Hirschsprung’s disease in children

What is Hirschsprung’s disease?
Hirschsprung’s disease is a disorder which affects the bowel. Most commonly only part of the large bowel (colon) is affected but in some cases part, or all, of the small bowel (also called ‘small intestine’ or ‘ileum’) is also affected. The majority of cases affect only the lower part of the colon (referred to as ‘short segment disease’). If most of the colon or any small bowel is involved this is called ‘long segment Hirschsprung’s disease’.

The colon moves digestive material through the gut by a series of contractions and relaxations called ‘peristalsis’. Peristalsis is controlled by nerves (called ‘ganglion cells’) which are found in between the layers of muscle tissue. Children who have Hirschsprung’s disease are missing the ganglion cells along part of the length of their bowel which prevents the bowel from relaxing and so results in difficulty passing faeces (‘stool’ or ‘poo’) or an obstruction in the bowel.

What causes Hirschsprung’s disease?
Hirschsprung’s disease is a congenital condition which means that it is present at birth. Before birth, while a baby is developing in the womb, ganglion nerve cells develop in the intestine, passing from top to bottom in the direction of the anus. In Hirschsprung’s disease ganglion cells stop developing at some point on that pathway meaning that the lower part of the bowel does not have ganglion cells. The cells most commonly stop relatively close to the anus meaning that patients have short segment disease.
At the current time we don’t know what causes this to occur but we do know:

- Hirschsprung’s disease affects one child in every 5000.
- There is nothing that a parent should have done or not done during pregnancy to prevent Hirschsprung’s disease in their child.
• Hirschsprung’s disease can run in families. A number of genes causing Hirschsprung's disease have now been identified but not all children have a specific genetic mutation.
• Hirschsprung’s disease affects boys more than girls with approximately five times more boys being affected than girls.
• Some children with other conditions such as Down syndrome or genetic heart conditions are more likely to have Hirschsprung’s disease.

What are the signs and symptoms?

Signs and symptoms will depend, in part, on the length of bowel affected and also on the age of the child at diagnosis:

In the newborn and neonatal period:
• Hirschsprung’s disease will be suspected if a newborn baby does not pass meconium (the dark, tar like stool) in the first 24 to 36 hours of life. This is an important sign as nearly half of infants with delayed passage of meconium have Hirschsprung’s disease.
• Constipation
• Swollen abdomen, bloating (called ‘abdominal distension’)
• Repeated vomiting which may include vomiting green bile

In older infants and children:
• Constipation which is poorly treated/ resistant to treatment with laxatives
• Abdominal swelling (called ‘abdominal distension’)
• Bowel obstruction
• Chronic abdominal pain
• Diarrhea
• vomiting, which may include vomiting green bile
• difficulty gaining weight
• soiling with overflow incontinence

How will we know that my child has Hirschsprung’s disease?

Hirschsprung’s disease is diagnosed based on symptoms and test results.

A doctor will talk to you about your child’s symptoms and examine your child by looking at their general appearance, feeling the abdomen and also by inserting a finger into your child’s anus to feel inside the bottom.

Hirschsprung's disease can be suggested on x-rays. In Hirschsprung’s disease the last segment of the bowel can look narrower than normal whilst the section above it will look distended (bulged) because of the blocked stool stretching the intestine.

The only way to reliably diagnose Hirschsprung’s disease is to take a small piece of tissue from the bowel to examine under a microscope. This is called a rectal biopsy. In babies a rectal biopsy can be undertaken on the ward (this is called a ‘suction rectal biopsy’) but
older babies and children will need to have a general anaesthetic and an ‘open rectal biopsy’ taken.

A specific information leaflet for parents and carers is available which discusses rectal biopsy in detail. If your child is to have a rectal biopsy and you have not been given this leaflet please ask your nurse or doctor to provide one for you.

**How is Hirschsprung’s disease treated?**

All children with Hirschsprung’s disease will require surgery. However, the type of surgery undertaken will depend on the age of the child and how well the child is at the time of diagnosis. In circumstances where children are unwell at the time of diagnosis, their acute problems need to be treated before any surgery is planned:

**Acute problems**

**Presence of intestinal (bowel) obstruction**

Infants and children who have symptoms of intestinal obstruction will be treated with the following:

- Intravenous rehydration – this means that fluid is given via a ‘drip’ to rehydrate and then keep the child hydrated.
- Gastric decompression – to decompress the upper gut (stomach) oral feeding will need to be temporarily stopped. A tube called a ‘nasogastric tube’ is passed down the child’s nose, down the food pipe (oesophagus) and into the stomach. Through this tube the stomach contents and any trapped gas can be cleared to decompress the upper gut and prevent further vomiting.
- Intestinal decompression – a member of the surgical team will need to perform a ‘digital rectal examination’. This involves passing a finger into the anus to help clear trapped wind and stool. In addition, stool will need to be washed out of the bowel via the anus and this will need to be undertaken once or twice a day. A specific parent/carer information leaflet is available on rectal washouts; please ask your nurse or doctor for a leaflet if your child needs rectal washouts and you have not been given a copy of this leaflet.

**Presence of enterocolitis**

Enterocolitis is an inflammation of the digestive tract caused by an infection. If your child has symptoms of enterocolitis at the time of Hirschsprung’s being suspected or diagnosed, they will need to receive treatment with intravenous antibiotics (antibiotics are given through a ‘drip’) in addition to the above.

**What does surgery involve?**

During surgery for Hirschsprung’s disease the aim is to remove the affected (‘aganglionic’) section of bowel. Surgery may be undertaken as a single stage or as in more than one stage depending on the age of your child and how well they are at the time of diagnosis:

**Single stage surgery**

If your child is newborn and generally well at the time of being diagnosed with Hirschsprung’s disease, your child’s surgical team are likely to advise that surgery to
remove the affected part of the bowel is undertaken as a single (one) stage operation which is called a ‘primary pull through procedure’.

A primary pull through procedure for Hirschsprung’s disease is performed under general anaesthetic. Small (2-5mm) incisions are made in the abdominal (tummy) wall and through these special instruments are passed and, through one of the incisions a camera (called a laparoscope) is passed so the surgeon can see the inside of your child’s abdomen. To confirm the exact length of affected bowel, a series of biopsies are taken using the laparoscopic instruments and these biopsies are sent immediately to the laboratory and examined under a microscope and results immediately passed back to the surgeon. Once the length of affected bowel (called the ‘aganglionic section’) has been identified, the blood supply is divided using the laparoscopic instruments. The affected bowel is then removed via your child’s anus and the bowel with the ganglion cells present within it is sewn in place.

**Advantages of one stage surgery:**

- Only one operation for child to undergo.
- Surgery is undertaken as a keyhole (‘laparoscopic’) operation.
- Keyhole surgery results in a quicker recovery for most children compared to those that have staged or open surgery; they start to take feeds again quicker, have less pain after surgery, spend less time in hospital after surgery and, in the longer term, have smaller scars.

**Disadvantages of one stage surgery:**

- To make the operation as technically easy as possible and to decrease complications at the time of surgery, babies usually need to weigh a minimum of five kilograms before single stage surgery can be undertaken.
- Whilst waiting to undergo the single stage operation your child will need to have their intestine decompressed at home and this is achieved by parents/ carers performing the rectal washouts at home. Parents/ carers are taught to perform rectal washouts by the nursing staff at the hospital and a specific leaflet is available for parents/ carers about rectal washouts so please ask your nurse or doctor for a leaflet if you have not received one.

**Staged surgery**

**Staged surgery is performed in:**

- Babies who are unwell at the time that Hirschsprung’s disease is diagnosed
- In babies where intestinal decompression has not been effective
- In older children

Babies and children who undergo staged surgery have two or three separate operations and there is usually a minimum of approximately six weeks between each operation. In the first operation a series of biopsies are taken and sent immediately to the laboratory and examined under a microscope to determine the length of affected bowel. A stoma is then formed in the part of the bowel that has ganglion cells present in it. Forming a stoma means that a small section of large bowel (colon) or small bowel (ileum) is brought onto the surface of the skin as an artificial opening. Faeces (stool, poo) will then pass through the stoma and
then into a bag which is stuck to the skin and so bypass the section of bowel that does not have the ganglion cells within in.

Having the stoma allows your child to recover and become healthy again and to then be able to go home.

During the second operation, which is usually undertaken a minimum of approximately six weeks later, the section of bowel that does not contain the ganglion cells (called the ‘aganglionic section’) is separated from the rest of the bowel and removed. The bowel with the ganglion cells present within it is then connected to your child’s anus and sewn in place. Sometimes the stoma can be closed (and so the bowel re-joined and stool therefore passed via the rectum) at the end of the second operation but in other cases the stoma needs to be closed at a third, separate operation. Your surgeon will discuss with you which is more likely to be necessary for your child.

**Advantages of staged surgery:**
- Allows the safe recovery of unwell children
- Home rectal washouts to decompress the bowel are not necessary

**Disadvantages of staged surgery:**
- More than one operation is needed.
- Child will need to have a stoma temporarily
- Scar will be larger than with keyhole surgery

**Are there any alternatives to surgery?**
No, all children with Hirschsprung’s disease will need to have surgery.

**Will losing a length of the bowel affect my child?**
The extent to which losing some of the bowel affects your child will depend on the length that needs to be removed during the pull through operation.

In adults without any problems related to their bowel the length of the bowel varies; men have slightly longer bowels than women. In an adult the small bowel (ileum) is, on average, seven metres long (approximately 23 feet) but can vary from 4.6m (15 feet) to 9.8m (32 feet). The large bowel (colon) meanwhile is, on average, about 1.5m (5 feet) long.

The length of bowel increases as a child grows so, whilst an adult’s colon is approximately 1.5m long (5 feet), at birth the large bowel is only about 0.6 m (2 feet) long.

Because the bowel continues to gain length throughout childhood, removing a short section of bowel during a pull through operation does not usually cause any long term affect. However, for children with long segment Hirschsprung’s disease more bowel needs to be removed and so children will be at greater risk of dehydration, diarrhoea, poor absorption of nutrients and poorer growth. To help ensure that these risks have only a minimal affect on your child, your child will be closely monitored by our specialist doctors and dietitians. This may include intermittent urine and blood tests, your child may need some medication including those that can slow the passage of stool down through the bowel and those that give nutritional supplements and it will be very important that your child drinks plenty of fluid.
What happens before surgery takes place?

Before any surgery takes place, any acute problems will need to be treated (see section ‘How is Hirschsprung’s disease treated’ above). What then happens depends on the type of surgery planned.

Before staged surgery

Your child will be transferred to theatre for the first stage of surgery that is, forming the stoma.

Once your child has recovered you will be taught how to care for your child’s stoma by the ward and specialist nurses and then you will be able to go home.

The second stage of surgery (the pull through procedure) will be undertaken, usually a minimum of six weeks later. Your child will be readmitted on the same day or the day before stage two.

Before single stage surgery

You will be taught to undertake rectal washouts for your child by our nursing staff. Once you are both competent and confident with undertaking these and, assuming your child’s bowel decompresses effectively with the washouts, you will be able to go home.

Once at home support is provided to you by a community children’s nursing team and also, from the hospital, via the surgical nurse specialist team. Once your child weighs over approximately five kilograms surgery will be planned.

Your child will be admitted one to two days before the single stage pull through procedure to be prepared for the operation. This involves:

- blood tests
- additional washouts being given to clear the bowel of faeces in preparation for surgery
- your baby receiving clear fluids only rather than milk feeds

What are the risks/complications of the surgery?

As with all operations there are risks but these are rare. They include:

- **Bleeding, bruising, infection** and/or **protrusion of tissue** through the wound.
- **Adhesions:** This is scar tissue formation which can occur after any abdominal operation. It is a small but lifelong risk which may result in an obstruction (‘blockage’) of the intestine. Symptoms of adhesion obstruction include cramping abdominal pain and green (bile) vomit.
- There can be **leakage** from the repair site in the bowel.
- **Stricture** in the bowel where the bowel was resected and rejoined.
- **Enterocolitis** – whilst the risk of enterocolitis is reduced after the pull through operation have been undertaken, the risk continues to be present (see section ‘What is enterocolitis and why is it important that I know about it?’ below).
What care will my child need after surgery?

**Short term:**
- After surgery your child will need intravenous fluid (through a drip) until they are able to start taking fluids and feed again. How long this is depends on the type of surgery undertaken and the child’s individual recovery time but can be 24 hours to a few days in total. Children are able to start taking fluids again once flatus (wind) is being passed.
- Your child will receive intravenous antibiotics through a drip.
- Pain killing medication will be given, initially through the drip and then, once feeds have been recommenced, by mouth.
- The nurses on the ward will carefully monitor your child’s heart rate, respiration rate and temperature and also the fluid in and output.
- Once your child starts to pass stool (poo) careful attention will need to be paid to provision of perianal skin care. The skin on your child’s bottom area will not be used to having stool on it and, in addition, after surgery it is quite common for children to open their bowels very frequently for the first days and weeks. To prevent the skin becoming sore, skin care should include the following:
  - Use of topical creams to the skin around the bottom. Creams which are commonly used in children include Cavilon (a barrier film which is used once per day and is available on prescription), Metanium (a barrier cream that should be applied at every nappy change and is available on prescription or without) and Ilex paste (available to purchase and on prescription). Your nurse specialist will discuss this in detail with you before discharge.
  - Using a hair dryer on its cold setting to dry the skin.
  - Keeping your child’s bottom exposed to air for periods during the day whenever possible.

**Medium term:**
In the weeks and first months after surgery has been completed, your child will need monitoring and reviews. Telephone consultations with the nurse specialist team will be provided as well as reviews on the ward and in the outpatient clinic. If you have any concerns or questions it is important that you contact the nurse specialist team (phone numbers are provided at the end of this leaflet). In particular:
- If your child’s skin starts to become sore despite receiving the skin care described above it is important that you contact the nurse specialist team for advice or review.
- Your child’s weight should be monitored every two weeks for the first few months after surgery and, if weight is static or falls, the nurse specialist team should be contacted.
- Children will be reviewed to assess for any signs of a stricture developing where the bowel was resected and rejoined. Some children will need to receive ‘anal dilatation’ to prevent or treat any narrowing (‘stenosis’). A specific information leaflet is available which discusses anal dilatation in detail; if your child requires anal dilatation and you have not received a leaflet please ask your nurse or doctor for a copy.

**Long term:**
- The majority of children born with Hirschsprung’s disease grow up to lead normal lives with normal bowel function.
• Your child will need ongoing outpatient monitoring, usually until a minimum age of five years.
• Many children are potty trained normally but for others potty training of faeces may be slightly delayed compared to children that were not born with Hirschsprung’s disease.
• Some children experience constipation and so need to receive laxatives. Laxative medication may be needed for some months or, in some cases, many years.
• Some children experience loose stools and some of these will need to be given medication to help slow the passage of stool down.
• It is important for us all to have a healthy diet and plenty of fluids each day but it is more important in children with Hirschsprung’s disease. There is no need to delay weaning a baby onto solids and no need to avoid any particular foods in older children; however, in most children some foods tend to help them poo by keeping their stools soft (this is often fruit except bananas) whilst other foods bulk the stool (high fibre, whole-grain foods). You will learn, from your child trying different foods, which ones are more likely to constipate them or to make stools loose. Drinking plenty of fluids is always important and will help prevent constipation. One of our dietitians will be pleased to give you advice if you have any concerns or questions.
• Children with long segment Hirschsprung’s disease will have lost a longer length of bowel during the pull through operation and so need specific monitoring of their growth to ensure they are absorbing enough nutrients from their food. One of our dietitians will see your child and be able to advise you.

What is enterocolitis and why is it important that I know about it?

Enterocolitis is an inflammation in the bowel in children with Hirschsprung’s disease caused by infection which is potentially very serious. Enterocolitis can present with symptoms of a simple gastroenteritis (vomiting and diarrhoea) illness but children with enterocolitis can become very unwell in a short space of time.
If your child experiences the following symptoms it is important that they receive urgent medical review:
• high temperature
• vomiting – this may be green in colour
• abdominal distension
• explosive, offensive smelling and profuse watery stools
• blood passed via the bottom (rectum)
• dehydration (so child is pale and floppy)

Children with suspected enterocolitis will be admitted to hospital to receive intravenous fluids and antibiotics (through a drip) and rectal washouts to help clear the infection. During the time that your child is at greatest risk of enterocolitis (that is, before the pull through operation has been undertaken) your child will have ‘open access’ to your nearest children’s ward. This means that you will need to call the relevant ward/ department in advance but then will be seen immediately by appropriate professionals who know how to assess and treat children with this condition rather than needing to be seen by a GP or general A&E doctor first. Your nurse specialist will explain the process for open access that...
is relevant to your child before discharge and you will be given a letter to keep with you and to provide should you need to utilise the open access system.

**As Hirschsprung’s disease can run in families, what does this mean for any future children I might have?**

If you have a child with Hirschsprung’s disease future children may be at increased risk of also having this condition. If you would like, we can arrange for you to meet the specialist genetics team to discuss this with you further.

**Who shall I contact if I have any queries, concerns or questions?**

For further information/queries please contact:

The ward you were on ______________________________________________________________

Your nurse specialist (Mon to Fri 0800 to 1800hrs) 01223 586973

Your children’s community nursing team ________________________________________________

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We are now a smoke-free site: smoking will not be allowed anywhere on the hospital site. For advice and support in quitting, contact your GP or the free NHS stop smoking helpline on 0800 169 0 169.

**Other formats:**

If you would like this information in another language, large print or audio, please ask the department where you are being treated, to contact the patient information team: patient.info@addenbrookes.nhs.uk.

Please note: We do not currently hold many leaflets in other languages; written translation requests are funded and agreed by the department who has authored the leaflet.

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