Anorectal anomalies and Hirschsprung disease (including stomas)

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Abstract
Anorectal malformations are congenital anomalies caused by a failure of the hindgut to open into an adequate position on the perineum. They are often associated with congenital anomalies in other systems. A thorough understanding of the anatomical aberration will allow surgical planning and correction of the defect. This article outlines the principle considerations in diagnosis, early and definitive management of these defects.

Hirschsprung disease is a congenital anomaly caused by a failure of development of the enteric nervous system and consequent absence of ganglia. Marked spasticity of the bowel and functional bowel obstruction ensue. This article discusses recognition, diagnosis, initial and definitive management.

In both conditions sequelae include life-long incontinence. Optimization of long-term outcomes is therefore important and meticulous surgical management in early life should be coupled with careful follow-up.

Keywords Anaganglionosis; anorectal malformation; Duhamel procedure; imperforate anus; PSARP; Soave procedure; Swenson procedure; transition zone; VACTERL association

Introduction
Anorectal malformations (ARM) and Hirschsprung disease (HD) together constitute a significant part of paediatric colorectal surgical practice. Both are congenital anomalies caused by anatomical (in the case of ARM) or physiological (in the case of HD) aberrations that require meticulous surgical management. The sequelae of congenital anomalies are likely to affect a neonatal patient for decades onwards and careful attention to follow-up is required if long-term functional outcomes are to be optimized.

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Anorectal malformations
An ARM is a congenital abnormality in which the terminal portion of the hindgut fails to open into the correct position on the perineum or does so but with an inadequate calibre. The incidence varies between areas and is approximately 1:3500—1:5000. The cause remains unknown.

Spectrum of disease
ARMs fall into a spectrum in both males and females.

Males: the spectrum of abnormalities and the relationship between the hindgut and the urethra are illustrated in Figure 1.

The most minor defect in a male is a perineal fistula. Here, the hindgut opens onto the perineum but, since it is anterior to the normal position, it is incompletely surrounded by sphincter. Next are malformations in which the hindgut terminates as a fistula into the urethra. This may be a recto-bulbar urethral fistula or a recto-prostatic fistula. At the most severe end of the spectrum are defects in which the hindgut ends in a fistula into the bladder neck (recto-bladder fistulae) or bladder (recto-vesical).

Of clinical importance is the anatomical relationship between the hindgut and the urinary tract; for the recto-perineal fistulae, there is always some distance between the two structures. For the other defects this is not the case. For the recto-bulbar urethra and the recto-prostatic fistulae, not only do the hindgut and urethra lie in close proximity, they are almost parallel. In these patients, the walls of the hindgut and the urethra effectively share a ‘common wall’ which may extend for some distance necessitating particular care in dissection. The trajectory of the hindgut relative to the urinary tract for the recto-bladder neck fistulae tends to be more perpendicular and a common wall is less of an issue.

In addition to this spectrum in males, there are some special situations worthy of mention:

- ‘ARM with no fistula’. This occurs when the hindgut is blind ended and is seen most frequently in Down syndrome. Although there is no fistula into the urethra, there is often a common wall, as per recto-bulbar urethral defects.
- Stenosis. Here the hindgut opens onto the perineum through the sphincters but is stenotic either at skin level (anal stenosis) or higher up at the level of the dentate (rectal stenosis).
- Bucket handle defect. Here, the hindgut does not enter the urinary tract but lies just under the skin surface. The overlying skin has a typical appearance in which the midline raphe becomes prominent (hence the name bucket-handle). This is similar to a perineal fistula.

Females: the spectrum of abnormalities in females is illustrated in Figure 2. At the minor end is the perineal fistula. In keeping with its male counterpart, the hindgut opens onto the perineum but is incompletely surrounded by sphincter, and may also be stenotic.

Since the introitus and vagina, rather than the urethra, lie anterior to the hindgut, the next most severe defects are the recto-posterior forchette fistula (in which the hindgut enters at the posterior corner of the introitus) and the recto-vestibular
fistulae (in which the hindgut enters introitus itself and so can be seen at a point distal to the hymen). As with fistulae that enter the urethra in males, there is often a common wall (here between hindgut and the vagina) that makes delicate dissection essential.

The most severe defect in females is a cloaca. Here, not only does the hindgut open directly into the vagina, but the urethral orifice is also abnormal, lying high on the anterior vaginal wall (‘female hypospadias’). In cloacae therefore, urinary, genital and intestinal structures all drain into a common channel. The point at which these two structures open into the vagina may be relatively close to the skin (producing a short common channel) or may be much more proximal (producing a long common channel). The length of the common channel correlates with prognosis.

Clinical presentation

Whilst ARMs are almost always clinically obvious when inspecting the perineum at neonatal examination, the frequency with which they are missed during the neonatal assessment is surprisingly high. In part this is because some abnormalities (e.g. recto-perineal fistulae or stenoses) have the potential to transmit stool onto the newborn perineal skin. If an unsuspecting clinician fails to clean away this meconium then the defect will remain undiagnosed. A less common scenario occurs when the abnormality lies at the level of the dentate line (for example rectal stenosis) but the external structures look normal. Delay in diagnosis is however seen even at the most severe end of the spectrum and one recent report from the UK demonstrated that up to 50% of newborns with ARM were missed at the neonatal examination.1

Assessment

Once an abnormality has been identified, the primary objectives are to:

- determine if a stoma is needed before definitive repair or not
- look for associated abnormalities.

Stoma or not? ARMs in which the position of the hindgut is clinically obvious and clearly separated from the urinary tract are deemed ‘low’ and considered amenable to definitive repair in the neonatal period. This effectively means recto-perineal fistulae.

In contrast, defects at the severe end of the spectrum have a close anatomical relationship between the hindgut and the urinary tract. Furthermore, the distance of the hindgut from the perineal skin may be significant (indeed the hindgut may be so high that it cannot be reached safely from a perineal incision). Surgical exploration without a clear understanding of anatomy in the neonatal period is hazardous. This group of defects is deemed to be ‘high’ and by definition requires a temporary stoma.

Categorization of abnormalities into low and high can be performed by clinical assessment alone in almost all patients. If a fistula is seen to enter the perineum, or a bucket handle is observed, one can be sufficiently confident of the position of the hindgut to consider definitive repair if the baby is otherwise well. If meconium is seen in the urinary stream (this typically takes 24 hours or so) then a fistula is proven and a stoma required. Other clinical signs indicating a high abnormality are poorly formed buttocks and perineal musculature, or a foreshortened sacrum on plain X-ray.

Figure 1 Recto-perineal, recto-bulbar urethral, and recto-bladder neck fistulae. (From Pena A, Surgical Management of Anorectal Malformations, 1989. With kind permission of Springer Science & Business Media.)

Figure 2 Recto-perineal, recto-vestibular and cloacal defects. (From Pena A, Surgical Management of Anorectal Malformations, 1989. With kind permission of Springer Science & Business Media.)
In only a few neonates does sufficient doubt exist as to the need for a stoma or not that radiology is needed. The most useful investigation is a prone lateral shoot through with a radio opaque pellet applied carefully to the point where the anus might have been expected. A catch for the unwary is the need for sufficient pressure upstream to a fistula for it to be delineated and the examination is best performed at around 24 hours of age.

In some centres, ultrasound has been used although this requires particular experience on the part of the radiologist; too much pressure on the perineum will compress tissues giving an erroneous appearance of a low defect. Other investigations such as MRI are not recommended as the lack of pressure above a fistula is likely to produce misleading but persuasive results.

When required, formation of a stoma for ARM will allow the child to decompress (and then feed) and also allows detailed investigation of the anatomy of the ARM to inform surgical planning using a distal loopogram. Here, water-soluble contrast is instilled into the terminal portion of the hindgut from above via a mucous fistula. If sufficient pressure is applied to the contrast, it will enter the urinary tract, demonstrating a fistula if present.

Although cloacae are at the most severe end of the spectrum they may be missed, or mislabelled as a recto-vaginal fistula. In reality, recto-vaginal fistulae are rare and are often cloacae in which the urethral abnormality has not been recognised. Since surgical correction of cloacae often entails mobilization of the urethra at the same time as the hindgut, and all cloacae require a stoma, correct diagnosis is of practical importance.

**Associated anomalies:** a well-recognized association is the VACTERL association which is comprised of Vertebral, Anorectal, Cardiac, Tracheo-Oesophageal, Renal tract and Limb abnormalities. The likelihood of these abnormalities co-existing increases with the severity of ARM but even minor ARMs can be affected.

All babies should have spinal and sacral plain films, spinal ultrasound (to look for evidence of bony anomalies or a tethered cord), a full cardiology assessment, renal tract ultrasound and limb examination. The degree of sacral foreshortening can help infer prognosis and is therefore an important clinical feature to guide discussion with families.

The presence of one congenital anomaly increases the likelihood of others and a careful neonatal examination is needed. Cloacae, and to a lesser extent, any other ARM in a female may be associated with gynaecological abnormalities such as bicornuate uterus, vaginal septum or even vaginal agenesis. In cloacae, collections of fluid in the uterus (hydrometrocolpos) are sometimes seen and may be so marked that ureteric compression and renal impairment occurs. Drainage of this fluid with a pigtail catheter placed through the abdominal wall will usually address this emergency.

Other less common associated abnormalities include the association of rectal or anal stenoses with an abnormal sacrum and the development of a presacral mass (Currarino triad). For this reason, MRI is recommended in these defects.

**Management**

**Neonatal placement of stoma:** for those abnormalities deemed to be high, a stoma is formed. The placement of the stoma at the junction of the descending colon and the sigmoid colon allows enough proximity for contrast to be injected with sufficient pressure to demonstrates a urinary fistula but not so low that it impedes surgical repair (Figure 3). This is the divided descending colostomy.

**Definitive surgical management:** once the neonate has been fully assessed clinically, had a stoma formed (if needed) and undergone a distal loopogram to delineate anatomy, definitive surgery is undertaken. The posterior sagittal anorectoplasty (PSARP), described by Pena in 1982, involves a midline incision though the perineum. Pelvic muscles and subtending innervation are already paired. A midline incision minimizes further iatrogenic injury to these structures whilst at the same time allowing excellent exposure.

By using muscle stimulation throughout the dissection, the paired musculature can be readily identified on both sides. As well as seeing levator ani, sphincteric muscle is also seen to run parallel to skin at a superficial level (probably representing external sphincter) and then perpendicular to the skin (probably representing an extension up to the levator).

PSARP allows safe exposure of the hindgut and its meticulous dissection from the urethra where necessary, mobilization of an adequate length of bowel and precise placement of the ‘neoanus’ within the sphincteric tissue with accurate re-apposition of pelvic musculature (Figure 4).

For the most severe defects, the distal loopogram demonstrates that the hindgut lies so high within the pelvis that it cannot safely be reached with a PSARP approach alone. For these patients, the PSARP can be combined with an abdominal approach. Although an open approach through a Pfannenstiel incision may be employed, a laparoscopic approach provides an

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**Figure 3** Divided descending colostomy. (From Pena A, Surgical Management of Anorectal Malformations, 1989. With kind permission of Springer Science & Business Media.)
excellent view into the pelvis and facilitates safe division of these high fistulae. Defects likely to benefit from this approach are recto-bladder neck and high recto-prostatic, not only because the fistulae are easily accessible from above but also because as described above, the length of the common wall is short. Such a combined approach means that the PSARP incision need only be long enough to allow an entrance to the peritoneum since a full exploration is not required. Precise placement of the neorectum and neoanus within the sphincteric mechanism, and tacking of the posterior wall to the sphincters, can then be performed.

The laparoscopic approach has also been combined with a pull through using a perineal laparoscopic port but this arguably does not allow sufficiently accurate placement of the bowel within the musculature, is often associated with prolapse and risks injury to the urethra and distal ureters. Pull though using a port is no longer recommended by the current authors.

One further consideration of laparoscopic ligation of the fistula is the importance of not leaving a significant stump of intestinal tissue on the urinary tract. Doing so predisposes to stone formation, potential sepsis and, given the risks of long-term exposure of colonic mucosa to urine, even malignancy.4

In most centres a programme of anal dilatations commences at approximately 2 weeks postoperatively. This reduces the likelihood of cicatrization the circular wound at the point where the neoanus is Anastomosed to the skin. Dilatations are undertaken by the parents with support from nurse specialists and continued until the faecal stream is rediverted to the perineum following stoma closure. This usually takes place after 6 weeks or so.

Long-term outcome

Long-term outcomes now exist for this condition which demonstrate that:

- Low abnormalities tend to be predisposed to constipation although almost all are continent of both faeces and urine.
- Approximately 60–80% of patients with a high abnormality have impaired continence. Management of this scenario must be proactive. If the child is to avoid the serious psychological consequences of soiling, symptoms should be controlled before the child reaches school age. In some, control can be achieved by artificially thickening the stool with medication and/or dietary manipulation. In others, rectal irrigations can be employed to empty the bowel fully once every 24 hours or some form of antegrade continence enema procedure is performed such as a Malone procedure.
- Cloacae are worthy of particular mention; in addition to a relatively high risk of impaired continence, the degree and frequency of associated urinary tract and gynaecological abnormalities is such that a multidisciplinary approach and involvement of experts in both urological and gynaecological aspects is essential for good outcome. The relative infrequency with which these abnormalities appear means that such a team does not always exist in every centre.
- The long-term sequelae do not tend to improve spontaneously with time. In all patients, early detection and correction of symptoms is crucial.

Hirschsprung disease

Hirschsprung disease (HD) is a congenital motility disorder affecting the intestine. It is characterized by an absence of ganglion cells (aganglionosis). The role of ganglion cells is as a relay point for the enteric nervous system (ENS) and an absence leads to spasticity of the bowel and functional obstruction. The incidence is about 1:5000 live births.

Spectrum of disease

The distribution of HD is such that the most distal part of the rectum is always affected. The length of the segment varies and although the proximal extent of aganglionosis most often lies.

Figure 4 PSARP allows safe identification of the hindgut through a midline dissection. Muscle fibres of sphincter complex are clearly seen. Stay sutures have been placed on the hindgut wall to allow it to be safely opened, fistula identified and divided. The hindgut can then be dissected away from the urinary tract. (From Pena A, Surgical Management of Anorectal Malformations, 1989. With kind permission of Springer Science & Business Media.)
within the rectosigmoid (classic segment), it may involve the whole colon (total colonic aganglionosis) and even small bowel. Above the aganglionic zone lies the transition zone (TZ) in which ganglion cells are present but other ENS abnormalities are still seen. Above the TZ lies the ganglionic bowel. Because of the downstream functional obstruction, the ganglionic bowel is often distended just above the TZ (Figure 5).

The eyes only see what the mind is prepared to comprehend

-Henri Bergson

When Harald Hirschsprung first recognized this disease as an entity he believed the obviously distended segment to be the cause of symptoms. Since this is the ganglionic segment, no histological abnormality was found within it. Although spastic, the aganglionic segment looks almost normal in comparison to the distended bowel above. The aganglionic segment was not recognized as the true cause of the problem for a further 60 years. This vignette underlines the need to think laterally when trying to understand as yet unexplained disease processes.

Clinical presentation

A majority of patients with HD present in the neonatal period with a distal obstruction evidenced by abdominal distention, bilious (i.e. green) vomiting and delayed passage of first stool (or meconium). Since more than 98% of term neonates will have passed meconium within 24 hours, a delay beyond this in conjunction in association with other clinical features is considered significant. The risk of transmission to offspring is 1–33% and varies with length of segment and gender of the index case. Since there is a degree of hereditability a positive family history is sometimes seen and must always be sought.

Other congenital abnormalities must be excluded on clinical assessment, as there are a number of syndromes associated with HD, most notably Down syndrome. A small group of genetic defects have now also been described including the RET mutation. Identification of a RET mutation raises the possibility of multiple endocrine neoplasia type 2b (MEN 2b) and therefore medullary thyroid cancer.

HD may also be associated with enterocolitis characterized by systemic sepsis, distention and offensive loose stool. Although some clinical features of enterocolitis are shared with infective gastroenteritis, the severity and rapidity of progress are such that this can be a lethal condition and must be considered as a diagnosis in any child known to have HD or indeed in a newborn presenting with these features.

Once other differential diagnosis of a distal obstruction have been excluded such as neonatal sepsis, missed ARM (see above), intestinal atresia and meconium ileus, rectal washouts are commenced using warm saline delivered though a soft rectal tube and delivered by experienced staff. As long as the volume instilled on each washout (perhaps 50 ml) equates reasonably closely to that returned there is no upper limit on the total volume of volume of washouts to be used. This technique produces adequate decompression in most patients and has radically changed the management of HD by reducing the need for a stoma. If severe enterocolitis is present, the segment is too long to allow washouts, or in the relatively rare event that washouts fail in classic segment disease, a stoma should be considered.

Although in the recent past, stoma formation was considered the norm following HD diagnosis, stomas are associated with frequent complications and additional surgery. They are to be avoided in HD where possible.

A less common presentation is the older child with constipation. Since the number of older children with constipation without an obvious cause is high, indications as to which of these patients require biopsy have been produced; patients with a history of delayed passage of meconium, constipation from the first few days of life, a positive family history, previous enterocolitic symptoms, failure to thrive and an explosive decompression should be considered as good indications in a constipated child.

Diagnosis and investigation

The gold standard is rectal biopsy. For neonates, this minor procedure can be performed on the ward, but with antibiotic cover and consent. Whilst an absence of ganglion cells is diagnostic, the presence of thickened nerve trunks, staining brown with acetylcholinesterase (AchE) is considered to be a further feature supporting the diagnosis (Figure 5).

Anorectal manometry is said by some to be of value in the diagnosis but is insufficiently accurate and does not obviate the need for biopsy. A contrast enema is performed in the neonatal phase in some but not all centres. Whilst it too does not reduce the need for biopsy, it can give information about the position of the TZ. When the TZ is in the classic position i.e. recto sigmoid, this investigation may help surgical planning. If the whole of the colon is affected, contrast enemas are notoriously misleading and must be interpreted in the context of the clinical setting. One further benefit of the contrast enema is to inform the management of the rectal washouts by providing an estimate of the volume needed to reach the distended bowel above the TZ.

Definitive management

Following a period of washouts to reduce proximal distention, definitive surgery is performed. This is typically undertaken after 4–8 weeks although in some centres this now takes place in the neonatal period. Broadly there are three main operations in current use (Figure 6). In all procedures, the principles are to remove aganglionic bowel, to accurately identify and preserve the anal canal and to pull though the ganglionic bowel to a point at or just above the dentate line.

- Swenson procedure. This was the first described procedure and involves the mobilization of the aganglionic colon from above the dentate line. Dissection continues proximally in a plane outside the serosa until distended bowel is reached. Full-thickness biopsies are then sent to a waiting pathologist for frozen section. If histological appearances demonstrate that a point above the TZ has been reached, this section of bowel is pulled through and anastomosed to the dentate line and the abnormal bowel downstream is resected.

This has become increasingly popular again in recent years, especially in the USA, as previous concerns about potential iatrogenic injury to the sacral outflow have been addressed with strict maintenance of a plane of dissection immediately on the serosa of the rectum. This procedure is now performed transanally with or without laparoscopic assistance (see below).
**Figure 5** (a) Resection specimen and histology from bowel with Hirschsprung disease demonstrating distended, but histologically normal, ganglionic bowel (on left), tapering Transition Zone (TZ) (central) and spastic aganglionic bowel (right). The ganglionic zone has a normal ENS (b) with ganglion cells (c) and no hypertrophic nerves. The Transition Zone (d) has ganglion cells but also hypertrophied nerves. The aganglionic zone has no ganglion cells but has hypertrophied nerves (e). The hypertrophied nerves stain brown with ACHE (f).
Soave procedure. This technique was developed in order to protect the sacral outflow previously considered to be at risk with the Swenson. Again the dissection begins at a point just above the dentate but proximal dissection occurs in the submucosal plane (i.e. the circular and longitudinal muscles are left in situ in the early phases of the dissection). This means that the layer of bowel wall that produces the spasticity is the layer that remains. In order to mitigate the effect of spasticity, the cuff of muscle is split. Failure to adequately divide this tissue can lead to obstructive features (including constipation) and therefore a predisposition to enterocolitis. Again this procedure is now generally performed transanally (with or without laparoscopic assistance). This is now the most common procedure performed in the UK.

Since the majority of the dissection in both of these procedures is transanal, the sphincteric mechanism is at risk of iatrogenic injury caused by excessive or prolonged retraction and the surgeon must be meticulous about preserving and dentate line and not starting the dissection too low.

Duhamel procedure. This technique is involves mobilization of the ganglionic bowel down to the dentate line through a plane created retro-rectally. An incision in the native rectum along the dentate line allows the ganglionic bowel to be anastomosed to the back of the native rectum. Using a stapling device, a pouch approximately 6 cm in length is formed which is intended to act as a reservoir for stool. The aganglionic bowel above the pouch is resected.

The pouch comprises of aganglionic bowel anteriorly and histologically normal, ganglionic bowel posteriorly. Since a section of aganglionic bowel remains there is an increased risk of obstructive features (including constipation) and therefore a predisposition to enterocolitis. In some patients, the pouch can become distended or a spur can form at the junction of the ganglionic and aganglionic bowel at the top of the pouch again leading to obstructive features.

There remain controversies; the choice of procedure to be undertaken is a source of debate; proponents of the Swenson procedure are keen to avoid leaving aganglionic bowel in situ and

Figure 6 Main operations in current use: (a) Swenson procedure, (b) Soave procedure and (c) Duhamel procedure.
have a lower incidence of obstructive problems post-operatively. Proponents of the Duhamel claim avoidance of injury to the sphincteric mechanism and an increased likelihood of preservation of continence.

Proponents of laparoscopic assistance advocate the avoidance of inadvertent twist when pulling through the bowel, the ability to mobilize at least some of the bowel from above, and the potential to obtain biopsies as advantages. Proponents of a pure transanal approach claim the addition of laparoscopy is not always needed.

The surgeon requires confirmation from the pathologist that the bowel from which the frozen section is sent is histologically normal (i.e. above the TZ). This must be a full-thickness specimen to confirm the presence of ganglion cells and normal-sized nerves (less than 40 μm). The boundary between histologically normal bowel and TZ is not, however, straight and it would be possible for one quadrant of the circumference of the bowel to be histologically normal but at the same level the other three quadrants to be abnormal. One approach is to examine all four quadrants histologically, but although histological abnormalities are more frequently seen, their relevance is not always known.

Outcomes
Outcome is predicated by length of segment (the shorter the segment, generally the better the outcome), the presence of syndromic features and technical factors such as adequate preservation of the anal canal and sphincteric mechanism. As described above, the choice of pull through technique has a direct impact on the profile of the postoperative problems encountered.

Even though the ganglionic bowel is theoretically normal, there are a number of patients in whom even this segment fails to function with entirely normal motility and ideally a functional, rather than a histological, evaluation of the bowel to predict motility would be used.

The cause of enterocolitis remains elusive and the condition incompletely understood. A number of patients have episodes of enterocolitis even following surgery. This is much more likely to be seen in children with obstructive features.

### REFERENCES


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