**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.

**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). BM1: 23.8 kg/m² (89% pc, WHO chart).

**Tanner stage**

**General appearance**
Shield chest, slightly dysmorphic features and onychopathy.

**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, X/X (X ring chromosome) and 45, X0, as confirmed by a subsequent karyotype.

**DISCUSSION**

The co-occurrence of Turner syndrome (TS) and MRKHS has been rarely described in literature. The resulting clinical picture includes congenital aplasia of the uterus and of the upper two-thirds of the vagina and ovarian dysgenesis.

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 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.

**REFERENCES**


**CONTACT INFORMATION**

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**LAB TESTS**

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

**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.

**MRKHS**

Severely hypoplastic vagina.

**GYN**

Coexistence of Turner syndrome (TS) and Mayer-Rokitansky-Küster-Hauser Syndrome: A Case Report

**REFERENCES**