Oregon Reproductive Medicine | ORM GENOMICS

Genetic Information + Family Tree (GIFT)

Donor #: XD155730

Donor's year of birth: 1987
Ethnicity: Caucasian/English (paternal)
English (maternal)

GIFT updated: August 2018
Valid for donations before: September 2019

Genetic Assessment

Genetic assessment is an important part of egg donor screening, matching, and identifying potential risk factors and disease susceptibilities for donor-conceived children. 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 recipient(s) genetic information, which should be evaluated separately.

Family history assessment is based on the information reported by the donor at the time of the genetic consultation. Family histories are dynamic and are expected to change over time. This assessment should be repeated on a yearly basis. This document will be considered out of date for egg donations occurring after August 2019. 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 recipients 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.

As with all ORM egg donors, this donor's personal and family histories meet 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 about which we anticipated intended parents might have questions. Please see the attached family tree for full details of this donor's family history.

Family History

Donor's medical history:

Donor XD155730 reported that she is in excellent health. She reported that she has no other major/chronic medical issues. She did not report having any birth defects, learning disabilities, or mental health issues.

Family history with possible increased risk to offspring:

Donor XD155730 reported that her father has hypertension (high blood pressure) and high cholesterol. She reported that her maternal grandfather also has a history of hypertension. Hypertension causes the heart to work harder to supply blood to the rest of the body, which can damage and weakens blood vessels, and increases the risk of blood clots. Cholesterol is a waxy substance that comes from the body and food. Cholesterol can form fatty, waxy deposits called plaques in the arteries, which reduces blood flow. Over time, high cholesterol and hypertension can damage your arteries, contribute to heart disease, and increase your risk for a stroke. Hypertension and high cholesterol are multifactorial, meaning both genetic and environmental risk factors have to be present in order to develop symptoms. Specific environmental factors such as a diet high in fat, a sedentary lifestyle or smoking can interact with a person's genetics to cause high cholesterol or hypertension. High cholesterol and high blood pressure are very common in the general population, so it is unlikely that this history would significantly increase the risks for offspring conceived with this donor's eggs to develop them. However, the donor is young and may develop these conditions as she ages; if she does, the risks to offspring may be higher. We therefore recommend sharing the family history with the child's medical providers with advancing age.

Donor XD155730 reported that her mother has degenerative arthritis. Degenerative arthritis (also known as osteoarthritis) is the most common form of arthritis, affecting millions of people worldwide. It occurs when the protective cartilage on the ends of the bones wears down over time, causing joint pain and stiffness (mainly in the hands, knees, hips and spine). Osteoarthritis often gradually worsens, and no cure exists. However, staying active, maintaining a healthy weight and other treatments may slow progression of the disease and help improve pain and joint function. Osteoarthritis is multifactorial, meaning both genetic and environmental factors must be present for someone to develop the condition. It is mostly related to aging and repetitive use of the joints over the years. About 1 in 5 (20%) adults in the U.S. population report a history of arthritis as diagnosed by a physician. Offspring conceived with this donor's eggs could have a slightly increased chance to have degenerative arthritis with age.

Family history with low/general population risk to offspring:

Donor XD155730 reported that her father had a sister who died around one week old. The donor was not aware of a cause of death. There are many potential causes of infant death including genetic diseases, birth defects, prematurity, low birth weight, infection, complications of pregnancy or delivery, and others. When the cause is truly unknown, it may be referred to as sudden infant death syndrome (SIDS). The cause of SIDS in some cases may relate to underlying brain abnormalities (related to the portion of the brain that controls breathing/sleep arousal), low birth weight/prematurity, or respiratory infections; however, additional causes and risk factors have yet to be identified. In this case, the donor did not know the cause of the infant death. Given that this was an isolated case in the family and that the donor's grandparents had four other healthy children, it is unlikely that this history would be associated with increased risks to offspring conceived with this donor's eggs Specific risks cannot be provided, however, since we do not have more detailed information.

Donor XD155730 reported that her paternal grandfather died of a brain aneurysm at age 69. A brain aneurysm is a bulge or ballooning in a blood vessel in the brain. A brain aneurysm can leak or rupture, causing bleeding into the brain (hemorrhagic stroke). While most brain aneurysms don't ever rupture, create health problems, or cause symptoms, a ruptured aneurysm quickly becomes life-threatening and requires prompt medical treatment. About 1/10,000 Americans experience a ruptured aneurysm per year, while about 1/50 has an un-ruptured aneurysm. A number of factors can contribute to weakness in an artery wall and increase the risk of a brain aneurysm. Genetic factors likely play a role, while additional risk factors include older age, smoking, high blood pressure, hardening of the arteries, drug or alcohol use, head injury, and others. Rarely, brain aneurysms are associated with the presence of an underlying genetic syndrome that can be inherited. Based on the information provided by the donor, there is no suggestion of a family history of these particular genetic syndromes. When two or more family members have experienced a brain aneurysm, this is called "familial intracranial aneurysms" and other family members are at increased risk to have an aneurysm. Fortunately, this donor did not report any other affected family members aside from her grandfather. Given the information that the donor provided, the risk for offspring conceived with this donor's eggs to experience a brain aneurysm is likely similar to the general population risk.

Family histories on both sides of donor XD155730'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.

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 XD155730: Negative 1.0 Screen (101 conditions)

Donor XD155730 was not found to be a carrier of any of the conditions included on the Counsyl Family Prep Screen 1.0. There is an updated version of the Counsyl screen now available. 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), or updating the donor's screening.

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

XD155730

Caucasian/English

d. 69 yrs brain aneurysm

d. 87 yrs old age

67 yrs
A+W

A+W

60 yrs
A+W

2

A+W

58 yrs
A+W

A+W

A+W

d. 1 wks ?COD

66 yrs
hypertension
high cholesterol

32 yrs
A+W

9 yrs
A+W

5 yrs
A+W

31 yrs
A+W

ectopic pregnancy

4 yrs
A+W

1 yrs
A+W

English

91 yrs
hypertension

94 yrs
A+W

may not know all minor health issues

60 yrs
degenerative arthritis

2

A+W

A+W

68 yrs
A+W

A+W

A+W

65 yrs
A+W

A+W

A+W

64 yrs
A+W

A+W

A+W

Recorded by GS 08/2018