

## A Lawyer's Perspective on Genetic Screening Performed by Cryobanks

As a lawyer practicing in the area of sperm bank litigation, I have, unfortunately, represented too many couples that conceived a child with donor sperm that was later born with a genetic disease that was should have been caught. While most Cryobanks represent that certain genetic testing is performed on all donor sperm, in some cases, the testing is either not done at all, done in error, or the results are improperly interpreted, thereby permitting the semen of a donor who tested positive for a genetic trait to be released to their clients seeking to conceive a child.

If you or a friend or family member conceived a child with donor sperm and the child was born with a genetic disease, like Cystic Fibrosis, then you may be entitled to substantial compensation to recover the life-long medical care costs that you will incur to take care of the child. Please feel free to contact me for a no-cost consultation about your individual scenario.

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Every woman has a 3 to 4% chance to have a child with a birth defect, regardless of whether she conceives naturally or uses assisted reproductive technologies. It is not possible to eliminate these risks or test for all of them before becoming pregnant. However, many Cryobanks perform some genetic testing on the donor applicants as part of the donor qualification process.

While it is not possible to test any one person for every genetic disorder, genetic testing is performed on donor applicants for a number of common genetic disorders. Donor applicants who have an abnormal genetic test result are not eligible to participate in a sperm donor program. Donor applicants are eligible to participate only if they have negative genetic test results. The genetic testing helps to reduce the risk for these specific disorders in the donors' offspring.

Some Cryobanks use practice guidelines developed by the American College of Medical Genetics and Genomics ([www.acmg.net](http://www.acmg.net)), American College of Obstetrics and Gynecology ([www.acog.org](http://www.acog.org)), and the American Society of Reproductive Medicine ([www.asrm.org](http://www.asrm.org)) to

determine the best practices for performing genetic testing on anonymous sperm donors.

Currently, some Cryobanks perform testing on their donors for the following genetic diseases:

### **Cystic fibrosis carrier screening**

Cystic fibrosis is a chronic illness that typically involves severe lung disease and gastrointestinal problems. People with CF often have a shortened lifespan. Cystic fibrosis is inherited in an autosomal recessive manner which means that both parents have to carry mutations for CF to be at risk of having a child affected with this disorder. CF occurs most often among individuals of Caucasian ancestry; approximately 1 out of every 25 healthy Northern European or Ashkenazi Jewish individuals carries a mutation for CF. Carrier screening for cystic fibrosis is performed by analyzing the donor's DNA for common mutations or changes in the gene for CF. CF carrier screening is performed on all of CCB's sperm donor applicants, regardless of the donor's ancestry.

### **Hemoglobin evaluation**

Hemoglobin is the molecule that carries oxygen to the cells of our bodies. Hemoglobin disorders are inherited in an autosomal recessive manner. This means that a child must inherit mutations from both parents to be at risk of developing a disorder such as alpha-thalassemia, beta-thalassemia, or sickle cell anemia. A hemoglobin evaluation on our donor applicants includes a complete blood count (CBC) which evaluates the size, shape, and number of a person's blood cells. In addition, a test called a hemoglobin fractionation is performed to detect many, but not all, clinically significant differences in a person's hemoglobin.

- **Alpha-thalassemia and Beta-thalassemia**

Individuals who are affected with thalassemia generally have severe anemia and may require frequent blood transfusions and other medical interventions over the course of their lifetimes. Some types of thalassemia may result in a shortened lifespan; more severe forms may result in fetal demise or death in infancy. Alpha-thalassemia occurs most often among individuals of Asian or African ancestry. Beta-thalassemia (also known as Cooley's anemia or Mediterranean anemia) occurs most often among people of Mediterranean descent.

- **Sickle cell anemia**

Hemoglobin S is the hemoglobin that is found in abnormal amounts in a person who is affected with sickle cell anemia. Sickle cell anemia causes painful episodes of joint and bone pain and increased risks for strokes, infections, and organ

damage. Approximately 1 out of every 10 individuals of African descent carries a mutation for sickle cell anemia.

### **Chromosome analysis**

Chromosomes are the structures that carry our DNA. A chromosome analysis looks at the number and the structure of an individual's chromosomes. Typically, an analysis of human chromosomes reveals a total of 46 chromosomes. If a sperm donor applicant has an abnormal chromosome numbers or structure he would not be eligible to participate in our program.

### **Spinal muscular atrophy (SMA) carrier screening**

Spinal muscular atrophy is a progressive neuromuscular disease that results in muscle wasting and, in its most common and severe form, death due to respiratory failure before two years of age. SMA is inherited in an autosomal recessive manner which means that both parents must be carriers of mutations for SMA to be at risk of having a child affected with this disorder. Approximately 1 out of every 50 individuals in the general population carries a mutation for SMA. California Cryobank began performing carrier screening for SMA on all active sperm donors and donor applicants in August 2008.

### **Additional carrier screening performed on donors with Jewish ancestry**

The following disorders occur most often among individuals of Ashkenazi Jewish ancestry. Each of these conditions is inherited in an autosomal recessive manner which means that both parents have to carry mutations for the same condition to be at risk of having a child affected with that disorder. Carrier screening for these conditions is performed by analyzing the donor's DNA for common mutations that cause these conditions in the Ashkenazi Jewish population.

### **Bloom syndrome**

Individuals affected with Bloom syndrome have growth problems, poor immune system function, and a high rate of cancer. Most affected individuals die from cancer before 30 years of age. The carrier frequency for Bloom syndrome is approximately 1 in 100 among individuals with Ashkenazi Jewish ancestry. California Cryobank began performing carrier screening for Bloom syndrome on Jewish donors in August 2008.

### **Canavan disease**

Symptoms of Canavan disease vary from person to person and the onset of symptoms can occur at different stages of life. The most common type occurs in infancy, with death occurring in early childhood. Affected infants have mental retardation, seizures, and other neuromuscular problems. The carrier frequency for Canavan disease in the Ashkenazi Jewish population is approximately 1 in 40.

### **Familial dysautonomia**

Familial dysautonomia is a disorder of the nervous system, characterized by insensitivity to pain, episodes of vomiting and sweating, and unstable blood pressure and body temperature. In addition, people with familial dysautonomia can have learning disabilities. The average life expectancy for someone with familial dysautonomia is approximately 30 years. The carrier frequency in the Ashkenazi Jewish population is approximately 1 in 30.

### **Fanconi anemia Type C**

Symptoms of Fanconi anemia Type C include short stature, significant bone marrow problems, and heart, kidney, gastrointestinal, spinal, or limb defects. Lifespan may be shortened, as individuals with this condition have an increased risk for leukemia and other cancers. The carrier frequency in the Ashkenazi Jewish population is approximately 1 in 89.

### **Gaucher disease**

Symptoms of Gaucher disease include frequent fevers, bone pain and fractures, problems with blood clotting, anemia, seizures, and enlargement of the spleen and liver. The symptoms vary in age of onset and severity and may result in a shortened lifespan. Treatment for Gaucher disease is currently available for many affected individuals. The carrier frequency for Gaucher disease in the Ashkenazi Jewish population is approximately 1 in 12.

### **Mucopolipidosis Type IV**

Mucopolipidosis Type IV (MLIV) affects the development of the brain and nervous system beginning in the first year of life. There is no treatment for MLIV at this time. The carrier frequency for Mucopolipidosis Type IV in the Ashkenazi Jewish population is approximately 1 in 120. California Cryobank began performing carrier screening for MLIV on Jewish donors in August 2008.

### **Niemann-Pick Disease Type A/B**

Individuals with Niemann-Pick disease Type A/B have enlargement of the liver and spleen with rapid neurological deterioration and death typically occurring by age 4. The carrier frequency for Niemann-Pick disease in the Ashkenazi Jewish population is approximately 1 in 90.

### **Tay-Sachs disease (TSD)**

Tay-Sachs disease is a progressive neurological disorder that begins in infancy with the loss of developmental milestones and typically results in death by age 5. Tay-Sachs disease occurs most often among individuals with Ashkenazi Jewish or French Canadian ancestry. Approximately 1 out of every 30 individuals of Ashkenazi Jewish ancestry carries a mutation for TSD. Routine carrier screening for TSD is performed by enzyme analysis: carrier screening by DNA analysis was added in November 2013.

### **Expanded Jewish screening panel**

In November 2013, California Cryobank started performing carrier screening for the following conditions on donors with Jewish ancestry. Please see the Genetic Test Summary (GTS) for the specific donors in which you are interested to confirm which tests were performed on an individual donor.

### **Dihydrolipoamide dehydrogenase deficiency (DLD):**

Dihydrolipoamide dehydrogenase deficiency can vary widely among affected individuals but common features include recurrent vomiting and abdominal pain, seizures, decreased muscle tone, lethargy, and can be life-threatening. The carrier frequency for DLD is approximately 1 in 107 among individuals of Ashkenazi Jewish ancestry.

### **Familial hyperinsulinism, *ABCC8*-related:**

Individuals with familial hyperinsulinism produce too much insulin which can lead to lethargy, low muscle tone, and episodes of hypoglycemia. If untreated it can cause serious complications such as seizures, brain damage and can be life threatening. The carrier frequency for familial hyperinsulinism among the Ashkenazi population is approximately 1 in 68.

### **Glycogen storage disease type 1a**

Individuals with glycogen storage disease type 1a (GSD1a) may have low blood sugar, enlarged livers, seizures and kidney disease. The carrier frequency for GSD1a in the Ashkenazi population is approximately 1 in 64.

### **Joubert syndrome Type 2:**

Joubert syndrome affects many parts of the body and includes symptoms such as jerky eye movements, abnormal breathing pattern, renal and liver disease and developmental delay. The carrier frequency for Joubert syndrome type 2 is 1 in 92 among Ashkenazi individuals.

### **Maple syrup urine disease, Type A/B (MSUD):**

This condition gets its name from the distinctive sweet odor of an affected child's urine and is characterized by poor feeding, lethargy, and developmental delay. The carrier frequency is 1 in 97 among the Ashkenazi population.

### **Nemaline myopathy, *NEB*-related:**

Nemaline myopathy is a disorder that mainly affects the skeletal muscle leading to weakness and poor muscle tone. The condition typically progresses over time and can lead to respiratory failure in severe cases. The carrier frequency within the Ashkenazi population is approximately 1 in 168.

### **Usher syndrome Type 1F:**

Individuals with Usher syndrome Type 1F have profound deafness at birth and a gradual loss of vision beginning in adolescence. They also have trouble with balance which can cause delayed motor skills. The carrier frequency is 1 in 147 within the Ashkenazi population.

### **Usher syndrome Type III:**

Usher syndrome Type III leads to progressive hearing loss that typically starts in adolescence. The rate at which hearing and vision decline can vary from person to person. It has a carrier frequency of 1 in 120 in Ashkenazi individuals.

**Walker-Warburg syndrome, *FKTN*-related:**

Considered a congenital muscular dystrophy, individuals with Walker-Warburg have low muscle tone, brain abnormalities leading to global developmental delays and eye malformations. Most affected individuals do not survive past age 3. The carrier frequency among Ashkenazi Jewish individuals is 1 in 150.