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Anorectal Malformation With Fistel

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ABSTRACT

Anorectal malformation (MAR) with fistula is a congenital abnormality in which patients do not have an anus and the formation of a fistula. In men, the types of fistula are rectovesica fistula, rectourethra fistula, and perineal fistula. Meanwhile, in women, the types of fistula are rectovaginal fistula, rectovestibular fistula, cloaca and perineal fistula. The diagnosis is made by the presence of meconium in the urine. On physical examination, a flat perineum was found, there was no anal dimple and there was meconium in the perineum. The treatment is colostomy in cases of high fistula then Posterior Sagittal Anorectoplasty (PSARP) and fistulectomy, while in low fistula cases PSARP and fistulectomy are immediately performed.

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INTRODUCTION

According to the 2019 Indonesian Health Profile, congenital abnormalities accounted for 12.5% of newborn deaths. This shows that congenital anomalies, including anorectal malformations, can cause risks to newborns in Indonesia. The incidence of anorectal malformations is approximately 1/5000 to 1/2000 in various countries and regions. The combined live birth prevalence of anorectal malformations from the study period (1974–2014) was 2.96 per 10,000 births, which equates to 8,583 (91% of all cases) live birth cases of anorectal malformations. The distribution of mortality in the first week of age was 12.5%, including 654 (7.6%) who died on the day of birth, and 418 (4.9%) who died between days 2-6 after birth, subsequently, 137 (2 .0%) died between days 7-27, and 197 (3.1%) between days 28-364.

More common in baby boys than girls. The majority of male babies with anorectal malformations are recto-urethral fistula type. Anorectal malformations are often combined with other congenital anomalies, such as congenital heart disease, urogenital anomalies, spinal anomalies, and chromosomal abnormalities.

The specific etiology of anorectal malformations is unknown, most likely because genetic factors play a role in the development of the incidence of anorectal malformations, and exposure to environmental factors is also associated with the incidence of anorectal malformations. Anorectal malformations have several classifications, including anorectal malformations with fistula, namely rectovesica fistula, bulbar and prostatic type rectourethral fistula, perineal fistula in male babies, rectovaginal fistula, rectovestibular fistula, cloaca and perineal fistula in female babies.

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METHOD

Research on anorectal malformations with fistulas has become a major focus in efforts to understand this complex disease. By adopting the literature review research method, the researcher has directed his attention to a broad and in-depth literature review. The initial step involves a systematic search for relevant sources of information, including scientific articles, books, and other reliable sources that discuss medical aspects, causes, diagnosis, and treatment of anorectal malformations with fistula. After rigorous selection, the selected literature is then carefully analyzed to understand the research methodology used, the main findings produced, as well as the recommendations proposed by previous researchers.

Through evaluation and synthesis of information from various sources, this study aims to compile a comprehensive picture of anorectal malformations with fistula. It is hoped that the resulting research report will provide a valuable contribution in deepening understanding of the pathogenesis, diagnosis, and effective therapeutic approaches in treating cases related to this condition. In addition, with the solid foundation of this literature review, it is hoped that it will also stimulate interest in further research and renewal of clinical approaches in facing this complex medical challenge.

RESULT AND DISCUSSIONS

Definition

Anorectal malformation is an umbrella term for a variety of diagnoses often referred to as imperforate anus. Anorectal malformations are a spectrum of structural congenital defects involving the anorectum and variable segments of the urogenital system in both boys and girls. Patients with this diagnosis do not have a normal anal canal, but rather an open fistulous tract into the perineum anterior to the anal muscle complex or to adjacent anatomical structures. In men the fistula tract may connect to the urinary system and in women to gynecological structures. The distance of the open fistula tract from the exact location of the anal canal usually determines the severity of the defect.

Etiology

The etiology of anorectal malformations is unknown. It is likely that genetic factors play a role in the occurrence of anorectal malformations. There are several genetic syndromes with an increased incidence of anorectal malformations such as Currarino triad indicating autosomal dominant inheritance, and patients with trisomy 21. Deviations in any stage of hindgut development, as a result of genetic or epigenetic changes, can lead to an anorectal malformation phenotype. Furthermore, genetic knock-out animal models show that exposure to trans-retinoic acid and ethylene thiourea is correlated with the incidence of anorectal malformations. Lastly, environmental factors may be involved in the development of anorectal malformations such as in vitro fertilization, thalidomide exposure.

Pathomechanism

Early in embryonic life, the terminal part of the hindgut, the primitive cloaca, is divided into dorsal and ventral parts by a sheet of coronal mesenchyme, the urorectal septum and is separated from the amniotic cavity by the cloacal membrane. Most anorectal malformations result from abnormal development of the urorectal septum.

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Embryologically, anorectal malformations can be divided into two main groups according to when the disorder occurs, namely those that manifest as a fistula caused by the initial abnormal development of the dorsal part of the cloaca and the cloacal membrane at 4-7 weeks, while those that manifest as an abnormal anus are caused by defective recanalization. later in life from secondary obstruction of the anal canal at 7 and 8 weeks.

Rectourethral fistulas and rectovaginal fistulas can be caused by abnormalities in the formation of the cloaca and/or urorectal septum. For example, if the cloaca is too small or if the urorectal septum does not extend far enough caudally, then the hindgut opening shifts anteriorly toward the hindgut opening into the urethra or vagina. Perineal fistulas vary in severity and may leave a narrow tube or fibrous remnant connected to the surface of the perineum. These defects may be due to misexpression of genes during epithelial-mesenchymal signaling.

Classification

Classification of Anorectal Malformations is useful for determining the management and prognosis of this disease. The 2005 Krickenbeck conference established a new classification, based primarily on the presence or absence of a fistula and its type and location, as well as the position of the anal sac. Types of anorectal malformations in men are perineal fistula, rectourethral fistula (bulbar, prostatic) and rectovesica fistula. Meanwhile, in women there are types of cloaca, perineal fistula, rectovestibular fistula and rectovaginal fistula. The most common defect in women is rectovestibular fistula. Meanwhile, in men, the most common defect is rectouretheral fistula.

Diagnosis

It is estimated that half of children with anorectal malformations have an associated anomaly. The reported incidence of anomalies varies, but most groups agree that genitourinary anomalies (40-50%) are the most common, followed by cardiovascular (30-35%), spinal cord tethering (25-30%), gastrointestinal anomalies (5 -10%), and anomalous vertebral defects, anal atresia, cardiac defects, tracheoesophageal fistula, renal anomalies, and limb abnormalities (VACTERL) as much as 4-9%. The higher the tip of the rectal pouch, the higher the probability of being associated with an anomaly. Therefore, anomalies are most often associated with rectovesical fistula type anorectal malformations, but spinal anomalies occur frequently in all groups of anorectal malformations including perineal fistulas.

Most anorectal malformations are diagnosed as newborns. The anus should be in the right location and size, based on age. The normal size of a full-term baby's anus is a 10 to 12 Hegar dilator (a tool used to measure the anus), and the anus size of a 12 month old baby should be around a 15 Hegar dilator. The correct location is at the anal canal which is in the middle of the anal muscle complex.

Patients with complaints of defecation along with urine or holes in other places such as perineal fistula type. The absence of the anal canal in the correct position requires further evaluation. If no anus or fistula is visible, there may be a low lesion or a closed anus. Within 24 hours after birth, meconium will appear which gives a blue or black appearance in low-lying lesions. On physical examination, a flat perineum is characterized by the absence of anal dimples, which indicates that there are poor perineal muscles in the

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patient. It is associated with a high-lying anorectal malformation and requires a colostomy. Supporting examinations that can be carried out are CT-Scan and MRI used as a structural evaluation of the pelvic floor muscles and their relationship to the pouch for evaluation after surgery, distal colostography or lopography to find fistulas using contrast and urinalysis to evaluate the presence or absence of meconium in the urine.

Management

The initial management of patients with anorectal malformations is to be treated in neonatal care, kept fasting with an orogastric tube, maintained adequate hydration intravenously, and given antibiotics to prevent sepsis. For male babies with a low rectum, the treatment given is Posterior Sagittal Anorectoplasty (PSARP) and fistulectomy, then if the rectum is high, a colostomy is performed first, then PSARP and fistulectomy. In baby girls with a low rectum, the treatment given is PSARP and fistulectomy and in babies with a low rectum colostomy was performed.

CONCLUSION

Based on the results of literature review research on anorectal malformations with fistula, it can be concluded that this medical condition is a complex subject and requires an in-depth understanding of various aspects, including pathogenesis, diagnosis and therapeutic management. Through a synthesis of information from relevant scientific literature, this research succeeded in constructing a comprehensive understanding of the disease. The implication is that this research report can provide a strong foundation for further research in deepening understanding of anorectal malformations with fistulas, as well as supporting efforts to develop more effective clinical approaches in treating them.

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