CASE REPORT

MAYER-ROKITANSKY-KUSTER-HAUSER (MRKH) SYNDROME TYPE 2: ATYPICAL PRESENTATION OF RARE CASE

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ABSTRACT

Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome is a congenital malformation characterized by an absence of the vagina associated with a variable abnormality of the uterus and the urinary tract but functional ovaries. We are reporting atypical presentation of rare case. Patient had obstructed left inguinal hernia with genitourinary and skeletal deformity. Latter it diagnosed as Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome which is a rare disease.

Key word: Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome;Mullerian Defects; Urinary Tract Anomalies.

INTRODUCTION

Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome consists of vaginal aplasia with other müllerian (ie, paramesonephric) duct abnormalities. Type I MRKH syndrome is characterized by an isolated absence of the proximal two thirds of the vagina, whereas type II is marked by other malformations, including vertebral, cardiac, urologic (upper tract), and otologic anomalies. This condition causes the vagina and uterus to be underdeveloped or absent. Affected women usually do not have menstrual periods due to the absent uterus. Often, the first noticeable sign of MRKH syndrome is that menstruation does not begin by age 16 (primary amenorrhea). Women with MRKH syndrome have a female chromosome pattern (46,XX) and normally functioning ovaries. They also have normal female external genitalia and normal breast and pubic hair development.Surgical correction of the vaginal anomaly permits normal sexual function Although women with this condition are usually unable to carry a pregnancy, they may be able to have children through assisted reproduction.

CASE REPORT

A 14year young female presents with irreducible left groin swelling for 2days with vomiting and abdominal pain.

On examination, all vital parameters are normal, short neck, low hair line (Fig 1, 2); restriction of movement of neck, sprengle's shoulders (Fig 3) was present. Abdomen is soft and localized 6x4x3cm, tender, warm, non-reducible left inguinal swelling present without impulse on coughing.

Figure 1: Short neck

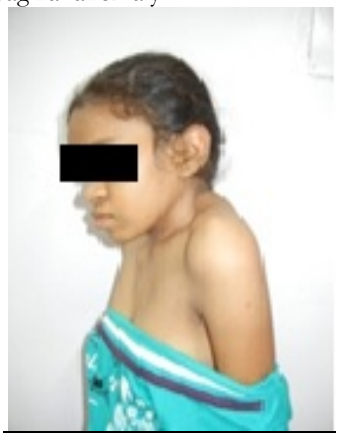


Figure 2: Low hair line

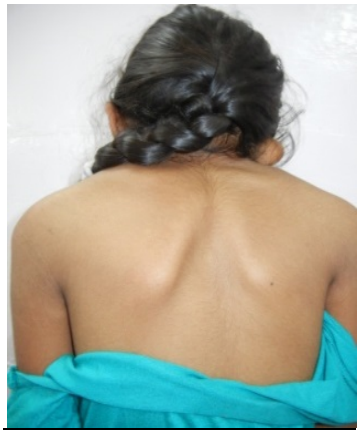
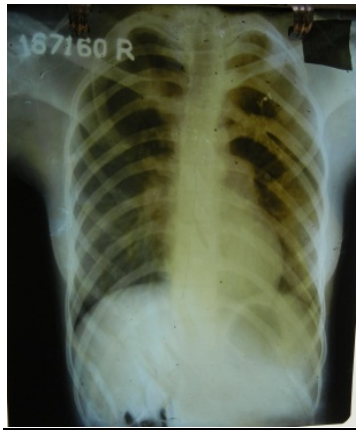
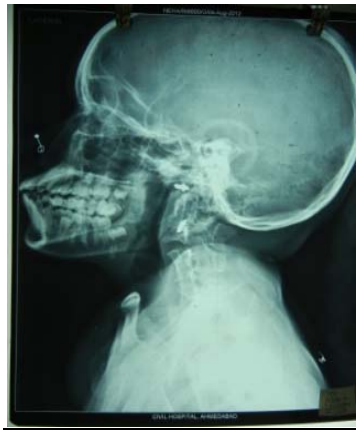


Figure 3: Sprengle's shoulders





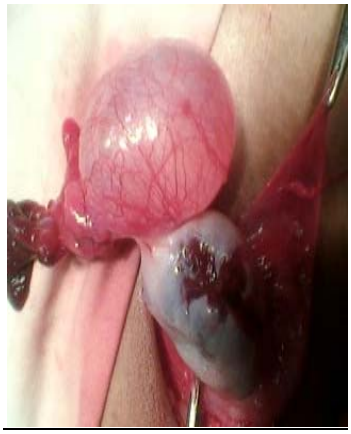


Figure 4 **Figure 5** **Figure6**

Figure 4: Fusion of posterior end of 5 &6th ribs
Figure 5: Fusion of C2-3-4 and C5-6-7 vertebrae
Figure 6: Gangrenous left ovary

On investigation, hemogram, renal function, electrolyte, liver function was within normal limit. X-ray chest showing fusion of posterior end of 5 &6th ribs (Fig 4) and x-ray cervical spine showing fusion of C2-3-4 and C5-6-7 vertebrae (Fig 5). Ultrasonography of abdomen shows absence of both uterus and left kidney and 17mm defect in left inguinal region showing herniating sac contain bowel loops and left ovary with compromised vascularity. Right ovary is in its place and normal.

On emergency exploration there was gangrenous left ovary with 3 x 3cm complex cyst present (Fig 6). Left oophorectomy with herniorraphy was done. USG findings lead to think us some congenital anomaly and on further work up it proved to be MRKH type II.

Patient was well in post-operative days and discharge on 3rd post-op days. Patient is investigated for hearing and cardiac anomaly but no abnormality detected. Counseling for further management of disease and associated abnormality of syndrome has been done.

DISCUSSION

The Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome is characterized by congenital aplasia of the uterus and the upper 2/3rdpartof the vagina in women showing normal development of secondary sexual characteristics and a normal 46, XX karyotype[1,2].

Other associated malformations include (type II or MURCS association)[3] Renal (unilateral agenesis, ectopic kidney or horseshoe kidney); Skeletal and, in particular, vertebral (Klippel-Feil anomaly; fused vertebrae, mainly cervical; scoliosis); Hearing defects and more rarely, cardiac and digital anomalies (syndactyly, polydactyly).

Isolated utero-vaginal aplasia is referred to type I (isolated) MRKH syndrome. Incomplete aplasia with other malformations is generally referred to as MURCS association (or type II MRKH syndrome). In this case, the term GRES (Genital Renal Ear Syndrome) can also be used[4].

The incidence of MRKH syndrome has been estimated as 1 in 4500 female births. The mode of inheritance seems to be autosomal dominant with an incomplete degree of penetrance and variable expressivity. Type I (isolated) MRKH is less frequent than MURCS association.

The first clinical finding is generally a primary amenorrhea in patients presenting with a normal female phenotype, normal 46, XX karyotype, and normal and functioning ovaries without sign of androgen excess. External examination reveals completed puberty with normal secondary female sexual characteristics (pubic hair and breast development are Tanner stage 5) and normal external genitalia. At the same time, the vagina is reduced to a more or less deep (2–7 cm) vaginal dimple.

Associated Urinary upper tract malformations[5]: Associated upper urinary tract malformations are found in about 40% of cases with MRKH syndromelike unilateral renal agenesis (23–28%), ectopia of one or both kidneys (17%), renal hypoplasia (4%), horseshoe kidney and hydronephrosis

Associated skeletal abnormalities[6]: These anomalies mainly involve the spine (30 to 40%) and less frequently, the face and the limb extremities. Spinal malformations encountered in MURCS association are scoliosis (20%), isolated vertebral anomalies (asymmetric, fused or wedged vertebrae), Klippel-Feil association (fusion of at least two cervical segments, short neck, low hair line, restriction of neck motion) and/or Sprengel's deformity, rib malformation or agenesis, and spina bifida. Face and limb malformations are mainly brachymesophalangy, ectrodactyly, duplicated thumb, absent radius, atrio-digital dysplasia (Holt-Oram like syndrome) and facial asymmetry.

Associated hearing impairment: Auditory defects or deafness are associated with 10 to 25% of MURCS

patients, they often concern conductive deafness due to middle ear malformations, such as stapedialankylosis, or sensorineural defects of varying severity.

Associated heart malformations: The association of MRKH with heart malformations is less common. All reports involved lethal or severe cardiac defects evocating Holt-Oram or velocardiofacial-like syndromes requiring surgery.

Sometimes patient presenting with hernia and accidentally diagnosed as MRKH syndrome.

Trans-abdominal ultrasonography[7] reveals an absence of the uterine structure between the bladder and the rectum. Magnetic resonance imaging (MRI) is used to diagnose uterine as well as renal and skeletal malformation

Treatment of utero-vaginal aplasia[8] is Nonsurgical creation of a neo-vaginais by Franck's dilator method. For Surgical creation of a neovagina three methods are currently in use. 1. The Abbe-McIndoe operation; 2. The Vecchietti operation; and 3. Sigmoidal colpoplasty.

Above all, a careful psychological preparation of the patient before any treatment or intervention is of major importance. Infertility will be the most difficult aspect of the disorder to accept but *in vitro* fertilization of patients own eggs and to use surrogate pregnancy can be helpful.

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