ANORECTAL MALFORMATION WITH VAGINAL AGENESIS: A CASE REPORT

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Abstract

Vaginal agenesis is characterised by primary amenorrhea in adolescent women and other abnormalities, such as the absence of the uterus. The incidence of vaginal agenesis or distal vaginal atresia associated with anorectal malformations is difficult to estimate. A 5-year-old patient, at birth, immediately cried, and faeces came out through an orifice in the skin under the vagina. Previously, the patient had planned for a posterior sagittal anorectoplasty (PSARP), but it was known that the patient did not have a vaginal opening at the time of surgery. The patient was diagnosed with a low-type anorectal malformation, vaginal agenesis, and a sigmoidostomy status. PSARP and vaginoplasty were performed while the patient was 7 years old. Clinical and radiographic examination in the first 3 days of age plays a role in determining the type of anorectal malformation and whether or not a colostomy is necessary. In children, MRI radiology plays a role in the preoperative examination to evaluate the size, morphology, and stage of development of the muscular sphincter.

Keywords: anorectal malformations, vaginal agenesis.

INTRODUCTION

One of the most frequent congenital anomalies seen in paediatric surgery is anorectal malformations, which are thought to affect between 1 in 2,000 and 1 in 5,000 live births. It is rare to diagnose isolated anorectal malformations before birth. The majority of instances are discovered in the early neonatal stage (1).

Congenital abnormalities known as vaginal agenesis, or the absence of a vagina, can occur by themselves or in conjunction with other severe anomalies. According to estimates, 1 in 4,000–5,000 live births of females result in vaginal agenesis (2). Vaginal agenesis is characterised by primary amenorrhea in an adolescent woman and other abnormalities such as the absence of the uterus (also called bilateral Mullerian tubercles and ductal malformations), commonly known as Mayer-Rokitansky-Kuster-Hauser syndrome (MRKH). In cases where vaginal agenesis is associated with an anorectal malformation, it is usually detected in the neonatal period or during the repair of the anorectal malformation (3,4). The incidence of vaginal agenesis or distal vaginal atresia associated with anorectal malformations is difficult to estimate (3).

Imaging plays an important role in the evaluation of anorectal malformations. The role of clinical examination and imaging in the early life of the baby is to detect anorectal malformations as soon as possible and to help decide what action to take with the patient (5). The purpose of this study was to determine the incidence, urgency, role, and prognosis of the occurrence of anorectal malformations and vaginal agenesis.

CASE DESCRIPTION
A 5-year-old patient was born normally with a weight of 2,900 grams. At birth, the patient immediately cried, and faeces came out through an orifice in the skin under the vagina. Currently, the patient has a good general condition with a weight of 16 kg, a GCS of 456, a pulse of 96 beats per minute, a respiratory rate of 24 beats per minute, and an oxygen saturation of 97% room air. The patient continued treatment because she had not had an anus since birth (Fig. 1), so the stool comes out through the orifice in the skin under the vagina. Previously, the patient had planned for a PSA procedure, but it was known that the patient did not have a vaginal opening at the time of surgery. At the age of 2 years, the patient underwent surgery to make a sigmoid stoma; one year later, she underwent a stoma repair operation due to prolapse; and, at the age of 5 years, an anus operation was planned but was postponed because it was known that there was no vaginal opening. This evaluation was carried out along with plastic surgery. The patient was diagnosed with low-lying anorectal malformation, vaginal agenesis, and sigmoidostomy status. Posterior sagittal anorectoplasty and vaginoplasty were performed when the patient was 7 years old.

![Fig 1. The clinical photograph of the patient](https://jiiri.ub.ac.id/27/06/023/1-6/e-ISSN_2830-506X_p-ISSN_2830-6007)

**DISCUSSION**

The distal anus and rectum, as well as the urinary and genital tracts, are frequently affected by anorectal malformations, a complicated collection of congenital defects. The urorectal septum develops abnormally early in fetal life, which causes the majority of anorectal malformations. The anus is typically not perforated, and the distal intestinal components either end blindly (atresia) or as fistulas into the genitourinary, urogenital, or perineal tracts. A substantial range of syndromes and correlations with congenital defects also include anorectal malformations among their symptoms (5).

The prevalence of anorectal malformations ranges from 1 in 5,000 live births (1). Wingspread classification divides anorectal malformations into 3 types: low, intermediate, and high, depending on the location of the rectal pouch, whether it is below or at the level of the puborectal sling, with special categories in the cloaca and rare malformations. The most recent classification comes from the Krikenbeck conference in 2005, which distinguishes types based on the presence or absence of fistulas and the type of rectal sac location (5).

This patient had an imperforate anus with a perineal fistula. The patient was born in a midwife's office at an advanced gestational age. The patient's family only found out that the patient did not have an anus when the patient was examined by a health worker when she was 9 months old with complaints of difficult and small bowel movements. Due to the cost factor, the patient just received a colostomy at the age of 2.5 years.

Patients with an anorectal malformation are both high- and low-risk; the initial treatment is enterostomy, and then a high-pressure colostography evaluation is carried out at least 2–3 months after enterostomy. All orifices (clean stoma, dirty stoma, anal projection, external urethral orifice, vaginal orifice, fistula) are marked on a laparography examination. Then, the patient is exposed to AP and lateral. If the patient has had colostography, it will be followed by a pelvic MRI examination to determine the prognosis of the perineal muscle and an MRI of the spine to determine the presence or absence of a tumour and a tethered cord. The woman patient was examined for 3 orifices: anal projection, external urethral orifice, and
vaginal orifice, to determine whether there was a cloaca or not.

The initial examination in patients with anorectal malformations (ARM) is performed on a prone cross table with the baby pronated, the knee not placed on the chest, the baby's abdomen padded, and the femurs positioned close together. The knee was flexed and given an anus marker, and then the patient was exposed. The babygram examination aims to evaluate the thoracolumbar spine, pelvic cavity, and extremities.

The first lopography examination (Fig. 2) was carried out at the age of 2.5 years, whereas this examination is ideally carried out at the age of 2–3 months when patients with a stoma will undergo a definitive repair procedure. The results of the lopography examination reveal high anal atresia with a rectocutaneous (perineal) fistula with a diameter of ± 4.4 mm and a tract fistula length of ± 1 cm. The second lopography examination was carried out at the age of 4 years for evaluation of the fistula before surgery. The results obtained describe low-lying imperforate anus with a rectocutaneous fistula ± 1.6 cm long (Fig. 3).

The prognosis in these patients depends on the sacral ratio (Fig. 4), the location of the fistula, and the presence or absence of abnormalities in the spinal cord. The patient's sacral ratio is 0.35, which shows a poor prognosis; the rectoperineal fistula is a poor prognosis because the fistula is close to the anatomical location of the anal, and the quality of the spinal cast is still unknown because a lumbosacral MRI has not been performed.

Furthermore, at the age of 5, the patient was scheduled for anal surgery. However, it was postponed because, from the physical examination, there was no vaginal opening, and an MRI examination was then planned. One month after the incident, the result of the MRI revealed vaginal agenesis, bilateral normal uterus, and ovaries, and a Tarlov cyst was found as high as S3. The patient was planned for PSARP and vaginoplasty when she was 7 years old.
An ARM can occur as a non-syndromic disorder or be associated with a syndrome. Environmental factors are considered to have played a slight role in the development of ARM (6). In one study, 80% of children with ARM had other associated congenital anomalies (7). Syndromes associated with ARM include VACTERL/VACTER (spinal deformities, anal atresia, heart defects, tracheoesophageal fistulas, and renal anomalies), Trisomy 18, Trisomy 21, and other syndromes (6). In the fourth and eighth weeks of pregnancy, the urorectal septum stops descending toward the cloacal membrane. It is believed that this phenomenon is what caused this abnormality. This abnormality may only impact one person (sporadic) or, in exceptional circumstances, multiple members of the same family (8).

In this study, the patient was associated with a vaginal agenesis syndrome. Furthermore, the patient's ARM was only discovered when she was 9 months old. Higher mortality and poorer outcomes are linked to delayed diagnosis. Except in low-resource environments, such as in many developing countries, severe delays after childhood are uncommon (9).

The diagnosis of vaginal agenesis in such patients is typically delayed due to a lack of awareness of the relationship between vaginal agenesis and anorectal abnormalities, which presents a diagnostic challenge to paediatric surgeons. The results of a thorough perineal examination guide the diagnostic procedure in the evaluation of women infants with anorectal malformations. However, it can be quite challenging to examine the neonate's perineum at the bedside while the infant is awake and active. The precise number of orifices is frequently ambiguous. Rectovestibular fistula continues to be the most prevalent anorectal malformation in women neonates, so while one would anticipate three orifices (urethral, vaginal, and rectal fistulas), in a classic rectovestibular fistula, conclusive identification of all three is not always attainable at the base (10).

When the diagnosis is made early, the majority of patients with vaginal agenesis or distal vaginal atresia associated with anorectal malformations have neo-anal and vaginal reconstruction during the first year of life, according to Weter et al (11). Due to the necessity of early surgical surgery for many anorectal malformations, these patients frequently appear at a young age. Typically, the absence of a vaginal opening is discovered by accident during a preoperative check (10).

There are several significant treatment implications of the relationship between anorectal malformations and vaginal agenesis. The use of the sigmoid colon during colostomy in the management of anorectal malformations may inhibit its use for vaginoplasty at a later stage, therefore early detection affects clinical care. Additionally, vaginal agenesis seriously affects a child's capacity for reproduction. Early detection of this ailment enables the evaluation and treatment of psychological issues and concerns in adolescents (12).

Suggestions that the author can give to clinicians are as follows: the prenatal examination is carried out after 12 weeks of gestation to see the presence of MAR, the postnatal examination is carried out with a babygram and prone cross table, and the MRI examination is carried out for MRI evaluation. Then, for radiologists, there are a number of suggestions: on babygram, examination and prone cross-table evaluation of the high and low location of imperforate anus; on high-pressure colostography, examination is to assess the presence of a fistula; spine and perineal MRI is to determine prognosis; and sacral ratio examination should be done on a babygram photo.

If the child has an ordinary growth rate, the definitive treatment can be done as soon as 4 to 8 weeks following the colostomy
procedure. Early definitive repair reduces the amount of time spent with an abdominal stoma, the size difference between the proximal and distal intestine at the time of stoma closure, and the psychological after-effects of uncomfortable perineal surgery (8). In our case, the patient's lack of knowledge of the relationship between ARM and vaginal agenesis was the cause of the delay in a decisive surgery.

CONCLUSION

Anorectal malformations are a group of congenital abnormalities that include the distal anus and rectum and can also involve the urinary tract and genital tract. In this case, the patient who was to be operated on by ARM was found to have vaginal agenesis, so the operation was postponed. This highlights the fact that clinical and radiographic examination in the first 3 days of age play a role in determining the type of anorectal malformation and whether or not a colostomy is necessary. In children, MRI radiology plays a role in the preoperative examination to evaluate the size, morphology, and stage of development of the muscular sphincter.

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REFERENCES

6. Rosas-Blum ED, Reddy A, Shaban MA,


