International Journal of Innovative Technologies in Social Science

e-ISSN: 2544-9435

Scholarly Publisher
RS Global Sp. z O.O.
ISNI: 0000 0004 8495 2390

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ARTICLE TITLE	FERTILITY AND PARENTING OPTIONS IN TURNER SYNDROME - A REVIEW OF CURRENT POTENTIAL AND LIMITATIONS
ARTICLE INFO	Aleksandra Dzwonkowska, Paulina Redel. (2025) Fertility and Parenting Options in Turner Syndrome - A Review of Current Potential and Limitations. *International Journal of Innovative Technologies in Social Science*. 3(47). doi: 10.31435/ijitss.3(47).2025.3505
DOI	https://doi.org/10.31435/ijitss.3(47).2025.3505
RECEIVED	20 June 2025
ACCEPTED	27 July 2025
PUBLISHED	31 July 2025
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FERTILITY AND PARENTING OPTIONS IN TURNER SYNDROME - A REVIEW OF CURRENT POTENTIAL AND LIMITATIONS

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ABSTRACT

Turner syndrome (TS) is a chromosomal disorder with a prevalence of 1 in 2,500 live births. The most common karyotype is 45.X, however, mosaic karyotypes are also present and are associated with a milder presentation of the syndrome. The main symptoms faced by patients with TS include growth deficiency, cardiovascular disorders, and reproductive disorders, including premature ovarian failure (POI). Fertility disorders are mainly due to the presence of dysgenetic gonads, which negatively affect the development of secondary sexual characteristics. Changes in the ovarian cells, including stunted follicle development, abnormal morphology, and follicle atresia, are also a significant problem. Measurements of FSH, LH, and AMH, which appear to be the most stable parameters of ovarian reserve, play an important role in the diagnosis of POI. Due to the fertility disorders in TS, spontaneous pregnancies are achieved in less than 6% of women. Among the fertility preservation methods in patients with preserved ovarian reserve are oocyte cryopreservation or cryopreservation of ovarian tissue, while oocyte donation is the method of choice when ovarian reserve is depleted. For women desiring offspring with contraindications to pregnancy, surrogacy or adoption are alternatives. Pregnancy in TS is associated with many risks for both the mother and the fetus. Patients should be managed by a specialised team experienced in the management of women with TS, including cardiac assessment, both preconceptional and postconceptional. This review discusses the available parenting methods for patients with TS, taking into account recent literature.

KEYWORDS

Turner Syndrome, Fertility, Cryopreservation, Oocyte Donation

CITATION

Aleksandra Dzwonkowska, Paulina Redel. (2025) Fertility and Parenting Options in Turner Syndrome - A Review of Current Potential and Limitations. *International Journal of Innovative Technologies in Social Science*. 3(47). doi: 10.31435/ijitss.3(47).2025.3505

Introduction.

Turner syndrome (TS) is a genetic disorder characterised by the complete or partial absence of the second X chromosome. The syndrome is named after Henry H. Turner, who described the syndrome in 1936 [1]. The prevalence of the syndrome in newborns has been estimated at 64 per 100 000 [2]. The underestimation of the prevalence may be due to the occurrence of mosaic TS, which is characterised by a milder phenotypic change, standard life expectancy, and absence of cardiovascular problems and has a prevalence of 76/100 000 [2,3]. Epidemiological studies have observed an increase in the diagnosis of TS, with a decrease in the proportion of women with karyotype 46, X among those with a diagnosis, while the proportion of other karyotypes has increased [4]. Diagnosis is most commonly made at 3 periods of life. The first is the prenatal and early postnatal period, during which a chorionic villus biopsy or amniocentesis is performed in the event of an abnormal ultrasound picture [2]. Features indicating the possible presence of TS on ultrasound imaging

include hygroma colli, fetal hydrops, cardiac defects (most commonly aortic coarctation), and increased nuchal translucency [5]. The second period in which the diagnosis is often made is between 5 and 20 years of age. At this time, growth deficiency, delayed puberty, or menstrual abnormalities may indicate the need to extend the diagnosis to Turner syndrome [2]. The third period is between the ages of 30 and 40, during which fertility disorders may indicate the diagnosis [2].

The most common karyotype in people with TS is karyotype 45,X, occurring in 40-50% of women with TS. This is followed by mosaicism with 46,XX (45,X/46,XX) occurring with a frequency of 15-25%, isochromosome Xq Isodicentric Xp (46,X,i(Xq); 46,X,idic(Xp)) with a frequency of 15% and mosaicism with 46,XY(45,X/46,XY) occurring in 10-12% of patients. Less common karyotype abnormalities include mosaicism with 47,XXX, ring X chromosome or proximal deletion of Xp [6]. The following should be excluded from the diagnosis of TS: those with deletion distal to Xq24 frequently and classified as having premature ovarian insufficiency (POI), symptomatic women over 50 years of age with less than 5% 45,X cells because the loss of one of the chromosomes may be due to age, and those with mosaicism 45,X/46,XY without typical female genitalia (however, for these individuals it is recommended to follow the guidelines for TS due to the similar profile of comorbidities) [6]. Phenotypic differences may vary depending on the individual's karyotype. For reproduction, women with mosaicism 45,X/46,XX undergo menarche and menopause at an average age and have no increased risk of pregnancy loss. In contrast, women with 47.XXX experience menopause five years earlier than those with a normal karyotype [3]. A study on a Swedish female population showed that 12% of women with TS became pregnant, of which 40% of pregnancies were spontaneous. In addition, a higher proportion of pregnancies occur in those with mosaicism versus monosomy, but pregnancy is also possible in these individuals [7].

Methodology

A thorough literature review was performed to analyse the current state of knowledge concerning fertility preservation and reproductive options in individuals diagnosed with TS. The search was conducted using the PubMed database, applying the keywords: “Turner Syndrome,” “Fertility,” “Cryopreservation,” and “Oocyte Donation.”.

Discussion

Phenotype in TS

The spectrum of symptoms in TS is wide, and the disease affects both the physical and psychological spheres. Among the abnormalities presented by people with TS are [8,9]:

- Short stature;
- Head and neck abnormalities (epicanthus, nearsightedness, strabismus, ptosis, infections of the middle ear, abnormal hearing and auricular structure, micrognathia, high arched palate, low posterior hairline, broad and short neck, pterygium colli, excess loose skin in the back of the neck of newborns);
- Broad chest, widely spaced nipples, inverted nipples;
- Hair, skin, and nail disorders (increased skin ridge count, lymphedema of hands and feet at birth, multiple pigmented nevi, nail hypoplasia, vitiligo, alopecia);
- Skeletal disorders (delayed bone age, valgus elbows and knees, reduced bone mineralisation, scoliosis, Magdelung's deformity, congenital hip dislocation);
- Endocrine disorders (glucose tolerance disorders and type I and II diabetes, hypothyroidism, hypertension);
- Gastroenterological disorders (elevated hepatic enzymes, celiac disease, and inflammatory bowel disease);
- Cardiovascular disorders (bicuspid aortic valves, coarctation of the aorta, and aortic dilation/aneurysm);
- Nephrological disorders (horseshoe kidney, renal pelvis, ureter, and vascular disorders, renal aplasia);
- Reproductive disorders (pubertal disorders, infertility, oestrogen deficiency);
- Psychosocial disorders (learning problems, emotional immaturity);

Fertility assessment

Hypergonadotropic hypogonadism, which results from gonadal dysgenesis leading to poor development of secondary sexual characteristics (breast and uterine development), is mentioned as an important component of the syndrome [10]. In individuals with TS, luteinising hormone (LH) and folliculotropic hormone (FSH) values are normal during childhood and elevate at puberty or when ovarian function is extinguished. An additional parameter useful in the diagnosis of ovarian failure is antimüllerian hormone (AMH) [6]. Small antral follicles produce it and appear to be a relatively stable marker of ovarian function in girls with TS compared to FSH and E2 [11,12]. In women, it indicates the pool of follicles in the ovaries so that reproductive lifespan can be estimated [6,13]. In a study on 120 female TS patients, it was observed that an AMH value < 4 pmol/l predicted failure to enter spontaneous puberty, whereas in adolescent girls, AMH values < 5 pmol/l were indicative of impending POI [14]. AMH values are also influenced by the karyotype, with the result that individuals with a mosaic karyotype tend to have higher values of AMH and thus also have a greater chance of entering puberty and maintaining ovarian function during young adulthood [11]. It is also important to keep in mind the high biological and inter-test variability of AMH [16]. Fertility problems in people with TS are mainly due to premature extinction of ovarian function. POI is diagnosed in individuals who experience amenorrhoea before the age of 40 years with high FSH and low oestradiol (E2) levels, and Turner syndrome is its most common genetic cause [17]. Spontaneous puberty occurs in only about one-third of patients and most often begins with pubarche, while menarche is observed in one in five [6,18]. This percentage increases up to twofold in the case of a mosaic karyotype, which may suggest that the presence of a second X chromosome affects the onset of spontaneous puberty [6,19]. Regular menstruation only occurs in up to 9 % of individuals with Turner syndrome [19].

In TS, it is recommended that luteinising hormone (LH), folliculotropic hormone (FSH), and antimüllerian hormone (AMH) levels be tested annually from 8 to 11-12 years of age to refer the patient for fertility preservation treatment if they increase [6].

According to international guidelines, in the case of persistently elevated FSH levels, it is recommended to start estrogen replacement therapy from very low doses at 11-12 years of age to reflect the physiological changes that occur during puberty. Starting therapy at low doses prevents the skeleton from maturing too quickly, and higher doses can be used and increased at a faster rate in those starting therapy after growth is complete. After the first vaginal bleeding has occurred, it is recommended to include cyclic progesterone, which protects against excessive endometrial hyperplasia [6].

The number of ovarian follicles also plays an important role in the reproduction of people with TS. In this syndrome, accelerated apoptosis of these cells starts as early as 18 weeks of gestation, which ultimately leads to fewer reproductive cells at birth [20]. The following are considered possible causes of germ cell loss in people with TS: abnormal chromosome pairing during meiosis, which hinders crossing-over between chromosomes and chromosome pairing leading to arrested meiosis, abnormal interactions between oocytes and granulosa cells and a reduced number of genes on the X chromosome, with genes on Xp playing a very important role in ovarian function. Among these genes are the BMP15 gene involved in folliculogenesis and PGRMC1 involved in the anti-apoptotic effect of progesterone on granulosa cells [21]. A study by Hreinsson et.al involving 9 pubertal girls with TS analysed the number and density of follicles in ovarian tissue. In ovarian biopsy tissue, follicles were observed in 8 of 9 patients, with the youngest subjects and those with mosaic karyotype having the highest number of follicles. In addition, a correlation between FSH and the number of follicles was shown, where those with the lowest FSH levels had the highest follicle density. The presence of follicles in the ovaries of adolescent girls suggests the possibility of fertility preservation methods in people with TS [22]. Examination of ovarian tissue in fetuses with TS showed the presence of oogonia in some ovaries in the absence of primordial, preantral, and antral follicles, indicating inhibition of follicular development [23]. TS also affects the quality of follicles, as shown in a retrospective study on a group of 15 individuals with TS. A high proportion of follicles with abnormal morphology (oocyte vacuolisation, lack of connection to the basement membrane and stroma), follicular atresia and abnormal expression of zona pellucida proteins were observed, and follicular fluid from small antral follicles had lower concentrations of oestrogen and testosterone (which may affect follicle development) and higher AMH levels. In a patient with a mosaic TS karyotype, in vitro oocyte maturation was performed, obtaining a maturation rate of 16%, which may indicate fertility potential [24].

Pregnancy risks

Another aspect that plays an important role in the aspect of TS pregnancy is the increased risk of maternal and fetal complications, regardless of the method of conception. During counselling, patients should be informed about the possible risks for pregnant women, such as hypertension, pre-eclampsia, aortic dissection, increased risk of caesarean section, or higher risk of preterm birth. Among the risks for the baby, the risk of fetal aneuploidy and congenital malformations, intrauterine growth abnormalities, low birth weight, or preterm birth complications should be kept in mind [6,25]. A study by Bernard et.al involving 480 women with TS, of whom 27 experienced labour, showed that none experienced aortic root dilatation or aortic dissection, while pregnancy-related hypertension (PHD) occurred in 13.3% of pregnant women [26]. A multicentre retrospective study in the UK, including 81 women with TS who became pregnant, reported two cases of aortic dissection. Interestingly, both of these women presented with a 45, X karyotype and a bicuspid aortic valve. In addition, one woman required aortic root replacement six months after delivery [27]. The method of delivery is adapted to the patient's health and condition and the course of the pregnancy and caesarean sections are performed in 46 - 67% of pregnancies in TS women compared to 12% in the general population, mainly due to cephalopelvic disproportion [6,26,27]. The overall miscarriage rate is 47.6% in those with TS compared to 15% in the general population [28,29]. Endometrial abnormalities, which may be due to the absence of genes on the X chromosome that account for endometrial receptivity, have been identified as one of the causes of miscarriage in women with TS. This is supported by a study by Jaron et.al in which, among women with premature ovarian failure, patients with TS had a higher rate of early miscarriage (60%) and a significantly lower rate of births per pregnancy (20%) compared to patients without TS (8.7% and 73.1%, respectively) [30]. Other factors favouring miscarriage include fetal chromosomal anomalies, uterine malformations, and the presence of autoantibodies [31].

In the pre-conceptual preparation of people with TS, a thorough cardiac examination is recommended, together with advice from a maternal-fetal medicine specialist and a cardiologist specialising in TS in relation to the risks of pregnancy in these individuals [6]. According to the recommendations of the Cincinnati International Turner Syndrome Meeting, imaging of the thorax (thoracic aorta and heart) by transthoracic echocardiography (TTE) and cardiac computed tomography/magnetic resonance (CMR) within 2 years before planned pregnancy is recommended in all women with TS [32]. Measurement of the aortic size index appears to be crucial in the cardiac examination, and an ASI > 2.5 cm/m2 is considered a contraindication to pregnancy, due to the risk of aortic dissection [33].

Methods of fertility preservation in TS

It is important to remember that early diagnosis of TS and early assessment of ovarian reserve are crucial for fertility preservation, due to the progressive accelerated depletion of ovarian reserve and the impossibility of predicting the exact time of complete fertility loss.

Spontaneous pregnancy - due to ovarian abnormalities and early follicular loss in women with TS, spontaneous pregnancy can be problematic, but possible. Factors that increase the likelihood of spontaneous pregnancy include: mosaic karyotype with 46,XX cell line, spontaneous maturation, measurable AMH or FSH <10 IU/L -1 [6,26,28]. Spontaneous pregnancies occur at a rate of 5.6% in the population of women with TS and the risk of miscarriage is 30.8%. Pregnancy in TS is associated with a higher risk of termination by caesarean section (46.7% compared with 21% in the general population). The median delay in conception in TS is three months longer than in the general population. Although the mosaic karyotype increases the chance of pregnancy, cases of individuals with a 45.X karyotype with spontaneous pregnancies have been described [7,26]. Chromosomal fetal anomalies, uterine malformations, impaired uterine vasculature, and reduced endometrial receptivity have been identified as factors that may contribute to the increased miscarriage rate in TS patients [31]. Spontaneous pregnancy is also associated with an increased risk of fetal aneuploidy and a lower incidence of maternal complications compared to pregnancies after OD. It is suggested that this may be due to the younger age of patients with spontaneous pregnancies and a milder phenotype. Nevertheless, hypertension affects 13.3% of pregnant women with spontaneous pregnancies [26].

Oocyte cryopreservation - this method is currently used for fertility preservation in oncology patients and its use in Turner syndrome is still poorly documented [34]. The use of this method is limited to the time between puberty and the end of ovarian function [10]. Due to improvements in the technique over the last few decades, currently, implantation and live pregnancy rates are comparable using vitrified oocytes and fresh oocytes [35]. In this process, mature oocytes at meiosis stage II are retrieved under ultrasound guidance after prior ovarian stimulation with gonadotropins. The mature oocytes obtained then undergo vitrification and

cryopreservation. Due to the low number of oocytes that can usually be obtained in patients with TS and the higher risk of miscarriages and aneuploidy in ovarian cells, this procedure is recommended to be performed several times [20,36]. The optimal number of oocytes required to achieve pregnancy in TS has still not been established. When immature oocytes are retrieved (at the embryonic follicle or meiosis I stage), stimulation with gonadotropins is not required [20]. This technique is considered to be effective in both 45,X karyotype, mosaic TS, and also in patients with reduced AMH [37]. In a study by Nadesapillai et.al, 52% of women succeeded in vitrification of at least 20 oocytes, and the median oocyte vitrification in the first cycle was 6. Furthermore, a correlation between 46.XX, FSH, AFC, and AMH cell percentage values and the number of vitrified oocytes were shown [36]. When deciding to cryopreserve oocytes, prenatal genetic testing should be considered due to the increased risk of chromosomal aberrations in the fetus [6]. In 2022, the first case of live birth after vitrification of oocytes in a woman with mosaic Turner syndrome was also described. In a 24-year-old female patient, a mosaic with TS, cryopreservation of 29 oocytes was performed. The patient had regular cycles, no endocrine disorders, and a normal uterine image on ultrasound. After 5 years, oocytes were thawed, of which 23 survived and were fertilised using intra-cytoplasmic sperm injection (ICSI). This resulted in 3 good-quality blastocysts, and they were then subjected to preimplantation genetic testing for aneuploidy (PGT-A). The test revealed 2 blastocysts to be euploid, of which one embryo was transferred into the uterus using the HRT preparation protocol. The pregnancy progressed without complications and ended with the birth of a healthy daughter at 40 weeks of gestation [38].

Ovarian tissue cryopreservation (OTC) - is still an experimental therapy, but appears to be a good alternative to oocyte cryopreservation because this method can be used at any age, does not require ovulation stimulation and preserves ovarian hormonal function and fertility [31,25]. Its disadvantage is its invasiveness, requiring surgical removal of part of the ovary, with the need to obtain as much material as possible. This necessity is because the thawed ovarian tissue does not show its full function, which may be due to its hypoxia and over-activation of follicle growth, and its possible damage through the use of coagulation during the procedure [25]. In this method, the harvested material is frozen and is used after thawing when the patient decides to become pregnant [20]. Ovarian tissue transplantation is performed in one of two locations, depending on whether the patient has managed to retain part of the ovary after the ovarian cortex retrieval procedure. Orthotopic transplantation is performed in patients with a retained ovary and involves transplanting tissue into the remaining ovary or mesentery of the ovary. In patients with no retained ovaries, the transplant is performed into a pelvic area with a large blood supply [20]. It is also possible to perform heterotopic transplantation, into sites such as the subcutaneous tissue of the abdominal wall or forearm [25]. More than 60 live births have already been achieved as a result of OTC, and at least two women have given birth to three children each. In addition, ovarian tissue transplantation has been shown to restore ovarian activity for up to five years and, with repeated procedures, this time can be extended to 11 years [39]. However, data on live births after OTC in patients with TS are still lacking. As previously mentioned, Hreinsson et.al analysed the number and density of follicles in ovarian cortical tissue taken from nine adolescent TS patients. The presence of follicles was demonstrated in eight patients, with the highest number of follicles found in younger patients and those with a mosaic karyotype, while the highest follicle density was found in patients with lower FSH levels [22]. Spontaneous maturation, mosaicism, and normal FSH and AMH levels are considered prognostic factors for obtaining follicles from ovarian tissue [40]. However, the literature reports a case of a patient who successfully underwent OTC despite very low AMH levels (0.1 ug/l; normal 0.56-8.4 μg/l) [41]. Another aspect is that a large proportion of ovarian follicles in TS show abnormal morphology. These include abnormalities in apoptosis and expression of zona pellucida proteins. Furthermore, the follicular fluid shows lower concentrations of oestrogen and testosterone and higher concentrations of anti-Müllerian hormone compared to the control group [24]. On a positive note, studies have shown that despite the presence of aneuploidy in granulosa layer cells and ovarian stroma cells, the vast majority of oocytes show a normal X chromosome number [6,42]. Furthermore, studies using mouse xenograft models have shown that aneuploidy of granulosa and stroma cells in patients with mosaic TS does not affect folliculogenesis [43]. OTC appears to be a fertility preservation option for girls with TS who cannot wait until puberty to undergo oocyte cryopreservation, with preserved ovarian reserve. However, this is an experimental method and, to date, none of the TS patients who have undergone OTC have come forward for transplantation of cryopreserved ovarian tissue.

Oocyte donation (OD) - is a third option for fertility preservation. It is an alternative for those women in whom ovarian reserve has been depleted and it is not possible to obtain their oocytes. Pregnancies achieved by oocyte donation have lower miscarriage rates and higher live birth rates compared to pregnancies achieved using their oocytes in patients with TS [7]. According to a study by Khastgir et.al, pregnancies from oocyte

donation in TS have a pregnancy rate of 41.2% per treatment cycle, and endometrial thickness greater than 6.5 mm, correlated with higher pregnancy rates [44]. In a study by Press et.al, pregnancy rates using ovum donation were comparable for women with TS and women with other causes of ovarian failure [45]. However, oocyte donation is associated with a higher risk of developing pregnancy-induced hypertension, low birth weight, and bleeding in the first trimester of pregnancy as observed in a study comparing in vitro fertilisation using own and donated oocytes [46,47].

Adoption - pregnancy with TS is associated with an increased risk of health complications for the mother and the fetus. Therefore, adoption appears to be a suitable option for women with TS who have contraindications to pregnancy, who have had ovarian function loss, and who are concerned about aneuploidy as a result of pregnancy with their own oocytes.

Surrogate motherhood (GS) - is an option for patients who have contraindications to pregnancy due to a medical condition. In this case, both own gametes (with the risk of aneuploidy) can be used, making it possible to become the biological parents of the child, and from donation, as an alternative for women with POI. However, GS is only available in some countries due to legal restrictions.

Conclusions

Premature extinction of ovarian function leading to infertility is an important part of life for women with TS. Early diagnosis and assessment of ovarian reserve are important so that fertility preservation methods can be implemented before ovarian function ceases. Women affected by TS can have spontaneous pregnancies, but this is rare and is associated with the risk of aneuploidy in the fetus. Among the fertility preservation methods available for post-pubertal women with preserved ovarian function is oocyte cryopreservation. For patients unable to wait until puberty, cryopreservation of ovarian tissue is an alternative, but this is an experimental method with no documented pregnancies among women with TS. Because of the risk of chromosomal defects in fetuses of patients who decide to use their oocytes, they should be informed about the possibility of prenatal genetic testing and preimplantation genetic diagnosis. If it is not possible to obtain own oocytes, oocyte donation is a good option, but it is associated with a higher risk of pregnancy-associated diseases. For women who have contraindications to pregnancy, surrogacy or adoption is an option. When managing a patient with TS, it is important to remember that pregnancy in these women is associated with a higher risk of both maternal and fetal complications and a higher risk of miscarriage. When choosing a method of fertility preservation, the emotional maturity of the patient and her readiness to undergo the necessary procedures should also be taken into account, and counselling should be carried out by a multi-specialist team specialising in the treatment of women with TS.

All authors have read and agreed with the published version of the manuscript.

Funding Statement: The study did not receive special funding.
Conflict of Interest Statement: The authors report no conflict of interests
Funding sources: There are no sources of funding to declare.

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