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"ANORECTAL MALFORMATIONS" EMBRYOLOGICAL BASISAND ITS CLINICAL SIGNIFICANCE

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ABSTRACT

Anorectal malformations (ARM) comprises, a wide spectrum of diseases, which can affect girls and boys, and involve the rectum and distal anus as well as the genital and urinary tracts. ARM occurs in around 1 in 5000 live births. Defects range varies from the very minor or "low" which are easily treated with an excellent functional prognosis, to those that are difficult to manage, complex, are often associated with other anomalies known as "high", and have a poor functional prognosis. The surgical approach to repairing these defects changed intensely in 1980 with the introduction of the posterior sagittal approach, Better imaging techniques, and a improved knowledge of the anatomy and physiology of the pelvic structures at the time of birth.

Introduction

Anus is an opening where the gastrointestinal tract ends and faecal matter exits the body. The anus starts at the bottom of the rectum, the last portion of the colon (large intestine). The anorectal line separates the anus and rectum. Tough tissues that are called fascia surrounds the anus and attaches it to nearby structures. Circular muscles called the external sphincter ani form the wall of the anus and hold it locked. anal glands release fluid into the anus to keep its surface moist. A plate-like band of muscles, called the levator ani muscles, surround the anus and form the base of the pelvis. There is a network of veins lines the skin of the anus. The rectum is the concluding part of the large intestine that terminates in the anus. The average length of the human rectum ranges between 10 and 15 cm. Diameter of rectum can be compared to that of the sigmoid colon (the part of the large intestine nearest the rectum) at its onset. However, it becomes bigger near the anus, where it forms the rectal ampulla [1].

The main role of the rectal ampulla is to act as a temporary storehouse for feces. The expansion of the rectal walls causes the stretch receptors within the walls that stimulate the urge to defecate. However, if the defecation process is delayed, it may result in constipation. When the storage site becomes full, the intra-rectal pressure causes the anal canal walls to dilate and expand. This results in the feces entering the canal. A rectal exam may be conducted to diagnose some diseases. Certain types of cancers may be diagnosed by performing an endoscopy of the rectum. An endoscopy is a procedure where a doctor uses an endoscope — a small, flexible tube with camera and light is used to examine areas inside the body. Body temperature may also be obtained from the rectal area. In case of infants and babies, this is generally the most accurate method for determining actual body temperature [1-6].

Anorectal malformations (ARMs) are a complex group of congenital anomalies involving the distal anus and rectum, as well as the urinary and genital tracts in a significant percentage cases. Prevalence is approximately one per 5000 live births, with a slight male preponderance. In most ARMs, the anus is not perforated and the distal enteric component may ends blindly (ie, atresia), or as fistula into the urinary tract, genital tract, or perineum. ARMs are associated with other congenital anomalies in up to 70% of cases. The final prognosis and quality of life for patients with ARMs will depend to a large extent on the presence and gravity of these associated anomalies. Urogenital abnormalities are most frequently observed and appears in up to 60% of patients with vesicoureteral reflux and hydronephrosis the most common findings. The spine and spinal cord are also often involved with agenesis and sacral dysplasia, vertebral dysplasia and tethered cord syndrome the most frequently detected problems. ARMs are also present in a great number of syndromes and associations of multi-systemic congenital anomalies. The classification of ARMs is mainly based on the position of the rectal pouch relative to the puborectal sling and the presence or absence of fistulas. Imaging studies play a important role in the initial evaluation of ARMs. In new born, an accurate diagnosis of the type of ARM, the presence or absence of fistulas and their locations and identification of associated anomalies is essential in deciding about immediate therapy. However, the real utility and the most appropriate sequence of performance of the different imaging diagnostic methods in the initial days of life remains subject of discussion. For patients with ARMs, the ultimate objective of therapy is to achieve a good quality of life, with acceptable levels of bowel control, normal sexual and reproductive capacities. In infants and older children, pelvic magnetic resonance (MR) imaging is the most effective imaging technique for defining anatomy and determining the grade of

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development of the sphincteric muscles which plays a significant role in continence. This information will help the medical team to make decisions about the definitive surgical approach and provide the orientation about the possible postoperative prognosis and remaining sequelae [11-19].

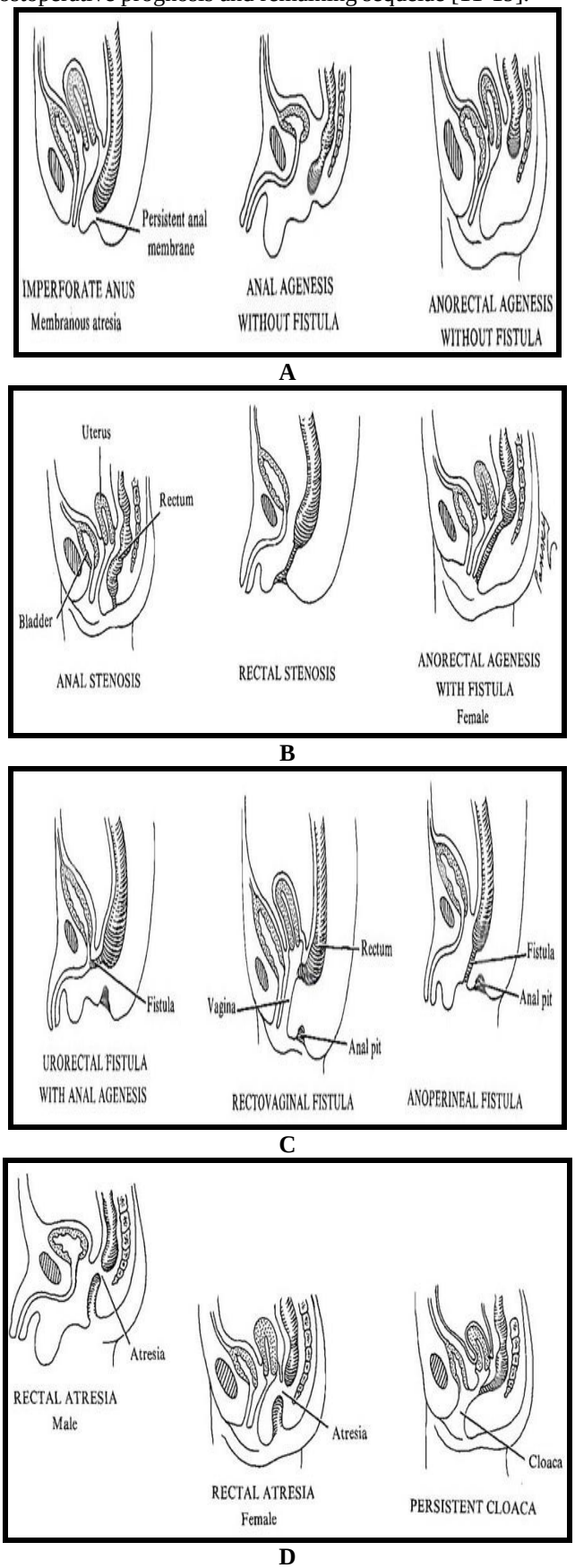


Fig 1. A, B, C, D Schematic representation shows the different type of anorectal anomalies.

Incidence

Anorectal malformations comprises of a wide spectrum of diseases, which can affect both male and female. This anomaly involves the distal anus and rectum as well as the genital and urinary tracts. They occur in around 1 in 5000 live births. Defects range from the minor and easily treated with an excellent functional prognosis, to those that are difficult to manage, are often associated with other anomalies, and often have a poor functional prognosis [18-26].

Ontogenesis for the normal development of the anus

An appropriate knowledge of embryology is crucial for understanding ARMs. The early embryologic development of the anorectal, the primitive urogenital sinus, and the caudal neural tube is closely related, which helps to explain the associated malformations of these systems [27].

In early embryonic life, the terminal portion of the hindgut i.e. the primitive cloaca is divided intoventral and dorsal parts by a coronal sheet of mesenchyme which is called the urorectal septum and separated from the amniotic cavity by the cloacal membrane. Most of ARMs result from abnormal development of the urorectal septum [28].

Between weeks 4 and 6 of gestation, both the yolk sac or primitive hindgut and primitive urogenital sinus or the allantois enters into the cloaca. Then urorectal septum the develops forklike infoldings (Rathke folds and Tourneux) of the lateral cloacal walls; at the same time, the embryo starts to curve as a result of the longitudinal growth of the developing neural tube and mesodermal compartment. With these kind of morphologic changes, the distance between the cloacal membrane and the tip of the urorectal septum is progressively reduced [28].

At the end of week 7, the cloacal membrane and the urorectal septum are located at the same level. The cloaca is thus divided into a ventral part (the urogenital sinus) and a dorsal part (the proximal anal canal and rectum). Between them, the tip of urorectal septum becomes the perineal area. At this time, the cloacal membrane ruptures by apoptosis, thus opening two orifices in the perineum: one dorsal or anal and one ventral or urogenital [29].

Also at the end of week 7, a secondary occlusion of the anorectal canal takes place, firstly by adhesion of the walls and afterward by the formation of an epithelial “plug” at the anal level. This secondary closed anal orifice will rupture and recanalize by apoptosis at the end of week 8 [30].

Embryologically, Anorectal malformations can thus be subdivided into two main groups according to when the disturbances occur: Those manifesting as an ectopic anal orifice or fistula are due to early abnormal development of the cloacal membrane and the dorsal part of the cloaca (at weeks 4–7), whereas those manifesting as an abnormal anus in a normal position are due to later defective recanalization of the secondary occluded anal orifice (at weeks 7 and 8) [31].

Ontogenesis of the abnormal development of the anus

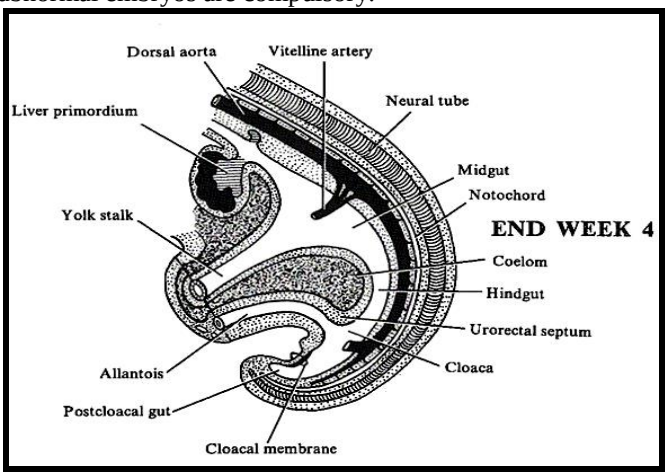
Some form of imperforate anus is frequently seen, occurs in 1/5000 births, and is more common in males than females. Most of the anorectal malformations are result of an abnormal development of the urorectal septum, which results in an incomplete or partial separation of the cloaca into the anorectal and urogenital parts [32].

Clinically three major types are seen. Absent or imperforated anus: A usual anal imperfection includes many varieties, and all of them require surgical intervention. Insufficient anus leads to problems of meconium evacuation and it should be treated without any delay. Ectopic sinus: less

serious than above since it does allow some intestinal passage, but that too, is usually functionally insufficient [34].

Superficial deformities are due to anomalies in development and fixation at the superficial perineal levels. Anal agenesis or insufficient anus (with or without fistula): the canal may end sightlessly, and there may be an ectopic anus (ectopic opening) or vulva or male urethra or fistula opening in the perineum due to partial separation of the cloaca by the urorectal septum. The abnormal communications normally called fistulae and it should be regarded as ectopic anal opening/orifices. Anorectal deformities with the anus in usual position are best described as late embryonic defects [35].

Now days, the abnormal and normal development of the hindgut is still a matter of speculation. However, as the result of certain recent studies in suitable animal models, most of the embryologic events that finally lead to abnormal hindgut development are better known than in the past: the process of maldevelopment starts in early embryonic stages; the cloacal membrane is permanently too short in its dorsal part, thus, the dorsal cloaca is absent; and as a result of this the hindgut remains attached to the urogenital sinus and forms the recto-urethral fistula. In the past, an impaired process of septation was supposed to be the main cause of abnormal hindgut development. In contrast to this, other results indicate that the development of the septum is more inactive. Furthermore, the results of some studies in abnormal and normal development indicate that the embryonic cloaca is never passes through a stage which is similar to any form of anorectal malformation in neonates, as well as the so-called "cloaca's" in female embryos, and to explain the abnormal development, studies in abnormal embryos are compulsory.



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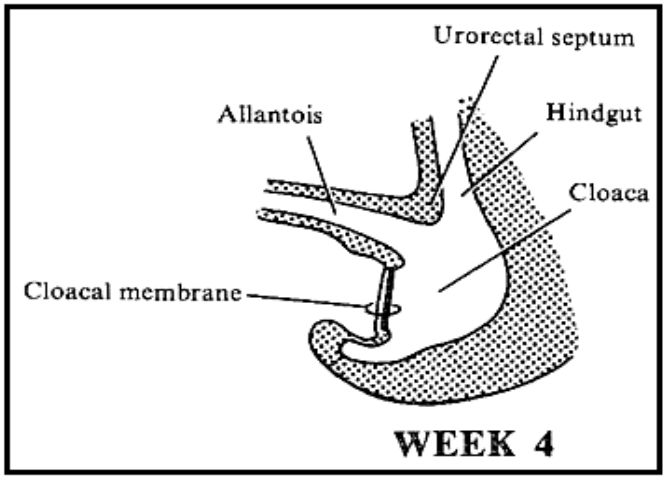

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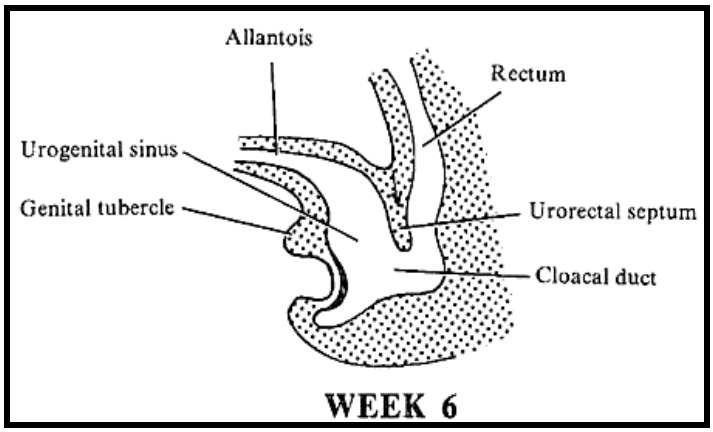

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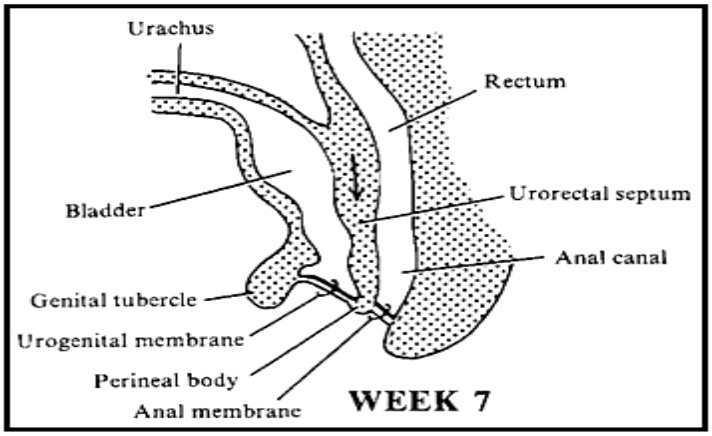

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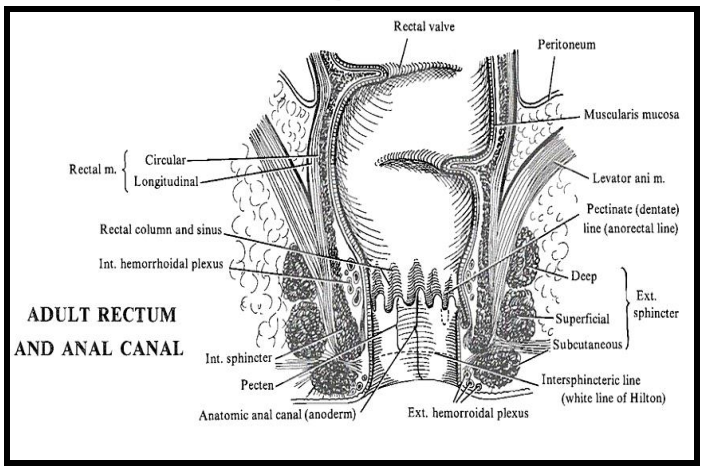

E

Fig 2. A, B, C, D, E Schematic representation shows the development of rectum and anus from hindgut.

Membranous atresia or covered anus either with or without fistula are very rare in which the anus is in normal place, but a thin and tiny layer of tissue separates the anal canal from the outside. This is due to a failure of the anal membrane to perforate at the end of week 8. Anorectal agenesis either with or without fistula: rectum ends sightlessly above the anal canal, but there is usually a fistula to the vagina in the female or urethra in the male. This defect is similar to membranous atresia. Deep malformations, the anomaly affects the septation of the cloaca by unusual migration of the urorectal septum. Pure rectal atresia is complete failure of the development of the inferior part of the rectum and anal canal. Rectal atresias with fistula are always insufficient. The degree and length of anastomosis differentiate the various types of anomalies. Mixed malformations comprise with all forms of ectopic anus.

Discussion

If there is a opening (fistula) to the skin, vagina or urethra, a newborn will pass meconium (a baby's first stool), unless a careful examination is done, the imperforate anus may not be suspected. However, if there is no anal fistula and there is no anal opening, the baby will not be able to pass stool after birth and this will lead to a "enlarged" or swollen abdomen and vomiting. Since in each child's specific anatomic abnormality will variable, it is important for you to understand your patient's particular anatomy. If the anal opening is in the wrong spot or absent, it can be seen on examination. If there is stool coming out of the vagina orurethra, instead of the anus, it will be visible soon after birth. In females with anorectal malformation, we should have a careful examination of the vestibule (area between the labia) made to ensure that there is separate openings of the urethra and vagina. In the males with imperforated anus, careful examination of the perineum is must to identify any anomalous passage of stool. Children with anorectal deformity may also have other congenital anomalies. The following anomalies can be found to occur together and these are described by the acronym VACTERL association. Vertebral anomalies in which vertebrae (bones of the spine) can be abnormally formed in children with imperforate anus, Anorectal malformations, Cardiac or heart defects, TE which stands for tracheoesophageal fistula/atresia. This is an abnormality of the oesophagus in which there is a unsighted ending or abnormal connection to the trachea (windpipe), R-Renal means kidney. Some children have abnormalities related to urinary system which includes kidneys, bladder, ureters and the urethra and Limb or irregularities of the radial bones in the arms that can lead to displaced hands.

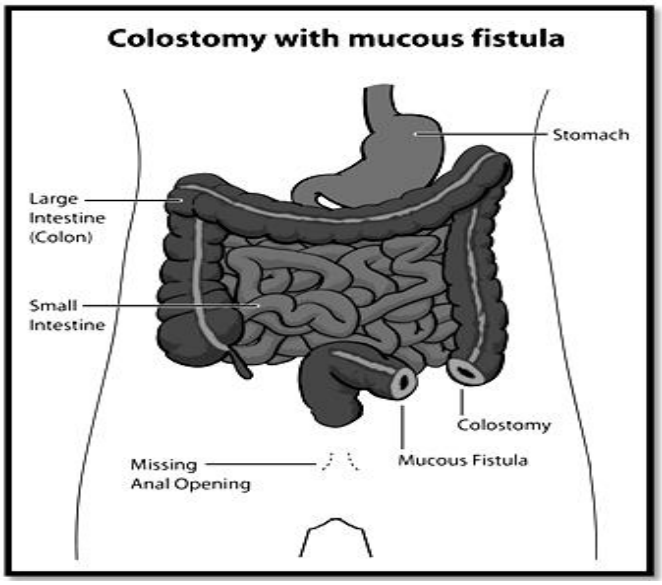

Fig 3. Schematic representation shows the imperforated anus colostomy.

Some Important associated anomalies contain genitourinary defects, which can be seen in approximately 50% of all patients with anorectal malformations. All patients must be carefully examined at birth to rule out one of these defects, and the most important screening test is abdominal and pelvic ultrasound. Urologic examination prior to colostomy gives the surgeon the essential information which is needed to address the urologic problem during the colostomy. The surgeon must be ready to perform a urologic diversion if it is necessary.

Unfortunately, sometimes a common error in diagnosis and analysis occurs during the perineal examination. When a female is thought to have "imperforate anus with rectovaginal fistula" when in actuality, all three structures, the urinary tract, rectum and vagina all meet in a common channel and the baby has a cloaca. The presence of a solitary perineal orifice is the clinical evidence of a patient with persistent cloaca. children with these anomalies also have small genitalia. In children with persistent cloaca, examination of the abdomen may show an abdominal mass which probably represents a distended vagina (hydrocolpos), present in almost 50% of patients with cloaca. An abdominal ultrasound determines the presence of an disruptive uropathy as well as the presence of hydrocolpos. This misconception has significant therapeutic implications that will be discussed below. It is vital to make the exact determination of cloaca because nearly 90% of babies have an associated urologic anomaly, and nearly 50% have hydrocolpos. Both the distended vagina and the urinary tract may need to be dealt with in the new-born period to avoid serious complications. Missing the diagnosis of cloaca regularly means that an obstructive uropathy is ignored. The patient may then receive only a colostomy and subsequently may suffer from acidosis, sepsis, and sometimes death.

The other implication of missing the diagnosis of cloaca includes repairing only the rectal component of the anomaly and leaving the patient with a persistent urogenital sinus. Perineal fistulas in both males and females have usually been called "low" defects. In these cases, the rectum opens in a small orifice, generally stenotic and situated anterior to the center of the sphincter. Most of these patients have brilliant sphincter mechanisms and a normal sacrum. In males, the perineum may exhibit some other features that help in recognition of this defect, such as a prominent midline skin bridge which is known as 'bucket handle' or a sub epithelial midline raphe fistula that looks like a black ribbon because it is full of meconium. These features are superficially visible and help diagnose a perineal fistula [32-40].

A simple anoplasty expands the stenotic orifice and relocates the rectal orifice posteriorly within the limits of the sphincter complex. The procedure is called a "minimal posterior sagittal anoplasty". It is executed with the patient positioned prone with the pelvis elevated; multiple fine silk sutures are the places where the mucocutaneous junction of the bowel opens for traction. A short midsagittal incision of 1–2 cm is made posterior to the fistula site which divides the entire external sphincter complex. The lower part of the rectum and fistula are carefully dissected to allow mobilization of the rectum for backward placement, within the limits of sphincter complex. The perineal body is the area where the fistula was situated and is repaired with some long-term absorbable sutures.

In the rare cases of a true supralevator malformation called recto-bladder neck fistula, there is a operation which involves both an abdominal component and a posterior sagittal incision, which can be done with laparotomy or laparoscopy.In patients with imperforate anus without a fistula, the same scrupulous dissection is required to separate the distal rectum from the urinary tract in patients with recto urinary fistulae because the urethra and rectum still share a common wall.

In cases of recto-vestibular fistula, the posterior sagittal incision can be shorter than rectourethral fistulae in male patients. Often the entire levator mechanism needs not be divided and only the muscle complex, external sphincter, and

part of the lower portion of the levator mechanism need to be divided. The posterior vagina and rectum share a common wall, and it is this separation that is the most tough part of the operation. Once the rectum is completely mobilized, a perineal body is built, and the rectum is placed within the limits of the sphincter mechanism.

Rectal atresia a very rare malformation occurs in 1% of cases. In rectal atresia, the anal canal is normal and the anus appear normal externally. However, there is a small blockage 1–2 cm from the anal skin which is usually found when the nurse tries to pass a thermometer. These babies should undergo colostomy at the time of birth, and then their definitive repair involves a posterior sagittal approach and an end-to-end anastomosis is made between the anal canal and the upper rectal pouch.

The repairing of persistent cloaca represents a serious technical challenge that should be performed in some specialized centres by pediatric surgeons dedicated to the care of these complicated cases. This malformation characterizes a wide spectrum of defects by itself. The defect involves fusion of the vagina, rectum, and urethra together to form a common channel. The length of this common channel ranges from 1 to 10 cm. The vagina and rectum share a common wall and the urinary tract and vagina likewise have a common wall. The goals of surgical treatment are to attain urinary control, bowel control, and normal sexual function. Sometimes all three goals are achieved but sometimes only two, often only one, and occasionally none.

Conclusion

In conclusion, surgery of perianal abscesses and fistulae show numerous possible variations. As shown in the literature the surgeon's knowledge about anatomy and function, technical skills, experience and patience of both patient and surgeon is needed to achieve satisfying results. Anorectal malformations of the rectum and anal canal are best classified into "high" and "low" anomalies. The "low" anomalies are comparatively easy to diagnose, can be approached and treated through a perineal route and have a favourable prognosis. The "high" anomalies are difficult to manage, require a combined abdominosacral or abdominoperineal approach and have generally a poor prognosis.

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