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P31. Rare and atypical cases of primary amenorrhea

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Context. Usual cases of primary amenorrhea (PA) are constitutional delay of puberty, gonadal dysgenesis, hypogonadotropic hypogonadism, hyperprolactinemia, Müllerian abnormalities.
Objective: to present rare and atypical causes of PA, i.e. an adolescent with complete androgen insensitivity and 3 patients with suspected abnormalities of gonadotropin receptors, evaluated in C.I. Parhon Institute of Endocrinology.
Patient 1. PA at age 15 y, 176 cm height, normal sexualisation and external genitalia, a 4 cm measured vagina, sparse axillary and reduced pubic hair. MRI- intra-abdominal small gonads but no uterus. High serum testosterone, LH and AMH, normal estradiol and adrenal hormones and 46 XY caryotype confirmed a complete androgen insensitivity syndrome; the patient had bilateral gonadectomy and thereafter transdermal estrogen.
Patient 2. PA treated from age 23 to 30 y with estro-progestins (EP), clinically normal at presentation. High FSH and LH, low estradiol, normal androgens, a 9/5 mm pituitary mass (suspected gonadotroph tumor). Monthly GnRH analog for 5 months decreased FSH LH and the pituitary mass to half. During suppression urofollitropin 150 IU/day was administered for 18 days and bhCG 10,000 IU induced ovulation (twice) but no pregnancy occurred. The pituitary mass was 8/4 mm 1 year after withdrawal of triptorelin. The patient withdrew EP 4 years later, had regular spontaneous menses and a pregnancy.
Patient 3. PA at age 21 y, with small uterus, ovaries, vagina and labia minor, normal breasts. High FSH 34.2 U/L, LH 32.3 U/L, positive sexual chromatin, a 5 mm pituitary mass; 2 years of EP, then menses occurred only with progestins, albeit normal FSH, LH, prolactin, androgens; no ovulation with Clomiphene up to 100 mg/day ± Metformin 1500 mg/day. Stable microincidentaloma after 11 years.
Patient 4. PA at 16 y, thelarche and pubarche from 15 y, Tanner stage 4. High FSH, low estradiol, but responsive to Triptorelin 100 µg s.c.(FSH from 25 to 45 U/L, LH from 6.4 to 33.9 U/L, E2 from 37 to 105 pg/mL). Positive sexual chromatin, open growth cartilages, other pituitary and adrenal hormones were normal. She was started on oestradiol. Genetic testing for suspected gonadotropin receptor abnormalities could not be done in patients 2,3,4.
Conclusion. A thorough clinical, hormonal, genetic and imaging evaluation of patients with PA is needed in order to identify and correctly treat the rare causes.

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