Anorectal Malformations and Hirschsprung disease

Article by Eunice Gribbin RN and co-founder of The Bowel Group for Kids

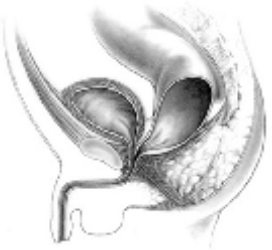
What is an anorectal malformation?
These are congenital abnormalities in which the anal opening in the baby's bottom is absent or abnormal in size or position *(also known as imperforate anus)*, so there may be limited or no exit for faeces. The condition varies greatly in severity from a somewhat tight anus, with little else wrong; to major malformations involving a single opening for anus, vagina and bladder known as a "cloaca" in girls, or the formation of a fistula between the bowel and bladder in boys.

The terminology used to describe these conditions has changed a number of times, but your doctor may talk of "high", intermediate" or low malformations. There may be other associated problems that also need to be dealt with; the most common associated problems are collectively known as the "VACTERL" or "VATER" association.

Diagnosis should be straightforward and made shortly after birth when a full newborn assessment of the baby is performed by a midwife or paediatrician. This will confirm the presence and position of the anus in the newborn baby and is essential to exclude the diagnosis of an anorectal malformation. In the case of a high malformation, a baby boy often passes urine stained with meconnium and a baby girl will pass meconium through the vagina. Because the abnormal connections are too small for the large bowel to empty, most babies will develop a bowel obstruction. If the condition is undiagnosed at birth, some or all of the signs to look for would be the baby developing an increasingly distending abdomen, poor feeding and possibly bilious vomiting.

The surgical treatment depends greatly on the severity of the condition. These babies mostly require surgery at birth: either a complete repair to fashion or reposition the anus, or a temporary stoma until definitive surgery can repair the defect when the baby is older.

As with Hirschsprung disease, it is important to make an early diagnosis to prevent the development of enterocolitis which can be life threatening. Therefore, any child that presents with a distended abdomen must be attended to immediately.



Male high imperforate anus

Female Cloaca

What is Hirschsprung Disease (HSCR)?

Hirschsprung disease is a relatively common form of bowel obstruction due to the inability of the gut to relax. This results in the baby being unable to pass faeces of variable severity, which causes the bowel to dilate. The underlying problem is the absence of ganglion cells, which are the nerve cells required to co-ordinate peristalsis, necessary to move faeces through the gut. There are varying degrees of Hirschsprung disease. The most common being short-segment which affects the rectosigmoid region

of the colon *(bottom part of the large bowel)*; total colonic involves the entire colon and about 10cm of Ileum *(last part of the small intestine)* and extremely rarely, the entire gut may be affected which is known as total intestinal aganglionosis.

The condition was only formally described in the 1950's, is largely genetic and inherited in a complex fashion, but is still not fully understood. A number of other congenital conditions may be inherited with it, most importantly Down syndrome.

The newborn baby with Hirschsprung disease nearly always requires surgery, which may be definitive, or else involve a temporary stoma before later repair when the baby is older. It is more common in boys than girls; a ratio of 4:1. The risk is higher in siblings with total colonic involvement.

A typical presentation of Hirschsprung disease is:

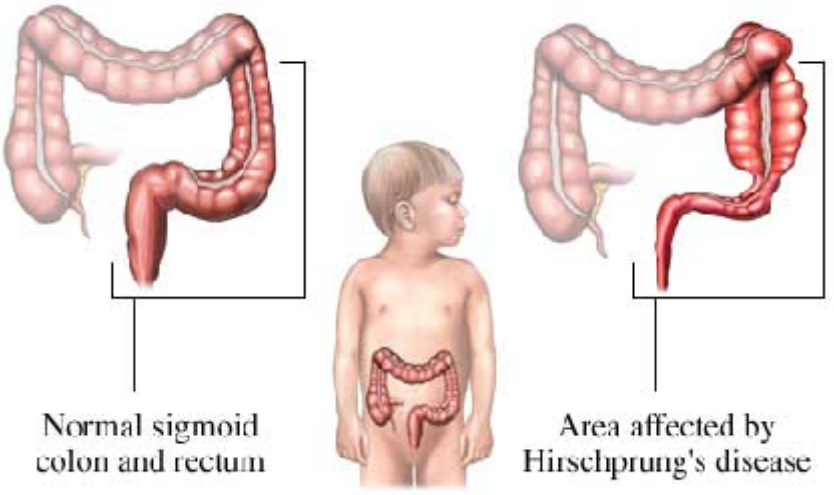
- failure to pass meconium in the first 24-48 hours after birth
- abdominal distension
- possible bilious vomiting
- this may be followed by infrequent, explosive bowel movements or difficulty in passing a bowel motion

The diagnosis should be made on the above-mentioned clinical grounds which would prompt the doctor to order any or all of the following tests which maybe suggestive of Hirschsprung disease:

- Abdominal x-ray
- Barium enema
- Manometry to measure the pressures of the rectum
- A rectal biopsy is essential to confirm the absence of ganglion cells under a microscope

It is important to make an early diagnosis to prevent the development of enterocolitis which can be life threatening. To relieve the obstruction, washouts are very effective in the vast majority of cases and they can be continued while waiting for the biopsy report.

Half of the children are diagnosed in the first three days, ¾ by the end of the first week, 7/8 by the end of the first month, and over 90% by the first birthday. Diagnosis has certainly got earlier over the last four decades of the 20th century, perhaps as a result of the development of paediatric surgery as a specialty and the improvement of services. Occasionally people are diagnosed at later ages. Audits from cases in the UK were that only 1% of rectal biopsies from children over a year of age were positive. It is very rare for people to be diagnosed in adult life.



Where to get support and information:

The Bowel Group for Kids (BGK):
The BGK is a registered national organisation dedicated to providing education, practical information and support, to families of children born with Hirschsprung's disease (HSCR); anorectal malformations (ARM) and associated conditions.

The organisation was formed in 1995 by a group of dedicated parents and professionals who were drawn together by their shared experience and frustration at the lack of information and support available to parents whose baby was born with one of these conditions. Even though they are very different in origin and presentation, they are congenital, rare and may result in very similar long term complications and problems.

Most families say what they like about being part of the BGK is no longer feeling alone; the newsletter gives insight into real stories and articles specific to the condition; support on the forum; national annual conferences and support when they need it with access to an impressive committee many of whom have a medical or nursing background.

When parents first learn that their child has HSCR/ARM they are naturally distressed. It can be confronting to be faced with the uncertainty of conditions they may not have heard of before and especially the need for surgery for their newborn baby, BUT, from that first moment, to sharing the everyday challenges of bringing up a child with HSCR/ARM, the Bowel Group for Kids is here to offer friendship, support and information throughout your child's life for the challenges they may encounter.

NB: Whilst your doctors provide the medical advice; the Bowel Group for Kids provides the day to day practical living experience. All BGK information and resources are neither intended nor implied to be a substitute for professional medical advice.

To find out more or to join the Bowel Group for Kids, contact us by:

Ph: **61 2 4659 6067**
Mobile: **0431 857 188**
Email: **enquiries@bgk.org.au**
Website: **www.bgk.org.au**

Glossary of terms:

Bowel:	consists of small and large Intestine (colon), sometimes referred to as colon, gut or gastrointestinal tract
Bilious vomiting:	vomiting bile
Congenital:	present at birth
Dilatation:	expansion of an organ or vessel
Distension:	uncomfortable swelling in the intestines usually caused by excessive amounts of gas and fluid
Enterocolitis:	potentially life threatening Inflammation of the intestines
Faeces:	poo, stools, motions (and any other terms used at home)
Fistula:	abnormal tube like connection between two hollow organs such as between bowel and bladder
Ganglion cells:	these cells are normally found in the bowel and are very important for the process of peristalsis
Meconium:	first stool passed by a newborn infant normally occurring in the first 24-48 hours after birth. It is characterised by its dark green sticky appearance
Peristalsis:	wave like movement of the bowel which pushes food along the intestines towards the rectum
Stoma:	Greek word meaning mouth or opening

VACTERL/VATER anomalies include:

V	**V**	Vertebral defects
A	**A**	Anal atresia
C		Cardiovascular
T	**T**	Trachea
E	**E**	(O)esophageal fistula/atresia
R	**R**	Renal
L		Limb