1. Introduction and History

Anorectal malformations comprise a wide spectrum of diseases, which can affect boys and girls, and involve the distal anus and rectum as well as the urinary and genital tracts. Defects range from the very minor and easily treated with an excellent functional result, to those that are complex, difficult to manage, are often associated with other anomalies and have a poor functional result.

Imperforate anus has been a well-known condition since antiquity. For many centuries, physicians, as well as individuals who practiced medicine, created an orifice in the perineum of children with imperforate anus. Those that survived most likely suffered from a type of defect that would now be recognized as “low”. Those with a “high” defect did not survive that treatment. The written accounts describe very few patients, so it is likely that most patients died without treatment. (1) Successful maneuvers involved rupturing an obstructing membrane with a finger or the point of a knife (2) and evolved to an incision to find the bowel in the perineum or in the hollow of the sacrum. The first inguinal colostomy was performed in 1783 (3), but most infants died, and colostomy was considered as a method of last resort. In the mid-1900’s, several authors suggested that the peritoneum be opened if the bowel was not encountered from below. (4-7) In the mid-1900’s, single-stage abdominoperineal procedures became popular (8-9) and usually involved resection of the rectosigmoid colon.

Shortly after these reports, Stephens described a procedure which emphasized passage of the rectum within the puborectalis sling. (10) Until the early 1980s, this surgery and its modifications was the
standard approach, but involved a blind dissection near the posterior urethra. A progressive increase in the length of the perineal incision to gain exposure eventually led to the posterior sagittal anorectoplasty, \(^{(11)}\) which was rapidly adopted because it allowed full visualization of the sphincteric complex and more clearly showed the relationship of the rectum to the urologic system and the surrounding vital structures.

Amussat, in 1835 was the first to suture the rectal wall to the skin edges, which could be considered the first anoplasty. During the first 60 years of the 20th century, surgeons performed a perineal operation without a colostomy for so-called low malformations. High imperforate anus was usually treated with a colostomy performed in the newborn period, followed by an abdomino-perineal pull-through some time later in life, but surgeons lacked objective anatomic guidelines. Unfortunately this left many patients incontinent and was not an appropriate solution for the spectrum of malformations.

The surgical approach to repairing these defects changed dramatically in 1980 with the introduction of the posterior sagittal approach, which allowed surgeons to view the anatomy of these defects clearly, to repair them under direct vision, and to learn about the complex anatomic arrangement of the junction of rectum and genitourinary tract \(^{(12-17)}\). This has become the predominant surgical method for anorectal anomalies. In cases in which the rectum is very high, an abdominal approach is also needed and laparoscopy is appropriate for certain cases.

2. Classification

The classification system shown in Table 1 is purely descriptive and has therapeutic and prognostic implications. The anatomic types are depicted in Figures 1-9. In boys, 85% have a rectourinary fistula. In terms of low lying anomalies, 35% of boys have a perineal fistula, where as 93% of girls have an external fistula. The most common defect in females is rectovestibular fistula. Most high anomalies in girls are cloacas; a high anomaly with a rectovaginal fistula is exceedingly rare \(^{(18)}\) and in the literature likely many such patients are misclassified. Cloacal malformations are more common than previously thought, most likely because patients were previously misdiagnosed as having a rectovaginal fistula. \(^{(19)}\)

### Table 1: Classification of Anorectal Malformations

<table>
<thead>
<tr>
<th>Gender</th>
<th>Male</th>
<th>Female</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Perineal fistula</td>
<td>Perineal fistula</td>
</tr>
<tr>
<td>Anatomic Type</td>
<td>Rectourethral fistula</td>
<td>(Recto)Vestibular fistula</td>
</tr>
<tr>
<td></td>
<td>Bulbar Prostatic</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Rectovesical fistula (bladder-neck)</td>
<td>Persistent cloaca</td>
</tr>
<tr>
<td></td>
<td></td>
<td>3 cm common channel</td>
</tr>
<tr>
<td></td>
<td></td>
<td>&gt; 3 cm common channel</td>
</tr>
<tr>
<td></td>
<td>Imperforate anus without fistula</td>
<td>Imperforate anus without fistula</td>
</tr>
<tr>
<td></td>
<td>Rectal atresia</td>
<td>Rectal atresia</td>
</tr>
<tr>
<td></td>
<td>Complex defects</td>
<td>Complex defects</td>
</tr>
</tbody>
</table>

**Figure 1:** Perineal fistula (low malformation) in male
Figure 2: Rectourethral fistula
A) Bulbar
B) Prostatic

Figure 3: Bladder-neck fistula

Figure 4: No

Figure 5: Perineal fistula in female

Figure 6: (Recto)Vestibular fistula
Figure 7: Vaginal fistula
A) Low                                      B) High

Figure 8: Cloaca
A) Short common channel (<3cm)   B) Long common channel (>3cm)

Figure 9: Rectal Atresia
3. Epidemiology and Incidence

Most authors have written that the average incidence worldwide is 1 in 5000 live births, (20) although the condition is more common in some areas such as Africa because of high fertility rate and associated malnutrition and poor antenatal care. Anorectal malformations are slightly more common in boys, and boys are twice as likely as girls to have higher anomalies. Some families have a genetic predisposition, with anorectal malformations being diagnosed in succeeding generations. (21)

In addition, imperforate anus occurs in association with several syndromes. (22) Although imperforate anus may occur as an isolated malformation, it coexists with duodenal atresia, tracheoesophageal fistula, vertebral and renal anomalies, Down syndrome, and congenital heart disease. Patients with Down syndrome usually have a unique anomaly - imperforate anus with no fistula. (23) Approximately 60% of patients have some form of associated urologic malformation. (19)

4. Embryology

The cause of anorectal malformation is unknown. Anorectal anomalies occur as a result of failure of development of the cloacal membrane or failure of recanalization of the secondarily closed anal canal during embryonic life. The cloaca in the embryo is a cavity into which open the hindgut, tailgut, allantois, and, later, the mesonephric ducts. The cloaca is first formed at around 21 days of gestation. It is U-shaped, with the allantois lying anteriorly and the hindgut posteriorly. The septum in the middle grows downward, fusing with lateral folds (Rathke’s plicae) until it joins the cloacal membrane. In this six week process, a urogenital cavity is created anteriorly and an anorectal cavity posteriorly. Rapid growth of the genital tubercle changes the shape of the cloaca and the orientation of the cloacal membrane, which is displaced posteriorly. The cloacal membrane breaks down at 7 week’s gestation, thereby creating two openings: the urogenital and the anal ones. The muscles that surround the rectum develop at the same time and are seen in the sixth and seventh weeks of gestation and by the ninth week, all relevant structures are in place. (24) At this stage, differentiation into male or female external genitalia has not yet occurred.

5. Clinical presentations

When one is called to see a newborn male with an anorectal malformation, a thorough perineal inspection must be performed. This usually gives the key clues to the patient’s type of malformation. It is important not to make a decision about a colostomy or a primary operation before 24 hours of life. The reason for this is that significant intraluminal pressure is required for the meconium to be forced through a fistulous orifice. Passage of meconium through a fistula is the most valuable sign of the location of that fistula. If meconium is seen on the perineum, that is evidence of a perineal fistula. If
there is meconium in the urine, that confirms the presence of a rectourinary fistula. Radiologic evaluations do not show the real anatomy before 24 hours because the rectum is collapsed and it takes a significant amount of intraluminal pressure to overcome the muscle tone of the sphincters that surround the lower part of the rectum. Therefore, radiologic evaluations done too early, (before 24 hours) most likely will show a “very high rectum” - giving an incorrect diagnosis.

6. Investigations and Management

The decision-making algorithm for the initial management of the male is shown in Figure 10. During the last few years, the tendency by the pediatric surgical community has been to operate on patients with anorectal malformations primarily, without a protective colostomy (12-15). We promote this trend, but also alert all to the potential negative consequences of doing these operations without the necessary preoperative evaluation and experience. Often, if babies present late with significant abdominal distention, the clinician must perform an urgent colostomy.

During the first 24 hours of life, the baby should receive intravenous fluids, antibiotics, and be evaluated for associated defects that may represent a risk to the baby’s life; mainly cardiac malformations, esophageal atresia, and urologic defects (16). A nasogastric or orogastric tube protects the baby from aspiration but does not really impact colonic distention. An echocardiogram of the heart can be taken, and the baby must be checked for the presence of esophageal atresia. An x-ray film of the lumbar spine and the sacrum should be taken, as well as a spinal ultrasound to rule out the presence of tethered cord or other spinal anomalies. An ultrasound of the abdomen will evaluate for presence of hydronephrosis.

Figure 10: Decision making algorithm for male newborns with anorectal malformations (30)

If the baby has signs of a perineal fistula, an anoplasty can be performed, without a protective colostomy. This can be done during the first 48 hours of life, or delayed for several months depending on the baby’s condition.

If, after 24 hours, there is no meconium on the perineum, we recommend performing a cross-table lateral x-ray with the baby in prone position. If air in the rectum is seen located below the coccyx...
(Figure 11 A, B) and the baby is in good condition with no significant associated defects one may consider performing a posterior sagittal operation without a protective colostomy. A more conservative alternative could be to perform the posterior sagittal repair and a protective colostomy in the same stage.

Conversely, if the rectal gas does not extend beyond the coccyx, the patient has meconium in the urine, an abnormal sacrum, or a flat bottom, we strongly recommend a colostomy. This allows for a future distal colostogram which will precisely delineate the anatomy. We would then perform a posterior sagittal anorectoplasty within the first few months of life, provided the baby is gaining weight normally.

After recovery from the colostomy, the patient is discharged from the hospital.

Figure 11:
A) Cross-table lateral position B) Cross-table lateral radiograph

Performing the definitive repair within the first six months of life has important advantages, including less time with an abdominal stoma, less size discrepancy between proximal and distal bowel at the time of colostomy closure, and easier anal dilations (because the baby is smaller). In addition, at least theoretically, placing the rectum in the right location early in life may represent an advantage in terms of the potential for acquired local sensation (17).

All of these potential advantages of an early operation must be weighed against the possible disadvantages of surgery by an inexperienced surgeon, who is not familiar with the minute anatomic structures of an infant’s pelvis.

A temptation to repair these defects, without a protective colostomy, always exists (12, 13). Repair without a colostomy limits the anatomic information (provided by a distal colostogram) that may be very helpful to the surgeon. The worst complications involve patients operated on without a colostomy and without a properly done distal colostogram (25). Proceeding with the posterior sagittal approach looking blindly for the rectum has resulted in a spectrum of serious injuries including damage to the urethra, complete division of the urethra, pull-through of the urethra, pull through of the bladder neck, injury to the ureters, and division of the vas deferens or seminal vesicles (25).

Figure 12 shows the decision-making algorithm for the initial management of newborn females. Again, the perineal inspection is the most important step to guide diagnosis and decision making. The first 24 hours should also be used to rule out serious, associated defects as previously described. The perineal inspection may disclose the presence of a single perineal orifice. This single finding establishes the diagnosis of a cloaca which carries a high risk of an associated urologic defect. The patient needs a complete urologic evaluation including an abdominal and pelvic ultrasound to look for hydronephrosis and hydrocolpos.

Figure 12: Decision making algorithm for female newborns with anorectal malformations. (30)
Cloacas require a colostomy. It is important to perform the divided sigmoid colostomy in such a manner as to leave enough redundant, distal rectosigmoid colon to allow for the pull through (Figure 13 and 14) and even a vaginal replacement if needed. When performing the colostomy, it is mandatory to drain the hydrocolpos, when present, with a rubber tube.

Because a significant number of these patients have two hemivaginas, the surgeon must be certain that both hemivaginas are drained. Occasionally, a vaginovaginostomy in the vaginal septum will have to be created to drain both hemivaginas with one catheter. At times, the hydrocolpos is so large that it may produce respiratory distress. It is the hydrocolpos which compresses the trigone and causes bilateral hydronephrosis. Drainage of the hydrocolpos allows for decompression of the urologic system. A vesicostomy in most circumstances will not decompress the ureters. Rarely, the baby may require a vesicostomy or suprapubic cystostomy as well to decompress the bladder, if the common channel is very narrow and does not allow the bladder to drain.

However, in the vast majority of cases drainage of the hydrocolpos is all that is required. Endoscopic examination of the cloaca is recommended to delineate the anatomy. This is best done later (in several months) during a separate anesthetic because the newborn perineum is swollen and endoscopy is difficult. This endoscopic examination, along with contrast studies, allows the surgeon to formulate a clear reconstructive plan.

The perineal inspection may show the presence of a perineal fistula. In that kind of defect, we recommend performing a primary anoplasty without a colostomy. A protective colostomy to help avoid perineal infection is a conservative approach and appropriate in many clinical circumstances.

**Figure 13: Ideal colostomy for anorectal malformation**
Figure 14: Colostogram in a patient with an ideally placed colostomy, in which plenty of length was left for the pull through.

The presence of a vestibular fistula represents the most common finding in female patients. *(Figure 15)* When newborns with a vestibular fistula are repaired primarily at our institution, we keep the patient five to seven days with nothing by mouth, receiving parenteral nutrition. Conversely, when the patient has a primary repair of a vestibular fistula or perineal fistula without colostomy later in life, we are very strict about a preoperative bowel clean out 24 hours in advance, to be sure that the intestine is completely clean. We then keep the patient seven to ten days on parenteral nutrition with nothing by mouth, all in an attempt to avoid a perineal infection. If this NPO period with TPN is not possible in a given clinical circumstance, protection of the repair with a colostomy is indicated.

In fewer than 5% of girls, no visible fistula exists and there is no evidence of meconium after 24 hours of observation. This small group of patients requires a cross table lateral prone x-ray *(Figure 11 A, B)*. If the radiograph shows gas in the rectum very close to the skin, it means that the patient very likely has a very narrow perineal fistula. Conversely, if the distal extent of the rectal gas is located about 1-2 cm above the skin, it is most likely the patient has an imperforate anus with no fistula. If the patient is in good condition, a primary operation without a colostomy can be performed depending on the surgeon’s experience. Most of these patients with no fistula (in both boys and girls) also have Down’s syndrome *(26)*. In the event that associated conditions make the rectal repair unfeasible in the newborn period, a colostomy should be done, with definitive repair deferred. Occasionally, if the baby, with rectoperineal or rectovestibular fistula- has severe associated defects or is ill, the surgeon may elect to dilate the fistula to facilitate emptying of the colon while these other issues are addressed, with a plan for a future...
definitive repair.

**Figure 15: Rectovestibular fistula**

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7. Complications and Outcomes

When evaluating the results of the treatment of anorectal defects, we feel that one cannot group patients according to the traditional nomenclature into “high,” “intermediate,” and “low” defects, as malformations classified this way have different treatments and different prognoses. For instance, rectoprostatic fistula and bladder-neck fistula, both traditionally considered as “high” defects are actually very different. We believe that an anatomic classification has more clinical value. (Table 1) The functional results of the repair of anorectal anomalies seem to have significantly improved since the advent of the posterior sagittal approach. However, the results of this approach are difficult to compare with those of other methods because terminology and classification are not consistent (27, 28).

7.1. Fecal continence

Fecal continence depends on three main factors: voluntary sphincter muscles, anal canal sensation, and colonic motility.

7.1.1. Voluntary muscle structures

In the normal patient, the voluntary muscle structures are represented by the levator muscle complex, and external sphincter. Normally, they are used only for brief periods, when the rectal fecal mass reaches the anorectal area, pushed by the involuntary peristaltic contraction of the rectosigmoid motility. This voluntary contraction occurs only in the minutes prior to defecation, and these muscles are used only occasionally during the rest of the day and night. Patients with anorectal malformations have abnormal voluntary striated muscles with different degrees of hypo-development. Voluntary muscles can be used only when the patient has the sensation that it is necessary to activate them. To appreciate that sensation, the patient needs information that can only be derived from an intact anal sensory mechanism, a mechanism that many patients with anorectal malformations lack.
7.1.2. Anal Canal

Exquisite sensation in normal individuals resides in the anal canal. Except for patients with rectal atresia, most patients with anorectal malformations are born without an anal canal; therefore, sensation does not exist or is rudimentary.

It seems that patients can perceive distention of the rectum, but this requires a rectum that has been properly located within the muscle structures. This sensation seems to be a consequence of stretching of the voluntary muscle (proprioception). The most important clinical implication of this is that liquid stool or soft fecal material may not be felt by the patient as it does not distend the rectum. Thus, to achieve some degree of sensation and bowel control, the patient must have the capacity to form solid stool.

7.1.3. Bowel motility

Perhaps the most important factor in fecal continence is bowel motility; however, the impact of motility has been largely underestimated. In a normal individual, the rectosigmoid remains quiet for variable periods of time (one to several days), depending on specific defecation habits. During that time, sensation and voluntary muscle structures are almost unnecessary because the stool, if it is solid, remains inside the colon. The patient feels the peristaltic contraction of the rectosigmoid that occurs prior to defecation. Voluntarily, the normal individual can relax the striated muscles which allow the rectal contents to migrate down into the highly sensitive area of the anal canal. There, accurate information is provided by the anal canal concerning the consistency and quality of the stool. The voluntary muscles are used to push the rectal contents back up into the rectosigmoid and to hold them if desired, until the appropriate time for evacuation. At the time of defecation, the voluntary muscle structures relax.

The main factor that provokes emptying of the rectosigmoid is a massive involuntary peristaltic contraction helped sometimes by a Valsalva maneuver. Most patients with an anorectal malformation suffer from a disturbance of this sophisticated bowel motility mechanism. Patients who have undergone a posterior sagittal anorectoplasty or any other type of sacroperineal approach, in which the most distal part of the bowel was preserved, show evidence of an over-efficient bowel reservoir (megarectum). The main clinical manifestation of this is constipation, which seems to be more severe in patients with lower defects.

Constipation that is not aggressively treated, in combination with an ectatic distended colon, eventually leads to severe constipation. A vicious cycle ensues, with worsening constipation leading to more rectosigmoid dilation, leading to worse constipation. The enormously dilated rectosigmoid, with normal ganglion cells, behaves like a myopathic type of hypomotile colon. Those patients with anorectal malformation treated with techniques in which the most distal part of the bowel was resected behave clinically as individuals without a rectal reservoir. This is a situation equivalent to a perineal colostomy. Depending on the amount of colon resected, the patient may have loose stools. In these cases, medical management consisting of enemas plus a constipating diet, and medications to slow down the colonic motility is indicated.

7.1.4. Management of true fecal incontinence

For patients with true fecal incontinence, the ideal approach is a bowel management program consisting of teaching the patient and his/her parents how to clean the colon once daily so as to stay completely clean for twenty-four hours. This is achieved by keeping the colon quiet in between enemas. These patients cannot have voluntary bowel movements and require an artificial mechanism to empty their
colon, a daily enema. The program, although simplistic, is implemented by trial and error over a period of one week. The patient is seen each day and an x-ray film of the abdomen is taken so that they can be monitored on a daily basis for the amount and location of any stool left in the colon, as well as the presence of stool in the underwear. The decision as to whether the type and/or quality of the enemas should be modified as well as changes in their diet and/or medication can be made daily. (29)

Approximately 75% of all patients with anorectal malformations have voluntary bowel movements. (30) About 50% of them have voluntary bowel movements, but soil their underwear occasionally. Episodes of soiling are usually related to constipation, and when constipation is treated properly, the soiling frequently disappears. Approximately 40% of the group have voluntary bowel movements and never soil, thus making them totally continent. 25% of patients suffer from fecal incontinence and must receive a bowel management regimen to artificially keep them clean.

Once the diagnosis of the specific defect is established, the functional prognosis can be rapidly predicted, which is vital in order to avoid raising false expectations in the parents. Factors such as the status of the spine, sacrum, and perineal musculature influence the counselling of the parents. Patients with a hypo-developed sacrum are much more likely to be incontinent. A hypo-developed sacrum is also a good predictor of associated spinal problems such as tethered cord. If the patient’s defect is of the type pointing to a good prognosis such as vestibular fistula, perineal fistula, rectal atresia, rectourethral bulbar fistula, or imperforate anus without fistula, one should expect that that child will have voluntary bowel movements by the age of 3. Such children need supervision to avoid fecal impaction, constipation, and soiling. If a patient’s defect points to a poor prognosis, such as a high cloaca (common channel greater than 3 cm) or a recto-bladder neck fistula, the parents should be informed of the likelihood that that child will need a bowel management program to remain clean, which should be implemented at the age of 3 or 4. Patients with rectoprostatic fistulas have almost equal chance of having voluntary bowel movements or being incontinent. Toilet training should be attempted at age 3, and if unsuccessful, a bowel management program should be initiated. Each year, during summer vacation, an attempt should be made to try to achieve bowel control, and if unsuccessful, the bowel management should be restarted. As the child grows older and more cooperative, the likelihood of achieving bowel control will improve. Once bowel management is successful with a daily enema, a Malone appendicostomy for antegrade enemas can be considered. (25) This changes the route of enema administration to an easier and more efficient one.

7.2. Urinary continence

Urinary incontinence occurs in male patients with anorectal malformations only when they have an extremely defective sacrum, absent sacrum, abnormal spine, or when the basic principles of surgical repair are not followed and important nerves are damaged during the operation. The overwhelming majority of male patients have urinary control. This is also true for female patients, not including the cloaca group.

For patients with cloaca, functional prognosis with regard to achieving fecal continence depends on the complexity of the defect and the status of the spine and sacrum. Urinary control varies based on the length of the common channel. 70% of patients with cloaca, with a common channel greater than 3 cm, require intermittent catheterization; as compared to 20% in the group with a common channel less than 3 cm. The bladder neck in most patients is competent, and those patients that require catheterization remain dry in between. If catheterization is not performed, overflow incontinence occurs. Occasionally, the bladder neck is not competent or is non-existent, and in these cases, urinary diversion such as a Mitrofanoff procedure is considered. Careful, regular follow-up is necessary to accurately review the prognosis and to avoid problems, which
can dramatically impact the ultimate, functional result.

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8. References


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