

Fertility preservation in a young woman with Turner-like phenotype due to chromosomal aberration

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Objective

Reporting a clinical case of fertility preservation in a young woman affected by an abnormal karyotype and a Turner like phenotype.

Methods

Our Department started a fertility preservation project for an early interception of clinical conditions at risk of Premature Ovarian Insufficiency (POI).

Turner's syndrome is one of these conditions, usually with no chance of future pregnancy except for some forms which allows residual ovarian function after menarche and, if soon intercepted, may allow oocyte preservation through controlled ovarian stimulation.

Patient

A 16-yo (karyotype 46XXqter?Xp22.3) came to our observation. Her menarche appeared at 14 years with irregular menses (20-35 days). The ovarian reserve study showed hormonal values of AMH 0.39 ?g/L and FSH 7.8 UI/L and an ultrasonographic follicles count of 2+1. A counselling on intervention method of fertility preservation and on possible future preimplantation diagnosis was performed.

Intervention

Ovarian stimulation, with ?-follitropin at a dose of 300UI/die (total 2700UI), was started on 3rd day of a spontaneous menstrual cycle with a subsequent use of GnRH antagonist. The ovulation induction occurred with gonadotropin chorionic at a single dose administration of 10000UI.

Main outcome measures

The pick-up of stimulated oocyte was performed, obtaining 3 oocytes. Only 2 oocytes were in Metaphase II and were cryopreserved.

Results

Fertility preservation even in patient with a very dramatic ovarian reserve seems feasible.

Conclusions

This case shows how, in patients with chromosomal pathologies, counselling and evaluation of the ovarian reserve are fundamental. Fertility preservation strategies at pre-existing POI are very important to offer patients with genetic risk the ability to have their own biological progeny.