Original Research Paper

Volume - 10 | Issue - 6 | June - 2020 | PRINT ISSN No. 2249 - 555X | DOI : 10.36106/ijar

Gynecology

MAYER ROKITANSKY-KUSTER-HAUSER SYNDROME: SYNDROME OF AGENESIS- A CASE REPORT

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ABSTRACT Mayer-Rokitansky-Kuster-Hauser syndrome is the complete failure in the development of the mullerian duct resulting in the absence of the fallopian tubes, uterus and most of the vagina .It affects atleast 1 in 4,500 women. The patients have normal female karyotype .Renal ectopy and agenesis, skeletal abnormalities, as well as cardiac abnormalities are associated. The MRKH syndrome has been linked with decreased galactose-1-phosphate uridyl transferase activity leading to increased galactose exposure. Here we report a case of MRKH syndrome in a 17yr old female who presented with primary amenorrhoea.

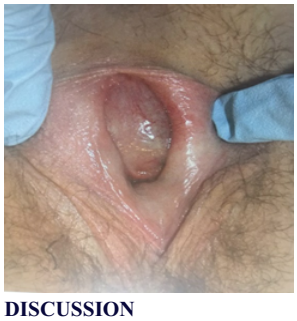
KEYWORDS :

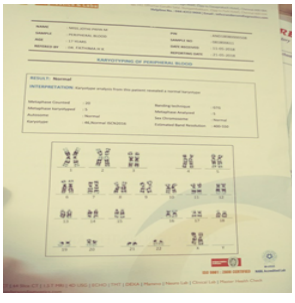
INTRODUCTION:

Mayer–Rokitansky–Kuster–Hauser syndrome (MRKH syndrome) is named after its most famous discoverer Baron Karl von Rokitansky , a physician and professor at the University of Vienna. In 1829 and in 1838, Mayer–Rokitansky described a syndrome that includes agenesis of the uterus and vagina, while Kuster then observed a correlation with urological defects. For this reason, this condition is also known as MRKH syndrome .[1] . Mayer-Rokitansky-Kuster-Hauser syndrome is an autosomal dominant inheritance with incomplete penetrance and variable expressivity. Because of the variable inheritance, penetrance and expressivity it is divided into two types: type 1:inwhich only the structures developing from the mullerian duct is affected and type2: where the same structures are affected but is characterized by additional malformations of the other body systems most often including renal and skeletal and cardiac systems.

CASE REPORT:

A 17-year-old girl accompanied by her mother came with the complaints of primary amenorrhoea. Patient was a known caseof hypothyroidism on T.Thyronorm 25 micrograms for the past 2yrs.She was the only child had no siblings. Patient gave H/O taking Estradiol 4 mons back. Her mother attained menarche at the age of 15yrs. H/O delayed menarche on the paternal side. No H/Oany cranial tumours or any other malignancy in the family. On examination shewas 146cm and her weight was38kgs,a butterfly shaped swelling present in the anterior aspect of the neck her IQ was normal. Tanner staging- Breast development- B4,Pubic hairdevelopement-P4,Axillary hair-A2,Externalgenitalia- Labia majora and labia minora – Infantile. Ultrasonography and MRI showed that the uterus was hypoplastic with nonvisualization of the ovaries in the ovarian fossa. Kidney and urinary bladder appeared normal. Karyotype was 46, XX (female). Hormonal study for AMH was 0.85 ng/ml, follicle-stimulating hormone and luteinizing hormone was within normal limits. Based on clinical features, radiological, biochemical,our first case was diagnosed as MRKH syndrome Type 1.





DISCUSSION

MRKH syndrome is subdivided into two types: type I (isolated) or Rokitansky and Type II or MURCS association.[1] MRKH syndrome is the second most common cause of primary amenorrhea after gonadal pathology.[2] The first sign of MRKH syndrome is a primary amenorrhea in young women presenting otherwise with normal development of the secondary sexual characteristics and normal external genitalia, normal and functional ovaries, and karyotype 46, XX, without visible chromosomal anomaly. In our study, this case presented with primary amenorrhea with MRKH syndrome (Mullerian agenesis), a type of developmental anomaly.[1]

Based on the clinical features, biochemical findings and the ultrasound and MRI showing absence of uterus and cervix with nonvisualization of ovaries in the ovarian fossa, a final diagnosis of MRKH syndrome (Type I) was made in the this case.

Associated upper urinary tract malformations are found in about 40% of the cases with MRKH syndrome. Most commonly, they include unilateral renal agenesis (23–28%), ectopia of one or both kidneys (17%), renal hypoplasia (4%), horseshoe kidney, and hydronephrosis. Auditory defects or deafness are associated with 10–25% of the MURCS patients. Associated skeletal abnormalities of the spine are found in 30–40% of the cases. The association of MRKH with heart malformations is less common. Cases of polycystic ovaries and ovarian tumors have been described in women presenting otherwise with normal 46, XX karyotype.[3],[4],[5],[6],[7],[8],[9]

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