



3.7.2 Molecular Hepatology

Publications:9 | Q1:3

COMPOSITION

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- **Luiz Stark Aroeira.** Investigador Postdoctoral. Hospital Universitario La Paz
- **David Vicent López.** Investigador Senior (Contrato Miguel Servet - I2). Hospital Universitario La Paz

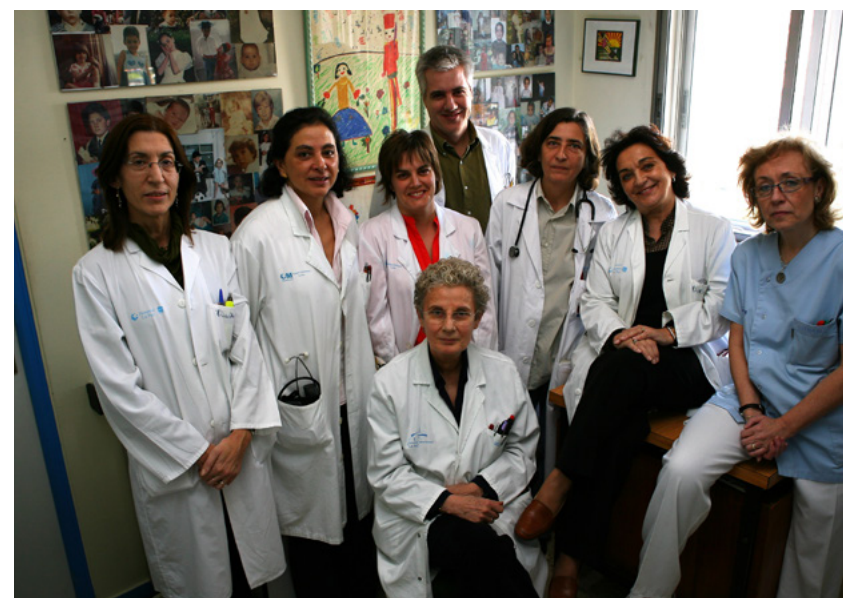
STRATEGIC OBJECTIVE

- Our research interest is focused on the study of the molecular mechanisms underlying the most severe paediatric liver disorders, namely cholestasis, which results from the impaired secretion of bile from the liver to the intestine.
- As such, it represents a clinical and biochemical syndrome that is produced by a wide variety of disease processes that affect the liver. Individuals with cholestasis manifest jaundice, severe itching, malabsorption of fats and lipidsoluble vitamins and, in many cases, progressive liver damage. These clinical manifestations are due to the accumulation in blood and tissues of substances normally secreted in the bile, such as bilirubin, bile acids, and cholesterol and to the absence of bile from the intestine.
- When manifested in early infancy, cholestasis is often life threatening and usually requires liver transplantation. Extrahepatic biliary atresia (EHBA), Alagille syndrome and progressive familial intrahepatic cholestasis (PFIC) constitute the main paediatric cholestatic disorders. EHBA is an enigmatic disease of unknown aetiology, characterised by a precocious and accelerated obstruction of the biliary tree. Alagille syndrome is associated with mutations in the Jag1 gene and is characterised by a paucity or absence of intrahepatic bile ducts. PFIC encompasses a heterogeneous group of autosomal recessive diseases that exhibit similar clinical features. These diseases are caused by mutations in proteins located in the canalicular membrane of the hepatocyte and in proteins involved in bile secretion, such as the bile salt export pump (BSEP; ABCB11), the phospholipid transport protein MDR3 (ABCB4) and the aminophospholipid translocase FIC1 (ATP8B1). These cholestatic disorders constitute the most common indication for liver transplantation in childhood.



3. Information groups by area

3.7 Maternal Infant Child and Youth Research Area



RESEARCH LINES

- Molecular basis of paediatric liver diseases
- Liver Pathobiology
- Biomarker identification

RESEARCH ACTIVITY

Publications

- **Delgado-Miguel C, Triana P, Miguel-Ferrero M, Díaz M, Hierro L, Jara P, López-Gutiérrez JC, Oliveros FH.** Mortality predictive factors in congenital hepatic hemangioma: a case-control study. Eur J Pediatr. 2023; 182(4): 1657-63. Article. IF: 3.0; Q1



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- Hierro Llanillo L. Diagnóstico molecular de enfermedades hepáticas infantiles de carácter hereditario (PI-426). Fundación ACS. 2012-Ongoing. *Managment centre: FIBHULP*
- Hierro Llanillo L. Estableciendo una base de datos como parte de la red europea de referen-

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- Ruiz de Valbuena R, Hierro Llanillo L. A prospective and retrospective cohort study to refine and expand the knowledge on patients with chronic forms of Acid Sphingomyelinase Deficiency (ASMD) (Estudio PIR16183). Sanofi . 2021-. *Managment centre:*
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- Jara Vega P. Support to Generic Services for the ERN TransplantChild (ICT 2372000). UE. 2021-2023. *Managment centre: FIBHULP*

Cibers and Retics

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3. Information groups by area

3.7 Maternal Infant Child and Youth Research Area