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MAYER-ROKITANSKY-KUSTER-HAUSER SYNDROME. A STUDY OF FIFTEEN CASES

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Although Mayer Rokitansky Kuster Hauser (MRKH) is a rare entity, it is a fairly common cause of primary amenorrhoea. The purpose of the study is to establish guideline for the investigation of a patient suspected to have a MRKH and to look for some congenital defects which may be associated with this syndrome. We reviewed fifteen cases of MRKH seen at the King Fahd Hospital of the University, Al-Khobar, and Al-Madina Maternity and Children's Hospital, Al-Madina, Saudi Arabia. The age of the patients ranged between 14 to 24 years old (mean 17.3 years old). Eight of the patients complained of delayed menstruation, five of whom were unable to conceive, and two complained of dyspareunia. All patients were investigated by cytogenetic studies, hormonal profile, intravenous pyelography, pelvic ultrasound and a diagnostic laparoscopy. Cytogenetic studies revealed that all fifteen patients were normal females of 46XX karyotype. Hormonal studies in nine patients were within normal range, but in the other six the hormonal values correlated well with polycystic ovary syndrome and this were confirmed by pelvic ultrasound. Bahrain Med Bull 1995;17:

Mayer Rokitansky Kuster (MRKH) Syndrome is a Mullerian abnormality, characterised by the absence of the vagina, associated with abnormal or absent uterus, and the presence of apparently normal tubes and ovaries[1]. Physical growth development and secondary sexual characters as well as the external genitalia are also normal. These patients usually present with primary amenorrhea. Although MRKH syndrome is a rare entity, it is a common cause of primary amenorrhea and indeed in some reports ranked second to gonadal dysgenesis as a cause of primary amenorrhea[2,3,4].

We describe fifteen patients with MRKH syndrome who attended King Faisal University Hospital, Al-Khobar, and Al Madina Maternity and Children Hospital, Al Madina, Saudi Arabia.

Our report describes our methods of investigation, diagnosis and the findings in these patients.

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METHODS

The study is based on the review of records of all of 15 patients seen at the Gynaecology Clinic of King Fahd University Hospital, Al-Khobar, and Al Madina Maternity and Children Hospital, Al Madina, Saudi Arabia diagnosed as having MRKH syndrome. The records were examined to find out the age of the patients, complaint, physical examination and radiologic and endoscopic findings.

All patients were investigated by cytogenic studies and hormonal profile which include FSH, LH, prolactin, oestrogen, progesterone and thyroid function tests. Intravenous pyelography, pelvic ultrasound and diagnostic laparoscopy were also done.

RESULTS

The age of the patients ranged from 14 to 24 years old (mean 17.3 years old). Eight patients complained of delayed menstruation, five of inability to conceive, and two of dyspareunia.

The average height and weight were that of normal women, and all had well-developed secondary sexual characters. Physical examination revealed no abnormality apart from two patients with who had coarctation of the aorta.

Cytogenetic studies revealed that all fifteen patients were normal females of 46XX karyotype with no discernible anomaly.

In nine patients the pituitary gonadotropins, thyroid hormones and gonadal steroids were within normal range. But in six patients the steroid hormones and FSH were normal with elevated LH and LH/FSH ratio of > 3:1 in serial measurements. The diagnosis of polycystic ovary syndromes in these six patients was confirmed by ultrasound examination.

Pelvic examination of all patients showed normal vulva and a short blind vagina with non-palpable uterus, except in two patients who had complete abscess of the vagina. The ultrasonic scan of the pelvis showed absence of uterus and upper vagina in thirteen patients, and the presence of bilateral normal looking ovaries in nine patients and polycystic ovaries in the other six patients. In two patients, pelvic ultrasonic scan showed a tiny structure which looked like a uterine rudiment with no cavity with absence of cervix and vagina.

Intravenous pyelography was normal in thirteen patients. Two patients had renal anomalies with one having ectopia of the left kidney while the other had mobile right kidney. The patient with ectopia complained of recurrent urinary tract infection, and culture of the midstream urine specimen showed E.Coli.

Laparoscopy confirmed the ultrasonic scan diagnosis of absence of uterus in all patients and the presence of slender rudimentary structures which looked like a uterine rudiment, which were seen high in the pelvis with no cervix or vagina in two patients only. Laparoscopy also confirmed the presence of normal looking ovaries in nine patients and polycystic ovaries in the other six patients. In two patients with absent vaginas, an artificial reconstruction of the vagina was carried out successfully by McIndoe and Read techniques.

DISCUSSION

MRKH syndrome is a genital tract anomaly which may be associated with further congenital defects involving urinary or skeletal system[4,5]. In its classical form, it presents as a rudimentary solid, bipartite uterus with solid non-canalized vagina. A shallow depression or vaginal dimple is noted at the lower end. The ovaries are usually normal. The disorder can be atypical with tiny Mullerian ridges or small uterine rudiments placed high in the pelvis, along with the absence of part or whole of the vagina.

The aetiology of MRKH syndrome is unknown. Griffin and colleagues in an extensive review postulated that the disorder may reflect a variable manifestation of a single genetic defect in some cases, while in others may result from a multifactorial cause rather than a single gene defect[5]. Tarry et al studied the pathogenesis of MRKH and traced the association of system defects to errors of formation of the Wolffian body and defined MRKH syndrome as a spectrum of Mullerian anomalies including vaginal agenesis, renal and ovarian defects in different embryological combinations[6]. In our fifteen cases, two

patients had rudimentary uterus while thirteen had no uterus. Vaginal agenesis was noted in two cases while thirteen had functioning vaginas and was leading a normal sexual life.

The diagnosis of MRKH syndrome was made on the basis of ultrasonography and confirmed by laparoscopy. Exploratory laparotomy is not indicated in these cases and under normal circumstances it is not necessary to remove the Mullerian rudiments. However, very rarely rudimentary uterus may contain functioning endometrium that may lead to haematometra and thus necessitate surgical intervention.

Two of our patients had renal anomalies with one having ectopia of the left kidney while the other had mobile right kidney. This observation was not unusual as a high incidence of urinary abnormalities have been reported in these cases[4,5,6]. Renal ectopy and agenesis of one or both kidneys are the most frequently reported renal anomalies[5,7]. This point out to the importance of renal assessment before any operative procedures in these patients as the presence of pelvic kidney may pose technical problems and great care needs to be taken in the surgical reconstruction of vagina. Also, structurally, abnormal kidneys are more prone to urinary tract infection[8]. Our patient with renal ectopy had recurrent urinary infections.

We noted a minor bony anomaly in the lumbar vertebra of one of our patients. Griffin and colleagues noted 12% incidence of skeletal anomalies in patients with MRKH syndrome with two-thirds having spinal abnormalities[5].

All our patients were phenotypically and genotypically females and this observation is in agreement with other reports[2,5]. In nine patients the pituitary gonadotrophin and steroid hormones levels were within normal range along with normal ovaries, while in the other six patients, the ultrasonic findings of polycystic ovaries along with high LH/FSH ratio (> 3:1) in serial measurements were diagnostic of polycystic ovary syndrome. Tzingounis et al noted an increased prevalence of major Mullerian anomalies in patients with polycystic ovaries while MacDougall et al believed the association to be a coincidental, as in their experience the incidence of Mullerian anomalies in relation to polycystic ovaries was similar to that in the general population[9,10].

CONCLUSION

In evaluating patients with suspected Mayer Rokitansky syndrome, the first investigation to be performed should be pelvic ultrasonic scanning because of its simplicity, and low cost. Laparoscopy should be delayed until all investigations have been completed to confirm the diagnosis. Magnetic resonance imaging can also be used but the procedure is expensive and should only be employed if sonographic findings are inconclusive.

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