



the  
**Oncofertility**<sup>®</sup>  
Consortium



## Center for Reproductive Health After Disease

### Fertility Preservation Fact Sheet

## Fragile X- associated Primary Ovarian Insufficiency (FXPOI)

### 5 Questions to Ask Your Health Care Provider

1. As a Fragile X carrier, what is my risk of developing POI?
2. How does FXPOI affect my health and fertility?
3. What therapies may help me regain or prolong ovarian function?
4. What fertility options are available for women with FXPOI?
5. Can I have children in the future?

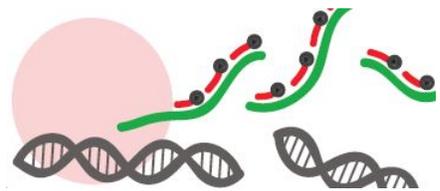
#### What is Fragile X-associated Primary Ovarian Insufficiency (FXPOI)?

Women have two copies of the *FMRI* gene, which contains important instructions that tell the body how to grow and function. Approximately 1.3% of women in the United States have a mistake in one copy of their *FMRI* gene. Mistakes in *FMRI* are caused by three chemical letters, “CGG”, repeated over and over.

Women with 55-200 “CGG” repeats are called fragile X *premutation carriers*; they have ~20% risk to develop Fragile X-associated Primary Ovarian Insufficiency (FXPOI). FXPOI

is a type of *premature ovarian insufficiency* (POI), which is a medical condition where menstruation stops prior to age 40 years. Women with greater than 200 “CGG” repeats, also called *full mutation carriers*, do not have an increased risk of developing POI.

Repeat number also correlates with a woman’s risk of having a child with fragile X, the most common heritable form of mental retardation in males. Genetic counselors are health care professionals who can discuss your specific risk of having a child with fragile X. Learn more at: <http://oncofertility.northwestern.edu/your-genetic-counselor>.



## How Does Fragile X-associated Primary Ovarian Insufficiency Effect Fertility?

POI is also called *premature menopause*, because menstruation stops prior to age 40 years.

Approximately one third of women with FXPOI will have their final menstrual cycle before age 29 years and 1% will have their final menstrual cycle before age 18. The perimenopausal transition makes it difficult to get pregnant, and once menopause is truly reached women can no longer achieve pregnancy.

Early in reproductive life, *FMRI* premutation carriers should discuss their risk of developing FXPOI with their health care provider and consider available fertility preservation options if interested in biological children.



Currently, there are no successful therapies to regain ovarian function in women with POI. However, there are hormone replacement therapy (HRT) and fertility preservation options available to women with FXPOI to consider when family planning.

## Hormone Replacement Therapy (HRT) for FXPOI

The American Society for Reproductive Medicine (ASRM) and the International Menopause Society recommend women with POI

begin estrogen replacement therapy. Hormone replacement in girls and young women provides the hormones their ovaries should naturally produce at their age. The hormones can promote development of secondary sexual characteristics, like: breast development, menstruation, body hair growth, and widening hips.



## Female Fertility Preservation Options

### *Embryo Banking*

Embryo banking is an option for females who have gone through puberty. First, a woman's ovaries are stimulated to mature multiple eggs, which are then removed and fertilized with sperm using *in vitro fertilization* (IVF) to create embryos. The embryos are frozen for future use. Embryo banking can take up to 1 month.

### *Egg Banking*

Egg banking is an option for females who have gone through puberty. It is very similar to embryo banking, except the eggs are not fertilized before freezing. Egg banking is a good option for women who do not have a male partner and do not want to use a sperm donor at the time of the procedure. Egg banking can take up to 1 month.

### *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. Part or all of an ovary is surgically removed and frozen for future use. The procedure can be done any time, as no stimulation of eggs is needed.

Information found here or elsewhere on the [oncofertility.northwestern.edu](http://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>

### Natural Pregnancy in Women with POI

It is important to not assume infertility when you are diagnosed with FXPOI. Research studies estimate a 5-10% chance of pregnancy with POI in the general population. If you are not actively trying to pregnant, use contraception.

### Alternative Options

Women with FXPOI may choose to use donor eggs, 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!

