

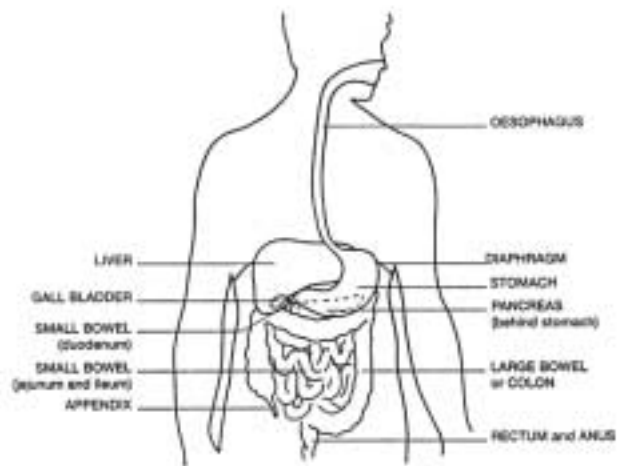
# Gastrointestinal problems in children with Down's syndrome

*by Dr Liz Marder*

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## The gastrointestinal tract

The gastrointestinal tract comprises the parts of the body involved in taking in food, processing it to make use of the nutrients and disposing of the solid waste. It includes the mouth, the oesophagus, stomach, small intestines or bowel (duodenum, jejunum, ileum), the large intestines (colon, rectum) and the anus - see diagram. The liver and pancreas are attached to the gastrointestinal tract and produce enzymes which help with the digestion of food.



Problems with the gastrointestinal tract can either be due to abnormal structure i.e the organs are formed differently from usual, or may be because part of the tract is not functioning properly. Children with Down's syndrome are more likely to have problems in both of these areas than the general population. Some of these problems are serious and life threatening and are likely to cause immediate problems in the new born period. Other problems may not be so dramatic, but nevertheless cause considerable problems. In some of the conditions, problems will develop slowly and may not be picked up by parents or health professionals for some time.

Gastrointestinal problems are a common cause of illness in all children. The more common problems include gastroenteritis (an infection affecting the stomach and intestines) and appendicitis. Children with Down's syndrome may get any of the problems that occur in other children. If a child with Down's syndrome does seem to have a problem relating to the gastrointestinal tract it is important that they have a medical assessment which takes into account both the common conditions that may affect any child as well as those that particularly affect children with Down's syndrome. In this article I am only going to describe some of the problems that are more common in Down's syndrome.

## **Symptoms of gastrointestinal disorders**

Problems arising within the gastrointestinal tract may cause a number of different symptoms.

### **Vomiting**

All children vomit from time to time. In babies this may just be a small amount of undigested milk that is brought back after a feed, sometimes with wind. This is called possetting and is not usually a sign of underlying disease. It can often be helped by careful attention to feeding technique, avoiding taking in too much air and keeping the baby upright after feeds for about half an hour. Children may also vomit when they have a generalised illness - eg flu, chicken-pox or other viruses - , ear or urine infections, when travelling, or if they are very upset or excited. In these situations the vomiting is usually short lived and goes away when the underlying problem is resolved. If vomiting is severe, frequent (i.e. more than a few times a day) or prolonged (lasting more than a few days) you should seek the advice of a doctor. Other signs that the problem is more serious include bile stained vomit, or blood in the vomit. (This may look dark brown or like coffee grounds mixed in the vomit.)

### **Diarrhoea**

This is when there is frequent passage of loose stools. It is impossible to specify how loose, or how often is abnormal, as every person is different. Many infants and toddlers (and some adults) will pass several loose stools everyday as their normal pattern. There will also be normal healthy variation depending on what we eat and drink. The important thing is to notice if there is a significant change from the child's normal pattern that lasts more than a few days. Sometimes stools may be particularly bulky, foul smelling or look frothy or greasy. This may be a sign that something is not being absorbed properly in the diet.

### **Constipation**

Most people get constipated from time to time. Again it is impossible to say what is the normal frequency for passing stools as there is a lot of variation in healthy people. Some people open their bowels several times a day and others only once or twice a week. It is the change from their usual pattern that may be important. Very hard stools can be a problem in themselves as they can be painful to pass and cause a small amount of bleeding on the way out. Sometimes constipation is not identified because the child is continuing to pass small amounts of liquid stools. This happens when hard dry stool is retained in the rectum, while small amounts of liquid stool seep around the sides, often resulting in soiling. This is called constipation with overflow.

Constipation is a common problem in Down's syndrome, and in most cases it won't be due to underlying disease. It may be due to a combination of low muscle tone, poor mobility, diet and inadequate fluid intake. Giving extra fluid, fruit and cereals may solve the problem, or it may be necessary for the doctor to prescribe a stool softening laxative. If constipation is severe or persists despite these measures, then other causes should be considered. Underactivity of the thyroid gland (hypothyroidism) can cause constipation, and is more common in Down's syndrome. A blood test can be done to check on this. Hirschprung's disease also causes constipation and is discussed below.

## **Pain**

Children often complain of tummy ache. In many cases it is nothing to do with the gastrointestinal tract but is a general symptom of being unwell, or it may be the child's way of explaining a pain elsewhere in the body. It may also be a symptom of anxiety. Most tummy aches disappear after a few hours and a few cuddles! It is often caused by constipation, and may be relieved by opening the bowels. Advice should be sought from a doctor if it persists, is associated with other symptoms such as vomiting, or if severe.

Pain may also be related to the oesophagus (heartburn or indigestion). This is likely to be due to reflux (see below) It is most likely to occur in babies who are unable to tell us they are in pain, so is always worth thinking about in a baby who seems in discomfort, especially after feeds.

## **Poor weight gain**

This can be a symptom of many childhood disorders. It is often associated with gastrointestinal problems. The other symptoms above may not be very serious, but should always be considered more seriously if associated with poor weight gain, or weight loss.

Children with Down's syndrome do grow slowly when compared with other children. There are now charts available specifically for children with Down's syndrome. Like all children there is considerable variation in size and weight so a child's actual weight is not as important as the rate of weight gain or the comparison with previous weight or height measurements for that child.

## **Structural problems**

Around 10% of children born with Down's syndrome will have one of these problems. Not all the problems that occur are discussed, but these are a few of the main examples.

### **Small bowel obstruction**

This is where there is obstruction to the small bowel so that food cannot pass from the stomach to the large bowel. This can be complete, where part of the bowel has failed to form at all (duodenal or jejunal atresia) or partial, where the bowel has formed but is narrower than it should be. (duodenal stenosis). A similar problem can occur with annular pancreas. The pancreas should normally lie behind the lower part of the stomach and the first part of the small bowel. In this condition the pancreas encircles the duodenum causing narrowing or blockage.

When the blockage is severe, it may be detectable before birth on an ultrasound scan. If not detected before birth problems will present in the first few hours or days of life with vomiting or failure to pass stools. Less severe obstruction may not present so dramatically, though vomiting is still likely to be the main problem. Diagnosis will generally be made by X-ray. Treatment is usually surgical. This involves removing the blocked part of the bowel and then joining it up again. It is a major surgical procedure, but without this treatment most babies will not survive. In a minority of milder cases the problem can be managed without surgery by manipulating the diet.

### **Abnormalities of the anus**

Sometimes babies are born without an anal opening (Imperforate anus). This is not common but occurs more often in Down's syndrome. This will be noticed at birth and usually requires immediate action. The extent of corrective surgery will depend on how severe the abnormality is. Less severe problems occur when the anal opening is narrower than usual (anal stenosis). This is likely to cause constipation. The opening can sometimes be stretched under anaesthetic but severe cases may still require surgery

### **Hirschprung's disease**

Again this is a relatively rare condition that is more common in Down's syndrome (approximately 2% of children with Down's syndrome). It is an abnormality of the lower part of the large bowel, whereby part of the bowel wall has nerve cells missing. This means it cannot do its normal work of pushing stools along to the anus. Sometimes a long segment of bowel wall is affected. In this case it may be obvious in the newborn period because the baby does not pass any stools. More often these babies have chronic constipation, poor weight gain, vomiting and a swollen abdomen. If however only a short part of the bowel is involved (short segment Hirschprung's disease) symptoms are less severe. It is in these children that the diagnosis may be easily missed. It is important to consider the possibility of short segment Hirschprung's in any child whose constipation persists despite dietary measures and simple laxatives.

Diagnosis is made by a combination of medical examination, X-ray and biopsy of the bowel. Treatment usually involves surgery to remove the abnormal part of the bowel. Sometimes it is necessary to let the remaining lower part of the bowel 'rest' by using a colostomy. This is where the upper end of the remaining bowel is temporarily attached to an opening or stoma in the abdominal wall, through which stools are passed into a bag. Once the bowels have healed up, usually several months, they can be joined up again.

## **Problems of function**

### **Feeding difficulties**

Babies with Down's syndrome quite commonly have difficulties with feeding, particularly if they are born prematurely. This may be due to their generally low muscle tone and difficulties coordinating sucking and swallowing. It may also be due to other medical problems. For instance babies with heart problems may tire easily, or be short of breath and not able to feed adequately. There are a whole number of things that may help including different ways of positioning during feeds, 'tricks' to help stimulate the baby's mouth movements, and, in bottle fed babies, trying different teats, bottles or other equipment. Health visitors, specialist nurses and speech and language therapists are able to provide this sort of advice.

Sometimes the baby is not able to feed adequately despite these measures and it is necessary to feed the baby via a nasogastric tube for a while. This is a small, flexible tube that is passed into the nostril, down the oesophagus and into the stomach. This is usually only necessary for a short time.

### **Gastro-oesophageal reflux**

This occurs when food that had already passed into the stomach and beyond comes back up into the oesophagus and may be vomited up. Most healthy people experience this from time to time. It is more

common in babies because their food is liquid and therefore more easily brought back they spend less of their time upright the muscle at the top of the stomach that should prevent this is not yet well established.

Some also have a hiatus hernia where the top part of the stomach is pushed just above the diaphragm into the chest. Babies with Down's syndrome are more likely to have reflux, probably because the muscles of the stomach and oesophagus that work to push food along seem to work less effectively. Symptoms may be very mild and merely a nuisance. Simple measures mentioned above may help. However, vomiting may be considerable and the child may not gain weight. Also the acid contents of the stomach irritate the lower oesophagus causing discomfort, and sometimes bleeding from the oesophageal wall. This can cause anaemia. In these cases medical treatment is necessary. Several different kinds of medicine are used, often in combination. They work in a number of ways - by preventing the stomach contents flowing back, by neutralising the stomach acid and by improving the gastrointestinal motility. Very occasionally these measures won't be sufficient. and an operation to tighten up the junction between the oesophagus and stomach will be necessary.

### **Malabsorption**

This is a condition in which the bowels are unable to absorb particular nutrients from food. This can cause the body to run short of some nutrients, and the stools to be abnormal. Possible malabsorption of a number of different vitamins and minerals has been described in Down's Syndrome from time to time. However the evidence for this is inconsistent and whether the malabsorption leads to any health problems is uncertain.

There is, however, one important type of malabsorption that is more common in Down's syndrome called Coeliac Disease. In this, the body develops an allergy to part of a protein called gluten, which is found in wheat and some other cereal grains. Symptoms include poor growth, abnormal stools (diarrhoea, frothy, foul smelling or bulky stools are typical), swollen stomach, tiredness and irritability. Anaemia may also result. Special blood tests are available which may help with diagnosis, but a jejunal biopsy may be necessary. In this test a small tube is swallowed, and a sample of the wall of the jejunum is removed for examination under a microscope. Treatment is by special diet excluding gluten. This should be supervised by a dietitian.

In this article I have considered some of the gastrointestinal problems that occur in Down's syndrome. Many children will, happily, have none of these problems. Some will have one and an unfortunate minority will have several at various times in their life. As I stated early on in the article, children with Down's syndrome may also get any of the problems that occur in other children. As in all medical conditions, the symptoms should never be accepted as "just part of Down's syndrome," without first considering other, treatable, conditions.

Dr. Liz Marder,

Consultant Community Paediatrician and Medical Advisor to the DSA. August 1996