



Maternal Causes of Infertility and Miscarriage

Have you been diagnosed with premature ovarian failure (POF), polycystic ovarian syndrome (PCOS), or hypogonadism (low or no hormone production for ovaries)? In some cases, there can be an underlying genetic explanation for this, including:

- A missing X chromosome ([Turner syndrome](#)) in some (called [mosaicism](#)) or all of an individual's cells.
- A woman with a Y chromosome instead of a second X chromosome (Swyer syndrome).
- Changes in a gene called *CYP17A1*, which causes a condition called congenital adrenal hyperplasia, which can cause fertility issues among other health concerns.
- Being a carrier for a genetic condition called [fragile X syndrome](#). Women who carries for this are at a higher risk for premature ovarian failure.
- Harmful changes (called pathogenic variants) in a gene called *GALT*, which causes a condition called galactosemia.
- Chromosome [translocations](#) or rearrangements (when pieces of two different chromosomes break off and switch spots with each other).

Meeting with a healthcare professional, such as a genetic counselor, to walk through your personal and [family history](#) can be helpful to determine the likelihood of a genetic cause for these concerns. For the majority of cases of POF, PCOS, and hypogonadism, testing for all of the above things will likely come back normal, and we're unable to determine the specific underlying cause.

Click [here](#) to learn more about scheduling a genetic counseling appointment for infertility or preconception questions.