POSTER PRESENTATIONS

REPRODUCTIVE HEALTH

PP-RH-01

45, X/47, XY, +13 MOSAICISM IN A 15-YEAR-OLD GIRL

https://doi.org/10.15605/jafes.037.AFES.109

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BACKGROUND

Approximately 45% of postnatal Turner syndrome patients have a pure 45,X cell line. Other karyotypes that may be mosaic with 45,X most commonly include:46,XX,; 47,XXX; or 46,XY;The presence of a 46,XY cell line may occur in 5–10%. Only a few instances of mosaicism with X-monosomic and trisomic cell lines of 21, 18, 13 were described.

CASE

We report the case of a 15-year-old female consulting for pubertal delay. Physical examination revealed the following signs of Turner syndrome: short stature, cubitus valgus lack of secondary sexual signs, brachymetacarpia and cutaneous nevi while no features of trisomy 13 were present. She showed female type of external genitalia. LH and FSH were elevated (100 and 21.5 mUI/l), serum estradiol: <10 pg/ml, serum testosterone: 0.1 ng/ml. Chromosomal analysis revealed a complicated karyotype: 45,X/47,XY,+ 13 mosaicism. The culture of skin fibroblasts, however, showed only 45,X cells. The coelioscopy showed a hypoplastic uterus and the histological examination of the castrated gonads showed ovarian agenesis. Our patient had no complication of Turner syndrome nor of 13 trisomy except for the agenesis of her corpus callosum.

CONCLUSION

Despite her pathological karyotype, this patient had a Turner syndrome phenotype with no complication, no evidence of any androgen effect and no detected malformations. The phenotype in a 45,X/46,XY mosaic patient likely depends on the distribution of mosaicism percentage in different tissues. We suggest that additional cells should be analyzed and more molecular genetic studies should be conducted.

PP-RH-02

CLINICAL AND HORMONAL CHARACTERISTICS OF PATIENTS WITH POLYCYSTIC OVARY SYNDROME IN A PERUVIAN TERTIARY CENTER

https://doi.org/10.15605/jafes.037.AFES.110

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OBJECTIVES

To describe the clinical and hormonal characteristics of patients with PCOS in a Peruvian center.

METHODOLOGY

Descriptive study that evaluated the clinical, biochemical and ultrasound characteristics of patients with PCOS at the Hospital Edgardo Rebagliati Martins, Lima-Peru in 2019.

RESULTS

We evaluated 100 consecutive patients, age range between 14 to 43 years (x ± SD: 27.2±7.3). Menarche at 12.2 ± 1.5 years. Oligomenorrhea that began during adolescence was present in 84%. The most frequent reason for consultation was oligomenorrhea, obesity and infertility (primary in 11 and secondary in 4 women), 21% had a history of abortions. Hirsutism (modified Ferriman-Gallwey Score >8), nodular-cystic acne, and androgynous alopecia were present in 89%, 44%, and 21%, respectively. LH and FSH concentrations (early follicular phase) were 8.9 ± 5.7 and 4.4 ± 2.4 IU/L, respectively; LH/FSH ratio >2 in 61%. The mean concentration of free testosterone: 2.8 ± 1.2 (0.6 to 6.5 pg/mL), 47% had values above the upper limit of our laboratory (>2.6 pg/mL). Androstenedione was from 0.20 to 6.60 (mean value of 3.0 ± 1.1 ng/mL), 45% had values above the upper normal value of our laboratory (>2.7 ng/mL). On ultrasonography, 78% presented with the morphology of polycystic ovaries (follicular cysts >12, 2 – 9 mm in diameter, ovarian volume ≥10 mL); endometrial thickness >5 mm in 33%.

CONCLUSION

The most frequent reason for consultation was ovarian dysfunction, and hirsutism was the most prevalent clinical alteration. Hyperandrogenemia was present in 50% and polycystic morphology in 75%.