ORM GENOMICS

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A Patient Guide

ABOUT ORM Oregon Reproductive Medicine (ORM) is a world-class fertility center that is passionately committed to helping people grow their families. ORM is dedicated to achieving the highest pregnancy success rates while providing a customized, compassionate patient experience. Unrivaled expertise, outstanding results and personalized care make ORM highly sought after around the world. ORM has the largest in-house genomics program of any fertility center in the U.S., with four board-certified genetic counselors dedicated to supporting our PGD patients.

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PREIMPLANTATION GENETIC DIAGNOSIS:

HEREDITARY CANCER SYNDROMES

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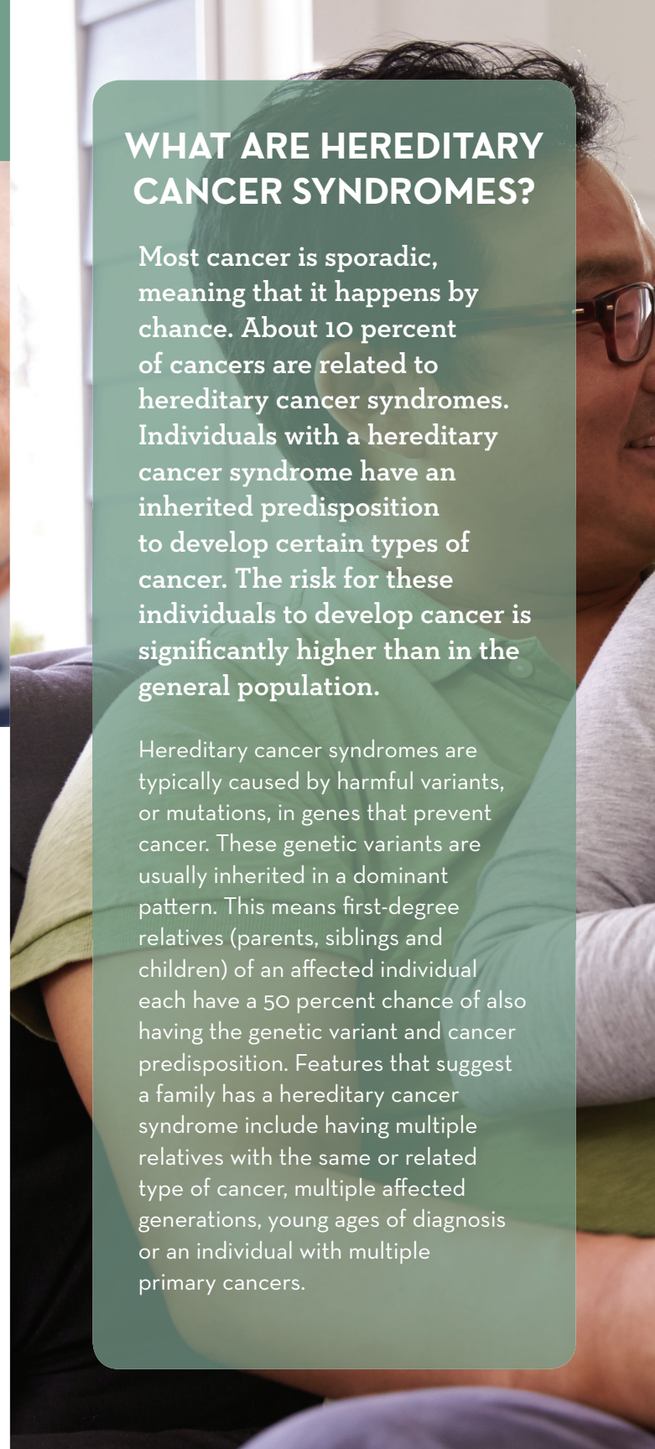

WHAT ARE HEREDITARY CANCER SYNDROMES?

Most cancer is sporadic, meaning that it happens by chance. About 10 percent of cancers are related to hereditary cancer syndromes. Individuals with a hereditary cancer syndrome have an inherited predisposition to develop certain types of cancer. The risk for these individuals to develop cancer is significantly higher than in the general population.

Hereditary cancer syndromes are typically caused by harmful variants, or mutations, in genes that prevent cancer. These genetic variants are usually inherited in a dominant pattern. This means first-degree relatives (parents, siblings and children) of an affected individual each have a 50 percent chance of also having the genetic variant and cancer predisposition. Features that suggest a family has a hereditary cancer syndrome include having multiple relatives with the same or related type of cancer, multiple affected generations, young ages of diagnosis or an individual with multiple primary cancers.

Does my family have a hereditary cancer syndrome?

Genetic counseling and testing is available for families with a history suggestive of a hereditary cancer syndrome. Genetic testing is typically done with either a blood test or a saliva test. If this testing identifies a genetic variant, other family members have the option of having testing done to determine whether they also have an increased risk of developing cancer. There are pros and cons to genetic testing, and it is best to discuss your family history and this testing in detail with a genetic counselor or your physician.

What are my reproductive options?

Most individuals with a hereditary cancer syndrome have a 50 percent chance of passing the gene to their offspring. Those who wish to decrease the chance of passing the gene on may consider prenatal diagnosis, preimplantation genetic diagnosis (PGD), or using an egg or sperm donor.

Prenatal diagnosis is typically done by chorionic villus sampling or amniocentesis. Individuals who have prenatal diagnosis would have the option of continuing or terminating an affected pregnancy.

If you choose not to have prenatal diagnosis or PGD, your children can have genetic testing after they are born.

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What is preimplantation genetic diagnosis (PGD)?

PGD is the testing of embryos for a specific genetic or chromosomal condition in a family, such as a hereditary cancer syndrome. Patients undergo in vitro fertilization (IVF), a process in which eggs are retrieved from a woman's ovaries and fertilized with sperm in an embryology laboratory. The resulting embryos develop in a sterile and womb-like environment for five to six days. Next, three to eight cells are safely removed from each embryo for genetic testing, which can identify the embryos that inherited the gene mutation and those that did not. An embryo that did not inherit the genetic condition is transferred into a woman's uterus in the hopes of achieving pregnancy. Any resulting children are at very low risk of inheriting the genetic condition.

Why do people choose PGD?

PGD offers hope to individuals or couples at risk of passing on a condition that could impact the quality of life of their offspring. Patients report that preventing their child from inheriting a hereditary cancer gene is one of the most important things they can do as a parent. As all pregnancies conceived spontaneously would be at risk of having the heritable condition, invasive prenatal diagnosis is the only other option to learn the genetic status of offspring prior to delivery. Many people wish to avoid being in the stressful and emotional position of learning this information while pregnant. PGD provides reassurance that the risk for their child to be affected is very low. PGD can provide peace of mind during pregnancy and after birth, and can lead to better long-term outcomes for the family and their children.

For which conditions can PGD be offered?

Generally, PGD can be offered for any hereditary cancer syndrome if the genetic mutation(s) in the family is known. PGD has been successfully performed for many hereditary cancer syndromes, including the following:

- Hereditary breast and ovarian cancer (BRCA 1 & 2)
- Lynch syndrome (hereditary nonpolyposis colon cancer)
- Familial adenomatous polyposis
- Hereditary diffuse gastric cancer
- Li-Fraumeni syndrome
- Multiple endocrine neoplasia
- Von Hippel-Lindau
- Retinoblastoma

What if I live outside of the U.S.?

Some countries have strict laws regarding genetic testing on embryos, egg donation and surrogacy. In addition, some countries do not provide coverage or access to IVF to same-sex couples or single individuals. ORM welcomes all global patients who do not have access to PGD or IVF clinics with high success rates.

Who can access PGD?

PGD is generally available to patients with any condition in which the genetic mutation responsible for the condition in the family is known. Some patients come to us already having identified the mutation in their family. Others need help facilitating the genetic testing process. In either case, we can help.

ORM offers complimentary case reviews with a board-certified genetic counselor.

How much does PGD cost?

The costs vary depending on the IVF clinic and the specific situation. We recommend that you reach out to our team if you would like to receive a personalized estimate.

Where can I find more information?

Contact an ORM Genetic Counselor for a complimentary PGD case review at:

geneticcounselor@ormgenomics.com

ormgenomics.com

877.567.4994 or 503.274.2994