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Original article

Radiology Section

Mayer Rokistanky Kuster Hauser Syndrome-The Psychological and Socio-cultural Implications In Nigerians

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ABSTRACT

Background: Most cases of reported Mayer-Rokinstanky-Kuster-Hauser Syndrome(MRKH) were from developed countries. The relevance of procreation in less developed nations, prevalent illiteracy and poverty made it necessary to examine MRKH in our context.

Material And Methods: Socio-demographic information was collected after informed consent from patients with the aid of questionnaires. The findings on clinical and radiological examinations were analysed and reviewed.

Results: Most of the patients presented rather late in our environment with the mean age of presentation being 31 years. Most of the patients were poorly educated with meagre income. Renal anomaly was the commonest anomaly and ectopic kidney was the commonest of all renal anomalies. The commonest skeletal anomaly was spinal bifida occulta.

There was a very high incidence of divorce/separation and depression amongst the patients after diagnosis despite counselling and follow-up.

Conclusion: MRKH diagnosis has a devastating psychological and socio-cultural effects on the patients. There is a need to further educate the populace, improve the economy and make health services including assisted reproductive facilities accessible and affordable.

Key Words: MRKH Renal Anomalies, Pschochological Sociocultural

INTRODUCTION

Mayer-Rokinstanly-Kuster-Hauser syndrome (MRKH) is a rare disorder characterized by congenital aplasia of the uterus and upper 2/3rd vaginal in women showing normal development of secondary sexual characteristics. Most of the reported cases were from advanced and developed continents. We therefore decided to review all the cases of MRKH seen by us with the background of our peculiar socio-cultural values, beliefs and prevalent poor education and poverty.

METHODOLOGY

The Radiologists prospectively review all the cases of MRKH syndrome seen by them within the past ten years. The socio-demographic data was collected by means of questionare; clinical examination was done and documented and radiological investigations results reviewed and analysed. The patients were managed, reviewed and follow-up by a team of specialists including radiologists, qynaecologist, urologist and psychiatrist.

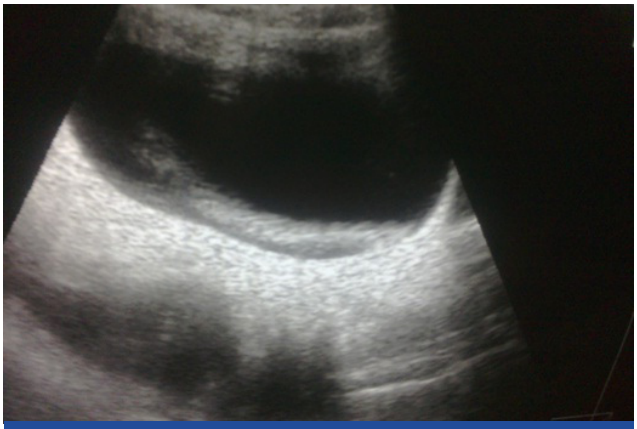
RESULTS

A total number of thirty patients were seen within a ten-year period but only twenty-two (22) patients were included in the study. Excluded from the study were patients who refused consent or failed to undergo basic investigations. The mean age was 31 years. Most of the patients presented within the age range of 30-35 years that is 63.6% (14) followed by 27% within the age range of 26-30years; one patients each (4.5%) presented before 20 years of age and after 30 years respectively.

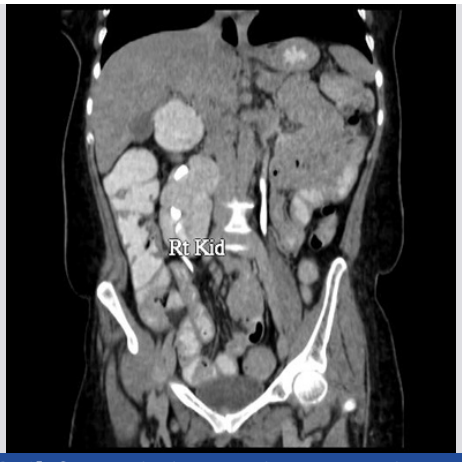
Fourteen of the patients (63.6%) were married as at the time of presentation. Out of this fourteen, only two (14.3%) were still married six months after the diagnosis was made. 8 patients (57.1%) were separated while 4 (28.6%) were divorced.

None of the eight patients (36%) that were single at the time of presentation got married through the follow-up period. Primary amenorrhea alone was the indication for pelvic ultrasound in six patients (27.3%) while the remaining 16 (72.7%) presented because of primary infertility and primary amenorrhea. Two (9.10%) were first degree relatives and there was no similar history/diagnosis in relatives of the other twenty patients. General physical examinations were essentially normal in all the patients, also external genitalia appeared normal but findings of small, narrow/shallow vagina was common to all the patients. Eleven patients (50%) were unemployed, 10 (45.5%) were employed and 1 (4.5%)) was a secondary school student. The average monthly and yearly family income was less than 62.5 US dollars and 750 US dollars respectively in 90% of the patients. The remaining 10% had monthly family income ranging between 100 US dollars -150 US dollars (that is, 1,200-1,800 US dollars yearly). Ten (10) of the patients (45.4%) were uneducated, 8 (36.4%) and 4 (18.2%) had secondary and post secondary school education respectively

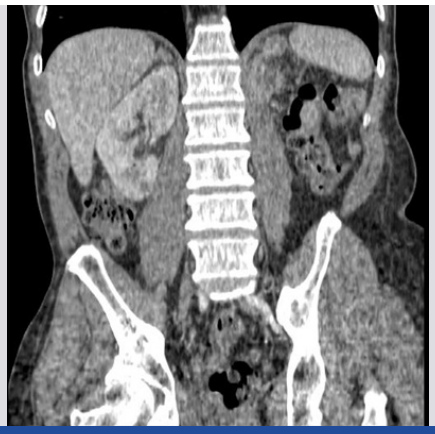
All of them had pelvic ultrasound done and a diagnosis of congenital absence of the uterus made [Table/Fig-1]. However, ovaries were visualized in 12 (54.5%) of the patients while ovaries were absent in 8 (45.5%). Renal anomaly was the commonly associated abnormality: 11 (50%) had normal right and left kidneys; six (27.3%) patients had ectopic kidneys-it was the solitary kidney in 3 patients, right sided ectopic pelvic kidney in two patients and left sided ectopic pelvic kidney in 1 patient [Table/Fig-2]; unilateral congenitally absent kidney was diagnosed in four patients (two on either side) [Table/Fig-3 & 4] and ptosis/wandering right kidney in one patient. Spinal bifida occulta was the only skeletal anomaly seen in the patients: this was seen in 7 patients (31.8%) and it was made up of lumbar in three patients (42.9%); sacral in three patients (42.9%) and both sacral and lumbar vertebra in one patients (14.2%).



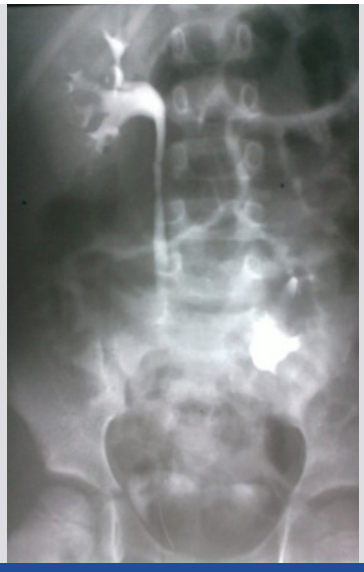
[Table/Fig-1]: Transabdominal pelvic ultrasound showing the urinary bladder but absent uterus



[Table/Fig-2]: Computerized tomography scan,coronal reconstruction showing right kidney,urinary bladder ,absent uterus and left kidney in another patient



[Table/Fig-3]: Computerized tomography scan,coronal reconstruction showing absent uterus and left kidney



[Table/Fig-4]: Intravenous urography showing left(ectopic) pelvic kidney and a normal right kidney

DISCUSSION

In Africa, like most other less developed continents, a woman status and acceptance are solely dependent on her motherhood. Infact, marriages are not successful if not immediately followed by procreation. MRKH is a syndrome characterized by aplastic uterus and upper part of vagina in women showing normal development and secondary sexual characteristics. The karyotype is often normal, 46XX [1, 2].

It is rare, seen in about 1 out of 4500 women [1, 2]. It is said to be mainly sporadic [3] in most cases but familiar cases have also been described [2, 4]. The mode of inheritance is said to be autosomal dominant with an incomplete degree of penetrance and variable expressivity. They are often associated with unilateral agenesis, ectopic of one or both kidneys and horseshoe kidney [1, 2, 5].

Also, skeletal abnormalities such as defective vertebral segments, joint malformation and spinal stiffness may be seen [6]. Auditory and cardiac abnormalties of variable severity are less common defects. In most of the reported studies, the patients presented between birth and early adolescence [5, 6] but in our study, more than 95% of the patients presented after 25 years of age. Infact the mean age of presentation in this study was 31 years.

Although, strubbe et al., [7] reported a mean age of 25 years in a study conducted in Netherlands. The late presentation is not unusual in our environment and may not be unconnected with the poverty level, all the patients had monthly family income of less than 100 US dollars per month; low level of education and ignorance obviously demonstrated in our result. Strubbe et al., [7], also suggested designation of the typical and atypical forms as type A and type B respectively and concluded that upper urinary tract anomalies are common in type B. Our findings also confirmed that familiar cases are not as common as sporadic cases as only two of our patients are first degree relatives while no similar history was elucidated in first degree relatives of other patients.

Renal anomaly was the commonest associated anomaly in our study. It was seen in 50% of the patient with ectopic kidney responsible for the highest renal anomaly. Other renal anomalies seen were unilateral absent kidney (18.2%) and Ptosis kidney (4.5%). Also, 7 patients (31.8%) had just one kidney; in 3 patients, the single kidney was ectopic pelvic;two patients had their solitary kidney in the Rt and Lt renal fossa respectively. Our finding was similar to previous reports of about 40%-60% of renal anomalies found in several patients.

Although,various associated skeletal anomalies had been reported by different authors [1, 5, 6], 31.8% of our patients had associated skeletal anomalies. Despite adequate counseling, all our patients except one rejected surgical option of vaginal dilation because ''it would not lead to procreation". Also, the counselling did not prevent their marriages from collapsing as 12 patients (85.72%) of the married patients became divorced or separated from their husbands after the diagnosis. Much more worrisome was the fact that more than 80% of the divorced or separated wives/patients were sent out by the in-laws since their ''son cannot be married to a man".

The fact that surrogate motherhood and other means of assisted conception are yet to be culturally acceptable in our environment and are also beyond the reach of the poor lead to the defects to attitude of the patients an their husbands. The psychiatrist eventually commenced treatment for depression for twelve (55%) of our patient within three months of the diagnosis.

CONCLUSION

The significance placed on motherhood in less developed countries, combined with prevalent illiteracy and poverty have made the diagnosis of MRKH a devastating one. There is need to improve education, re-orientate the populace on the other reasons for marriages and make assisted reproduction more accessible.

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