Evolution of management of anorectal malformation through the ages
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INTRODUCTION
Anorectal malformations include a wide spectrum of defects in the development of the lowest portion of the intestinal and urogenital tracts. Many children with these malformations are said to have an imperforate anus because they have no opening where the anus should be. The diagnosis is usually made shortly after birth by a routine physical examination. Imperforate anus occurs in about 1 in 3500 live births and its cause is unknown. In spite of advancement in knowledge and better techniques, the condition still carries considerable morbidity in the form of incontinence of stool after surgery due to improper placement of the rectum with reference to the controlling muscles of the perineum. Traditional surgical dictum did not allow for division of the posterior midline because this division of the muscle was believed to cause incontinence in the child. Therefore, surgeons approached these malformations using a combined abdominal, sacral, and perineal approach, with limited visibility. Such approaches have put continence at greater risk than simply cutting sphincter muscles to adequately visualize the malformation. This principle was the centre of attraction for the development of the surgical techniques currently used to repair these malformations.

ANATOMY
The levator ani muscle lies in a plane between the symphysis pubis and the coccyx (PC Plane). This muscle comprises ileococcygeus and pubococcygeus including puborectalis. The puborectalis forms the most medial part of the levator hammock. The external anal sphincter (EAS) has three components which are the superficial, subcutaneous and deep sphincter muscles. The deep fibres of the EAS blend imperceptibly into the inferior portion of the puborectalis. These anatomically inseparable muscle entities function in-vivo as a single coherent unit and all are important in normal continence.

In 1971, Stephens compared normal anatomy to anorectal malformation for the first time and emphasized the preservation of the puborectalis muscle sling. In 1972, Pena paid particular attention to puborectalis where operating anatomy seemed different from that in the textbook and said, “Do not trust diagrams – trust the real thing”. He considers it to be a striated muscle complex, stating that there is no separate puborectalis sling.
PATHOPHYSIOLOGY
Understanding the true anatomy is helpful to prevent damage to important structures during surgical repair and to preserve the best potential for bowel control. Anatomic visualization has allowed surgeons to eliminate many previous misconceptions. For instance, the previous classification of these defects into high, intermediate, and low malformations was a misleading oversimplification that did not adequately demonstrate the spectrum of anorectal anomalies.

Improved imaging techniques and a more thorough knowledge of the anatomy and physiology of the pelvic structures at birth have refined diagnosis and early treatment. Analysis of large series of patients has allowed better prediction of associated anomalies and functional prognosis.

CLASSIFICATIONS
The variety of classification and differences in terminology has caused considerable confusion in describing the pathology of anorectal anomalies.

1. GROSS CLASSIFICATION:
According to this classification the anorectal malformations were divided in two groups depending upon the levator muscle, the supralelevator and infralelevator anorectal anomalies.

2. INTERNATIONAL CLASSIFICATION (1970):
This classification was described in 1970, it described low, intermediate, high and miscellaneous lesions both in males and females.

3. WINGSPREAD CLASSIFICATION (1984):
The most common previous international classification was referred to as the Wingspread classification of anorectal malformations, elaborated in Wingspread, Wisconsin, in 1984. This classification distinguished between high, intermediate, and low anomalies in the male and female, with special groups established for cloacal and rare malformations. High-type anorectal malformations were subdivided into anorectal agenesis with and without fistula, and rectal atresia. The intermediate malformations were classified as rectovestibular and rectovaginal fistula in the female and rectobulbar fistula in the male as well as anal agenesis without fistula in both sexes. The low-type malformations were classified as anovestibular fistula in the female and, in both sexes, as anocutaneous fistula and anal stenosis. This classification was widely accepted over the years and was based on detailed embryological and anatomic studies performed especially by Stephens et al., Kelly on anatomic sections and radiographic investigations.

4. PENA CLASSIFICATION (1995):
Some details of the Wingspread classification remained questionable. Some types of anorectal malformations such as rectovaginal fistulas are very rare, and from the surgical point of view, using PSARP in about two thirds of all anorectal malformations, the sex of the patient did not seem important in the choice of the surgical approach. Therefore, in 1995, Peña proposed a classification which was based on the relationship of the terminal colon to the levator sling muscles of the pelvic floor. He distinguished between perineal, vestibular, bulbar, prostatic, and bladder neck fistulas; imperforate anus without fistula; vaginal fistulas; cloacal fistulas; and rectal atresia or stenosis. This descriptive and fistula-related grouping became widely accepted over the past decade. The advantage of the classification of Peña is that the type of the fistula provides information not only about localization of the blind pouch but also on the anticipated extent of mobilization of the atretic rectal segment necessary to perform a sacro- or abdominosacropereineal pull-through.

5. KRICKENBECK CLASSIFICATION (2005):
A number of rare anomalies, not previously recognized and included in the Wingspread classification, were also reported; perineal groove, H type of anorectal anomalies, rectal ectasia, rectal atresia, and most importantly the pouch colon were reported from the Indian subcontinent. Thus, a need was felt for revision of the previous classification, review of recent surgical approaches and developing international standards for assessing postoperative outcome. For this, a conference was held in Krickenbeck (Germany) from May 17-20, 2005, with the aim to have a thorough discussion on the international classification of ARMs, form international criteria for their treatment and develop a uniform international scoring system for comparable follow-ups.

ETIOLOGY
The etiology of such malformations remains unclear and is likely multifactorial. There are, however, reasons to believe there is a genetic component. As early as in the 1950s, it was recognized that there was an increased risk for a sibling of a patient with ARM to be born with a malformation, as much as 1 in 100, compared with the incidence of about 1 in 3500 in the general population. It is likely that the mutation of a
variety of different genes can result in ARM, or that the etiology of ARM is multigenic.

**DIAGNOSIS**

An absent or anomalous anus is usually so obvious that it is discovered in the delivery room. The diagnosis of low or translevator lesions can be made by physical examination of the perineum. In the female the number of openings in the perineum is highly significant. Three openings means that the problem can be tackled from the perineum while the presence of two or only one opening means staged surgery. Insipite of tremendous evolution in the diagnosis of anorectal malformations the female anomalies are still presenting at a later age group in India. At our center, we have treated a patient of rectovestibular fistula even at age of 22 years. Similarly, the detection of a visible fistulous communication in a male child means that the anomaly is of low or infralevator type. Clinical inspection of the buttocks is important. Perineal signs found in patients with low malformations include the presence of meconium at the perineum, a “bucket-handle” malformation (a prominent skin tag located at the anal dimple below which an instrument can be passed), and an anal membrane (through which one can see meconium). A flat “bottom” or flat perineum, as evidenced by the lack of a midline gluteal fold and the absence of an anal dimple indicates that the patient has very poor muscles in the perineum. These findings are associated with a high malformation and therefore a colostomy should be performed.

**INVESTIGATIONS**

The aims of investigative procedures are:

1. To determine the nature of the anomaly and the level of termination of the bowel whether low, intermediate or high.

2. To detect a fistulous communication.

3. To determine the presence of associated anomalies having a direct bearing on the immediate outcome of treatment, e.g., gastrointestinal anomalies like duodenal and intestinal atresias, midgut volvulus, short colon and others.

4. Biochemical and bacteriological studies to check the effects of the anorectal malformation on the body.

5. Later radiological studies to further determine the actual position of the blind pouch (distal cologram) and assess the urinary system (micturating cystourethrogram, intravenous urography and ultrasonography)

**INVERTOGRAM (1930)**

The upside-down x-ray (invertogram) originally described by Wangensteen and Rice (1930) has been considered the classic method for determining the distance from the blind rectal pouch to a marker placed on or within the anal dimple. The baby is carefully held upside down for at least 3 minutes; decision should be made about the level of the rectal pouch at least 24 hours after births.

**PRONE CROSS-TABLE LATERAL VIEW (1983)**

A ‘prone cross-lateral view’ modification as described by Narasimharao was employed. The infant would be placed in a prone position with the hip flexed and elevated up to 45 degrees. The radiographic center was placed around the greater trochanter. A radiologic marker was routinely placed at the perineal area where there should be anal dimpling. Because of the retrospective nature of the present study, the timing of the investigation was not protocolized and varied from hours to days after birth.

A distal cologram was performed prior to a definitive anorectal reconstruction in infants for whom colostomy had been established. A water-soluble contrast medium was injected into the distal limb of a sigmoid loop colostomy via a plain catheter. The level of anomalies and possible presence of a fistula was observed under fluoroscopy in a true lateral position.

**ULTRASONIC EXAMINATION (1996 & 2003)**

Few studies were conducted to know the pouch-perineal distance through transperineal route (1996) but the most recent one is on infracoccygeal ultrasonography (US) (2003) which can directly demonstrate the puborectalis muscle in neonates with imperforate anus. In contrast, conventional transperineal US cannot depict the puborectalis muscle. The differentiation of low- from high-type imperforate anus has been indirectly performed with the measurement of the distance from the distal rectal pouch to the perineum, which is now used routinely. The puborectalis muscle was identified as a hypoechoic U-shaped band at the level of the anorectal flexure. US finding of the distal rectal pouch passing through the puborectalis muscle suggests a low-type imperforate anus. The puborectalis muscle is the innermost portion of the levator ani muscle and is considered to have an important role in the control of bowel function.
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MRI AND CT SCAN
Though it can demonstrate the structural aberration in great details, it has not been practiced routinely in the majority of the centers. Nowadays, it is used to assay the normality of the perineal region after repair of anorectal malformation.

TREATMENT
Throughout the centuries, doctors have seen and attempted to treat babies born with imperforate anus. Paulus Aegineta recorded the earliest account of successful surgery for imperforate anus in the 7th century AD. He suggested rupturing an obstructing membrane with the finger or knifepoint and then dilating the tract until healing was complete. In 1660, Scultet used dilatation to treat an infant with anal stenosis. In 1676, Cooke used incision and dilatation and advised care of the sphincter muscles. In 1783, acting on Littre’s suggestion from 1710, Dubois performed an inguinal colostomy for imperforate anus. Other surgeons followed suit, but almost all of the infants died; thus, colostomy remained unpopular and a procedure only of last resort. In 1835, Amussat described formal perineal proctoplasty (i.e., mobilization of the bowel through a perineal incision and suturing to the skin). This technique gained rapid acceptance. Strictures were less common than observed in earlier procedures. In addition to Amussat, Dieffenbach described anal transposition (1826); Chassaignac used a probe through a stoma to guide the perineal dissection (1856); and Leisrink (1872), McLeod (1880), and Hadra (1884) recommended opening the peritoneum if the bowel was not encountered from below. In 1930, Wangensteen and Rice first advocated imaging to delineate the abnormality. Single-stage abdominoperineal procedures became widely used after reports by Rhoads, Pipes, Randall, Norris, Brophy, and Brayton (1948-1949). In 1953, Stephens proposed an initial sacral approach followed by an abdominoperineal operation, when necessary. The purpose of the sacral stage of the procedure was to preserve the puborectalis sling, considered a key factor in maintaining fecal continence. This surgery and its modifications were the standard approach until 1980. In 1980, the surgical approach to repairing anorectal malformations altered dramatically with the introduction of the posterior sagittal approach described by Pena. This approach allowed pediatric surgeons to clearly view the anatomy of anorectal malformations and to repair them under direct vision, with better visualization and understanding of the anatomy than previous approaches allowed and is now the most preferred approach for the management of all types of anorectal malformations. Having determined whether the anomaly is low, intermediate or high the treatment is as follows:

1. In males, low anomalies are treated with single-stage perineal surgery while intermediate and high anomalies require a preliminary colostomy.

2. In females, low and intermediate anomalies can be treated through the perineal route without a colostomy while high anomalies require staging.

3. When in doubt as to the nature of the anomaly inspite of all possible investigations, it is always better to do a colostomy rather than explore the perineum.

4. Pelvic colostomy is physiologically sounder compared to transverse colostomy for the following reasons: (a) more solid stool consistency, (b) less area for the absorption of urine refluxing from the colourinary fistula.

The most widely accepted approach for the management of anorectal malformation is staged surgery. Staged surgery requires three operations; firstly colostomy at birth, than definitive operation after 2-3 months of age and finally colostomy closure at the age of around 6 months. All these cases need regular follow-up in a bowel management treatment center in order to achieve better continence.

The recent research suggests that the sooner the correction of congenital malformation the better will be the result. Albanese et al. thought that early restoration of gastrointestinal continuity would “train” the perineal musculature and improve long-term fecal continence. On the contrary, if the repair of anorectal anomalies is delayed, the critical time may be lost, in which neuronal networks and synapses would have formed resulting in normal or near-normal function. Moore also thought that it was very important to establish brain-defecation reflexes early. So it is very important to restore the gastrointestinal continuity in the neonate. Few questions arise while using single-stage PSARP procedure in neonates. First: Is the procedure safe? – Yes it is safe, well documented in few series. Second: Questions arise as to how to distinguish types of malformations, how to ascertain the precise location of the rectal pouch without a good distal cologram, and how to decide operation methods, via posterior sagittal or abdominal route; the technique of prone lateral x-ray is carried out on patients without obvious fistulae to show the level of the rectal pouch and sacrum in most of the cases. Third: Is it
easy to perform the PSARP procedure in a neonate? The answer is: Yes, it is easy to perform in a neonate provided the surgeon is experienced. Forth: Can the same results be achieved with fewer complications in the one-stage PSARP procedure than in the three-stage PSARP procedure? Yes, comparable result can be obtained by this approach.

With this view, few centers opted for the neonatal single-staged definitive repair with gratifying results. At our centre, we are doing neonatal single-stage repair for almost all types of anorectal malformations except the common cloacae malformations since 1995. We have published the largest series of anorectal malformation pouch colon as single-stage management in the newborn and single-stage repair for vestibular anus in the neonatal age group.

Why do we prefer single-stage management of anorectal malformations?

We feel that the cerebral cortical fibers develop in the first few years of life and sensations of rectal fullness are essential for these fibers to develop fully so that continence can be achieved to its maximal potential.

We feel the dissection to be easier in the neonatal period due to virgin tissue planes with no fibrosis due to pouchitis as seen in the older patients of the staged procedure.

There was no need for tapering of the rectal pouch when the operation was performed in the neonatal period.

Primary PSARP has the advantages of relieving the alimentary tract obstruction at birth, eliminating urinary tract contamination and establishing anorectal continuity, thus giving maximal potential for normal defecation reflexes at birth.

Our own experiences for treatment of all types of anorectal malformations:

ANORECTAL MALFORMATIONS

Primary single-stage procedure (PSSP) was done over a period of ten years (1996–2006) in 735 cases, which have been compared with the records of 458 out of 763 cases of staged procedure that underwent all three stages done from 1989 to 1996. The method used was a modification of Pena's PSARP with extra-luminal dissection of the rectal pouch well above the fistula site to facilitate its separation from the urethra followed by ligation of the fistula without opening and tapering of the rectal pouch. The fistula was transfixed towards the rectal pouch and divided and closed with interrupted sutures towards the urethra. The rectal pouch was opened only at the last phase of the operation before its fixation with the skin at the proposed site of the anus.

The mean age for the staged procedure was 3.2 days for colostomy, 9.6 months for definitive surgery and 34.4 months for colostomy closure. In the single-stage repair group, the mean age was 3.1 days. The duration of hospital stay for staged procedure (SP) was 22-49 days (mean 34.8 days) with 5-10 days for colostomy, 10-16 days for definitive surgery, and 7-10 days for colostomy closure and 3-26 days for readmissions due to complications (colostomy associated diarrhea, bleeding, massive prolapse, stenosis, adhesive obstruction, etc). In the single-stage group (PSSP), the duration of hospital stay was 10-16 days (mean 11.2 days). Continence was assessed by Kelly's clinical method (continence, staining and sphincter squeeze) at the age of three years. It was good in 45%, fair in 33% and poor in 22% in the SP group while it was good in 68%, fair in 22% and poor 10% in the PSSP group. The mortality was 4.5% in the PSSP group. In the SP group, the mortality was high as only 40% cases completed all the three stages of the operation with early colostomy, the mortality was 15-20%. A mortality rate of 4.7% occurred with the definitive procedure in Group A while the mortality rate for colostomy closure was 0.3% (being a historical control group).

POUCH COLON

Congenital pouch colon is an unusual type of ARM most common in India, particularly in North India. No exact etiology and embryogenesis could be found. We have operated 143 (1996-2006) cases of pouch colon in newborns as a single-stage procedure. In our centre, we excise the entire pouch colon, unlike other centres where the pouch is being tubularized for pull-through as a staged procedure. We have compared our results of single-stage repair for pouch colon with that of staged procedure and observed that the continence of single-stage was better than staged procedure. In our series, continence was good, fair, and poor in 43%, 31% and 26%, respectively, for single-stage procedure and in 29%, 20% and 51% for staged procedure.

Anatomical normalcy was attained with primary single-stage procedure (PSSP) at the age of 5.4 days and at 26 months with staged procedure (SP). Physiological normalcy (near-normal bowel frequency of <3 stools per day) was attained at the age of 6 months with PSSP and at puberty with SP. Mortality was high (46.93%) in SP (52/98 turned up, 42 in
follow-up) whereas in PSSP, it was only 10.78%.

Based on our experience, we think the trend for repair of anorectal malformation should move toward primary single-stage repair in the neonatal period which has equivalent results with that of staged procedure with minimal morbidity.

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