

Research team discovers gallstone gene

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Scientists at the University of Bonn, together with colleagues from Romania, have discovered a gene variant that significantly increases the risk of developing gallstones.

It is estimated that one in ten Europeans has this variant in their hereditary disposition. For those affected, the likelihood of developing a gallstone in the course of their life is two to three times higher. The relevant gene contains the instructions for building a molecular pump which transports cholesterol from the liver into the bile ducts – cholesterol being the substance from which most gallstones are formed. The genetic modification appears to cause this pump to work permanently at high speed. Gallstones are a common disorder: in Germany alone more than 170,000 gallbladder operations a year are performed.

Gallstones occur very frequently, affecting 15 to 20 per cent of all Germans, usually without noticeable symptoms. But in around a quarter of cases the stones will announce their presence at some time in the form of a painful colic. In the end they often have to be operated. "Gallstones are among the gastrointestinal problems that incur the highest treatment costs," says Professor Dr. Frank Lammert, the Bonn-based specialist for internal medicine.

Gallstones tend to be found at high levels within certain families. In particular, studies of twins provide evidence of a genetic component that boosts risk levels. "We reckon that environmental influences, like the wrong diet, are 70 to 80 per cent responsible for the disorder," explains

Lammert who works in the university hospital's Department I headed by Professor Dr. Tilman Sauerbruch. "The rest is caused by genes."

Professor Lammert – working together with his colleagues Dr. Frank Grünhage, Maja Walier and Professor Dr. Thomas Wienker as well as scientists at the University Clinic of Cluj-Napoca in Romania – has been searching for the specific genes involved. And he has succeeded, thanks to a study covering 178 women and men from 84 families. They all suffer from gallstones. In 21.4 per cent of cases the subjects were found to be carrying a particular gene variant. In healthy individuals studied as a control group, this variant also occurred, but only at a frequency of 8.6 per cent.

Cholesterol pump at full speed

"The mutation concerns what is known as the ABCG8-gene," Dr. Grünhage explains. "It contains the instructions for building a pump responsible for transporting the blood lipid cholesterol from the liver into the bile ducts." Most of the gallstones consist to a high degree of crystallised cholesterol. The medical researcher concludes that, "The genetic alteration probably makes the pump run permanently at high speed."

The researchers now hope that their finding will have positive consequences for prevention and therapy. Professor Lammert thinks that, "It may be possible for certain patients to be helped with drug treatments in future, thus avoiding the need for an operation." However, the genetic contribution to the common problem of gallstones has not been fully explained by this study: "We believe there are at least three or four other gene variants that increase gallstone risk," says the medical scientist.

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