Vall d'Hebron Barcelona Hospital Campus | Aula Vall d'Hebron

COURSE

GENETIC CHOLESTATIC LIVER DISEASES:

From Diagnosis to Precision Medicine

26 November 2026

BARCELONA

COURSE DIRECTOR
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Genetic cholestatic liver diseases comprise a heterogeneous group of inherited disorders characterized by impaired bile formation or transport, leading to the accumulation of bile acids within the liver. These conditions are typically caused by pathogenic variants in genes involved in hepatocellular transport and bile secretion—such as ABCB11, ATP8B1, ABCB4, and others—and most follow an autosomal recessive inheritance pattern. They often present in infancy or childhood, although the clinical spectrum can vary widely, ranging from mild, intermittent cholestasis to severe, progressive liver disease. Despite their individual rarity, collectively they represent a significant cause of pediatric cholestasis and an important indication for liver transplantation. Notably, these disorders are increasingly being recognized in the adult population, likely due to greater awareness and advances in genetic testing, which have allowed the identification of milder or previously undiagnosed phenotypes.

Due to their genetic basis and clinical heterogeneity, diagnosis can be challenging and frequently requires a combination of biochemical, histological, and genetic testing. Delayed or missed diagnosis may lead to disease progression, including fibrosis, cirrhosis, and liver failure. Early recognition is therefore essential to enable appropriate management, including medical therapies, nutritional support, and timely referral for advanced treatments when needed. The care of patients with genetic cholestatic liver diseases requires a multidisciplinary approach involving hepatologists, geneticists, nutritionists, surgeons, and transplant specialists to ensure comprehensive and individualized patient management. At Vall d'Hebron Barcelona Hospital Campus, there are over 100 specialist professionals dedicated to the care of more than 2,000 rare diseases. In addition to being the hospital in the country that treats the most rare diseases, it is one of Europe's leading hospitals in this field. In fact, Vall d'Hebron Hospital is part of 20 European Reference Networks (ERNs). This makes us a highly specialized center for treating rare diseases, from birth to adulthood, thanks to a network system that allows for the sharing of resources and knowledge with other top-tier hospitals.

The implementation of a theoretical and practical course on genetic cholestatic liver diseases will equip healthcare professionals with the knowledge and skills required for the early recognition, diagnosis, and management of these rare disorders. Particular emphasis will be placed on advances in molecular diagnosis, genotype–phenotype correlations, and the rapidly evolving therapeutic landscape, including recently approved and emerging targeted therapies. By promoting a multidisciplinary approach, the course aims to improve patient outcomes, facilitate timely referral to expert centers, and foster collaboration among specialists involved in the care of these patients.

COURSE OBJECTIVES

1. Understand the Pathophysiology:
   - o Gain a comprehensive understanding of the molecular and genetic basis of genetic cholestatic liver diseases.
   - o Review the key pathways involved in bile formation and transport, and how their disruption leads to cholestasis.

2. Identify Clinical Manifestations:
   - o Recognize the spectrum of clinical presentations across different age groups, from infancy to adulthood.
   - o Differentiate between major entities such as PFIC and other inherited cholestatic disorders, including their phenotypic variability

3. Diagnostic Approaches:
   - o Understand the diagnostic work-up, including biochemical markers, imaging, histology, and genetic testing.
   - o Explore the role of next-generation sequencing in identifying underlying genetic defects.

4. Treatment and Management:
   - o Review current therapeutic strategies, including medical treatments, surgical approaches, and indications for liver transplantation.
   - o Discuss the role of novel therapies, such as IBAT inhibitors, and emerging treatment options.

5. Case Studies and Clinical Practice:
   - o Analyze representative clinical cases to address diagnostic challenges and management decisions.
   - o Engage in interactive discussions to apply knowledge to real-world patient scenarios.
   - o Discuss multidisciplinary management and transition from pediatric to adult care

COURSE FORMAT

- Lectures: Delivered by experts in the field, covering fundamental and advanced topics.
- Workshops: Hands-on sessions focusing on diagnostic techniques and treatment protocols.
- Case Discussions: Interactive sessions analyzing patient cases to apply theoretical knowledge to clinical practice.
- Research Presentations: Updates on the latest research and advancements in the treatment og Genetic Cholestatic Liver Diseases

TARGET AUDIENCE

- Physicians and healthcare providers in genetics, pediatrics, hepatology, gastroenterology, and metabolic diseases.
- Medical students and trainees interested in genetic disorders.

COURSE FORMAT

The course faculty consists of senior consultants from Vall d'Hebron University Hospital, a tertiary referral centre with extensive expertise in pediatric hepatology, liver transplantation and multidisciplinary management of pediatric liver diseases. All faculty members have substantial clinical and teaching experience in their respective fields, ranging from more than 5 to over 20 years of professional practice.

GENETIC CHOLESTATIC LIVER DISEASE COURSE SCHEDULE

GROUP A:

08.00 - 08.15. Welcome (Dr. Quintero; Floor 11, Room 2, General Hospital)

08.15 - 09.00. Introduction to Genetic Cholestatic Liver Diseases; Dr. Quintero (Floor 11, Room 2, General Hospital)

09.00 - 10.00. How to Perform a Liver Biopsy; Dr. Molino (Multipurpose Rooms; Floor -1, Maternal and Child Hospital)

10.00 - 10.30. Coffee Break

10.30 - 13.30. Practical Modules

Module A: Interpretation of a Liver Biopsy; Dr. Salcedo
(Pathology Building)
Module B: Clinical case presentations; Dr. Mercadal / Dr. Padrós
(Multipurpose Pediatric Day Hospital Floor 0, Maternal and Child Hospital)
Module C: Imaging Tests; Anna Coma
(Department of Diagnostic Imaging, Floor -2, Maternal and Child Hospital)

13.30 - 15.00. Lunch Break

15.00 - 16.00. Biochemical Diagnosis; Dr. Quintero (Hepatology and Liver Transplant Unit, Floor 2, Maternal and Child Hospital)

16.00 - 17.00. Genetic Diagnosis; Dr Irene Valenzuela (Clinical Genetics Unit, Genetics Building)

17.00 - 17.15. Conclusions and Course Closure; Dr. Quintero (Clinical Genetics Unit, Genetics Building)

GROUP B:

08.00 - 08.15. Welcome (Dr. Quintero; Floor 11, Room 2, General Hospital)

08.15 - 09.00. Introduction to the Disease; Dr. Quintero (Floor 11, Room 2, General Hospital)

09.00 - 10.00. How to Perform a Liver Biopsy; Dr. Molino (Multipurpose Rooms; Floor -1, Maternal and Child Hospital)

10.00 - 10.30. Coffee Break

10.30 - 13.30. Practical Modules

Module A: Clinical case presentations; Dr. Mercadal / Dr. Padrós
(Multipurpose Pediatric Day Hospital Floor 0, Maternal and Child Hospital)
Module B: Imaging Tests; Anna Coma
(Department of Diagnostic Imaging, Floor -2, Maternal and Child Hospital)
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Theoretical-Practical Course on Genetic Cholestatic Liver Diseases
Module C: Interpretation of a Liver Biopsy; Dr. Salcedo
(Pathology Building)

13.30 - 15.00. Lunch Break

15.00 - 16.00. Biochemical Diagnosis; Dr. Quintero (Hepatology and Liver Transplant Unit, Floor 2, Maternal and Child Hospital)

16.00 - 17.00. Genetic Diagnosis; Irene Valenzuela (Clinical Genetics Unit, Genetics Building)

17.00 - 17.15. Conclusions and Course Closure; Dr. Quintero (Clinical Genetics Unit, Genetics Building)

Faculty Member	Current Position	Institution
Jesús Quintero Bernabeu, MD, PhD	Head of the Pediatric Hepatology and Liver Transplant Unit	Vall d'Hebron University Hospital, Barcelona
José Andrés Molino Gahete, MD	Consultant Pediatric Surgeon	Vall d'Hebron University Hospital, Barcelona
Cristina Padrós Fornieles, MD	Consultant, Pediatric Hepatology and Liver Transplant Unit	Vall d'Hebron University Hospital, Barcelona
María Mercadal Hally, MD	Consultant, Pediatric Hepatology and Liver Transplant Unit	Vall d'Hebron University Hospital, Barcelona
María Teresa Salcedo Allende, MD	Consultant Pathologist	Vall d'Hebron University Hospital, Barcelona
Anna Coma Muñoz, MD	Consultant Pediatric Radiologist	Vall d'Hebron University Hospital, Barcelona
Irene Valenzuela Palafolls, MD	Consultant Clinical Geneticist	Vall d'Hebron University Hospital, Barcelona

ACCREDITATION

Accreditation has been requested from the **Catalan Council for Continuing Education of the Health Professions – Commission for Continuing Education of the Spanish National Health System**.

A certificate of attendance will be issued to participants who complete **100% of the course**.

TECHNICAL SECRETARIAT

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