TRIPLE X AND PREMATURE OVARIAN INSUFFICIENCY CASE REPORT

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Introduction

Premature Ovarian Insufficiency (POI) is characterized by precocious depletion of ovarian follicles (before age 40 years old), in association with amenorrhea, hypoestrogenism and high gonadotrophin levels.
It affects 1% of the female population and in 5% of cases it is caused by anomalies in the X chromosome.

Case Report

Female gender, 22 year old. Previously healthy.

Menarche at age 11. Normal pubertal development.
No history of previous pregnancy.
No family history of amenorrhea.

Referred to the Endocrinology Clinic due to transitory amenorrhea (6 months duration) that had already spontaneously resolved.

At the first visit she was menstruating, although irregularly, and she denied any other complaints. She expressed the will of future pregnancy.
Clinical examination was normal.

The following studies and complementary exams were performed

Laboratory analysis

FSH 67.6 mUI/ml, LH 35.9 mUI/ml, E2 13.5 pg/ml.
TSH, Total testosterone, D4-AE, DHEA-SO4, 17-HO-Progesterone and PRL within reference ranges. β-HCG <1 U/L.

Pelvic US (transvaginal approach)

Uterus of normal size with regular endometrium.
The right ovary had appropriate dimensions and 3 microfollicles.
The left ovary had 15x7 mm with no follicular activity.

On a second evaluation, hypergonadotrophic hypogonadism was confirmed: FSH 86.0mUI/ml, LH 73.8mUI/ml, E2 12.9pg/ml.

At this time she complained of progressively worsening hot flushes, and substitutive therapy was started with amelioration of symptoms.

Bone densitometry

Dual-energy x-ray absorptiometry: Osteopneia.
T score of the lumbar spine -1,3 and T score of the fémur neck -2,1.

Anti-Mullerian Hormone

Not detectable.

Anti-Ovarian antibodies

Negative.

Genetic studies

Study for the identification of mutations at the CGG unstable repetition sequence of the gene FMR1: not sugestive of Fragile X.

Kariotype

Mosaicism 46, XX (10%)/ 47, XXX (90%).

The patient was reffered to a fertility clinic with the intent of oocyte retrieval and preservation, althought there were none viable.
She entered an oocyte donation program.

Conclusion

Females with Triple X abnormalities do not show any manifestations other than irregular menses.
In this context, POI is associated with rapid depletion of ovarian follicles.
Genetic testing is part of the etiological study and unexpected kariotype findings have an important impact in fertility.

Bibliography

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