The Secret Disease

HIRSCHSPRUNG'S

Hirschsprung's & Motility Disorders

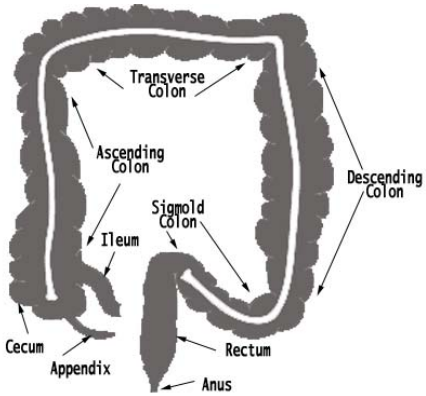
Support Network

What is Hirschsprung's?

Hirschsprung's is also known by several different names, such as

- Congenital megacolon
- Aganglionic megacolon
- Megacolon congenitum
- Congenital aganglionosis

The simplest definition is that Hirschsprung's is the lack of Ganglion cells in the bowel. This results in the stool being unable to pass through the bowel to the rectum.



You may hear the terms **Short Segment, Long Segment, Total Colon, Total Intestinal, or Ultra Short**. These refer to the length of colon where these cells are absent from and so they are fairly self explanatory but if you have any doubt do not hesitate in asking your consultant. Figure 1 shows the different parts of the bowel.

Ganglion Cells!!!! What are they?

Ganglion cells allow the bowel to relax so that the stool can pass through the bowel. If the bowel cannot relax then the stool or bowel motion cannot pass down the bowel and the bowel becomes distended and blocked.

These cells arrive in the bowel when the baby is in the womb. The arrival is part of the process called the 'Neural Crest Cells Migration'. The ganglion cell travel down the bowel to the anus. In Hirschsprung's this migration is told to stop before the whole bowel has been populated. Why the migration is stopped is not yet clear but is one of the areas of research.

What Effect does this have?

Well basically, the result is that the baby cannot go to the toilet. Without the

Ganglion Cells the bowel cannot relax and push the stool down the bowel. The pushing of the stool through the bowel happens in stages, like a wave washing debris onto a beach. As the stool tries to pass down the bowel, where it reaches the bad part of bowel there is a build up of stool making the bowel stretch.. Just after this blockage the bowel looked smaller as it will not relax, this is what is looked for in a contrast x-ray.

What are the Symptoms?

The most common sign of Hirschsprung's is that the baby does not pass the meconium within the usual 24 to 48 hours. Mind you this may have been passed by the baby just before birth but this is unusual. The meconium is the substance that lines the baby's intestines during pregnancy.

Other Signs are

- Vomiting
- Tiredness
- Explosive or Difficult Bowel Movements
- Jaundice
- Swollen Tummy
- Poor Feeding
- Poor Weight Gain
- Slow Growth in the first 5 years

If your baby has some of these, don't worry the odds are it is not Hirschsprung's but what you must do is to get your baby examined by a specialist doctor

How will it be Diagnosed?

In some cases it can be very hard to diagnosis Hirschsprung's, which is why some children can go many years without it being diagnosed.

The diagnosis will be dependant on how old your child is. In a young or newly born baby, the diagnosis will generally be on bowel movements or lack of them, the condition of the tummy and **full thickness or suction biopsy**

This is where tiny snip-its of the bowel are taken and tested for the presence ganglion cells and if found how many? In the affected part of the bowel there is an increase in the nerve trunk density & thickness. Ganglion cells can be found in Hirschsprung's children but they maybe so few in numbers that the bowel will not function properly. Sometimes it may take more than one biopsy to obtain a definitive diagnosis.

The older children will have a **contrast x-ray** to show the state of the bowel. Depending on the age of the child an **anorectal manometry** may be performed. This will tests the strength of the muscles around the rectum to see if they are working properly. It is painless but does feel a little uncomfortable or strange.

Surely I could have prevented this?

Please be assured that there was nothing that you did while pregnant or even before pregnancy would have caused or contributed to your child having this disease. Hirschsprung's is a genetic disease. To be technical it is caused by a fault in the Ret proto-oncogene. In a number of cases this faulty gene is hereditary but in the most common form of Hirschsprung's there seems to be no family history.
So far there is no test that can be performed to show that your unborn child has Hirschsprung's or not.

My child is not a baby but still has bowel problems?

In some instances it can be very hard to diagnose HD. It is easier to prove that a child has Hirschsprung's instead of proving they do not. The biopsy may have shown ganglion cells but not enough biopsies were taken to show the areas without cells. The doctor may of thought that the problem was the child holding in the stool as does happen.

If after this length of time, your child is still having problems then they must be checked by a specialist consultant. It has been known for adults, later on in their lives to be diagnosed with Hirschsprung's but thank goodness as diagnostic techniques and knowledge of Hirschsprung's improves late diagnosis is becoming rarer.

What is the treatment, laxatives will work?

Hirschsprung's is a very serious disease and the only current effective treatment is to have a surgical procedure called a Pullthrough. The affected part of the bowel must to be removed as it will never function properly. In the operation, the affected bowel is removed and the good bowel is pulled down and attached to the anus.

This may be performed in a one, two or three stage operation. In the two stage operation the pullthrough is performed but part of the bowel is attached to a bag. The reason for this is usually to allow the bowel to recover from all of its stretching and infections, such as enterocolitis that may have occurred previously. The attaching of the bowel to a bag, through a hole in the tummy is

called a stoma. Depending on where in the bowel the stoma is taken from it is the called either an illeostomy or a colostomy. At a later date agreed with the surgeon the stoma will be closed and your child will then start to pass motions normally.

In the one stage procedure there is no stoma and your child will pass motions normally soon after the operation.

The operation which was first performed by Dr Swenson in 1946, has had subtle variations developed over time and are known as the **Swenson, Duhamel and Soave procedures**. The newest procedures are the Transanal & Laparoscopic pullthrough's. The Transanal is where the pullthrough is performed via the anus and no abdominal incisions are necessary. The Laparoscopic method reduces the abdominal incisions and therefore reduces the pain involved .

Your doctor will perform the surgery that they feel most comfortable with and which they consider the best for your child. Remember that the doctor does want the best for child.

What will happen after the operation?

If your child has had the two or three stage pullthrough then you will need to learn how to look after the stoma. The biggest concern is that the stoma stays moist and red, showing good blood flow. To help you in this you will be allocated a Stoma Nurse who will teach you all you need to know. Stoma Nurses are a mine of information about Stoma's and will reassure you on any fears that you may have.

Once your child starts to pass motions this is normally the first step to recovery. This rate of recovery varies from one child to another. if your child's recovery seems slow do not despair, you will get there in the end. Usually your child will want to pass motions a lot more frequently than you expected, this is normal for this time. The motions maybe watery and very acidy. This will lead to one of the most common problems where the bottom gets very sore. There are numerous barrier creams to help this.
Hirschsprung's children also tend to have lactose allergies but usually out grow this.

You may also find that your child has a problem controlling the passing of motion but again this will improve in time but can be very distressing for both you and your child, especially in the older child
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Enterocolitis, what's this?

This is the most serious complication associated with Hirschsprung's. The symptoms can at times be vague but are usually abdominal distension, fever and foul smelling stools Enterocolitis must be diagnosed and treated quickly to prevent very serious complications. If caught early then this can be treated with antibiotics and re-hydration. An important part of the treatment of enterocolitis is irrigations. This is where saline is pushed up through the colon to remove the gas and stool, this is similar to an enema.

The chances of enterocolitis reduce as the child gets older but can occur both post and pre-operative.

What Next?

The good news is that 80% or more Hirschsprung's children go on to have full control of their bowels and lead a very satisfying life.

There is a slightly increased chance of your child also having Hirschsprung's children and that you may have more Hirschsprung's children. The risk is low but must be thought of. Genetic Counselling is available to those who feel the need but as this is a new field the outcome cannot be guaranteed.

A small percentage of children do continue to have problems and a second or third pullthrough maybe necessary. This is because there are still parts of the bowel that do not have ganglion cells. **Why is this?** To be honest sometimes there is no explanation. In very rare cases the grouping cells may have breaks in them and when the original pullthrough was performed the biopsies were not taken high enough up to take this into account. This is still a little understood disease, although frequent advances are being made and our knowledge is advanced regularly. Because of this you should firstly seek out an expert in this field. Then secondly do not get depressed when things do not go smoothly, there seems to be no standard progress for Hirschsprung's children except that in time most get there.

There is another 2 smaller groups of Hirschsprung's children who have total intestinal Hirschsprung's and Total Colon Hirschsprung's. In Total Intestinal, some of these children do not respond to treatment and generally their only option is a multi organ transplant. This again is a very new form of transplant and therefore has great dangers attached to it and therefore will only be performed rarely. Total Colon Hirschsprung's occurs in around 5% of HD children and is usually treated with a ileostomy and when the child is older then the pull through is performed.

The Future of Hirschsprung's Treatment?

When Harald Hirschsprung's defined the disease, apart from rectal washouts there was no real treatment. Then Dr Swenson developed the pullthrough in the 1940's. This procedure has been developed ever since and the outcome is much better than was ever thought.

New treatments for bowel control are being brought out; new medicines are being developed to firm motions. Research is ongoing at Alder Hay in the UK into Stem Cells to repopulating the bowel. There is also a large genetic study being performed by Johns Hopkins Medicine in Baltimore, Md. USA.

There is a long way to go before this Secret Disease is understood and the severe cases don't need transplants but huge steps have been made in the passed decades.

Why do we not know more of Hirschsprung's?

As we have seen, Hirschsprung's is still a disease that children can in rare instances die from. So why is it not taken seriously by people? Well let's face it what happens when we talk about going to the toilet, people giggle, look disgusted or just pretend not to hear. A lot of people don't want to know about such 'dirty' things as bowels and poo.

In the UK a baby with Hirschsprung's is born every 72 hours, in the USA it is every 15 hours and worldwide 1 in every 5,000 children have Hirschsprung's. It is 4 times rarer in girls than in boys. You are not alone; there are others out there like you.

How do I find these other Parents?

The Hirschsprung's & Motility Disorders Support Network is a charity that can put you in contact with parents like yourself**.** Through them you will be able to talk to parents worldwide who are having the same fears and problems you are having. They will also try to put you in contact with parents near you as well as arrange annual meetings.

They have various means of contact

- Web: www.hirschsprungs.info
- Email: info@hirschsprungs.info
- Phone: (+44) (0)7935 787776