de Laboratorio. Hospital Universitario La Paz*

- **Karen Elise Heath.** *Investigadora Senior (Contrato Ramón y Cajal- I3). Jefe de Laboratorio. Hospital Universitario La Paz*
- **Juan Andrés Jiménez Estrada.** *Investigador Postdoctoral. Hospital Universitario La Paz*
- **Teresa López Timénez.** *Técnico de Laboratorio. Hospital Universitario La Paz*
- **Alicia Llorente Alonso.** *Técnico de Laboratorio. Hospital Universitario La Paz*
- **María Elena Mansilla Aparicio.** *Genetista. Hospital Universitario La Paz*
- **Pilar Martínez González.** *Bióloga. Hospital Universitario La Paz*
- **José Carlos Moreno Navarro.** *Facultativo Especialista de Área en Genética. Hospital Universitario La Paz*
- **Julián Nevado Blanco.** *Biólogo. Hospital Universitario La Paz*
- **María Palomares Bralo.** *Bióloga. Hospital Universitario La Paz*
- **Víctor Luis Ruiz Pérez.** *Investigador Científico de OPIs. Jefe de laboratorio. IIB Alberto Sols*
- **Elena Vallespín García.** *Jefa de Grupo de la Sección de Ofalmogenética Molecular. Hospital Universitario La Paz*

STRATEGIC OBJECTIVES

- This is a basic and clinical research team that carries out several lines of research in early diagnosis, disease mechanisms and new therapeutic strategies for genetic diseases, developmental disorders and congenital defects.
- It is a heterogeneous group composed of medical professionals, pharmacists, molecular biologists, bioinformaticians, technicians and administrators



1 Introduction

2 Executive summary

3 Research areas and groups

4 Associated clinicians

RESEARCH LINES

- Subtelomeric restructurings in patients with idiopathic intellectual disability
- Genetic and functional analysis of the SHOX and SHOX2 genes in human growth
- Overgrowth syndromes. Clinical and molecular analysis
- Genetic determinants of harmonious undergrowth
- Genetic aspects of monogenic diabetes mellitus.
- Genetics of the Ghrelin axis in childhood-onset obesity.
- SNP matrices in pharmacogenetics of HIV patients.
- CGArrays and genomic rearrangements in patients with congenital malformations, intellectual disability and tumors.
- Molecular genetics of hypertrophic cardiomyopathy.
- Functional analysis of mutations in CLCN1 causing congenital myotonia.
- Molecular study of endothelial dysfunction in diabetes and aging.
- Molecular characterization of the 22q11.2 region using MLPA, microsatellites and FISH.
- Pharmacogenetics and Pharmacogenomics
- Autosomal recessive osteogenesis imperfecta.
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- Autosomal recessive osteogenesis imperfecta
- Genomic tools for diagnosis: BAC, oligos and SNP arrays.
- Macrocephaly-capillary malformations and syndromes with macrocephaly.
- Next-generation sequencing as a new tool in the diagnosis of genetic diseases.
- Classification and ontologies of diseases
- Primary pulmonary hypertension
- Global genomic analysis in the invdup(15) syndrome [idic(15)syndrome]
- Molecular characterization of inv dup del (8p) by karyotyping, SNP arrays 2023 and FISH

RESEARCH ACTIVITY

Publications

- Andres-Ibarrola P, Maestro IM, Santamaría AFD, Alfonso CB, Rodríguez ES, Almaraz RL, Merino MJB, Rodríguez-Laguna L, Martínez-Glez V, Gutiérrez JCL, Eckert MM, González-Hermosa MR. Multifocal PIK3CA related congenital hemangioma. *Pediatr Dermatol*. 2025; 42(6): 1198-201. Article. IF: 1.2; Q3
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- Bel-Fenellós C, Biencinto-López C, Orío-Aparicio C, da Silva-Mori X, Tenorio-Castaño JA, Lapunzina P, Nevado J. Family well-being in families with children and young people with Wolf-Hirschhorn Syndrome. *Res Dev Disabil*. 2025; 160: 104974. Article. IF: 2.9; D1
- Blanco-Lago R, Málaga I, Tenorio-Castaño JA, Álvarez-Álvarez N, Lapunzina P, Nevado J. Epilepsy in Wolf-Hirschhorn syndrome: clinical insights from a pediatric cohort and a review of the literature. *J Clin Med*. 2025; 14(22): 8044. Article. IF: 3.3; Q1
- Bruel AL, Vulto-vanSilfhout AT, Bilan F, Le Guyader G, Gilbert-Dussardier B, Le Guillou X, Rondeau S, Rio M, Lee KN, Beil A, Suri M, Guerin F, Ruault V, Goldenberg A, Lecoquierre F, Bertsch N, Anderson R, Yang XR, Inness M, Rikeros-Orozco E,

- Palomares-Bralo M, Hayek JC, Cech J, Jhuraney A, Kumar RD, Mercimek-Andrews S, Ambrose A, Wakeling EN, Wentzensen IM, Torti E, Gooch C, Faivre L, Philippe C, Duffourd Y, Vitobello A, Thauvin-Robinet C. Heterozygous CELF4 variants in the N-term region crucial for the RNA-binding activity lead to neurodevelopmental disorder and obesity. *Eur J Hum Genet*. 2025; 33(7): 852-9. Article. IF: 4.6; Q1
- Cabrera-Alarcon JL, Cruz R, Rosa-Moreno M, Latorre-Pellicer A, de Almeida SD, Riancho JA, Rojas-Martínez A, Flores C, Lapunzina P, Sánchez-Cabo F, Carracedo A, Enriquez JA. Shaping current European mitochondrial haplogroup frequency in response to infection: the case of SARS-CoV-2 severity. *Commun Biol*. 2025; 8(1): 33.. Article. IF: 5.8; D1
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- Guzmán SG, Ruggiero SM, Ganesan S, Ellis CA, Harrison AG, Sullivan KR, Stark Z, Brown NJ, Kana SL, Tuttle A, Tenorio J, Lapunzina P, Nevado J, McDonald MT, Jensen C, Wheeler PG, Stange L, Morrison J, Keren B, Heide S, Keating MW, Butler KM, Lyons MA, Jain S, Yeganeh M, Thompson ML, Schroeder M, Nguyen H, Granadillo J, Johnston KM, Murali CN, Bosanko K, Burrow TA, Morgan S, Watson DJ, Hakonarson H, Helbig I. Variants in BSN, encoding the presynaptic protein Bassoon, result in a distinct neurodevelopmental disorder with a broad phenotypic range. *Am*

3.5.Cancer and Human Molecular Genetics Area

1 Introduction

2 Executive summary

3 Research areas and groups

4 Associated clinicians

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 - Jolitz L, Helbig I, Fitzgerald MP, Ruggiero SM, Cohen S, Angelini C, Vallespin E, Michaud V, Gerasimenko A, Cogne B, Isidor B, Keren B, Dymont D, Heron D, Karstensen HG, Cuppen I, Christodoulou J, Wilson M, Lake NJ, Biskup S, Syrbe S, Mori T, Becker LL, Kaindl AM. Phenotype Spectrum of TRPM3-Associated Disorders. *Ann Neurol.* 2025; 97(3): 561-570. Article. IF: 7.5; D1
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3.5.Cancer and Human Molecular Genetics Area

1 Introduction

2 Executive summary

3 Research areas and groups

4 Associated clinicians

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Research projects

- **Heath KE.** Análisis genético y funcional de las displasias esqueléticas (PID2020-116263RB-I00). MICINN. 2021-2025. **Managment centre: FIBHULP**
- **Moreno Navarro JC.** Biomarcadores de disrupción tiroidea gestacional y daño cerebral fetal en el ratón ko para dehal1 y en mujeres embarazadas y sus hijos. ISCIII. 2023-2026. **Managment centre: FIBHULP**
- **Rodríguez Pérez MR, Pedrosa Delgado M.** Caracterización alérgica y determinación de la relevancia clínica del polen de encina como alérgeno de interés en pacientes policlínicos y su relación con el desarrollo de síndrome de alergia oral con alimentos. ISCIII. 2023-2027. **Managment centre: FIBHULP**
- **Vallespin García E.** Desarrollo e implementación de un protocolo integral combinado con la búsqueda de nuevos genes de cataratas congénitas bilaterales no sindrómicas. Ferrer Internacional S.A. 2020-Ongoing. **Managment centre: FIBHULP**
- **Lapunzina Badía PD.** Dinamización de ciencia disruptiva en predictores de enfermedades tumorales, infecciosas y hereditarias como base de la medicina personalizada (FORT23/00006_GRUPO I). ISCIII. 2024-2027. **Managment centre: FIBHULP**
- **Heath KE.** ERN BOND 2023-2027_European Reference Network on Rare Bone Diseases (BOND2023-2027_Heath). UE. 2023-2027. **Managment centre: FIBHULP**
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- **Palomares Bralo M.** Paving the way for personalized medicine in neonatal and pediatric intensive care units (PI22/01743). ISCIII. 2023-2027. **Managment centre: FIBHULP**
- **Lapunzina Badía PD.** PROTECT CHILD_A PRivacy-prOTecting Environment for Child Transplants health-related and genomic data integration in the European Reference Network. UE. 2024-2028. **Managment centre: FIBHULP**
- **Nevado Blanco J.** Implementación de una red pediátrica para la medicina personalizada en enfermedades raras pediátricas. Un proyecto piloto (PMP22/00008). ISCIII. 2023-2026. **Managment centre: FIBHULP**

Ciber and Retics

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Patents and trademarks

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- **INGEMM, author; FIBHULP, assignee.** Brand name: INGEMM INSTITUTO DE GENÉTICA MÉDICA Y MOLECULAR DEL HOSPITAL UNIVERSITARIO LA PAZ; ESM 2.884.849, USM 77.891.143, CM 8.746.869; 2009 July 16, 2009 December 11, 2009 December 10.



- **INGEMM, author; FIBHULP, assignee.** Brand name: KaryoArray; USM 79.078.751, CM 8.512.907; 2010 January 07, 2009 August 27.



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- **Lapunzina Badía PD, Borobia Pérez AM, Carcas Sansuán AJ, Tenorio Castaño JA, Arias Lajara P, Frías Inieta J, Dapia García I, Muñoz M, authors; FIBHULP, UAM, assignees.** Brand name: ClinPharmarray., CM15489511; 2016 May 31, 2016 September 07.



- **Palomares Bralo M, Vallespin García E, del Pozo Mate A, Santos Simarro F, authors; FIBHULP, assignee.** Brand name: RD-Seq Sequencing Rare Diseases; CM 17.911.029; 2018 May 31.

