

Half of all young children with a rare inherited liver disease need a liver transplant

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Pediatric liver disease. Credit: The European Society for Paediatric Gastroenterology Hepatology and Nutrition

An international research team has today reported the first results of a study investigating the natural history of progressive familial intrahepatic cholestasis (PFIC)—a rare genetic liver disease that predominantly affects children. Most alarmingly, the team reported that, by the age of



10 years, approximately half of the children with two different forms of PFIC had already received a liver transplant.

The study presented today at the 51st Annual Congress of the European Society for Paediatric Gastroenterology Hepatology and Nutrition, was undertaken by the NAPPED (NAtural course and Prognosis of PFIC and Effect of biliary Diversion) Consortium—an international study group whose aim is to characterise globally the natural history of two types of PFIC: PFIC1 (FIC1 protein deficiency) and PFIC2 (BSEP protein deficiency). These conditions result from inherited genetic mutations that lead to impaired bile flow through the liver (cholestasis), resulting in the accumulation of bile, progressive liver damage and, potentially, end-stage liver disease and death2. Children with PFIC may benefit from medical treatment with ursodeoxycholic acid (UDCA) or from surgical biliary diversion techniques, however, many children require a liver transplant at a young age.

"PFIC1 and PFIC2 are very rare conditions and, because of this, very little is known about their <u>natural history</u>, the impact of different genetic mutations on phenotype, and the overall effectiveness of the <u>treatment</u> options available," said Daan van Wessel from the University Medical Center Groningen in The Netherlands, who presented the study findings in Geneva today. "The NAPPED Consortium, which involves 28 specialist centres across the world, hopes to better define the natural course of disease, the outcomes of different treatment options, and, hopefully, find biomarkers that can help target treatment for these conditions more effectively."

The study involved a retrospective analysis of data from 42 children with PFIC1 and 184 children with PFIC2. Children in the PFIC1 group attended their first specialist centre at a median age of 6 months (range: 0 months to 201 months); and 33% of these had already received UDCA treatment. Children in the PFIC2 group attended their first specialist



centre at a median age of 9 months (range: 0 months to 195 months) and 47% had received UDCA. By the age of 5 years, 27% and 36% of children with PFIC1 and PFIC2, respectively, had already received a liver transplant and, by 10 years of age, 49% and 52% of children, respectively, had been transplanted.

The results of this study also shed important new light on the benefits of surgical biliary diversion in children with PFIC—a procedure used to reduce the amount of bile entering the liver. Although the procedure did not appear to reduce the need for liver transplantation in children with PFIC1, among those with mild or moderately severe PFIC2 mutations, surgical biliary diversion led to a 63% increase in the percentage of children surviving with their own livers compared with those who did not have the procedure (hazard ratio: 0.37; 95% confidence interval: 0.24-0.68; p=0.001).

"This study has demonstrated for the first time that surgical biliary diversion is a highly effective treatment option for children with mild or moderate PFIC2 mutations. Present data on PFIC1 are still inconclusive, but we expect to assess the value of surgical biliary diversion for PFIC1 shortly, based on the ongoing collection of patients' data" said Daan van Wessel. "We hope this information will help healthcare professionals to inform the children and their parents about their possible prognosis and enable clinicians to select treatment options more rationally based on the evidence. The current data also provide strong support for the recent development of oral drugs that accomplish similar effects as surgical biliary diversion" he adds.

Provided by The European Society for Paediatric Gastroenterology Hepatology and Nutrition

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