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CO-OCCURRENCE OF TURNER (46,X-ring/45,X0 MOSAICISM) AND MAYER-ROKITANSKY-KUSTER-HAUSER SYNDROMES: A CASE REPORT

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INTRODUCTION

Mayer-Rokitansky-Kuster-Hauser Syndrome **(MRKHS)** has been historically defined as the finding of congenital aplasia of the uterus and the upper two-thirds of the vagina in an otherwise phenotypically normal woman with a 46, XX karyotype.
Nevertheless, a growing body of literature has shed light on the potential co-occurrence of MRKHS and different syndromic conditions[1,2,3,4,5].

CASE REPORT

Age and gender
14.3-year-old female patient

Family history
Unremarkable. No family history consistent with pubertal delay.

Previous medical history
- Born at term after uneventful pregnancy
- Subsequent onset of mild-to-severe intellectual disability
- Strabismus
- Recent-onset epilepsy requiring anticonvulsant therapy.

Reason for referral
Pubertal arrest.
Breast budding at the age of 11 years, with no subsequent progression of pubertal signs.

PHYSICAL EXAMINATION

Auxology
Severe short stature (height: -4.25 SDS, WHO growth chart).
BMI: 23.8 kg/m^2 (89°pc, WHO chart)

Tanner stage
B2, PH3, AH1.
Complete lack of oestrogenization.

General appearance
Shield chest, slightly dysmorphic features and onychopathy.

LAB TESTS

Findings consistent with **primary ovarian failure:**
- FSH 97.4 mUI/ml
- LH 16.5 mUI/ml
- Undetectable oestradiol.

RADIOLOGY

Pelvis ultrasound: streak ovaries, but - unexpectedly - no visible uterus was detected.
Pelvis MRI confirmed these findings.
No additional abnormalities involving the spine and the kidneys.

GENETICS

ArrayCGH detected two cellular lines, in the setting of a mosaic genotype: 46, XrX (X ring chromosome) and 45, X0, as confirmed by a subsequent **karyotype**.

Turner

GYNO

Severely hypoplastic vagina.

MRKHS

THERAPY

Progressively increasing doses of transdermal **oestradiol** were started to prompt pubertal development.

Despite a progression of secondary sexual features, the sequential sonographic assessment of internal genitalia did not show any signs consistent with oestrogenization
and no uterus was detected upon last evaluation, **12 months later**.

DISCUSSION

The co-occurrence of **Turner syndrome (TS)** and **MRKHS** has been rarely described in literature[3]. The resulting clinical picture includes congenital aplasia of the uterus and of the upper two-thirds of the vagina and ovarian dysgenesis[4,5].
Though the relationship between TS and MRKHS may be regarded as **coincidental**, the fact that the co-occurrence of the latter with several syndromic conditions[1,2] has been already described, may support the hypothesis of a **causative** association.

CONCLUSION

We aimed at increasing the awareness about a possible association between TS and MRKHS.

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