**Turner Syndrome**

**What is Turner syndrome?**

Turner syndrome is a genetic condition caused by a missing X chromosome. Chromosomes contain the instructions that tell our bodies how to grow, develop, and function. A missing chromosome may result in health problems.

Usually females have two copies of the X chromosome in most of the cells in their bodies. Females with classic Turner syndrome have one copy of the X chromosome in the cells of their body. Some females with Turner syndrome may have *mosaic* Turner syndrome, which means that some cells in their body have one X chromosome while other cells may have two or three X chromosomes or an X and a Y chromosome.

Females with Turner syndrome often have multiple health problems. Some females are born with heart or kidney problems. Additionally, some females with Turner syndrome may be shorter than expected based on their family member’s heights; they may have learning disabilities; they may have low bone mineral density; and they may experience delayed or absent puberty.

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How does Turner syndrome affect puberty?

Some girls with Turner syndrome have normal puberty. However, many girls with Turner syndrome have delayed or absent puberty due to ovarian failure. This means that their ovaries are not producing the estrogen required to enter puberty. Girls with delayed or absent puberty may be treated with Estrogen replacement therapy (ERT) by their endocrinologist.

What is Estrogen Replacement Therapy (ERT)?

ERT is a standard treatment recommended for girls with Turner syndrome who have not started puberty by age 12-14 years. ERT is often a successful way to promote puberty because it replaces the estrogen the body can’t make on its own. ERT may be taken as a pill or patch, depending on what is prescribed by the endocrinologist. It is important to discuss the side effects of ERT with an endocrinologist before beginning treatment.

Does ERT help fertility?

Although ERT helps girls with Turner syndrome enter puberty, it has not been shown to increase fertility.

How does Turner syndrome affect Fertility?

Approximately 95% of all women with Turner syndrome develop ovarian failure, usually because they have far fewer eggs than women without Turner syndrome. The ovaries in women with Turner syndrome are often not capable of releasing eggs regularly, or in some cases, at all. When and how quickly ovarian failure occurs in females with Turner syndrome is unclear and may be different for each woman with Turner syndrome. Research suggests that ovarian failure in some females with Turner syndrome may start as early as before birth. There may be time before complete ovarian failure occurs to preserve fertility and allow women with Turner syndrome to have biological children.

Fertility preservation options should be discussed with females with Turner syndrome and their families at all life stages, as it may be best to pursue fertility preservation early in life.

Female Fertility Preservation Options

Ovarian Tissue Banking

Ovarian tissue banking is an experimental option for females of any age. This is the only option for girls who have not started puberty and the recommended option for girls with Turner syndrome. Ovarian tissue banking involves surgical removal of part or all of an ovary, which is then frozen for future use. The procedure can be done any time, as no stimulation of eggs is needed.

Egg and Embryo Banking

Embryo and egg banking are options only for females who have gone through puberty. Therefore, they may not be applicable to most women with Turner syndrome. First, a woman’s ovaries are stimulated to mature multiple eggs, which are then removed. They can be frozen at this point (egg banking) or fertilized with sperm using in vitro fertilization (IVF) to create embryos. The embryos are frozen for future use. Embryo and egg banking can take up to one month.

Information found here or elsewhere on the oncofertility.northwestern.edu website should not be considered medical advice, diagnosis, or treatment. Any information on this document or website should not be used in lieu of consultation with your healthcare provider or physician. Before starting any course of treatment, always consult a qualified health care provider. Do not delay seeking or disregard medical advice because of anything you have read or seen here. For information regarding fertility options contact the FERT line at 866-708-FERT (3378).
Online Resources for More Information on Fertility Preservation Options

MyOncofertility.org
http://www.myoncofertility.org

SAVEMYFERTILITY
http://www.savemyfertility.org

http://oncofertility.northwestern.edu

Turner Syndrome

Can women with Turner syndrome get pregnant naturally?
Yes. Approximately 1-2% of women with Turner syndrome get pregnant naturally.

Is pregnancy dangerous in some women with Turner syndrome?
Pregnancy is not recommended for women with Turner syndrome and certain heart complications. Approximately 10% of pregnancies in women with Turner syndrome result in maternal aortic dissection (rupture of major artery that connects to heart) and 2% result in maternal death. Doctors recommend surveillance for high blood pressure and periodic echocardiograms (heart ultrasound) throughout pregnancies in women with Turner syndrome. For these women, gestational surrogacy using their own preserved ovarian tissue or eggs is an option that still allows for biological children.

Alternative Options
Women with Turner syndrome may choose to use donor eggs (if pregnancy is a safe option for them), or consider adoption when family planning.

Questions?
Call the 24-hour FERTLINE to ask your fertility preservation questions, get connected with a fertility preservation program near you, and access resources, tools, and support!