Tufting enteropathy

Tufting enteropathy (also known as intestinal epithelial dysplasia) is a very rare congenital (present at birth) condition affecting the inner surface of the intestines. It causes severe life threatening diarrhoea in the first few days after birth.

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 tufting enteropathy?

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. Children with tufting enteropathy may be missing some of the microvilli and where microvilli are present, the cells on the surface may be arranged to form tufts rather than being smooth.

Doctors think that there is a genetic component to tufting enteropathy, probably caused by mutation (change) to a specific gene. This mutation can be passed on from parent to child but in many cases develops sporadically (out of the blue). If it is inherited, it is passed on as an autosomal recessive condition. This means that a child has to inherit the faulty gene from both parents to develop the condition. The gene causing the disorder has now been identified so prenatal (before birth) diagnosis may be possible.

Tufting enteropathy is a very rare condition, affecting one child in every 50,000 to 100,000. It seems to be more common in certain races, particularly those of Arabic descent.

What are the signs and symptoms of tufting enteropathy?

The main symptom is that a baby develops severe diarrhoea within hours of birth. This continues whether or not the baby has a feed. The diarrhoea can be life-threatening as it leads to severe dehydration and imbalanced mineral and salt levels.

How is tufting enteropathy diagnosed?

Once a baby has been stabilised with intravenous fluids, samples of diarrhoea 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. As the microvilli are so tiny, the sample needs to be examined using an electron microscope to confirm the diagnosis.

How is tufting enteropathy 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 tufting enteropathy 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. However, as a transplant is a potentially risky procedure, it is currently only offered to children who have developed complications as a consequence of long term TPN and would not be done as first line treatment.
What happens next?

Unfortunately, tufting enteropathy 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 and many children with tufting enteropathy have good quality of life on TPN given at home.