Hirschsprung's Disease

The Secret Disease

Contents

- Let us Introduce Ourselves
- Hirschsprung's Disease, What is it?
- Ganglion Cells, What's their function?
- How will it be Diagnosed?
- What will happen after the operation?
- What's next for my child?
- My child is not a baby but still has bowel problems?
- The Future of Hirschsprung's Treatment?
- Why do we not know more of Hirschsprung's?
- How do I find these other Parents?

Let us Introduce Ourselves

The Hirschsprung's & Motility Disorders Support Network (HMDSN) is a charity whose aim is to support and inform parents of children with gastrointestinal conditions such as Hirschsprung's. We are based in the UK but have members living in most of the continents of the world This means wherever you are we should be able to offer you support.

We work with doctors, nurses & consultants to better understand these disabling conditions. By working together we can combine a wealth of knowledge to improve the lives of the children suffering from these conditions.

Hirschsprung's Disease, What is it?

Hirschsprung's (pronounced HURSH-sprungz) Disease is a condition affecting the large intestine. It is also known as congenital megacolon & congenital intestinal aganglionosis.

When the baby starts growing in the womb, nerves called Ganglion cells grow in the intestine, starting at the top, working their way down the intestine to the anus. In HD, these cells do not grow the whole way down the bowel but stop before reaching the anus. This disease affects 1 in 5,000 babies and is more common in boys than in girls.

You may hear various terms for HD, Short Segment, Long Segment, Total Colon, Total Intestinal & Ultra-Short Hirschsprung's. These terms describe how much of the bowel is aganglionic or lacking these cells. Do not be too confused by these terms as there is no strict definition and some specialists do not recognised the difference between Long & Short Segment HD.

Ganglion Cells. What's their function?

The intestine pushes the stool down the bowel to the anus by wave movements, properly called peristaltic movements, of contracting and then relaxing. The ganglion cells job is to allow the bowel to relax. Without being able to relax, the bowel remains constricted and narrow. No stool can pass this point in the bowel and accumulates back up the bowel. The bowel then swells giving the appearance of the tummy looking bloated. When a contrast or barium x-ray is taken, the bowel looks like a 'Y' or ice cream cone, narrow at the bottom getting wider at the top where the stool is accumulating.

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 symptoms, don't worry, the odds are it is not Hirschsprung's but what you must do is to get your baby examined by a gastrointestinal 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 a full thickness or suction biopsy of the rectum.

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 and thickness. Ganglion cells can be found in Hirschsprung's children but they may be so few in numbers that the bowel will not function properly. Sometimes more than one biopsy to may be necessary to obtain a definitive diagnosis.

The older child may have a contrast x-ray to show the state of the bowel. Depending on the age of the child anorectal manometry may be performed. This will test 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 that 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. Why this gene is faulty is the subject of a lot of research but nothing points to it being environmental and therefore caused by the parent.

You can have genetic counselling to try to determine the risk of you having other children with HD. Once you have an HD child the risk of having another is increased but most parents go on to have other healthy children. So far there is no test that can be performed to show whether your unborn child has Hirschsprung's or not.

If you have a child with HD then the odds of having another are about 4%, depending on whether the new baby is a boy or girl and whether your HD child has long or short segment. Therefore, the odds of having a healthy child next is extremely good.

What is the treatment? Will laxatives cure it?

Hirschsprung's is a very serious disease and the only current effective treatment is a surgical procedure called a pullthrough operation. This is where the affected or aganglionic part of the bowel must be removed as it will never function properly 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 called either an ileostomy 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 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 pull-through'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 the stay in hospital. Your doctor will perform the surgery that they feel most comfortable with and which they

consider give the best results for your child. Remember that the doctor does want the best for your 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 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. The 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 may be watery and very acidic. This will lead to one of the most common problems where the bottom gets very sore. There are numerous barrier creams to help this. Again, speak to your Gastro or Stoma Nurse for help.

After the period of diarrhoea, there may be constipation. It is vital that this is looked at but do not worry too much if this occurs. Again it is not too uncommon and will pass.

Later, you may also find that your child has a problem controlling the passing of motions. Again this will improve in time but can be very distressing for both you and your child, especially in the older child.

Hirschsprung's children also tend to have lactose allergies but

usually grow out of this. Some HD children do not put the expected weight on, again this is normal. Just make sure your child seems active and alert. You can discuss this with your surgical team if you have any fears.

What's next for my child?

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

A small percentage of children do continue to have problems and a second or third pullthrough may be necessary. This may be 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 ganglion cells may have breaks in them and when the original pullthrough was performed the biopsies were not taken high enough up the bowel to take this into account. This is still a little understood disease. Frequent advances are being made and our knowledge is advancing regularly. Because of this, you should firstly seek out an expert in this field. 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. They grow into healthy adults and have families of their own, mostly HD free.

If your child still has problems controlling their bowel movements and continues to have accidents when older, then an ACE or Malone may be necessary. This is where a tube is inserted from the tummy, to usually, the appendix and then into the bowel. This then allows the bowel to be cleaned with saline, from the top down, on a nightly basis. This may sound unpleas-

ant but during this problem period the child is usually clean and therefore does not feel socially excluded. Children usually do not find this a problem and can look after their ACE from an early age, some as young as 4 or 5.

There are another two smaller groups of Hirschsprung's children who have total intestinal Hirschsprung's and Total Colon Hirschsprung's. In Total Intestinal HD, some of these children do not respond to treatment and sometimes their option is a multi-organ transplant. This is a very new form of transplant and, therefore, has great dangers attached to it and therefore will only be performed as a last resort. Total Colon Hirschsprung's occurs in around 5% of HD children and is normally treated with an ileostomy and when the child is older then the pull through is performed. This can vary depending on the consultant involved and what they feel is in the best interests of the child.

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 it does not. The biopsy taken may have shown ganglion cells but not enough biopsies were taken to show the areas without cells. The doctor may have thought that the problem was the child withholding the stool, which 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 has improved. This makes

late diagnosis rarer.

If your child is having bowel problems, the odds are it is not Hirschsprung's. Whatever the problem is, it must be fully investigate and diagnosed by a gastrointestinal specialist.

Enterocolitis, what is this?

This is the most serious complication associated with Hirschsprung's and is also called HEC or HAEC, Hirschsprung's entercolitis. The symptoms can at times be vague but are usually

- **Abdominal distension or Swollen Tummy,**
- **Fever**
- **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 reduces as the child gets older but can occur both post and pre-operative.

If you suspect your child has HEC then please take them to your gastrointestinal consultant or, if they are not available then to A & E at a main hospital.

The Future of Hirschsprung's Treatment?

When Harald Hirschsprung's in the 19th century, defined the disease, apart from rectal washouts, there was no real treatment. Then Dr Swenson developed the pullthrough in the late 1940's. This procedure has been developed ever since and the outcome is much better than was ever thought possible. There is still a long way to go before this disease is properly understood and the outcome can be properly predicted.

New treatments for bowel management routines are being developed. If your child is having problems after the operation talk to your surgical team about it. There are new medicines which are being developed to firm bowel motions. Research is ongoing into getting Stem Cells to repopulate the bowel. There is also a large genetic study being performed.

There is a long way to go before this Secret Disease is understood and the severe cases do not need transplants but huge steps have been made in the past decades. Being a Secret Disease, research funding is scarce.

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 more 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 pooh. Everyone goes to the toilet but who wants to talk about? People think that if a child is constipated then just give it some laxatives, more roughage or prunes and things will be fine. This can in fact be the wrong thing to give an HD child.

In the U.K. 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.

As you may of found out already, no one really understands the problems of these children and the families involved unless you have experienced it yourself.

We have various means of contact

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