An atypical case of Mayer-Rokitansky-Kuster-Hauser syndrome with hyperandrogenemia

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Objectives:

Mayer-Rokitansky-Kuster-Hauser syndrome (MRKH) is characterized by utero-vaginal atresia in patients with a normal female phenotype and 46 XX karyotype. Various anomalies may accompany MRKH. The number of cases with accompanying hyperandrogenemia is limited.

Case :

A 17.5-year-old girl presented to our medical outpatient department with primary amenorrhea. Her Tanner staging was B5 P5. Physical exam revealed mild hirsutism without clitoromegaly. There was no scoliosis and her hearing was normal. Hormonal analysis revealed normal follicle-stimulating hormone(3.66 mIU/ml, normal range, 1.5–11.7),normal luteinizing hormone (9.23 mIU/ml, normal range 0.4-11.7), slightly elevated dehydroepiandrosterone sulfate (342 µg/dl, normal range 44-332), elevated 17 hydroxy progesterone (3.6 ng/ml,highest normal level: 2) and slightly elevated total testosterone level (0.85ng/ml, highest normal level: 0.6). Karyotypewas normal (46 XX). Magnetic resonance imaging(MRI) showeda hypoplastic vagina and uterine agenesis with multiple bilateral millimetric ovarian cystic lesions. Renal ultrasound and echocardiogram performed to investigate the presence of any accompanying renal or cardiac anomaly, respectively, were normal.

Discussion:

Mullerian agenesis is the second most common cause of primary amenorrhea after gonadal dysgenesis. The incidence of hyperandrogenism in patients with Mullerian anomaly is unknown. We encountered only seven cases presenting with varying degrees of clinical and biological hyperandrogenemia since the combination was first described in 2004 . The only genetic defect identified in the cases reported to date is WNT4 mutation , although there are also cases without this mutation and of unknown cause. Overexpression of two enzymes necessary for testosterone biosynthesis, 3β-hydroxysteroid dehydrogenase and CYP17A1, has been shown together with Mullerian agenesis inWNT 4 deficient mice , and this accounts for the hyperandrogenism accompanying the anomaly in them. We were unable to investigate WNT4 mutation in our patient.

Conclusions:

Androgen levels should be investigated in patients with Mullerian agenesis when even mild findings of clinical hyperandrogenemia are present.

References:

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