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Fertility Preservation in Turner Syndrome Females

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ABSTRACT

This article examines the existing fertility preservation options for females with Turner syndrome and provides practitioners with practical guidelines. Turner syndrome is the most common sex chromosome abnormality in females, affecting one in every 2500 live births. Women with Turner syndrome are at an extremely high risk of developing POI and infertility. Although 70-80% of women have no spontaneous pubertal development and 90% have primary amenorrhea, the remainder may have a small number of ovarian follicles at birth or in early childhood. The current challenge is to identify these women as early in life as possible so that they can take advantage of the existing fertility preservation options.

Keywords: Fertility preservation; Turner syndrome; Embryo cryopreservation; Adoption

INTRODUCTION

Sixty-five years have passed since Henry Turner described the association of a cell line with the phenotypic findings he described 21 years earlier. Turner syndrome is one of the most common chromosomal disorders, affecting about one out of every 2500 newborn female infants. A female infant is born with approximately two million oocytes within her ovaries under normal circumstances, but this is not the case in females with TS, who are born with significantly fewer3. How do oocytes develop prior to birth during embryonic life, and how is this process altered in the absence of two intact X chromosomes? The progenitors of a female's oocytes emerge outside the embryonic tissue that will become the ovary, migrate along the dorsal mesentery from the region of the junction of the ovary and the uterus, and then mature.

There is a lot of variation in the size of this pool of primordial follicles between people. This could be due to variation between females during the process that leads to follicular atresia, but it could also be due to the presence of sex chromosome aneuploidy at the ovum level, which would not be visible in a leukocyte karyotype. If the ovary contains two populations of oogonia, one with two X chromosomes and one without an X chromosome, i.e. a mosaic karyotype, the number of oocytes at birth will be significantly higher than in ovaries from a single line of 45,X. The pool of primordial follicles in mosaics may then be large enough for a young woman to go through menstruation and normal pubertal development [1].

DESCRIPTION

Spontaneous conceptions are uncommon in women with TS, and infertility due to insufficient or exhausted ovarian reserve is the norm. The reproductive phenotype of TS varies depending on the genotype. Ovarian dysgenesis is common in the context of a full,X genotype, and the use of donated oocytes or embryos can increase the chances of a successful conception. In contrast, those with the mosaic genotype can retain varying degrees of ovarian function for varying time intervals, though POI is unfortunately unavoidable. Salvage of existing viable oocytes through the use of assisted reproductive technologies is possible in this latter subgroup of young women with TS. We will go over the currently available and experimental methods for preserving fertility in females diagnosed.

While ovarian primordial follicle reserve may be depleted well before puberty in nearly all TS girls with non-mosaic karyotype, ovarian reserve may persist for a variable period post menarche23 in mosaic cases and depending on the degree

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of mosaicism. However, because the reserve would be low and depletion would occur at an accelerated rate, the majority of women may not reach adulthood with sufficient ovarian reserve. It is therefore critical to preserve oocytes in these girls as soon as possible after menarche [2].

The first human birth attributed to the use of cryopreserved and thawed oocytes was reported. For nearly three decades, vitrification freezing protocols were considered "experimental," but refinements in the last decade have significantly advanced the field. Oocyte cryopreservation is now considered a standard procedure in adults, and it is increasingly being offered to and used by reproductive-age women prior to the start of gonadotoxic therapy for both cancerous and non-cancerous conditions. [3,4]

However, despite existing technology and the potential for applicability and utilisation of oocyte freezing early in reproductive life as a fertility preserving strategy for TS patients, possible oocyte aneuploidy and the risk to the progeny cannot be overlooked. There is also a lack of experience with quantitative ovarian response to exogenous gonadotropins, as well as data on qualitative aspects of oocytes retrieved in this population. The financial and emotional costs of potentially freezing genetically abnormal oocytes harvested from the ovaries of women with TS cannot be underestimated, and until the safety of autologous oocytes is established.

Maternal oocyte donation and cryopreservation for later use by a TS daughter is a viable option for girls with POI32. However, because the egg donor would be the biological parent of her grandchild, this approach raises ethical concerns. Sibling-to-sibling oocyte donation is a less risky and more acceptable option. When considering a mother-child or sibling-sibling egg donation approach for preserving procreative potential, a careful psychosocial evaluation of all participants and family dynamics is required [5].

CONCLUSION

Adoption is a viable option for women with TS who want to be parents, and it is one that many women with TS have pursued. Multiple factors, such as the low rates of spontaneous pregnancy; potential health risks of pregnancy to the mother and genetic outcomes of the foetus; and highly personal desires, such as the desire to carry a pregnancy; and or the desire for a biological child; and future parenting goals, can result in complex decisions. Such decisions should be made with the help of a personal physician, a reproductive endocrinologist, a partner, and family members.

To summarize, women with TS are extremely vulnerable to POI and infertility. There are existing techniques, some of which are still in the experimental stage that can preserve fertility potential for some girls with TS if timely consideration is given. To maximize the benefits of fertility preservation, all girls with TS should be evaluated by an expert as early in childhood as possible, because the vast majority will have their ovarian reserve depleted before adulthood. For those who have already lost their ovarian reserve, donor oocytes, donor embryos, and adoption are options for fulfilling their desire to be a parent. For those with TS-related cardiac contraindications to pregnancy, the use of GS may allow for biological parenting using their own oocytes; alternatively.

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CONFLICT OF INTEREST

There are no conflicts of interest by author.

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