International Journal of Reproduction, Contraception, Obstetrics and Gynecology
Ndiade A et al. Int J Reprod Contracept Obstet Gynecol. 2022 Dec;11(12):3402-3404
www.ijrcog.org
pISSN 2320-1770 | eISSN 2320-1789

DOI: https://dx.doi.org/10.18203/2320-1770.ijrcog20223141

Case Report

Rare association of Turner syndrome and Mayer-Rokitansky-Kuster-Hauser syndrome

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Received: 27 September 2022
Revised: 29 October 2022
Accepted: 31 October 2022

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ABSTRACT

Turner syndrome and Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome is a rare association. The incidences of Turner and MRKH syndromes are estimated at 1/2000 and 1/4500 female births respectively. This is a 23-year-old patient, born of a consanguineous marriage, who was referred to us for exploration of primary amenorrhoea. The karyotype, performed three times, from peripheral blood lymphocytes was 45X0. The diagnosis of Turner syndrome associated with MRKH syndrome was retained.

Keywords: Turner syndrome, MRKH syndrome, Senegal

INTRODUCTION

Turner syndrome is a common chromosomal disorder resulting from the complete or partial absence of the second sex chromosome, with or without cell line mosaicism.[1] Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome is characterised by congenital aplasia of the uterus and upper 2/3 of the vagina, but with physiological development of secondary sex characteristics with a normal female karyotype.[2]

The incidences of Turner and MRKH syndromes are estimated at 1/2000 and 1/4500 female births respectively.[3] Few cases of association of these 2 syndromes have been reported.[4]

CASE REPORT

This is a 23-year-old patient, born of a second-degree consanguineous marriage, who was referred to us for exploration of primary amenorrhoea.

She had no particular pathological history. The examination revealed a good general condition with well-stained mucous membranes. The weight was 40 kg and the height 147 cm corresponding to a body mass index (BMI) of 18. The blood pressure was 12/08 and the respiratory rate was 18 cycles/min. On neurological examination, the patient was lucid and coherent with normal motor skills and tone.

Sensitivity and reflexes were present with good coordination of gestures. At the perineal level, there was an absence of pubic hair y and poorly developed external genitalia with infantile labia majora. The breast was flattened with breast development classified as stage 2 according to the TANNER classification (Figure 1a). The speculum examination was not performed, but the vaginal examination showed a shallow vagina and no cervix.

Osteoarticular examination showed prognathism and bowed legs (Figure 1b). The rest of the examination was unremarkable.

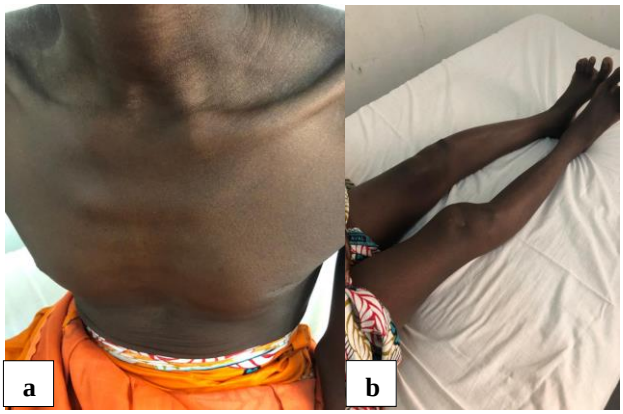

Figure 1: Picture of the patient (a) flattened chest, and (b) bow legs.

The biological tests showed normal kidney and liver functions

Ultrasound showed a hypoechoic fibrous band with no individualised uterus and well-differentiated kidneys. The ovaries were not visualised. Abdominal and pelvic computed tomography (CT) scan confirmed the ultrasound data (Figure 2).

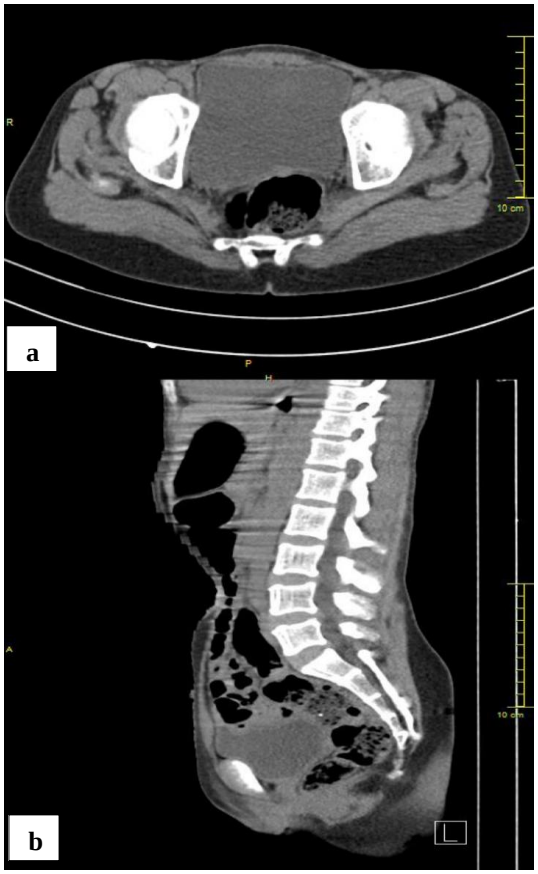

Figure 2: Abdominal and pelvic CT (a) sagittal reconstruction, and (b) axial section.

The karyotype, performed three times, from peripheral blood lymphocytes was 45X0 (Figure 3). The diagnosis of Turner syndrome associated with MRKH syndrome was retained.

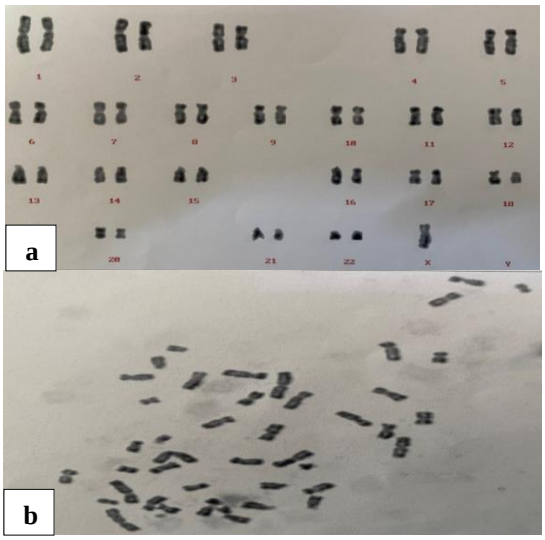

Figure 3: Karyotype (a) chromosome classification, (b) x1000 GIEMSA stain.

Hormone replacement therapy with estrogen and progesterone, vaginoplasty and breasts implants were offered to the patient. Due to lack of financial means the patients were not able to benefit those treatments.

DISCUSSION

Gonadal dysgenesis is the most common cause of primary amenorrhoea with absence of secondary sex characteristics.[5] Patients with Turner syndrome have a characteristic phenotype related to structural abnormalities of one of the sex chromosomes. Turner syndrome is related to the complete or partial absence of an X chromosome. Our patient had a complete absence of an X chromosome confirmed by karyotype. It is important to remember that people with Turner syndrome are diagnosed very early before puberty in developed countries. Usualy the reasons for consultation concern anomalies related to bone deformation, as an 11-year-old girl referred for orthodontic consultation, then the diagnosis was made early.[6] The reason for referral did not concern the patient's genito-reproductive life.

Our patient had dental anomalies and facial dysmorphia. Several authors have shown that these anomalies are due to chromosomal aberration which is a common feature with other patients with Turner syndrome.[7-9]

Our patient had the particularity of presenting an absence of the posterior 2/3 of her external genital organs. This absence corresponds to Rockitansky's disease. She had primary amenorrhea as a reason for consultation, which corresponds to the main reason for consultation of patients with MRKH syndrom. That was the case of a 19-year-old female patient who came to the clinic for primary amenorrhoea.[10]

The consaguineous marriage is considered as a risk factor for the occurence of these syndroms.[10,11] In our study, the patient was born in a second-degree consanguineous marriage.

The combination of Turner and MRKH syndromes was confirmed by radiological images and karyotype.

CONCLUSION

Turner syndrome and MRKH syndrome is a rare association, the diagnostic approach although simple can sometimes escape the specialists. Patients with this disease should always receive psychological support.

Funding: No funding sources
Conflict of interest: None declared
Ethical approval: Not required

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Cite this article as: Ndiade A, Thiam M, Dia SAM, Sy M. Rare association of Turner syndrome and Mayer-Rokitansky-Kuster-Hauser syndrome. Int J Reprod Contracept Obstet Gynecol 2022;11:3402-4.