



Microvillus atrophy

This information sheet from Great Ormond Street Hospital (GOSH) explains the causes, symptoms and treatment of microvillus atrophy and where to get help.

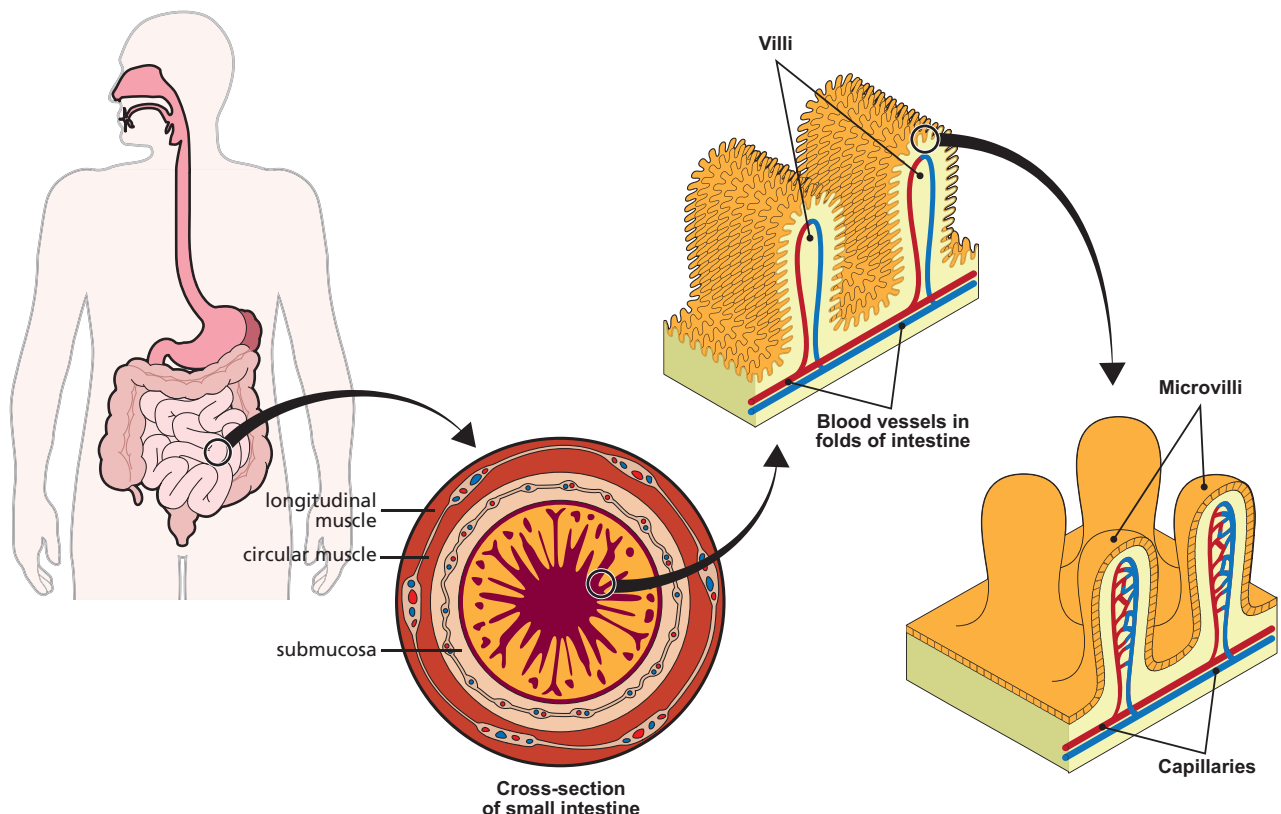
What is microvillus atrophy?

Microvillus atrophy is a life threatening condition causing severe diarrhoea in the first few days or weeks after birth. It is caused by an abnormality in the cells in the small intestine that make it impossible for them to absorb any fluid or nutrients from food.

The gastrointestinal (GI) tract is a complex organ that extends as a hollow tube from the mouth to the anus. Its main function is to break down food so it can be absorbed into the bloodstream. The process of moving

food through the GI tract involves a complex interaction between hormones (chemical messengers) and nerves, so that food is squeezed rhythmically through the system (peristalsis).

Once food has been processed in the stomach, it empties into the small intestine where the majority of digestion and absorption occurs. Here it is mixed with bile and pancreatic juice containing enzymes (proteins that cause or speed up a chemical reaction).





The broken down nutrients are then small enough to pass through the wall of the small intestine, which contains small finger-like structures called villi, and then absorbed by the blood. The blood is carried away from the small intestine through the hepatic portal vein to the liver, where it is filtered, toxins are removed and the nutrients are processed. The residue then passes into the large intestine, where water is absorbed to form solid faeces (poo).

What causes microvillus atrophy?

Normally, each cell in the small intestine is covered with tiny finger-like structures called villi and microvilli. These increase the surface area of the cell so improve the rate of absorption. The cells of a child with microvillus atrophy do not have microvilli, which means that fluid and nutrients cannot be absorbed by the small intestine.

Microvillus atrophy was first described in 1978 with less than 200 cases identified in the world to date. In 2008, the faulty gene causing most cases of microvillus atrophy was identified as the MYO5B gene. Microvillus atrophy is passed on in an autosomal recessive manner, that is, both parents have to have the faulty gene and there is a 1 in 4 chance with each pregnancy that the child will have the condition. Microvillus atrophy affects twice as many boys than girls.

What are the signs and symptoms of microvillus atrophy?

There are two types of microvillus atrophy: congenital and late-onset. The symptoms of both types are the same but the age at which they appear is different. In congenital microvillus atrophy, a baby develops severe diarrhoea within hours of birth. This continues whether or not the baby has a feed. Severe diarrhoea appears later in late-onset microvillus atrophy, usually around six to eight weeks after birth. The diarrhoea can be life-

threatening as it leads to severe dehydration and imbalanced mineral and salt levels.

How is microvillus atrophy diagnosed?

Initially stool (faeces or poo) samples may suggest a diagnosis of microvillus atrophy but the definitive test is an intestinal biopsy (tiny tissue sample). However, as the microvilli are so tiny, the sample needs to be examined using an electron microscope to confirm the diagnosis.

How is microvillus atrophy treated?

The initial task is to re-hydrate the baby by infusing fluid directly into the bloodstream through an intravenous cannula (thin plastic tube) inserted into a vein. The fluid usually contains minerals and salts to correct these levels as well.

Once the baby is stabilised, intravenous feeding (total parenteral nutrition or TPN) is the only way for the baby to receive nutrients as in most cases oral (by mouth) feeding will be impossible. A liquid solution that contains nutrients (vitamins, minerals, carbohydrates, proteins and fats) needed for growth and development is given directly into a vein through a central venous catheter. The catheter will need to be inserted in a short procedure with a general anaesthetic. Long-term TPN can have side effects, such as complications associated with the central venous catheter, so may not be suitable for everyone.

The only 'cure' for microvillus atrophy is a small bowel transplant. This involves replacing the damaged small intestine with a donated one. This procedure is still fairly new in the UK but one that is offered jointly between GOSH and King's College Hospital in South London. After a small bowel transplant, children are able to eat by mouth, building up to a normal diet and feeding regime over time.



What happens next?

Unfortunately, microvillus atrophy cannot be treated in every child and those who do survive may have long term problems, such as developmental delay due to the initial period of dehydration before diagnosis and treatment. The complications associated with long-term TPN are also problematic for some children. However, as small bowel transplant becomes more commonplace, outcomes are improving all the time.

Further information and support

The Daniel Courtney Trust – Making it better is a UK-based support group for anyone affected by microvillus atrophy. Visit their website at www.makingitbetter.org.uk for further details.