

FRAGILE X-ASSOCIATED PRIMARY OVARIAN INSUFFICIENCY (FXPOI)

WHAT IS FXPOI?

FXPOI stands for **Fragile X-associated Primary Ovarian Insufficiency**. It is a condition that impacts some women who carry the Fragile X premutation.

FXPOI occurs when the ovaries do not function at full capacity. The ovaries may:

- Release eggs less often or regularly
- Produce lower amounts of hormones like estrogen

About 20% (1 in 5) women with an *FMR1* premutation develop primary ovarian insufficiency (POI), compared to 1% of the general population. Women with the *FMR1* premutation in the 80-100 CGG repeat range have the highest chance (38%) of developing FXPOI.

Women with a full mutation are not at risk for FXPOI.

WHAT ARE SIGNS AND SYMPTOMS OF FXPOI?

Symptoms of FXPOI can mirror symptoms of menopause. These symptoms can occur on a spectrum, so some women may have mild symptoms while other women may have more severe symptoms.



Changes in Your Menstrual Cycle

- Irregular periods
- Skipped periods
- Very light periods
- Cycles shorter than 21 days
- Cycles longer than 35 days
- Periods stopping completely before age 40





Fertility Problems

- Difficulty getting pregnant
- Changes in fertility test results (like elevated Follicle Stimulating Hormone (FSH) or low Anti-Mullerian Hormone (AMH))

Pregnancy is still possible with FXPOI. Unlike complete menopause, ovarian function can sometimes come and go. Your ovaries may occasionally release eggs, and regular periods can return.

Early Menopause



- Hot flashes
- Night sweats
- Vaginal dryness
- Trouble sleeping
- Mood changes
- Irritability
- Anxiety
- Trouble concentrating (“brain fog”)
- Lower sex drive

These symptoms are generally due to lower levels of estrogen. Women with a premutation, on average, experience menopause five years earlier (ages 40-45) than women without a premutation.

WHAT DO I DO NEXT?

If you notice changes in your menstrual cycle, symptoms like hot flashes, or have concerns about fertility (especially if you know you carry the Fragile X premutation), you should speak with your healthcare provider. A primary care provider or OB/GYN can review FXPOI with you and help you understand what evaluations or next steps might be appropriate.

If you are having difficulty getting pregnant, you may want to discuss your family planning options with a reproductive endocrinologist or genetic counselor. Some women also find it helpful to join support groups or connect with other women with similar experiences.



Want to know more?

- fragilex.org
- contact@fragilex.org

