第69回人類遺伝学会

P1-08-04

北海道、2024.10.9.-10.12.

Title:

A novel approach to fertility preservation in females with Turner syndrome

Yoshiko Asai, Yoshie Nagatakidani, Tomoko Inoue, Yoshiharu Morimoto HORAC Grand Front Osaka Clinic

Abstract

Premature ovarian failure is inevitable in Turner syndrome (TS), leading to infertility due to depleted ovarian reserves at young ages. We hypothesized that fertility preservation strategies could beneficial for TS patients. Recently, five TS women (ages 22 to 36) sought fertility preservation at our hospital. Four underwent oocyte retrieval, with two successful oocyte cryopreservation, while the other two showed no follicle growth.

Case 1: A 30yrs old, TS mosaicism (45, X/46, XX) and irregular menstrual cycles since menarche. Her height, 156 cm and weight, 51 kg. Anti-Mullerian hormone (AMH) was 0.03 ng/ml. Over three cycles in eight months, two mature oocytes were cryopreserved.

Case 2: A 27yrs old, 47, XXX/45, X TS mosaicism. She received growth hormone treatment and spontaneously entered puberty with regular menstruation at age 13. Her height, 150 cm and weight, 50 kg, and her AMH was 1.79 ng/ml. Following two ovarian stimulations, 10 mature occytes were cryopreserved.

We suggest that oocyte cryopreservation soon after puberty could be a viable fertility preservation option for girls with TS. Despite regular menstrual cycles, TS patients may exhibit diminished ovarian reserves and may not respond effectively to fertility preservation treatments in adulthood. Oocyte retrieval can also be successful in mosaic TS cases.

It is important to advise parents to consider early fertility preservation options for girls diagnosed with TS. Additionally, informed consent is essential due to the risks of oocyte chromosomal abnormalities and perinatal cardiovascular disease.