



DONOR NUMBER:
7108

This donor is a healthy carrier for a genetic disease.
Please see his [Genetic Testing Summary](#) and [Acknowledgment of Genetic Risk](#) for details



PHYSICAL

Height:
5'9"
(175 cm)

Weight:
170 lb
(77 kg)

Eye Color:
Green

Hair:
Brown/
Straight

Skin Tone:
Medium Light

Ancestry:
Latino



Blood Type:
O+

Ethnic Background:
Ecuadorian/Ecuadorian

Education:
Pursuing EdD/Org. Leadership

Occupation:
Science Teacher

Interests: Piano, Video games



MEDICAL

QUESTION RESPONSE

Have you or any of your family members been diagnosed with alcoholism or drug addiction? If yes, relation and age affected:

No

QUESTION	RESPONSE
Any dietary restrictions? If yes, explain:	No
Do you wear glasses or contact lenses? Are you near or far-sighted?	Yes - Near-sighted, -6.0/-6.0
Allergies (medicines, food, pollens)? If yes, please list substance and reaction caused:	No
<u>CMV IgG Antibody</u>	<u>Positive</u>
<u>CMV IgM Antibody</u>	<u>Negative</u>
Note any comments regarding above items:	N/A



FAMILY MEDICAL HISTORY

See list of questions asked [here](#)

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YOUR MOTHER

QUESTION	RESPONSE
Current age or age at death	52
Living / Dead	Living
Cause of death and any treatment prior to death	N/A

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HEALTH PROBLEMS

Healthy

—

YOUR FATHER

QUESTION	RESPONSE
Current age or age at death	49
Living / Dead	Living
Cause of death and any treatment prior to death	N/A

HEALTH PROBLEMS

Healthy

BROTHERS

Your Brother 1

QUESTION	RESPONSE
Current age or age at death	21
Living / Dead	Living
Cause of death and any treatment prior to death	N/A

HEALTH PROBLEMS

Healthy

YOUR MOTHER'S FATHER

QUESTION	RESPONSE
Current age or age at death	67
Living / Dead	Living
Cause of death and any treatment prior to death	N/A

HEALTH PROBLEMS

Healthy

YOUR MOTHER'S MOTHER

QUESTION	RESPONSE
Current age or age at death	65
Living / Dead	Dead
Cause of death and any treatment prior to death	COVID-19 - Respiratory issues

HEALTH PROBLEMS

DISEASE	AGE DIAGNOSED	TREATMENT FOR CONDITION
High blood pressure	50	Medication

YOUR MOTHER'S BROTHERS 1

QUESTION	RESPONSE
Current age or age at death	48
Living / Dead	Living
Cause of death and any treatment prior to death	N/A



HEALTH PROBLEMS

Healthy

YOUR MOTHER'S BROTHERS 2

QUESTION	RESPONSE
Current age or age at death	37
Living / Dead	Living
Cause of death and any treatment prior to death	N/A



HEALTH PROBLEMS

Healthy

YOUR FATHER'S FATHER

QUESTION	RESPONSE
Current age or age at death	74
Living / Dead	Dead
Cause of death and any treatment prior to death	Heart attack



HEALTH PROBLEMS

DISEASE	AGE DIAGNOSED	TREATMENT FOR CONDITION
Other heart disease	65	Heart strokes, due to lack of health care, no specific diagnosis or treatment

YOUR FATHER'S MOTHER

QUESTION	RESPONSE
Current age or age at death	86
Living / Dead	Living
Cause of death and any treatment prior to death	N/A

HEALTH PROBLEMS

Healthy

YOUR FATHER'S SISTERS 1

QUESTION	RESPONSE
Current age or age at death	64
Living / Dead	Living
Cause of death and any treatment prior to death	N/A

HEALTH PROBLEMS

Healthy

YOUR FATHER'S SISTERS 2

QUESTION	RESPONSE
Current age or age at death	61
Living / Dead	Living
Cause of death and any treatment prior to death	N/A

HEALTH PROBLEMS

Healthy

YOUR FATHER'S SISTERS 3

QUESTION	RESPONSE
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QUESTION	RESPONSE
Current age or age at death	59
Living / Dead	Living
Cause of death and any treatment prior to death	N/A

HEALTH PROBLEMS

Healthy

YOUR FATHER'S SISTERS 4

QUESTION	RESPONSE
Current age or age at death	51
Living / Dead	Living
Cause of death and any treatment prior to death	N/A

HEALTH PROBLEMS

Healthy

YOUR FATHER'S BROTHERS 1

QUESTION	RESPONSE
Current age or age at death	55
Living / Dead	Living
Cause of death and any treatment prior to death	N/A

HEALTH PROBLEMS

Healthy

YOUR FATHER'S BROTHERS 2

QUESTION	RESPONSE
Current age or age at death	52

QUESTION	RESPONSE
Living / Dead	Living
Cause of death and any treatment prior to death	N/A

HEALTH PROBLEMS

Healthy

YOUR FATHER'S BROTHERS 3

QUESTION	RESPONSE
Current age or age at death	35
Living / Dead	Dead
Cause of death and any treatment prior to death	Car Accident

HEALTH PROBLEMS

DISEASE	AGE DIAGNOSED	TREATMENT FOR CONDITION
Other		No diagnosed health problems at time of death.

RELIGION:

Faith	Other
Denomination	Agnostic theism

UPDATES TO PROFILE

Update Available	Yes
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Updates - Personal	Update received September 2024: Donor reports his religion is now Christian (Catholic).
Updates - Medical	N/A
Updates - Family Medical History	N/A



Donor 7108

Genetic Testing Summary

Fairfax Cryobank recommends reviewing this genetic testing summary with your healthcare provider to determine suitability.

Last Updated: 09/05/23

Donor Reported Ancestry: Ecuadorian

Jewish Ancestry: No

Genetic Test*	Result	Comments/Donor's Residual Risk**
Chromosome analysis (karyotype)	Normal male karyotype	No evidence of clinically significant chromosome abnormalities
Hemoglobin evaluation	Normal hemoglobin fractionation and MCV/MCH results	Reduced risk to be a carrier for sickle cell anemia, beta thalassemia, alpha thalassemia trait (aa/-- and a-/a-) and other hemoglobinopathies
Expanded Genetic Disease Carrier Screening Panel attached- 514 diseases by gene sequencing.	Carrier: Biotinidase deficiency (BTD) Carrier: Glycogen storage disease type II (Pompe disease) (GAA) Negative for other genes sequenced.	Partner testing recommended before using this donor. Residual risks for negative results can be seen here: https://www.invitae.com/carrier-residual-risks/

*No single test can screen for all genetic disorders. A negative screening result significantly reduces, but cannot eliminate, the risk for these conditions in a pregnancy.

**Donor residual risk is the chance the donor is still a carrier after testing negative.

Patient name:	Donor 7108	Sample type:	Blood	Report date:	08-FEB-2023
DOB:	██████████	Sample collection date:	26-JAN-2023	Invitae #:	██████████
Sex assigned at birth:	Male	Sample accession date:	31-JAN-2023	Clinical team:	██████████
Gender:					██████████
Patient ID (MRN):					

Reason for testing

Gamete donor

Test performed

Invitae Comprehensive Carrier Screen without X-linked Disorders

- Primary Panel (CF, SMA)
- Add-on Comprehensive Carrier Screen without X-linked Disorders genes
- Add-on genes with variable presentation


RESULT: POSITIVE

This carrier test evaluated 514 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 below. No other clinically significant changes were identified in the remaining genes evaluated with this test.

RESULTS	GENE	VARIANT(S)	INHERITANCE	PARTNER TESTING RECOMMENDED
Carrier: Biotinidase deficiency	BTB	c.1330G>C (p.Asp444His)	Autosomal recessive	Yes
Carrier: Glycogen storage disease type II (Pompe disease)	GAA	c.-32-13T>G (Intronic)	Autosomal recessive	Yes



Patient name: Donor 7108

DOB: [REDACTED]

Invitae #: [REDACTED]

Next steps

- See the table above for recommendations regarding testing of this individual's reproductive partner.
- 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.
- Individuals can register their tests at <https://www.invitae.com/patients/> to access online results, educational resources, and next steps.

Clinical summary



RESULT: CARRIER

Biotinidase deficiency

A single Pathogenic variant, c.1330G>C (p.Asp444His), was identified in BTBD.

What is biotinidase deficiency?

Biotinidase deficiency (BTD) is a condition in which the body has difficulty recycling a B vitamin called biotin. Symptoms of BTD are variable and typically involve neurologic and skin findings. If untreated, profound BTD typically presents during the first few months of life, and the symptoms may be severe. There is also a milder form of BTD, called partial biotinidase deficiency. Individuals with partial BTD typically do not have any signs or symptoms of the condition (asymptomatic). However, if untreated, symptoms of partial BTD may appear during times of illness or stress and may include low muscle tone (hypotonia), skin rashes, and hair loss (alopecia). BTD is readily treatable, and early treatment, including biotin supplementation, may prevent or reduce the severity of symptoms.

Individuals with partial BTD have one copy of the c.1330G>C (p.Asp444His) variant and a second disease-causing variant in the BTBD gene on the opposite chromosome. Some individuals have 2 copies of the c.1330G>C (p.Asp444His) variant (homozygous). These individuals have mild enzyme deficiency, but do not have clinical symptoms of partial BTD.

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.



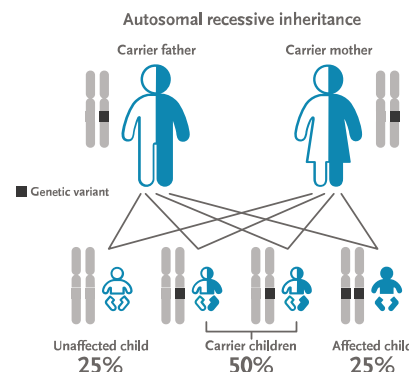
If your partner tests positive:

The various forms of biotinidase deficiency are inherited in an autosomal recessive fashion. In autosomal recessive inheritance, an individual must have disease-causing genetic changes in each copy of the BTBD 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. The form of biotinidase deficiency depends on the specific BTBD variants inherited from the reproductive parents.



If your partner tests negative:

A negative carrier test result reduces, but does not eliminate, the chance that a person may be a carrier. The risk that a person could still be a carrier, even after a negative test result, is called a residual risk. See the table below for your partner's hypothetical residual risk after testing negative for biotinidase deficiency. These values are provided only as a guide, are based on the detection rate for the condition as tested at Invitae, 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
Biotinidase deficiency (AR) NM_000060.3	BTBD	Pan-ethnic	1 in 125	1 in 12400


RESULT: CARRIER

Glycogen storage disease type II (Pompe disease)

A single Pathogenic variant, c.-32-13T>G (Intronic), was identified in GAA.

What is glycogen storage disease type II (Pompe disease)?

Glycogen storage disease (GSD) is a group of conditions in which individuals have difficulty breaking down a complex sugar called glycogen. A buildup of glycogen impairs the function of certain organs and tissues. The symptoms of glycogen storage disease type II (GSD II), also called Pompe disease, vary in age of onset and severity. Classical Pompe disease typically presents in infancy and is characterized by low muscle tone (hypotonia), poor growth (failure to thrive), muscle weakness (myopathy), an enlarged heart (cardiomegaly) and thickened heart muscle (hypertrophic cardiomyopathy). The condition is often fatal in infancy or early childhood due to heart or breathing problems. Non-classical forms of Pompe disease can present in infancy, childhood, adolescence, or adulthood, often with milder symptoms and slower disease progression. Symptoms may include weakness in the arm and leg muscles that are closest to the body (proximal myopathy) and breathing difficulties, with little to no heart muscle involvement. Enzyme replacement therapy is available and early initiation may delay the onset of the symptoms and reduce their severity. 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.



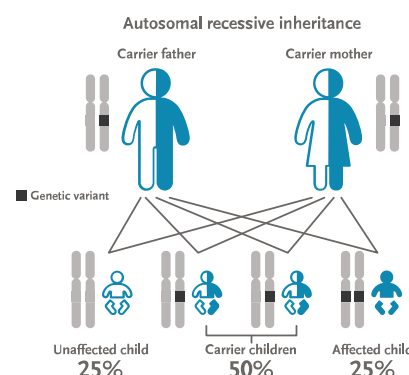
If your partner tests positive:

In autosomal recessive inheritance, an individual must have disease-causing genetic changes in each copy of the GAA 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 a person may be a carrier. The risk that a person could still be a carrier, even after a negative test result, is called a residual risk. See the table below for your partner's hypothetical residual risk after testing negative for glycogen storage disease type II (Pompe disease). These values are provided only as a guide, are based on the detection rate for the condition as tested at Invitae, 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
Glycogen storage disease type II (Pompe disease) (AR) NM_000152.3	GAA	Pan-ethnic	1 in 100	1 in 9900

Results to note

SMN1

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

Pseudodeficiency allele(s)

- Benign changes, c.550C>T (p.Arg184Cys) and c.1685T>C (p.Ile562Thr), known to be pseudodeficiency alleles, identified in the GALC gene. Pseudodeficiency alleles are not known to be associated with disease, including Krabbe disease.
- The presence of a pseudodeficiency allele does not impact this individual's risk to be a carrier. Individuals with pseudodeficiency alleles may exhibit false positive results on related biochemical tests, including newborn screening. However, pseudodeficiency alleles are not known to cause disease, even when there are two copies of the variant (homozygous) or when in combination with another disease-causing variant (compound heterozygous). Carrier testing for the reproductive partner is not indicated based on this result.

Variant details

BTD, Exon 4, c.1330G>C (p.Asp444His), heterozygous, PATHOGENIC

- This sequence change replaces aspartic acid, which is acidic and polar, with histidine, which is basic and polar, at codon 444 of the BTD protein (p.Asp444His).
- This variant is present in population databases (rs13078881, gnomAD 6%), and has an allele count higher than expected for a pathogenic variant.
- In the homozygous state this variant does not cause biotinidase deficiency or partial biotinidase deficiency (PMID: 28682309, 9654207). However, this variant in conjunction with another pathogenic variant is a common cause of partial biotinidase deficiency (PMID: 10206677, 9654207, 12227467, 23644139). This variant has also been observed in individuals affected with profound biotinidase deficiency when this variant is in cis with the p.A171T variant and in trans with a third variant (PMID: 10206677, 9654207).
- In individuals affected with partial biotinidase deficiency who harbor this variant in combination with another BTD variant, serum biotinidase activity was approximately 24% of the mean normal control activity (PMID: 9654207). In individuals affected with profound biotinidase deficiency who harbor this variant in cis with p.A171T and in trans with another BTD variant, serum biotinidase activity was <10% of the mean normal control activity (PMID: 10206677, 9654207). Individuals who are homozygous for this variant typically have an enzyme activity that is approximately 50% of normal (PMID: 20539236, 28682309, 9654207), similar to what is seen for a carrier of a profound allele.
- ClinVar contains an entry for this variant (Variation ID: 1900).
- Advanced modeling of protein sequence and biophysical properties (such as structural, functional, and spatial information, amino acid conservation, physicochemical variation, residue mobility, and thermodynamic stability) performed at Invitae indicates that this missense variant is expected to disrupt BTD protein function.
- For these reasons, this variant has been classified as Pathogenic.

GAA, Intron 1, c.-32-13T>G (Intronic), heterozygous, PATHOGENIC

- This sequence change falls in intron 1 of the GAA gene. It does not directly change the encoded amino acid sequence of the GAA protein. RNA analysis indicates that this variant induces altered splicing and is likely to result in the loss of the initiator methionine.
- This variant is present in population databases (rs386834236, gnomAD 0.6%), including at least one homozygous and/or hemizygous individual.
- This variant is a well documented cause of late-onset Pompe disease among individuals of European ancestry, and it has also been reported in affected individuals of other ethnicities (PMID: 7881425, 24590251, 21439876, 22613277, 26231297, 24245577). It has also been observed to segregate with disease in related individuals.
- ClinVar contains an entry for this variant (Variation ID: 4027).



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- Experimental studies have shown that this variant interferes with mRNA splicing of exon 1 of the GAA gene. The effect is such that it allows for some normally spliced transcript to be produced, which is thought to contribute to its role in late-onset as opposed to early-onset Pompe disease (PMID: 7881425, 2510307).
- For these reasons, this variant has been classified as 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 and are inferred from published carrier frequencies and estimated detection rates based on testing technologies used at Invitae. You can view Invitae's complete Carrier detection rates and residual risks document (containing all carrier genes) online at <https://www.invitae.com/carrier-residual-risks/>. Additionally, the order-specific information for this report is available to download in the portal (under this order's documents) or can be requested by contacting Invitae Client Services. The complete Carrier detection rates and residual risks document will not be applicable for any genes with specimen-specific limitations in sequencing and/or deletion/duplication coverage. Please see the final bullet point in the Limitations section of this report to view if this specimen had any gene-specific coverage gaps.

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	GENE	TRANSCRIPT	GENE	TRANSCRIPT
AAAS	NM_015665.5	AP1S1	NM_001283.3	CBS	NM_000071.2
ABCA12	NM_173076.2	AQP2	NM_000486.5	CC2D1A	NM_017721.5
ABCA3	NM_001089.2	ARG1	NM_000045.3	CC2D2A	NM_001080522.2
ABCA4	NM_000350.2	ARL6	NM_177976.2	CCDC103	NM_213607.2
ABCB11	NM_003742.2	ARSA	NM_000487.5	CCDC39	NM_181426.1
ABCB4	NM_000443.3	ARSB	NM_000046.3	CCDC88C	NM_001080414.3
ABCC2*	NM_000392.4	ASL	NM_000048.3	CD3D	NM_000732.4
ABCC8	NM_000352.4	ASNS	NM_133436.3	CD3E	NM_000733.3
ACAD9	NM_014049.4	ASPA	NM_000049.2	CD40	NM_001250.5
ACADM	NM_000016.5	ASS1	NM_000050.4	CD59	NM_203330.2
ACADVL	NM_000018.3	ATM*	NM_000051.3	CDH23	NM_022124.5
ACAT1	NM_000019.3	ATP6V1B1	NM_001692.3	CEP152	NM_014985.3
ACOX1	NM_004035.6	ATP7B	NM_000053.3	CEP290	NM_025114.3
ACSF3	NM_174917.4	ATP8B1*	NM_005603.4	CERKL	NM_001030311.2
ADA	NM_000022.2	BBS1	NM_024649.4	CFTR*	NM_000492.3
ADAMTS2	NM_014244.4	BBS10	NM_024685.3	CHAT	NM_020549.4
ADAMTSL4	NM_019032.5	BBS12	NM_152618.2	CHRNE	NM_000080.3
ADGRG1	NM_005682.6	BBS2	NM_031885.3	CHRNA	NM_005199.4
ADGRV1	NM_032119.3	BBS4	NM_033028.4	CIITA	NM_000246.3
AGA	NM_000027.3	BBS5	NM_152384.2	CLCN1	NM_000083.2
AGL	NM_000642.2	BBS7	NM_176824.2	CLN3	NM_001042432.1
AGPS	NM_003659.3	BBS9*	NM_198428.2	CLN5	NM_006493.2
AGXT	NM_000030.2	BCKDHA	NM_000709.3	CLN6	NM_017882.2
AHI1	NM_017651.4	BCKDHB	NM_183050.2	CLN8	NM_018941.3
AIPL1*	NM_014336.4	BCS1L	NM_004328.4	CLRN1	NM_174878.2
AIRE	NM_000383.3	BLM	NM_000057.3	CNGB3	NM_019098.4
ALDH3A2	NM_000382.2	BLOC1S3	NM_212550.4	COL11A2*	NM_080680.2
ALDH7A1	NM_001182.4	BLOC1S6	NM_012388.3	COL17A1	NM_000494.3
ALDOB	NM_000035.3	BMP1	NM_006129.4;NM_001199.3	COL27A1	NM_032888.3
ALG1	NM_019109.4	BRIP1	NM_032043.2	COL4A3	NM_000091.4
ALG6	NM_013339.3	BSND	NM_057176.2	COL4A4	NM_000092.4
ALMS1	NM_015120.4	BTBD	NM_000060.3	COL7A1	NM_000094.3
ALPL	NM_000478.5	CAD	NM_004341.4	COX15	NM_004376.6
AMN*	NM_030943.3	CANT1	NM_138793.3	CPS1	NM_001875.4
AMT	NM_000481.3	CAPN3	NM_000070.2	CPT1A	NM_001876.3
ANO10*	NM_018075.3	CASQ2	NM_001232.3	CPT2	NM_000098.2


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GENE	TRANSCRIPT
CRB1	NM_201253.2
CRTAP	NM_006371.4
CTNS	NM_004937.2
CTSA	NM_000308.3
CTSC	NM_001814.5
CTSD	NM_001909.4
CTSK	NM_000396.3
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
CYP7B1	NM_004820.3
DBT	NM_001918.3
DCAF17	NM_025000.3
DCLRE1C	NM_001033855.2
DDX11*	NM_030653.3
DFNB59	NM_001042702.3
DGAT1	NM_012079.5
DGUOK	NM_080916.2
DHCR7	NM_001360.2
DHDDS	NM_024887.3
DLD	NM_000108.4
DLL3	NM_016941.3
DNAH11	NM_001277115.1
DNAH5	NM_001369.2
DNAI1	NM_012144.3
DNAI2	NM_023036.4
DNMT3B	NM_006892.3
DOK7	NM_173660.4
DUOX2*	NM_014080.4
DYNC2H1	NM_001080463.1
DYSF	NM_003494.3
EIF2AK3	NM_004836.6

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
EPG5	NM_020964.2
ERCC2	NM_000400.3
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
FAH*	NM_000137.2
FAM161A	NM_001201543.1
FANCA	NM_000135.2
FANCC	NM_000136.2
FANCD2*	NM_033084.3
FANCE	NM_021922.2
FANCG	NM_004629.1
FANCI	NM_001113378.1
FANCL*	NM_018062.3
FBP1	NM_000507.3
FBXO7	NM_012179.3
FH*	NM_000143.3
FKBP10	NM_021939.3
FKRP	NM_024301.4
FKTN	NM_001079802.1
FMO3	NM_006894.6
FOXN1	NM_003593.2
FOXRED1	NM_017547.3
FRAS1	NM_025074.6
FREM2	NM_207361.5

GENE	TRANSCRIPT
FUCA1	NM_000147.4
G6PC	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
GBA*	NM_001005741.2
GBE1	NM_000158.3
GCDH	NM_000159.3
GCH1	NM_000161.2
GDF5	NM_000557.4
GFM1	NM_024996.5
GHR*	NM_000163.4
GJB2	NM_004004.5
GLB1	NM_000404.2
GLDC	NM_000170.2
GLE1	NM_001003722.1
GNE*	NM_001128227.2
GNPAT	NM_014236.3
GNPTAB	NM_024312.4
GNPTG	NM_032520.4
GNS	NM_002076.3
GORAB	NM_152281.2
GRHPR	NM_012203.1
GRIP1	NM_021150.3
GSS	NM_000178.2
GUCY2D	NM_000180.3
GUSB	NM_000181.3
HADH	NM_005327.4
HADHA	NM_000182.4
HADHB	NM_000183.2
HAMP	NM_021175.2
HAX1	NM_006118.3


Patient name: Donor 7108

DOB: [REDACTED]

Invitae #: [REDACTED]

GENE	TRANSCRIPT
HBA1*	NM_000558.4
HBA2	NM_000517.4
HBB	NM_000518.4
HEXA	NM_000520.4
HEXB	NM_000521.3
HGSNAT	NM_152419.2
HJV	NM_213653.3
HLCS	NM_000411.6
HMGCL	NM_000191.2
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
HSD3B2	NM_000198.3
HYAL1	NM_153281.1
HYLS1	NM_145014.2
IDUA	NM_000203.4
IGHMBP2	NM_002180.2
IKBB	NM_001556.2
IL7R	NM_002185.3
INVS	NM_014425.3
ITGA6	NM_000210.3
ITGB3	NM_000212.2
ITGB4	NM_001005731.2
IVD	NM_002225.3
JAK3	NM_000215.3
KCNJ1	NM_000220.4
KCNJ11	NM_000525.3
LAMA2	NM_000426.3
LAMA3	NM_000227.4
LAMB3	NM_000228.2
LAMC2	NM_005562.2
LARGE1	NM_004737.4

GENE	TRANSCRIPT
LCA5	NM_181714.3
LDLR	NM_000527.4
LDLRAP1	NM_015627.2
LHX3	NM_014564.4
LIFR*	NM_002310.5
LIG4	NM_002312.3
LIPA	NM_000235.3
LMBRD1	NM_018368.3
LOXHD1	NM_144612.6
LPL	NM_000237.2
LRAT	NM_004744.4
LRP2	NM_004525.2
LRPPRC	NM_133259.3
LYST	NM_000081.3
MAK	NM_001242957.2
MAN2B1	NM_000528.3
MANBA	NM_005908.3
MCEE	NM_032601.3
MCOLN1	NM_020533.2
MCPH1	NM_024596.4
MECR	NM_016011.3
MED17	NM_004268.4
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
MOCS1	NM_001358530.2
MOCS2A	NM_176806.3
MOCS2B	NM_004531.4
MPI	NM_002435.2
MPL	NM_005373.2
MPV17	NM_002437.4
MRE11	NM_005591.3

GENE	TRANSCRIPT
MTHFR*	NM_005957.4
MTR	NM_000254.2
MTRR	NM_002454.2
MTTP	NM_000253.3
MUSK	NM_005592.3
MUT	NM_000255.3
MVK	NM_000431.3
MYO15A	NM_016239.3
MYO7A	NM_000260.3
NAGA	NM_000262.2
NAGLU	NM_000263.3
NAGS	NM_153006.2
NBN	NM_002485.4
NCF2	NM_000433.3
NDRG1	NM_006096.3
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
NPC1	NM_000271.4
NPC2	NM_006432.3
NPHP1	NM_000272.3
NPHS1	NM_004646.3
NPHS2	NM_014625.3
NR2E3	NM_014249.3
NSMCE3	NM_138704.3
NTRK1	NM_001012331.1
OAT*	NM_000274.3
OCA2	NM_000275.2
OPA3	NM_025136.3
OSTM1	NM_014028.3
OTOA*	NM_144672.3
OTOF	NM_194248.2;NM_194323.2
P3H1	NM_022356.3

GENE	TRANSCRIPT
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
PDHB	NM_000925.3
PEPD	NM_000285.3
PET100	NM_001171155.1
PEX1*	NM_000466.2
PEX10	NM_153818.1
PEX12	NM_000286.2
PEX13	NM_002618.3
PEX16	NM_004813.2
PEX2	NM_000318.2
PEX26	NM_017929.5
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_00103183.5.2
PHKG2	NM_000294.2
PHYH	NM_006214.3
PIGN	NM_176787.4
PKHD1*	NM_138694.3
PLA2G6	NM_003560.2
PLEKHG5	NM_020631.4
PLOD1	NM_000302.3
PMM2	NM_000303.2
PNPO	NM_018129.3
POLG	NM_002693.2
POLH	NM_006502.2
POMGNT1	NM_017739.3
POMT1	NM_007171.3
POMT2	NM_013382.5

GENE	TRANSCRIPT
POR	NM_000941.2
POU1F1	NM_000306.3
PPT1	NM_000310.3
PRCD	NM_001077620.2
PRDM5	NM_018699.3
PRF1	NM_001083116.1
PROP1	NM_006261.4
PSAP	NM_002778.3
PTPRC*	NM_002838.4
PTS	NM_000317.2
PUS1	NM_025215.5
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
RDH12	NM_152443.2
RLBP1	NM_000326.4
RMRP	NR_003051.3
RNASEH2A	NM_006397.2
RNASEH2B	NM_024570.3
RNASEH2C	NM_032193.3
RPE65	NM_000329.2
RPGRIP1L	NM_015272.2
RTEL1	NM_001283009.1
RXYLT1	NM_014254.2
RYR1	NM_000540.2
SACS	NM_014363.5
SAMD9	NM_017654.3
SAMHD1	NM_015474.3
SCO2	NM_005138.2
SEC23B	NM_006363.4
SEPSECS	NM_016955.3
SGCA	NM_000023.2
SGCB	NM_000232.4
SGCD	NM_000337.5
SGCG	NM_000231.2

GENE	TRANSCRIPT
SGSH	NM_000199.3
SKIV2L	NM_006929.4
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
SLC35A3	NM_012243.2
SLC37A4	NM_001164277.1
SLC38A8	NM_001080442.2
SLC39A4	NM_130849.3
SLC45A2	NM_016180.4
SLC4A11	NM_032034.3
SLC5A5	NM_000453.2
SLC7A7	NM_001126106.2
SMARCA1	NM_014140.3
SMN1*	NM_000344.3
SMPD1	NM_000543.4
SNAP29	NM_004782.3
SPG11	NM_025137.3
SPR	NM_003124.4
SRD5A2	NM_000348.3
ST3GAL5	NM_003896.3
STAR	NM_000349.2
STX11	NM_003764.3
STXBP2	NM_006949.3
SUMF1	NM_182760.3
SUOX	NM_000456.2
SURF1	NM_003172.3


Patient name: Donor 7108

DOB: [REDACTED]

Invitae #: [REDACTED]

GENE	TRANSCRIPT
SYNE4	NM_001039876.2
TANGO2	NM_152906.6
TAT	NM_000353.2
TBCD	NM_005993.4
TBCE*	NM_003193.4
TCIRG1	NM_006019.3
TCN2	NM_000355.3
TECPR2	NM_014844.3
TERT	NM_198253.2
TF	NM_001063.3
TFR2	NM_003227.3
TG*	NM_003235.4
TGM1	NM_000359.2
TH	NM_199292.2
TK2	NM_004614.4
TMC1	NM_138691.2
TMEM216	NM_001173990.2
TMEM67	NM_153704.5
TMPRSS3	NM_024022.2
TPO	NM_000547.5
TPP1	NM_000391.3
TREX1	NM_033629.4
TRIM32	NM_012210.3
TRIM37	NM_015294.4
TRMU	NM_018006.4
TSEN54	NM_207346.2
TSFM*	NM_001172696.1
TSHB	NM_000549.4
TSHR	NM_000369.2
TTC37	NM_014639.3
TTPA	NM_000370.3
TULP1	NM_003322.4
TYMP	NM_001953.4
TYR*	NM_000372.4
TYRP1	NM_000550.2
UBR1	NM_174916.2
UNC13D	NM_199242.2
USH1C*	NM_005709.3
USH2A	NM_206933.2

GENE	TRANSCRIPT
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
WISP3	NM_003880.3
WNT10A	NM_025216.2
WRN*	NM_000553.4
XPA	NM_000380.3
XPC	NM_004628.4
ZBTB24	NM_014797.2
ZFYVE26	NM_015346.3
ZNF469	NM_001127464.2

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 $\geq 50\times$ 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 section. 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. Invitae utilizes a classification methodology to identify next-generation sequencing (NGS)-detected variants that require orthogonal confirmation (Lincoln, et al. J Mol Diagn. 2019 Mar;21(2):318-329.). Confirmation of the presence and location of reportable variants is performed based on stringent criteria established by Invitae (1400 16th Street, San Francisco, CA 94103, #05D2040778), as needed, using one of several validated orthogonal approaches (PubMed ID 30610921). The following analyses are performed if relevant to the requisition. For GBA 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 GBA, 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. In some cases, it may not be possible to disambiguate between the gene and pseudogene. 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 which generates diplotype calls. If sequence data for a sample does not support a unique high confidence match from among hypotheses tested, that sample is flagged for manual review. Copy number variation is only reported for coding sequence of HBA1 and HBA2 and the HS-40 region. This assay does not distinguish among the $\alpha 3.7$ subtypes, and all $\alpha 3.7$ variants are called as HBA1 deletions. This assay may not detect overlapping copy gain and copy loss events when the breakpoints of those events are similar. For FMR1, triplet repeats are detected by 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. Technical component of confirmatory sequencing is performed by Invitae Corporation (1400 16th Street, San Francisco, CA 94103, #05D2040778).
- 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 are not included in this report. However, if additional evidence becomes available to indicate that the clinical significance of a variant has changed, Invitae may update this report and provide notification.
- 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 (<http://exac.broadinstitute.org>), gnomAD (<http://gnomad.broadinstitute.org>), and dbSNP (<http://ncbi.nlm.nih.gov/SNP>).

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 Invitae. It has not been cleared or approved by the FDA. The laboratory is regulated under the Clinical Laboratory Improvement Act (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 exon-level deletions and duplications. Invitae's methods also detect insertions and deletions larger than 15bp but smaller than a full exon but sensitivity for these may be marginally reduced. Invitae's deletion/duplication analysis determines copy number at a single exon resolution at virtually all targeted exons. However, in rare situations, single-exon copy number events may not be analyzed 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 not be detected. 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.
- TBCE:** Sequencing analysis for exons 2 includes only cds +/- 10 bp. **BBS9:** Deletion/duplication analysis is not offered for exon 4. **SMN1:** Systematic exon numbering is used for all genes, including SMN1, and for this reason the exon typically referred to as exon 7 in the literature (PMID: 8838816) is referred to as exon 8 in this report. This assay unambiguously detects SMN1 exon 8 copy number. The presence of the g.27134T>G variant (also known as c.*3+80T>G) is reported if SMN1 copy number = 2. SMN1 or SMN2: NM_000344.3:c.*3+80T>G variant only. **AMN:** Deletion/duplication analysis is not offered for exon 1. **MLC1:** Sequencing analysis for exons 11 includes only cds +/- 10 bp. **CFTR:** Sequencing analysis for exons 7 includes only cds +/- 10 bp. **PTPRC:** Sequencing analysis is not offered for exons 3, 15. **DUOX2:** Deletion/duplication and sequencing analysis is not offered for exons 6-7. **USH1C:** Deletion/duplication analysis is not offered for exons 5-6. **VPS53:** Sequencing analysis for exons 14 includes only cds +/- 5 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. **CYP21A2:** Analysis includes the most common variants (c.92C>T (p.Pro31Leu), c.293-13C>G (intronic), c.332_339delGAGACTAC (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.923dupT (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. Full gene deletion analysis is not offered. Sensitivity to detect these variants, if they result from complex gene conversion/fusion events, may be reduced. **EYS:** Sequencing analysis for exons 30 includes only cds +/- 0 bp. **GNE:** Sequencing analysis for exons 8 includes only cds +/- 10 bp. **LIFR:** Sequencing analysis for exons 3 includes only cds +/- 5 bp. **OAT:** Deletion/duplication analysis is not offered for exon 2. **PEX1:** Sequencing analysis for exons 16 includes only cds +/- 0 bp. **PKHD1:** Deletion/duplication analysis is not offered for exon 13. **COL11A2:** Deletion/duplication analysis is not offered for exon 36. **ANO10:** Sequencing analysis for exons 8 includes only cds +/- 0 bp. **GHR:** Deletion/duplication and sequencing analysis is not offered for exon 3. **TYR:** Deletion/duplication and sequencing analysis is not offered for exon 5. **FANCL:** Sequencing analysis for exons 4, 10 includes only cds +/- 10 bp. **FAH:** Deletion/duplication analysis is not offered for exon 14. **FH:** Sequencing analysis for exons 9 includes only cds +/- 10 bp. **TSFM:** Sequencing analysis is not offered for exon 5. **GALE:** Sequencing analysis for exons 10 includes only cds +/- 5 bp. **WRN:** Deletion/duplication analysis is not offered for exons 10-11. Sequencing analysis for exons 8, 10-11 includes only cds +/- 10 bp. **ATM:** Sequencing analysis for exons 6, 24, 43 includes only cds +/- 10 bp. **VPS13A:** Deletion/duplication analysis is not offered for exons 2-3, 27-28. **NEB:** Deletion/duplication analysis is not offered for exons 82-105. NEB variants in this region with no evidence towards pathogenicity are not included in this report, but are available upon request. **ATP8B1:** Sequencing analysis for exons 19 includes only cds +/- 10 bp. **AIPL1:** Sequencing analysis for exons 2 includes only cds +/- 10 bp. **DDX11:** NM_030653.3:c.1763-1G>C variant only. **ABCC2:** Deletion/duplication analysis is not offered for exons 24-25. **OTOA:** Deletion/duplication and sequencing analysis is not offered for exons 20-28. **TG:** Deletion/duplication analysis is not offered for exon 18. Sequencing analysis for exons 44 includes only cds +/- 0 bp. **GALC:** Deletion/duplication analysis is not offered for exon 6. **GBA:** c.84dupG (p.Leu29Alafs*18), c.115+1G>A (Splice donor), c.222_224delTAC (p.Thr75del), c.475C>T (p.Arg159Trp), c.595_596delCT (p.Leu199Aspfs*62), c.680A>G (p.Asn227Ser), c.721G>A (p.Gly241Arg), c.754T>A (p.Phe252Ile), c.1226A>G (p.Asn409Ser), c.1246G>A (p.Gly416Ser), c.1263_1317del (p.Leu422Profs*4), c.1297G>T (p.Val433Leu), c.1342G>C (p.Asp448His), c.1343A>T (p.Asp448Val), c.1448T>C (p.Leu483Pro), c.1504C>T (p.Arg502Cys), c.1505G>A (p.Arg502His), c.1603C>T (p.Arg535Cys), c.1604G>A (p.Arg535His) variants only. Rarely, sensitivity to detect these variants may be reduced. When sensitivity is reduced, zygosity may be reported as "unknown". **HBA1/2:** This assay is designed to detect deletions and duplications of HBA1 and/or HBA2, resulting from the -alpha20.5, --MED, --SEA, --FIL/--THAI, -alpha3.7, -alpha4.2, anti3.7 and anti4.2. Sensitivity to detect other copy number variants may be reduced. Detection of overlapping deletion and duplication events will be limited to combinations of events with significantly differing boundaries. In addition, deletion of the enhancer element HS-40 and the sequence variant,



Patient name: Donor 7108

DOB:

Invitae #:

Constant Spring (NM_000517.4:c.427T>C), can be identified by this assay. 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 in our primary report.

This report has been reviewed and approved by:



Matteo Vatta, Ph.D., FACMG
Clinical Molecular Geneticist

Client/Sending Facility:
FAIRFAX CRYOBANK [REDACTED]

LCLS Specimen Number: [REDACTED]

Patient Name: 7108, DONOR

Date of Birth: [REDACTED]

Gender: M

Patient ID: [REDACTED]

Lab Number: [REDACTED]

Indications: NOT GIVEN

Account Number: [REDACTED]

Ordering Physician: [REDACTED]

Specimen Type: BLOOD

Client Reference:

Date Collected: 02/11/2023

Date Received: 02/14/2023

Date Reported: 03/07/2023

Test: Chromosome, Blood, Routine

Cells Counted: 20

Cells Analyzed: 20

Cells Karyotyped: 2

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 GTG 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.

LCLS Specimen Number: [REDACTED]
Patient Name: 7108, DONOR
Date of Birth: [REDACTED]
Gender: M
Patient ID: [REDACTED]
Lab Number: [REDACTED]

Account Number: [REDACTED]
Ordering Physician: [REDACTED]
Specimen Type: BLOOD
Client Reference:
Date Collected: 02/11/2023
Date Received: 02/14/2023





Client/Sending Facility:
FAIRFAX CRYOBANK

LCLS Specimen Number:

Patient Name: 7108, DONOR

Date of Birth:

Gender: M

Patient ID:

Lab Number:

Account Number:

Ordering Physician:

Specimen Type: BLOOD

Client Reference:

Date Collected: 02/11/2023

Date Received: 02/14/2023

Changqing Xia, PhD

Anjen Chenn, M.D., Ph.D.
Medical Director

Technical component performed by Laboratory Corporation of America Holdings,
1904 TW Alexander Drive, RTP, NC, 27709-0153 (800) 345-4363

Professional Component performed by LabCorp CLIA 34D1008914, 1904 TW Alexander Dr, Research Triangle Park, NC 27709. Medical Director, Anjen Chenn, M.D., Ph.D.
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If you have received this document in error, please call 800-533-0567.

Patient ID: [REDACTED]
Specimen ID: [REDACTED]

DOB: [REDACTED]
Age: [REDACTED]
Sex: Male

Patient Report

Account Number: [REDACTED]
Ordering Physician: [REDACTED]



Date Collected: 02/11/2023

Date Received: 02/11/2023

Date Reported: 03/07/2023

Fasting: No

Ordered Items: **CBC With Differential/Platelet; Chromosome, Blood, Routine; Hgb Fractionation Cascade;**

Date Collected: 02/11/2023

CBC With Differential/Platelet

[illegible]

Chromosome, Blood, Routine

Test	Current Result and Flag	Previous Result and Date	Units	Reference Interval
Specimen Type ⁰²	Comment: BLOOD			
Cells Counted ⁰²	20			
Cells Analyzed ⁰²	20			
Cells Karyotyped ⁰²	2			
GTG Band Resolution Achieved ⁰²	500			
Cytogenetic Result ⁰²	Comment: 46, XY			
Interpretation ⁰²	Comment: NORMAL MALE KARYOTYPE Cytogenetic analysis of PHA stimulated cultures has revealed a MALE karyotype with an apparently normal GTG banding pattern in all cells observed. This result does not exclude the possibility of subtle			



7108, Donor

Patient ID:

Specimen ID

DOB:

Age:

Sex: Male

Patient Report

Account Number:

Ordering Physician



Date Collected: 02/11/2023

Chromosome, Blood, Routine (Cont.)

rearrangements below the resolution of cytogenetics or congenital anomalies due to other etiologies.

Director Review:⁰²

Comment:

Changqing Xia, PhD

PDF

Hgb Fractionation Cascade

Test	Current Result and Flag	Previous Result and Date	Units	Reference Interval
Hgb Fractionation by CE: ⁰¹				
Hgb F ⁰¹	0.0		%	0.0-2.0
Hgb A ⁰¹	97.6		%	96.4-98.8
Hgb A2 ⁰¹	2.4		%	1.8-3.2
Hgb S ⁰¹	0.0		%	0.0

Interpretation:⁰¹

Normal hemoglobin present; no hemoglobin variant or beta thalassemia identified.

Note: Alpha thalassemia may not be detected by the Hgb Fractionation Cascade panel. If alpha thalassemia is suspected, Labcorp offers Alpha-Thalassemia DNA Analysis (#511172).

Disclaimer

The Previous Result is listed for the most recent test performed by Labcorp in the past 5 years where there is sufficient patient demographic data to match the result to the patient. Results from certain tests are excluded from the Previous Result display.

Icon Legend

▲ Out of Reference Range ■ Critical or Alert

Performing Labs

01: TA - Labcorp Tampa 5610 W LaSalle Street, Tampa, FL, 33607-1770 Dir: Sean Farrier, MD

02: YU - Labcorp RTP 1904 TW Alexander Drive Ste C, RTP, NC, 27709-0153 Dir: Anjen Chenn, MDPhD

For Inquiries, the physician may contact Branch: 713-856-8288 Lab: 800-877-5227

Patient Details**7108, Donor**

Phone:

Date of Birth:

Age:

Sex: Male

Patient ID:

Alternate Patient ID:

Physician Details**Specimen Details**

Specimen ID:

Control ID:

Alternate Control Number:

Date Collected: 02/11/2023 1202 Local

Date Received: 02/11/2023 0000 ET

Date Entered: 02/11/2023 1304 ET

Date Reported: 03/07/2023 1510 ET

Date Created and Stored 03/07/23 1520 ET **Final Report** Page 2 of 2



DONOR NUMBER:
7108



PERSONAL INFORMATION

TALENTS:	
Voice:	Baritone
Tell us about your interests/talents and how long you have been pursuing. Provide several.	Piano (started early 2020) - My mother always loved "Canon in D" and wished to listen to it on the Piano for her Birthday. I bought a keyboard on Amazon and I played it for her. Competitive E-Sports (started mid-2015) - I played in "contenders" teams that recruited the top 1% of the player population for games like Guild Wars and Overwatch. While I do not play in the pro leagues anymore, I play casually (3-5 hours a month).

FAVORITE:	
Color	Blue
Food	Ribeye Steak
Music	Rock and Roll (1950s - 1990s)
Animal	Dogs (Labradors)
Pet	Mixed Labrador
Car	Tesla
Movie	Star Wars - Empire Strikes Back
Song	Johnny B. Goode

GOALS:	
Academic	EdD in Organizational Leadership (expected graduation 2026)
Professional	Private School Headmaster

RELIGION:

Faith

Other

Denomination

Agnostic theism

LANGUAGES:

Speak

English and Spanish

SPORTS:

Play

N/A

Watch

N/A

SIBLINGS:

Are you or any of your siblings twins?
Please specify who and if they are
fraternal or identical

No

ACADEMICS

STANDARD TEST SCORE: PROVIDE SCORE OR DENOTE N/A IF NOT TAKEN

SAT
on scale of

1400 - 1600

ACT
on scale of

N/A - N/A

GRE
on scale of

N/A - N/A

MCAT
on scale of

N/A - N/A

LSAT
on scale of

N/A - N/A

GMAT

N/A - N/A

on scale of	
TOEFL on scale of	N/A - N/A
Other on scale of	N/A - N/A

HIGH SCHOOL		
GPA on scale of	3.8	4.0

COLLEGE		
GPA on scale of	2.92	4.0

GRADUATE SCHOOL		
GPA on scale of	4.0	4.0

SUBJECTS STUDIED:	
Major subjects	Science and Education
Minor subjects	Psychology

DEGREES	
Bachelor's	Biology
Master's	Science Education
Doctorate	EdD/Organizational Leadership
Other(s)	N/A

AWARDS & ACTIVITIES	
High school	FBLA (Future Business Leaders of America) and HOSA (Future Health Professionals) competitions

College	Mostly HOSA public speaking competitions
Academic	HOSA Advisor
Professional	Advisor of the Year, Alumni of the Year, Outstanding Educator Award
Political	Left Leaning Centrist
Personal	Piano, E-Sports
Charitable	HOSA, STEM Education
Others	Bacterial Antibiotic Research



MILITARY

Served in military	No
Branch	N/A
Rank	N/A
Years of service	N/A



PHYSICAL FEATURES

Hands	Right-Handed
Fingers	Medium



SIZES & MEASUREMENTS

Neck (inches)	14
Chest (inches)	38
Inseam (inches)	30
Waist (inches)	35

Sleeve (inches)

32

Hat size

7 ¾

Shoe size

10



PHYSICAL AIDS



EYES:

Vision Near-Sighted

Glasses Single - -6.0/-6.0

Astigmatism No

Age started wearing glasses/contacts 12

Laser surgery? No



DENTAL:

Device Retainer

Reason Cosmetic

Age range during use Braces finished around 17. Used retainer every night since then



OTHER PHYSICAL AIDS:

List N/A

Reason/cause N/A



FACIAL FEATURES



EYES:

Set	 Narrow	 Average	 Wide
Size	 Small	 Average	 Large
Shape	 Almond	 Oval	 Round
Color	- Black - Blue - Green - Hazel - Brown		
Shade	- Light - Medium - Dark		

EYEBROWS:

Arch	■ Flat ■ Medium ■ High
Thickness	 Thin  Average  Thick
Set	

- Narrow
- Average
- Wide

NOSE:

Size

- Small
- Medium
- Large

Shape



Round



Straight



Wide

Width

- Narrow
- Average
- Wide

Length

- Short
- Average
- Long

Bridge



Convex



Straight



Concave

Nostril flare

- Small
- Average
- Wide

CHEEBONES:

Set

- Low
- Average
- High

MOUTH:

	Small	Average	Large
Size			
	Thin	Average	Full
Lips			
	☐ Downward ☒ Straight ☐ Upward		
Turn			

TEETH:

	Average	Large	Small
Size			
	☐ Underbite ☒ Even ☐ Overbite		
Set			
Gap	No		
	☐ Poor ☐ Fair ☒ Good		
Condition			

CHIN:

	Round	Oval	Square
Shape			

Prominence

- Slight
- Average
- Strong

Cleft



None



Slight



Strong

EARS:

Size



Small



Average



Large

Set

- Low
- Average
- High

Angle

- Close
- Average
- Far

Lobe size

- Small
- Medium
- Large

Lobe attachment



Detached



Attached

HAIR:

Color at birth

	Blond Brown Auburn Red Black grey
Natural color in adulthood	Blond Brown Auburn Red Black grey
Shade	Light Medium Dark
Type	Straight Wavy Curly
Fullness	Thin Medium Thick
Texture	Fine Coarse Medium

SKIN:

Tone

[More about skin tones](#)

	- Ivory - Porcelain - Pale ivory - Warm ivory - Sand - Rose beige - Limestone - Beige - Sienna - Honey - Band - Almond - Chestnut - Bronze - Umber - Golden - Espresso - Chocolate
Tan ability	- None - Slight - Medium - Easy
Condition	- Oily - Medium - Combination - Dry

HAIRLINE:	
Forehead	- Low - Average - High
Contour	- Straight - Slight Curve - Widow Peak

OTHER FACIAL FEATURES:	
Moles	

	None One Several
Freckles	None Several Moderate Numerous
Dimples	None Slight Medium Deep
Adam's apple	Slight Medium Strong

FACIAL HAIR

Thickness	Thin Medium Dense
Color	Auburn Black Blond Brown Grey Red
Shade	Light Medium Dark

FAMILY INFORMATION

DONOR'S PARENTS

Donor's Father Personal Information

Occupation	Produce Distributor
------------	---------------------

Donor's Father Education:

Level	High school diploma
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Degree	Other
--------	-------

Subject	General Studies
---------	-----------------

Donor's Father Religion:

Faith	Christian
-------	-----------

Denomination	Non-practicing
--------------	----------------

Donor's Father Languages:

Speak	English, Spanish
-------	------------------

Donor's Father Talents:

Voice	Bass
-------	------

Instrument	N/A
------------	-----

Other	N/A
-------	-----

Donor's Father Physical Information

Body

Age	49
-----	----

Height	6' 0"
--------	-------

Weight	280 lbs.
--------	----------

Hands	Right-Handed
-------	--------------

Donor's Father Facial Features

Eyes:

Eye Color	Brown
-----------	-------

Eye shade	Light
Corrective Lenses	No

— Hair:

Original hair color	Brown
Type	Wavy

— Skin:

Skin Tone More about skin tones	Honey
--	-------

— Other:

Chin cleft	Slight
Ear lobe	Detached
Facial Dimples	None

— Donor's Mother Personal Information

Occupation	Homemaker
------------	-----------

— Donor's Mother Education:

Level	College
Degree	Bachelor
Subject	Business Management

— Donor's Mother Religion:

Faith	Christian
Denomination	Non-practicing

— Donor's Mother Languages:

Speak	English and Spanish
-------	---------------------

Donor's Mother Talents:

Voice	Soprano
Instrument	N/A
Other	N/A

Donor's Mother Physical Information

Body

Age	52
Height	5' 7"
Weight	140 lbs.
Hands	Right-Handed

Facial Features

Eyes:

Eye Color	Brown
Eye shade	Light
Corrective Lenses	No

Hair:

Original hair color	Brown
Type	Straight

Skin:

Skin Tone More about skin tones	Porcelain
--	-----------

Other:

Chin cleft	Slight
Ear lobe	Detached
Facial Dimples	None

DONOR'S SIBLINGS

Donor's Sibling(s) (1-4)

Donor's Sibling #1

Full/Half sibling Full

Sibling #1 Physical Information

Body

Age 21

Height 5' 11"

Weight 176 lbs.

Hands Right-Handed

Sibling #1 Personal Information

Occupation Produce Distributor (Works in family business)

Education:

Level College

Degree Bachelor

Subject Business Management

Languages:

Speak English and Spanish

Talents:

Voice Baritone

Instrument N/A

Other N/A

Sibling #1 Children:

Boys 0
Ages:

Girls 0
Ages:

Sibling #1 Facial Features

Eyes:

Eye color Brown

Eye shade Light

Corrective Lenses Yes

Hair:

Original hair color Brown

Type Straight

Skin:

Skin tone Ivory
[More about skin tones](#)

Other:

Chin cleft Slight

Ear lobe Detached

Facial dimples None

FATHER'S SIBLINGS

Father's Brothers

of Brothers 3

Ages of Brothers: 55, 52, 35

Education levels for each: High School, High School, High School

Occupations for each: Taxi Driver, Taxi Driver, Accountant

Father's Brothers' Total Children:

Boys 3

Girls 6

Father's Sisters

of Sisters

4

Ages of Sisters:

64, 61, 59, 51

Education levels for each:

High School, High School, High School, High School

Occupations for each:

Homemaker, Homemaker, Homemaker, Homemaker

Father's Sisters' Total Children:

Boys

5

Girls

7

MOTHER'S SIBLINGS

Mother's Brothers

Brothers

2

Ages of Brothers:

48, 37

Education levels for each:

Some High School, High School

Occupations for each:

Car Sales, Hot Dog Vendor

Mother's Brothers' Total Children:

Boys

4

Girls

2

FATHER'S PARENTS

Father's Father

Education

Level

High school diploma

Degree

Other

Occupation:

Taxi Car Company Owner

Siblings:

Brothers

1

Sisters

1

Other:	
Languages	Spanish
Religion	Christian

Father's Mother	
Education	
Level	High school diploma
Degree	Other
Occupation:	Homemaker

Siblings:	
# Brothers	1
# Sisters	2

Other:	
Languages	Spanish
Religion	Christian

MOTHER'S PARENTS

Mother's Father	
Education	
Level	Some college
Degree	Bachelor
Occupation:	Car Sales Company Owner

Siblings:	
# Brothers	2
# Sisters	4

Other:	
--------	--

Languages	Spanish
Religion	Christian

— Mother's Mother

— Education

Level	Some college
Degree	Bachelor
Occupation:	Nurse Aid

— Siblings:

# Brothers	1
# Sisters	1

— Other:

Languages	Spanish, English
Religion	Christian



DONOR NUMBER:
7108

PHYSICAL APPEARANCE:

Donor 7108 is an attractive guy, with a head full of thick brown hair and warm green eyes that are enhanced by his golden skin. He has an incredible smile that conveys his friendly nature, along with a confident voice and infectious laugh. He has a slender build and is always nicely dressed for his role as an educator in stylish business-casual attire.

PERSONALITY:

Donor 7108 exudes positivity, confidence, and ambition. He is very outspoken about issues that are important to him. He is respectful of others and has excellent manners. He is a happy person and conversations with him are generally cheerful in nature. This donor is very passionate about his career as a teacher and loves working in the field of education, where he is pursuing a doctorate degree.



DONOR NUMBER:
7108



ESSAY QUESTIONS

What is your most memorable childhood experience?	I vividly remember my mother home-schooling me before fourth grade. She was determined to give me the best education possible and would spend hours teaching me relatively advanced math and science. She was extremely patient with all the questions I would ask her, even when she didn't have the answers. If I passed all her oral exams, my father would then agree to play the first Spiderman game on the PlayStation 1 with me. Despite it being just a game, he would consistently compete against me, using the pain of defeat to drive me to improve. It wouldn't be until my teenage years that I would experience the bittersweet moment of victory against my dad. As a result of their outstanding tutelage, the mentality of "Work Hard, Play Hard" was engraved in my personality. This inspired my ambition in my professional career to go into educational leadership and my drive for success in hobbies like competitive online games.
What immediate family member do you closely identify with and why?	My mom has always been there for me, providing guidance and support throughout my whole life. She taught me the value of effective study habits, morality, and being passionate about anything I put my mind to. She always had a vision of raising an ambitious and strong son, but was still compassionate. She had worked with and seen several successful men become powerful, but lack basic empathy toward others. Due to this vision, I turned out as she describes: 'Un Buen hombre'.
What character traits do you admire in an individual?	The character traits I admire most in an individual are aspiration, adaptability, and kindness. These character traits show that an individual is well-rounded, capable of success, and most importantly, helps to make the world a better place. Aspiration allows individuals to unlock their potential and reach the limits of their abilities. Adaptability allows individuals to thrive in any situation, handling change no matter how serious a problem might appear. Kindness ensures that this individual uses their abilities for the best reasons, improving the world, one person at a time. Overall, the best character traits I admire are any that ensure a person is responsible, resilient, and empathetic, since these traits help an individual be successful in their personal and professional lives.
What is the funniest thing ever to happen to you?	Dating for the first time in High School is always an anxiety-inducing experience. One of the funniest moments I have experienced is when I was invited over to my crush's house for the first time. It was a typical Friday night and we had both spent hours getting ready to see each other. I remember Kiara's mom telling me how she spend days picking the perfect outfit and organizing all the make-up needed. As I arrived through the front door, Kiara's pet Parrot, Peckers, flew down from his home and landed on my shoulder. The initial sharp pain of his claws immediately went away as he adjusted himself as if he knew he could hurt others. I didn't mind the company as we sat down to watch movies and eat. However, things took a turn for the mischievous as the Peckers announced he was bored. "I can't believe Kiara loves 'my name' and wants to get married!" Both our faces turned red as we tried to laugh it off. Immediately after he comments, "Unless she still wants to marry her old English teacher!" Kiara started shooping away the bird, "Go away you Parrotted piece of crap". She chased the bird around the house as Peckers squawked, almost as if he was enjoying the hunt. Eventually, he flew back to his cage and said, "You are sorry". She was overwhelmed with anger as she returned to me, only to eventually redirect her frustration to me as I couldn't stop laughing at the situation.

If time and money were not an issue, where would you travel and why?	If finances and time were not an issue, I would love to go to Tokyo. The location is renowned for its wonderful traditional Japanese cuisine and international foods. It is also a dramatically different and new culture I would expose to, it's not every day that one can have countless temples, shrines, and museums to explore. I also always wanted to witness sumo wrestling and the kabuki theaters in person. Additionally, the location is tourist friendly because it has amazing and reliable public transportation, making it easy to go anywhere without expansive planning. Ultimately, I love trying out new and tasty food, I would travel anywhere that is known for excellent meals.
When and if you ever have children, what would you like to pass on to them?	As a parent, the final goal for my children is that they grow up to be confident, self-sufficient, kind, and adaptive individuals. This ensures that they are ready to effectively face any challenge in their professional or personal lives and thrive while they are at it. This can be achieved by passing on life skills, essential values, and good habits through love and support. Teaching children life skills like cooking, finances, and home maintenance allows them to become independent and self-effective. Essential values like learning the importance of education, empathy, and ambition for success carves a path to a successful career. Teaching children effective habits like exercise/sleep schedules and conflict resolution skills will help them create a healthy personal life. Most importantly, I need my children to use these skills wisely. That is why ensuring they grow up in a loving and compassionate environment is vital for a strong sense of self-worth and morality.

Donor # 7108 Personality Type: ENTJ

Our donors have completed a personality test based on the work of Isabel Briggs Myers, Katherine Cook Briggs and originator of the system, Carl Jung.

THE TEST MEASURES PREFERENCES FOR:

Extrovert	Sensing	Thinking	Judging
E	S	T	J
vs	vs	vs	vs
I	N	F	P
Introvert	iNtuitive	Feeling	Perceiving

THE COMMANDER

E | N | T | J

An ENTJ's primary mode of living focuses on external aspects and all things are dealt with rationally and logically. Their secondary mode of operation is internal, where intuition and reasoning take effect. ENTJs are natural born leaders and like being in charge. They live in a world of possibilities and they often see challenges and obstacles as great opportunities to push themselves. The ENTJ personality seems to have a natural gift for leadership, making decisions, and considering options and ideas quickly yet carefully. They are "take charge" people who do not like to sit still.

Being extroverts, people of this personality type like to be with people and need connections and contact with others, but they can find it challenging to get really close to someone or to truly understand what other people are thinking or feeling. ENTJs can have difficulty seeing things from outside their own perspective.

Common traits:

Natural born leader and likes being in charge

Lives in a world of possibilities

Project-oriented and focused on the task at hand

Logical and thoughtful before taking action

Can be a bit of a perfectionist

Well-developed communication skills

Self-confident and sure of themselves and their abilities

The Lifetime Series of Donor 7108



Age 3



Age 4

From Our Staff

"Donor 7108 exudes positivity, confidence, and ambition. This donor is very passionate about his career as a teacher and loves working in the field of education, where he plans to pursue a doctorate degree."



Age 6

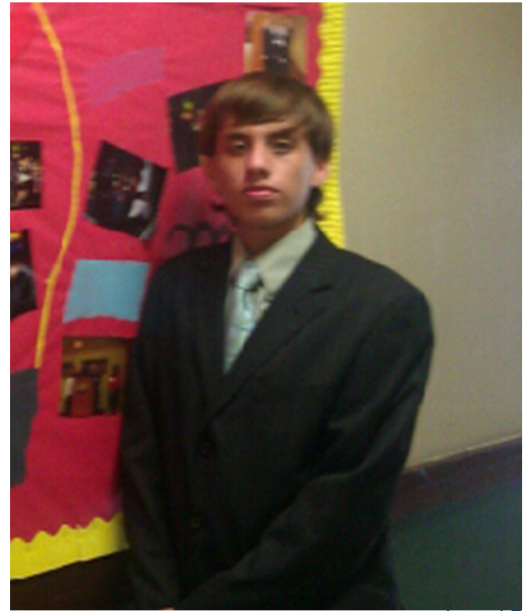
Fairfax Cryobank

The Trusted Choice for Donor Sperm

The Lifetime Series of Donor 7108



Age 12



Age 15



Age 18



Adult