



RESULTS RECIPIENT  
**SEATTLE SPERM BANK**  
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Report Date: 11/09/2021

MALE  
**DONOR 12806**  
DOB:  
Ethnicity: Northern European  
Sample Type: EDTA Blood  
Date of Collection: 11/02/2021  
Date Received: 11/04/2021  
Date Tested: 11/09/2021  
Barcode: 11004512971784  
Accession ID:  
CSLDJQW2EE3JDQ2  
Indication: Egg or sperm donor

FEMALE  
N/A

# Foresight® Carrier Screen

**POSITIVE: CARRIER**

## ABOUT THIS TEST

The **Myriad Foresight Carrier Screen** utilizes sequencing, maximizing coverage across all DNA regions tested, to help you learn about your chance to have a child with a genetic disease.

## RESULTS SUMMARY

| Risk Details                                                                                                                                                   | DONOR 12806                                                                                                           | Partner                                                                                                                                                             |
|----------------------------------------------------------------------------------------------------------------------------------------------------------------|-----------------------------------------------------------------------------------------------------------------------|---------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| Panel Information                                                                                                                                              | Foresight Carrier Screen<br>Universal Panel<br>Fundamental Plus Panel<br>Fundamental Panel<br>(175 conditions tested) | N/A                                                                                                                                                                 |
| <b>POSITIVE: CARRIER</b><br>Congenital Adrenal Hyperplasia, CYP21A2-related<br><br>Reproductive Risk: 1 in 220<br>Inheritance: Autosomal Recessive             | <b>+ CARRIER*</b><br>NM_000500.7(CYP21A2):c.844G>T(V282L) heterozygote                                                | The reproductive risk presented is based on a hypothetical pairing with a partner of the same ethnic group. Carrier testing should be considered. See "Next Steps". |
| <b>POSITIVE: CARRIER</b><br>Canavan Disease<br><br>Reproductive Risk: 1 in 630<br>Inheritance: Autosomal Recessive                                             | <b>+ CARRIER*</b><br>NM_000049.2(ASPA):c.854A>C (E285A) heterozygote                                                  | The reproductive risk presented is based on a hypothetical pairing with a partner of the same ethnic group. Carrier testing should be considered. See "Next Steps". |
| <b>POSITIVE: CARRIER</b><br>Congenital Disorder of Glycosylation Type Ia<br><br>Reproductive Risk: 1 in 630<br>Inheritance: Autosomal Recessive                | <b>+ CARRIER*</b><br>NM_000303.2(PMM2):c.422G>A (R141H) heterozygote                                                  | The reproductive risk presented is based on a hypothetical pairing with a partner of the same ethnic group. Carrier testing should be considered. See "Next Steps". |
| <b>POSITIVE: CARRIER</b><br>Glycogen Storage Disease Type Ia<br><br>Reproductive Risk: 1 in 710<br>Inheritance: Autosomal Recessive                            | <b>+ CARRIER*</b><br>NM_000151.3(G6PC1):c.247C>T (R83C) heterozygote                                                  | The reproductive risk presented is based on a hypothetical pairing with a partner of the same ethnic group. Carrier testing should be considered. See "Next Steps". |
| <b>POSITIVE: CARRIER</b><br>Hexosaminidase A Deficiency (Including Tay-Sachs Disease)<br><br>Reproductive Risk: 1 in 1,200<br>Inheritance: Autosomal Recessive | <b>+ CARRIER*</b><br>NM_000520.4(HEXA):c.1274_1277dupTATC(aka Y427Ifs*5) heterozygote                                 | The reproductive risk presented is based on a hypothetical pairing with a partner of the same ethnic group. Carrier testing should be considered. See "Next Steps". |

\*Carriers generally do not experience symptoms.

Continued on next page...

| Risk Details                                                                                                           | DONOR 12806                                                          | Partner                                                                                                                                                             |
|------------------------------------------------------------------------------------------------------------------------|----------------------------------------------------------------------|---------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| <b>POSITIVE: CARRIER</b><br>Usher Syndrome Type 3<br>Reproductive Risk: 1 in 1,600<br>Inheritance: Autosomal Recessive | <b>+ CARRIER*</b><br>NM_174878.2(CLRN1):c.144T>G (N48K) heterozygote | The reproductive risk presented is based on a hypothetical pairing with a partner of the same ethnic group. Carrier testing should be considered. See "Next Steps". |
| <b>POSITIVE: CARRIER</b><br>Familial Dysautonomia<br>Reproductive Risk: 1 in 2,000<br>Inheritance: Autosomal Recessive | <b>+ CARRIER*</b><br>NM_003640.3(ELP1):c.2204+6T>C heterozygote      | The reproductive risk presented is based on a hypothetical pairing with a partner of the same ethnic group. Carrier testing should be considered. See "Next Steps". |

\*Carriers generally do not experience symptoms.

No disease-causing mutations were detected in any other gene tested. A complete list of all conditions tested can be found on page 20.

#### CLINICAL NOTES

- None

#### NEXT STEPS

- Carrier testing should be considered for the diseases specified above for the patient's partner.
- Patients are recommended to discuss reproductive risks with their health care provider or a genetic counselor. Patients may also wish to discuss any positive results with blood relatives, as there is an increased chance that they are also carriers.

**POSITIVE: CARRIER**

# Congenital Adrenal Hyperplasia, CYP21A2-related

**Reproductive risk: 1 in 220**  
Risk before testing: 1 in 12,000

**Gene:** CYP21A2 | **Inheritance Pattern:** Autosomal Recessive

|                        |                                                                                                                                                                                                                                                 |                          |
|------------------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|--------------------------|
| <b>Patient</b>         | <b>DONOR 12806</b>                                                                                                                                                                                                                              | <b>No partner tested</b> |
| <b>Result</b>          | ⊕ Carrier                                                                                                                                                                                                                                       | N/A                      |
| <b>Variant(s)</b>      | NM_000500.7(CYP21A2):c.844G>T(V282L) heterozygote                                                                                                                                                                                               | N/A                      |
| <b>Methodology</b>     | Analysis of homologous regions (v3.2)                                                                                                                                                                                                           | N/A                      |
| <b>Interpretation</b>  | This individual is a carrier of congenital adrenal hyperplasia, CYP21A2-related. Carriers generally do not experience symptoms. NM_000500.7(CYP21A2):c.844G>T(V282L) is a non-classic congenital adrenal hyperplasia, CYP21A2-related mutation. | N/A                      |
| <b>Detection rate</b>  | 96%                                                                                                                                                                                                                                             | N/A                      |
| <b>Variants tested</b> | CYP21A2 deletion, CYP21A2 duplication, CYP21A2 triplication, G111Vfs*21, I173N, L308Ffs*6, P31L, Q319*, Q319*+CYP21A2dup, R357W, V282L, [I237N;V238E;M240K], c.293-13C>G.                                                                       | N/A                      |

## What Is Congenital Adrenal Hyperplasia, CYP21A2-Related?

Congenital adrenal hyperplasia (CAH) refers to a group of genetic disorders that affect the body's adrenal glands. The adrenal glands regulate essential functions in the body, including the production of several important hormones. CAH occurs when the adrenal glands are unable to produce these hormones properly, resulting in a hormone imbalance. CAH, CYP21A2-related is caused by mutations in the *CYP21A2* gene. The *CYP21A2* gene produces the 21-hydroxylase enzyme. Another name for this disorder is 21-hydroxylase-deficient CAH (21-OHD CAH).

When the 21-hydroxylase enzyme is missing or present at low levels, the adrenal glands are unable to produce two critical hormones, cortisol and aldosterone. The body responds to this deficiency by producing an excess of male sex hormones, called androgens. Collectively, the excess androgen production and hormone deficiencies can lead to a variety of medical problems, which vary in severity depending on the form of CAH. CAH associated with *CYP21A2* (21-OHD CAH) has two major forms: classic and non-classic.

### CLASSIC FORM

The most severe form referred to as classic 21-OHD CAH, can be further divided into two different subtypes: salt wasting and simple virilizing (non-salt wasting) types. The classic salt-wasting type is associated with near-to-complete deficiency of the 21-hydroxylase enzyme, resulting in the complete inability to produce the hormones cortisol and aldosterone. In this type, the body cannot retain enough sodium (salt) and when too much salt is lost in the urine, it may lead to dehydration, vomiting, diarrhea, poor growth, heart-rhythm abnormalities (arrhythmias), and shock (salt wasting). If not properly treated, salt wasting can lead to death in some cases.

Additionally, female newborns often have external genitals that do not clearly appear either male or female (ambiguous genitalia), whereas male newborns may present with enlarged genitals. Signs of early puberty and the exaggerated development of male characteristics (virilization) occur in both males and females with CAH. These symptoms may include: rapid growth and development

in early childhood, but shorter-than-average height in adulthood, abnormal menstruation cycles for females, excess facial hair for females, early facial-hair growth for males, severe acne, and infertility in both men and women. Male characteristics such as muscle bulk and a deep voice can occur in females and in boys (masculinization).

The simple virilizing type of CAH is associated with partial 21-hydroxylase deficiency. Unlike the salt-wasting type, individuals with this condition typically do not experience severe and life-threatening sodium-deficiency symptoms as newborns. However, the majority of female newborns with this type will have ambiguous genitalia, and both male and female children may show signs of early puberty.

#### NON-CLASSIC FORM

The non-classic type (late-onset type) is the the least-severe form of 21-OHD CAH and is caused by a mild deficiency of the 21-hydroxylase enzyme. Individuals with this type may start experiencing symptoms related to excess androgen production in childhood, adolescence, or adulthood. Both males and females may exhibit rapid growth in childhood, shorter-than-average stature in adulthood, virilization, and infertility. Additionally, girls may experience symptoms of masculinization and abnormal menstruation. However, some individuals with non-classic CAH may never know they are affected because the symptoms are so mild.

## How Common Is Congenital Adrenal Hyperplasia, CYP21A2-Related?

The incidence of 21-OHD CAH varies by type and ethnicity. The incidence for the classic form is approximately 1 in 15,000 births worldwide. The prevalence of the classic form varies from 1 in 300 for Yupik Eskimos in Alaska to 1 in 21,000 in Japanese. The non-classic form of 21-OHD CAH is much more common, with an incidence of approximately 1 in 1000 births. The prevalence of the non-classic form is much higher in some ethnicities, namely in the Ashkenazi Jewish (1 in 27), Hispanic (1 in 40), Slavic (1 in 50), and Italian (1 in 300) ethnicities. Mutations in *CYP21A2* account for about 90% of CAH cases.

## How Is Congenital Adrenal Hyperplasia, CYP21A2-Related Treated?

Currently, there is no cure for CAH. However, treatments are available to address some of the associated symptoms. Patients benefit from taking hormone-replacement medications, which work to increase levels of deficient hormones and suppress the overproduction of male hormones. Most individuals with classic CAH will need to take hormone medications for the rest of their lives. Those with the less-severe forms of CAH are sometimes able to stop taking these medications in adulthood and are typically treated with lower doses. Some individuals with non-classic CAH do not require any treatment. A multidisciplinary team of physicians, including an endocrinologist, will need to monitor the medication dosage, medication side effects, growth, and sexual development of patients who continue to receive treatment.

Newborn females with ambiguous genitalia may need surgery to correct the function and appearance of the external genitalia. Surgery, if needed, is most often performed during infancy, but can be performed later in life. Treatments provided during pregnancy may reduce the degree of virilization in female fetuses. However, because the long-term safety of prenatal treatment is unknown, these therapies are considered experimental and are not recommended by professional guidelines.

## What Is the Prognosis for an Individual with Congenital Adrenal Hyperplasia, CYP21A2-Related?

With early diagnosis and proper medication management, most individuals with 21-OHD CAH will have a normal life expectancy. Early death can occur during periods of significant sodium loss (salt crises) if medication dosage is not adequately adjusted, especially during times of illness or trauma. Problems with growth and development, ambiguous genitalia, and virilization are monitored by physicians on an ongoing basis. Females with 21-OHD CAH can become pregnant, but fertility is reduced.

## POSITIVE: CARRIER Canavan Disease

**Reproductive risk: 1 in 630**  
Risk before testing: 1 in 100,000

**Gene:** ASPA | **Inheritance Pattern:** Autosomal Recessive

| Patient        | DONOR 12806                                                                                     | No partner tested |
|----------------|-------------------------------------------------------------------------------------------------|-------------------|
| Result         | Carrier                                                                                         | N/A               |
| Variant(s)     | NM_000049.2(ASPA):c.854A>C(E285A) heterozygote                                                  | N/A               |
| Methodology    | Sequencing with copy number analysis (v3.1)                                                     | N/A               |
| Interpretation | This individual is a carrier of Canavan disease. Carriers generally do not experience symptoms. | N/A               |
| Detection rate | 98%                                                                                             | N/A               |
| Exons tested   | NM_000049:1-6.                                                                                  | N/A               |

## What Is Canavan Disease?

Canavan disease is an inherited condition that destroys the myelin sheath, the white matter that insulates nerve cells in the brain. This impairs the ability of nerve cells to communicate with other cells. Canavan disease is caused by mutations in the *ASPA* gene that result in a deficiency of an enzyme called aspartoacylase. This enzyme breaks down a material called N-acetyl-L-aspartic acid (NAA) in the brain. Without enough enzyme, the NAA builds up in the brain and destroys its white matter.

### NEONATAL/INFANTILE FORM

Neonatal/infantile Canavan disease is the most common form of Canavan disease. Affected children develop muscle weakness and developmental delay, leading to severe mental disability. Developmental delays typically begin at three to five months of age with poor muscle tone (hypotonia) which causes problems with an infant's ability to turn over, control head movements, and to sit up. The infant's head also becomes rapidly larger (macrocephaly). Over time, children with the condition may become unable to swallow and may develop sleep disturbances, seizures, and blindness.

### MILD/JUVENILE FORM

Individuals with the mild/juvenile form of Canavan disease may have mild speech or motor delays beginning in childhood. However, for some affected individuals, the delays may be so mild that they go unrecognized or so non-specific that affected individuals may be undiagnosed.

## How Common Is Canavan Disease?

The prevalence of Canavan disease in the general population is unknown. Among people of Ashkenazi Jewish descent, the prevalence is 1 in 6,400 to 1 in 13,500 individuals.



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MALE  
**DONOR 12806**  
DOB:  
Ethnicity: Northern European  
Barcode: 11004512971784

FEMALE  
N/A

## How Is Canavan Disease Treated?

At this time, there is no cure for Canavan disease. Treatment for the neonatal/infantile form of the disease focuses on keeping the affected person comfortable with proper nutrition and hydration and controlling seizures with medication. Physical therapy can be beneficial to maximize ability and to minimize contractures. Individuals with the mild/juvenile form of Canavan disease may need speech therapy or tutoring but generally, do not require special medical care.

## What Is the Prognosis for a Person with Canavan Disease?

Most people with the neonatal/infantile form of Canavan disease die in childhood, although some survive into their teens or early twenties and beyond, depending on the medical care provided. Individuals with the mild/juvenile form of the disease do not typically have a shortened lifespan.

## POSITIVE: CARRIER

# Congenital Disorder of Glycosylation Type Ia

Reproductive risk: 1 in 630

Risk before testing: 1 in 100,000

Gene: PMM2 | Inheritance Pattern: Autosomal Recessive

| Patient        | DONOR 12806                                                                                                                                                                                      | No partner tested |
|----------------|--------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|-------------------|
| Result         | Carrier                                                                                                                                                                                          | N/A               |
| Variant(s)     | NM_000303.2(PMM2):c.422G>A(R141H) heterozygote                                                                                                                                                   | N/A               |
| Methodology    | Sequencing with copy number analysis (v3.1)                                                                                                                                                      | N/A               |
| Interpretation | This individual is a carrier of congenital disorder of glycosylation type Ia. Carriers generally do not experience symptoms. In the homozygous state, R141H may be associated with fetal demise. | N/A               |
| Detection rate | >99%                                                                                                                                                                                             | N/A               |
| Exons tested   | NM_000303:1-8.                                                                                                                                                                                   | N/A               |

## What is Congenital Disorder of Glycosylation Type Ia?

Congenital disorder of glycosylation type Ia (CDG-Ia) is an inherited metabolic disorder that impairs the production of glycoproteins, which are proteins that have attached carbohydrates. In CDG-Ia, there is a defect in an enzyme called phosphomannomutase.

CDG-Ia affects many systems of the body, notably the nervous system. The disease causes developmental delay that can lead to mental disability. It can also cause seizures and stroke-like episodes. It impairs both the ability to move physically and the ability coordinate that movement. Approximately 20% of infants with the disease die within the first year of life. Most people with CDG-Ia will be wheelchair bound.

A person with CDG-Ia may show some or all of the following symptoms: a failure to grow at the normal rate; a particular type of limited vision which leads to blindness (retinitis pigmentosa); an underactive thyroid (hypothyroidism); an enlarged liver and/or liver disease; heart and kidney problems; low blood sugar; a decreased ability for blood to clot following an injury; thickened, swollen, pitted skin (peau d'orange); and bone abnormalities. Some people have many of the above symptoms while others have few.

In people with CDG-Ia, certain distinct features are apparent at birth. These may include inverted nipples, poor muscle tone, almond shaped eyes which are crossed, a large forehead, an unusual distribution of body fat, and abnormal genitals. Part of the brain called the cerebellum is often partially wasted away. Females with the disease often do not reach sexual development.

## How common is Congenital Disorder of Glycosylation Type Ia?

CDG-Ia accounts for 70% of the congenital disorders of glycosylation, which combined affect 1 in every 50,000 to 100,000 births. Cases of CDG-Ia have been reported worldwide, with about half coming from Scandinavian countries.

## How is Congenital Disorder of Glycosylation Type Ia treated?

There is no cure for CDG-Ia, however some measures may be taken to improve the lives of people affected by the disease. Parents of a young child with CDG-Ia should ensure the child gets the best possible nutrition to help with growth. Some children will require a feeding tube. Early use of occupational, physical, and speech therapy may be helpful in improving the child's long-term abilities in these areas. Surgical or non-surgical measures may correct crossed eyes and ensure better vision. Infusion of blood plasma may be necessary before a surgery to help clotting. Medications may help to control seizures. Various hormones may be useful if the person has an underactive thyroid gland. Wheelchairs and other movement aids are often useful as well.

## What is the prognosis for a person with Congenital Disorder of Glycosylation Type Ia?

Twenty percent of people with CDG-Ia die within the first year of life, often due to infection, liver problems, or heart disease. Others with CDG-Ia may live into adulthood. Most are wheelchair bound throughout their life. Some are able to speak and converse, albeit with some impairment. People with CDG-Ia are unable to live independently, but may accomplish certain tasks independently.



## POSITIVE: CARRIER

# Glycogen Storage Disease Type Ia

Reproductive risk: 1 in 710

Risk before testing: 1 in 130,000

Gene: G6PC1 | Inheritance Pattern: Autosomal Recessive

| Patient        | DONOR 12806                                                                                                      | No partner tested |
|----------------|------------------------------------------------------------------------------------------------------------------|-------------------|
| Result         | Carrier                                                                                                          | N/A               |
| Variant(s)     | NM_000151.3(G6PC1):c.247C>T(R83C) heterozygote                                                                   | N/A               |
| Methodology    | Sequencing with copy number analysis (v3.1)                                                                      | N/A               |
| Interpretation | This individual is a carrier of glycogen storage disease type Ia. Carriers generally do not experience symptoms. | N/A               |
| Detection rate | 98%                                                                                                              | N/A               |
| Exons tested   | NM_000151:1-5.                                                                                                   | N/A               |

## What is Glycogen Storage Disease Type Ia?

Glycogen Storage Disease Type I (GSDI), also called von Gierke disease, is an inherited disorder that impairs the body's ability to break down a stored form of sugar, called glycogen. There are two genes associated with GSDI, and harmful genetic changes (mutations) in *G6PC1* (formerly known as *G6PC*) cause GSDIa. In GSDIa, the body cannot maintain normal blood-sugar levels between meals, leading to low blood sugar (hypoglycemia). Additionally, glycogen builds up in organs, leading to an enlarged liver (hepatomegaly) and enlarged kidneys (nephromegaly).

Infants with GSDIa appear normal at birth, but usually begin to show symptoms when they start to sleep longer through the night. Low blood sugar can cause tiredness, irritability, and seizures. If not properly diagnosed, infants typically experience a medical crisis within the first few months of life.

Children diagnosed with GSDIa have swollen abdomens due to an enlarged liver, as well as delayed or stunted growth. Non-cancerous (benign) tumors in the liver are often seen around the time of puberty. Rarely, these can become cancerous. Changes in kidney function may occur as a patient reaches his or her twenties and may include kidney stones and a decreased ability to filter waste products. In advanced cases, dialysis and/or a kidney transplant may be needed. Other symptoms or complications that may develop include delayed puberty, thinning of the bones (osteoporosis), and a form of arthritis caused by uric acid crystals in joints (gout). Mental function is not affected by GSDIa.

## How common is Glycogen Storage Disease Type Ia?

The incidence of glycogen storage disease type I (both Ia and Ib) is 1 in 100,000 live births. Approximately 80% of glycogen storage disease type I cases are GSDIa.

## How is Glycogen Storage Disease Type Ia treated?

Individuals with GSDIa should be followed by a team of specialists who are familiar with the long-term management of glycogen storage disease to ensure appropriate monitoring and treatment for potential complications of the condition. The treatment of GSDIa involves careful monitoring of the patient's diet, both in the type of foods eaten and the frequency of meals. Individuals with GSDIa should avoid foods with sucrose (table sugar), fructose (sugar from fruits), and lactose and galactose (sugars found in milk). To maintain healthy blood sugar levels, individuals need to eat every 1-3 hours during the day and every 3-4 hours at night.

Infants and young children often need a feeding tube to tolerate frequent eating. A feeding pump may be needed at night and for emergency feedings, should their blood sugar drop to dangerously low levels. Children with GSDIa often develop problems eating and swallowing food orally and may need therapy to relearn sucking, swallowing, and sometimes speech.

Physicians recommend that individuals with GSDIa drink cornstarch mixed with water, soy formula, or soy milk. Cornstarch is digested slowly and therefore releases its glucose gradually, helping to safely extend the time between meals. Due to the restricted nature of the diet, multivitamins, calcium, and vitamin D are necessary.

## What is the prognosis for an individual with Glycogen Storage Disease Type Ia?

With careful monitoring of diet and blood sugar levels, individuals with GSDIa typically have normal growth and live into adulthood. Without close monitoring of the diet, extremely low blood sugar levels can be fatal. Even with treatment, adolescence and adults may still develop kidney complications, high blood pressure, and cancerous liver tumors.

## POSITIVE: CARRIER

# Hexosaminidase A Deficiency (Including Tay-Sachs Disease)

**Reproductive risk: 1 in 1,200**  
Risk before testing: 1 in 360,000

Gene: HEXA | Inheritance Pattern: Autosomal Recessive

| Patient        | DONOR 12806                                                                                                                                                                                                                                                                           | No partner tested |
|----------------|---------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|-------------------|
| Result         | ⊕ Carrier                                                                                                                                                                                                                                                                             | N/A               |
| Variant(s)     | NM_000520.4(HEXA):c.1274_1277dupTATC(aka Y427Ifs*5) heterozygote                                                                                                                                                                                                                      | N/A               |
| Methodology    | Sequencing with copy number analysis (v3.1)                                                                                                                                                                                                                                           | N/A               |
| Interpretation | This individual is a carrier of hexosaminidase A deficiency. Carriers generally do not experience symptoms. The Y427Ifs*5 mutation is associated with Tay-Sachs disease. Disease phenotype is dependent on, but not necessarily predicted by, the combination of mutations inherited. | N/A               |
| Detection rate | >99%                                                                                                                                                                                                                                                                                  | N/A               |
| Exons tested   | NM_000520:1-14.                                                                                                                                                                                                                                                                       | N/A               |

## What Is Hexosaminidase A Deficiency?

Hexosaminidase A Deficiency (HEX A deficiency), caused by mutations in the *HEXA* gene, is an inherited disease that causes brain and other nerve cells to die. Mutations in the *HEXA* gene cause a deficiency of the beta-hexosaminidase A enzyme which can lead to severe neurological and mental problems. This enzyme helps break down a particular fatty acid called GM2 ganglioside. Without adequate amounts of functional enzyme, GM2 ganglioside will build up in nerve cells and cause them to die.

There are several forms of HEX A deficiency, including acute infantile (Tay-Sachs disease), juvenile, chronic, and adult-onset forms.

### ACUTE INFANTILE FORM (TAY-SACHS DISEASE)

Tay-Sachs disease is the most common and severe form of HEX A deficiency. Tay-Sachs disease is a progressive condition that results in the gradual loss of movement and mental function. It is typically fatal early in childhood.

The symptoms of Tay-Sachs disease usually appear in infants between three and six months of age. Initially, infants lose the ability to turn over, sit, or crawl. They also become less attentive and develop an exaggerated startle response to loud noise. As the disease progresses and nerve cells further degenerate, infants with Tay-Sachs develop seizures, vision and hearing loss, mental disabilities, and eventually become paralyzed. Death usually occurs by the age of four.

### JUVENILE-ONSET FORM

Those with juvenile-onset HEX A deficiency typically begin to show symptoms between the ages of 2 and 10. Early signs can include a decline in verbal skills, the ability to accomplish life skills, and overall thought processes. These symptoms become progressively worse over time. The child will also develop spastic movement, seizures, and vision loss. Most children with the juvenile-onset form of HEX A deficiency will enter a vegetative state by the age of 10 to 15.

### CHRONIC FORM

Symptoms of the chronic form of the disease can begin any time from early childhood until the age of 10. While this form of HEX A deficiency can cause a variety of movement problems, the decline of verbal skills and thought processes tend to occur later than in the juvenile form.

### ADULT-ONSET FORM

Adult-onset HEX A deficiency is rare, but it is thought that symptoms begin in one's twenties or thirties. This form of the disease has the greatest variation in symptoms from person to person, even among individuals in the same family. Often its symptoms appear identical to other more common diseases, leading to misdiagnoses. These symptoms can include some or all of the following: muscle weakness, involuntary muscle twitching, speech difficulties, altered thought, or severe mental disorders like psychosis or schizophrenia. Some individuals with adult-onset HEX A deficiency may develop movement disorders but be spared mental decline until their sixties or seventies. For some, adult-onset HEX A deficiency is not fatal.

## How Common Is Hexosaminidase A Deficiency?

Tay-Sachs disease is most common among Ashkenazi Jews from Central and Eastern Europe, certain French-Canadian communities in Quebec, Amish populations in Pennsylvania, and Louisiana Cajuns. Tay-Sachs disease is found in individuals of all ethnicities, though the incidence outside of the ethnic groups mentioned above is much lower.

The incidence of Tay-Sachs disease in Ashkenazi Jewish individuals is approximately 1 in 3,600, compared to 1 in 360,000 for the non-Jewish Caucasian population. Since 1970, an organized campaign in the Jewish community to educate potential parents about Tay-Sachs and test them for mutations causing this disease has dramatically lowered the number of children affected by the condition. Because of these successful screening programs, today the majority of children born in the U.S. with Tay-Sachs disease do not have an Ashkenazi Jewish background.

The incidence of other forms of HEX A deficiency is unknown.

## How Is Hexosaminidase A Deficiency Treated?

At this time there is no cure for Tay-Sachs disease, and treatment largely focuses on ensuring the child's proper nutrition and hydration, protecting his or her ability to breathe, managing any infections, and controlling seizures with medication.

There is also no cure for these later-onset forms of HEX A deficiency. Treatment largely addresses symptoms as they arise, such as aiding mobility with mechanical aids or controlling seizures and mental disorders with medication. Because the symptoms of these forms of the disease vary widely, treatment depends on the types of symptoms and their severity.

## What Is the Prognosis for an Individual with Hexosaminidase A Deficiency?

Even with the best care available, children affected by Tay-Sachs disease usually die by the age of four. They have worsening seizures and lapse into an unresponsive vegetative state.

The prognosis for an individual with the other forms of HEX A deficiency can vary widely, depending both the age of onset and the severity of symptoms. Those with juvenile-onset or chronic HEX A deficiency typically experience severe mental decline between the ages of 2 and 10 and often reach a vegetative state between the ages of 10 to 15, with death following several years later. In more severe cases, those with juvenile-onset HEX A deficiency can die in early childhood. Those with the adult-onset form of the disease face more varied outcomes. Some may develop severe mental problems such as psychosis by age 20, while others may reach their sixties or seventies with movement difficulty but without mental problems. The lifespans of individuals with adult-onset HEX A deficiency are not well studied and can be difficult to predict. In some cases, the disease has not impacted lifespan.

## POSITIVE: CARRIER

# Usher Syndrome Type 3

**Reproductive risk: 1 in 1,600**

Risk before testing: 1 in 680,000

**Gene:** CLRN1 | **Inheritance Pattern:** Autosomal Recessive

| Patient               | DONOR 12806                                                                                           | No partner tested |
|-----------------------|-------------------------------------------------------------------------------------------------------|-------------------|
| <b>Result</b>         | 👤 Carrier                                                                                             | N/A               |
| <b>Variant(s)</b>     | NM_174878.2(CLRN1):c.144T>G(N48K) heterozygote                                                        | N/A               |
| <b>Methodology</b>    | Sequencing with copy number analysis (v3.1)                                                           | N/A               |
| <b>Interpretation</b> | This individual is a carrier of usher syndrome type 3. Carriers generally do not experience symptoms. | N/A               |
| <b>Detection rate</b> | >99%                                                                                                  | N/A               |
| <b>Exons tested</b>   | NM_174878:1-3.                                                                                        | N/A               |

## What Is Usher Syndrome Type 3?

Usher syndrome type 3, caused by mutations in the *CLRN1* gene, is an inherited disease that causes progressive hearing loss and vision impairment. The rate at which hearing and vision decline varies greatly from person to person, even among those in the same family. In some individuals, the hearing and/or vision loss can be profound, while in others it can be milder.

Individuals with Usher syndrome type 3 are born with normal hearing and most commonly develop hearing loss in their teenage years, requiring hearing aids by mid-to-late adulthood. By middle age, they are often completely deaf.

Usher syndrome type 3 also causes an eye disease known as retinitis pigmentosa. Often arising during puberty, this causes night blindness that progresses to blind spots in the late teens or early adult years. Peripheral (side) vision is often the first to be reduced and often, by mid-life, the person is legally blind.

Unlike other forms of the disease, Usher syndrome type 3 does not usually cause major problems with balance though some problems may arise later in life.

The disease does not affect intelligence, nor does it cause any other health problems.

## How Common Is Usher Syndrome Type 3?

Usher syndrome type 3 is rare, making up just 2% of all cases of Usher syndrome. Usher syndrome type 3 is more common in Finland and among Ashkenazi Jews. One study showed that in the New York City area, 0.7% of Ashkenazi Jews are carriers of an Usher syndrome type 3-causing mutation, which would mean that 1.2 in 100,000 Ashkenazi Jewish children would be affected.

## How Is Usher Syndrome Type 3 Treated?

There is no cure for Usher syndrome, but there are ways to ameliorate the vision and hearing loss it causes.



RESULTS RECIPIENT  
**SEATTLE SPERM BANK**  
Attn: Jeffrey Olliffe  
NPI: 1306838271  
Report Date: 11/09/2021

MALE  
**DONOR 12806**  
DOB:  
Ethnicity: Northern European  
Barcode: 11004512971784

FEMALE  
N/A

Individuals with the disease will learn to speak normally before their hearing declines. They can explore a range of options including cochlear implants, hearing aids, or sign language.

An individual with Usher syndrome will eventually require low-vision aids and specialized instructions on how to cope with their limited vision. They can be prone to accidental injury due to their vision loss and may need to devise systems to avoid such problems.

Specialists in both hearing loss and vision loss can guide individuals to the best options to fit their needs.

## What Is the Prognosis for an Individual with Usher Syndrome Type 3?

Usher syndrome type 3 will cause severe hearing and vision impairment by mid-life. However, it does not affect one's lifespan or intelligence.

## POSITIVE: CARRIER

# Familial Dysautonomia

**Reproductive risk: 1 in 2,000**

Risk before testing: < 1 in 1,000,000

**Gene:** ELP1 | **Inheritance Pattern:** Autosomal Recessive

| Patient        | DONOR 12806                                                                                           | No partner tested |
|----------------|-------------------------------------------------------------------------------------------------------|-------------------|
| Result         | Carrier                                                                                               | N/A               |
| Variant(s)     | NM_003640.3(ELP1):c.2204+6T>C heterozygote                                                            | N/A               |
| Methodology    | Sequencing with copy number analysis (v3.1)                                                           | N/A               |
| Interpretation | This individual is a carrier of familial dysautonomia. Carriers generally do not experience symptoms. | N/A               |
| Detection rate | >99%                                                                                                  | N/A               |
| Exons tested   | NM_003640:2-37.                                                                                       | N/A               |

## What Is Familial Dysautonomia?

Familial dysautonomia, caused by harmful genetic changes (mutations) in the *ELP1* gene (formerly known as the *IKBKAP* gene), is an inherited condition that causes nerve cells to deteriorate. It affects the autonomic nervous system, which controls involuntary actions such as breathing, tear production, blood pressure, and body temperature. It also affects the sensory nervous system, which controls senses such as the abilities to perceive taste, pressure, pain, and temperature.

Symptoms associated with the condition may first appear in infancy. Examples of these symptoms include feeding problems, poor growth and muscle tone, lack of tears, frequent lung infections, and marked fluctuations in body temperature. Children with the condition may also hold their breath for long periods of time, which may cause fainting or make their lips or skin appear blue. This behavior usually ends by the age of six. While some children may not have any delays in development, individuals usually experience some delays in milestones such as walking and speech.

As children become older they may develop symptoms including bed-wetting, vomiting, reduced sensitivity to temperatures and pain, decreased ability to taste, poor balance, abnormal curvature of the spine, easily fractured bones, and kidney and heart problems. They can also have trouble regulating blood pressure and commonly experience a sharp drop in blood pressure when they stand up, which can cause blurred vision, dizziness, or fainting. This can also lead to episodes of high blood pressure when nervous or excited.

By adulthood, individuals with familial dysautonomia may have balance problems that prevent them from walking unaided. Other common complications include sleep apnea, lung damage due to repeated infections, poor vision as optic nerves atrophy, and kidney disease. Intellect is usually not affected in individuals with this condition.

## How Common Is Familial Dysautonomia?

Familial dysautonomia is found almost exclusively in individuals of Ashkenazi Jewish descent, where it affects approximately 1 in 3,800 individuals. Roughly 1 in 31 Ashkenazi Jews is a carrier of the disease. It is extremely rare in the general population.

## How Is Familial Dysautonomia Treated?

There is no cure for the cause of familial dysautonomia. Treatment focuses on relieving its symptoms.

Infants with the condition may need to be fed thickened formula to ensure adequate nutrition and prevent them from inhaling their food. Vomiting crises are treated with IV fluids and anti-nausea medication. Recurrent pneumonia caused by inhaling food or vomit requires daily chest physiotherapy. Older children who experience low blood pressure may require elastic stockings and leg exercises to improve muscle tone and prevent blood from pooling in leg veins. Corneal injuries caused by low tear production may be treated with regular eye drops, soft contact lenses, or in rare cases, surgery. Spinal fusion surgery may be necessary to correct scoliosis. Kidney disease may require dialysis. Sleep apnea is generally treated with a machine to support breathing. Many adults with the condition require walkers or wheelchairs.

## What Is the Prognosis for an individual with Familial Dysautonomia?

The average lifespan of an individual with familial dysautonomia is shortened with 60% of individuals with the disease living to age 20.



## Methods and Limitations

**DONOR 12806 [Foresight Carrier Screen]:** Sequencing with copy number analysis, spinal muscular atrophy, analysis of homologous regions, and alpha thalassemia (HBA1/HBA2) sequencing with targeted copy number analysis (Assay(s): DTS v3.2).

### Sequencing with copy number analysis

High-throughput sequencing and read-depth-based copy number analysis are used to analyze the genes listed in the Conditions Tested section of the report. Except where otherwise noted, the region of interest (ROI) comprises the indicated coding regions and 20 non-coding bases flanking each region. In a minority of cases where genomic features (e.g., long homopolymers) compromise calling fidelity, the affected non-coding bases are excluded from the ROI. The ROI is sequenced to a minimum acceptable read depth, and the sequences are compared to a reference genomic sequence (Genome Reference Consortium Human Build 37 [GRCh37]/hg19). On average, 99% of all bases in the ROI are sequenced at a read depth that is greater than the minimum read depth. Sequence variants may not be detected in areas of lower sequence coverage. Insertions and deletions may not be detected as accurately as single-nucleotide variants. Select genes or regions for which pseudogenes or other regions of homology impede reliable variant detection may be assayed using alternate technology, or they may be excluded from the ROI. *CFTR* and *DMD* testing includes analysis for exon-level deletions and duplications with an average sensitivity of ~99%. Only exon-level deletions are assayed for other genes on the panel and such deletions are detected with a sensitivity of ≥75%. Selected founder deletions may be detected at slightly higher sensitivity. Affected exons and/or breakpoints of copy number variants are estimated from junction reads, where available, or using the positions of affected probes. Only exons known to be included in the region affected by a copy number variant are provided in the variant nomenclature. In some cases, the copy number variant may be larger or smaller than indicated. If *GJB2* is tested, large upstream deletions involving the *GJB6* and/or *CRYL1* genes that may affect the expression of *GJB2* are also analyzed.

### Spinal muscular atrophy

Targeted copy number analysis via high-throughput sequencing is used to determine the copy number of exon 7 of the *SMN1* gene. Other genetic variants may interfere with this analysis. Some individuals with two copies of *SMN1* are “silent” carriers with both *SMN1* genes on one chromosome and no copies of the gene on the other chromosome. This is more likely in individuals who have two copies of the *SMN1* gene and are positive for the g.27134T>G single-nucleotide polymorphism (SNP) (PMID: 9199562, 23788250, and 28676062), which affects the reported residual risk; Ashkenazi Jewish or Asian patients with this genotype have a high post-test likelihood of being carriers for SMA and are reported as carriers. The g.27134T>G SNP is only reported in individuals who have two copies of *SMN1*.

### Analysis of homologous regions

A combination of high-throughput sequencing, read-depth-based copy number analysis, and targeted genotyping is used to determine the number of functional gene copies and/or the presence of selected loss-of-function variants in certain genes that have homology to other genomic regions. The precise breakpoints of large deletions in these genes cannot be determined but are instead estimated from copy number analysis. Pseudogenes may interfere with this analysis, especially when many pseudogene copies are present.

If *CYP21A2* is tested, patients who have one or more additional copies of the *CYP21A2* gene and a pathogenic variant may or may not be a carrier of 21-hydroxylase deficient CAH, depending on the chromosomal location of the variants (phase). Benign *CYP21A2* gene duplications and/or triplications will only be reported in this context. Some individuals with two functional *CYP21A2* gene copies may be “silent” carriers, with two gene copies resulting from a duplication on one chromosome and a gene deletion on the other chromosome. This and other similar rare carrier states, where complementary changes exist between the chromosomes, may not be detected by the assay. Given that the true incidence of non-classic CAH is unknown, the residual carrier and reproductive risk numbers on the report are based only on the published incidence for classic CAH. However, the published prevalence of non-classic CAH is highest in individuals of Ashkenazi Jewish, Hispanic, Italian, and Yugoslav descent. Therefore, the residual and reproductive risks are likely an underestimate for CAH, especially in the aforementioned populations, as they do not account for non-classic CAH.

## Alpha thalassemia (HBA1/HBA2) sequencing with targeted copy number analysis

High-throughput sequencing and read-depth-based copy number analysis are used to identify sequence variation and functional gene copies within the region of interest (ROI) of *HBA1* and *HBA2*, which includes the listed exons plus 20 intronic flanking bases. In a minority of cases where genomic features (e.g., long homopolymers) compromise calling fidelity, the affected intronic bases are not included in the ROI. The ROI is sequenced to a minimum acceptable read depth, and the sequences are compared to a reference genomic sequence (Genome Reference Consortium Human Build 37 [GRCh37]/hg19). On average, 99% of all bases in the ROI are sequenced at a read depth that is greater than the minimum read depth. Sequence variants may not be detected in areas of lower sequence coverage. Insertions and deletions may not be detected as accurately as single-nucleotide variants. For large deletions or duplications in these genes, the precise breakpoints cannot be determined but are instead estimated from copy number analysis. This assay has been validated to detect up to two additional copies of each alpha globin gene. In rare instances where assay results suggest greater than two additional copies are present, this will be noted but the specific number of gene copies observed will not be provided.

Extensive sequence homology exists between *HBA1* and *HBA2*. This sequence homology can prevent certain variants from being localized to one gene over the other. In these instances, variant nomenclature will be provided for both genes. If follow-up testing is indicated for patients with the nomenclature provided for both genes, both *HBA1* and *HBA2* should be tested. Some individuals with four functional alpha globin gene copies may be "silent" carriers, with three gene copies resulting from triplication on one chromosome and a single gene deletion on the other chromosome. This and other similar rare carrier states, where complementary changes exist between the chromosomes, may not be detected by the assay.

## Interpretation of reported variants

The classification and interpretation of all variants identified in this assay reflects the current state of Myriad's scientific understanding at the time this report was issued. Variants are classified according to internally defined criteria, which are compatible with the ACMG Standards and Guidelines for the Interpretation of Sequence Variants (PMID: 25741868). Variants that have been determined by Myriad to be disease-causing or likely disease-causing (i.e. pathogenic or likely pathogenic) are reported. Benign variants, variants of uncertain clinical significance (VUS), and variants not directly associated with the specified disease phenotype(s) are not reported. Variant classification and interpretation may change for a variety of reasons, including but not limited to, improvements to classification techniques, availability of additional scientific information, and observation of a variant in more patients. If the classification of one or more variants identified in this patient changes, an updated report reflecting the new classification generally will not be issued. If an updated report is issued, the variants reported may change based on their current classification. This can include changes to the variants displayed in gene specific 'variants tested' sections. Healthcare providers may contact Myriad directly to request updated variant classification information specific to this test result.

## Limitations

The MWH Foresight Carrier Screen is designed to detect and report germline (constitutional) alterations. Mosaic (somatic) variation may not be detected, and if it is detected, it may not be reported. If more than one variant is detected in a gene, additional studies may be necessary to determine if those variants lie on the same chromosome or different chromosomes (phase). This test is not designed to detect sex-chromosome copy number variations. If present, sex-chromosome abnormalities may significantly reduce test sensitivity for X-linked conditions. Variant interpretation and residual and reproductive risk estimations assume a normal karyotype and may be different for individuals with abnormal karyotypes. The test does not fully address all inherited forms of intellectual disability, birth defects, or heritable diseases. Furthermore, not all forms of genetic variation are detected by this assay (i.e., duplications [except in specified genes], chromosomal rearrangements, structural abnormalities, etc.). Additional testing may be appropriate for some individuals. Pseudogenes and other regions of homology may interfere with this analysis. In an unknown number of cases, other genetic variation may interfere with variant detection. Rare carrier states where complementary changes exist between the chromosomes may not be detected by the assay. Other possible sources of diagnostic error include sample mix-up, trace contamination, bone marrow transplantation, blood transfusions, and technical or analytical errors.

Detection rates are determined using published scientific literature and/or reputable databases, when available, to estimate the fraction of disease alleles, weighted by frequency, that the methodology is predicted to be able or unable to detect. Detection rates are approximate and only account for analytical sensitivity. Certain variants that have been previously described in the literature may not be reported, if there is insufficient evidence for pathogenicity. Detection rates do not account for the disease specific rates of *de novo* variation.

This test was developed, and its performance characteristics determined by, Myriad Women's Health, Inc. It has not been cleared or approved by the US Food and Drug Administration (FDA). The FDA does not require this test to go through premarket review. This test is used for clinical purposes. It should not be regarded as investigational or for research. This laboratory is certified under the Clinical Laboratory Improvement Amendments of 1988 (CLIA) as qualified to perform high-complexity clinical testing. These results are adjunctive to the ordering physician's evaluation. CLIA Number: #05D1102604.



RESULTS RECIPIENT  
**SEATTLE SPERM BANK**  
Attn: Jeffrey Olliffe  
NPI: 1306838271  
Report Date: 11/09/2021

MALE  
**DONOR 12806**  
DOB:  
Ethnicity: Northern European  
Barcode: 11004512971784

FEMALE  
N/A

### Incidental Findings

Unless otherwise indicated, these results and interpretations are limited to the specific disease panel(s) requested by the ordering healthcare provider. In some cases, standard data analyses may identify genetic findings beyond the region(s) of interest specified by the test, and such findings may not be reported. These findings may include genomic abnormalities with major, minor, or no, clinical significance.

If you have questions or would like more information about any of the test methods or limitations, please contact (888) 268-6795.

## Resources

**GENOME CONNECT** | <http://www.genomeconnect.org>

Patients can share their reports using research registries such as Genome Connect, an online research registry building a genetics and health knowledge base. Genome Connect provides patients, physicians, and researchers an opportunity to share genetic information to support the study of the impact of genetic variation on health conditions.

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### SENIOR LABORATORY DIRECTOR

A handwritten signature in black ink that reads "Karla R. Bowles".

Karla R. Bowles, PhD, FACMG, CGMB

Report content approved by Karla Bowles, PhD, FACMG, CGMB on Nov 9, 2021

# Conditions Tested

**6-pyruvoyl-tetrahydropterin Synthase Deficiency** - Gene: PTS. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000317:1-6. **Detection Rate:** Northern European >99%.

**Adenosine Deaminase Deficiency** - Gene: ADA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000022:1-12. **Detection Rate:** Northern European 98%.

**Alpha Thalassemia, HBA1/HBA2-related** - Genes: HBA1, HBA2. Autosomal Recessive. Alpha thalassemia (HBA1/HBA2) sequencing with targeted copy number analysis. Exons: NM\_000517:1-3; NM\_000558:1-3. Variants (16): -(alpha)20.5, --BRIT, --MEDI, --MEDII, --SEA, --THAI or --FIL, -alpha3.7, -alpha4.2, HBA1+HBA2 deletion, Hb Constant Spring, Poly(A) AATAAA>AATA--, Poly(A) AATAAA>AATAAG, Poly(A) AATAAA>AATGAA, anti3.7, anti4.2, del HS-40. **Detection Rate:** Not calculated due to rarity of disease in this individual's reported ethnicity.

**Alpha-mannosidosis** - Gene: MAN2B1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000528:1-23. **Detection Rate:** Northern European >99%.

**Alpha-sarcoglycanopathy** - Gene: SGCA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000023:1-9. **Detection Rate:** Northern European >99%.

**Alstrom Syndrome** - Gene: ALMS1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_015120:1-23. **Detection Rate:** Northern European >99%.

**Andermann Syndrome** - Gene: SLC12A6. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_133647:1-25. **Detection Rate:** Northern European >99%.

**Argininemia** - Gene: ARG1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000045:1-8. **Detection Rate:** Northern European 97%.

**Argininosuccinic Aciduria** - Gene: ASL. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_001024943:1-16. **Detection Rate:** Northern European >99%.

**Aspartylglucosaminuria** - Gene: AGA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000027:1-9. **Detection Rate:** Northern European >99%.

**Ataxia with Vitamin E Deficiency** - Gene: TTPA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000370:1-5. **Detection Rate:** Northern European >99%.

**Ataxia-telangiectasia** - Gene: ATM. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000051:2-63. **Detection Rate:** Northern European 96%.

**ATP7A-related Disorders** - Gene: ATP7A. X-linked Recessive. Sequencing with copy number analysis. Exons: NM\_000052:2-23. **Detection Rate:** Northern European 90%.

**Autoimmune Polyglandular Syndrome Type 1** - Gene: AIRE. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000383:1-14. **Detection Rate:** Northern European >99%.

**Autosomal Recessive Osteopetrosis Type 1** - Gene: TCIRG1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_006019:2-20. **Detection Rate:** Northern European 96%.

**Autosomal Recessive Polycystic Kidney Disease, PKHD1-related** - Gene: PKHD1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_138694 2-67. **Detection Rate:** Northern European >99%.

**Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay** - Gene: SACS. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_014363 2-10. **Detection Rate:** Northern European 99%.

**Bardet-Biedl Syndrome, BBS1-related** - Gene: BBS1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_024649:1-17. **Detection Rate:** Northern European >99%.

**Bardet-Biedl Syndrome, BBS10-related** - Gene: BBS10. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_024685:1-2. **Detection Rate:** Northern European >99%.

**Bardet-Biedl Syndrome, BBS12-related** - Gene: BBS12. Autosomal Recessive. Sequencing with copy number analysis. Exon: NM\_152618:2. **Detection Rate:** Northern European >99%.

**Bardet-Biedl Syndrome, BBS2-related** - Gene: BBS2. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_031885:1-17. **Detection Rate:** Northern European >99%.

**BCS1L-related Disorders** - Gene: BCS1L. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_004328:3-9. **Detection Rate:** Northern European >99%.

**Beta-sarcoglycanopathy** - Gene: SGCB. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000232:1-6. **Detection Rate:** Northern European >99%.

**Biotinidase Deficiency** - Gene: BTD. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000060:1-4. **Detection Rate:** Northern European >99%.

**Bloom Syndrome** - Gene: BLM. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000057:2-22. **Detection Rate:** Northern European >99%.

**Calpainopathy** - Gene: CAPN3. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000070:1-24. **Detection Rate:** Northern European 99%.

**Canavan Disease** - Gene: ASPA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000049:1-6. **Detection Rate:** Northern European 98%.

**Carbamoylphosphate Synthetase I Deficiency** - Gene: CPS1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_001875:1-38. **Detection Rate:** Northern European >99%.

**Carnitine Palmitoyltransferase IA Deficiency** - Gene: CPT1A. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_001876:2-19. **Detection Rate:** Northern European >99%.

**Carnitine Palmitoyltransferase II Deficiency** - Gene: CPT2. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000098:1-5. **Detection Rate:** Northern European >99%.

**Cartilage-hair Hypoplasia** - Gene: RMRP. Autosomal Recessive. Sequencing with copy number analysis. Exon: NR\_003051:1. **Detection Rate:** Northern European >99%.

**Cerebrotendinous Xanthomatosis** - Gene: CYP27A1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000784:1-9. **Detection Rate:** Northern European >99%.

**Citrullinemia Type 1** - Gene: ASS1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000050:3-16. **Detection Rate:** Northern European >99%.

**CLN3-related Neuronal Ceroid Lipofuscinosis** - Gene: CLN3. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_001042432 2-16. **Detection Rate:** Northern European >99%.

**CLN5-related Neuronal Ceroid Lipofuscinosis** - Gene: CLN5. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_006493:1-4. **Detection Rate:** Northern European >99%.

**CLN8-related Neuronal Ceroid Lipofuscinosis** - Gene: CLN8. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_018941:2-3. **Detection Rate:** Northern European >99%.

**Cohen Syndrome** - Gene: VPS13B. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_017890:2-62. **Detection Rate:** Northern European 97%.

**COL4A3-related Alport Syndrome** - Gene: COL4A3. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000091:1-52. **Detection Rate:** Northern European 94%.

**COL4A4-related Alport Syndrome** - Gene: COL4A4. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000092:2-48. **Detection Rate:** Northern European >99%.

**Combined Pituitary Hormone Deficiency, PROP1-related** - Gene: PROP1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_006261:1-3. **Detection Rate:** Northern European >99%.

**Congenital Adrenal Hyperplasia, CYP11B1-related** - Gene: CYP11B1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000497:1-9. **Detection Rate:** Northern European 97%.

**Congenital Adrenal Hyperplasia, CYP21A2-related** - Gene: CYP21A2. Autosomal Recessive. Analysis of homologous regions. **Variants (13):** CYP21A2 deletion, CYP21A2 duplication, CYP21A2 triplication, G111Vfs\*21, I173N, L308Ffs\*6, P31L, Q319\*, Q319\*+CYP21A2dup, R357W, V282L, [I237N;V238E;M240K], c.293-13C>G. **Detection Rate:** Northern European 96%.

**Congenital Disorder of Glycosylation Type Ia** - Gene: PMM2. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000303:1-8. **Detection Rate:** Northern European >99%.

**Congenital Disorder of Glycosylation Type Ic** - Gene: ALG6. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_013339:2-15. **Detection Rate:** Northern European >99%.

**Congenital Disorder of Glycosylation, MPI-related** - Gene: MPI. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_002435:1-8. **Detection Rate:** Northern European >99%.

**Costeff Optic Atrophy Syndrome** - Gene: OPA3. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_025136:1-2. **Detection Rate:** Northern European >99%.

**Cystic Fibrosis** - Gene: CFTR. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000492:1-27. IVS8-5T allele analysis is only reported in the presence of the R117H mutation. **Detection Rate:** Northern European >99%.

**Cystinosis** - Gene: CTNS. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_004937:3-12. **Detection Rate:** Northern European >99%.

**D-bifunctional Protein Deficiency** - Gene: HSD17B4. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000414:1-24. **Detection Rate:** Northern European 98%.

**Delta-sarcoglycanopathy** - Gene: SGCD. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000337:2-9. **Detection Rate:** Northern European 96%.

**Dihydrolipoamide Dehydrogenase Deficiency** - Gene: DLD. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000108:1-14. **Detection Rate:** Northern European >99%.

**Dysferlinopathy** - Gene: DYSF. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_003494:1-55. **Detection Rate:** Northern European 98%.

**Dystrophinopathy (Including Duchenne/Becker Muscular Dystrophy)** - Gene: DMD. X-linked Recessive. Sequencing with copy number analysis. Exons: NM\_004006:1-79. **Detection Rate:** Northern European 99%.

**ERCC6-related Disorders** - Gene: ERCC6. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000124:2-21. **Detection Rate:** Northern European 96%.

**ERCC8-related Disorders** - Gene: ERCC8. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000082:1-12. **Detection Rate:** Northern European 97%.

**EVC-related Ellis-van Creveld Syndrome** - Gene: EVC. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_153717:1-21. **Detection Rate:** Northern European 96%.

**EVC2-related Ellis-van Creveld Syndrome** - Gene: EVC2. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_147127:1-22. **Detection Rate:** Northern European 98%.

**Fabry Disease** - Gene: GLA. X-linked Recessive. Sequencing with copy number analysis. Exons: NM\_000169:1-7. **Detection Rate:** Northern European 98%.

**Familial Dysautonomia** - Gene: ELP1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_003640:2-37. **Detection Rate:** Northern European >99%.

**Familial Hyperinsulinism, ABCC8-related** - Gene: ABCC8. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000352:1-39. **Detection Rate:** Northern European >99%.

**Familial Hyperinsulinism, KCNJ11-related** - Gene: KCNJ11. Autosomal Recessive. Sequencing with copy number analysis. Exon: NM\_000525:1. **Detection Rate:** Northern European >99%.

**Familial Mediterranean Fever** - Gene: MEFV. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000243:1-10. **Detection Rate:** Northern European >99%.

**Fanconi Anemia Complementation Group A** - Gene: FANCA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000135:1-43. **Detection Rate:** Northern European 92%.

**Fanconi Anemia, FANCC-related** - Gene: FANCC. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000136:2-15. **Detection Rate:** Northern European >99%.

**FKRP-related Disorders** - Gene: FKRP. Autosomal Recessive. Sequencing with copy number analysis. Exon: NM\_024301:4. **Detection Rate:** Northern European >99%.

**FKTN-related Disorders** - Gene: FKTN. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_001079802:3-11. **Detection Rate:** Northern European >99%.

**Free Sialic Acid Storage Disorders** - Gene: SLC17A5. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_012434:1-11. **Detection Rate:** Northern European 98%.

**Galactokinase Deficiency** - Gene: GALK1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000154:1-8. **Detection Rate:** Northern European >99%.

**Galactosemia** - Gene: GALT. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000155:1-11. **Detection Rate:** Northern European >99%.

**Gamma-sarcoglycanopathy** - Gene: SGCG. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000231:2-8. **Detection Rate:** Northern European 87%.

**Gaucher Disease** - Gene: GBA. Autosomal Recessive. Analysis of homologous regions. **Variants (10):** D409V, D448H, IVS2+1G>A, L444P, N370S, R463C, R463H, R496H, V394L, p.L29Afs\*18. **Detection Rate:** Northern European 60%.

**GJB2-related DFNB1 Nonsyndromic Hearing Loss and Deafness** - Gene: GJB2. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_004004:1-2. **Detection Rate:** Northern European >99%.

**GLB1-related Disorders** - Gene: GLB1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000404:1-16. **Detection Rate:** Northern European >99%.

**Glutaric Acidemia, GCDH-related** - Gene: GCDH. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000159:2-12. **Detection Rate:** Northern European >99%.

**Glycine Encephalopathy, AMT-related** - Gene: AMT. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000481:1-9. **Detection Rate:** Northern European >99%.

**Glycine Encephalopathy, GLDC-related** - Gene: GLDC. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000170:1-25. **Detection Rate:** Northern European 94%.

**Glycogen Storage Disease Type Ia** - Gene: G6PC1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000151:1-5. **Detection Rate:** Northern European 98%.

**Glycogen Storage Disease Type Ib** - Gene: SLC37A4. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_001164277 3-11. **Detection Rate:** Northern European >99%.

**Glycogen Storage Disease Type III** - Gene: AGL. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000642:2-34. **Detection Rate:** Northern European >99%.

**GNE Myopathy** - Gene: GNE. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_001128227:1-12. **Detection Rate:** Northern European >99%.

**GNPTAB-related Disorders** - Gene: GNPTAB. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_024312:1-21. **Detection Rate:** Northern European >99%.



**HADHA-related Disorders** - Gene: HADHA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000182:1-20. **Detection Rate:** Northern European >99%.

**Hb Beta Chain-related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease)** - Gene: HBB. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000518:1-3. **Detection Rate:** Northern European >99%.

**Hereditary Fructose Intolerance** - Gene: ALDOB. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000035:2-9. **Detection Rate:** Northern European >99%.

**Hexosaminidase A Deficiency (Including Tay-Sachs Disease)** - Gene: HEXA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000520:1-14. **Detection Rate:** Northern European >99%.

**HMG-CoA Lyase Deficiency** - Gene: HMGCL. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000191:1-9. **Detection Rate:** Northern European >99%.

**Holocarboxylase Synthetase Deficiency** - Gene: HLCS. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000411:4-12. **Detection Rate:** Northern European >99%.

**Homocystinuria, CBS-related** - Gene: CBS. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000071:3-17. **Detection Rate:** Northern European >99%.

**Hydrolethalus Syndrome** - Gene: HYL1. Autosomal Recessive. Sequencing with copy number analysis. Exon: NM\_145014:4. **Detection Rate:** Northern European >99%.

**Hypophosphatasia** - Gene: ALPL. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000478:2-12. **Detection Rate:** Northern European >99%.

**Isovaleric Acidemia** - Gene: IVD. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_002225:1-12. **Detection Rate:** Northern European >99%.

**Joubert Syndrome 2** - Gene: TMEM216. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_001173990:1-5. **Detection Rate:** Northern European >99%.

**Junctional Epidermolysis Bullosa, LAMA3-related** - Gene: LAMA3. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000227:1-38. **Detection Rate:** Northern European >99%.

**Junctional Epidermolysis Bullosa, LAMB3-related** - Gene: LAMB3. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000228:2-23. **Detection Rate:** Northern European >99%.

**Junctional Epidermolysis Bullosa, LAMC2-related** - Gene: LAMC2. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_005562:1-23. **Detection Rate:** Northern European >99%.

**Krabbe Disease** - Gene: GALC. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000153:1-17. **Detection Rate:** Northern European >99%.

**Leigh Syndrome, French-Canadian Type** - Gene: LRPPRC. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_133259:1-38. **Detection Rate:** Northern European >99%.

**Lipoid Congenital Adrenal Hyperplasia** - Gene: STAR. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000349:1-7. **Detection Rate:** Northern European >99%.

**Lysosomal Acid Lipase Deficiency** - Gene: LIPA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000235:2-10. **Detection Rate:** Northern European 98%.

**Maple Syrup Urine Disease Type Ia** - Gene: BCKDHA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000709:1-9. **Detection Rate:** Northern European >99%.

**Maple Syrup Urine Disease Type Ib** - Gene: BCKDHB. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_183050:1-10. **Detection Rate:** Northern European >99%.

**Maple Syrup Urine Disease Type II** - Gene: DBT. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_001918:1-11. **Detection Rate:** Northern European 97%.

**Medium Chain Acyl-CoA Dehydrogenase Deficiency** - Gene: ACADM. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000016:1-12. **Detection Rate:** Northern European >99%.

**Megalencephalic Leukoencephalopathy with Subcortical Cysts** - Gene: MLC1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_015166 2-12. **Detection Rate:** Northern European >99%.

**Metachromatic Leukodystrophy** - Gene: ARSA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000487:1-8. **Detection Rate:** Northern European >99%.

**Methylmalonic Acidemia, cblA Type** - Gene: MMAA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_172250:2-7. **Detection Rate:** Northern European >99%.

**Methylmalonic Acidemia, cblB Type** - Gene: MMAB. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_052845:1-9. **Detection Rate:** Northern European >99%.

**Methylmalonic Acidemia, MMUT-related** - Gene: MMUT. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000255:2-13. **Detection Rate:** Northern European >99%.

**Methylmalonic Aciduria and Homocystinuria, cblC Type** - Gene: MMACHC. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_015506:1-4. **Detection Rate:** Northern European >99%.

**MKS1-related Disorders** - Gene: MKS1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_017777:1-18. **Detection Rate:** Northern European >99%.

**Mucopolidosis III Gamma** - Gene: GNPTG. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_032520:1-11. **Detection Rate:** Northern European 98%.

**Mucopolidosis IV** - Gene: MCOLN1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_020533:1-14. **Detection Rate:** Northern European >99%.

**Mucopolysaccharidosis Type I** - Gene: IDUA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000203:1-14. **Detection Rate:** Northern European >99%.

**Mucopolysaccharidosis Type II** - Gene: IDS. X-linked Recessive. Sequencing with copy number analysis. Exons: NM\_000202:1-9. **Detection Rate:** Northern European 89%.

**Mucopolysaccharidosis Type IIIA** - Gene: SGSH. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000199:1-8. **Detection Rate:** Northern European >99%.

**Mucopolysaccharidosis Type IIIB** - Gene: NAGLU. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000263:1-6. **Detection Rate:** Northern European >99%.

**Mucopolysaccharidosis Type IIIC** - Gene: HGSNAT. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_152419:1-18. **Detection Rate:** Northern European >99%.

**Muscular Dystrophy, LAMA2-related** - Gene: LAMA2. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000426:1-43,45-65. **Detection Rate:** Northern European 98%.

**MYO7A-related Disorders** - Gene: MYO7A. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000260:2-49. **Detection Rate:** Northern European >99%.

**NEB-related Nemaline Myopathy** - Gene: NEB. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_001271208:3-80,117-183. **Detection Rate:** Northern European 92%.

**Nephrotic Syndrome, NPHS1-related** - Gene: NPHS1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_004646:1-29. **Detection Rate:** Northern European >99%.

**Nephrotic Syndrome, NPHS2-related** - Gene: NPHS2. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_014625:1-8. **Detection Rate:** Northern European >99%.

**Neuronal Ceroid Lipofuscinosis, CLN6-related** - Gene: CLN6. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_017882:1-7. **Detection Rate:** Northern European >99%.

**Niemann-Pick Disease Type C1** - Gene: NPC1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000271:1-25. **Detection Rate:** Northern European >99%.

**Niemann-Pick Disease Type C2** - Gene: NPC2. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_006432:1-5. **Detection Rate:** Northern European >99%.

**Niemann-Pick Disease, SMPD1-related** - Gene: SMPD1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000543:1-6. **Detection Rate:** Northern European >99%.

**Nijmegen Breakage Syndrome** - Gene: NBN. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_002485:1-16. **Detection Rate:** Northern European >99%.

**Ornithine Transcarbamylase Deficiency** - Gene: OTC. X-linked Recessive. Sequencing with copy number analysis. Exons: NM\_000531:1-10. **Detection Rate:** Northern European 97%.

**PCCA-related Propionic Acidemia** - Gene: PCCA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000282:1-24. **Detection Rate:** Northern European 95%.

**PCCB-related Propionic Acidemia** - Gene: PCCB. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000532:1-15. **Detection Rate:** Northern European >99%.

**PCDH15-related Disorders** - Gene: PCDH15. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_033056:2-33. **Detection Rate:** Northern European 93%.

**Pendred Syndrome** - Gene: SLC26A4. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000441:2-21. **Detection Rate:** Northern European >99%.

**Peroxisome Biogenesis Disorder Type 1** - Gene: PEX1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000466:1-24. **Detection Rate:** Northern European >99%.

**Peroxisome Biogenesis Disorder Type 3** - Gene: PEX12. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000286:1-3. **Detection Rate:** Northern European >99%.

**Peroxisome Biogenesis Disorder Type 4** - Gene: PEX6. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000287:1-17. **Detection Rate:** Northern European 97%.

**Peroxisome Biogenesis Disorder Type 5** - Gene: PEX2. Autosomal Recessive. Sequencing with copy number analysis. Exon: NM\_000318:4. **Detection Rate:** Northern European >99%.

**Peroxisome Biogenesis Disorder Type 6** - Gene: PEX10. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_153818:1-6. **Detection Rate:** Northern European >99%.

**Phenylalanine Hydroxylase Deficiency** - Gene: PAH. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000277:1-13. **Detection Rate:** Northern European >99%.

**POMGNT-related Disorders** - Gene: POMGNT1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_017739:2-22. **Detection Rate:** Northern European 96%.

**Pompe Disease** - Gene: GAA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000152:2-20. **Detection Rate:** Northern European 98%.

**PPT1-related Neuronal Ceroid Lipofuscinosis** - Gene: PPT1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000310:1-9. **Detection Rate:** Northern European >99%.

**Primary Carnitine Deficiency** - Gene: SLC22A5. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_003060:1-10. **Detection Rate:** Northern European >99%.

**Primary Hyperoxaluria Type 1** - Gene: AGXT. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000030:1-11. **Detection Rate:** Northern European >99%.

**Primary Hyperoxaluria Type 2** - Gene: GRHPR. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_012203:1-9. **Detection Rate:** Northern European >99%.

**Primary Hyperoxaluria Type 3** - Gene: HOGA1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_138413:1-7. **Detection Rate:** Northern European >99%.

**Pycnodysostosis** - Gene: CTSK. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000396:2-8. **Detection Rate:** Northern European >99%.

**Pyruvate Carboxylase Deficiency** - Gene: PC. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000920:3-22. **Detection Rate:** Northern European >99%.

**Rhizomelic Chondrodysplasia Punctata Type 1** - Gene: PEX7. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000288:1-10. **Detection Rate:** Northern European >99%.

**RTEL1-related Disorders** - Gene: RTEL1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_032957:2-35. **Detection Rate:** Northern European >99%.

**Sandhoff Disease** - Gene: HEXB. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000521:1-14. **Detection Rate:** Northern European 98%.

**Short-chain Acyl-CoA Dehydrogenase Deficiency** - Gene: ACADS. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000017:1-10. **Detection Rate:** Northern European >99%.

**Sjogren-Larsson Syndrome** - Gene: ALDH3A2. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000382:1-10. **Detection Rate:** Northern European 96%.

**SLC26A2-related Disorders** - Gene: SLC26A2. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000112:2-3. **Detection Rate:** Northern European >99%.

**Smith-Lemli-Opitz Syndrome** - Gene: DHCR7. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_001360:3-9. **Detection Rate:** Northern European >99%.

**Spastic Paraplegia Type 15** - Gene: ZFYVE26. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_015346:2-42. **Detection Rate:** Northern European >99%.

**Spinal Muscular Atrophy** - Gene: SMN1. Autosomal Recessive. Spinal muscular atrophy. Variant (1): SMN1 copy number. **Detection Rate:** Northern European 95%.

**Spondyl thoracic Dysostosis** - Gene: MESP2. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_001039958:1-2. **Detection Rate:** Northern European >99%.

**TGM1-related Autosomal Recessive Congenital Ichthyosis** - Gene: TGM1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000359 2-15. **Detection Rate:** Northern European >99%.

**TPP1-related Neuronal Ceroid Lipofuscinosis** - Gene: TPP1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000391:1-13. **Detection Rate:** Northern European >99%.

**Tyrosine Hydroxylase Deficiency** - Gene: TH. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_199292:1-14. **Detection Rate:** Northern European >99%.

**Tyrosinemia Type I** - Gene: FAH. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000137:1-14. **Detection Rate:** Northern European >99%.

**Tyrosinemia Type II** - Gene: TAT. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000353:2-12. **Detection Rate:** Northern European >99%.

**USH1C-related Disorders** - Gene: USH1C. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_005709:1-21. **Detection Rate:** Northern European >99%.

**USH2A-related Disorders** - Gene: USH2A. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_206933:2-72. **Detection Rate:** Northern European 98%.

**Usher Syndrome Type 3** - Gene: CLRN1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_174878:1-3. **Detection Rate:** Northern European >99%.



RESULTS RECIPIENT  
**SEATTLE SPERM BANK**  
Attn: Jeffrey Olliffe  
NPI: 1306838271  
Report Date: 11/09/2021

MALE  
**DONOR 12806**  
DOB:  
Ethnicity: Northern European  
Barcode: 11004512971784

FEMALE  
N/A

**Very-long-chain Acyl-CoA Dehydrogenase Deficiency** - Gene: ACADVL. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000018:1-20.

**Detection Rate:** Northern European >99%.

**Wilson Disease** - Gene: ATP7B. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000053:1-21. **Detection Rate:** Northern European >99%.

**X-linked Adrenal Hypoplasia Congenita** - Gene: NR0B1. X-linked Recessive. Sequencing with copy number analysis. Exons: NM\_000475:1-2. **Detection Rate:** Northern European 97%.

**X-linked Adrenoleukodystrophy** - Gene: ABCD1. X-linked Recessive. Sequencing with copy number analysis. Exons: NM\_000033:1-6. **Detection Rate:** Northern European 77%.

**X-linked Alport Syndrome** - Gene: COL4A5. X-linked Recessive. Sequencing with copy number analysis. Exons: NM\_000495:1-51. **Detection Rate:** Northern European 96%.

**X-linked Juvenile Retinoschisis** - Gene: RS1. X-linked Recessive. Sequencing with copy number analysis. Exons: NM\_000330:1-6. **Detection Rate:** Northern European 98%.

**X-linked Myotubular Myopathy** - Gene: MTM1. X-linked Recessive. Sequencing with copy number analysis. Exons: NM\_000252:2-15. **Detection Rate:** Northern European 96%.

**X-linked Severe Combined Immunodeficiency** - Gene: IL2RG. X-linked Recessive. Sequencing with copy number analysis. Exons: NM\_000206:1-8. **Detection Rate:** Northern European >99%.

**Xeroderma Pigmentosum Group A** - Gene: XPA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000380:1-6. **Detection Rate:** Northern European >99%.

**Xeroderma Pigmentosum Group C** - Gene: XPC. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_004628:1-16. **Detection Rate:** Northern European 97%.



## Risk Calculations

Below are the risk calculations for all conditions tested. Negative results do not rule out the possibility of being a carrier. Residual risk is an estimate of each patient's post-test likelihood of being a carrier, while the reproductive risk represents an estimated likelihood that the patients' future children could inherit each disease. These risks are inherent to all carrier-screening tests, may vary by ethnicity, are predicated on a negative family history, and are present even given a negative test result. Inaccurate reporting of ethnicity may cause errors in risk calculation. In addition, average carrier rates are estimated using incidence or prevalence data from published scientific literature and/or reputable databases, where available, and are incorporated into residual risk calculations for each population/ethnicity. When population-specific data is not available for a condition, average worldwide incidence or prevalence is used. Further, incidence and prevalence data are only collected for the specified phenotypes (which include primarily the classic or severe forms of disease) and may not include alternate or milder disease manifestations associated with the gene. Actual incidence rates, prevalence rates, and carrier rates, and therefore actual residual risks, may be higher or lower than the estimates provided. Carrier rates, incidence/prevalence, and/or residual risks are not provided for some genes with biological or heritable properties that would make these estimates inaccurate. A '+' symbol indicates a positive result. See the full clinical report for interpretation and details. The reproductive risk presented is based on a hypothetical pairing with a partner of the same ethnic group.

| Disease                                                      | DONOR 12806<br>Residual Risk                                   | Reproductive Risk |
|--------------------------------------------------------------|----------------------------------------------------------------|-------------------|
| 6-pyruvoyl-tetrahydropterin Synthase Deficiency              | < 1 in 50,000                                                  | < 1 in 1,000,000  |
| Adenosine Deaminase Deficiency                               | 1 in 22,000                                                    | < 1 in 1,000,000  |
| Alpha Thalassemia, HBA1/HBA2-related                         | Alpha globin status: aa/aa.                                    | Not calculated    |
| Alpha-mannosidosis                                           | 1 in 35,000                                                    | < 1 in 1,000,000  |
| Alpha-sarcoglycanopathy                                      | < 1 in 50,000                                                  | < 1 in 1,000,000  |
| Alstrom Syndrome                                             | < 1 in 50,000                                                  | < 1 in 1,000,000  |
| Andermann Syndrome                                           | < 1 in 50,000                                                  | < 1 in 1,000,000  |
| Argininemia                                                  | 1 in 12,000                                                    | < 1 in 1,000,000  |
| Argininosuccinic Aciduria                                    | 1 in 15,000                                                    | < 1 in 1,000,000  |
| Aspartylglucosaminuria                                       | < 1 in 50,000                                                  | < 1 in 1,000,000  |
| Ataxia with Vitamin E Deficiency                             | < 1 in 50,000                                                  | < 1 in 1,000,000  |
| Ataxia-telangiectasia                                        | 1 in 4,200                                                     | < 1 in 1,000,000  |
| ATP7A-related Disorders                                      | < 1 in 1,000,000                                               | 1 in 250,000      |
| Autoimmune Polyglandular Syndrome Type 1                     | 1 in 15,000                                                    | < 1 in 1,000,000  |
| Autosomal Recessive Osteopetrosis Type 1                     | 1 in 8,900                                                     | < 1 in 1,000,000  |
| Autosomal Recessive Polycystic Kidney Disease, PKHD1-related | 1 in 8,100                                                     | < 1 in 1,000,000  |
| Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay    | < 1 in 44,000                                                  | < 1 in 1,000,000  |
| Bardet-Biedl Syndrome, BBS1-related                          | 1 in 32,000                                                    | < 1 in 1,000,000  |
| Bardet-Biedl Syndrome, BBS10-related                         | 1 in 42,000                                                    | < 1 in 1,000,000  |
| Bardet-Biedl Syndrome, BBS12-related                         | < 1 in 50,000                                                  | < 1 in 1,000,000  |
| Bardet-Biedl Syndrome, BBS2-related                          | < 1 in 50,000                                                  | < 1 in 1,000,000  |
| BCS1L-related Disorders                                      | < 1 in 50,000                                                  | < 1 in 1,000,000  |
| Beta-sarcoglycanopathy                                       | 1 in 39,000                                                    | < 1 in 1,000,000  |
| Biotinidase Deficiency                                       | 1 in 13,000                                                    | 1 in 650,000      |
| Bloom Syndrome                                               | < 1 in 50,000                                                  | < 1 in 1,000,000  |
| Calpainopathy                                                | 1 in 13,000                                                    | < 1 in 1,000,000  |
| Canavan Disease                                              | E285A heterozygote <sup>†</sup>                                | 1 in 630          |
| Carbamoylphosphate Synthetase I Deficiency                   | < 1 in 57,000                                                  | < 1 in 1,000,000  |
| Carnitine Palmitoyltransferase IA Deficiency                 | < 1 in 50,000                                                  | < 1 in 1,000,000  |
| Carnitine Palmitoyltransferase II Deficiency                 | 1 in 25,000                                                    | < 1 in 1,000,000  |
| Cartilage-hair Hypoplasia                                    | < 1 in 50,000                                                  | < 1 in 1,000,000  |
| Cerebrotendinous Xanthomatosis                               | 1 in 11,000                                                    | < 1 in 1,000,000  |
| Citrullinemia Type 1                                         | 1 in 14,000                                                    | < 1 in 1,000,000  |
| CLN3-related Neuronal Ceroid Lipofuscinosis                  | 1 in 8,600                                                     | < 1 in 1,000,000  |
| CLN5-related Neuronal Ceroid Lipofuscinosis                  | < 1 in 50,000                                                  | < 1 in 1,000,000  |
| CLN8-related Neuronal Ceroid Lipofuscinosis                  | < 1 in 50,000                                                  | < 1 in 1,000,000  |
| Cohen Syndrome                                               | < 1 in 15,000                                                  | < 1 in 1,000,000  |
| COL4A3-related Alport Syndrome                               | 1 in 3,400                                                     | < 1 in 1,000,000  |
| COL4A4-related Alport Syndrome                               | 1 in 35,000                                                    | < 1 in 1,000,000  |
| Combined Pituitary Hormone Deficiency, PROP1-related         | 1 in 6,100                                                     | < 1 in 1,000,000  |
| Congenital Adrenal Hyperplasia, CYP11B1-related              | 1 in 8,400                                                     | < 1 in 1,000,000  |
| Congenital Adrenal Hyperplasia, CYP21A2-related              | NM_000500.7(CYP21A2):c.844G>T(V282L) heterozygote <sup>†</sup> | 1 in 220          |
| Congenital Disorder of Glycosylation Type Ia                 | R141H heterozygote <sup>†</sup>                                | 1 in 630          |
| Congenital Disorder of Glycosylation Type Ic                 | < 1 in 50,000                                                  | < 1 in 1,000,000  |

| Disease                                                                                     | DONOR 12806<br>Residual Risk | Reproductive Risk |
|---------------------------------------------------------------------------------------------|------------------------------|-------------------|
| Congenital Disorder of Glycosylation, MPI-related                                           | < 1 in 50,000                | < 1 in 1,000,000  |
| Costeff Optic Atrophy Syndrome                                                              | < 1 in 50,000                | < 1 in 1,000,000  |
| Cystic Fibrosis                                                                             | 1 in 3,000                   | 1 in 360,000      |
| Cystinosis                                                                                  | 1 in 22,000                  | < 1 in 1,000,000  |
| D-bifunctional Protein Deficiency                                                           | 1 in 9,000                   | < 1 in 1,000,000  |
| Delta-sarcoglycanopathy                                                                     | < 1 in 13,000                | < 1 in 1,000,000  |
| Dihydrolipoamide Dehydrogenase Deficiency                                                   | < 1 in 50,000                | < 1 in 1,000,000  |
| Dysferlinopathy                                                                             | 1 in 11,000                  | < 1 in 1,000,000  |
| Dystrophinopathy (Including Duchenne/Becker Muscular Dystrophy)                             | Not calculated               | Not calculated    |
| ERCC6-related Disorders                                                                     | 1 in 8,500                   | < 1 in 1,000,000  |
| ERCC8-related Disorders                                                                     | < 1 in 16,000                | < 1 in 1,000,000  |
| EVC-related Ellis-van Creveld Syndrome                                                      | 1 in 7,800                   | < 1 in 1,000,000  |
| EVC2-related Ellis-van Creveld Syndrome                                                     | 1 in 9,800                   | < 1 in 1,000,000  |
| Fabry Disease                                                                               | < 1 in 1,000,000             | 1 in 220,000      |
| Familial Dysautonomia                                                                       | c.2204+6T>C heterozygote †   | 1 in 2,000        |
| Familial Hyperinsulinism, ABCC8-related                                                     | 1 in 17,000                  | < 1 in 1,000,000  |
| Familial Hyperinsulinism, KCNJ11-related                                                    | < 1 in 50,000                | < 1 in 1,000,000  |
| Familial Mediterranean Fever                                                                | 1 in 11,000                  | < 1 in 1,000,000  |
| Fanconi Anemia Complementation Group A                                                      | 1 in 2,800                   | < 1 in 1,000,000  |
| Fanconi Anemia, FANCC-related                                                               | < 1 in 50,000                | < 1 in 1,000,000  |
| FKRP-related Disorders                                                                      | 1 in 16,000                  | < 1 in 1,000,000  |
| FKTN-related Disorders                                                                      | < 1 in 50,000                | < 1 in 1,000,000  |
| Free Sialic Acid Storage Disorders                                                          | < 1 in 30,000                | < 1 in 1,000,000  |
| Galactokinase Deficiency                                                                    | 1 in 37,000                  | < 1 in 1,000,000  |
| Galactosemia                                                                                | 1 in 8,600                   | < 1 in 1,000,000  |
| Gamma-sarcoglycanopathy                                                                     | 1 in 3,300                   | < 1 in 1,000,000  |
| Gaucher Disease                                                                             | 1 in 260                     | 1 in 110,000      |
| GJB2-related DFNB1 Nonsyndromic Hearing Loss and Deafness                                   | 1 in 2,500                   | 1 in 260,000      |
| GLB1-related Disorders                                                                      | 1 in 17,000                  | < 1 in 1,000,000  |
| Glutaric Acidemia, GCDH-related                                                             | 1 in 16,000                  | < 1 in 1,000,000  |
| Glycine Encephalopathy, AMT-related                                                         | 1 in 26,000                  | < 1 in 1,000,000  |
| Glycine Encephalopathy, GLDC-related                                                        | 1 in 2,500                   | < 1 in 1,000,000  |
| Glycogen Storage Disease Type Ia                                                            | R83C heterozygote †          | 1 in 710          |
| Glycogen Storage Disease Type Ib                                                            | 1 in 35,000                  | < 1 in 1,000,000  |
| Glycogen Storage Disease Type III                                                           | 1 in 16,000                  | < 1 in 1,000,000  |
| GNE Myopathy                                                                                | 1 in 23,000                  | < 1 in 1,000,000  |
| GNPTAB-related Disorders                                                                    | 1 in 20,000                  | < 1 in 1,000,000  |
| HADHA-related Disorders                                                                     | 1 in 20,000                  | < 1 in 1,000,000  |
| Hb Beta Chain-related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease) | 1 in 3,700                   | 1 in 560,000      |
| Hereditary Fructose Intolerance                                                             | 1 in 7,900                   | < 1 in 1,000,000  |
| Hexosaminidase A Deficiency (Including Tay-Sachs Disease)                                   | Y427Ifs*5 heterozygote †     | 1 in 1,200        |
| HMG-CoA Lyase Deficiency                                                                    | < 1 in 50,000                | < 1 in 1,000,000  |
| Holocarboxylase Synthetase Deficiency                                                       | 1 in 15,000                  | < 1 in 1,000,000  |
| Homocystinuria, CBS-related                                                                 | 1 in 9,400                   | < 1 in 1,000,000  |
| Hydroletharus Syndrome                                                                      | < 1 in 50,000                | < 1 in 1,000,000  |
| Hypophosphatasia                                                                            | 1 in 30,000                  | < 1 in 1,000,000  |
| Isovaleric Acidemia                                                                         | 1 in 32,000                  | < 1 in 1,000,000  |
| Joubert Syndrome 2                                                                          | < 1 in 50,000                | < 1 in 1,000,000  |
| Junctional Epidermolysis Bullosa, LAMA3-related                                             | < 1 in 50,000                | < 1 in 1,000,000  |
| Junctional Epidermolysis Bullosa, LAMB3-related                                             | 1 in 32,000                  | < 1 in 1,000,000  |
| Junctional Epidermolysis Bullosa, LAMC2-related                                             | < 1 in 50,000                | < 1 in 1,000,000  |
| Krabbe Disease                                                                              | 1 in 14,000                  | < 1 in 1,000,000  |
| Leigh Syndrome, French-Canadian Type                                                        | < 1 in 50,000                | < 1 in 1,000,000  |
| Lipoid Congenital Adrenal Hyperplasia                                                       | < 1 in 50,000                | < 1 in 1,000,000  |
| Lysosomal Acid Lipase Deficiency                                                            | 1 in 14,000                  | < 1 in 1,000,000  |
| Maple Syrup Urine Disease Type Ia                                                           | 1 in 39,000                  | < 1 in 1,000,000  |
| Maple Syrup Urine Disease Type Ib                                                           | 1 in 39,000                  | < 1 in 1,000,000  |
| Maple Syrup Urine Disease Type II                                                           | 1 in 16,000                  | < 1 in 1,000,000  |
| Medium Chain Acyl-CoA Dehydrogenase Deficiency                                              | 1 in 4,400                   | 1 in 790,000      |
| Megalencephalic Leukoencephalopathy with Subcortical Cysts                                  | < 1 in 50,000                | < 1 in 1,000,000  |
| Metachromatic Leukodystrophy                                                                | 1 in 16,000                  | < 1 in 1,000,000  |

| Disease                                                | DONOR 12806<br>Residual Risk                              | Reproductive Risk |
|--------------------------------------------------------|-----------------------------------------------------------|-------------------|
| Methylmalonic Acidemia, cblA Type                      | < 1 in 50,000                                             | < 1 in 1,000,000  |
| Methylmalonic Acidemia, cblB Type                      | 1 in 48,000                                               | < 1 in 1,000,000  |
| Methylmalonic Acidemia, MMUT-related                   | 1 in 26,000                                               | < 1 in 1,000,000  |
| Methylmalonic Aciduria and Homocystinuria, cblC Type   | 1 in 16,000                                               | < 1 in 1,000,000  |
| MKS1-related Disorders                                 | < 1 in 50,000                                             | < 1 in 1,000,000  |
| Mucopolipidosis III Gamma                              | < 1 in 20,000                                             | < 1 in 1,000,000  |
| Mucopolipidosis IV                                     | < 1 in 50,000                                             | < 1 in 1,000,000  |
| Mucopolysaccharidosis Type I                           | 1 in 16,000                                               | < 1 in 1,000,000  |
| Mucopolysaccharidosis Type II                          | < 1 in 1,000,000                                          | 1 in 300,000      |
| Mucopolysaccharidosis Type IIIA                        | 1 in 19,000                                               | < 1 in 1,000,000  |
| Mucopolysaccharidosis Type IIIB                        | 1 in 27,000                                               | < 1 in 1,000,000  |
| Mucopolysaccharidosis Type IIIC                        | < 1 in 50,000                                             | < 1 in 1,000,000  |
| Muscular Dystrophy, LAMA2-related                      | 1 in 5,700                                                | < 1 in 1,000,000  |
| MYO7A-related Disorders                                | 1 in 15,000                                               | < 1 in 1,000,000  |
| NEB-related Nemaline Myopathy                          | 1 in 1,200                                                | 1 in 400,000      |
| Nephrotic Syndrome, NPHS1-related                      | < 1 in 50,000                                             | < 1 in 1,000,000  |
| Nephrotic Syndrome, NPHS2-related                      | 1 in 35,000                                               | < 1 in 1,000,000  |
| Neuronal Ceroid Lipofuscinosis, CLN6-related           | 1 in 20,000                                               | < 1 in 1,000,000  |
| Niemann-Pick Disease Type C1                           | 1 in 19,000                                               | < 1 in 1,000,000  |
| Niemann-Pick Disease Type C2                           | < 1 in 50,000                                             | < 1 in 1,000,000  |
| Niemann-Pick Disease, SMPD1-related                    | 1 in 25,000                                               | < 1 in 1,000,000  |
| Nijmegen Breakage Syndrome                             | 1 in 16,000                                               | < 1 in 1,000,000  |
| Ornithine Transcarbamylase Deficiency                  | < 1 in 1,000,000                                          | 1 in 140,000      |
| PCCA-related Propionic Acidemia                        | 1 in 4,200                                                | < 1 in 1,000,000  |
| PCCB-related Propionic Acidemia                        | 1 in 22,000                                               | < 1 in 1,000,000  |
| PCDH15-related Disorders                               | 1 in 3,300                                                | < 1 in 1,000,000  |
| Pendred Syndrome                                       | 1 in 8,200                                                | < 1 in 1,000,000  |
| Peroxisome Biogenesis Disorder Type 1                  | 1 in 16,000                                               | < 1 in 1,000,000  |
| Peroxisome Biogenesis Disorder Type 3                  | 1 in 44,000                                               | < 1 in 1,000,000  |
| Peroxisome Biogenesis Disorder Type 4                  | 1 in 9,300                                                | < 1 in 1,000,000  |
| Peroxisome Biogenesis Disorder Type 5                  | < 1 in 71,000                                             | < 1 in 1,000,000  |
| Peroxisome Biogenesis Disorder Type 6                  | < 1 in 50,000                                             | < 1 in 1,000,000  |
| Phenylalanine Hydroxylase Deficiency                   | 1 in 4,800                                                | 1 in 940,000      |
| POMGNT-related Disorders                               | < 1 in 12,000                                             | < 1 in 1,000,000  |
| Pompe Disease                                          | 1 in 4,000                                                | < 1 in 1,000,000  |
| PPT1-related Neuronal Ceroid Lipofuscinosis            | 1 in 7,700                                                | < 1 in 1,000,000  |
| Primary Carnitine Deficiency                           | 1 in 11,000                                               | < 1 in 1,000,000  |
| Primary Hyperoxaluria Type 1                           | 1 in 17,000                                               | < 1 in 1,000,000  |
| Primary Hyperoxaluria Type 2                           | < 1 in 50,000                                             | < 1 in 1,000,000  |
| Primary Hyperoxaluria Type 3                           | 1 in 13,000                                               | < 1 in 1,000,000  |
| Pycnodysostosis                                        | 1 in 43,000                                               | < 1 in 1,000,000  |
| Pyruvate Carboxylase Deficiency                        | 1 in 25,000                                               | < 1 in 1,000,000  |
| Rhizomelic Chondrodysplasia Punctata Type 1            | 1 in 16,000                                               | < 1 in 1,000,000  |
| RTEL1-related Disorders                                | < 1 in 50,000                                             | < 1 in 1,000,000  |
| Sandhoff Disease                                       | 1 in 18,000                                               | < 1 in 1,000,000  |
| Short-chain Acyl-CoA Dehydrogenase Deficiency          | 1 in 11,000                                               | < 1 in 1,000,000  |
| Sjogren-Larsson Syndrome                               | < 1 in 12,000                                             | < 1 in 1,000,000  |
| SLC26A2-related Disorders                              | 1 in 16,000                                               | < 1 in 1,000,000  |
| Smith-Lemli-Opitz Syndrome                             | 1 in 9,400                                                | < 1 in 1,000,000  |
| Spastic Paraplegia Type 15                             | < 1 in 50,000                                             | < 1 in 1,000,000  |
| Spinal Muscular Atrophy                                | Negative for g.27134T>G SNP<br>SMN1: 2 copies<br>1 in 770 | 1 in 110,000      |
| Spondylothoracic Dysostosis                            | < 1 in 50,000                                             | < 1 in 1,000,000  |
| TGM1-related Autosomal Recessive Congenital Ichthyosis | 1 in 22,000                                               | < 1 in 1,000,000  |
| TPP1-related Neuronal Ceroid Lipofuscinosis            | 1 in 30,000                                               | < 1 in 1,000,000  |
| Tyrosine Hydroxylase Deficiency                        | < 1 in 50,000                                             | < 1 in 1,000,000  |
| Tyrosinemia Type I                                     | 1 in 16,000                                               | < 1 in 1,000,000  |
| Tyrosinemia Type II                                    | 1 in 25,000                                               | < 1 in 1,000,000  |
| USH1C-related Disorders                                | 1 in 30,000                                               | < 1 in 1,000,000  |
| USH2A-related Disorders                                | 1 in 4,100                                                | < 1 in 1,000,000  |
| Usher Syndrome Type 3                                  | N48K heterozygote <sup>†</sup>                            | 1 in 1,600        |



RESULTS RECIPIENT  
SEATTLE SPERM BANK  
Attn: Jeffrey Olliffe  
NPI: 1306838271  
Report Date: 11/09/2021

MALE  
DONOR 12806  
DOB:  
Ethnicity: Northern European  
Barcode: 11004512971784

FEMALE  
N/A

| Disease                                           | DONOR 12806<br>Residual Risk | Reproductive Risk |
|---------------------------------------------------|------------------------------|-------------------|
| Very-long-chain Acyl-CoA Dehydrogenase Deficiency | 1 in 18,000                  | < 1 in 1,000,000  |
| Wilson Disease                                    | 1 in 6,500                   | < 1 in 1,000,000  |
| X-linked Adrenal Hypoplasia Congenita             | < 1 in 1,000,000             | < 1 in 1,000,000  |
| X-linked Adrenoleukodystrophy                     | 1 in 90,000                  | 1 in 42,000       |
| X-linked Alport Syndrome                          | Not calculated               | Not calculated    |
| X-linked Juvenile Retinoschisis                   | < 1 in 1,000,000             | 1 in 40,000       |
| X-linked Myotubular Myopathy                      | Not calculated               | Not calculated    |
| X-linked Severe Combined Immunodeficiency         | < 1 in 1,000,000             | 1 in 200,000      |
| Xeroderma Pigmentosum Group A                     | < 1 in 50,000                | < 1 in 1,000,000  |
| Xeroderma Pigmentosum Group C                     | 1 in 7,300                   | < 1 in 1,000,000  |

DOB  
[REDACTED]

Accession  
3101522

Submitter Patient ID(s) 11805766

Sample

Source: Blood in EDTA  
Date Collected: 9/7/2024  
Date Received: 9/9/2024

Testing

Date Started: 9/9/2024  
Date Reported: 9/23/2024

Provider

Account #: [REDACTED]  
Kuan, James

Test(s) Requested

Evaluate for duplication of at least 1.1 Mb within cytogenetic band 16p12.3 (chr16:16862945-18045110)(minimum coordinates) or 3.9 Mb extending from cytogenetic band 16p13.1 to 16p12.3 (chr16: 14216341-18069975)(maximum coordinates)

Test Indication(s)

The variant above was previously identified in this individual's relative by another facility.

Result: Uncertain Clinical Significance

| Result  | Cytogenetic Band | Type        | Zygosity     | Classification                    |
|---------|------------------|-------------|--------------|-----------------------------------|
| PRESENT | 16p12.3          | Duplication | Heterozygous | Variant of Uncertain Significance |

Interpretation

The variant is PRESENT in the submitted specimen. The duplicated interval in this patient corresponds with the minimal coordinates of the duplication previously reported by another laboratory. This is a copy number change of uncertain clinical significance.

The duplicated interval involves the entire XYLT1 gene. Biallelic variants in the XYLT1 gene have been reported in association with Desbuquois dysplasia type 2 (DBQD2), also known as Baratela-Scott syndrome (BSS) (PMID: 24581741, 27030147, 27481334, 27881841). DBQD2 is an autosomal recessive skeletal dysplasia characterized by severe pre- and postnatal short stature, short extremities, monkey wrench appearance of the femora, joint laxity and dislocations, and advanced carpal and tarsal ossification. Facial features described include flat face, prominent eyes, depressed nasal bridge, and long philtrum. Most individuals have been reported with developmental delay or intellectual disability that is typically mild. Blue sclerae, cleft palate, and obesity have been reported in some individuals (PMID: 24581741, 27030147, 27481334, 27881841). A wide range of sequence variants as well as expansion of a GGC repeat in the XYLT1 promoter region, associated with hypermethylation in exon 1 of the XYLT1 gene, have been reported in association with DBQD2 (PMID: 30554721). It is unclear whether this duplication has affected the expression or function of the gene(s) in this region. This region in its entirety is not known to vary in copy number in the normal population (Database of Genomic Variants).

Recommendation

Clinical correlation between the findings in this family and the clinical phenotype as well as genetic counseling are recommended.

Methods

Genomic DNA was extracted directly from the submitted specimen or, if applicable, from cultured fibroblasts. Copy number analysis for a known familial copy number variant(s) (CNVs) was performed using targeted microarray, multiplex ligation-dependent probe amplification (MLPA), and/or quantitative PCR (qPCR) with adequate coverage for the region(s) of interest. Probe sequences and locations were based on human genome build GRCh37/UCSC hg19. For single gene or intragenic CNVs, results are reported based on the probe coordinates, the exons involved, or precise breakpoints when known. For multigenic CNVs, results are typically reported as present/absent. In circumstances where multigenic CNVs are reported based on probe coordinates, the International System for Human Cytogenetic Nomenclature (ISCN) guidelines are followed. Available evidence for variant classification may change over time and variant(s) may be reclassified according to the ACMG/ClinGen standards (PMID: 31690835), which may lead to issuing a revised report.

Disclaimer

Genetic testing using the methods applied at GeneDx is expected to be highly accurate. Normal findings do not rule out the diagnosis of a genetic disorder since some genetic

DOB

[REDACTED]

Accession

3101522

Submitter Patient ID(s) 11805766

abnormalities may be undetectable by this test. Unless otherwise indicated, the methods used cannot reliably detect deletions of 20bp to 500bp in size, or insertions of 10bp to 500bp in size. Sequencing cannot detect low-level mosaicism. Copy number assessment methods cannot reliably detect mosaicism and cannot identify balanced chromosome aberrations. Regions of certain genes have inherent sequence properties (for example: repeat, homology, or pseudogene regions, high GC content, rare polymorphisms) that yield suboptimal data, potentially impairing accuracy of the results. Inaccurate results may occur in the setting of allogeneic bone marrow/stem cell transplantation, active or chronic hematologic conditions, recent blood transfusion, suboptimal DNA quality, or in other rare circumstances. DNA extracted at other laboratories may negatively affect test performance. Rarely, incidental findings of large chromosomal rearrangements outside the gene of interest may be identified. As the ability to detect genetic variants and naming conventions can differ among laboratories, rare false negative results may occur when no positive control is provided for testing of a specific variant identified at another laboratory. In addition, the chance of an erroneous result due to laboratory errors incurred during any phase of testing cannot be completely excluded. The clinical sensitivity of this test depends in part on the patient's clinical presentation and is expected to be highest for individuals with a clearly defined phenotype and/or family history. Interpretations are made with the assumption that any clinical information provided, including family relationships, are accurate. Consultation with a genetics professional is recommended for interpretation of results. This test was developed and its performance characteristics determined by GeneDx.

This test has not been cleared or approved by the U.S. Food and Drug Administration. The FDA has determined that such clearance or approval is not necessary. The test is used for clinical purposes and should not be regarded as investigational or for research. The laboratory is certified under the Clinical Laboratory Improvement Amendments of 1988 (CLIA) as qualified to perform high-complexity clinical testing.

## References

ClinVar (<https://www.ncbi.nlm.nih.gov/clinvar/>); ClinGen Dosage Sensitivity (<https://search.clinicalgenome.org/kb/gene-dosage>); Database for Genomic Variants (DGV) (<http://dgv.tcag.ca/dgv/app/home>); Decipher (<https://www.deciphergenomics.org/>); gnomAD (<https://gnomad.broadinstitute.org/>); MitoMap (<https://www.mitomap.org/MITOMAP>); OMIM (<https://www.omim.org/>); Provean (<https://www.jcvi.org/research/provean>); The Human Gene Mutation Database (HGMD) (<https://www.hgmd.cf.ac.uk/ac/index.php>)

## Report Electronically Signed By

Evica Rajcan-Separovic Ph.D., FACMG

Associate Director, Clinical Genetics

## Resources

These clinician and family resources are of an informational nature only. While GeneDx makes every effort to ensure its accuracy, this information is not meant to endorse a particular group, be a complete list, nor serve as medical advice. GeneDx presents this opportunity as a courtesy to clinicians and families who may wish to obtain more details. All GeneDx genetic test reports include this Resource page, no matter the positive or negative result.

- **General support and advocacy**

- Child Neurology Foundation  
[www.ChildNeurologyFoundation.org](http://www.ChildNeurologyFoundation.org)  
(859) 551-4977
- Global Genes  
[www.GlobalGenes.org](http://www.GlobalGenes.org)  
(949) 248-7273
- National Organization for Rare Disorders  
[www.RareDiseases.org](http://www.RareDiseases.org)  
(617) 249-7300

- **Information exchange and connection**

- GenomeConnect: [www.GenomeConnect.org](http://www.GenomeConnect.org)
- MyGene2: [www.MyGene2.org](http://www.MyGene2.org)

**Patient Information**

Patient Name: Donor 12806

Date Of Birth: [REDACTED]

Gender: Male

Ethnicity: Caucasian/Non-Hispanic  
White

Patient ID: N/A

Medical Record #: N/A

Collection Kit: 24862202-2-C

Accession ID: N/A

Case File ID: 8914460

**Test Information**

Ordering Physician: Jeffrey Olliffe, MD

Clinic Information: Seattle Sperm Bank

Phone: (206) 588-1484

Report Date: 04/21/2023

Sample Collected: 04/11/2023

Sample Received: 04/12/2023

Sample Type: Blood

**CARRIER SCREENING REPORT**

**ABOUT THIS SCREEN:** Horizon™ is a carrier screen for specific autosomal recessive and X-linked diseases. This information can help patients learn their risk of having a child with specific genetic conditions.

**ORDER SELECTED:** The Horizon Custom panel was ordered for this patient. Males are not screened for X-linked diseases

**FINAL RESULTS SUMMARY:****Negative for 2 out of 2 diseases**

No Pathogenic variants were detected in the genes that were screened. The patient's remaining carrier risk after the negative screening results is listed for each disease/gene on the Horizon website at <https://www.natera.com/panel-option/h-all/>. Please see the following pages of this report for a comprehensive list of all conditions included on this individual's screen.

Carrier screening is not diagnostic and may not detect all possible pathogenic variants in a given gene.

**RECOMMENDATIONS**

Individuals who would like to review their Horizon report with a Natera Laboratory Genetic Counselor may schedule a telephone genetic information session by calling 650-249-9090 or visiting [naterasession.com](https://naterasession.com). Clinicians with questions may contact Natera at 650-249-9090 or email [support@natera.com](mailto:support@natera.com).

Christine M. Eng, M.D.  
Medical Director, Baylor Genetics

J. Dianne Keen-Kim, Ph.D., FACMG  
Senior Laboratory Director, Natera

Urvan Meng, Ph.D.  
Laboratory Director, Baylor Genetics

Yang Wang, Ph.D., FACMG  
Laboratory Director, Natera



**Patient Information**

Patient Name: Donor 12806

**Test Information**

Ordering Physician: Jeffrey Olliffe, MD



Date Of Birth: [REDACTED]

Case File ID: 8914460

Clinic Information: Seattle Sperm Bank

Report Date: 04/21/2023

**DISEASES SCREENED**

Below is a list of all diseases screened and the result. Certain conditions have unique patient-specific numerical values, therefore, results for those conditions are formatted differently.

**Autosomal Recessive**

**F**  
FRIEDREICH ATAXIA (FXN) **negative**

**O**  
ORNITHINE AMINOTRANSFERASE DEFICIENCY  
(OAT) **negative**

**Patient Information**

Patient Name: Donor 12806

**Test Information**

Ordering Physician: Jeffrey Olliffe, MD



Date Of Birth:

Case File ID: 8914460

Clinic Information: Seattle Sperm Bank

Report Date: 04/21/2023

**Testing Methodology, Limitations, and Comments:****Next-generation sequencing (NGS)**

Sequencing library prepared from genomic DNA isolated from a patient sample is enriched for targets of interest using standard hybridization capture protocols. If RPGR is included in the panel, exon 15 (also known as ORF15) is enriched with PCR amplification. NGS is then performed to achieve the standards of quality control metrics, including a minimum coverage of 99% of targeted regions at 20X sequencing depth. Sequencing data is aligned to human reference sequence, followed by deduplication, metric collection and variant calling (coding region +/- 20bp). Variants are then classified according to ACMG/AMP standards of interpretation using publicly available databases including but not limited to ENSEMBL, HGMD Pro, ClinGen, ClinVar, 1000G, ESP and gnomAD. Variants predicted to be pathogenic or likely pathogenic for the specified diseases are reported. It should be noted that the data interpretation is based on our current understanding of the genes and variants at the time of reporting. Putative positive sequencing variants that do not meet Internal quality standards or are within highly homologous regions are confirmed by Sanger sequencing or gene-specific long-range PCR as needed prior to reporting.

Copy Number Variant (CNV) analysis is limited to deletions involving two or more exons for all genes on the panel, in addition to specific known recurrent single-exon deletions. This method does not detect gene inversions, single-exonic and sub-exonic deletions (unless otherwise specified), and duplications of all sizes (unless otherwise specified). Additionally, this method does not define the exact breakpoints of detected CNV events. Confirmation testing for copy number variation is performed by specific PCR, Multiplex Ligation-dependent Probe Amplification (MLPA), next generation sequencing, or other methodology.

This test may not provide detection of certain variants or portions of certain genes due to local sequence characteristics, high/low genomic complexity, or the presence of closely related pseudogenes. Variants within noncoding regions (promoter, 5'UTR, 3'UTR, deep intronic regions, unless otherwise specified), small deletions or insertions larger than 25bp, low-level mosaic variants, structural variants such as inversions, and/or balanced translocations may not be detected with this technology.

**Friedreich Ataxia (FXN)**

The GAA repeat region of the FXN gene is assessed by trinucleotide PCR assay and capillary electrophoresis. Variances of +/-1 repeat for normal alleles and up to +/-3 repeats for premutation alleles may occur. For fully penetrant expanded alleles, the precise repeat size cannot be determined, therefore the approximate allele size is reported. This analysis does not detect deletions or point mutations, which comprise about four percent of the FXN pathogenic variants.

**Friedreich Ataxia Repeat Categories**

| Categories  | GAA Repeat Sizes |
|-------------|------------------|
| Normal      | <34              |
| Premutation | 34 - 65          |
| Full        | >65              |

**Variant Classification**

Only pathogenic or likely pathogenic variants are reported. Other variants including benign variants, likely benign variants, variants of uncertain significance, or inconclusive variants identified during this analysis may be reported in certain circumstances. Our laboratory's variant classification criteria are based on the ACMG and Internal guidelines and our current understanding of the specific genes. This interpretation may change over time as more information about a gene and/or variant becomes available. Natera and its lab partner(s) may reclassify variants at certain intervals but may not release updated reports without a specific request made to Natera by the ordering provider. Natera may disclose incidental findings if deemed clinically pertinent to the test performed.

**Negative Results**

A negative carrier screening result reduces the risk for a patient to be a carrier of a specific disease but does not completely rule out carrier status. Please visit <https://www.natera.com/panel-option/h-all/> for a table of carrier rates, detection rates, residual risks and promised variants/exons per gene. Carrier rates before and after testing vary by ethnicity and assume a negative family history for each disease screened and the absence of clinical symptoms in the patient. Any patient with a family history for a specific genetic disease will have a higher carrier risk prior to testing and, if the disease-causing mutation in their family is not included on the test, their carrier risk would remain unchanged. Genetic counseling is recommended for patients with a family history of genetic disease so that risk figures based on actual family history can be determined and discussed along with potential implications for reproduction. Horizon carrier screening has been developed to identify the reproductive risks for monogenic inherited conditions. Even when one or both members of a couple screen negative for pathogenic variants in a specific gene, the disease risk for their offspring is not zero. There is still a low risk for the condition in their offspring due to a number of different mechanisms that are not detected by Horizon including, but not limited to, pathogenic variant(s) in the tested gene or in a different gene not included on Horizon, pathogenic variant(s) in an upstream regulator, uniparental disomy, de novo mutation(s), or digenic or polygenic inheritance.

**Additional Comments**

These analyses generally provide highly accurate information regarding the patient's carrier status. Despite this high level of accuracy, it should be kept in mind that there are many potential sources of diagnostic error, including misidentification of samples, polymorphisms, or other rare genetic variants that interfere with analysis. Families should understand that rare diagnostic errors may occur for these reasons.



**Client/Sending Facility:**  
Seattle Sperm Bank

4915 25th Ave Ne Ste 204  
SEATTLE, WA 98105  
Ph: (206)588-1484  
Fax: (206) 466-4696 WAB-55

**LCLS Specimen Number: 306-129-1307-0**

Patient Name: **12806, DONOR**

Date of Birth: [REDACTED]

Gender: M

Patient ID:

Lab Number: DCGWA21-4378 L

Indications: DONOR

Account Number: 46857540

Ordering Physician: **J OLLIFFE**

Specimen Type: **BLOOD**

Client Reference:

Date Collected: 11/02/2021

Date Received: 11/03/2021

Date Reported: **12/08/2021**

Test: **Chromosome, Blood, Routine**

Cells Counted: 20

Cells Analyzed: 5

Cells Karyotyped: 4

Band Resolution: 500

**CYTOGENETIC RESULT: 46,XY**

**INTERPRETATION: NORMAL MALE KARYOTYPE**

Cytogenetic analysis of PHA stimulated cultures has revealed a MALE karyotype with an apparently normal GTW banding pattern in all cells observed.

This result does not exclude the possibility of subtle rearrangements below the resolution of cytogenetics or congenital anomalies due to other etiologies.



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Date Received: 11/03/2021

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Hiba Risheg, PhD., FACMG

Daniel Toweill, MD  
Medical Director  
Stuart Schwartz, PhD  
National Director of Cytogenetics

Technical component performed by Laboratory Corporation of America Holdings,  
550 17th Ave Ste 300 , Seattle , WA , 98122-5789 (206) 861-7000

Professional Component performed by LabCorp CLIA 50D0630157, 550 17th Ave Suite 200, Seattle WA 98122-5789 Medical Director, Daniel Toweill, MD

This document contains private and confidential health information **protected by state and federal law.**

Ordered Items: LP+12AC+CBC/D/Plt+UA+Rh+ABO...

Date Collected: 11/02/2021

Date Received: 11/03/2021

Date Reported: 11/05/2021

Fasting: Not Given

## LP+12AC+CBC/D/Plt+UA+Rh+ABO...

| Test                                                                                                                                                                                                                                    | Current Result and Flag | Previous Result and Date | Units       | Reference Interval |
|-----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|-------------------------|--------------------------|-------------|--------------------|
| Glucose <sup>01</sup>                                                                                                                                                                                                                   | 91                      |                          | mg/dL       | 65-99              |
| Uric Acid <sup>01</sup>                                                                                                                                                                                                                 | 5.9                     |                          | mg/dL       | 3.8-8.4            |
| Therapeutic target for gout patients: <6.0                                                                                                                                                                                              |                         |                          |             |                    |
| BUN <sup>01</sup>                                                                                                                                                                                                                       | 12                      |                          | mg/dL       | 6-20               |
| Creatinine <sup>01</sup>                                                                                                                                                                                                                | 1.14                    |                          | mg/dL       | 0.76-1.27          |
| eGFR If NonAfricn Am                                                                                                                                                                                                                    | 88                      |                          | mL/min/1.73 | >59                |
| eGFR If Africn Am                                                                                                                                                                                                                       | 101                     |                          | mL/min/1.73 | >59                |
| **In accordance with recommendations from the NKF-ASN Task force,**<br>Labcorp is in the process of updating its eGFR calculation to the<br>2021 CKD-EPI creatinine equation that estimates kidney function<br>without a race variable. |                         |                          |             |                    |
| Calcium <sup>01</sup>                                                                                                                                                                                                                   | 10.2                    |                          | mg/dL       | 8.7-10.2           |
| Protein, Total <sup>01</sup>                                                                                                                                                                                                            | 7.8                     |                          | g/dL        | 6.0-8.5            |
| Albumin <sup>01</sup>                                                                                                                                                                                                                   | 5.1                     |                          | g/dL        | 4.1-5.2            |
| Bilirubin, Total <sup>01</sup>                                                                                                                                                                                                          | 0.5                     |                          | mg/dL       | 0.0-1.2            |
| Alkaline Phosphatase <sup>01</sup>                                                                                                                                                                                                      | 67                      |                          | IU/L        | 44-121             |
| **Please note reference interval change**                                                                                                                                                                                               |                         |                          |             |                    |
| LDH <sup>01</sup>                                                                                                                                                                                                                       | 151                     |                          | IU/L        | 121-224            |
| AST (SGOT) <sup>01</sup>                                                                                                                                                                                                                | 24                      |                          | IU/L        | 0-40               |
| ALT (SGPT) <sup>01</sup>                                                                                                                                                                                                                | 19                      |                          | IU/L        | 0-44               |
| ▲ Cholesterol, Total <sup>01</sup>                                                                                                                                                                                                      | 255 High                |                          | mg/dL       | 100-199            |
| ▲ Triglycerides <sup>01</sup>                                                                                                                                                                                                           | 184 High                |                          | mg/dL       | 0-149              |
| HDL Cholesterol <sup>01</sup>                                                                                                                                                                                                           | 44                      |                          | mg/dL       | >39                |
| ▲ LDL Chol Calc (NIH)                                                                                                                                                                                                                   | 177 High                |                          | mg/dL       | 0-99               |
| ▲ LDL/HDL Ratio                                                                                                                                                                                                                         | 4.0 High                |                          | ratio       | 0.0-3.6            |
| Please Note: <sup>01</sup>                                                                                                                                                                                                              |                         |                          |             |                    |

## LDL /HDL Ratio

|              | Men | Women |
|--------------|-----|-------|
| 1/2 Avg.Risk | 1.0 | 1.5   |
| Avg.Risk     | 3.6 | 3.2   |
| 2X Avg.Risk  | 6.2 | 5.0   |
| 3X Avg.Risk  | 8.0 | 6.1   |

|                                        |                                                                           |   |           |
|----------------------------------------|---------------------------------------------------------------------------|---|-----------|
| Hgb Fractionation by CE: <sup>02</sup> |                                                                           |   |           |
| Hgb F <sup>02</sup>                    | 0.3                                                                       | % | 0.0-2.0   |
| Hgb A <sup>02</sup>                    | 97.1                                                                      | % | 96.4-98.8 |
| Hgb A2 <sup>02</sup>                   | 2.6                                                                       | % | 1.8-3.2   |
| Hgb S <sup>02</sup>                    | 0.0                                                                       | % | 0.0       |
| Interpretation: <sup>02</sup>          | Normal hemoglobin present; no hemoglobin variant or thalassemia observed. |   |           |
| ABO Grouping <sup>01</sup>             | A                                                                         |   |           |

## LP+12AC+CBC/D/Plt+UA+Rh+ABO... (Cont.)

Rh Factor<sup>01</sup>

Positive

Please note: Prior records for this patient's ABO / Rh type are not available for additional verification.

|                                          |                                              |          |                |
|------------------------------------------|----------------------------------------------|----------|----------------|
| CBC, Platelet Ct, and Diff <sup>01</sup> |                                              |          |                |
| WBC <sup>01</sup>                        | 5.4                                          | x10E3/uL | 3.4-10.8       |
| RBC <sup>01</sup>                        | 4.83                                         | x10E6/uL | 4.14-5.80      |
| Hemoglobin <sup>01</sup>                 | 14.0                                         | g/dL     | 13.0-17.7      |
| Hematocrit <sup>01</sup>                 | 40.9                                         | %        | 37.5-51.0      |
| MCV <sup>01</sup>                        | 85                                           | fL       | 79-97          |
| MCH <sup>01</sup>                        | 29.0                                         | pg       | 26.6-33.0      |
| MCHC <sup>01</sup>                       | 34.2                                         | g/dL     | 31.5-35.7      |
| RDW <sup>01</sup>                        | 13.3                                         | %        | 11.6-15.4      |
| Platelets <sup>01</sup>                  | 274                                          | x10E3/uL | 150-450        |
| Neutrophils <sup>01</sup>                | 54                                           | %        | Not Estab.     |
| Lymphs <sup>01</sup>                     | 37                                           | %        | Not Estab.     |
| Monocytes <sup>01</sup>                  | 7                                            | %        | Not Estab.     |
| Eos <sup>01</sup>                        | 1                                            | %        | Not Estab.     |
| Basos <sup>01</sup>                      | 1                                            | %        | Not Estab.     |
| Neutrophils (Absolute) <sup>01</sup>     | 2.9                                          | x10E3/uL | 1.4-7.0        |
| Lymphs (Absolute) <sup>01</sup>          | 2.0                                          | x10E3/uL | 0.7-3.1        |
| Monocytes(Absolute) <sup>01</sup>        | 0.4                                          | x10E3/uL | 0.1-0.9        |
| Eos (Absolute) <sup>01</sup>             | 0.1                                          | x10E3/uL | 0.0-0.4        |
| Baso (Absolute) <sup>01</sup>            | 0.1                                          | x10E3/uL | 0.0-0.2        |
| Immature Granulocytes <sup>01</sup>      | 0                                            | %        | Not Estab.     |
| Immature Grans (Abs) <sup>01</sup>       | 0.0                                          | x10E3/uL | 0.0-0.1        |
| Urinalysis Gross Exam <sup>01</sup>      |                                              |          |                |
| Specific Gravity <sup>01</sup>           | 1.015                                        |          | 1.005-1.030    |
| pH <sup>01</sup>                         | 6.0                                          |          | 5.0-7.5        |
| Urine-Color <sup>01</sup>                | Yellow                                       |          | Yellow         |
| Appearance <sup>01</sup>                 | Clear                                        |          | Clear          |
| WBC Esterase <sup>01</sup>               | Negative                                     |          | Negative       |
| Protein <sup>01</sup>                    | Negative                                     |          | Negative/Trace |
| Glucose <sup>01</sup>                    | Negative                                     |          | Negative       |
| Ketones <sup>01</sup>                    | Negative                                     |          | Negative       |
| Occult Blood <sup>01</sup>               | Negative                                     |          | Negative       |
| Bilirubin <sup>01</sup>                  | Negative                                     |          | Negative       |
| Urobilinogen,Semi-Qn <sup>01</sup>       | 0.2                                          | mg/dL    | 0.2-1.0        |
| Nitrite, Urine <sup>01</sup>             | Negative                                     |          | Negative       |
| Microscopic Examination <sup>01</sup>    | Microscopic not indicated and not performed. |          |                |