

Reproductive Future of a Healthy Son in Women with Genetic Syndromes: Cryopreservation and PGD Breaking Barriers

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For many years women with some genetic syndromes burden the fact of being infertile and not be able to have a health sibling. One example is Turner's syndrome (45, XO), which among other things manifest with a premature ovarian failure (POV). Mosaicism in some Turner's syndrome patients can allow them to have their first period but it doesn't avoid the decrease in the ovarian reserve, ending the chances of reaming fertile when patients achieve the age to start a family [1,2].

Spontaneous pregnancy is possible and has been reported, nevertheless due to young age as most patients present the POV, making them physiologically immature to conceive, and the fact that some of them do not even reach the full physical reproductive maturity, relay on that outcome is sometimes impossible.

In that scenario Assisted Reproductive Techniques could represent hope for patients those patients with reproductive future. In vitro fertilization (IVF) and Intra Cytoplasmatic Sperm Injection (ICSI) are known establish procedures to help infertile patients, with or without genetic syndromes. In order to help young female with genetic syndrome. IVF or ICSI, are some tolls that could be used, such as cryopreservation and PGD.

Cryopreservation

Cryopreservation of the embryo and the oocyte are two well-established procedures in the literature for post pubertal women. The results from its use are similar, in some cases better, then the fresh components used in IVF/ICSI. Their use could be tricky when considering genetic syndromes patients. For the embryo cryopreservation process, a male partner is required and ovarian hyper stimulation as well. For the oocyte cryopreservation ovarian hyper stimulation is needed.

Genetic syndromes patients may develop POV in a very young age, when it is rare to have a male partner. The young age is also a barrier, considering that these patients do not present the physiological matureness to understand everything evolved in the whole process. Considering the need for ovarian hyper stimulation in both techniques, such patients require maturation of the hypothalamic-pituitary axis.

For those patients ovarian tissue cryopreservation technique seems to be the best alternative. Such procedure need a surgery intervention and it is still limited to reference centers and is applied most for pre-pubertal cancer patients for their fertility preservation. It consists in taking a part of the ovarian tissue with primordial follicles to be stored frozen. When the patient decides to restores their fertility, through laparoscopic surgery the tissue is implanted in the abdominal cavity. The literature showing promising results especially for returning the women's ovarian function [3].

To offer ovarian tissue cryopreservation for genetic syndromes patients is to offer an option for them to take control of their reproductive future, giving back the chance of a natural pregnancy when the patient is ready.

PGD

PGD stands for Pre Genetic Diagnosis, a useful tool to diagnose

some possible genetic condition on the embryo before it is implanted in the uterus. It is useful for couples that have a chance of conceiving a child with some genetic condition that can be detected through a single mutation on a gene or chromosome. That fits right in to a genetic syndrome patient, considering that more than 300 conditions can be detected. Using the example of Turner's syndrome again, there's a chance for the baby to be 45, XO. The mother to be could have her embryo diagnosed before the implantation.

Nowadays PDG can be done in a lot of different embryo development stages by multiple types of biopsy methods. The first step is to take some DNA sample from the embryo been developed. The oocyte that is going to be used could be the source for the DNA sample too, although the embryo sample could provide a better diagnosis chance once both, mother and father, DNA is analyzed. The sample is then sequenced to show the mutations [4].

The biopsy done in the embryo for DNA sample, it is a very delicate process and requires some ability to be done. The literature still discuss if these kind of intervention could bring some damage for the embryo as the rate of those who suffer the biopsy and do not develop any further is 20%. Another point in discussion is that once you have the diagnosis, what is done with that embryo, knowing that it is not a 100% accurate method.

Major progress has been done in the assisted reproductive field for these patients with known genetic syndromes giving them back the chance of building a genetic disease free family. Cryopreservation of the ovarian tissue could represent their chance of having restored their fertility and PGD could help them not conceive a child with known genetic condition and also with another possible genetic abnormalities.

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