

A Patient Guide to Estrogen Replacement Therapy in Turner Syndrome

What is HRT?

HRT is the abbreviation for hormone replacement therapy, also referred to as ERT, or estrogen replacement therapy. In females, the sex hormones estrogen and progesterone are secreted by the ovaries. If the ovaries are not fully developed, or not functioning properly, these hormones will be missing, causing what is called hypogonadism. Thankfully, these important hormones can be replaced with medication.

Sex Hormones and Turner Syndrome

Many girls with TS will experience ovarian failure early in life, resulting in a deficiency of the hormones, or chemicals, necessary to initiate sexual development/puberty. While a small percentage of girls with TS will show some signs of breast development in their early teens, and some will menstruate on their own, these usually stop sooner than usual. Ultimately, around 90% of women with TS will experience ovarian failure and require estrogen replacement.

Puberty in Turner Syndrome: What to Expect

All girls are born with a supply of eggs in their ovaries. These slowly die over the course of a lifetime. For girls with TS, this loss occurs more rapidly. The pace at which eggs are lost will determine whether or not a girl will begin puberty on her own. Girls with mosaicism are more likely to enter puberty spontaneously.

Pubic hair, body odor, underarm hair, and acne develop under the influence of hormones from the adrenal glands. Estrogen is not the primary force behind them, so girls with TS could develop these characteristics normally.

How Can We Predict Puberty?

There are ways to determine whether or not a girl with TS will experience spontaneous puberty. There are two main lab tests that can help predict future ovarian function. These are typically done around ages eleven and twelve.



- ▶ **FSH (Follicle Stimulating Hormone)**
- ▶ **AMH (Anti-Mullerian Hormone)** Lower AMH levels indicate a smaller number of eggs and earlier loss of ovarian function.

A **high** FSH and a **low** AMH indicates ovarian failure. If your daughter's lab results show normal levels of FSH/AMH, then you can watch for spontaneous puberty, expecting to add estrogen later if needed.

DID YOU KNOW?

Up to 30% of girls with Turner syndrome may spontaneously enter puberty.

The Role of Estrogen in Puberty

Brain signals ovaries to begin producing estrogen

First sign is usually breast development (breast buds)

Breast tissue develops under the influence of estrogen

Estrogen causes maturation of uterus, uterine lining, and vagina

Menstrual periods begin 2-2.5 years after breast development starts

How is Estrogen Given?

Estrogen can be taken orally (in pill form) or by way of a small transdermal patch, that adheres to the skin and delivers the estrogen directly into the bloodstream, bypassing the liver. Both work well. It just depends upon your preference, however the patch application is becoming more common and might offer some advantages over the tablet form. Some patches are applied weekly, while some are applied twice per week. For very small dose delivery, your doctor may recommend cutting the patch into smaller portions.

If you are a woman with TS and have had trouble with side effects of ERT, **don't give up**. Estrogen is **essential** for a healthy body and should be continued at least until the usual time of menopause, around **50 years of age**.

Timing of Hormone Replacement Therapy

- Since the goal is to mimic natural puberty, estrogen may be started if there is no breast development at all by age eleven to twelve.
- Start at a very low dose and gradually increase over 2-3 years to a normal young adult dose. This will allow for natural breast development and maturation of the uterus. Since higher doses of estrogen will cause bone age to advance more quickly, a lower dose is important for preserving height whether or not growth hormone has been started.
- If reaching a taller height is still possible, girls may remain on the lower dose for a longer period of time.
- After two years of estrogen, or when "breakthrough bleeding" occurs, another hormone, **progestin**, is added. This can be given as a pill or combined with estrogen in an oral contraceptive.
- Progestin is given for 10 days per month, resulting in a monthly menstrual period.
- Progestin is extremely important for maintaining a healthy uterus. It regulates periods, allowing the lining to be shed fully and preventing an unhealthy overgrowth of the uterine lining.
- Some girls and women may choose to have a menstrual period by adding progestin every other month or every three months to maintain uterine health.
- You and your doctor can decide when to start estrogen and what type will best suit your or your daughter's needs.

Should I Use the Patch or Pill?

Estrogen Patch:

- ❖ May decrease risk of blood clots
- ❖ May improve blood pressure control
- ❖ May result in better bone mass
- ❖ May improve effectiveness of growth hormone
- ❖ Bypasses the liver

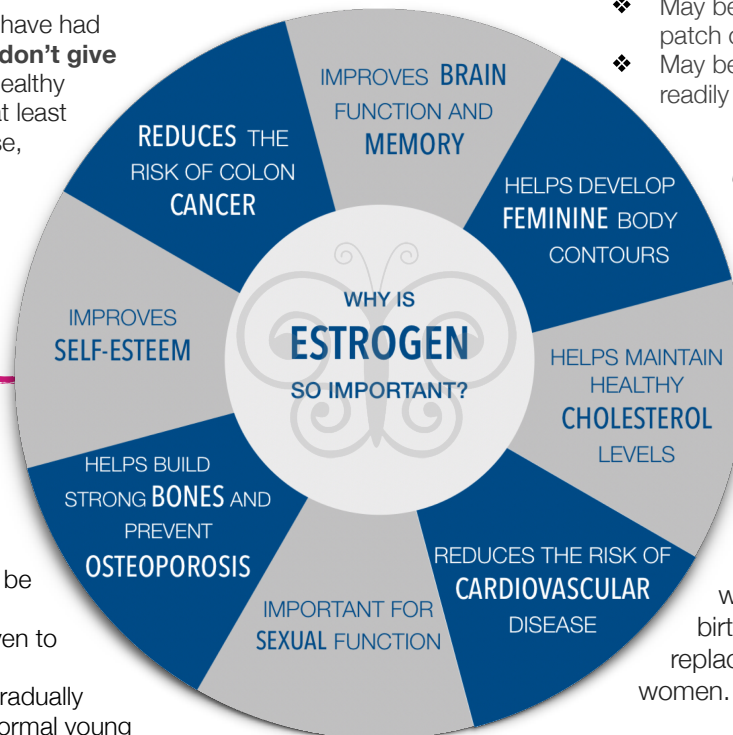
Estrogen Pill:

- ❖ May be more convenient or easier to remember and maintain
- ❖ May be less irritating than an adhesive patch on the skin
- ❖ May be more affordable and more readily available

Ultimately, you should do your research, talk with your doctor, and choose which option works best for you or your daughter.

Important Note

This hormone therapy is replacing a **pure deficiency** and is not the same as hormone treatment in a woman without TS who takes birth control pills or estrogen replacement in postmenopausal women.



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