INFORMATION SHEET FOR PARENTS WHOSE SPERM DONOR MADE MULTIPLE DONATIONS

I learned that my child's sperm donor had donated to dozens of other families and may be a carrier of tyrosinemia type 1. What are the genetic risks for my child and potential future grandchildren? What should I do?

A FEW BASIC NOTIONS ABOUT GENETICS

DNA contains the code that makes all life possible. Found in the nucleus of every cell in the body, it's made up of sequences called genes, which provide instructions for all the processes that allow living things to function. Humans have over 20,000 genes! For the vast majority of them, we have two copies: one from the egg and one from the sperm.

Sometimes, changes can be introduced into genes. These are called genetic variants.

! Some genetic variants appear spontaneously, while others are passed down from a person's biological parents.

! Some variants have no impact on health, while others can have a major impact, causing what are known as hereditary or genetic diseases.

GENETIC TESTING

Using genetic testing, scientists can read DNA and detect variants associated with certain diseases. In some cases, there may be several different variants that can cause the same disease.

Some genetic tests can only target a limited number of variants associated with a particular disease. In these cases, a negative result from an incomplete analysis can create a false sense of security.

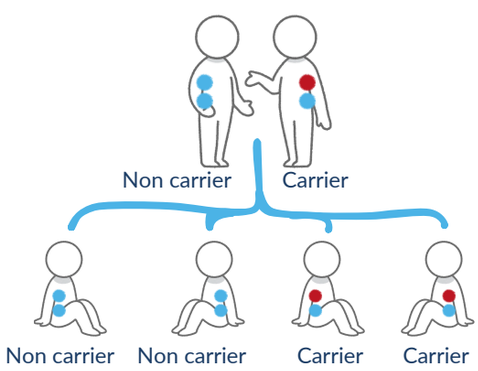
WHAT DOES IT MEAN TO BE A "CARRIER" OF A DISEASE?

In genetics, carriers are people who:

- Have one copy of the gene with a disease-causing variant and one normal copy of that same gene.
- Don't have the disease associated with the genetic variant and are not at risk of developing it in the future.
- Can have children who do have the disease, but only if the other biological parent also carries a variant in the same gene associated with the disease.

This kind of disease transmission is called "autosomal recessive disease". That means that if only one biological parent is a carrier of the variant, there's no risk of the child having the disease.

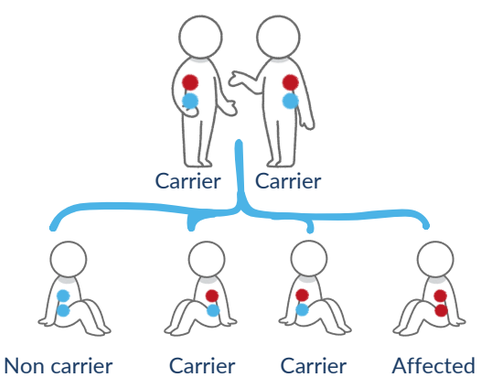
Only one of the two biological parents is a carrier of the variant



There's a 1 in 2 probability that the child will be a carrier and no possibility of being affected.

Normal gene
Variant

Both biological parents are carriers of the variant



There's a 1 in 2 probability that the child will be a carrier and one in 4 to be affected by the desease.

Votre gouvernement

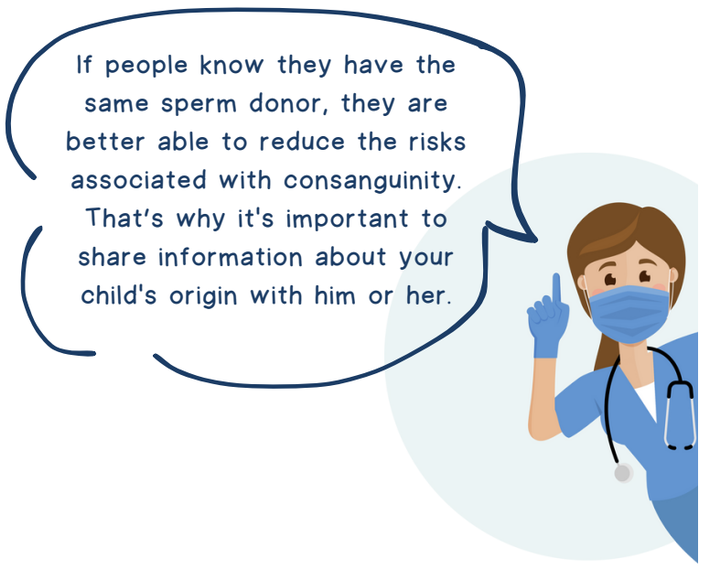
Québec

WHAT IS CONSANGUINITY?

Consanguinity refers to a union between people who share close biological ancestors, usually up to third-degree relatives (first cousins).

If two people have ancestors in common who could be carriers of genetic diseases, a consanguineous union may increase the likelihood of certain conditions in their children beyond the risk in the general population. This can include recessive genetic diseases and congenital malformations (birth defects).

Congenital malformations are more common in children of consanguineous couples (5%–6% of births in third-degree couples, such as first cousins) than in children of non-consanguineous couples (2% of births).



WHAT IS TYROSINEMIA TYPE 1?

Tyrosinemia type 1 is an autosomal recessive disease that affects mainly the liver, but also the kidneys and nervous system. Its worldwide prevalence—the proportion of children affected by it—is one in every 100,000 to 120,000 births.

In Québec, due in part to the founder effect, the prevalence is higher and is estimated at one in 16,000 births, with the number of carriers estimated at one in 66. In Saguenay–Lac-Saint-Jean, Charlevoix, and Haute-Côte-Nord, the prevalence is even higher, at one in 1,500 births and one in 19 carriers.

For more information on tyrosinemia type 1, visit https://www.quebec.ca/en/health/health-issues/a-z/recessive-hereditary-diseases/recessive-genetic-disorders-in-people-from-saguenay-lac-saint-jean-charlevoix-and-haute-cote-nord/hereditary-tyrosinemia-type-1.

WHAT IS FOUNDER EFFECT

When a small group of individuals moves from its place of origin to start a new population, everyone in that group carries some of the genetic characteristics of the original population. If some of them carry genes for hereditary diseases, those diseases will tend to become more frequent in the new population.

The majority of Québec's population is descended from less than 10,000 common ancestors who came from France in the 17th century, and an even smaller number of those ancestors settled in Saguenay–Lac-Saint-Jean, Charlevoix, and Haute-Côte-Nord, where genetic traits resulting from the founder effect can still be observed today.

HOW DO I KNOW IF MY CHILD IS A CARRIER OF A VARIANT ASSOCIATED WITH TYROSINEMIA TYPE 1?

YOUR ARE PREGNANT

Talk to your doctor or midwife, who can refer you to a genetics clinic to determine whether you are a carrier of tyrosinemia type 1.

- If you're not a carrier, you don't need to do anything else, as your child cannot have the disease.
- If you are a carrier and the sperm donor's variant is known and documented, your genetic counsellor may be able to offer you prenatal diagnosis either through a biopsy of the placenta or amniocentesis. The options available vary depending on the stage of your pregnancy. The prenatal genetics team will arrange the test and discuss the benefits, limitations and risks with you.

YOU HAVE A NEWBORN

In Québec, the Neonatal Blood and Urine Screening Program test for a number of rare diseases, including tyrosinemia type 1, in newborns as young as a few days old.

- Screening tests are free of charge for all newborns in Québec.
- In the event of a positive test result, the parents are contacted and the newborn is assigned to a medical care team.
- If the test results are negative and no symptoms are present, the newborn does not have tyrosinemia type 1 and will never develop it.

It's important to note that for tyrosinemia type 1, the screening program can determine whether a child has the disease, but not whether the child is a carrier.

For more information on the screening program, visit https://www.quebec.ca/en/health/advice-and-prevention/screening-and-carrier-testing-offer/blood-and-urine-screening-newborns.

YOU HAVE A HEALTHY CHILD

Healthy children will not develop tyrosinemia type 1 even if they and their sperm donors are both carriers.

Once your child is an adult, a carrier test may be a good idea if they want to start a family. When the time comes, your child can consult their doctor or a genetics clinic and have a professional determine the most appropriate tests for their situation.

The healthcare system offers some carrier tests to people from Saguenay–Lac-Saint-Jean, Charlevoix, and Haute-Côte-Nord who are planning to have a child. However, these tests only target the most common variants in these specific regions.

If you don't know where one of the biological parents or the sperm donor is from, and therefore which variant they might have, the wrong carrier test could produce an incomplete analysis and give you a false sense of security. That's why it's crucial to have a physician or genetic counsellor determine which carrier test to administer.

GLOSSARY

AUTOSOMAL RECESSIVE HEREDITARY DISEASE

Two parents who carry a genetic variant in the same gene can each pass on a copy of the variant to their child. When that happens, the resulting hereditary disease is said to be “recessive”. That’s the case with tyrosinemia type 1: to have the disease, a child must receive a copy of the variant from each biological parent, both of whom would be carriers. If only one biological parent has the variant and passes it on to the child, the child will also be a carrier, but will never develop the disease.

CONGENITAL MALFORMATION OR ABNORMALITY

Congenital anomalies, also known as congenital disorders or congenital malformations, are, in general, abnormalities that affect the development or functioning of organs. Dwarfism and cleft lip and palate (harelip) are examples. They can be detected before or at birth, or sometimes not until later in life. The causes of most of them are unknown, but there are certain known risk factors, including genetics, infections, nutrition, and environmental factors.

CLINICAL GENETIC TEST

This is a type of medical test used by healthcare professionals. Clinical genetic tests can be used to diagnose hereditary diseases in a person or their siblings. They can also be used to determine the genetic status (carrier or non-carrier) of adults with a known family history of a genetic condition, for example if one of their biological parents is a tyrosinemia carrier. Clinical genetic tests are prescribed by healthcare professionals, and some carrier tests are available online. Consult a physician or genetics clinic for more information. For a list of genetics clinics in Québec, visit: https://accgq-qagc.ca/genetics-clinics/.

CARRIER TEST

A carrier test is a clinical genetic test to determine whether an adult is a carrier of a variant in a gene associated with a genetic disease.

DIRECT-TO-CONSUMER GENETIC TEST

Direct-to-consumer tests can assess your risk of developing certain diseases. There are over a hundred companies that sell this type of test online. Some of them offer carrier tests. However, their analyses are often incomplete, targeting only a limited number of genes or variants associated with the diseases in question. And a negative result from an incomplete analysis can create a false sense of security. Direct-to-consumer tests should never be used as a replacement for clinical tests or medical care.

FAMILY RELATIONSHIP TEST

Family relationship tests analyze the DNA of two people to determine a person’s relationship to a biological parent, sibling (brother, sister, half-brother or half-sister), grandparent, uncle or aunt. For reliable results, it’s important to have the test done by a reputable company.

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