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Characteristics of anorectal malformations

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ABSTRACT
Anorectal malformation is a congenital abnormality that shows a condition without an anus or with an imperfect anus. Anorectal malformation is a common congenital problem that occurs in 1 in 5,000 births. Anorectal malformation is a classification where anorectal malformations are divided into high and low locations, as well as whether or not a colostomy is necessary. Anamesis and physical examination are very important in establishing a diagnosis of anorectal malformations. Plain radiographs of the lumbar spine and sacrum should be taken to evaluate the hemivertebrae and sacruml spine. Generally, low-lying malformations can be corrected directly with Posterior Sagitttal Anorectoplasty (PSARP) or anoplasty, while high-lying malformations require 3 stages of surgery, including the creation of a temporary colostomy. A colostomy is needed for decompression and protection against the possibility of intestinal obstruction.

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INTRODUCTION

Anorectal malformation is a congenital abnormality that shows a condition without an anus or with an imperfect anus. Anorectal malformations are congenital abnormalities that we often encounter in pediatric surgical cases. The incidence of anorectal malformations is 1 in 4,000 to 5,000 live births. The incidence of anorectal malformations in Europe between regions varies between 1.14 to 5.96 per 10,000 people and can change every year. In general, anorectal malformations are more common in men (58%) than women.

Anorectal malformations have several types of classification. One of them is the classification by Alberto Pena (1995), who introduced a classification where anorectal malformations are divided into high and low locations, as well as whether or not a colostomy is necessary. Being able to identify anorectal malformations before birth can redirect sending the baby to a pediatric surgery center. Anorectal malformations without perineal fistula and intestinal atresia causing obstruction are urgent surgical indications.

Management of anorectal malformations is based on classification, whether a one-stage operation can be carried out, for example an opalstomy or whether a colostomy is required first for a three-stage operation. Generally, low-lying malformations can be corrected directly with Posterior Sagittal Anorectoplasty (PSARP) or anoplasty, while high-lying malformations require 3 stages of surgery, including the creation of a temporary

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colostomy. A colostomy is needed for decompression and protection against possible intestinal obstruction.

Definition

Anorectal malformations are a common congenital problem occurring in 1 in 5,000 births and have a spectrum of anatomic presentations, requiring individualized care for the newborn, sophisticated approaches to definitive reconstruction, and management of long-term care and outcomes. Rectourethral fistulas most often occur in men and rectovestibular fistulas in women. Having no fistula at all is rare (5% of patients) and is associated with Down syndrome.

Etiology

The etiology of anorectal malformations is not yet known with certainty. Some experts believe that this disorder is a result of abnormalities in the embryological development of the anus, rectum and urogenital tract, where the septum does not divide the cloacal membrane completely. There are several prognostic factors that influence the occurrence of morbidity in anorectal malformations, such as abnormalities in the sacrum, imperfect perirenal muscle system and impaired colonic motility.

To date, factors associated with the presence of additional congenital abnormalities in anorectal malformation patients have not been fully identified. Some literature suggests that there is a correlation between gender and type of anorectal malformation with the presence of additional congenital abnormalities. Male patients are more likely to have additional congenital abnormalities compared with female patients. Patients who have anorectal malformations in higher locations are also more likely to have additional congenital abnormalities compared with those whose anorectal malformations are in intermediate or low locations. The relationship between prematurity and birth weight and the presence of additional congenital abnormalities in anorectal malformation patients still requires further research. However, it is known that babies born prematurely and with low birth weight usually tend to cause anomalies, VACTERL (abnormalities of the vertebrae, anus, heart, trachea-esophagus, kidneys and limbs).

Epidemiology

According to the World Health Organization (WHO), every year around 8 million babies worldwide experience congenital abnormalities. In the United States, the number of babies born with congenital abnormalities is up to 120,000 every year. About 3% of babies, or one in 33 babies, are reported to be born with structural congenital abnormalities. The prevalence of births of babies with congenital abnormalities in England is up to 2%, while in South Africa it is around 1.49%. In Lebanon, the incidence of congenital abnormalities was reported to be up to 1.64% in the South Beirut area.

According to Basic Health Research (RISKESDAS) data in Indonesia, congenital abnormalities are one of the factors causing infant death. In the age range of 0-6 days, the percentage of infant deaths caused by congenital abnormalities is up to 1.4%, and there is a significant increase to 18.1% in the age range of 7-28 days.

The death rate due to anorectal malformations varies between 16% and 29.4% based on several studies conducted in the period 1995-2014. Neonatal deaths caused by anorectal malformations still occur frequently, especially due to delays in diagnosis or

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delays in obtaining surgery or a colostomy, especially in developing countries. Obstacles such as the location of health facilities far from where one lives, limited facilities and a lack of pediatric surgical specialists are still the main challenges.

METHOD

This research uses a literature review method to explore the characteristics of anorectal malformations. Anorectal malformation is a congenital disorder involving anomalies in the lower digestive tract. Through analysis of relevant literature, this study aims to understand the clinical characteristics, pathophysiology, risk factors, and current diagnostic and management approaches to anorectal malformations. It is hoped that the findings from this literature review will provide a deeper understanding of this disorder, which can support efforts for early diagnosis, intervention and appropriate management for individuals affected by anorectal malformations.

RESULT AND DISCUSSION

Classification of Anorectal Malformations

The most common international classification of anorectal malformations is the Wingspread classification in 1984.

Table 1. Classification of Anorectal Malformations According to Wingspread

Location of Anorectal Malformations	Man	Woman
Tinggi	1. Agenesis anorectal a. Fistula Rektovesika b. Tanpa fistula 2. Atresia fistula	1. Agenesis anorectal a. Fistula Rektovesika b. Tanpa fistula 2. Atresia Fistula
Intermediate	1. Fistula Rektouretra 2. Agenesis anus tanpa fistula	1. Fistula Rektouretra 2. Fistula Rektovagina 3. Agenesis anus tanpa fistula
Rendah	1. Fistula perineal 2. Stenosis anus	1. Fistula anovestibular 2. Fistula anokutan 3. Stenosis anus
Lain-lain	Malformasi jarang	Kloaka Malformasi jarang

(Sources:Lokananta I, Gadjah M. Malformasi Anorektal)

Nevertheless, the implications of anorectal malformations have significant consequences, and the Wingspread classification is considered not to provide sufficient prognostic and therapeutic value. So, Pena in 1995 compiled a simpler classification.

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TABLE 103-1
Classification of Anorectal Malformations

Males	*Females*
Perineal fistula	Perineal fistula
Rectourethral fistula	Vestibular fistula
Bulbar	Persistent cloaca
Prostatic	≤3 cm common channel
Rectobladder neck fistula	>3 cm common channel
Imperforate anus without fistula	Imperforate anus without fistula
Rectal atresia	Rectal atresia
Complex defects	Complex defects

Figure 1. Classification of Anorectal Malformations According to Pena

Malborne divides based on the pubococcygeus line and the line that passes through the ischii abnormalities called:

a. High position if the rectum ends above the levator ani muscle (Musculus pubococcygeus)
b. The intermediate location is when the end of the rectum is located at the levator ani muscle.
c. Low position if the end of the rectum ends below the levator ani muscle.

Diagnosis

The importance of history taking and physical examination cannot be underestimated in the process of diagnosing anorectal malformations. The baby is placed in a lithotomy position with adequate lighting, then the anal canal is probed using methods such as a thermometer, probe, nasal speculum, or lacrimal duct probe. Newborns with anorectal malformations are usually identified at the first physical examination. Malformations in newborns that are missed on initial examination are often discovered within 24 hours when the newborn is observed to be distended and fails to pass meconium and a more thorough examination is carried out.

In order for the anus to be normal, a mandatory examination determines its correct location and correct size, based on age. The normal size of the anus in full-term babies is a Hegar 10 to 12 dilator (a tool used to measure the anus), and children aged 12 months must use a Hegar 15 dilator. The basis for the correct location is the anal canal which is in the middle of the anal muscle complex . The position of the anal opening to the muscle complex cannot always be seen in the clinic and often requires examination under anesthesia.

If the child has a normal urethra and no vestibular fistula, he may have an imperforate anus without a fistula. If she appears to have trisomy 21, the chances increase that she does not have a fistula. Girls with a normal urethra and no visible fistula were observed for 24 hours to allow for the appearance of a perineal fistula before surgery was necessary.

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This waiting period is useful in differentiating between children with perineal fistulas who can be treated electively with only minimal anoplasty from those who require a colostomy with further evaluation using distal colostography.

Alberto Pena, a pediatric surgeon from the United States, introduced a method for diagnosing anorectal malformations that applies to both male and female patients.

a. Man

   Anorectal malformations in lower positions can be identified when a perianal fistula, bucket handle, anal stenosis, or anal membrane is found. Meanwhile, anorectal malformations in a higher position can be confirmed by findings such as meconium, air in the urinary bladder, and a flat bottom shape. If there are still doubts, supporting examinations such as radiography (invertogram) can be carried out. Anorectal malformations in a low position can be established if the invertogram results prove that the end of the rectum is less than 1 cm from the surface of the skin, while anorectal malformations in a high position can be established if the end of the rectum is more than 1 cm.

b. Woman

   90% of anorectal malformations in women are accompanied by fistulas, if no fistula is found then an invertogram is performed. Low position if the end of the rectum is < 1 cm and minimum PSARP surgery can be performed immediately, while high position if the end of the rectum is > 1 cm and a colostomy must be performed first.

Supporting investigation

Radiological examination cannot show the actual anatomy before 24 hours because the rectum collapses due to the tone of the surrounding sphincter muscles. Thus, performing a radiological examination too early, i.e. before 24 hours, may prove a “very high rectum” and may ultimately result in an inaccurate diagnosis.

An echocardiogram may be performed, and the baby should be checked for esophageal atresia. Plain radiographs of the lumbar spine and sacrum are mandatory to evaluate the hemivertebrae and sacrum. Spinal ultrasound helps screen for umbilical cord and other spinal problems. Abdominal ultrasound evaluates for hydronephrosis.

Governance

To prevent morbidity and mortality, careful clinical examination of the perineum in newborns is crucial and provides crucial clues about the type of anorectal malformation. It is crucial to assess the perineum and anal canal carefully, as meconium discharge does not always prove normal anatomy. In most newborns with anorectal malformations, clinical examination will be sufficient to determine the correct diagnosis, with more complex malformations requiring additional studies and imaging.

The two main determinants of whether a child has an anorectal malformation are;

a. The anal canal is inadequate in size;
b. The anal canal is not completely within the sphincter complex.

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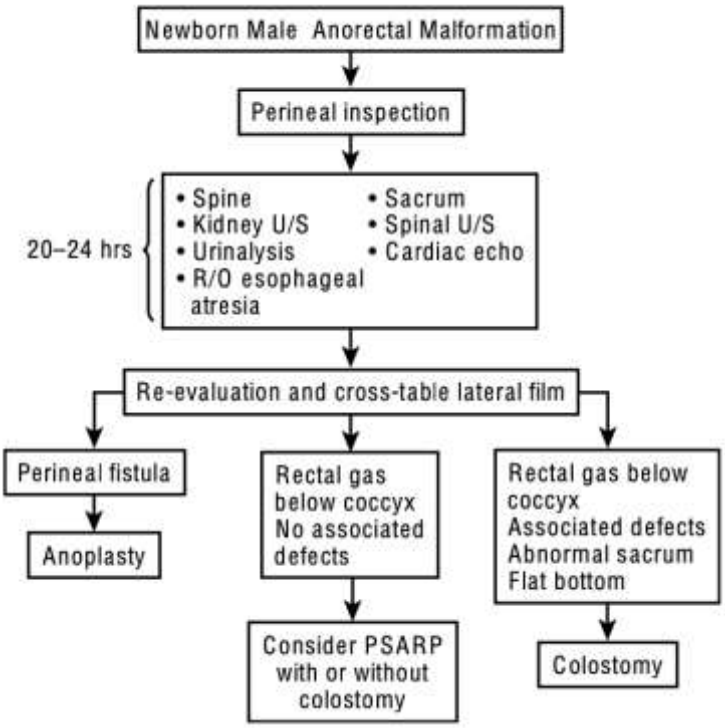

Figure 2. Algorithm for the management of male newborns with anorectal malformations based on physical examination and radiography. PSARP, posterior anorecto-sagittal

First, the doctor must perform a thorough examination of the perineum, which usually provides the most crucial clue about the type of malformation the patient has. It is crucial not to make a decision about a colostomy or primary surgery before 20 to 24 hours of age. The reason for waiting is that significant intraluminal pressure is required to push the meconium through the fistula which is the most valuable sign of distal rectal location in these infants. If meconium is visible in the perineum, it is evidence of a rectoperineal fistula. If there is meconium in the urine, the diagnosis of rectourinary fistula is clear. Radiological examination cannot depict the actual anatomy before 24 hours because the rectum collapses due to the tone of the sphincter muscle that surrounds the lower part. Thus, performing a radiological examination too early, i.e. before 24 hours, will likely prove a “very high rectum” and, as a result, may result in an inaccurate diagnosis. In the first 24 hours after birth, it is crucial for the newborn to receive intravenous fluids, antibiotics, and nasogastric decompression to prevent aspiration.

If the baby has signs of a perineal fistula, anoplasty can be performed without a colostomy in the newborn. Cases should not be postponed for more than a few months. After 24 hours, if no meconium is seen in the perineum or in the urine. If the gas in the rectum is below the coccyx and the baby is in good condition without significant associated defects, depending on the surgeon's experience, a protective posterior sagittal anorectoplasty (PSARP) may be considered. A more conservative alternative is to perform a colostomy, with definitive repair planned for the second stage.

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If rectal air is visible above the coccyx or the patient has meconium in the urine, significant associated defects, and/or an abnormal sacrum or flat bottom, a colostomy is recommended with delay of primary repair to subsequent surgery. This can be done 2 to 3 months ago after a distal colostogram depicting the anatomy is performed, provided the baby is gaining weight.

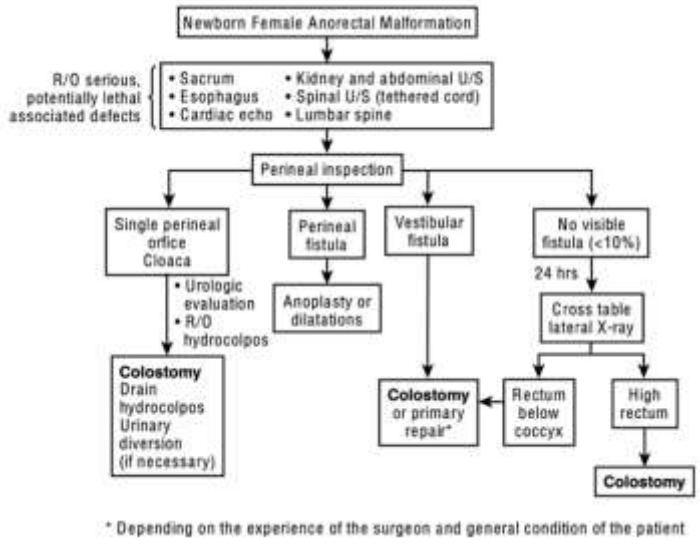

Figure 3. Decision-making algorithm for female newborns with anorectal malformations

Initial management of female patients. For example, in men, the most crucial step in diagnosis and decision making is inspection of the perineum. The first 24 hours should also be used to rule out serious associated defects. Inspection of the perineum may reveal a solitary perineal opening. These findings establish the diagnosis of cloaca. The doctor must know that the patient has a high probability of urological defects. The presence of hydrocolpos must be ruled out by ultrasound. For example, a baby needs a colostomy. It is crucial to perform a colostomy proximal enough to the sigmoid to allow repair of the malformation without interference from the colostomy. In other words, the surgeon is obliged to leave enough distal rectosigmoid to allow retraction and even vaginal replacement if necessary.

Abdominal ultrasound to evaluate for associated urologic problems and echocardiography are mandatory during the initial evaluation of the newborn. It is also convenient at this time to perform a spinal ultrasound to evaluate associated spinal anomalies e.g. myelomeningocele or umbilical cord. Ultrasound for example is an excellent examination for this anomaly, but must be performed before sacral ossification, which occurs at 3 months of age. If screening is performed later, magnetic resonance imaging is required. From plain anteroposterior and lateral radiographs of the sacrum, sacral ratios can be calculated which can help in predicting the prognosis for continence with low ratios predicting poor potential for bowel control and high ratios (>7) predicting good potential. If a hemisacrum is identified, a presacral mass may be present and should be sought with magnetic resonance imaging. During the assessment carried out before carrying out an

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anoplasty or colostomy, the baby is still given intravenous fluids, with a nasogastric tube inserted to decompress the stomach. Oral feeding is not permitted. Antibiotics are given after surgery.

Complications

Several complications associated with surgical intervention in anorectal malformations may arise. Wound infections can occur post-surgery, but generally only involve the skin and subcutaneous tissue, so they are not dangerous. These complications tend to resolve without leaving significant implications on body function. In addition, the tissue recovery process can occur within a relatively short period of time after surgery. Anal stricture can occur due to abnormal anal dilatation or less than optimal blood circulation after an anoplasty procedure.

Prognosis

Patients with perineal fistulas and rectal atresia of both sexes are expected to achieve optimal functional outcomes after the repair procedure, including the ability to achieve complete bowel continence. Male patients with anorectal malformations and patients of both sexes who do not have a fistula also have a good prognosis, with approximately 80% of them achieving bowel control between the ages of 3 and 4 years. However, most sufferers can still face some functional problems in the early stages after repair. Male patients with a rectourethral prostatic fistula have an approximately 60% chance of achieving bowel control by age 3 years. Meanwhile, male patients with rectovesical fistula have a less favorable functional prognosis, with only around 20% of them able to achieve voluntary bowel movements by the age of 3 years.

The presence of abnormalities in the sacrum bone generally indicates that the patient is likely to experience fecal incontinence, which is often accompanied by a rectovesical fistula or rectoprostatic fistula. Types of defects that have a good prognosis, for example perineal fistula or vestibular fistula, which are also associated with an abnormal sacrum, are rare. More than 90% of female patients with rectovestibular fistula can achieve voluntary bowel movements by the age of 3 years.

Patients who experience persistent cloaca with a common canal length of less than 3 cm have an approximately 80% chance of achieving voluntary bowel movements by the age of 3 years, and can generally control urination. On the other hand, if the length of the joint channel exceeds 3 cm, most sufferers experience fecal incontinence and require catheterization to empty the bladder. Patients who have a persistent cloaca with a channel more than 3 cm generally also have evidence of abnormalities in the sacrum bone.

A large number of sufferers experience fecal incontinence, sometimes accompanied by urinary incontinence. If the sufferer is old enough to be active in social life, a medical program is needed to manage bowel and bladder control. The use of enemas, suppositories, colonic irrigation, special diets, and sometimes medications to regulate colonic motility can help them maintain 24-hour hygiene, which in turn improves quality of life. Patients who experience fecal incontinence that does not respond to medical management and suffer from prolonged diarrhea usually require a permanent colostomy.

Most patients who undergo surgery to treat anorectal malformations will likely experience varying degrees of constipation. These symptoms tend to be more severe in

defects that are at a lower level and that are more simple in nature. Patients who experience a suboptimal colostomy (colostomy formation that allows feces to exit from the proximal to the distal end of the intestine) can experience a more serious level of constipation. These sufferers need to follow a diet rich in fiber and are sometimes given laxatives to help empty the rectum every day. Use of ineffective medical treatments can make the problem worse; The rectosigmoid continues to enlarge and the emptying process becomes inefficient, making the task of treatment difficult

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