This booklet has been designed to help those involved with the care of children with Hirschsprung's disease to understand what it is, the treatment involved and why long term good bowel management is so important.

This booklet is part of a series for children with bowel problems.

Titles of other booklets currently available in this series:

'Talk about going to the toilet'
'Talk about constipation'
'Understanding constipation in infants and toddlers'
'Understanding toilet refusal – the child who will only poo in a nappy'
'Understanding Anorectal Malformations
'Understanding bowel management' - in press

Bladder and Bowel UK

Illustrations - Les Eaves

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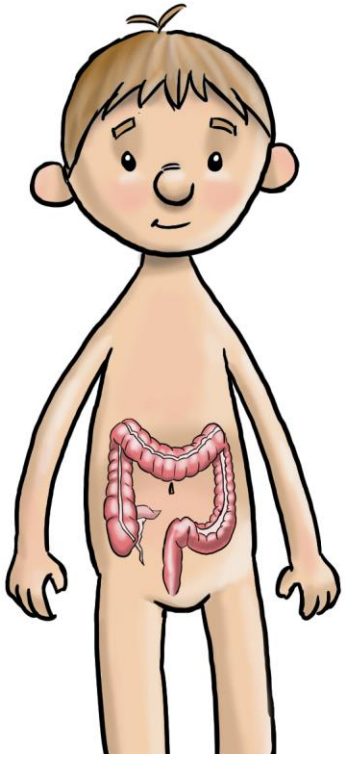
Down's Syndrome Association and the Platinum Trust

Bladder and Bowel UK
formerly PromoCon
part of Disabled Living

Understanding Hirschsprung's Disease

A guide for parents and carers



What is Hirschsprung's disease?

Hirschsprung's Disease is a rare congenital abnormality (it is present from birth), which affects specific nerve cells, called parasympathetic ganglion cells, in the large bowel (colon).

Normally, as the baby is developing in the womb, the nerve cells grow along the bowel towards the rectum. In children with Hirschsprung's Disease the nerve cells stop growing too soon. This happens before the 12th week of development and the reason for this is not yet known.

The result is a lack of these ganglion cells in the rectum and also in varying degrees along the length of the large bowel. Absence of the ganglion cells means that signals to the muscles are not sent and the affected part of the bowel is unable to relax - this prevents stools (poo) moving along the bowel, resulting in constipation, or in severe cases complete obstruction of the bowel.

How severe the Hirschsprung's disease is, depends on how much of the colon is affected. Short-segment Hirschsprung's Disease means only the last part of the bowel is affected, while long-segment Hirschsprung's means that most or all of the large bowel is affected.

How common is it?

Hirschsprung's is a rare disease. The incidence is 1 in 5000 live births and it affects more boys than girls. However, for children with Down's Syndrome there is a 40% increased risk of Hirschsprung's Disease occurring.

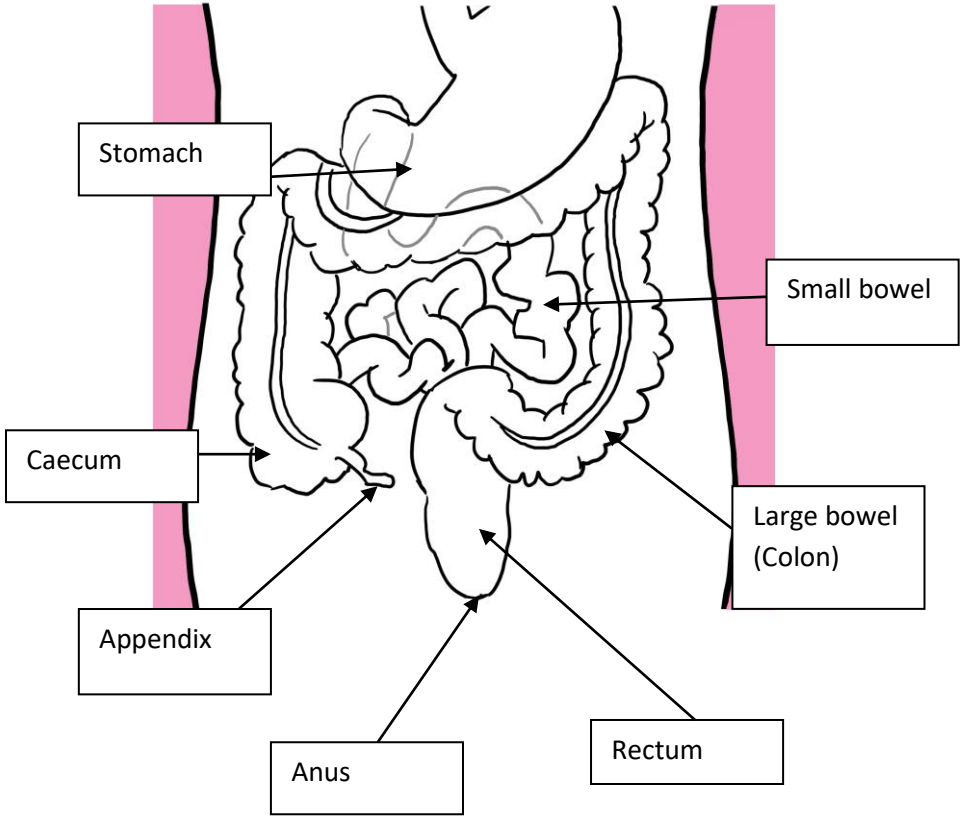


Diagram of Digestive System

What are the presenting symptoms?

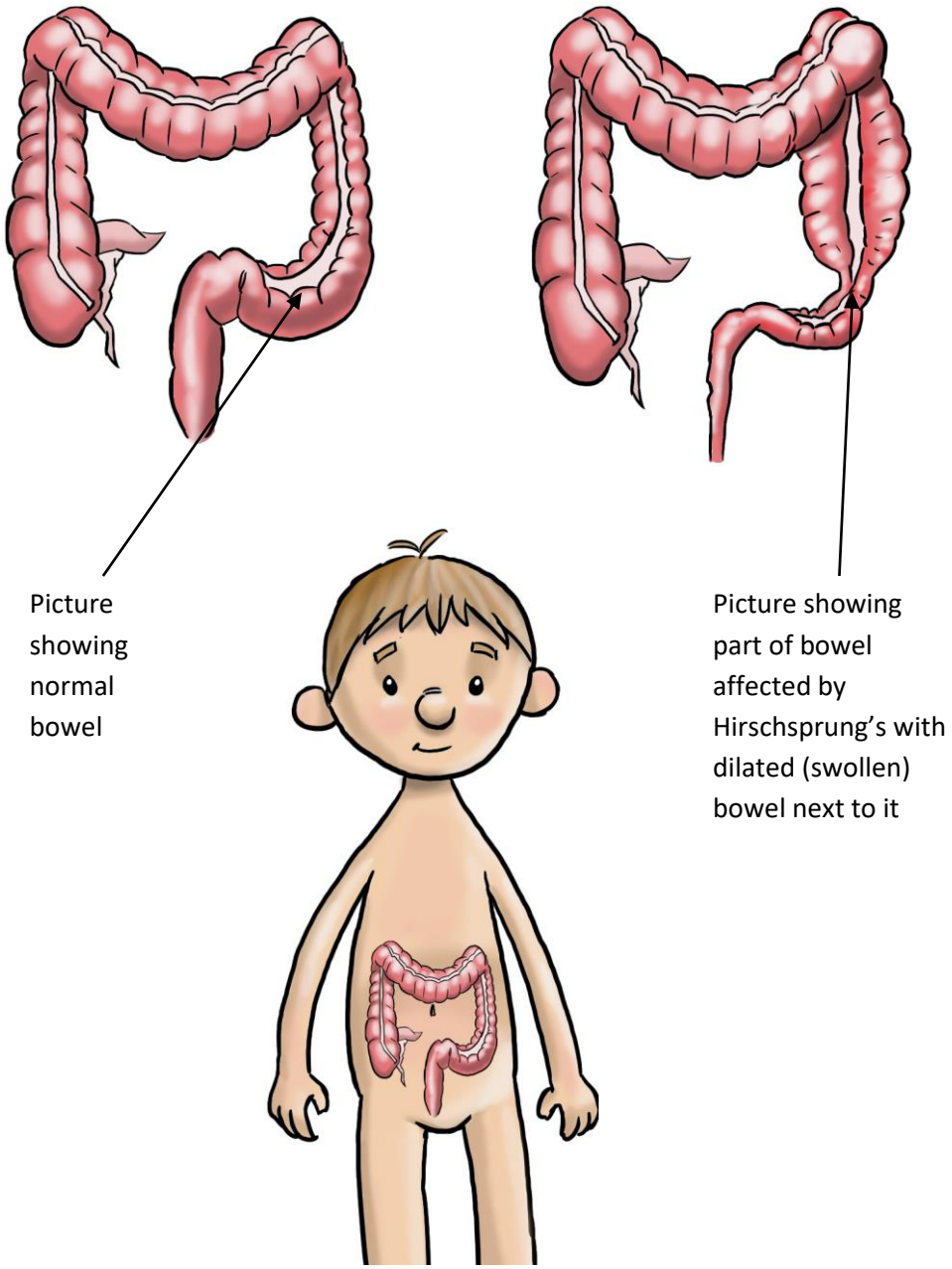
Hirschsprung's disease is usually identified shortly after birth. Delay in the passage of meconium (the first sticky, black stool) which is usually passed in the first 48 hours following birth, is a common symptom. A distended abdomen (bloated tummy) or vomiting may also occur.
Older infants and children with short-segment Hirschsprung's may present later, with chronic constipation which is resistant to the usual treatments and is often accompanied by poor weight gain.

How is it diagnosed?

The doctor will usually carry out a physical examination, including feeling the baby's' tummy and putting a finger in their bottom. However, the only definitive way to test for Hirschsprung's disease is a rectal biopsy – this involves taking a sample of cells from the rectum and looking at them under a microscope. If Hirschsprung's disease is present, there will be no ganglion cells in the sample biopsied. Other tests may include blood tests and x-rays. An abdominal x-ray will show a distended (swollen) bowel above the affected section, but a barium enema may be used to determine the extent of the problem.

What is the long term outcome?

The majority of children who are born with Hirschsprung's disease do very well. However, a number of children may have ongoing problems with diarrhoea or constipation, soiling and abdominal pain. Some children may take longer to toilet train, because of the problems with bowel management. Sometimes long term medication and further interventions, including surgical operations, are needed to help with these problems.



Pictures showing the position of the bowel in the body and how Hirschsprung's Disease affects it

How is it treated?

The immediate management of the baby will depend very much on the extent of the Hirschsprung's disease and the particular hospital unit the infant has been admitted to.

Some infants will be managed in the short term with rectal washouts before surgery is undertaken, to remove the affected bowel and attach the healthy bowel to the anus, called a 'pull through' operation. Sometimes the surgery needs to be done in two stages, with a colostomy or ileostomy being formed soon after birth and the second stage, of connecting the healthy bowel to the anus, is done later .

A colostomy is where the large bowel is brought to the surface of the abdomen (tummy) and an opening is formed. An ileostomy is where the small bowel is brought to the tummy wall. In both cases the poo is collected in a special bag (stoma bag), stuck over the opening.

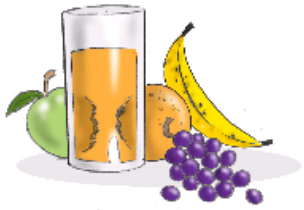
A stoma can feel scary to start with, but there is support available from stoma nurses or paediatric community nurses. They can show you how to look after a stoma and will also tell you how you can get the supplies, which you will need.

What happens when the stoma is reversed?

Following the reversal of a stoma, some children, especially those left with a short amount of large bowel left (because they have had a large part of their bowel removed or because it was absent), may initially have very loose, watery stools. As stools are then more alkaline than normal, if they come into prolonged contact with skin, they will 'burn' the skin and make it sore. The use of special barrier creams can help make this better or prevent this from happening – your health care professional can advise you further.

What is Enterocolitis?

Enterocolitis is an infection of the large bowel. It is more common in children with Hirschprung's disease. The cause of enterocolitis is not fully understood, but it is thought that poor movement of the intestinal contents and bacterial invasion are contributory factors. It presents as severe inflammation of the bowel and is often accompanied by explosive diarrhoea, foul smelling stool (poo), vomiting, abdominal distension (bloated tummy)and fever. The risk for enterocolitis is higher in children with Down's Syndrome. If this is suspected the child should be referred for urgent medical treatment.



Laxatives, fluids, fibre intake and toileting programmes are all important in maintaining health bowels

Caring for children with Hirschsprung's disease.

Notes

It does depend on how much bowel has been affected, and the results of the surgery, as to what problems the child will have. Some children will be toilet trained and fully continent by the time they get to school age. They may still need laxatives and a structured bowel programme including easy access to the toilet, because their need to poo may be very unpredictable.

Others children may struggle with constipation and soiling until they are in their teens. Some may need further surgery to enable them to be continent. Obviously there is a strong link between becoming continent of faeces (poo) and an improvement in quality of life. This makes it really important for children to receive practical support from health care professionals, schools, parents and carers, to stand the best chance in being successful at toilet training.

A structured bowel training programme should always be introduced

Further sources of information

Bladder and Bowel UK (Formally PromoCon)

Bladder and Bowel UK, part of the charity Disabled Living, provides qualified impartial advice and information regarding continence issues, products and services for children and adults with bowel and/or bladder problems.

Helpline Tel: **0161 607 8219**
Email: bladderandboweluk@disabledliving.co.uk
Website: www.bladderandboweluk.co.uk

Breakaway Foundation

Breakaway are are a UK wide charity offering support, holidays and activities to children from birth to eighteen, with bladder and/or bowel diversions/dysfunction and their families.
http://www.breakawayfoundation.org.uk/

Champs Appeal – raising Hirschprung's Disease awareness

CHAMPS is an appeal set up to raise awareness of children with Bowel and Bladder disorders, dysfunctions and diversions. They raise funds that will directly benefit children by supporting research, awareness campaigns and support groups
http://www.champsappeal.co.uk

Contact a Family

Contact a Family is a UK charity for families with disabled children. They offer information and support on specific conditions and rare disorders as well as advice regarding related issues such as benefits and finance. http://www.cafamily.org.uk/

Contact a Family, 209-211 City Road, London, EC1V 1JN
Tel: 020 7608 8700
Fax: 020 7608 8701
National freephone helpline: 0808 808 3555
Monday-Friday, 9.30am-5pm
e-mail: info@cafamily.org.uk

Diversions

Diversions is a support network, based in the North West of England, for families with a child or young person living with a bladder or bowel diversion/dysfunction. http://diversions.org.uk/home

Contact:
Melissa – 07816513889
Rachael – 07814613669
Email: diversions@live.co.uk

Down's Syndrome Association

Provides information and support regarding all aspects of Down's Syndrome to all those who need it.
http://www.downs-syndrome.org.uk/
Langdon Down Centre
2a Langdon Park
Teddington
Middlesex
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Tel: 0333 1212 300.
Email: info@downs-syndrome.org.uk