A Case of Mayer-Rokitansky-Küster-Hauser Syndrome with Coexisting Gonadal Dysgenesis

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Background

Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome is a congenital disorder characterized by agenesis or aplasia of the uterus and upper part of the vagina in a phenotypically and karyotypically normal female (46,XX). Patients present with primary amenorrhea and normal secondary sexual characteristics. Ovarian anomalies with MRKH are rare and only found in ~5–10% of cases (21 reported cases). These patients present with both primary amenorrhea and absent secondary sexual characteristics.

Case presentation

An 18-year-old girl, with delayed puberty and a history of repeated pathological fractures and bilateral cataract surgeries, presented with her first attack of hematemesis and melena and dyspnea at rest with oxygen saturation of 90% on room air.

On examination, the patient appeared pale, with no facial dysmorphism, thin and short with span>height and upper segment to lower segment ratio <1. She was tachycardic (HR 110 b/min, regular), normotensive (100/70), tachypneic (33-36) and afebrile. Cardiac examination showed accentuated heart sounds, S3 gallop and hemic murmur. Chest examination showed signs of bilateral pleural effusion. Deep palpation of the abdomen showed a shrunken liver and splenomegaly. Her pubic hair and breast development were in Tanner stage 1.

Routine laboratory works up showed an elevated ESR (60), macrocytic anemia (Hb: 4.4 g/dL, MCV: 106 fl, MCH: 29 pg), and platelets 136000, elevated INR (1.3), hypokalemia (K: 3.1 mmol/L), hypocalcemia (corrected Ca: 8.4 mg/dL), hypoalbuminemia (2.5 g/dL), slightly elevated liver enzymes (ALT 156 U/L, AST 79 U/L) and normal kidney function tests. The patient also had severe vitamin D deficiency (3.2 ng/mL), and a relatively poor response of the parathyroid gland (PTH 39 pg/mL). Hormonal evaluation showed hypergonadotropic hypogonadism (FSH 80 mIU/mL, LH 52 mIU/mL, E2 19 pg/mL), normal thyroid functions and normal prolactin level.

Abdominal and pelvic ultrasound showed liver cirrhosis, splenomegaly, and did not show the uterus or ovaries. The upper endoscopy showed esophageal varices and portal hypertensive gastropathy. Causes of liver cirrhosis were thoroughly investigated, and after exclusion of other causes the most likely cause was determined to be autoimmune hepatitis based on positive ANA, positive ASMA, and high level of serum IgG. For her unexplained dyspnea and hypoxia, an agitated saline study was done and showed evidence of hepato-pulmonary shunts. MRI pelvis showed a marked hypoplastic uterus as well as a picture suggestive of ovarian agenesis. FISH karyotyping showed a normal female karyotype of 46,XX. Thus, we confirmed the coexistence of two disorders, namely gonadal dysgenesis and MRKHS in this patient.

Management and outcomes

The patient received multiple blood transfusions, replacement for hypokalemia and hypocalcemia, vit D injection (200.000 units) and carvedilol 6.25mg twice daily. Anemia improved and hemoglobin level reached 11.3 g/dL; electrolytes, liver enzymes and vitamin D level normalized.

Discussion

MRKHS is a heterogeneous disorder characterized by uterovaginal atresia in a 46XX female. Gonadal dysgenesis is the most common cause of primary amenorrhea and absent secondary sexual characteristics. An association between both disorders has been reported but very rare. There has been no clear link between MRKHS and liver affection.

Conclusion

This case report demonstrates the rare coexisting gonadal dysgenesis in MRKH syndrome, where the patient not only presents with primary amenorrhea but with absent secondary sexual characteristics as well.

Keywords

Mayer-Rokitansky-Kuster-Hauser Syndrome, gonadal dysgenesis, hypogonadism, primary amenorrhea, liver cirrhosis