

# New genetic cause of children's liver disease discovered

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(Medical Xpress)—The discovery of a 'faulty gene' in children with liver disease could pave the way for new treatments for children with a range of serious and life-threatening liver conditions.

The findings – published on Sunday, 9 March in *Nature Genetics* – follow two years of research by doctors and scientists at King's College London and King's College Hospital, both part of King's Health Partners AHSC.

Cholestatic liver disease (CLD) describes a number of conditions caused by impairment of bile formation, or bile flow. Normal bile production is essential for absorption of food and the body's ability to dispose of waste. CLD can be fatal, and many patients require [liver transplantation](#). At least 12,000 people suffer from CLD in the UK.

Using the latest gene sequencing technology, the team at King's discovered that 12 patients with CLD had a mutation in both copies of the TJP2 gene, which means it doesn't produce the TJP2 protein.

This discovery of a new disease, called 'TJP2 deficiency', is a significant breakthrough. Crucially, it now means liver experts understand the mechanism underlying the disease, and can start treatment earlier and in a targeted fashion.

Dr Richard Thompson, a paediatric liver specialist at King's who led the research, said the findings should increase our understanding of the way other types of liver disease develop. He said:

'This is extremely exciting. By understanding the disease better, we are a step closer to one day finding a cure. In the short-term, it also makes the disease much easier to diagnose –we have gone from basic science discovery to routine diagnostic testing in less than a year, which is amazing. Crucially, this means we can start logical treatment for the disease sooner. The basic principles of the discovery could also be applied to other, more common forms of liver disease, which is also very exciting.'

King's, a major centre for the treatment of adult and paediatric [liver disease](#), is one of three centres currently trialling a new drug for the treatment for CLD in children.

**More information:** "Mutations in TJP2 cause progressive cholestatic liver disease." Melissa Sambrotta, et al. *Nature Genetics* (2014) [DOI: 10.1038/ng.2918](#). Received 21 September 2013 Accepted 14 February 2014 Published online 09 March 2014

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