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Female infant with apert syndrome and high imperforate anus without fistula

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ABSTRACT

Apert syndrome (AS) is a rare type of congenital craniofacial dysmorphic and severe syndactyly of the hands and feet. Fibroblast growth factor receptor (FGFR) gene mutations are suspect to be involved in this anomaly. The distinct features are craniosynostosis-a condition of premature closure of skull's sutures-, midface hypoplasia-an incomplete development of the middle of the face-, and syndactyly-webbed fingers or toes-. The anorectal malformations (ARMs) associations with AS is rare, and the genetic link is highly complex. Only 12.7% of ARMs cases were associated with a syndrome that has a well-known impact on intellectual development, including AS. To our knowledge, this is the first reported case of AS with a high imperforate anus without fistula.

1. Background

AS is a rare congenital disorder of type 1 acrocephalosyndactyly characterized by craniosynostosis, midface hypoplasia, and syndactyly. The prevalence is about 1–65,000 to 200,000 newborns with no difference incidence between genders [1]. It is an autosomal dominant disorder due to fibroblast growth factor receptor-2 gene (FGFR2) mutation [2]. We present a two-day-old infant diagnosed with AS and imperforate anus without fistula. The anorectal malformations (ARMs) without fistula occurs in approximately 5% of all ARMs cases. This malformation is closely related to Trisomy 21. Only 12.7% of ARMs cases were associated with the intellectual development-related syndrome, including Apert syndrome [3]. The genetic link between these two abnormalities is complex, and the relationship is still doubtful. The report about an infant with Apert syndrome and gastrointestinal abnormalities is rare [2,4]. To our knowledge, this is the first reported case of AS with a high imperforate anus. The literature is review systematically to assess the AS and imperforate anus clinical presentation, management, prognosis, and the operations of this type of ARM.

2. Case presentation

A two-day-old female infant weighing 3000 g was referred to our tertiary hospital with an imperforate anus. She did not pass stool since birth, and her stomach is distended. The yellow liquid is coming out from the orogastric tube. Meconium or stool was not found on her diapers. The patient's family history and mother pregnancy were unremarkable.

The abdominal area looks distended with prominent gut contour and dilated paraumbilical vein. The low abdominal sound was

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Fig. 1. Absence of anal opening.



Fig. 2. Babygram shown multiple dilated loops.



Fig. 3. Lateral view x-ray in prone position shown the distance of rectal gas shadow and anal lead marker is more than 2 cm, indicating a high imperforate anus.



Fig. 4. Craniofacial dysmorphism and syndactyly in patient, Fig. 4A. Coronal sutures fusion, depressed and broad nasal bridge, low-set ear, Fig. 4B. Prominent forehead, flat occipitalis, Fig. 4C. Narrow high arched palate, Fig. 4D–G. Syndactyly of both hands and foot.



Fig. 5. Post-operative presentation.

heard during auscultation and the tympanic sound during percussion. Genitourinary examination revealed normal genitals. The anal area showed hyperpigmentation anal area with an anal dimple, without orifice and without fistula in the perineal region (shown in Fig. 1). The babygram shown multiple dilated bowel loops (shown in Fig. 2). On the lateral abdominal x-ray evaluation of the prone position revealed the distance between the anus and the rectal gas shadow was more than 1 cm, suggesting a high imperforate anus (shown in Fig. 3). On further evaluation, she had a prominent forehead, coronal sutures fusion, and flat occipitalis. She had distinct facial and intraoral features: a depressed and broad nasal bridge, narrow high arched palate, and low-set ear (shown in Fig. 4). Hypertelorism, ocular proptosis, strabism, down sliding lateral palpebral fissures were absent. Syndactyly with complete fusion of all five digits on both hands and feet was present. No other apparent anomalies could be identified and no evidence of VACTERL association was present. The current examination suggests a case of high imperforate anus and AS.

The patient underwent an emergency sigmoidostomy. The dilated sigmoid contains meconium was exposed. Fascia and peritoneum were anchored with four sides sutures. Surgical decompression was performed. After decompression was complete, the sigmoid was delivered through the abdominal wall without twisting it then anchored to the muscle using “spurring” sutures in both sides. The fixation suture of sigmoid seromuscular to fascia-peritoneum using watertight closure technique was performed. The end of colostomy was opened and matured by sutures between the full-thickness colon and the dermis of the abdominal wall (shown in Fig. 5).

The distal colostogram using contrast examination that was carried out 4 weeks after the surgery shown normal result. The contrast material enters the gut smoothly until transverse colon and fill the sigmoidostomy. No stricture or abnormal indentation was found (shown in Fig. 6). Until this article was submitted, the anorectal reconstruction has not been performed. Due to the COVID-19 situation, the surgical activity is limited. The patient is planned for anorectal reconstruction in 8–12 weeks after sigmoidostomy.

3. Discussion

The premature fusion of cranial sutures in AS occurs before three months of age [5]. The consequences of early closure of coronal sutures cause shorter anteroposterior diameter, with a high and prominent forehead associated with an acrocephalic head (cone-shaped) [5]. Facial dysmorphic in AS patients include midfacial hypoplasia, hypertelorism, proptosis, down slanting palpebral fissures, short and wide nasal root, and low-set ears [1,5]. Oral abnormalities including dental crowding, high-arched palate, narrow palate, and pseudo-clefts [1]. Other prominent abnormalities of AS are syndactyly of hands and feet [5]. Our patient had the most marked symptoms, including early closure of coronal sutures, prominent forehead (frontal bossing), and syndactyly on both hands and feet [5]. The maxillo-facial dysmorphic presentation were narrow high arched palate, short and broad nasal root, and low-set ears.

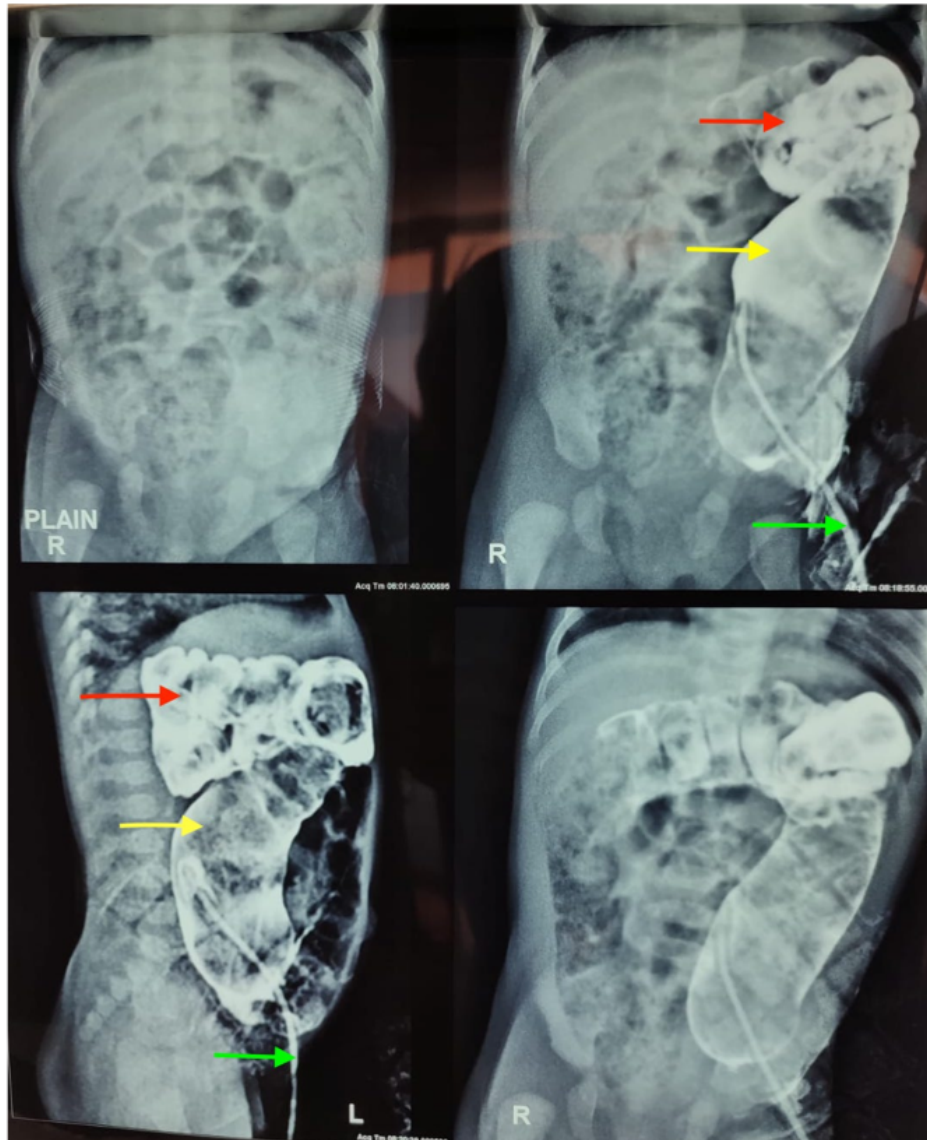


Fig. 6. The contrast material enters the gut smoothly (transverse colon [red arrow], descending colon [yellow arrow]) and fill the sigmoidostomy (green arrow). No stricture or abnormal indentation was found. (For interpretation of the references to colour in this figure legend, the reader is referred to the Web version of this article.)

Ultrasonography diagnosis of AS is difficult before third semester. Many cases remain undiagnosed until late gestation when the deformities are more apparent or until delivery [6]. Werner et al. suggest using 3D reconstruction ultrasound and MRI, that provides clearer visualization of the fetal anatomy [7]. The prenatal molecular genetic testing for FGFR2 may be performed at 16 weeks of gestation [5]. However, it is not widely available and not cost-effective, especially in developing countries like Indonesia where the genetic diagnostic resources and facilities are limited [8].

Other symptoms suggesting internal organ abnormalities apart from the imperforate anus were not present. The ARM occurs during 8–12 weeks of gestation due to incomplete development of hindgut while the exact etiology is still unknown. It is commonly associated with other congenital anomalies known as VACTERL syndrome (Vertebral defect, Anal defect, Cardiac defect, Tracheoesophageal fistula, Renal defect, Limb defect) which the diagnosis of VACTERL is when at least 3 of the defects presents. The imperforate anus should be evaluated during routine postpartum examination or shortly when the newborn fails to pass meconium or stool within 24 after birth. Lateral view x-ray in prone position should be done to determine the distance of rectal gas shadow from the anal dimple (usually have been marked by a lead marker). Our patient recto-anal distance was more than 1 cm, indicates high imperforate anus. The patients need to undergo sigmoidostomy and brought out through the left lower quadrant of abdomen. The definitive repair may

be done after 4–8 weeks of first surgery, depends on patient condition [9].

Since definitive treatment is not available, correcting the anatomical deformities may improve the patient condition and quality of life. The treatment is complex that requires a multidisciplinary team including neonatologist, pediatric surgeon, anesthesiologist, neurosurgeon, plastic and reconstructive surgeon, psychiatrist, ophthalmologist, neurologist, geneticist, and orthodontic [8]. The earliest skull surgery is done in the first 18 months of life, midface surgery is typically done after 6–8 years old, and first stage of syndactyly repairs is done between 9 and 12 months of age while the following stage is approximately 3 months later [10]. Although most patients with Apert syndrome have normal intellectual to mild intellectual disability, some individuals have been reported suffer from moderate to severe intellectual disability and emotional problem [8]. A psychiatric approach is needed for the patient who shows the tendency of emotional and behavioural disorder [5].

The differential diagnoses of AS are other genetic disorders in the acrocephalosyndactyly spectrum, including Crouzon, Carpenter, Chotzen, and Pfeiffer syndromes [11]. AS especially demonstrates similar dysmorphic characteristics with Crouzon syndrome [5]. However, AS is more asymmetric in nature and has more severe clinical manifestations than Crouzon syndrome [12].

Statement of ethics

Patient's family gives permission for patient information and photograph to be published in scientific journal anonymously.

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Author contributions

All authors attest that they meet the current ICMJE criteria for Authorship.

1. Setya Mithra Hartiastuti: conception and design, drafting, writing manuscript to final paper.
2. Risa Etika: revision and final approval.
3. Martono Tri Utomo: drafting, revision, final approval.
4. Quri Meihaerani Savitri: drafting, writing manuscript to final paper.

Declaration of competing interest

The authors declare that they have no known competing financial interests or personal relationships that could have appeared to influence the work reported in this paper.

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