ORIGINAL RESEARCH PAPER

Genetics

KEY WORDS:

AN ACCIDENTALLY DIAGNOSED CASE OF MAYER ROKITANSKI-KUSTER-HUSER SYNDROME

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ABSTRACT

A non-dysmorphic eighteen-year girl presented in outpatient with chief complaints of cold, headache and was having primary amenorrhoea. On detail examination and imaging she was found to have atrophic uterus; and was eventually diagnosed as Mayer-Rokitanski-Kuster-Huser syndrome type 1.

INTRODUCTION

An 18-year-old female has complaint of throat pain for a year with expectoration, headache and functional constipation without impaction. On extended history it was found that she has never had menarche and had primary amenorrhea. Approximately 32.5% of girls presented as primary amenorrhoea has Mayer-Rokitanski-Kuester-Huser Syndrome (MRKHS) (Anita G.S et al, 2015). The syndrome has two types viz. typical/isolated (type 1/or type A), Atypical (type 2/or type B) and associations (MURCS) (Herlin, M.K et al, 2020). Barffour, et al, 2021 reported that the GREB1L (Growth Regulation by Estrogen in Breast cancer 1-like gene) gene is associated with both sporadic as well as familial cases of MRKHS.

Case Study

An 18-year-old presented with throat pain and expectoration for a year, and un-impacted functional constipation. Patient has never had menstrual bleeding, though the secondary sexual characteristics were well-developed. In a pubertal girl with well-developed secondary sexual characteristics with primary amenorrhoea is highly suspected of MRKHS (Kiniya et al, 2023). Furthermore, patients of MRKHS are frequently associated with bowel symptoms and approximately quarter of them have constipation (Pennesi, C.M et al, 2021).

On initial vaginal and vulval examination; vaginal introitus and hymen was not appreciable and it was being wrongly diagnosed as case of imperforate hymen, however, after abdominal ultrasound imaging diagnosis of Mayer Rokitanski syndrome was confirmed due to presence of atrophied uterus, since the patient has no dysmorphism and was not associated with other malformations; hence type 1 was diagnosed.

On X Ray imaging of PNS showed right maxillary sinusitis, though no universal association of this condition was found in literature with sinusitis. Nevertheless, recurrent UTI was co-existed with the syndrome, in one such study type 2 MRKHS with renal anomaly, urinary incontinence leading to recurrent UTI (Torres de et, al, 2016).

She also has some of the components of primary cephalgia; tension type headache (some of ICHD-3 criteria), however, mostly it was secondary cephalgia due to sinusitis. There were no complaints of urinary system.

Piciu et, al, 2012 has mentioned that imaging in the form of MRI and ultrasound has the main role in the diagnosis of this syndrome. On further history it was being informed that she earlier had MRI abdomen and showed uterine agenesis, however, on USG abdomen atrophic uterus and normal ovaries were visualised at our centre. Herlin et, al, 2020 reported in his study about the syndrome that ovaries are usually normal, ovarian anomalies are present only in 5-10 % of cases and ovulation usually occurs (Morcel et, al, 2007).

On growth chart; height & weight were under third centile for age and sex whereas BMI was on 25th centile (CDC growth charts), Tanners SMR was A5B5P5. No universal association of short stature and faltering growth has been described as yet in these patients. Patient's development and learning were normal. Patient was treated for cephalgia, PEG for constipation (managed according to NICE/CHI guidelines) and medications for sinusitis and was subsequently referred to gynaecologist for therapeutic laparoscopy and surgical intervention.

DISCUSSION

This patient of MRKH syndrome type 1 had multiple diagnoses at the time of presentation viz. Right maxillary sinusitis, cephalgia (primary/secondary), and un-impacted functional constipation. She was incidentally diagnosed as MRKHS type 1 a rare entity.

No universal association of sinusitis, persistent cephalgia have been described in these patients as yet. The patient was treated symptomatically and was referred to gynaecologist for further management.

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Abbreviations: MURCS (Müllerian duct aplasia–renal agenesis–cervicothoracic somite dysplasia)
PEG: poly ethylene glycol

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