Genetic Information and Family Tree (GIFT)

Donor #: XD277191

Donor's year of birth: 1990
Ethnicity: German/English/French (paternal)
Italian/Irish/French (maternal)

Genetic assessment is an important part of egg donor screening, matching, and identifying potential risk factors and disease susceptibilities for children who are donor-conceived. While our risk assessment evaluates the personal medical history, family history, and carrier screening results of the egg donor, it does not take into account the intended parent(s) genetic information, which should be evaluated separately.

Family History

As with all ORM egg donors, this donor's personal and family history meets the 2012 gamete donation guidelines of the American Society of Reproductive Medicine (available upon request). This assessment focuses on conditions in the donor's family history that may result in some increased risk to genetic offspring of this donor, as well as conditions with low risk that we anticipated intended parents may have questions about. Please see the attached family tree for full details of this donor's family history.

Personal medical history:
Donor XD277191 considers herself to be in excellent health. She reported that she has no major/chronic medical issues. She did not report having any birth defects, learning disabilities, or mental health issues.

Possible increased risk:
Donor XD277191 reported that her mother and maternal grandfather have high cholesterol. High cholesterol is a common, multifactorial condition that is caused by both genetic and environmental risk factors. Specific environmental factors such as a diet high in saturated fat, cholesterol, or trans-fatty acids, a sedentary lifestyle, or smoking can interact with a person's genetics to cause high cholesterol. Because genetic factors that contributed to the egg donor's mother and grandfather's diagnoses of high cholesterol may have been passed on, offspring from this donor's eggs may have an increased risk for high cholesterol as they get older.

Low/general population risk:
Donor XD277191 reported that her sister has eczema. Eczema is a chronic inflammation of the skin that tends to occur mostly in children, but can occur at any age. Symptoms include itching, dryness, redness, rash, and/or blistering which may flare up periodically and then subside. Many children with eczema (up to 80%) will develop hay fever and/or asthma. Eczema is multifactorial, meaning both genetic and environmental risk factors must be present for someone to develop the condition. Some examples of triggers are dry skin, immune problems, bacteria (such as Staphylococcus aureus, which can block sweat glands), exposure to allergens such as dust

mites, pets, pollens and molds, food allergies and stress. In the general population, 15-20% will have symptoms of eczema. Given that the affected relative is a second degree relative to offspring created from this donor's eggs, the risk for eczema and/or another allergic problem is likely similar to the general population risk.

Donor XD277191 reported that her mother has anemia. Anemia is a condition in which there are not enough healthy red blood cells to carry adequate oxygen to the body's tissues. Having anemia may cause symptoms of fatigue and weakness. There are many forms of anemia, each with its own cause; it can be temporary or long term, and it can range from mild to severe. Considering that the donor's mother is otherwise healthy, this was most likely a temporary anemia caused by vitamin or iron deficiency. All egg donors are screened for anemia prior to their IVF cycle, and certain genetic forms of anemia (such as being a carrier of thalassemia) are screened for on the Counsyl screen.

Donor XD277191 reported that her maternal grandfather was diagnosed with Parkinson disease at age 73. Parkinson disease is a progressive disorder of the nervous system that can cause tremor, stiffness, slow movements, depression, and dementia. Researchers have identified specific genetic mutations that can cause Parkinson disease, but these are uncommon except in rare cases where many family members are affected. Most cases of Parkinson disease are caused by a combination of genetic factors that are inherited and can make a person susceptible, but certain environmental factors also have to be present in order to develop symptoms. Specific environmental factors include exposure to certain toxins like herbicides and pesticides or head trauma. The biggest risk factor for Parkinson disease is age. Over 3% of the general population will develop Parkinson disease throughout their lifetime. At this time, the risk for Parkinson disease is to the egg donor's mother; her cumulative lifetime risk of developing Parkinson disease is increased to between 3% and 7%. Based on the egg donor's maternal grandfather being the only person in the family who has been diagnosed with Parkinson disease, and his older age at diagnosis, the risk for Parkinson disease in offspring created from this donor's eggs is likely not increased above the general population risk.

Donor XD277191 reported that her paternal grandfather died of lung cancer at age 78. Smoking causes the majority of lung cancers, both in smokers and in people exposed to secondhand smoke. The risk of lung cancer increases with the length of time and number of cigarettes someone has smoked. Given that the donor's paternal grandfather was a lifetime smoker, the risk for lung cancer to develop in the donor's offspring would not be expected to be higher than general population risk, assuming that the offspring do not smoke.

Family histories on both sides of donor XD277191's family were otherwise negative for genetic conditions, chronic health conditions, birth defects, multiple miscarriages, and learning disabilities that would significantly increase the risks to offspring created using her eggs. Her family history was also otherwise negative for early-onset adult conditions that would be clearly associated with significantly increased risks above the general population. Family history assessment is based on the information reported by the donor at the time of her application. Family histories are dynamic and are expected to change over time. Donors are instructed to contact ORM should there be any significant changes to her personal or family histories prior to or following egg donation so that we may inform intended parents of any important changes.

We suggest that the donor's family history in its entirety be shared with your child's pediatrician and future medical providers, as early diagnosis and treatment are important to long-term outcomes of most conditions. While many of the conditions seen in donors' families are adolescent- or adult-onset, protective measures

(healthy diet, regular exercise, and maintaining normal weight) and avoidance of risk factors (obesity, poor diet, and smoking) may help to prevent symptoms of certain conditions from ever developing in your child.

Genetic Carrier Screening

Carrier screening is genetic (DNA) testing that investigates whether an egg donor "carries" gene changes (mutations) that may cause a disease in their genetic offspring. We typically have two copies of every gene, one from the egg provider and one from the sperm provider. Carriers of a genetic disorder have one gene copy that is not working correctly. Because the other copy is working fine, carriers usually have no signs of the disorder. If an egg donor and the sperm provider are carriers of mutations in the same gene, there is a 25% chance for the offspring to have that disorder due to *recessive* inheritance.

ORM follows the carrier screening recommendations of the American College of Obstetrics and Gynecology and the American College of Medical Genetics (ACOG/ACMG), and in addition performs "expanded" carrier testing for >100 genes on the **Counsyl Family Prep Screen** (www.counsyl.com). Being a carrier is common; about 50% of our donors/patients who undergo Counsyl screening test positive for at least one of the disorders on the panel, even when there are no genetic conditions in their family history. It is important to recognize that screening "negative" (normal) for a gene does not ensure that one is not a carrier. The risk that one could still be a carrier of a gene mutation, even after a negative screen, is specific to each disease and is provided with all negative results.

Counsyl results on Donor XD277191 : **Negative 2.0 Screen**

Donor XD277191 was not found to be carrier of any of the conditions included on the Counsyl Family Prep Screen 2.0. These results may be reviewed with a Counsyl genetic counselor to receive information about the conditions screened for as well as to discuss options for screening the male reproductive partner(s).

Risks for Chromosomal Abnormalities

Every pregnancy has a small risk for a randomly occurring chromosome abnormality. When using an egg donor who is 20-30 years old, the risk for a baby to have a chromosome abnormality such as Down syndrome (trisomy 21) is ≤ 1/380 or 0.3%. However, as chromosome abnormalities are very common in embryos and can affect the ability of an embryo to implant and progress to a healthy baby, testing called comprehensive chromosome screening (CCS) is made available to all patients undergoing donor egg IVF cycles at ORM. Information about a donor's previous IVF cycles, if any, and any CCS testing that may have been done in those cycles, is available upon request.

Background Risks

The vast majority of babies are born healthy. However, the background rate of birth defects and genetic conditions in any pregnancy/baby is 3-4%. Some studies have suggested an increased risk for birth defects or genetic conditions in pregnancies conceived through IVF. This information is outlined further in ORM's IVF consent form.

If you have any questions regarding this information, please contact us at geneticcounselor@portlandivf.net

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PEDIGREE KEY

	Male
	Female
	Gender Unknown
	Number of children of sex indicated, where 'n' is an unknown number.
	Spontaneous abortion (miscarriage)
	The arrow refers to the patient providing their family history, known as the 'proband.'
	Deceased
	Age of individual
Dx.	Diagnosed
d.	Died of
	Donor
	Surrogate
A&W	Alive and well

XD277191

German/English/French

Italian/Irish/French

d. 78 yrs lung cancer
(heavy smoker)

82 yrs
A+W

82 yrs
high cholesterol
Parkinson dx. 73

78 yrs
emphysema dx. 60's
(smoker)
A+W

59 yrs
A+W

55 yrs
A+W

56 yrs
A+W

55 yrs
anemia
high cholesterol

55 yrs
A+W

57 yrs
hyperthyroidism

n

A+W

n

A+W

26 yrs
A+W

28 yrs
eczema

n

A+W

n

A+W

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