Reproductive Technologies, Inc.
THE SPERM BANK OF CALIFORNIA
2115 MILVIA STREET, BERKELEY 94704 PHONE 510.841.1858 www.thespermbankofca.org A 501(C)(3) CORPORATION

Acknowledgement of Positive Carrier Screening Results: Donor 5926

I, the undersigned recipient, understand that this donor has tested **POSITIVE** as a carrier for the following condition(s). More information regarding these conditions can be found in his Expanded Carrier Screening results.

- **CFTR-related conditions (CFTR)**
- **CLRN1-related conditions (CLRN1)**
- **Neuronal ceroid lipofuscinosis type 6 (CLN6)**
- **Primary carnitine deficiency (SLC22A5)**

I intend to use sperm samples from this donor for insemination or other assisted conception procedure(s).

I acknowledge that The Sperm Bank of California (TSBC) has made the donor's genetic testing results available to me and my medical providers, and that I have reviewed these results. I understand that TSBC **strongly recommends** that I review these genetic testing results with a Genetic Counselor and my medical providers. I understand that TSBC can refer me to genetic counseling services if desired.

I understand that recipient testing is strongly recommended when a donor has positive carrier screening results and that such testing can reduce but not eliminate risks.

I acknowledge that I personally assume all risks associated with use of semen samples provided by a donor who has tested **POSITIVE as a carrier for CFTR-related conditions (CFTR), CLRN1-related conditions (CLRN1), Neuronal ceroid lipofuscinosis type 6 (CLN6), Primary carnitine deficiency (SLC22A5).**

On behalf of myself and my spouse, heirs, representatives, I hereby release and forever hold harmless TSBC and its current and former officers, directors, employees, attorneys, insurers, consultants, agents, and representatives (collectively "Releases") from any liability or responsibility whatsoever for any and all outcomes, and hereby release and forever discharge Releases from any and all actions, causes of action, demands, damages, losses, liabilities, suits, expenses, including attorneys' fees and costs, of whatever character, in law or in equity, whether currently known, suspected, unknown or unsuspected, matured or unmatured, arising out of my use of sperm donated by a donor who has tested **POSITIVE as a carrier for CFTR-related conditions (CFTR), CLRN1-related conditions (CLRN1), Neuronal ceroid lipofuscinosis type 6 (CLN6), Primary carnitine deficiency (SLC22A5). This release involves the waiver of all rights and benefits that I may have under California Civil Code section 1542, which states: "A general release does not extend to claims that the creditor or releasing party does not know or suspect to exist in his or her favor at the time of executing the release and that, if known by him or her, would have materially affected his or her settlement with the debtor or released party."**

Please select one of the following:

- ☐ I have been tested for the above named condition(s) and/or I plan to be tested prior to using the samples.
- ☐ I understand that TSBC **strongly recommends** that I discuss these results with a Genetic Counselor and my medical providers and consider testing for the above named condition(s). At this time I have **declined** testing and/or **do not anticipate being tested**.

I understand that if I transfer my vials (or embryos if applicable) to any other person, including my spouse, that TSBC requires that person (1) register with TSBC and (2) complete an **Acknowledgement of Positive Carrier Screening Results**.

I understand that any and all questions as to the legal interpretation, validity or any other aspect of this agreement shall be determined by the laws of the State of California, regardless of the location or residence of any of the parties.

Recipient's signature	Recipient's printed name	Date

Reproductive Technologies, Inc.
THE SPERM BANK OF CALIFORNIA
2115 Milvia Street, Berkeley Ca 94704 Phone 510.841.1858 Fax: 510.841.0332 Email: staff@tsbca.org

GENETIC TESTING: POSITIVE CARRIER STATUS

This donor tested **POSITIVE** as a carrier for one or more autosomal recessive conditions as described on the prior page and in the attached genetic testing results.

What does it mean to be a carrier?

All people carry genetic mutations in their DNA. Genetic testing can help to identify some, but not all, of these mutations. While this donor carries a mutation for one or more recessively inherited condition(s), **offspring from this donor are not expected to be at risk of developing these condition(s) unless the recipient (or egg provider if different from the recipient) also carries a genetic mutation for the same condition(s).** For this reason, we strongly encourage you to discuss carrier screening for yourself (or your egg provider) with your physician and a genetic counselor. Genetic testing can reduce but not eliminate risks.

What are my next steps?

1. **Download the genetic test results and review with your medical providers**
   We **strongly recommend** that you discuss this donor's genetic test results with your physician **PRIOR TO SCHEDULING A SHIPMENT OR PICK-UP**, to confirm the donor is suitable for your use. Vials retrieved from the building cannot be exchanged or refunded. The donor's genetic test results are available for free download on the donor's page at https://www.thespermbankofca.org/donor-catalog.

2. **We recommend scheduling a genetic counseling session.**
   A genetic counselor can explain the results in detail including the inheritance pattern, potential risks to your children, and the available testing options that you may want to consider for yourself (or your egg provider). Phone or in person consultations are available for a fee with TSBC's Genetic Counselors at San Francisco Genetic Counseling (https://www.sfgenetics.org/) or you can locate a genetic counselor at www.findageneticcounselor.com.

3. **Complete and return the Acknowledgement of Positive Carrier Screening Results**
   TSBC requires that all recipients selecting this donor complete this acknowledgement form **PRIOR TO SCHEDULING A SHIPMENT OR PICK-UP**. Completing this form documents that you have been informed about this donor's genetic test results and that you are aware of TSBC's recommendation to discuss the genetic test results with your medical providers as noted above.

DATE: 5/26/26

Reproductive Technologies, Inc.

THE SPERM BANK OF CALIFORNIA

EXPANDED CARRIER SCREENING RESULTS DONOR 5926

Expanded carrier screening for 719 autosomal recessive conditions was completed by Labcorp and reported on 5/22/26. The results were **POSITIVE for CFTR-related conditions (CFTR), CLRN1-related conditions (CLRN1), Neuronal ceroid lipofuscinosis type 6 (CLN6), Primary carnitine deficiency (SLC22A5).** Donor 5926 is a carrier for these conditions. **It is strongly recommended that recipients who use this donor's sperm undergo carrier screening for these specific conditions.** Testing was negative for the remainder of genes screened. Please refer to the donor's Labcorp expanded carrier test report for more information on the testing completed and the donors test results.

The specific mutation in CFTR for CFTR-related conditions is predicted to be a variant that can cause a range of symptoms when inherited in combination with another pathogenic variant. The donor was only found to have one pathogenic variant in CFTR. Labcorp states, "Additionally, individuals with a single disease-causing CFTR variant (heterozygous carriers) may have an approximately 4-10 fold increased risk for chronic pancreatitis, although the absolute risk of pancreatitis remains low (less than 1 in 100). Due to this potential increased risk for chronic pancreatitis, heterozygous carriers may consider follow-up with a medical provider."

Disease	Result	Residual risk to be a carrier (based on Pan-ethnic ancestry)
CFTR-related conditions (CFTR)	POSITIVE	n/a
CLRN1-related conditions (CLRN1)	POSITIVE	n/a
Neuronal ceroid lipofuscinosis type 6 (CLN6)	POSITIVE	n/a
Primary carnitine deficiency (SLC22A5)	POSITIVE	n/a
Spinal Muscular Atrophy (SMN1)	Negative: 2 copies exon 7 c.*3+80T>G variant not detected	1 in 880
HBB Hemoglobinopathies (HBB)	Negative	1 in 4,800
Alpha-Thalassemia (HBA1/ HBA2)	Negative	Reduced

Genetic screening tests can significantly reduce, but never completely eliminate, the chance that a person is a carrier for a particular disorder.

Please also see the Health Problems List for a summary of the information that this donor has provided to us regarding personal and family medical history.

Sincerely,
Casandra Pacheco
Certified Genetic Counselor
Arbor Genetic Counseling

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LABCORP CARRIER SCREEN

Patient name:	5926 Donor	**Sample type:**	Saliva	**Report date:**	22-MAY-2026
DOB:		**Sample collection date:**	24-APR-2026	**Invitae #:**	RQ8090210
Sex assigned at birth:	Male	**Sample accession date:**	04-MAY-2026	**Clinical team:**	L BONNER
Gender:		**Specimen ID:**	12263100050		
Patient ID (MRN):					

Reason for testing

Carrier screening

Test performed

Sequence analysis and deletion/duplication testing of the 719 genes listed in the Genes Analyzed section.

- Labcorp 700 PLUS Carrier Panel (No X-Linked)

RESULT: POSITIVE

This carrier test evaluated 719 gene(s) for genetic changes (variants) that are associated with an increased risk of having a child with a genetic condition. Knowledge of carrier status for one of these conditions may provide information that can be used to assist with family planning and/or preparation. Carrier screening is not intended for diagnostic purposes. To identify a potential genetic basis for a condition in the individual being tested, diagnostic testing for the gene(s) of interest is recommended.

This test shows the presence of clinically significant genetic change(s) in this individual in the gene(s) indicated in this report. No other clinically significant changes were identified in the remaining genes evaluated with this test.

RESULTS	GENE	VARIANT(S)	INHERITANCE	PARTNER TESTING RECOMMENDED
Carrier: CFTR-related conditions	CFTR	c.1210-34TG[12]T[5] (Intronic) (5T/12TG) PERSONAL RISK ▲	Autosomal recessive	Yes
Carrier: CLRN1-related conditions	CLRN1	c.149_152delinsTGTCCAAT (p.Ser50Leufs*12)	Autosomal recessive	Yes
Carrier: Neuronal ceroid lipofuscinosis type 6	CLN6	c.13C>T (p.Arg5Trp)	Autosomal recessive	Yes
Carrier: Primary carnitine deficiency	SLC22A5	c.761G>A (p.Arg254Gln)	Autosomal recessive	Yes

▲ This result may impact this person's health. See Clinical summary on following pages for more information.

Results to note

- SMN1: Negative result. SMN1: 2 copies; c.*3+80T>G not detected.

Laboratory Director: Jeana DaRe, PhD, FACMG
Labcorp Genetics 1400 16th Street. San Francisco. CA 94103 · clientservices@invitae.com · 800.436.3037

Patient name: 5926 Donor **DOB:**
Invitae #: RQ8090210

Next steps

- Even for genes that have a negative test result, there is always a small risk that an individual could still be a carrier. This is called "residual risk". See the Carrier Detection Rates and Residual Risks document.
- Discussion with a physician and/or genetic counselor is recommended to further review the implications of this test result and to understand these results in the context of any family history of a genetic condition.
- All patients, regardless of result, may wish to consider additional screening for hemoglobinopathies by complete blood count (CBC) and hemoglobin electrophoresis, if this has not already been completed.
- See the table in this report for recommendations regarding testing of this individual's reproductive partner.
- Genetic counseling is available to discuss the potential clinical and/or reproductive implications of this result, as well as recommendations for testing family members and, when applicable, this individual's partner. To access Labcorp Genetic Counselors please visit **https://womenshealth.labcorp.com/genetic-counseling** or call (855) GC-CALLS (855-422-2557).

Patient name: 5926 Donor **DOB:**
Invitae #: RQ8090210

Clinical summary

RESULT: CARRIER

CFTR-related conditions

A single Pathogenic variant, c.1210-34TG[12]T[5] (Intronic), was identified in CFTR. This variant has unique interpretation considerations. See "What are CFTR-related conditions?" and Variant details for additional information.

What are CFTR-related conditions?

The c.1210-34TG[12]T[5] cystic fibrosis (CF) variant was identified in this individual. There are multiple forms of the 5T variant, which are classified by the number of TG repeats. Each form of the 5T variant is associated with a different degree of risk for CFTR-related symptoms when inherited in combination with a pathogenic variant from the other parent, ranging from a healthy individual to congenital absence of the vas deferens (CAVD) in males to an individual with mild/atypical CF. The combination of the c.1210-34TG[12]T[5] variant with a severe pathogenic CFTR variant from the other parent is associated with symptoms in the majority of individuals; however, most individuals who are homozygous for the c.1210-34TG[12]T[5] variant are asymptomatic (see Variant details section).

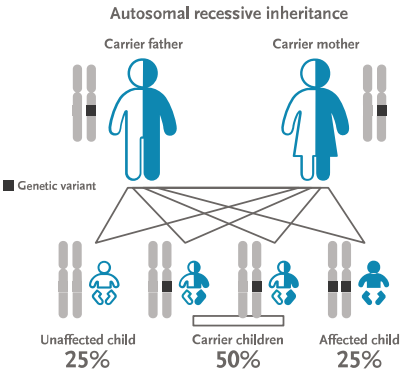
CFTR-related conditions encompass a spectrum of disorders that typically impact the respiratory and/or digestive systems, and cause male infertility. Cystic fibrosis (CF) is typically a childhood-onset disease in which abnormally thick mucus production can cause a variety of symptoms including recurrent respiratory infections and progressive lung disease, as well as nutritional deficiencies and poor growth due to deficiency of enzymes produced by the pancreas to digest food (pancreatic insufficiency). Symptoms range from mild to severe. Prognosis depends on the severity of symptoms as well as response to treatments; many affected individuals live well into adulthood. Milder forms of CFTR-related conditions include CAVD associated with male infertility, variable respiratory manifestations, and hereditary pancreatitis. Life span is not typically impacted with less severe CFTR-related conditions. Intellect is not affected with the various CFTR-related conditions. The combination of variants identified in an affected individual impacts the observed clinical features and severity of the symptoms. Additional genetic and environmental factors are believed to play a role in determining the risk of developing these complex CFTR-related conditions.

Additionally, individuals with a single disease-causing CFTR variant (heterozygous carriers) may have an approximately 4-10 fold increased risk for chronic pancreatitis, although the absolute risk of pancreatitis remains low (less than 1 in 100). Hereditary pancreatitis is characterized by recurrent episodes of acute inflammation of the pancreas (pancreatitis) beginning in childhood or adolescence, leading to chronic pancreatitis. Chronic pancreatitis is a risk factor for pancreatic cancer. Due to this potential increased risk for chronic pancreatitis, heterozygous carriers may consider follow-up with a medical provider.

Follow-up depends on each affected individual's specific situation, and discussion with a healthcare provider should be considered.

Next steps

Carrier testing for the reproductive partner is recommended.



If your partner tests positive:

In autosomal recessive inheritance, an individual must have disease-causing genetic changes in each copy of the CFTR gene to be affected. Carriers, who have a disease-causing genetic change in only one copy of the gene, typically do not have symptoms. When both reproductive partners are carriers of an autosomal recessive condition, there is a 25% chance for each child to have the condition.

If your partner tests negative:

A negative carrier test result reduces, but does not eliminate, the chance that an individual may be a carrier. The risk that an individual could still be a carrier, even after a negative test result, is called a residual risk. See the table in this report for your partner's hypothetical residual risk after testing negative for CFTR-related conditions. These values are provided only as a guide, are based on the detection

Patient name: 5926 Donor **DOB:**
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rate for the condition as tested at Labcorp Genetics, and assume a negative family history, the absence of symptoms, and vary based on the ethnic background of an individual. For genes associated with both dominant and recessive inheritance, the numbers provided apply to the recessive condition(s) associated with the gene.

DISORDER (INHERITANCE)	GENE	ETHNICITY	CARRIER FREQUENCY BEFORE SCREENING	CARRIER RESIDUAL RISK AFTER NEGATIVE RESULT
CFTR-related conditions (AR) NM_000492.3	CFTR *	Pan-ethnic	1 in 23	1 in 2200

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RESULT: CARRIER

CLRN1-related conditions

A single Pathogenic variant, c.149_152delinsTGTCCAAT (p.Ser50Leufs*12), was identified in CLRN1.

What are CLRN1-related conditions?

The CLRN1 gene is associated with multiple conditions that can have both distinct and overlapping symptoms. To understand which condition a genetic change is associated with, a review of the entire report, including the variant details section, is recommended.

CLRN1-related conditions include Usher syndrome type IIIA (USH3A) and autosomal recessive nonsyndromic retinitis pigmentosa (RP). Usher syndrome is a group of related conditions that causes deafness, progressive vision loss due to an eye disease called RP, and, in certain forms, balance difficulties due to inner ear problems (vestibular dysfunction). RP is a group of related conditions that affects the retina, which is the light-sensitive tissue that lines the back of the eye.

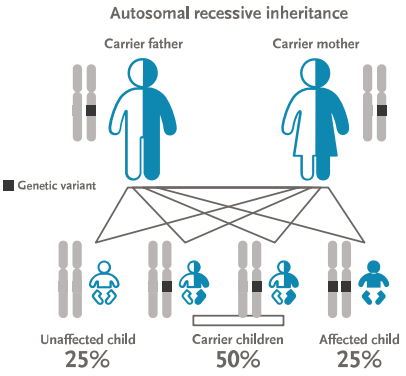
Individuals with USH3A are usually born with normal hearing and vision. Hearing and vision loss typically begin during late childhood or adolescence and worsen over time. Affected individuals typically have profound deafness by middle age. Some affected individuals develop balance problems later in life. Severity of symptoms can vary, even between family members with the same genetic change. Digenic inheritance, which occurs when an individual has a genetic change in two different Usher syndrome-associated genes, has been reported (PMID: 15537665); however, the evidence available at this time is insufficient to confirm this as a mode of inheritance.

The first symptom of RP is often difficulty seeing in low light settings (night blindness), which usually occurs during childhood or adolescence. Vision loss continues over years or decades and typically progresses to a loss of side (peripheral) vision, causing tunnel vision. Ultimately, central vision loss occurs. Many affected individuals are legally blind by adulthood, though the severity of symptoms and age of onset varies by individual. Intelligence and life expectancy are not typically affected.

For CLRN1-related conditions, early initiation of medical, educational, and social services is recommended to maximize outcomes.

Next steps

Carrier testing for the reproductive partner is recommended.



If your partner tests positive:

In autosomal recessive inheritance, an individual must have disease-causing genetic changes in each copy of the CLRN1 gene to be affected. Carriers, who have a disease-causing genetic change in only one copy of the gene, typically do not have symptoms. When both reproductive partners are carriers of an autosomal recessive condition, there is a 25% chance for each child to have the condition.

If your partner tests negative:

A negative carrier test result reduces, but does not eliminate, the chance that an individual may be a carrier. The risk that an individual could still be a carrier, even after a negative test result, is called a residual risk. See the table in this report for your partner's hypothetical residual risk after testing negative for CLRN1-related conditions. These values are provided only as a guide, are based on the detection rate for the condition as tested at Labcorp Genetics, and assume a negative family history, the absence of symptoms, and vary based on the ethnic background of an individual. For genes associated with both dominant and recessive inheritance, the numbers provided apply to the recessive condition(s) associated with the gene.

DISORDER (INHERITANCE)	GENE	ETHNICITY	CARRIER FREQUENCY BEFORE SCREENING	CARRIER RESIDUAL RISK AFTER NEGATIVE RESULT
CLRN1-related conditions (AR) NM_174878.2	CLRN1	Pan-ethnic	≤1 in 500	Reduced

Patient name: 5926 Donor **DOB:**
Invitae #: RQ8090210

RESULT: CARRIER

Neuronal ceroid lipofuscinosis type 6

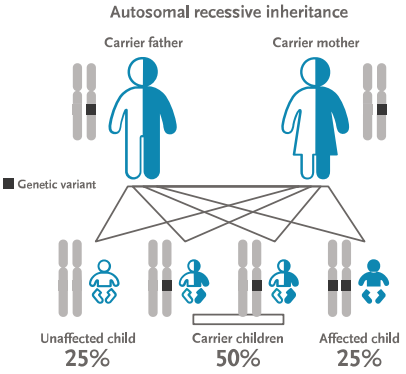
A single Pathogenic variant, c.13C>T (p.Arg5Trp), was identified in CLN6.

What is neuronal ceroid lipofuscinosis type 6?

Neuronal ceroid lipofuscinosis (NCL) is a group of related conditions resulting from dysfunction of lysosomes, which are structures in the cell that break down and recycle other molecules. NCLs primarily affect the brain. Ceroid lipofuscinosis, neuronal type 6 (CLN6) is a neurodegenerative condition resulting from storage material damaging brain cells (cerebral and cerebellar atrophy). Age of onset can vary. Late-infantile CLN6 typically presents between the ages of 18 months and eight years with seizures and with progressive loss of motor skills which causes problems with balance and coordination (ataxia), followed by jerky muscle contractions (myoclonus), loss of cognitive abilities, difficulty coordinating speech (dysarthria), and vision loss. Most affected individuals lose the ability to walk (nonambulatory) and need a wheelchair by late childhood. Life span is reduced, with most individuals living into the third decade. Adult CLN6 typically presents near age 30 with progressive myoclonus epilepsy. Prognosis depends on the severity of symptoms. Follow-up depends on each affected individual's specific situation, and discussion with a healthcare provider should be considered.

Next steps

Carrier testing for the reproductive partner is recommended.



If your partner tests positive:

In autosomal recessive inheritance, an individual must have disease-causing genetic changes in each copy of the CLN6 gene to be affected. Carriers, who have a disease-causing genetic change in only one copy of the gene, typically do not have symptoms. When both reproductive partners are carriers of an autosomal recessive condition, there is a 25% chance for each child to have the condition.

If your partner tests negative:

A negative carrier test result reduces, but does not eliminate, the chance that an individual may be a carrier. The risk that an individual could still be a carrier, even after a negative test result, is called a residual risk. See the table in this report for your partner's hypothetical residual risk after testing negative for neuronal ceroid lipofuscinosis type 6. These values are provided only as a guide, are based on the detection rate for the condition as tested at Labcorp Genetics, and assume a negative family history, the absence of symptoms, and vary based on the ethnic background of an individual. For genes associated with both dominant and recessive inheritance, the numbers provided apply to the recessive condition(s) associated with the gene.

DISORDER (INHERITANCE)	GENE	ETHNICITY	CARRIER FREQUENCY BEFORE SCREENING	CARRIER RESIDUAL RISK AFTER NEGATIVE RESULT
Neuronal ceroid lipofuscinosis type 6 (AR) NM_017882.2	CLN6	Pan-ethnic	≤1 in 500	Reduced

Patient name: 5926 Donor **DOB:**
Invitae #: RQ8090210

RESULT: CARRIER

Primary carnitine deficiency

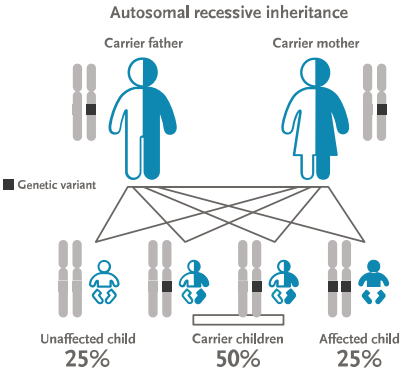
A single Likely Pathogenic variant, c.761G>A (p.Arg254Gln), was identified in SLC22A5.

What is primary carnitine deficiency?

Primary carnitine deficiency (PCD) is a condition in which individuals have difficulty breaking down fats for energy, leading to a variety of possible symptoms. The severity of symptoms of PCD varies widely among affected individuals. The infantile form typically presents with symptoms such as poor feeding, low blood sugar (hypoglycemia), lack of energy (lethargy), enlarged liver (hepatomegaly), and buildup of ammonia in the blood (hyperammonemia). The symptoms are triggered by fasting or concurrent illness (decompensation); symptoms can lead to coma, and may be fatal. The childhood onset form typically presents with weakened heart muscle (cardiomyopathy), and individuals with this form may also have weakness of the muscles used for movement (skeletal muscle myopathy). Adults with PCD may have susceptibility to fatigue (fatiguability). Other affected individuals may never experience any overt signs or symptoms (asymptomatic). Additionally, many minimally or asymptomatic women with PCD have been identified after having a child with an abnormal newborn screen for carnitine deficiency. Prognosis depends the severity of symptoms. Treatment with carnitine supplementation may help prevent or reduce the severity of symptoms. Follow-up depends on each affected individual's specific situation, and discussion with a healthcare provider should be considered.

Next steps

Carrier testing for the reproductive partner is recommended.



If your partner tests positive:

In autosomal recessive inheritance, an individual must have disease-causing genetic changes in each copy of the SLC22A5 gene to be affected. Carriers, who have a disease-causing genetic change in only one copy of the gene, typically do not have symptoms. When both reproductive partners are carriers of an autosomal recessive condition, there is a 25% chance for each child to have the condition.

If your partner tests negative:

A negative carrier test result reduces, but does not eliminate, the chance that an individual may be a carrier. The risk that an individual could still be a carrier, even after a negative test result, is called a residual risk. See the table in this report for your partner's hypothetical residual risk after testing negative for primary carnitine deficiency. These values are provided only as a guide, are based on the detection rate for the condition as tested at Labcorp Genetics, and assume a negative family history, the absence of symptoms, and vary based on the ethnic background of an individual. For genes associated with both dominant and recessive inheritance, the numbers provided apply to the recessive condition(s) associated with the gene.

DISORDER (INHERITANCE)	GENE	ETHNICITY	CARRIER FREQUENCY BEFORE SCREENING	CARRIER RESIDUAL RISK AFTER NEGATIVE RESULT
Primary carnitine deficiency (AR) NM_003060.3	SLC22A5	Pan-ethnic	1 in 76	1 in 7500

Patient name: 5926 Donor **DOB:**
Invitae #: RQ8090210

Variant details

CFTR, Intron 9, c.1210-34TG[12]T[5] (Intronic), heterozygous, PATHOGENIC

- This sequence change, also referred to as 5T;TG12 or TG12-5T in the literature, consists of 12 TG and 5 T sequence repeats on the same chromosome, and is located in intron 9 of the CFTR gene. It does not directly change the encoded amino acid sequence of the CFTR protein.
- The frequency data for this variant in the population databases is considered unreliable, as metrics indicate poor data quality at this position in the gnomAD database.
- The TG[12]T[5] allele has been observed in males with congenital bilateral absence of the vas deferens (CBAVD) and in both males and females with cystic fibrosis (CF) when present on the opposite chromosome (in trans) from a severe pathogenic CFTR variant (PMID: 14685937). When this allele is observed in trans with a severe pathogenic CFTR variant, the penetrance of CFTR-related conditions (CBAVD and/or mild CF) is expected to be high (>90%); however, the percentage of individuals who receive a diagnosis of CF (including individuals with mild lung disease and pancreatic sufficiency) has been reported to range from 6-35% (PMID: 14685937, 27447098, 35523714). Individuals who are homozygous for this variant, or who have this variant in combination with TG[11]T[5], are likely to be asymptomatic (PMID: 34196078).
- Algorithms developed to predict the effect of variants on gene product structure and function are not available or were not evaluated for this variant.
- Experimental studies demonstrate that the 5T allele leads to exclusion of exon 10 (referred to as exon 9 in some publications) from the mRNA, which ultimately results in a non-functional CFTR protein (PMID: 7691356, 7684641, 10556281, 14685937, 21658649). Importantly, the number of TG repeats (11, 12 or 13) modifies the extent of exon 10 skipping when in cis with the 5T allele (PMID: 14685937, 10556281, 9435322). In a mini-gene assay, the percentage of CFTR mRNA without exon 10 was 54% for TG[11]T[5], 72% for TG[12]T[5] and 100% for TG[13]T[5] (PMID: 10556281).
- Algorithms developed to predict the effect of sequence changes on RNA splicing suggest that this variant is not likely to affect RNA splicing.
- For these reasons, this variant has been classified as Pathogenic.

CLN6, Exon 1, c.13C>T (p.Arg5Trp), heterozygous, PATHOGENIC

- This sequence change replaces arginine, which is basic and polar, with tryptophan, which is neutral and slightly polar, at codon 5 of the CLN6 protein (p.Arg5Trp).
- The frequency data for this variant in the population databases is considered unreliable, as metrics indicate poor data quality at this position in the gnomAD database.
- This missense change has been observed in individual(s) with neuronal ceroid lipofuscinosis (PMID: 21990111, 28831385, 33024953). In at least one individual the data is consistent with being in trans (on the opposite chromosome) from a pathogenic variant. It has also been observed to segregate with disease in related individuals.
- ClinVar contains an entry for this variant (Variation ID: 457969).
- Invitae Evidence Modeling of protein sequence and biophysical properties (such as structural, functional, and spatial information, amino acid conservation, physicochemical variation, residue mobility, and thermodynamic stability) indicates that this missense variant is not expected to disrupt CLN6 protein function with a negative predictive value of 80%.
- For these reasons, this variant has been classified as Pathogenic.

CLRN1, Exon 1, c.149_152delinsTGTCCAAT (p.Ser50Leufs*12), heterozygous, PATHOGENIC

- This sequence change creates a premature translational stop signal (p.Ser50Leufs*12) in the CLRN1 gene. It is expected to result in an absent or disrupted protein product. Loss-of-function variants in CLRN1 are known to be pathogenic (PMID: 11524702, 24498627).
- Information on the frequency of this variant in the gnomAD database is not available, as this variant may be reported differently in the database.
- This premature translational stop signal has been observed in individual(s) with Usher syndrome (PMID: 12145752, 22135276).
- This variant is also known as c.149delCAGGinsTGTCCAAT.
- For these reasons, this variant has been classified as Pathogenic.

SLC22A5, Exon 4, c.761G>A (p.Arg254Gln), heterozygous, Likely Pathogenic

- This sequence change replaces arginine, which is basic and polar, with glutamine, which is neutral and polar, at codon 254 of the SLC22A5 protein (p.Arg254Gln).

- This variant is present in population databases (rs200699819, gnomAD 0.02%).
- This missense change has been observed in individual(s) with low plasma carnitine levels and primary carnitine deficiency (PMID: 26828774, 28711408, 37510298; internal data; http://www.arup.utah.edu/database/OCTN2/OCTN2_display.php).
- ClinVar contains an entry for this variant (Variation ID: 25389).
- Invitae Evidence Modeling of protein sequence and biophysical properties (such as structural, functional, and spatial information, amino acid conservation, physicochemical variation, residue mobility, and thermodynamic stability) indicates that this missense variant is expected to disrupt SLC22A5 protein function with a positive predictive value of 95%.
- Experimental studies have shown that this missense change does not substantially affect SLC22A5 function (PMID: 28841266).
- In summary, the currently available evidence indicates that the variant is pathogenic, but additional data are needed to prove that conclusively. Therefore, this variant has been classified as Likely Pathogenic.

Residual risk

No carrier test can detect 100% of carriers. There still remains a small risk of being a carrier after a negative test (residual risk). Residual risk values assume a negative family history. Carrier frequencies are derived from population databases including gnomAD (**https://gnomad.broadinstitute.org**) and ClinVar (**https://www.ncbi.nlm.nih.gov/clinvar**), or published literature and detection rates are estimated based on testing technologies used at Labcorp. Carrier Detection Rates and Residual Risks can be found at **womenshealth.labcorp.com/carrier-detection-rates-risks**. If this specimen had any gene-specific coverage gaps it will be listed in the Limitations section of this report and therefore the provided detection rates and residual risks may not be applicable.

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Patient name: 5926 Donor **DOB:**
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Genes analyzed

This table represents a complete list of genes analyzed for this individual, including the relevant gene transcript(s). If more than one transcript is listed for a single gene, variants were reported using the first transcript listed unless otherwise indicated in the report. An asterisk (*) indicates that this gene has a limitation. Please see the Limitations section for details. Results are negative, unless otherwise indicated in the report.

GENE	TRANSCRIPT
AAAS	NM_015665.5
ABCA12	NM_173076.2
ABCA3	NM_001089.2
ABCA4	NM_000350.2
ABCB11	NM_003742.2
ABCB4	NM_000443.3
ABCC2*	NM_000392.4
ABCC8	NM_000352.4
ABCD4	NM_005050.3
ACAD9	NM_014049.4
ACADM	NM_000016.5
ACADSB	NM_001609.3
ACADVL	NM_000018.3
ACAT1	NM_000019.3
ACOX1	NM_004035.6
ACSF3	NM_174917.4
ADA	NM_000022.2
ADAMTS2	NM_014244.4
ADAMTSL4	NM_019032.5
ADGRG1	NM_005682.6
ADGRV1	NM_032119.3
ADK	NM_001123.3
AGA	NM_000027.3
AGL	NM_000642.2
AGPAT2	NM_006412.3
AGPS	NM_003659.3
AGXT	NM_000030.2
AHCY	NM_000687.3
AHI1	NM_017651.4
AICDA	NM_020661.2
AIMP1	NM_004757.3
AIPL1*	NM_014336.4
AIRE	NM_000383.3
AK2*	NM_001625.3
AKR1D1*	NM_005989.3
ALDH3A2	NM_000382.2
ALDH4A1	NM_003748.3
ALDH7A1	NM_001182.4
ALDOB	NM_000035.3
ALG1	NM_019109.4
ALG12	NM_024105.3
ALG3	NM_005787.5
ALG6	NM_013339.3
ALMS1	NM_015120.4
ALOX12B	NM_001139.2
ALOXE3	NM_021628.2
ALPL	NM_000478.5
AMH	NM_000479.4
AMHR2	NM_020547.2
AMN*	NM_030943.3
AMPD2	NM_001257360.1
AMT	NM_000481.3
ANO10*	NM_018075.3
ANO5	NM_213599.2
ANTXR2*	NM_058172.5
AP1S1	NM_001283.3
AP3B1	NM_003664.4
AQP2	NM_000486.5
ARG1	NM_000045.3
ARL13B	NM_182896.2
ARL6	NM_177976.2
ARSA	NM_000487.5
ARSB	NM_000046.3
ASL	NM_000048.3
ASNS	NM_133436.3
ASPA	NM_000049.2
ASS1	NM_000050.4
ATM*	NM_000051.3
ATP13A2	NM_022089.3
ATP6V0A2	NM_012463.3
ATP6V0A4	NM_020632.2
ATP6V1B1	NM_001692.3
ATP6V1E1	NM_001696.3
ATP7B	NM_000053.3
ATP8B1*	NM_005603.4
B9D1	NM_015681.3
B9D2	NM_030578.3
BBS1	NM_024649.4
BBS10	NM_024685.3
BBS12	NM_152618.2
BBS2	NM_031885.3
BBS4	NM_033028.4
BBS5	NM_152384.2
BBS7	NM_176824.2
BBS9*	NM_198428.2
BCKDHA	NM_000709.3
BCKDHB	NM_183050.2
BCS1L	NM_004328.4
BLM	NM_000057.3
BLOC1S3	NM_212550.4
BLOC1S6	NM_012388.3
BMP1	NM_006129.4;NM_001199.3
BMPER	NM_133468.4
BRIP1	NM_032043.2
BSND	NM_057176.2
BTD	NM_000060.3
C19orf12	NM_001031726.3
CAD	NM_004341.4
CANT1	NM_138793.3
CAPN3	NM_000070.2
CASQ2	NM_001232.3
CASR	NM_000388.3
CAVIN1	NM_012232.5
CBS	NM_000071.2
CC2D1A	NM_017721.5
CC2D2A	NM_001080522.2
CCDC39	NM_181426.1
CCDC8	NM_032040.4

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GENE	TRANSCRIPT
CCDC88C	NM_001080414.3
CCN6	NM_003880.3
CD247	NM_198053.2
CD3D	NM_000732.4
CD3E	NM_000733.3
CD3G	NM_000073.2
CD40	NM_001250.5
CD59	NM_203330.2
CD8A	NM_001768.6
CDAN1	NM_138477.2
CDCA7	NM_031942.4
CDH23	NM_022124.5
CEP104	NM_014704.3
CEP152	NM_014985.3
CEP290	NM_025114.3
CERKL	NM_001030311.2
CERS3*	NM_178842.4
CFAP418	NM_177965.3
CFTR*	NM_000492.3
CHAT	NM_020549.4
CHMP1A	NM_002768.4
CHRNE	NM_000080.3
CHRNG	NM_005199.4
CHST6	NM_021615.4
CIB2	NM_006383.3
CIITA	NM_000246.3
CLCF1	NM_013246.2
CLCN1	NM_000083.2
CLCNKB*	NM_000085.4
CLN3	NM_001042432.1
CLN5	NM_006493.2
CLN6	NM_017882.2
CLN8	NM_018941.3
CLP1	NM_006831.2
CLRN1	NM_174878.2
CNGA1	NM_000087.3
CNGA3	NM_001298.2
CNGB1	NM_001297.4
CNGB3	NM_019098.4
CNTNAP2	NM_014141.5
COA8	NM_032374.4
COASY	NM_025233.6
COL11A2*	NM_080680.2
COL17A1	NM_000494.3
COL27A1	NM_032888.3
COL4A3	NM_000091.4
COL4A4	NM_000092.4
COL7A1	NM_000094.3
COLQ	NM_005677.3
COQ4	NM_016035.4
CORO1A*	NM_007074.3
COX10*	NM_001303.3
COX15	NM_004376.6
COX20	NM_198076.5
COX6B1	NM_001863.4
CP	NM_000096.3
CPLANE1	NM_023073.3
CPS1	NM_001875.4
CPT1A	NM_001876.3
CPT2	NM_000098.2
CRADD	NM_003805.4
CRB1	NM_201253.2
CRLF1	NM_004750.4
CRTAP	NM_006371.4
CTC1	NM_025099.5
CTNS	NM_004937.2
CTSA	NM_000308.3
CTSC	NM_001814.5
CTSD	NM_001909.4
CTSF	NM_003793.3
CTSK	NM_000396.3
CUL7	NM_014780.4
CWC27	NM_005869.3
CYBA	NM_000101.3
CYP11A1	NM_000781.2
CYP11B1*	NM_000497.3
CYP11B2*	NM_000498.3
CYP17A1	NM_000102.3
CYP19A1	NM_031226.2
CYP1B1	NM_000104.3
CYP21A2*	NM_000500.7
CYP27A1	NM_000784.3
CYP27B1	NM_000785.3
CYP4F22	NM_173483.3
CYP7B1	NM_004820.3
DBT	NM_001918.3
DCAF17	NM_025000.3
DCLRE1C	NM_001033855.2
DDB2	NM_000107.2
DDC*	NM_000790.3
DDR2	NM_006182.2
DDX11*	NM_030653.3
DGAT1	NM_012079.5
DGUOK	NM_080916.2
DHCR24	NM_014762.3
DHCR7	NM_001360.2
DHDDS	NM_024887.3
DLAT	NM_001931.4
DLD	NM_000108.4
DLL3	NM_016941.3
DNAAF19	NM_213607.2
DNAH11	NM_001277115.1
DNAH5	NM_001369.2
DNAI1	NM_012144.3
DNAI2	NM_023036.4
DNAL1	NM_031427.3
DNMT3B	NM_006892.3
DOCK8	NM_203447.3
DOK7	NM_173660.4
DOLK	NM_014908.3
DTNBP1	NM_032122.4
DUOX2*	NM_014080.4
DUOXA2	NM_207581.3
DYNC2H1	NM_001080463.1
DYSF	NM_003494.3
EFEMP2	NM_016938.4
EIF2AK3	NM_004836.6

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Labcorp Genetics 1400 16th Street. San Francisco. CA 94103 · clientservices@invitae.com · 800.436.3037

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GENE	TRANSCRIPT
EIF2B1	NM_001414.3
EIF2B2	NM_014239.3
EIF2B3	NM_020365.4
EIF2B4	NM_015636.3
EIF2B5	NM_003907.2
ELP1	NM_003640.3
EPB42	NM_000119.2
EPG5	NM_020964.2
ERBB3	NM_001982.3
ERCC2	NM_000400.3
ERCC3	NM_000122.1
ERCC4	NM_005236.2
ERCC5	NM_000123.3
ERCC6	NM_000124.3
ERCC8	NM_000082.3
ESCO2	NM_001017420.2
ETFA	NM_000126.3
ETFB	NM_001985.2
ETFDH	NM_004453.3
ETHE1	NM_014297.3
EVC	NM_153717.2
EVC2	NM_147127.4
EXOSC3	NM_016042.3
EYS*	NM_001142800.1
F7	NM_000131.4
FA2H	NM_024306.4
FAH*	NM_000137.2
FAM161A	NM_001201543.1
FANCA	NM_000135.2
FANCC	NM_000136.2
FANCD2*	NM_033084.3
FANCE	NM_021922.2
FANCF	NM_022725.3
FANCG	NM_004629.1
FANCI	NM_001113378.1
FANCL*	NM_018062.3
FBP1	NM_000507.3
FBXL4	NM_012160.4
FBXO7	NM_012179.3

GENE	TRANSCRIPT
FH*	NM_000143.3
FKBP10	NM_021939.3
FKRP	NM_024301.4
FKTN	NM_001079802.1
FMO3	NM_006894.6
FOLR1	NM_016725.2
FOXN1	NM_003593.2
FOXRED1	NM_017547.3
FRAS1	NM_025074.6
FREM2	NM_207361.5
FTCD	NM_006657.2
FUCA1	NM_000147.4
G6PC1	NM_000151.3
G6PC3	NM_138387.3
GAA	NM_000152.3
GALC*	NM_000153.3
GALE*	NM_000403.3
GALK1	NM_000154.1
GALNS	NM_000512.4
GALNT3	NM_004482.3
GALT*	NM_000155.3
GAMT	NM_000156.5
GATM	NM_001482.2
GBA1*	NM_001005741.2
GBE1	NM_000158.3
GCDH	NM_000159.3
GCH1	NM_000161.2
GDAP1	NM_018972.2
GDF5	NM_000557.4
GFM1	NM_024996.5
GFPT1*	NM_001244710.1
GHR*	NM_000163.4
GHRHR	NM_000823.3
GJB2	NM_004004.5
GLB1	NM_000404.2
GLDC	NM_000170.2
GLE1	NM_001003722.1
GNE*	NM_001128227.2
GNPAT	NM_014236.3

GENE	TRANSCRIPT
GNPTAB	NM_024312.4
GNPTG	NM_032520.4
GNRHR	NM_000406.2
GNS	NM_002076.3
GORAB	NM_152281.2
GPHN	NM_020806.4
GRHPR	NM_012203.1
GRIP1	NM_021150.3
GSS	NM_000178.2
GUCY2D	NM_000180.3
GUSB	NM_000181.3
GYS2	NM_021957.3
HADH	NM_005327.4
HADHA	NM_000182.4
HADHB	NM_000183.2
HAMP	NM_021175.2
HAX1	NM_006118.3
HBA1*	NM_000558.4
HBA2	NM_000517.4
HBB	NM_000518.4
HELLS	NM_018063.4
HEXA	NM_000520.4
HEXB	NM_000521.3
HGSNAT	NM_152419.2
HINT1	NM_005340.6
HJV	NM_213653.3
HLCS	NM_000411.6
HMGCL	NM_000191.2
HMGCS2	NM_005518.3
HMOX1	NM_002133.2
HOGA1	NM_138413.3
HPD	NM_002150.2
HPS1	NM_000195.4
HPS3	NM_032383.4
HPS4	NM_022081.5
HPS5	NM_181507.1
HPS6	NM_024747.5
HSD17B3	NM_000197.1
HSD17B4	NM_000414.3

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GENE	TRANSCRIPT
HSD3B2	NM_000198.3
HSD3B7	NM_025193.3
HYAL1	NM_153281.1
HYCC1	NM_032581.3
HYLS1	NM_145014.2
IDH3B	NM_006899.4
IDUA	NM_000203.4
IFT140	NM_014714.3
IGHMBP2	NM_002180.2
IKBKB	NM_001556.2
IL2RA	NM_000417.2
IL7R	NM_002185.3
INPP5E	NM_019892.4
INVS	NM_014425.3
ITGA2B	NM_000419.3
ITGA6	NM_000210.3
ITGB3	NM_000212.2
ITGB4	NM_001005731.2
ITPA	NM_033453.3
IVD	NM_002225.3
IYD	NM_203395.2
JAK3	NM_000215.3
KCNJ1	NM_000220.4
KCNJ11	NM_000525.3
KCTD7	NM_153033.4
KIF14	NM_014875.2
LAMA2	NM_000426.3
LAMA3	NM_000227.4
LAMB3	NM_000228.2
LAMC2	NM_005562.2
LARGE1	NM_004737.4
LARS1	NM_020117.10
LCA5	NM_181714.3
LCK	NM_001042771.2
LDLR	NM_000527.4
LDLRAP1	NM_015627.2
LHCGR	NM_000233.3
LHX3	NM_014564.4
LIFR*	NM_002310.5

GENE	TRANSCRIPT
LIG4	NM_002312.3
LIPA	NM_000235.3
LMAN1*	NM_005570.3
LMBRD1	NM_018368.3
LOXHD1	NM_144612.6
LPAR6	NM_005767.5
LPL	NM_000237.2
LRAT	NM_004744.4
LRP2	NM_004525.2
LRPPRC	NM_133259.3
LTBP4	NM_003573.2
LYST	NM_000081.3
MAK	NM_001242957.2
MALT1	NM_006785.3
MAN2B1	NM_000528.3
MANBA	NM_005908.3
MCCC1	NM_020166.4
MCCC2	NM_022132.4
MCEE	NM_032601.3
MCOLN1	NM_020533.2
MCPH1	NM_024596.4
MECR	NM_016011.3
MED17	NM_004268.4
MEFV	NM_000243.2
MEGF8*	NM_001410.2
MESP2	NM_001039958.1
MFSD8	NM_152778.2
MKKS	NM_018848.3
MKS1	NM_017777.3
MLC1*	NM_015166.3
MLYCD	NM_012213.2
MMAA	NM_172250.2
MMAB	NM_052845.3
MMACHC	NM_015506.2
MMADHC	NM_015702.2
MMUT	NM_000255.3
MOCS1	NM_001358530.2
MOCS2A	NM_176806.3
MOCS2B	NM_004531.4

GENE	TRANSCRIPT
MPI	NM_002435.2
MPL	NM_005373.2
MPV17	NM_002437.4
MRE11	NM_005591.3
MTHFD1	NM_005956.3
MTHFR*	NM_005957.4
MTMR2	NM_016156.5
MTR	NM_000254.2
MTRR	NM_002454.2
MTTP	NM_000253.3
MUSK	NM_005592.3
MVK	NM_000431.3
MYO15A	NM_016239.3
MYO7A	NM_000260.3
NAGA	NM_000262.2
NAGLU	NM_000263.3
NAGS	NM_153006.2
NBAS	NM_015909.3
NBEAL2	NM_015175.3
NBN	NM_002485.4
NCF2	NM_000433.3
NCF4	NM_013416.3
NDRG1	NM_006096.3
NDUFA11	NM_175614.4
NDUFAF2	NM_174889.4
NDUFAF5	NM_024120.4
NDUFS4	NM_002495.3
NDUFS6	NM_004553.4
NDUFS7	NM_024407.4
NDUFV1	NM_007103.3
NEB*	NM_001271208.1
NEU1	NM_000434.3
NGLY1	NM_018297.3
NHEJ1	NM_024782.2
NIPAL4	NM_001099287.1
NPC1	NM_000271.4
NPC2	NM_006432.3
NPHP1	NM_000272.3
NPHP3	NM_153240.4

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GENE	TRANSCRIPT
NPHS1	NM_004646.3
NPHS2*	NM_014625.3
NR2E3	NM_014249.3
NSMCE3	NM_138704.3
NTRK1	NM_001012331.1
OAT*	NM_000274.3
OBSL1	NM_015311.2
OCA2	NM_000275.2
ODAD3	NM_145045.4
OPA3	NM_025136.3
OSTM1	NM_014028.3
OTOA*	NM_144672.3
OTOF	NM_194248.2;NM_194323.2
P3H1	NM_022356.3
PAH	NM_000277.1
PANK2	NM_153638.2
PC	NM_000920.3
PCBD1	NM_000281.3
PCCA	NM_000282.3
PCCB	NM_000532.4
PCDH15	NM_033056.3
PCNT	NM_006031.5
PDE6A	NM_000440.2
PDHB	NM_000925.3
PDHX	NM_003477.2
PDP1	NM_018444.3
PEPD	NM_000285.3
PET100	NM_001171155.1
PEX1*	NM_000466.2
PEX10	NM_153818.1
PEX11B	NM_003846.2
PEX12	NM_000286.2
PEX13	NM_002618.3
PEX14	NM_004565.2
PEX16	NM_004813.2
PEX19	NM_002857.3
PEX2	NM_000318.2
PEX26	NM_017929.5
PEX3	NM_003630.2

GENE	TRANSCRIPT
PEX5	NM_001131025.1
PEX6	NM_000287.3
PEX7	NM_000288.3
PFKM	NM_000289.5
PGM3	NM_001199917.1
PHGDH	NM_006623.3
PHKB	NM_000293.2;NM_001031835.2
PHKG2	NM_000294.2
PHYH	NM_006214.3
PIGN	NM_176787.4
PIP5K1C	NM_012398.2
PJVK	NM_001042702.3
PKHD1*	NM_138694.3
PLA2G6	NM_003560.2
PLEKHG5	NM_020631.4
PLOD1	NM_000302.3
PLOD2	NM_182943.2
PMM2	NM_000303.2
PNP	NM_000270.3
PNPLA1	NM_001145717.1
PNPO	NM_018129.3
POC1A	NM_015426.4
POLG	NM_002693.2
POLH	NM_006502.2
POLR1C	NM_203290.3
POMGNT1	NM_017739.3
POMT1	NM_007171.3
POMT2	NM_013382.5
POR	NM_000941.2
POU1F1	NM_000306.3
PPIB	NM_000942.4
PPT1	NM_000310.3
PRCD	NM_001077620.2
PRDM5	NM_018699.3
PRF1	NM_001083116.1
PRICKLE1	NM_153026.2
PRKDC	NM_006904.6
PROP1	NM_006261.4
PSAP	NM_002778.3

GENE	TRANSCRIPT
PTPRC*	NM_002838.4
PTS	NM_000317.2
PUS1	NM_025215.5
PYCR1	NM_006907.3
PYGL	NM_002863.4
PYGM	NM_005609.3
QDPR	NM_000320.2
RAB23	NM_183227.2
RAG1	NM_000448.2
RAG2	NM_000536.3
RAPSN	NM_005055.4
RARS2	NM_020320.3
RAX	NM_013435.2
RD3	NM_183059.2
RDH12	NM_152443.2
RDH5	NM_002905.3
RFX5	NM_000449.3
RFXANK	NM_003721.3
RFXAP	NM_000538.3
RHAG	NM_000324.2
RLBP1	NM_000326.4
RMRP	NR_003051.3
RNASEH2A	NM_006397.2
RNASEH2B	NM_024570.3
RNASEH2C	NM_032193.3
ROGDI	NM_024589.2
RPE65	NM_000329.2
RPGRIP1	NM_020366.3
RPGRIP1L	NM_015272.2
RSPH9	NM_152732.4
RTEL1	NM_001283009.1
RXYLT1	NM_014254.2
RYR1	NM_000540.2
SACS	NM_014363.5
SAG	NM_000541.4
SAMD9	NM_017654.3
SAMHD1	NM_015474.3
SARS2	NM_017827.3
SCO1	NM_004589.3

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GENE	TRANSCRIPT
SCO2	NM_005138.2
SDCCAG8	NM_006642.3
SDR9C7	NM_148897.2
SEC23B	NM_006363.4
SELENON*	NM_020451.2
SEPSECS	NM_016955.3
SERPINF1	NM_002615.6
SGCA	NM_000023.2
SGCB	NM_000232.4
SGCD	NM_000337.5
SGCG	NM_000231.2
SGSH	NM_000199.3
SH3TC2	NM_024577.3
SKIC2	NM_006929.4
SKIC3	NM_014639.3
SLC12A1	NM_000338.2
SLC12A3	NM_000339.2
SLC12A6	NM_133647.1
SLC17A5	NM_012434.4
SLC19A2	NM_006996.2
SLC19A3	NM_025243.3
SLC1A4	NM_003038.4
SLC22A5	NM_003060.3
SLC25A13	NM_014251.2
SLC25A15	NM_014252.3
SLC25A20	NM_000387.5
SLC26A2	NM_000112.3
SLC26A3	NM_000111.2
SLC26A4	NM_000441.1
SLC27A4	NM_005094.3
SLC2A10	NM_030777.3
SLC2A2	NM_000340.1
SLC34A3	NM_080877.2
SLC35A3	NM_012243.2
SLC37A4	NM_001164277.1
SLC38A8	NM_001080442.2
SLC39A4	NM_130849.3
SLC3A1	NM_000341.3
SLC45A2	NM_016180.4

GENE	TRANSCRIPT
SLC46A1	NM_080669.5
SLC4A1	NM_000342.3
SLC4A11	NM_032034.3
SLC5A5	NM_000453.2
SLC6A19	NM_001003841.2
SLC7A7	NM_001126106.2
SLC7A9	NM_014270.4
SMARCAL1	NM_014140.3
SMN1*	NM_000344.3
SMPD1	NM_000543.4
SNAP29	NM_004782.3
SNX10	NM_001199835.1
SP110	NM_004509.3
SPATA7	NM_018418.4
SPG11	NM_025137.3
SPG21	NM_016630.6
SPG7	NM_003119.3
SPINK5	NM_006846.3
SPR	NM_003124.4
SRD5A2	NM_000348.3
ST3GAL5	NM_003896.3
STAR	NM_000349.2
STK4	NM_006282.3
STX11	NM_003764.3
STXBP2	NM_006949.3
SUCLA2	NM_003850.2
SUMF1	NM_182760.3
SUOX	NM_000456.2
SURF1	NM_003172.3
SYNE4	NM_001039876.2
TANGO2	NM_152906.6
TAT	NM_000353.2
TBCD	NM_005993.4
TBCE*	NM_003193.4
TBX19*	NM_005149.2
TCIRG1	NM_006019.3
TCN2	NM_000355.3
TCTN1	NM_001082538.2
TCTN2	NM_024809.4

GENE	TRANSCRIPT
TCTN3	NM_015631.5
TECPR2	NM_014844.3
TERT	NM_198253.2
TF	NM_001063.3
TG*	NM_003235.4
TGM1	NM_000359.2
TH	NM_199292.2
TK2	NM_004614.4
TMC1	NM_138691.2
TMEM138	NM_016464.4
TMEM216	NM_001173990.2
TMEM231	NM_001077416.2
TMEM237	NM_001044385.2
TMEM38B	NM_018112.2
TMEM67	NM_153704.5
TMEM70	NM_017866.5
TMPRSS3	NM_024022.2
TNFSF11	NM_003701.3
TNXB*	NM_019105.8
TPO	NM_000547.5
TPP1	NM_000391.3
TRAPPC11	NM_021942.5
TRDN	NM_006073.3
TREX1	NM_033629.4
TRHR	NM_003301.5
TRIM32	NM_012210.3
TRIM37	NM_015294.4
TRMU	NM_018006.4
TRPM6	NM_017662.4
TSEN2	NM_025265.3
TSEN54	NM_207346.2
TSFM*	NM_001172696.1
TSHB	NM_000549.4
TSHR	NM_000369.2
TTC7A	NM_020458.3
TTC8	NM_198309.3
TTPA	NM_000370.3
TULP1	NM_003322.4
TYMP	NM_001953.4

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Labcorp Genetics 1400 16th Street. San Francisco. CA 94103 · clientservices@invitae.com · 800.436.3037

LABCORP CARRIER SCREEN

Patient name: 5926 Donor **DOB:**
Invitae #: RQ8090210

GENE	TRANSCRIPT
TYR*	NM_000372.4
TYRP1	NM_000550.2
UBR1	NM_174916.2
UNC13D	NM_199242.2
UNG	NM_080911.2
USH1C*	NM_005709.3
USH1G	NM_173477.4
USH2A	NM_206933.2
VDR	NM_001017535.1
VLDLR	NM_003383.4
VPS11	NM_021729.5
VPS13A*	NM_033305.2
VPS13B	NM_017890.4
VPS45	NM_007259.4
VPS53*	NM_001128159.2
VRK1	NM_003384.2
VSX2	NM_182894.2
WHRN	NM_015404.3
WNT1	NM_005430.3
WNT10A	NM_025216.2
WRN*	NM_000553.4
XPA	NM_000380.3
XPC	NM_004628.4
ZAP70	NM_001079.3
ZBTB24	NM_014797.2
ZFYVE26	NM_015346.3
ZNF469	NM_001367624.2

Patient name: 5926 Donor **DOB:**
Invitae #: RQ8090210

Methods

- Genomic DNA obtained from the submitted sample is enriched for targeted regions using a hybridization-based protocol, and sequenced using Illumina technology. Unless otherwise indicated, all targeted regions are sequenced with a >=50x depth or are supplemented with additional analysis. Reads are aligned to a reference sequence (GRCh37), and sequence changes are identified and interpreted in the context of a single clinically relevant transcript, indicated in the Genes Analyzed table. Enrichment and analysis focus on the coding sequence of the indicated transcripts, 20bp of flanking intronic sequence, and other specific genomic regions demonstrated to be causative of disease at the time of assay design. Promoters, untranslated regions, and other non-coding regions are not otherwise interrogated. Exonic deletions and duplications are called using an in-house algorithm that determines copy number at each target by comparing the read depth for each target in the proband sequence with both mean read-depth and read-depth distribution, obtained from a set of clinical samples. Markers across the X and Y chromosomes are analyzed for quality control purposes and may detect deviations from the expected sex chromosome complement. Such deviations may be included in the report in accordance with internal guidelines. No other incidental findings will be reported. Variants are reported according to the Human Genome Variation Society (HGVS) guidelines. Confirmation of the presence and location of reportable variants is performed as needed based on stringent criteria, using one of several validated orthogonal approaches (PubMed ID 30610921). Primary and confirmatory testing is performed by Labcorp Genetics (1400 16th Street, San Francisco, CA 94103, #05D2040778).

  The following additional analyses are performed if relevant to the requisition. For FMR1, cytosine-guanine-guanine (CGG) triplet repeats in the 5' untranslated region (5' UTR) of the FMR1 gene are detected by triplet repeat-primed PCR (RP-PCR) with fluorescently labeled primers followed by capillary electrophoresis. Reference ranges: Normal: <45 CGG repeats, intermediate: 45-54 CGG repeats, premutation: 55-200 CGG repeats, full mutation: >200 CGG repeats. For alleles with 55-90 triplet repeats, the region surrounding the FMR1 repeat is amplified by PCR. The PCR amplicons are then processed through PacBio SMRTBell library prep and sequenced using PacBio long read technology. The number of AGG interruptions within the 55-90 triplet repeat is read directly from the resulting DNA sequences. For GBA1 the reference genome has been modified to mask the sites of polymorphic paralog sequence variants (PSVs) in both the gene and pseudogene. For CYP21A2 and GBA1, if one or more reportable variants, gene conversion, or fusion event is identified via our NGS pipeline (see Limitations), these variants are confirmed by PacBio sequencing of an amplicon generated by long-range PCR and subsequent short-range PCR. For GJB2, the reportable range includes large upstream deletions overlapping GJB6. For HBA1/2, the reference genome has been modified to force some sequencing reads derived from HBA1 to align to HBA2, and variant calling algorithms are modified to support an expectation of 4 alleles in these regions. HBA1/2 copy number calling is performed by a custom hypothesis testing algorithm. Copy number losses are only reported for coding sequence of HBA1 and HBA2 and the HS-40 region. This assay does not distinguish among the -alpha3.7 or -alpha4.2 subtypes. -alpha3.7 variants are reported as HBA1 deletions.

- This report only includes variants that have a clinically significant association with the conditions tested as of the report date. Variants of uncertain significance, benign variants, and likely benign variants may also have been detected but are not included in this report. If interested, these can be released upon request. Additional evidence may become available to indicate that the clinical significance of a variant has changed; updated classifications are available upon request.
- A PMID is a unique identifier referring to a published, scientific paper. Search by PMID at http://www.ncbi.nlm.nih.gov/pubmed.
- An rsID is a unique identifier referring to a single genomic position, and is used to associate population frequency information with sequence changes at that position. Reported population frequencies are derived from a number of public sites that aggregate data from large-scale population sequencing projects, including ExAC, gnomAD, and dbSNP.

Disclaimer

DNA studies do not constitute a definitive test for the selected condition(s) in all individuals. It should be realized that there are possible sources of error. Errors can result from trace contamination, rare technical errors, rare genetic variants that interfere with analysis, recent scientific developments, and alternative classification systems. This test should be one of many aspects used by the healthcare provider to help with a diagnosis and treatment plan, but it is not a diagnosis itself. This test was developed and its performance characteristics determined by Labcorp Genetics. It has not been cleared or approved by the FDA. The laboratory is regulated under the Clinical Laboratory Improvement Amendments (CLIA) as qualified to perform high-complexity clinical tests (CLIA ID: 05D2040778). This test is used for clinical purposes. It should not be regarded as investigational or for research.

Limitations

- Based on validation study results, this assay achieves >99% analytical sensitivity and specificity for single nucleotide variants, insertions and deletions <15bp in length, and multi-exon deletions and duplications. The methods also detect insertions and deletions larger than 15bp and

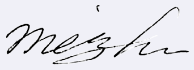
smaller than two exons, but sensitivity for these may be marginally reduced. In some situations, single-exon copy number events may not be confidently determined due to inherent sequence properties or isolated reduction in data quality. Certain types of variants, such as structural rearrangements (e.g. inversions, gene conversion events, translocations, etc.) or variants embedded in sequence with complex architecture (e.g. short tandem repeats or segmental duplications), may cause false positive or false negative results. Additionally, it may not be possible to fully resolve certain details about variants, such as mosaicism, phasing, or mapping ambiguity. Unless explicitly guaranteed, sequence changes in the promoter, non-coding exons, and other non-coding regions are not covered by this assay. Please consult the test definition on our website for details regarding regions or types of variants that are covered or excluded for this test. This report reflects the analysis of an extracted genomic DNA sample. While this test is intended to reflect the analysis of extracted genomic DNA from a referred patient, in very rare cases the analyzed DNA may not represent that individual's constitutional genome, such as in the case of a circulating hematolymphoid neoplasm, bone marrow transplant, blood transfusion, chimerism, culture artifact or maternal cell contamination. Interpretations are made on the assumption that any clinical information provided, including specimen identity, is accurate.

- LMAN1: Sequencing analysis for exon 7 includes only cds +/- 0 bp. ANO10: Sequencing analysis for exon 8 includes only cds +/- 0 bp. ATP8B1: Sequencing analysis for exon 19 includes only cds +/- 10 bp. AIPL1: Sequencing analysis for exon 2 includes only cds +/- 10 bp. GHR: Deletion/duplication and sequencing analysis is not offered for exon 3. TBCE: Sequencing analysis for exon 2 includes only cds +/- 10 bp. AKR1D1: Sequencing analysis for exon 4 includes only cds +/- 0 bp. CERS3: Sequencing analysis for exon 8 includes only cds +/- 10 bp. CYP21A2: Analysis includes the most common variants (c.92C>T (p.Pro31Leu), c.293-13C>G (intronic), c.332_339del (p.Gly111Valfs*21), c.518T>A (p.Ile173Asn), c.710T>A (p.Ile237Asn), c.713T>A (p.Val238Glu), c.719T>A (p.Met240Lys), c.844G>T (p.Val282Leu), c.923dup (p.Leu308Phefs*6), c.955C>T (p.Gln319*), c.1069C>T (p.Arg357Trp), c.1360C>T (p.Pro454Ser) and the 30Kb deletion) as well as select rare HGMD variants only (list available upon request). Full gene duplications are reported only in the presence of a pathogenic variant(s). When a duplication and a pathogenic variant(s) is identified, phase (cis/trans) cannot be determined. Identification of full gene deletions is not offered. Sensitivity to detect and/or accurately name variants may be reduced if they arise from whole gene deletions and/or complex gene conversion, fusion, or deletion/duplication events. TYR: Deletion/duplication and sequencing analysis is not offered for exon 5. PTPRC: Sequencing analysis is not offered for exons 3, 15. ABCC2: Deletion/duplication analysis is not offered for exons 24-25. OTOA: Deletion/duplication and sequencing analysis is not offered for exons 20-28. COX10: Deletion/duplication and sequencing analysis is not offered for exon 6. CORO1A: Deletion/duplication and sequencing analysis is not offered for exon 11. DUOX2: Deletion/duplication and sequencing analysis is not offered for exons 6-7. TG: Deletion/duplication analysis is not offered for exon 18. Sequencing analysis for exon 44 includes only cds +/- 0 bp. FANCD2: Deletion/duplication analysis is not offered for exons 14-17, 22 and sequencing analysis is not offered for exons 15-17. Sequencing analysis for exons 6, 14, 18, 20, 23, 25, 34 includes only cds +/- 10 bp. TBX19: Sequencing analysis for exon 3 includes only cds +/- 5 bp. SELENON: Deletion/duplication analysis is not offered for exon 1. FANCL: Sequencing analysis for exons 4, 10 includes only cds +/- 10 bp. GFPT1: Sequencing analysis for exon 20 includes only cds +/- 10 bp. DDC: Deletion/duplication analysis is not offered for exons 10-11. ATM: Sequencing analysis for exons 6, 24, 43 includes only cds +/- 10 bp. CFTR: Sequencing analysis for exon 7 includes only cds +/- 10 bp. CYP11B1: The presence of a gene conversion may reduce the sensitivity to detect variants in the involved regions. Gene fusions will be reported as individual copy number variants. However, the exact fusion breakpoints cannot be determined by this assay. CYP11B2: The presence of a gene conversion may reduce the sensitivity to detect variants in the involved regions. Gene fusions will be reported as individual copy number variants. However, the exact fusion breakpoints cannot be determined by this assay. EYS: Sequencing analysis for exon 30 includes only cds +/- 0 bp. FAH: Deletion/duplication analysis is not offered for exon 14. FH: Sequencing analysis for exon 9 includes only cds +/- 10 bp. GALC: Deletion/duplication analysis is not offered for exon 6. GALT: The NM_000155.3:c.-119_-116del (Non-coding) (aka Duarte allele) variant is not included on the report unless requested. If requested and present, it would be reported in the Results to Note section of the report. GBA1: Analysis includes the most common variants: c.84dup (p.Leu29Alafs*18), c.115+1G>A (splice donor), c.222_224del (p.Thr75del), c.680A>G (p.Asn227Ser), c.887G>A (p.Arg296Gln), c.1226A>G (p.Asn409Ser), c.1342G>C (p.Asp448His), c.1448T>C (p.Leu483Pro), c.1504C>T (p.Arg502Cys), c.1604G>A (p.Arg535His) and others. Full list available upon request. Rarely, sensitivity to detect these variants may be reduced. When sensitivity is reduced, zygosity may be reported as "unknown". GNE: Sequencing analysis for exon 8 includes only cds +/- 10 bp. HBA1: This assay is designed to detect deletions of the alpha globin gene(s), resulting from the -alpha20.5, --MED, --SEA, --FIL/--THAI, -alpha3.7, or -alpha4.2. Additional copies of the alpha globin gene(s) are not included on the report unless requested. If requested and present, it would be reported in the Results to Note section of the report. Sensitivity to detect other copy number variants may be reduced. This assay may not detect the co-occurrence of a deletion and a duplication. In addition, deletion of the enhancer element HS-40 and the sequence variant, Constant Spring (NM_000517.4:c.427T>C), can be identified by this assay. LIFR: Sequencing analysis for exon 3 includes only cds +/- 5 bp. MLC1: Sequencing analysis for exon 11 includes only cds +/- 10 bp. MTHFR: The NM_005957.4:c.665C>T (p.Ala222Val) (aka 677C>T) and c.1286A>C (p.Glu429Ala) (aka 1298A>C) variants are not reported. NEB: Deletion/duplication analysis is not offered for exons 82-105. Sequence variants in this region with no evidence towards pathogenicity are not included in this report, but are available upon request. NPHS2: The NM_014625.3:c.686G>A (p.Arg229Gln) variant is not reported. OAT: Deletion/duplication analysis is not offered for exon 2. PEX1: Sequencing analysis for exon 16 includes only cds +/- 0 bp. PKHD1: Deletion/duplication analysis is not offered for exon 13. SMN1 or SMN2: NM_000344.3:c.*3+80T>G variant only. SMN1: This assay unambiguously detected copy number for exon 8 (also known as exon 7 in the literature; PMID: 8838816). The presence of the g.27134T>G variant (also known as c.*3+80T>G) is reported if copy number = 2 for this gene.

Patient name: 5926 Donor **DOB:**
Invitae #: RQ8090210

TSFM: Sequencing analysis is not offered for exon 5. USH1C: Deletion/duplication analysis is not offered for exons 5-6. VPS13A: Deletion/duplication analysis is not offered for exons 2-3, 27-28. VPS53: Sequencing analysis for exon 14 includes only cds +/- 5 bp. AMN: Deletion/duplication analysis is not offered for exon 1. ANTXR2: Sequencing analysis for exon 8 includes only cds +/- 0 bp. MEGF8: Deletion/duplication analysis is not offered for exon 39. Sequencing analysis for exon 39 includes only cds +/- 10 bp. GALE: Sequencing analysis for exon 10 includes only cds +/- 5 bp. DDX11: Deletion/duplication analysis is not offered for this gene. BBS9: Deletion/duplication analysis is not offered for exon 4. CLCNKB: Deletion/duplication analysis is not offered for this gene. COL11A2: Deletion/duplication analysis is not offered for exon 36. AK2: Deletion/duplication and sequencing analysis is not offered for exon 6. TNXB: Sensitivity and specificity for deletion/duplication and sequencing analysis is reduced due to homology and other structural complexities. Due to high genomic complexity, confirmation by an orthogonal method may not be performed for variants detected in this gene. Deletion/duplication and sequencing analysis is not offered for exons 32-44. WRN: Deletion/duplication analysis is not offered for exons 10-11. Sequencing analysis for exons 8, 10-11 includes only cds +/- 10 bp.

This report has been reviewed and approved by:



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