

February 2025



Dear members, partners and friends of ERN RARE-LIVER,

It was a pleasure to meet so many of you at our Members' Meeting in Barcelona. Our next Members' Meeting will take place virtually this autumn, more details will be announced soon. The subsequent Onsite Members' Meeting is planned for 13-14 February 2026 in Prague.

We look forward to a productive and collaborative period ahead!

The ERN RARE-LIVER Office

News

Clinical Exchange Programme 2025 & 2026

ERN RARE-LIVER offers the opportunity to apply for and participate in our Clinical Exchange Programme with a focus on rare liver diseases.

This programme is available for Clinicians and other health professionals and is for a maximum duration of 2 weeks. The available periods for the Clinical Exchange start in summer 2025 and in summer 2026. This opportunity is limited to our member centres. For further details, please visit our [website](#) or reach out to us directly via [email](#).

Release of CPMS 2.0 (Clinical Patient Management System)

We are pleased to announce that the latest version of the Clinical Patient Management System, **CPMS 2.0**, is now available for use. Following a comprehensive data migration process conducted by the EC, all essential steps have been successfully completed. CPMS 2.0 is designed to be more user-friendly, faster, and offers additional advantages. For further details, please visit our [website](#). You may access the system [here](#).

Please note that some minor enhancements are still in progress to further improve usability. Should you have any questions or encounter any issues while using the system, kindly reach out to our [ERN RARE-LIVER Helpdesk](#).

Call to host the ERN RARE-LIVER ACADEMY 2026

The call to host the ERN RARE-LIVER ACADEMY 2026 with a focus on paediatric rare liver diseases is now open. Applications can be submitted **until 30 April 2025**, and the organiser is expected to be announced in June 2025. Please visit our [website](#) for further information.

ERN RARE-LIVER Workshops 2026

We would like to encourage Disease Working Groups to develop specific topics by organising short on-site workshops. (Limited) funding is available through ERN RARE-LIVER. Please visit our [website](#) for information on the application process and submit the proposal form by 30 May 2025.

New ERN RARE-LIVER Flyers

We have designed new ERN RARE-LIVER flyers, including flyers about CPMS, the Youth Panel, as well as Patient and Parent representatives. To ensure that these flyers are distributed and reach the right target group, we would like to send you a number of flyers per centre. Please use [this link](#) to indicate the type and number of flyers you would like to receive.

Meetings & Events 2025



The **ERN RARE-LIVER EASL ACADEMY 2025** will be hosted from **10-11 April** by the University Hospital of Heidelberg. The focus of the academy will be on translating clinical practice guidelines into real-life care. For more information, the detailed programme and speakers, please visit our website: [ERN RARE-LIVER ACADEMY](#).



The **EASL Congress 2025** will take place from **7-10 May 2025** in Amsterdam, where ERN RARE-LIVER will have a congress booth. There will also be an **ERN RARE-LIVER Reception**, which is traditionally on the Thursday evening of the EASL conference (**8 May**). Registration options for the reception will be open soon and further information will follow shortly.



ERN RARE-LIVER will also be represented at [ESPGHAN](#) from **14-17 May 2025**. We look forward to meeting and networking with our paediatric members and partners during the conference. Additionally, we plan to have an **ERN RARE-**

LIVER Meeting, which is anticipated to take place on Thursday (**15 May**), the second day of the meeting, at noon. Further updates will be provided in due course.

Publications

AATD



Longitudinal Evaluation of Individuals With Severe Alpha-1 Antitrypsin Deficiency (Pi*ZZ Genotype)

The severe alpha-1 antitrypsin deficiency (AATD) (Pi*ZZ genotype) predisposes to pulmonary loss-of-function and hepatic gain-of-function injury. To improve the clinical management of Pi*ZZ subjects, the authors put together an international, ERN-associated, multicenter, longitudinal Pi*ZZ cohort evaluating the natural disease course and surrogates for future liver- and lung-related endpoints. During 2634 follow-up years, 39 individuals died, with liver and lung being responsible for 46% and 36% of deaths, respectively.

Liver-related endpoints within five years were most accurately predicted by liver stiffness measurement (LSM) (area under the curve [AUC] 0.95) followed by APRI (0.92). Baseline lung parameters displayed only a moderate predictive utility for lung-related endpoints within five years (FEV1 AUC 0.76). Fibrosis progression in those with no/mild fibrosis at baseline was rare and primarily seen in those with preexisting risk factors. Overall, the comparison of liver and lung surrogated should facilitate patient management as well as selection into clinical trials that typically target only one organ.

Study to Check the Safety of Fazirsiran and Learn if Fazirsiran Can Help People With Liver Disease and Scarring (Fibrosis) Due to an Abnormal Version of Alpha-1 Antitrypsin Protein

There is currently no approved treatment available for AATD – associated Liver Disease (AATD-LD).

Promising Phase 2 results have been previously published and support further clinical development of fazirsiran.

This clinical trial, known as The Redwood Study, is a Phase 3 randomized, double-blind, placebo-controlled study, evaluating the safety and efficacy of fazirsiran in adults with Alpha-1 Antitrypsin Deficiency-associated Liver Disease (AATD-LD) and the Pi*ZZ genotype. The study involves 11 ERN member sites and aims to determine the impact of fazirsiran on liver health and any potential side effects.

AIH



Quo vadis autoimmune hepatitis? - Summary of the 5th international autoimmune hepatitis group research workshop 2024

Autoimmune Hepatitis (AIH) is a rare chronic autoimmune liver disease. Experts within the field of AIH research gathered in June 2024 to identify research gaps and opportunities and set an agenda for current AIH research. The current report highlights the outcomes of the research workshop. Four key areas of interest were identified, namely the application of artificial intelligence to existing data sets to improve diagnosis and outcome prediction, the identification and validation of new/existing diagnostic markers, the evaluation of chimeric antigen receptor (CAR) regulatory T cell therapies in patients with difficult to treat AIH as well as the definition of core outcome sets of clinical trials in AIH and the improvement of understanding of the diseases' pathogenesis by leveraging state-of-the-art technologies like spatial transcriptomics and single cell sequencing techniques.

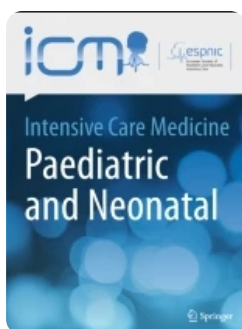


Update in Clinical Science: Autoimmune Hepatitis

Autoimmune hepatitis (AIH) is a complex disease with a wide range of presentations. Its pathogenesis, diagnosis, and management pose significant challenges. This review

focuses on developments from 2020 onwards, aiming to provide an updated perspective on the challenges and unanswered questions that are essential for improving patient care and outcomes in AIH. Areas of ongoing debate are also highlighted, offering insights into these discussions..

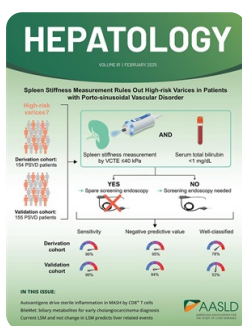
PALF



The management of paediatric acute liver failure: results from an international multi-centre survey across Europe

In this collaboration between paediatricians in the ERN-RARE-LIVER and ESPNIC networks, the authors invited paediatric liver transplant centres across Europe to reply to a survey about paediatric liver failure. The results highlighted varied aetiologies in different settings and variations in key aspects of management for patients with PALF. The paper concludes that international collaboration is essential to standardise guidelines, further improve equity of access to treatment, and, lastly, to foster research collaborations to improve outcomes for this high-risk patient group.

VLD



Performance of spleen stiffness measurement to rule out high-risk varices in patients with porto-sinusoidal vascular disorder

Current guidelines recommend screening for high-risk varices with endoscopy in all patients with porto-sinusoidal vascular disorder (PSVD). In this retrospective study (derivation cohort from Beaujon Hospital: n=154; validation cohort from 21 VALDIG centers: n=207), the authors observed that in patients with PSVD and ≥ 1 sign of portal hypertension, without a history of variceal bleeding, spleen

stiffness measurement (SSM) by vibration-controlled transient elastography (VCTE) is strongly associated with high-risk varices. A model combining SSM by VCTE ≤ 40 kPa and total serum bilirubin < 1 mg/dL identifies patients with a very low probability of having high-risk varices ($< 5\%$), in whom screening endoscopy can be spared.

Surveys



[Dietary habits of individuals living with PSC - ERN RARE-LIVER Survey for Patients](#)

[Wilson Disease and Pregnancy - ERN RARE-LIVER Survey for Physicians \(Deadline extended\)](#)

[Wilson Disease and Pregnancy - ERN RARE-LIVER Survey for Patients \(Deadline extended\)](#)

[Barriers to sexual function in patients with liver disease and Ltx - Survey for Physicians](#)

Webinars

04/03/2025

Novel developments in **alpha1-antitrypsin deficiency-associated liver disease**

18/03/2025

Main points to remember from **EASL Vascular liver disease guidelines**

15/04/2025

Diagnostic pitfalls and differential diagnosis of **Wilson Disease**

If you are interested to host a webinar or take part as a speaker, please contact Juan Carlos García-Pagán who is the lead of the Training and Education Committee of ERN RARE-LIVER (JCGARCIA@clinic.cat).

Patient Representatives

ERN RARE-LIVER Patient Representatives at the Members' Meeting in Barcelona

Five of our Patient Representatives (ePAGs) participated in the Members' Meeting held in Barcelona. Their involvement included contributions to the plenary session, with Wiebke Papenthin (Morbus Wilson e.V.), as well as active participation in the Disease Working Group meetings. Additionally, Gema Iribar (ALBI Spain) and Aida Regi Cosculluela (AEFE de Wilson) presented details on projects led by their respective patient organisations and shared valuable perspectives.

For more information on their projects, please click [here](#).





“Talk to Heal” - Project at the Paediatric Hospital of Coimbra

The “Talk to Heal” project aims to address various topics related to managing specific pathologies, fostering communication between patients of different ages and experiences. Therefore the Hepaturix Association in Portugal is planning a series of “Talk to Heal”-Events in 2025, in which the Paediatric Hospital of Coimbra and members of our ERN RARE-LIVER Youth Panel are involved. Please check our website for further dates.

Former ERN RARE-LIVER ePAG Lead on the ERDERA Multistakeholder Advisory Board

We are very happy for José Willemse, former lead of the ERN RARE-LIVER Patient Representatives, who has been selected to be part of the ERDERA Multistakeholder Advisory Board. The mandate will be for an initial period of 3 years. Congratulations!



Visit us on our website



ERN RARE-LIVER

University Medical Centre Hamburg-Eppendorf, Martinistraße 52, 20251, Hamburg
Sie haben die E-Mail erhalten, weil Sie sich für den Newsletter angemeldet haben.

[Im Browser öffnen](#) | [Abbestellen](#)