Case Report

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Vaginal Atresia-One Amongst the Manifestations of Rare Charge Syndrome

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Abstract

Introduction: Charge syndrome is a rare genetic disorder caused due to mutation in CHD7 gene and characterised by the presence of multiple congenital defects. Congenital genital defect is a minor abnormality of this syndrome. Here we are reporting a very rare case of Charge syndrome with vaginal atresia where vaginoplasty was done with uterovaginal tract recanalisation.

Case: A 15yr old girl presented with complaints of primary amenorrhoea and cyclical crampy lower abdominal pain for 7 months. It was also found that she had a poor scholastic performance, reduced vision in her left eye and difficulty in hearing since her childhood which however was unevaluated. Her cardio vascular system examination revealed pansystlic murmur. The ECHO cardiogram suggested a large restrictive ostium secundum atrial septal defect (ASD) with mild tricuspid regurgitation (TR) & mild pulmonary artery hyperplasia (PAH). Ultrasonography (USG) showed collections inside the uterine cavity suggestive of hematometra and examination of external genitalia revealed the presence of a blind vaginal pit suggestive of vaginal atresia. Presence of all the above clinical findings directed towards further evaluation which further revealed presence of multiorgan defects fullfilling all the criteria of Charge syndrome. She underwent ASD closure followed by Mc Indoe's vaginoplasty successfully.

Conclusion: While approaching a patient with multiorgan/multisystem abnormalities, a keen observation of its relation to any syndrome paves a path for its easy management, by prioritizing the immediate treatment and interval treatment. A multidisciplinary approach is always prudent. Vaginal atresia with presence of uterus could be managed successfully by surgical intervention, amongst which Mc Indoe's vaginoplasty may be preferred over other techniques.

Keywords: Charge syndrome; Vaginal atresia; Congenital defect.

Introduction

Charge syndrome is a rare genetic disorder caused due to mutation in CHD7 gene- an ATP dependant chromatin remodeler. Charge itself abbreviates its manifestation, C-Coloboma/Microphthalmia, H-Heart defects, A-Atresia Choana, R-Retardation of growth, G-Genital abnormalities, E-Ear abnormalities. However, the diagnosis of Charge syndrome can be made clinically, if there is atleast 2 major features and any number of minor features. Major features includes- coloboma of eye/microphthalmia, choanal atresia, conductive hearing loss, cranial nerve abnormalities. The minor features includes- heart defects, cleft lip/palate, trachea-esophageal fistula, kidney abnormalities, genital abnormalities, growth deficiency, hockey stick palmar crease etc. The commonly seen genital abnormalities are hypoplastic labia, small or

missing uterus in females, while lack of puberty without hormone intervention or delayed puberty seen both in male & females.[1]

Vaginal atresia is one of the congenital anomalies in development of mullerian structures and the defect being in the vertical fusion of the female genitourinary tract. In this syndrome the lower portion of the vagina is represented by fibrous tissue while the superior structures (uterus) are developed.[2] This condition is usually seen associated with other malformations of urinary system or specific syndromes viz. Mayor Rokitansky Kuster Hauser Syndrome (MRKH), Frasi syndrome etc, yet very rare association being established with Charge syndrome. Here we are reporting a very rare case of Charge syndrome with vaginal atresia who was managed successfully by a systematic multidisciplinary approach. We could not found any reported case of Charge syndrome where the vaginal atresia was managed and uterovaginal recanalisation was established. Hence probably it is the first such case to be reported.

Case Report

A 15yr old girl presented to our OPD in the month of February 2019 with complaints of primary amenorrhoea and cyclical crampy lower abdominal pain for 7 months. Her cyclical lower abdominal pain was incidious in onset, spasmodic, non progressive in nature, lasting for 4-5 days in a month and radiating to lower back. It was associated with bloating sensation and nausea. She was the second child of a non consanguineous marriage. Unlike her siblings, she was grossly short for her age. Her mother also gave history of poor scholastic performance and reduced vision in her left eye, and also revealed that she had difficulty in hearing since her childhood which however was unevaluated.

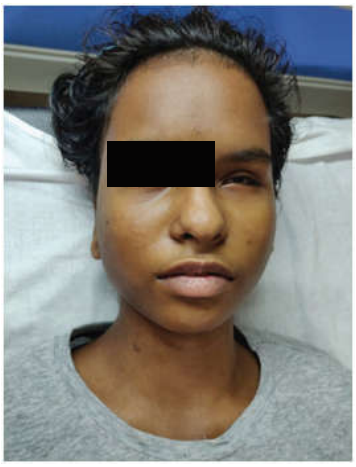

Fig. 1: Left microphthalmia with only perception to light.

She gave a history of being evaluated outside for the same with Ultrasonography(USG) of her pelvis which showed a bulky uterus with dimensions of 10.7 x 3.8 x 5.2cms and 113.2cc in volume. Moderate to gross amount of fluid collection noted within the endometrial cavity suggestive of hematometra. The patient was admitted to our hospital for further management. On admission, the detailed history was taken, general physical examination revealed left microphthalmia with only perception (Fig 1). of light. Secondary sexual characters, however revealed Tanner's stage 3, which was normal for her age. In her systemic examination, cardiovascular system (CVS) finding was suggestive of pansystolic murmur. Of all the above clinical finding directed towards multiorgan defects arouse a suspicion of an undiagnosed syndrome. Her repeat USG abdomen & pelvis showed the following findings: Left kidney is relatively smaller in size & measured (6.9 x 3.4) cms with mild dilatation of pelvicalceal system, while the right kidney was having normal dimensions, uterus was of size (10.2 x 3.6 x 4.5) cms with 89cc volume with heterogenous collection in endometrial cavity suggestive of hematometra.

Two dimensional ECHO cardiography finding was suggestive of large restrictive ostium secundum atrial septal defect (ASD) with mild tricuspid regurgitation (TR) & mild pulmonary artery hyperplasia (PAH). With this she was diagnosed with Charge syndrome. She was planned for two surgeries i.e closure of ASD and vaginoplasty. First of all surgery for closure of ASD was performed which was successful and vaginoplasty operation was planned after 3 months of it. Our team for vaginoplasty surgery was consisting of Gynaecologists, Plastic Surgeons and Anaesthesiologists. We planned for an abdomino pelvic approach. For vaginal atresia she underwent Mc Indoe's vaginoplasty.

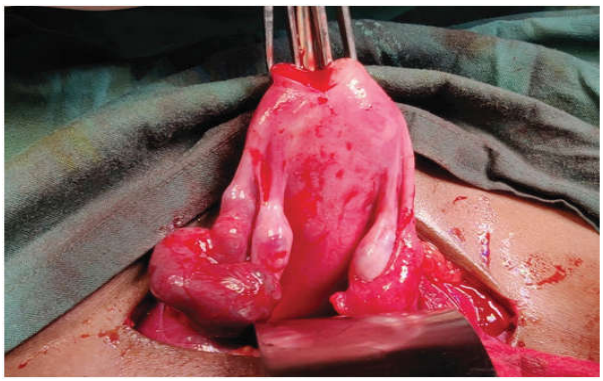

Fig. 2: Uterus (8 weeks size) with bilateral hematosalphinx and bilateral normal ovaries.

Abdomen was opened with a pfanensteil incision. The intraoperative findings are: uterus was around 8 weeks size, bilateral hematosalphinx was present while the bilateral ovaries appeared normal (Fig. 2).

A small vertical incision was given on the fundus and hematometra of around 60cc was drained. A Hegar's dilator was introduced through this fundal opening upto the cervical canal and kept there. Further, UV fold was dissected and bladder was released to facilitate vaginal procedure. Vaginal examination revealed presence of urethral opening & a blind vaginal pit of around one centimeter depth (Fig 3).

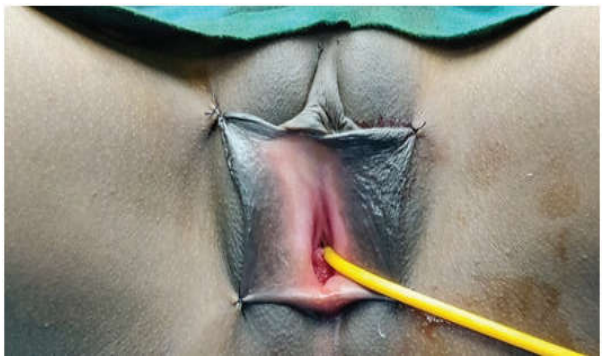

Fig. 3: Perineum showing, urethral opening and anal opening with a blind vaginal pit.

A 'Y' shaped incision given posterior to the urethral opening & 3 triangular flaps elevated via slow dissection between urethra & anal canal. A finger introduced into the anal canal to ensure the right plane.

Neovagina was created along the direction of the tip of the hegar's dilator placed from above. The uterocervical canal was opened into the cavity of the neo vagina by dissecting the lowermost part of the atretic cervix. A foley's catheter was passed from vaginal opening through the neovagina and cervical opening and was placed inside the uterine cavity. The balloon was inflatted with 10 cc of distilled water and was left in situ with sterile foam & double condom.

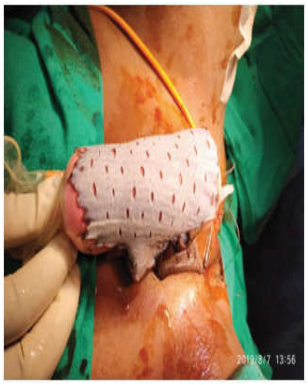

Fig. 4: Soft sponge mould. **Fig. 5:** soft sponge mould with split thickness skin graft.

A soft sponge mould was prepared by using a sterile foam & covering it with a double condom (Fig. 4). The distal end of the foley's catheter was introduced by the side of the mould below the condom cover.A split thickness skin graft was harvested from posterior surface of left thigh. The graft was perforated in multiple places with a scalpel and was covered around the sponge mould as shown in fig. 5 This prepared mould with skin graft over it was introduced inside the neo vaginal canal. The mould was kept in place by suturing labial folds over the mould by interrupted non absorbable sutures and applying tight dressing. Both intra operative and post operative periods were uneventful. Daily dressing was done, Around 15 ml of dark coloured fluid was collected in the drainage bag through the foley's catheter within 24 hours of surgery which is suggestive of the remaining hematometra drained subsequently. Broadspectrum injectable antibiotics were given for five days following operation. The vaginal mould was removed on day six of surgery, when the skin graft was found adherent to the raw surfaces of the neo vagina. She was discharged with the advice regarding the use of a hollow acrylic vaginal mould. The frequency of changing of the mould and its hygiene was explained. She was reviewed after 15 days when a diagnostic hysteroscopy showed the cavity of the neovagina with intact skin graft and the cervical os being normal in morphology.

She reviewed back after another 15 days with history of spotting per vagina for 1 day and bleeding pv for 1 day. In following months, she started to have her cyclical menstruation.

Discussion

The incidence of Charge syndrome was reported to range from 0.1 - 1.2 per 10,000 births.[3] Though genital manifestations including hypoplastic external genetalia is seen, it is profoundly appreciated in males than females. Our suspicion of syndromic presentation arouse when multisystem abnormalities were observed, the details of which has already been described. All of this directed towards making a diagnosis of Charge syndrome.

As the syndrome was diagnosed, we had a management strategy that included multispeciality intervention which was executed priority wise. She underwent ASD correction by CTVS surgeons first, and 3 months later her vaginal atresia was managed.

The genital abnormality seen in our case i.e vaginal atresia is very rarely found in this syndrome. Vaginal atresia/transverse vaginal septum/partial

vaginal agenesis as such having an incidence of 1 in 70,000 live births and is most commonly seen associated with MRKH syndrome, Frasi syndrome, Winter syndrome, Bardet-Biedl syndrome etc.[4,5] Vaginal atresia is the second most common cause of primary amenorrhoea. A uterus is absent in 2% to 7% of patients with vaginal agenesis.[6] In such cases the treatment would be mostly to provide sexual function by using vaginal dilators, a non surgical technique as the first line of management. It is usually preferred in women who are ready to start their sexual activity.[7] Unlike our case who had a uterus and presented with hematometra for which she was managed surgically. Various surgical procedures performed in such conditions to create a neo vagina are, Mc Indoe Reed vaginoplasty, Vecchietti procedure, Davydov procedure. In some of the procedures the principle of surgery is to create a neovagina based on the tissues used as a graft, for example- skin is used to create neovagina in Mc Indoe's procedure, peritoneum in Davydov procedure and so on.[8] On the other hand in Vecchietti procedure, traction is used as a principle to create neovagina.[6] However most commonly used procedure amongst this is the Mc Indoe's Reed's vaginoplasty.

A retrospective study done by Karapinar os et. al., observed seven cases who presented with complaints of either primary amenorrhoea or unable to perform sexual intercourse.[9] All of them underwent modified Mc Indoe's vaginoplasty and was found that only one among them had a post operative infection, dyspareunia occurred in two people (including one post op infected) & the rest five people had no intra, post operative complications and experienced comfortable sexual life. In another publication by Bastu E et. al. where a long term follow up of 23 patients who underwent Modified Mc Indoe's vaginoplasty shows that, out of 14 patients who used moulds regularly, only one experienced pain during intercourse and two of three patients who used dilators or moulds irregularly had severe pain during intercourse.[10] Thus the role of regular use of vaginal dilators or moulds should be emphasised.

Hence in our case, who underwent Mc Indoe's vaginoplasty, primarily for establishing outflow tract between uterus and vagina was successfully treated.

Conclusion

While approaching a patient with multiorgan/ multisystem abnormalities, a keen observation of its relation to any syndrome paves a path for its easy management, by prioritizing the immediate treatment and interval treatment. A multidisciplinary approach is always prudent. Vaginal atresia with presence of uterus could be managed successfully by surgical intervention, amongst which Mc Indoe's vaginoplasty may be considered over other techniques. With regular usage of dilators/moulds the outcome of such surgeries could be advantageous in providing a woman a healthy menstrual cycle as well as her sexual life.

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Conflict of Interest: None.

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