Premature Menarche Associated with McCune-Albright Syndrome in an Infant

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Introduction

McCune-Albright syndrome (MAS) is a rare syndrome, classically defined as the triad of gonadotropin-independent sexual precocious puberty, fibrous dysplasia of bone and cafe au lait lesions. We report a girl with MAS presenting with vaginal bleeding.

Case Report

A 2-year-old girl presented to our paediatric endocrinology clinic with vaginal bleeding for three days. She was born at 38 weeks of gestation with a birth weight of 2130 g and a body length of 45 cm. She was the second child of non-consanguineous parents with no remarkable family history. On physical examination, the weight was 9 kg [-2.7 standard deviation score (SDS)] and the height was 75 cm (-3.4 SDS). Cutaneous examination revealed irregular shaped café au lait skin spots. The pubertal stage was breast II, axillary and pubic hair I, according to Tanner score. The laboratory evaluation revealed a follicle stimulating hormone (FSH) level of 0.1 mIU/mL; luteinizing hormone (LH), 0.1 mIU/mL; estradiol (E2), 341 pg/mL; total testosterone, 2.5 ng/dL; dehydroepiandrosterone sulfate, 13.6 µg/dL; morning (08^{00}) cortisol, 10 µg/dL; prolactin, 8 ng/mL; free thyroxine (fT4), 1.1 ng/dL and thyroid stimulating hormone (TSH), 0.6 mIU/mL, gonadotropin-releasing hormone stimulating test revealed a peak LH and FSH levels of 0.1 mIU/mL and 0.2 mIU/mL, respectively. Pelvic ultrasound showed an enlarged uterus (31x26x43 mm) and ovaries (10x15 mm and 18x16 mm) with a follicle cyst (12 mm) in the left ovary. Bone scintigraphy demonstrated increased focal osteoblastic activity on the skull. Bone age was 3 years and 6 months at the chronological age of 2 years. Considering the clinical and laboratory findings, the patient was diagnosed with MAS. As an aromatase inhibitor, anastrazol (third-generation aromatase inhibitor, 0.06 mg/kg/day) was commenced. At 6-month follow-up visit, it was observed that ovarian cyst has regressed, serum E2 level were suppressed and no episode of vaginal bleeding was reported. After 9 months, the patient was admitted to our clinic with a complaint of palpitation, sweating and weight loss. The investigations revealed hyperthyroidism with a fT4 and TSH level of 2.1 ng/dL and 0.009 mIU/mL, respectively. The patient received methimazole therapy. The findings of hyperfunction of other endocrine glands such as acromegaly, hyperprolactinoma, Cushing syndrome or hyperparathyroidism were not observed in our patient.

Conclusion

MAS is associated with heterogeneous endocrine dysfunctions. Gonadal hyperfunction is the most common one and vaginal bleeding may be the initial clinical sign of this syndrome. In addition, the presence of accompanying disorders that may manifest during follow-up should be kept in mind.