ORM GENOMICS

PREIMPLANTATION GENETIC DIAGNOSIS FOR HEREDITARY CANCER SYNDROMES

A Patient Guide

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WHAT IS HERIDITARY CANCER SYNDROME?

Most cancer is sporadic, meaning that it happens by chance. About 10% 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 of developing cancer in these individuals 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% chance to also have 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 heriditary 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 would have the option of having testing 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?

Your options include prenatal diagnosis, preimplantation genetic diagnosis (PGD), using an egg or sperm donor, or proceeding without genetic testing.

Prenatal diagnosis is typically done by chorionic villus sampling or amniocentesis. These tests are associated with a risk of miscarriage (typically about 0.5%). 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. If the risk is for adult-onset cancer only, your children would have the option of having genetic testing when they are adults. Individuals who are at increased risk of cancer may have access to additional screening or preventative options (such as prophylactic surgery).

What is preimplantation genetic diagnosis?

PGD is the testing of embryos for a specific genetic or chromosomal condition in a family. Patients undergo in vitro fertilization (IVF), a process in which eggs are retrieved from a woman's ovaries and fertilized with sperm in the lab. The resulting embryos develop in a sterile and womb-like environment for 5-6 days. Next, about 3-8 cells are safely removed from each embryo for genetic testing. 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 familial cancer syndrome.

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. 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 couples with reassurance that the risk for their child to be affected is extremely 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.

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Find a Local Genetic Counselor

www.findageneticcounselor.com

Speak to your physician or oncologist

Contact an online genetic testing lab such as counsyl.com or color.com

Contact an ORM genetic counselor at geneticcounselor@portlandivf.net

Where can I find more information?

View our PGD Webinar at: ormgenomics.com

Contact an ORM genetic counselor at: geneticcounselor@portlandivf.net

Refer yourself (or a patient) at: ormgenomics.com (click the blue REFER button)

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 of 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.

ORM offers complementary consultations with a board-certified genetic counselor and an ORM reproductive endocrinologist to patients who live outside of the U.S who are considering PGD.

Who can access PGD?

Generally, PGD can be offered for any condition in which the genetic mutation(s) responsible for the condition in the family are known. Some patients come to us already having identified the mutations in their family. Others need help facilitating the genetic testing process. In either case, we can help.

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 an estimate.

ABOUT ORM Oregon Reproductive Medicine (ORM) is a world-class fertility center that is passionately committed to helping people grow their families. ORM is committed to achieving the highest 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 4 board-certified genetic counselors.

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