



Autoimmune enteropathy

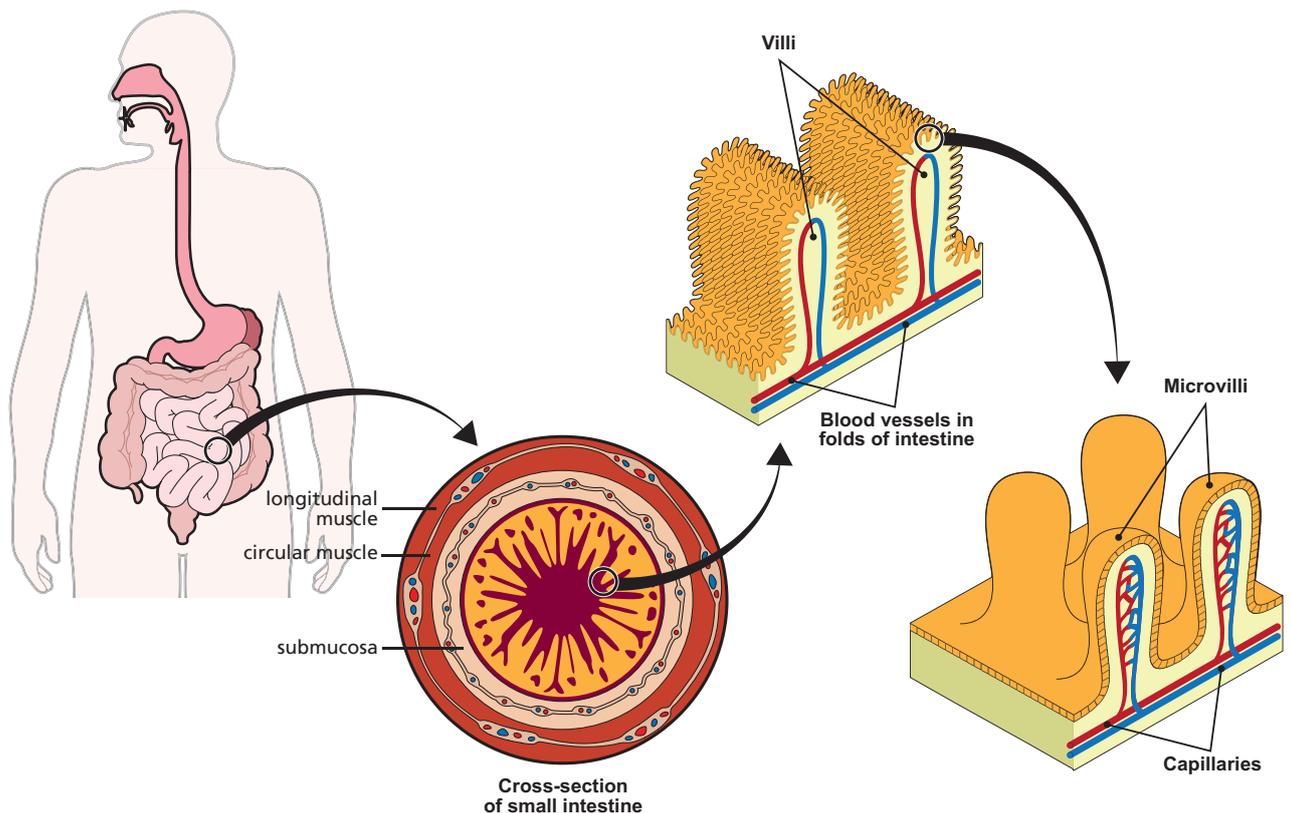
This information sheet from Great Ormond Street Hospital (GOSH) explains the causes, symptoms and treatment of autoimmune enteropathy and where to get help.

What is autoimmune enteropathy?

Autoimmune enteropathy is a rare condition affecting young babies causing severe long-lasting diarrhoea. It is caused by the immune system becoming mis-programmed, activating immune cells to attack the lining mainly of the small intestine, but it may also affect the stomach and the large intestines. This damages the cells so that the lining becomes inflamed and goodness from food cannot be absorbed. This leads to poor weight gain and growth.

What causes autoimmune enteropathy?

Autoimmune enteropathy is thought to be an autoimmune disorder although further research is needed to prove the actual mechanism of how the condition develops. Autoimmune disorders occur when the body mistakenly attacks itself rather than a foreign invader such as a bacterium or virus. We do not know what triggers this mis-programmed immune response. In some cases of autoimmune enteropathy a genetic cause can be found.





What are the signs and symptoms of autoimmune enteropathy?

The main sign is severe diarrhoea developing usually within the first six months of life. The diarrhoea develops regardless of the feed given and may lead to dehydration in a short time. Over time, the malabsorption of goodness from food leads to poor weight gain and growth problems. Frequently, children with autoimmune enteropathy can be affected by other autoimmune disorders and this will be checked with a blood test.

How is autoimmune enteropathy diagnosed?

The doctor will usually start by taking a clinical history of which symptoms are present and when they developed. A number of tests and procedures will be needed to rule out more common causes of diarrhoea. Samples of stool will be examined in a laboratory to exclude other causes of diarrhoea. Small samples of tissue – biopsies – will also be taken from various locations in the intestines as well. The biopsies will usually show destruction of the villi and areas of sore tissue. Blood tests will also be needed to look at the child's immune function and nutritional status.

How is autoimmune enteropathy treated?

If dehydration has developed, 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) may be the only way for the baby to receive nutrients as in most cases oral (by mouth) feeding may 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.

Medicines to reduce the autoimmune reaction will be needed. In many cases, this will include steroid medicines and immunosuppressant medicines that damp down the immune system so that the body no longer attacks itself. In many children, these medicines will have to be taken on a long term basis, but regular blood tests will monitor for any side effects and ensure that the minimum dose needed is given. Damage to the intestines is usually monitored with regular endoscopies.

What happens next?

The outlook is variable, depending on the severity of symptoms. Children with autoimmune enteropathy need nutritional support and adequate hydration to ensure optimal growth and development. In some severe cases, TPN may be required long term. Immunosuppression therapy has been used successfully but in more severe cases, a bone marrow transplant may be required.

Further information and support

There is no dedicated support group for autoimmune enteropathy but the umbrella organisation **Contact a Family** may be able to put you in touch with another family affected by the same or similar condition. Call them on 0808 808 3555 or visit their website at www.cafamily.org.uk

PINNT (Patients on Intravenous and Naso-gastric Nutrition Therapy) is the support group for anyone using TPN. Call them on 01202 481 625 (9.30am - 4.30pm, answerphone at other times) or visit their website at www.pinnt.com.

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