# Wilson disease: Genetic mutation "protects" against fatty liver and ensures better prognosis

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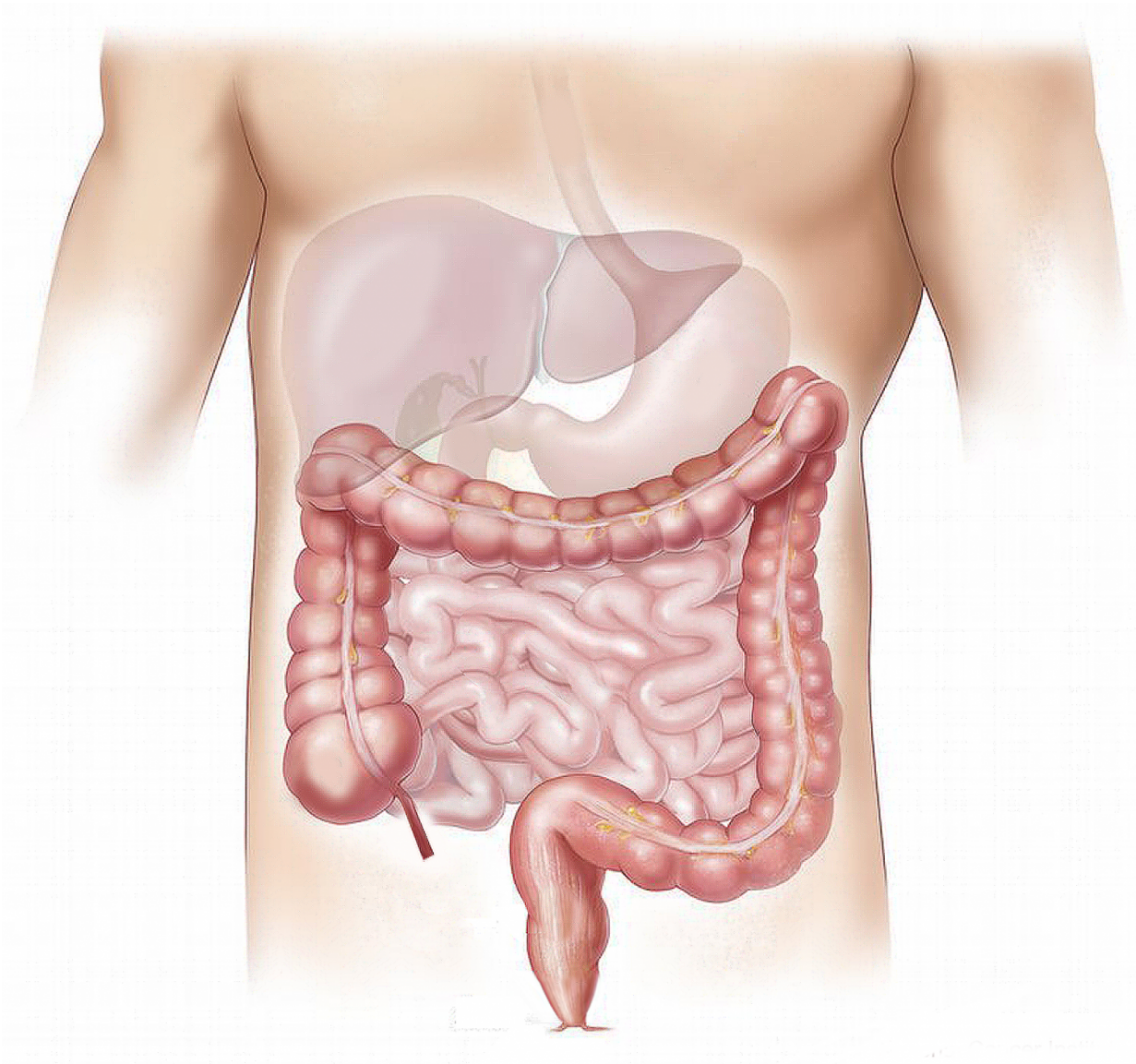

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Wilson disease (copper storage disease) is a rare genetic disorder in which one or more genetic mutations disrupt copper metabolism in the liver. At some point, the liver becomes incapable of eliminating copper with bile, and copper accumulates in the liver, eyes and central nervous system. This results in serious liver damage and neurological problems inter alia. MedUni Vienna researchers led by hepatologist Peter Ferenci have now discovered that a recently described genetic mutation protects against fatty liver – apparently via vitamin A metabolism –and also ensures a better outcome for Wilson disease patients. In future, this finding could prove very useful in the personalised treatment of such patients.

The gene that has been identified is "HSD17B13," which, according to Ferenci, plays an important role in vitamin A metabolism. Follow-up studies are planned to establish whether Wilson disease patients could benefit from this knowledge and potential treatment through the administration of vitamin A. Overall, this genetic mutation was found in every fourth person (around 26 percent). "We can assume that the HSD17B13 gene plays a critical role in the progression of Wilson disease," explains Ferenci. "If patients do not have this mutation, their prognosis is poorer. So, in the spirit of precision medicine, we are able to predict much more accurately how the disease will progress.

In a highly regarded study published in *Hepatology* in 2018, the researchers showed that the protein ATP7B, known as the Wilson disease gene, and of which there are hundreds of mutations, did not provide any definitive information regarding prognosis.

Wilson disease can go undetected for years, and is usually discovered by

accident. The age range is huge: The youngest known case in Vienna is a two-year-old child and the oldest patient was 74 years old. Even the symptoms are diverse: The disease can be symptomless for a long time or manifest in the form of severe liver damage, Kayser-Fleischer rings in the eyes (corneal changes) and neurological problems including movement disorders such as twitching or tremor in the limbs or even slurred speech and difficulty in swallowing. "In addition to that, there are often psychiatric disorders such as compulsive behaviours through to psychosis," adds Ferenci.

**More information:** Peter Ferenci et al. Age,sex, but not ATP7B genotype effectively influences the clinical phenotype of Wilson disease, *Hepatology* (2018). DOI: 10.1002/hep.30280

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