CASE REPORT

Vaginal Atresia in a Case of Fraser Syndrome

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Introduction

Vaginal atresia is a congenital anomaly of the female genitourinary tract and may occur as an isolated developmental defect or as part of a complex of anomalies like Mayer Rokitansky Kuster Hauser (MRKH) syndrome, Bardet-Biedl syndrome, Kaufman-McKusick syndrome, Fraser syndrome and Winter syndrome. The situation is most urgent where the normal uterovaginal outflow is obstructed and the patient presents with progressive abdominal distension and cyclic pain, which necessitates prompt action.

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Fraser syndrome (cryptophthalmos-syndactyly syndrome, Fraser-Francois syndrome, Meyer-Schwickerath's syndrome) is named after a geneticist G.R. Fraser, who first described the syndrome in 1962 [1]. It is an autosomal recessive congenital disorder. Major criteria consist of cryptophthalmos, syndactyly, abnormal genitalia and positive family history. Minor criteria are congenital malformation of nose, ears or larynx, cleft lip and/or palate, skeletal defects, umbilical hernia, renal agenesis and mental retardation. Diagnosis is based on Thomas criteria; presence of at least 2 major and 1 minor criteria or at least 4 minor criteria [2]. This disease has been linked to a gene *FRAS1* that is involved in skin epithelial morphogenesis during early development [3].

Abnormalities of the genital tract have been reported in patients with Fraser syndrome of which the commonest abnormality in female patients has been clitoral hypertrophy [4]. The other abnormalities seen are bicornuate uterus, cystic ovaries, rudimentary uterus, vaginal atresia and indeterminate sex. This case report shows a rare occurrence of vaginal atresia in Fraser syndrome presenting with primary amenorrhea.

Case Report

A 15-year-old female presented to our OPD with complaints of cyclic pain in lower abdomen with a mass increasing in size gradually for 5 months. She was the fourth child of a non-consanguineous marriage and had not

Fig. 1 A 15-year-old girl with Fraser syndrome and right cryptophthalmos

Fig. 4 Syndactyly of both feet

Fig. 2 Syndactyly of left hand

Fig. 5 Contracted ear lobule with deficient right ear helix

Fig. 3 Syndactyly of right hand

Fig. 6 Contracted left ear lobule

attained menarche. Her height was 143 cm, weight 30 kg, BMI 14.6 and her IQ was normal. She was found to have multiple congenital anomalies such as right-sided cryptophthalmos, absence of right eyebrow, deficient right alar cartilage, bilateral contracted ear lobule with deficient right ear helix, deviated nasal septum to right, high arched palate

and syndactyly of both hands and feet (Figs. 1, 2, 3, 4, 5, 6). No other congenital abnormality was detected in lips, larynx, heart, and lungs. There was no history of any congenital anomalies and Fraser syndrome in the family. There was no thyromegaly. Abdominal examination revealed congenital umbilical hernia and a mass arising from pelvis up to 28 weeks size. Kidney, Liver and spleen

Fig. 7 Umbilical Hernia and well developed breasts

Fig. 10 Pubic hair Tanner stage 2 and per abdomen mass 28 weeks size

Fig. 8 Axillary hair sparse

Fig. 11 Blind vagina

Fig. 9 Uterus 28 weeks size

Fig. 12 TAS showing Hematocolpos with hematometra

were not palpable. The Tanner staging for breast was stage 3; pubic hair stage 2, axillary hair was sparse. Genital examination revealed a blind vagina-admitting tip of finger but no bulge, normal anus and urethral opening (Figs. 7, 8, 9, 10, 11).

Routine blood and urine examination were within normal range. Special investigation like pituitary gonadotropins was also within normal range. Transabdominal scan showed hematometra, hematocolpos (11.6×7.7 cm) and wall defect measuring 30 mm at the umbilical region with

Fig. 13 Blind vagina with no bulge of hymen

Fig. 14 Drainage of chocolate coloured menstrual blood on dividing the transverse vaginal septum

normal genitourinary system (Figs. 12, 13). Both ovaries and tubes were normal. Transperineal scan revealed a transverse vaginal septum of size 1.3 cm in the mid-vagina. There were no apparent skeletal defects in radiological examination. Echocardiogram was found to be normal.

Surgery was planned after taking parents consent to create a neovagina and excise the septum making the uterovaginal outflow tract patent. She underwent an elective surgery under spinal anaesthesia. A space was created between rectum and urethra; septum incised avoiding injury to adjacent structures and around 2 L of altered chocolate colored menstrual blood drained out (Fig. 14). This division of the complete septum in the upper half was combined by a vaginal pull through procedure getting the proximal part of vagina more distally to allow its marsupialization to the introitus to avoid stenosis. Post operative period was uneventful and she was discharged on the fourth post operative day with the advice to use a mould as vaginal dilator for 3–4 h each day to avoid vaginal stenosis. On follow up, she reported normal menstrual cycles without any pain (Figs. 15, 16).

Discussion

The incidence of transverse vaginal septum or partial vaginal agenesis is 1 in 70,000 live births [5]. Isolated

Fig. 15 Cervix seen after formation of neovagina

Fig. 16 Immediate post-op with disappearance of abdominal swelling

vaginal atresia is an extremely rare finding and is the second most common cause of primary amenorrhea [6]. Most cases occur as Mullerian agenesis or are associated with the more common MRKH syndrome. Our case fulfilled the Thomas criteria; 3 major criteria—cryptophthalmos, syndactyly and abnormal genitalia and 3 minor criteria—congenital malformation of nose, ears and umbilical hernia thus establishing the diagnosis to be Fraser syndrome. The incidence of Fraser syndrome is 0.043 per 10,000 live births and 1.1 in 10,000 still births [7]. By autozygosity mapping, McGregor et al. (2003) located the Fraser syndrome locus to chromosome 4q21. The life expectancy of Fraser syndrome is <1 year and diagnosed most often during fetal autopsy.

Variants of vaginal atresia, formerly termed partial vaginal agenesis currently are classified as variants of transverse vaginal septum. Canalization of the uterovaginal canal is believed to occur from the caudal to the cephalic aspect, with an epithelial lining derived from the urogenital sinus. So, failure of vaginal canalization at various levels of

the vaginal plate results in the transverse septum, the most common location of transverse septum is the upper vagina, followed by middle, with least common location being the lower third [8]. There are two kinds of vaginal septa, transverse and longitudinal. Transverse vaginal septum may be complete resulting in retained secretion of genital tract in neonate, or partial with pin point opening allowing for menstrual flow. If it is located in lower part of vagina, it is more likely to be complete. Longitudinal vaginal septum is usually benign, though obstruction can occur when septum is associated with other mullerian anomalies and surgical excision may be required [8].

In spite of the high frequency of genital abnormalities, there have been no endocrinal studies in Fraser syndrome [4]. In this patient, gonadotropins were normal. This probably means that the genital abnormalities are structural in origin and not hormone dependent. The role of medical management of vaginal atresia is limited. In patients with functioning endometrium and an obstructed outflow tract, temporary hormonal manipulation of the menstrual cycle may be necessary until a patent genital tract can be created. Most patients with functional endometrial tissue do not have a fully functioning uterus. In patients who have a patent cervix and absent vagina (very rare), creation of a neovagina will allow for outflow of the menstrual blood as in this case. On the other hand, the creation of a cervix in patients without a patent cervix is associated with a very high rate of infection and mortality.

A series of patients with uterovaginal obstruction in infancy was reported by Omar Mansour and Hani Morsi (2008) [6]. They described 11 cases of hydrometrocolpos resulting from transverse vaginal septum and managed by incision of septum with marsupialization. Incision of the

septum together with marsupialization is the standard treatment. Whenever anatomical evaluation is not precisely done, vaginostomy, and staged repair is recommended. Postoperative complications include stenosis, dyspareunia, and decreased vaginal lubrication.

Conclusion

This is a very rare case of vaginal atresia associated with Fraser syndrome managed surgically resulting in good outcome.

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