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CASE REPORT

A Rare Case of Fraser Syndrome with Partial Vaginal Agenesis and Its Successful Reconstructive Cosmetic Management: A Case Report

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Abstract
Fraser syndrome is a rare disorder with autosomal recessive inheritance having a wide spectrum of phenotypic appearances. A fourteen-year-old female presented to us with a chief complaint of acute retention of urine without any cyclical abdominal pain with syndromic appearance. She had partial vaginal agenesis which was treated by successful reconstructive surgery by lotus petal flap technique followed by recanalisation. Objective of this report is to highlight the rare disease of Fraser syndrome along with successful rare surgical management.

Keywords Fraser syndrome · Vaginal agenesis · Cryptophthalmos · Lotus flap

Introduction

Fraser syndrome is a very rare disorder with autosomal recessive inheritance having a wide spectrum of phenotypic appearances [1]. The incidence of this syndrome is 0.043 per 10,000 live born infants and 1.1 in 10,000 stillbirths, making it a rare syndrome [2]. In 1962 George Fraser established the complete description of Fraser syndrome including cryptophthalmos, cutaneous syndactyly, genital anomalies, laryngeal stenosis, ear malformations, and renal abnormalities. The diagnostic criteria of Fraser syndrome were defined by Thomas et al. in 1986, in which they classified them as major and minor criteria (Table 1) [1]. For diagnosis either two major criteria and one minor criterion, or one major criterion and four minor criteria must be met. Cryptophthalmos is considered to be the single most important malformation of Fraser syndrome and occurs in approximately 88% of cases [1]. In the absence of cryptophthalmos, Fraser syndrome can easily be confused with other malformation syndromes, such as Walker-Warburg or Peters' Plus syndromes, MURCS (M¨ullerian duct aplasia, renal aplasia and cervical dysplasia) association, and Bardet–Biedl Syndrome. Agenesis of vagina is a congenital anomaly of the female genitourinary tract and may occur commonly 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. It is an emergency situation where the normal utero-vaginal outflow is obstructed and the patient presents with progressive abdominal distension and cyclic pain, which necessitates urgent action. 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. The purpose of our article is to discuss a rare, unreported case of Fraser Syndrome with partial vaginal agenesis in a fourteen-year-old girl, its diagnostic criteria and its successful reconstructive surgery of partial vaginal agenesis by lotus petal flap technique and recanalisation.

Case Report

A fourteen-year-old girl presented to us with a chief complaint of retention of urine and pain lower abdomen for two days with syndromic appearance without any history of previous catheterisation. She was the second daughter of non-consanguineous parents without any history of birth asphyxia or developmental delay with normal intelligent quotient (IQ). She had not attained menarche. Her elder

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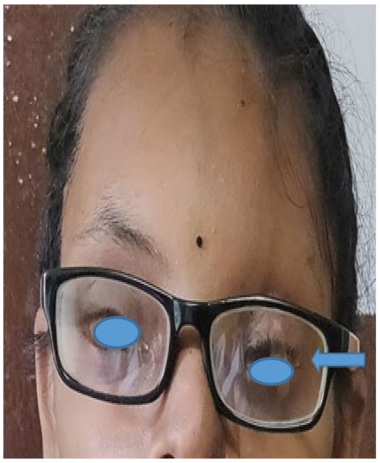
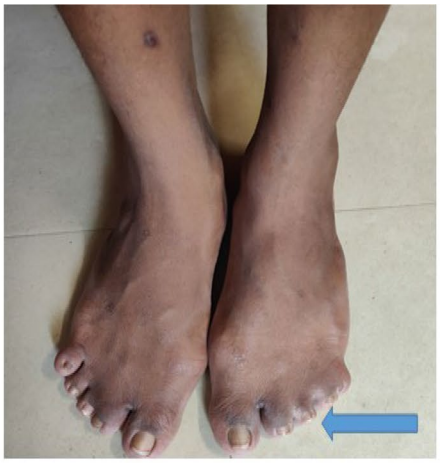
Table 1 Diagnostic criteria of Fraser syndrome

Major criteria	Minor criteria
Cryptophthalmos	Congenital malformation of nose
Syndactyly	Congenital malformation of ears
Genital abnormality	Congenital malformation of larynx
Sibling with Fraser syndrome	Cleft lip +/− palate
	Skeletal defects
	Umbilical hernia
	Renal agenesis
	Mental retardation

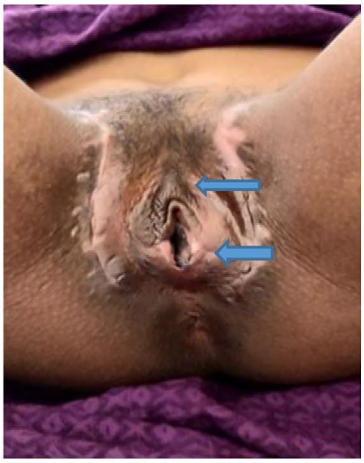
sister had similar syndromic profile with cryptopthalmia and imperforate hymen, which was corrected by minor surgery. Her facial profile revealed low set ears, small eyes along with sparse eye lashes and eyebrows, syndactyly in bilateral foot (Fig. 1; Pic. 1, 2). She had breast development tanner stage 3, Pubic hair stage 3, and Axillary hair stage 3. She had complete closure of eyes at birth, later surgically the eye lids were separated (cryptopthalmia). On abdominal examination she was having distended bladder with tenderness in lower abdomen and a low seated umbilicus. There were no skeletal defects in radiological examination. Other investigations were normal.



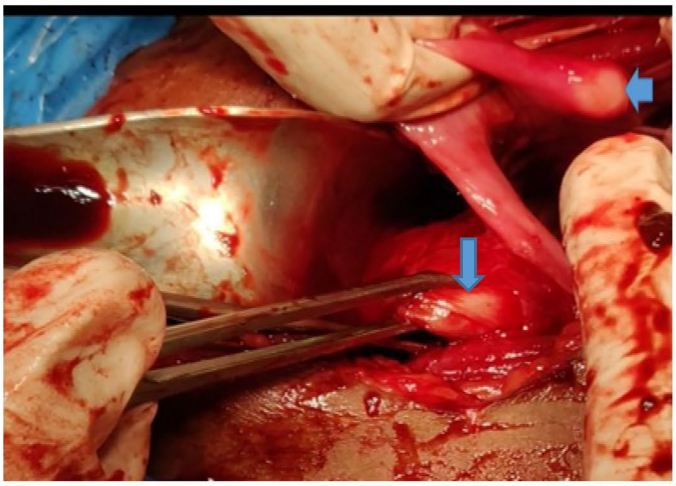
(PIC-1:Corrected cryptopthalmus)



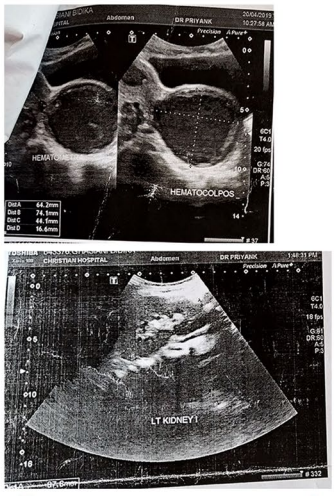
(PIC-2: Bilateral syndactyly)



(PIC-3: Neovagina after lotus petal flap vaginoplasty)



(PIC-4:Intraoperative view upper arrow- Blind end of fallopian tube ;lower arrow- Unicornuate uterus)



(PIC 5: USG showing Haematocolpos)

Fig. 1 Pictures of patient with intraoperative photography

Vaginal Examination Perineal skin healthy and pubic hair was present. Urethra was patent, anus appeared normal, dimple like vaginal opening. Clitoris and labia appeared normal (pic. 3).

Ultrasonography (USG) USG Abdomen and pelvis showed that she had haematocolpos with partial vaginal agenesis in lower two thirds of the vagina and solitary left kidney and ureter with corticomedullary differentiation preserved (Fig. 1; Pic. 5).

Fraser syndrome criteria She had all major criteria and one minor criteria confirming her as a case of Fraser syndrome.

Surgical procedure (Fig. 2; flow chat 1: Fig. 1; Pic. 3, 4) Surgery was planned after counselling regarding prognosis of operation and taking parents' consent to create a neovagina making the uterovaginal outflow tract patent in a two sitting procedure. She underwent a successful Partial vaginal reconstruction with a Lotus petal flap technique with consultation from plastic surgery department of our hospital. The procedure was selected because it was safe, with good functional and cosmetic results in young women. She was discharged with continuous oral combined oestrogen and progesterone supplements and planned for review after two months. Unfortunately she missed her pills and had an episode of painful cryptomenorrhoea.

Transperineal Ultrasound: Transverse vaginal tissue gap between neovagina and blind upper vagina of thirteen millimetre thickness. Magnetic Resonance Imaging (MRI) was not done due to claustrophobic symptoms of patient.

She underwent a septal resection and drainage of haematocolpos and haematometra followed by anastomosis of lower neo-vagina and upper blind vagina by abdominovaginal route (Fig. 2; flow chart 2). It was noted that a unicornuate uterus was present with a normal cervix. Both ovaries were long and tubular. Left side fallopian tube had no fimbrial end. Right side fallopian tube was absent. A Foleys catheter was used as a drain which was removed after three week follow-up.

Follow-up She was followed up subsequently every 2 months over one year. She had normal cyclical menstruation without any hormonal support and had no further complaints. There was no need of periodic dilation after surgery.

Discussion

Fraser syndrome, in a living healthy female is a very rare autosomal recessive multiple malformations syndrome. The diagnosis of Fraser syndrome is done on clinical examination using the diagnostic criteria proposed by Thomas et al. [1]. All the major criteria were observed in our case, i.e. cryptophthalmos, syndactyly, and abnormal genitalia along with sibling with cryptophthalmos. The minor features observed are congenital malformations of the ear and renal agenesis.

The incidence of transverse vaginal septum or partial vaginal agenesis is 1 in 70,000 live births [3]. Vaginal agenesis is a rare condition with devastating repercussions on fertility

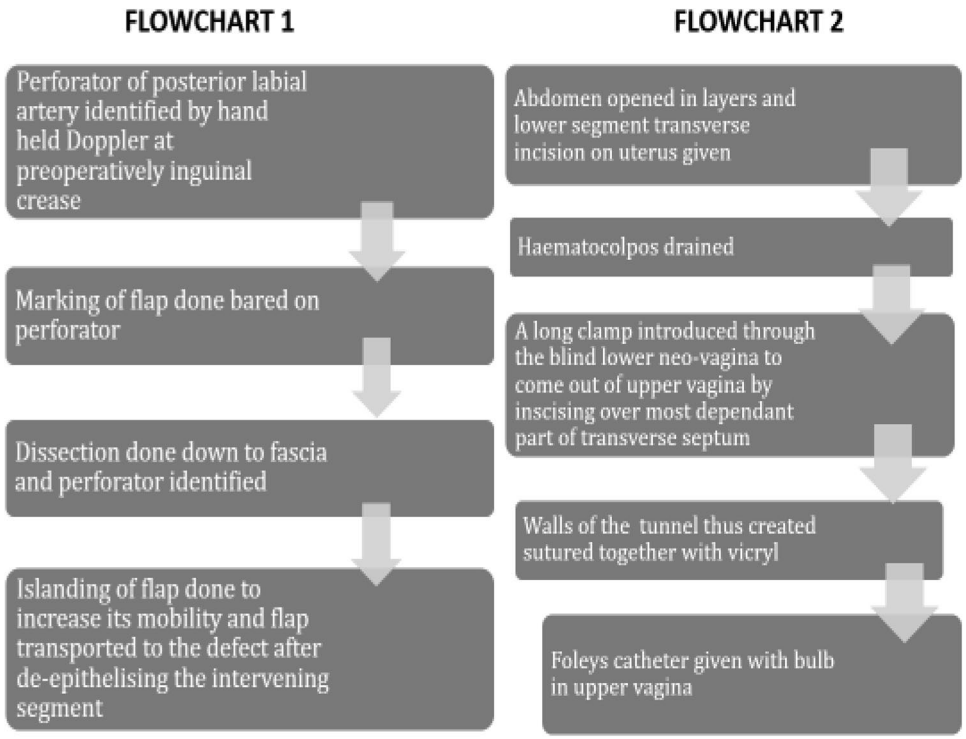


Fig. 2 Operative flow chart

and sexual function. The most common location of transverse septum is the upper vagina, followed by middle, with least common location being the lower third. The reconstruction of vaginal atresia is challenging, as one has to maintain the important excretory, menstrual and sexual functions. The ideal time for intervention is at or after adolescence, when the woman has reached physical and psychological maturity. Vaginal dilation should always be recommended as the first line of treatment in vaginal agenesis. Whenever anatomical evaluation is not precisely done staged repair is recommended. There are several options for reconstruction for perineal defects like Williams vulvovaginoplasty, Vecchietti procedure, McIndoe Reed, Davydov but lotus petal flap technique was adopted in this case.

Zhihong Xie et al. [4] in a retrospective study of 67 patients with congenital vaginal atresia (from March 1984 to March 2015) observed for lower vaginal atresia, 25 patients successfully underwent vaginoplasty at the lower portion of the vagina. 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.

Adoption of a staged procedure was decided upon in consultation with plastic surgery department to allow for proper graft acceptance before establishment of menses. This helped reduce infection rates. Postoperative complications include stenosis, decreased vaginal lubrication, graft rejection, dyspareunia and infections. The overall life expectancy in Fraser Syndrome as reported in literature is less than 10 surviving cases of women over the age of 20 years.

Conclusion

A very rare case of partial vaginal aplasia associated with Fraser syndrome was managed surgically and resulted in good outcome. Though it's a rare case but the syndromic association with good outcome after surgery makes it very important for academicians. Meticulous planning and stepwise surgery by lotus flap technique can increase the success rate in vaginal atresia.

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About the Author

Sambedana Panigrahi Dr. Sambedana Panigrahi has done her graduation and post-graduation from SCB Medical College and Hospital. At present she is working as senior resident in SCBMCH, Cuttack, Odisha. She has presented papers in many national and international conferences and is interested in clinical research.