Fairfax Cryobank
The Trusted Choice for Donor Sperm

Summary Profile

DONOR NUMBER:
7240

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:	Weight:	Eye Color:	Hair:	Skin Tone:	Ancestry:
5'8" (172 cm)	159 lb (72 kg)	Blue	Blond/ Straight	Light	Caucasian

Blood Type:	Ethnic Background:
O+	German/Norwegian

Education:	Occupation:
BS/IT Management	IT Lead

Interests: Curling, Cycling, Euphonium, Guitar, Trumpet

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?	No
Allergies (medicines, food, pollens)? If yes, please list substance and reaction caused:	No
CMV IgG Antibody	Positive
CMV IgM Antibody	Negative
Note any comments regarding above items:	N/A

FAMILY MEDICAL HISTORY

See list of questions asked here

YOUR MOTHER

QUESTION	RESPONSE
Current age or age at death	65
Living / Dead	Living
Cause of death and any treatment prior to death	N/A

HEALTH PROBLEMS

DISEASE	AGE DIAGNOSED	TREATMENT FOR CONDITION
Breast cancer	60	Lumpectomy and chemotherapy

YOUR FATHER

QUESTION	RESPONSE
Current age or age at death	68
Living / Dead	Living

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

HEALTH PROBLEMS

DISEASE	AGE DIAGNOSED	TREATMENT FOR CONDITION
High cholesterol	58	Medication management and diet

SISTERS

Your Sister 1

QUESTION	RESPONSE
Current age or age at death	39
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	56
Living / Dead	Dead
Cause of death and any treatment prior to death	Heart Attack

HEALTH PROBLEMS

DISEASE	AGE DIAGNOSED	TREATMENT FOR CONDITION
Other		No other diagnosed health issues at time of death

YOUR MOTHER'S MOTHER

QUESTION	RESPONSE
Current age or age at death	90

QUESTION	RESPONSE
Living / Dead	Dead
Cause of death and any treatment prior to death	Undiagnosed Cancer and Broken Hip

HEALTH PROBLEMS

DISEASE	AGE DIAGNOSED	TREATMENT FOR CONDITION
Other		Cancer was discovered when he underwent surgery for a broken hip, no treatment

YOUR MOTHER'S SISTERS 1

QUESTION	RESPONSE
Current age or age at death	65
Living / Dead	Living
Cause of death and any treatment prior to death	N/A

HEALTH PROBLEMS

Healthy

YOUR MOTHER'S SISTERS 2

QUESTION	RESPONSE
Current age or age at death	60
Living / Dead	Living
Cause of death and any treatment prior to death	N/A

HEALTH PROBLEMS

Healthy

YOUR MOTHER'S BROTHERS 1

QUESTION	RESPONSE
Current age or age at death	63

QUESTION	RESPONSE
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	98
Living / Dead	Dead
Cause of death and any treatment prior to death	Accidental Fall

HEALTH PROBLEMS

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

YOUR FATHER'S MOTHER

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

HEALTH PROBLEMS

DISEASE	AGE DIAGNOSED	TREATMENT FOR CONDITION
Colon cancer	65	Chemotherapy and surgery

YOUR FATHER'S SISTERS 1

QUESTION	RESPONSE

QUESTION	RESPONSE
Current age or age at death	60
Living / Dead	Living
Cause of death and any treatment prior to death	N/A

HEALTH PROBLEMS

Healthy

RELIGION:

Faith	None
Denomination	N/A

UPDATES TO PROFILE

Update Available	N/A
Updates - Personal	N/A
Updates - Medical	N/A
Updates - Family Medical History	N/A

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Personal Profile

DONOR NUMBER:
7240

PERSONAL INFORMATION

TALENTS:

Voice:	Tenor
Tell us about your interests/talents and how long you have been pursuing. Provide several.	(1) Curling: I've been curling for about 20 years and play competitively. I've Played Junior Nationals, Men's Nationals, Mixed Fours Nationals, Club Nationals, College Nationals, and at the World University Games. I play in many tournaments throughout the year and play in a few different leagues during the winter. (2) Downhill skiing: I've been downhill skiing for about 27 years. I love going out west to ski, though I don't get out there enough. (3) Road biking: 2 years ago, I decided to buy a road bike and start riding and fell in love with it. This year my goal is to ride at least 100 miles each month. (4) Working out: I get to the gym to lift or do other exercises 5-6 days a week. (5) Trumpet and Guitar: I don't play my trumpet much these days, but I played steadily from middle school through college so around 12 years. I also play guitar a little and have probably been playing for around 12 years, but not as steadily. It's more of an on again off again thing. (6) Computers and PC Gaming: I've been big into computer building and PC gaming for about 20 years. I've built many computers for myself and for others over the years. I'm also quite interested in enterprise computer infrastructure and have my own home lab made up of enterprise grade servers and networking hardware which I use to tinker and learn. I've turned this hobby into a side business where I consult on and install computers and networking equipment for small businesses and consumers. For PC Gaming, role playing games, such as the Elder Scrolls series and Grand Theft Auto games, are my all-time favorites, but I also enjoy team-based shooters, such as Overwatch, and I also play too much Rocket League. I will occasionally hop into indie games like Stardew Valley, Ori and the Blind Forest, and the Trine series. I get together with a small group of friends a couple times a year for LAN parties. (7) Dungeons and Dragons: I've recently gotten into Dungeons and Dragons with some work friends, and this is becoming a bigger hobby for me.

FAVORITE:

Color	Blue
Food	Pizza
Music	Alt Rock, Post Hardcore, Indy
Animal	Dog
Pet	Dog

Car	Kia Stinger
Movie	Anchorman / Step Brothers
Song	I'm Not Ok by My Chemical Romance

GOALS:

Academic	MBA in IT Management
Professional	Chief Information Officer
Personal	Win a national curling championship

RELIGION:

Faith	None
Denomination	N/A

LANGUAGES:

Speak	English

SPORTS:

Play	Curling, Cycling
Watch	Curling, Football, Soccer, BMX, Skateboarding, Drifting

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	N/A - N/A

on scale of	
ACT on scale of	27 - 36
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 on scale of	N/A - N/A
TOEFL on scale of	N/A - N/A
Other on scale of	N/A - N/A

HIGH SCHOOL

GPA on scale of	3.9	4.0

COLLEGE

GPA on scale of	3.054	4.0

GRADUATE SCHOOL

GPA on scale of	N/A	N/A

SUBJECTS STUDIED:

Major subjects	IT Management
Minor subjects	N/A

DEGREES

Bachelor's	IT Management
Master's	N/A
Doctorate	N/A
Other(s)	N/A

AWARDS & ACTIVITIES

High school	Concert Band, Jazz Band, Pep Band, Marching Band, Drum Major senior year, 1st Chair trumpet junior and senior year, Rocketry Team, Most Improved Curler (freshman year), MVP (Curling Senior Year), Lettered in Curling all 4 years.
College	Curling Club, US Curling Association College Nationals
Academic	National Honor Society
Professional	Certified Salesforce Administrator
Political	N/A
Personal	Many Curling Awards
Charitable	N/A
Others	Cyling, Video Games, PC Building, IT Home Lab (servers, virtual machines, containers, networking, etc.), Dungeons and Dragons, board and card games

MILITARY

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

PHYSICAL FEATURES

Hands	Right-Handed
Fingers	Short

SIZES & MEASUREMENTS

Neck (inches)	14
Chest (inches)	37
Inseam (inches)	29
Waist (inches)	32
Sleeve (inches)	30
Hat size	7 ¼
Shoe size	10

PHYSICAL AIDS

EYES:

Vision	Normal
Glasses	None
Astigmatism	No
Age started wearing glasses/contacts	N/A
Laser surgery?	No

DENTAL:

Device	None
Reason	N/A
Age range during use	N/A

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

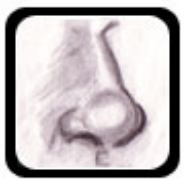
- Narrow
- **Average**
- Wide

NOSE:

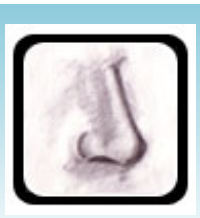
Size

- Small
- **Medium**
- Large

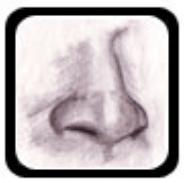
Shape



Round



Straight



Wide

Width

- Narrow
- **Average**
- Wide

Length

- Short
- **Average**
- Long

Bridge

Convex

Straight

Concave

Nostril flare

- Small
- Average
- Wide

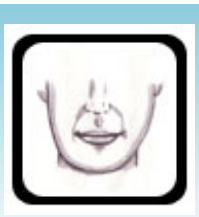
CHEEKBONES:

Set
- Low
- Average
- High

MOUTH:

Size


Small


Average


Large

Lips


Thin


Average


Full

Turn
- Downward
- Straight
- Upward

TEETH:

Size


Average


Large

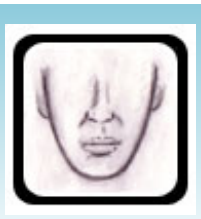

Small

Set
- Underbite
- Even
- Overbite

Gap: No

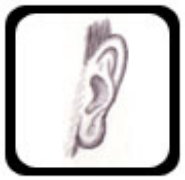
Condition	Poor Fair **Good**

CHIN:

Shape	 **Round**	 Oval	 Square
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	Self-employed (Construction)

Donor's Father Education:

Level	Some college
Degree	Not Completed
Subject	Engineering

Donor's Father Religion:

Faith	Christian
Denomination	Methodist

Donor's Father Languages:

Speak	English

Donor's Father Talents:

Voice	Tenor
Instrument	N/A
Other	Working on his equipment and land, big football fan.

Donor's Father Physical Information

Body

Age	68
Height	5' 8"
Weight	170 lbs.
Hands	Right-Handed

Donor's Father Facial Features

Eyes:

Eye Color	Brown

Eye shade	Medium
Corrective Lenses	No

Hair:

Original hair color	Brown
Type	Straight

Skin:

Skin Tone More about skin tones	Warm ivory

Other:

Chin cleft	None
Ear lobe	Attached
Facial Dimples	None

Donor's Mother Personal Information

Occupation	Small Business Owner

Donor's Mother Education:

Level	Some college
Degree	Not Completed
Subject	Accounting

Donor's Mother Religion:

Faith	Christian
Denomination	Lutheran

Donor's Mother Languages:

Speak	English

Donor's Mother Talents:

Voice	Soprano
Instrument	N/A
Other	Gardening, Pontooning, Traveling

Donor's Mother Physical Information

Body

Age	65
Height	5' 4"
Weight	130 lbs.
Hands	Right-Handed

Facial Features

Eyes:

Eye Color	Brown
Eye shade	Medium
Corrective Lenses	No

Hair:

Original hair color	Brown
Type	Straight

Skin:

Skin Tone More about skin tones	Bronze

Other:

Chin cleft	None
Ear lobe	Detached
Facial Dimples	Slight

DONOR'S SIBLINGS

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

Donor's Sibling #1

Full/Half sibling	Half

Sibling #1 Physical Information

Body

Age	39
Height	5' 5"
Weight	130 lbs.
Hands	Right-Handed

Sibling #1 Personal Information

Occupation	Abstract Administration

Education:

Level	College
Degree	Bachelor
Subject	Medical Terminology

Languages:

Speak	English

Talents:

Voice	Soprano
Instrument	N/A
Other	N/A

Sibling #1 Children:

# Boys Ages:	0
# Girls Ages:	0

Sibling #1 Facial Features

Eyes:

Eye color	Blue
Eye shade	Medium
Corrective Lenses	Yes

Hair:

Original hair color	Blond
Type	Wavy

Skin:

Skin tone More about skin tones	Warm ivory

Other:

Chin cleft	None
Ear lobe	Attached
Facial dimples	Slight

FATHER'S SIBLINGS

Father's Sisters

# of Sisters	1
Ages of Sisters:	60
Education levels for each:	Graduate School
Occupations for each:	Consultant

Father's Sisters' Total Children:

# Boys	0
# Girls	0

MOTHER'S SIBLINGS

Mother's Brothers

# Brothers	1
Ages of Brothers:	63
Education levels for each:	College
Occupations for each:	Purchasing Manager

Mother's Brothers' Total Children:

# Boys	1
# Girls	1

Mother's Sisters

# Sisters	2
Ages of Sisters:	65, 60
Education levels for each:	Some College, Some College
Occupations for each:	Homemaker, Warehouse Manager

Mother's Sisters' Total Children:

# Boys	2
# Girls	2

FATHER'S PARENTS

Father's Father

Education

Level	Graduate school
Degree	Master
Occupation:	Lawyer

Siblings:

# Brothers	1
# Sisters	0

Other:

Languages	English
Religion	Christian

Father's Mother

Education

Level	Some college
Degree	Not Completed
Occupation:	Homemaker

Siblings:

# Brothers	2
# Sisters	0

Other:

Languages	English
Religion	Christian

MOTHER'S PARENTS

Mother's Father

Education

Level	College
Degree	Bachelor
Occupation:	Teacher

Siblings:

# Brothers	1
# Sisters	0

Other:

Languages	English
Religion	Christian

Mother's Mother

Education

Level	College
Degree	Bachelor
Occupation:	Teacher

Siblings:

# Brothers	1
# Sisters	0

Other:

Languages	English
Religion	Christian

Donor 7240

Genetic Testing Summary

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

Last Updated: 05/31/24

Donor Reported Ancestry: Norwegian, German 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: Bardet-Biedl syndrome (BBS7-related) Carrier: Congenital adrenal hyperplasia due to 21-hydroxylase deficiency (CYP21A2) Carrier: GJB2-related conditions (GJB2) Carrier: Oculocutaneous albinism type 2 (OCA2) Negative for other genes sequenced.	Partner testing is recommended before using this donor. Residual risks for negative results can be seen here: https://fairfaxcryobank.com/invitae-residual-risk-table

*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.

INVITAE

INVITAE CARRIER SCREEN RESULTS

Patient name:	Donor 7240	**Sample type:**	Blood	**Report date:**	22-SEP-2023
DOB:	[redacted]	**Sample collection date:**	29-AUG-2023	**Invitae #:**	[redacted]
Sex assigned at birth:	Male	**Sample accession date:**	30-AUG-2023	**Clinical team:**	[redacted]
Gender:	Man				
Patient ID (MRN):	[redacted]				

Reason for testing

Gamete donor

Test performed

Invitae Carrier Screen

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: Bardet-Biedl syndrome (BBS7-related)	BBS7	c.712_715del (p.Arg238Glufs*59)	Autosomal recessive	Yes
Carrier: Congenital adrenal hyperplasia due to 21-hydroxylase deficiency	CYP21A2	c.293-13C>G (Intronic) c.332_339del (p.Gly111Valfs*21) c.92C>T (p.Pro31Leu)	Autosomal recessive	Yes
Carrier: GJB2-related conditions	GJB2	c.101T>C (p.Met34Thr)	Autosomal recessive	Yes
Carrier: Oculocutaneous albinism type 2	OCA2	c.1327G>A (p.Val443Ile)	Autosomal recessive	Yes

Laboratory Director: Qing Zhang, MD, PhD, FACMG, NYCQ, CGMB
Invitae 1400 16th Street, San Francisco, CA 94103 · clientservices@invitae.com · 800.436.3037

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

Bardet-Biedl syndrome (BBS7-related)

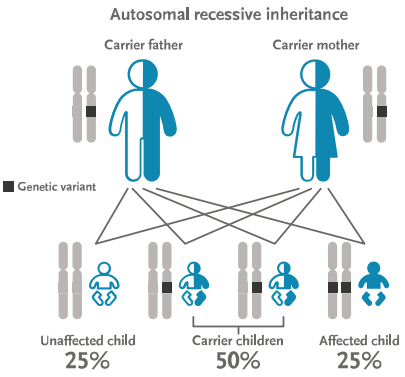
A single Pathogenic variant, c.712_715del (p.Arg238Glufs*59), was identified in BBS7.

What is Bardet-Biedl syndrome (BBS7-related)?

Bardet-Biedl syndrome (BBS) is part of a spectrum of conditions called ciliopathies, which involve defects in the microscopic, finger-like projections (cilia) that are located on the surface of cells and that are involved in cell movement and signaling. Ciliopathies affect many parts of the body. BBS can be caused by changes in many different genes. Symptoms of BBS generally include rod-cone dystrophy, an eye condition characterized by degeneration of the rods and cones (photoreceptors) which are the cells in the retina that respond to light, leading to progressive vision loss, as well as potentially life-threatening kidney problems, intellectual disability, genital abnormalities, and infertility. Affected individuals are commonly obese and have extra fingers and toes (polydactyly). They may also have a mirror-image reversal of their internal organs (situs inversus totalis), in which, for example, the heart is on the right side of the body instead of on the left, or the organs in their chest and abdomen may not be arranged as expected (heterotaxy). The atypical position of the organs may lead to a variety of health complications. Symptoms of BBS can vary widely, even within the same family. Some affected individuals may not have obvious symptoms (reduced penetrance). 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 BBS7 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 Bardet-Biedl syndrome (BBS7-related). 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
Bardet-Biedl syndrome (BBS7-related) (AR) NM_176824.2	BBS7	Pan-ethnic	≤1 in 500	Reduced

RESULT: CARRIER

Congenital adrenal hyperplasia due to 21-hydroxylase deficiency

Three Pathogenic variants, c.293-13C>G (Intronic), c.332_339del (p.Gly111Valfs*21), and c.92C>T (p.Pro31Leu) were identified in CYP21A2. Based on this analysis, these changes are located in the same copy of the CYP21A2 gene, and are derived from CYP21A1P, a nearby pseudogene, due to a conversion event between CYP21A2 and CYP21A1P.
This individual is a carrier for congenital adrenal hyperplasia due to 21-hydroxylase deficiency.

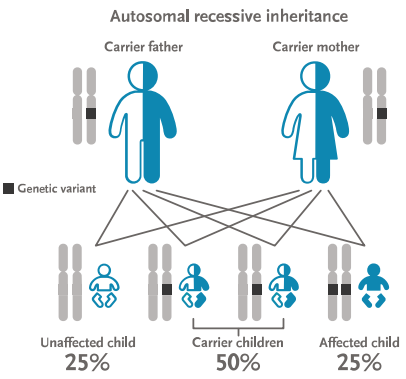
What is congenital adrenal hyperplasia due to 21-hydroxylase deficiency?

21-hydroxylase deficiency (21-OHD) is one of a group of conditions called congenital adrenal hyperplasia (CAH), which impair hormone production by the adrenal glands. The adrenal glands produce hormones that regulate many essential functions in the body, including sexual development and maturation. There are several types of CAH, which are caused by changes in different genes.

Symptoms of 21-OHD CAH range in severity, and are caused by the adrenal glands producing excess male sex hormones (androgens). There are three types of 21-OHD which include two classic forms, known as the salt-wasting and simple virilizing types, and the third is called the non-classic type. The salt-wasting type is the most severe, the simple virilizing type is less severe, and the non-classic type is the mildest form. Individuals with the salt-wasting type of 21-OHD lose large amounts of sodium in the urine, which can be life-threatening in early infancy. Infants with the simple virilizing type of 21-OHD do not experience salt-wasting. Female infants with classic 21-OHD usually have external genitalia that do not look clearly male or female (ambiguous genitalia). Male infants with classic 21-OHD usually have normal genitalia, although the testes may be smaller than typical. Individuals with a classic form of 21-OHD may have decreased fertility. Females with non-classic 21-OHD are born with typical external genitalia. They may experience irregular menstruation, decreased fertility, excess hair growth on the face and body (hirsutism), and male-pattern baldness. Males with non-classic 21-OHD may experience early beard growth and have small testes. Some individuals with non-classic 21-OHD may not have signs or symptoms of the condition (asymptomatic). The form(s) of 21-OHD CAH for which an individual would be at risk depends on the specific CYP21A2 variants inherited from the reproductive parents. 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 CYP21A2 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 congenital adrenal hyperplasia due to 21-hydroxylase 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
Congenital adrenal hyperplasia due to 21-hydroxylase deficiency (AR) NM_000500.7	CYP21A2 *	Pan-ethnic	1 in 61	1 in 751

RESULT: CARRIER

GJB2-related conditions

A single Pathogenic variant, c.101T>C (p.Met34Thr), was identified in GJB2.

What are GJB2-related conditions?

The GJB2 gene is associated with multiple conditions that can have both distinct and overlapping symptoms, as well as different inheritance patterns. GJB2-related conditions include autosomal recessive nonsyndromic deafness (DFNB1), as well as autosomal dominant nonsyndromic deafness (DFNA3) and several conditions involving deafness and skin findings. To understand which condition a genetic change is associated with, a review of the entire report, including the variant details section, is recommended.

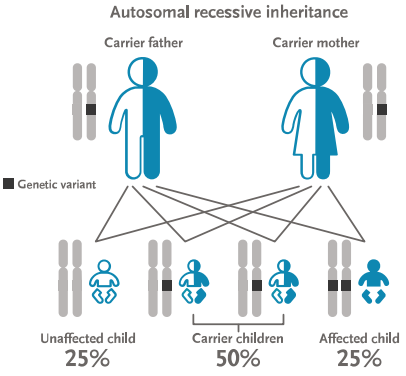
Please note that the GJB2 variant identified in this individual is expected to be associated with autosomal recessive nonsyndromic deafness (DFNB1).

Nonsyndromic deafness is a condition that affects an individual's ability to hear. It can be caused by changes in several different genes. Nonsyndromic deafness does not affect any other part of the body. Affected individuals are born with mild to profound deafness that typically does not worsen over time. Severity of deafness may vary, even among members of the same family. Intellect and life span are not impacted. Fewer than 1% of individuals with GJB2-related nonsyndromic deafness have been reported to have a variant in GJB2 on one chromosome and a deletion that includes both a region upstream of the GJB2 gene and a portion of GJB6, an adjacent gene, on the opposite chromosome. Follow-up depends on each affected individual's specific situation, and discussion with a healthcare provider should be considered.

Please note that the c.101T>C (p.Met34Thr) GJB2 variant identified in this individual has been associated with autosomal recessive nonsyndromic deafness with reduced penetrance. Reduced penetrance means that not all individuals with this genetic change will show signs or symptoms of the condition. See Variant details for additional information.

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 GJB2 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 GJB2-related conditions. 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
GJB2-related conditions (AR) NM_004004.5	GJB2	Pan-ethnic	1 in 50	1 in 4900

RESULT: CARRIER

Oculocutaneous albinism type 2

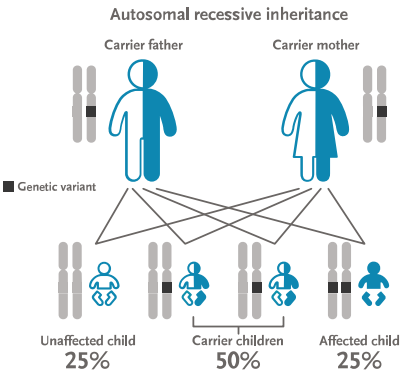
A single Pathogenic variant, c.1327G>A (p.Val443Ile), was identified in OCA2.

What is oculocutaneous albinism type 2?

Oculocutaneous albinism (OCA) is a condition that causes decreased color (hypopigmentation) of the hair, skin, and eyes. Affected individuals produce a reduced amount of melanin, the pigment that gives skin, hair, and eyes their color, resulting in hypopigmentation. Additional symptoms of OCA include reduced visual acuity (farsightedness or nearsightedness), increased sensitivity to light (photophobia), involuntary eye movements (nystagmus), and eyes that do not look in the same direction (strabismus). Other eye findings, such as reduced pigmentation of the light-sensitive tissue that lines the back of the eye (retina) and misrouting of the nerves of the eye (optic nerves), are seen on ophthalmologic exam. Individuals with fair complexions have an increased risk for skin cancers. Intelligence is not typically affected. Treatment is aimed at correcting vision and providing visual aids, or other visual resources. Sun protection is essential due to the increased risk for skin cancer.

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 OCA2 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 oculocutaneous albinism type 2. 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
Oculocutaneous albinism type 2 (AR) NM_000275.2	OCA2	Pan-ethnic	1 in 95	1 in 9400

Results to note

ABCA4

- c.5603A>T (p.Asn1868Ile) was identified in the ABCA4 gene.
- This benign variant is not known to cause disease and does not impact this individual's risk to be a carrier for ABCA4-related conditions. Carrier testing for the reproductive partner is not indicated based on this result. See Variant details for more information.

SMN1

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

Pseudodeficiency allele(s)

- Benign changes, 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

ABCA4, Exon 40, c.5603A>T (p.Asn1868Ile), heterozygous, Benign (reportable variant)

- This sequence change replaces asparagine, which is neutral and polar, with isoleucine, which is neutral and non-polar, at codon 1868 of the ABCA4 protein (p.Asn1868Ile).
- This variant is present in population databases (rs1801466, gnomAD 7%), including several hundred presumably unaffected homozygous individuals.
- This missense change has been observed in individual(s) with late onset Stargardt disease with foveal sparing. However, the vast majority (estimated 95%) of homozygous and compound heterozygous individuals remain unaffected with penetrance ranging from 0.24% to 9.54% across published studies. This variant may modify disease severity and/or age of onset when it is present in combination with additional known pathogenic variants (e.g., when this variant is on the same chromosome as one or more deleterious variants, such as c.2588G>C, c.5461-10T>C, c.4496G>A, and/or c.2564G>A, and also on the opposite chromosome with a pathogenic variant). In other cases, disease progression is not impacted when this variant is one component of other complex alleles, such as with c.769-784C>T (PMID: 11328725, 28446513, 29971439, 30204727, 30480704, 30670881, 31614660, 31618761, 31884623, 32037395, 32307445, 32815999, 34440414, 34874912).
- ClinVar contains an entry for this variant (Variation ID: 99390).
- 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 ABCA4 protein function.
- Experimental studies are conflicting or provide insufficient evidence to determine the effect of this variant on ABCA4 function (PMID: 11017087, 32845050, 33375396).
- For these reasons, this variant has been classified as a Benign reportable variant.

BBS7, Exon 7, c.712_715del (p.Arg238Glufs*59), heterozygous, PATHOGENIC

- This sequence change creates a premature translational stop signal (p.Arg238Glufs*59) in the BBS7 gene. It is expected to result in an absent or disrupted protein product. Loss-of-function variants in BBS7 are known to be pathogenic (PMID: 12567324, 19402160, 21209035, 31196119).
- This variant is present in population databases (rs760165634, gnomAD 0.01%).

- This premature translational stop signal has been observed in individuals with Bardet-Biedl Syndrome (PMID: 19402160, 26518167).
- ClinVar contains an entry for this variant (Variation ID: 281626).
- For these reasons, this variant has been classified as Pathogenic.

CYP21A2, Intron 2, c.293-13C>G (Intronic), heterozygous, PATHOGENIC

- This sequence change falls in intron 2 of the CYP21A2 gene. It does not directly change the encoded amino acid sequence of the CYP21A2 protein.
- The frequency data for this variant in the population databases (gnomAD) is considered unreliable due to the presence of homologous sequence, such as pseudogenes or paralogs, in the genome.
- This variant has been observed in individual(s) with classic salt-wasting, simple virilizing, and non-classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency (PMID: 20080860, 30995443). In at least one individual the data is consistent with being in trans (on the opposite chromosome) from a pathogenic variant.
- This variant is also known as IVS2-13A/C>G, I2G, c.293-13A/C>G, In2G.
- ClinVar contains an entry for this variant (Variation ID: 12155).
- Algorithms developed to predict the effect of sequence changes on RNA splicing suggest that this variant may disrupt the consensus splice site.
- For these reasons, this variant has been classified as Pathogenic.

CYP21A2, Exon 3, c.332_339del (p.Gly111Valfs*21), heterozygous, PATHOGENIC

- This sequence change creates a premature translational stop signal (p.Gly111Valfs*21) in the CYP21A2 gene. It is expected to result in an absent or disrupted protein product. Loss-of-function variants in CYP21A2 are known to be pathogenic (PMID: 10857554).
- The frequency data for this variant in the population databases (gnomAD) is considered unreliable due to the presence of homologous sequence, such as pseudogenes or paralogs, in the genome.
- This premature translational stop signal has been observed in individual(s) with classic salt-wasting, simple virilizing, and non-classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency (PMID: 7749410, 8081391, 23359698, 25227725, 26804566).
- This variant is also known as 706del8, 707_714del8, E3del8bp, c.711_718delGAGACTAC.
- ClinVar contains an entry for this variant (Variation ID: 12164).
- For these reasons, this variant has been classified as Pathogenic.

CYP21A2, Exon 1, c.92C>T (p.Pro31Leu), heterozygous, PATHOGENIC

- This sequence change replaces proline, which is neutral and non-polar, with leucine, which is neutral and non-polar, at codon 31 of the CYP21A2 protein (p.Pro31Leu).
- The frequency data for this variant in the population databases (gnomAD) is considered unreliable due to the presence of homologous sequence, such as pseudogenes or paralogs, in the genome.
- This missense change has been observed in individual(s) with classic salt-wasting, simple virilizing, and non-classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency (PMID: 2072928, 23142378, 23359698, 26804566, 31446012). In at least one individual the data is consistent with being in trans (on the opposite chromosome) from a pathogenic variant.
- This variant is also known as P30L.
- ClinVar contains an entry for this variant (Variation ID: 12153).
- 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 not expected to disrupt CYP21A2 protein function.
- Experimental studies have shown that this missense change affects CYP21A2 function (PMID: 2072928, 28539365).
- For these reasons, this variant has been classified as Pathogenic.

GJB2, Exon 2, c.101T>C (p.Met34Thr), heterozygous, PATHOGENIC

- This sequence change replaces methionine, which is neutral and non-polar, with threonine, which is neutral and polar, at codon 34 of the GJB2 protein (p.Met34Thr).

- This variant is present in population databases (rs35887622, gnomAD 2.0%), and has an allele count higher than expected for a pathogenic variant.
- This missense change has been observed in individual(s) with autosomal recessive nonsyndromic sensorineural deafness (PMID: 16077952, 22668073, 26117665; Invitae). 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. This variant has been reported to show reduced penetrance (PMID: 31160754).
- ClinVar contains an entry for this variant (Variation ID: 17000).
- 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 GJB2 protein function.
- For these reasons, this variant has been classified as Pathogenic.

OCA2, Exon 13, c.1327G>A (p.Val443Ile), heterozygous, PATHOGENIC

- This sequence change replaces valine, which is neutral and non-polar, with isoleucine, which is neutral and non-polar, at codon 443 of the OCA2 protein (p.Val443Ile).
- This variant is present in population databases (rs121918166, gnomAD 0.5%), and has an allele count higher than expected for a pathogenic variant.
- This missense change has been observed in individual(s) with oculocutaneous albinism (PMID: 8302318, 17960121, 20301410, 28976636, 29345414). In at least one individual the data is consistent with being in trans (on the opposite chromosome) from a pathogenic variant.
- ClinVar contains an entry for this variant (Variation ID: 955).
- 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) has been performed at Invitae for this missense variant, however the output from this modeling did not meet the statistical confidence thresholds required to predict the impact of this variant on OCA2 protein function.
- Experimental studies have shown that this missense change affects OCA2 function (PMID: 8980282, 25513726).
- 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
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
ACAD9	NM_014049.4
ACADM	NM_000016.5
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
AGA	NM_000027.3
AGL	NM_000642.2
AGPS	NM_003659.3
AGXT	NM_000030.2
AHI1	NM_017651.4
AIPL1*	NM_014336.4
AIRE	NM_000383.3
ALDH3A2	NM_000382.2
ALDH7A1	NM_001182.4
ALDOB	NM_000035.3
ALG1	NM_019109.4
ALG6	NM_013339.3
ALMS1	NM_015120.4
ALPL	NM_000478.5
AMN*	NM_030943.3
AMT	NM_000481.3
ANO10*	NM_018075.3
AP1S1	NM_001283.3
AQP2	NM_000486.5
ARG1	NM_000045.3
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
ATP6V1B1	NM_001692.3
ATP7B	NM_000053.3
ATP8B1*	NM_005603.4
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
BRIP1	NM_032043.2
BSND	NM_057176.2
BTD	NM_000060.3
CAD	NM_004341.4
CANT1	NM_138793.3
CAPN3	NM_000070.2
CASQ2	NM_001232.3
CBS	NM_000071.2
CC2D1A	NM_017721.5
CC2D2A	NM_001080522.2
CCDC103	NM_213607.2
CCDC39	NM_181426.1
CCDC88C	NM_001080414.3
CD3D	NM_000732.4
CD3E	NM_000733.3
CD40	NM_001250.5
CD59	NM_203330.2
CDH23	NM_022124.5
CEP152	NM_014985.3
CEP290	NM_025114.3
CERKL	NM_001030311.2
CFTR*	NM_000492.3
CHAT	NM_020549.4
CHRNE	NM_000080.3
CHRNG	NM_005199.4
CIITA	NM_000246.3
CLCN1	NM_000083.2
CLN3	NM_001042432.1
CLN5	NM_006493.2
CLN6	NM_017882.2
CLN8	NM_018941.3
CLRN1	NM_174878.2
CNGB3	NM_019098.4
COL11A2*	NM_080680.2
COL17A1	NM_000494.3
COL27A1	NM_032888.3
COL4A3	NM_000091.4
COL4A4	NM_000092.4
COL7A1	NM_000094.3
COX15	NM_004376.6
CPS1	NM_001875.4
CPT1A	NM_001876.3
CPT2	NM_000098.2

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

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
IKBKB	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_001031835.2
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
SMARCAL1	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

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 ≥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. 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 as needed based on stringent criteria using one of several validated orthogonal approaches (PubMed ID 30610921). Sequencing is performed by Invitae Corporation (1400 16th Street, San Francisco, CA 94103, #05D2040778). Confirmatory sequencing is performed by Invitae Corporation (1400 16th Street, San Francisco, CA 94103, #05D2040778).

  The following additional 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 -α3.7 subtypes, and all -α3.7 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, 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.

- 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.
- 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. DUOX2: Deletion/duplication and sequencing analysis is not offered for exons 6-7. GALE: Sequencing analysis for exons 10 includes only cds +/- 5 bp. DDX11: NM_030653.3:c.1763-1G>C variant only. GNE: Sequencing analysis for exons 8 includes only cds +/- 10 bp. 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. PKHD1: Deletion/duplication analysis is not offered for exon 13. VPS13A: Deletion/duplication analysis is not offered for exons 2-3, 27-28. TBCE: Sequencing analysis for exons 2 includes only cds +/- 10 bp. BBS9: Deletion/duplication analysis is not offered for exon 4. WRN: Deletion/duplication analysis is not offered for exons 10-11. Sequencing analysis for exons 8, 10-11 includes only cds +/- 10 bp. GHR: Deletion/duplication and sequencing analysis is not offered for exon 3. OAT: Deletion/duplication analysis is not offered for exon 2. CFTR: Sequencing analysis for exons 7 includes only cds +/- 10 bp. EYS: Sequencing analysis for exons 30 includes only cds +/- 0 bp. FH: Sequencing analysis for exons 9 includes only cds +/- 10 bp. ANO10: Sequencing analysis for exons 8 includes only cds +/- 0 bp. ATP8B1: Sequencing analysis for exons 19 includes only cds +/- 10 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. COL11A2: Deletion/duplication analysis is not offered for exon 36. 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. TSFM: Sequencing analysis is not offered for exon 5. VPS53: Sequencing analysis for exons 14 includes only cds +/- 5 bp. 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, 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. AIPL1: Sequencing analysis for exons 2 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. LIFR: Sequencing analysis for exons 3

includes only cds +/- 5 bp. AMN: Deletion/duplication analysis is not offered for exon 1. MLC1: Sequencing analysis for exons 11 includes only cds +/- 10 bp. PEX1: Sequencing analysis for exons 16 includes only cds +/- 0 bp. USH1C: Deletion/duplication analysis is not offered for exons 5-6. FAH: Deletion/duplication analysis is not offered for exon 14. GALC: Deletion/duplication analysis is not offered for exon 6. TYR: Deletion/duplication and sequencing analysis is not offered for exon 5. TG: Deletion/duplication analysis is not offered for exon 18. Sequencing analysis for exons 44 includes only cds +/- 0 bp. FANCL: Sequencing analysis for exons 4, 10 includes only cds +/- 10 bp. ATM: Sequencing analysis for exons 6, 24, 43 includes only cds +/- 10 bp.

This report has been reviewed and approved by:

Arunkanth Ankala, Ph.D., FACMG
Clinical Molecular Geneticist

PATIENT INFORMATION
7240,DONOR

REPORT STATUS **Final**

Nichols Institute, Chantilly

DOB: [redacted] Age: [redacted]
SEX: M

ORDERING PHYSICIAN
[redacted]

CLIENT INFORMATION
[redacted]

SPECIMEN INFORMATION
SPECIMEN: [redacted]
REQUISITION: [redacted]
LAB REF NO: [redacted]

ID: 7240-[redacted]

COLLECTED: 08/29/2023 00:00
RECEIVED: 08/30/2023 15:00
REPORTED: 09/07/2023 19:23

Test Name	In Range	Out of Range	Reference Range	Lab
Hemoglobinopathy Evaluation				AMD
Red Blood Cell Count	5.29		4.20-5.80 Mill/uL	
HEMOGLOBIN	16.0		13.2-17.1 g/dL	
Hematocrit				
Hematocrit		**50.1 H**	38.5-50.0 %	
MCV	94.7		80.0-100.0 fL	
MCH	30.2		27.0-33.0 pg	
RDW	12.5		11.0-15.0 %	
Hemoglobin A	97.8		>96.0 %	
Hemoglobin F	0.0		<2.0 %	
Hemoglobin A2 (Quant)	2.2		2.2-3.2 %	
Interpretation	NORMAL PATTERN There is a normal pattern of hemoglobins and normal levels of Hb A2 and Hb F are present. No variant hemoglobins are observed. This is consistent with A/A phenotype. If iron deficiency coexists with a mild/silent beta thalassemia trait Hb A2 may be in the normal range. Rare variant hemoglobins have no separation from hemoglobin A by capillary zone electrophoresis (CZE) or high-performance liquid chromatography (HPLC). If clinically indicated, Thalassemia and Hemoglobinopathy Comprehensive (TC 17365) should be considered.			
CBC (includes Differential and Platelets)				AMD
CBC (includes Differential and Platelets)				
White Blood Cell Count	5.5		3.8-10.8 Thous/uL	
Red Blood Cell Count	5.29		4.20-5.80 Mill/uL	
HEMOGLOBIN	16.0		13.2-17.1 g/dL	
Hematocrit		**50.1 H**	38.5-50.0 %	
MCV	94.7		80.0-100.0 fL	
MCH	30.2		27.0-33.0 pg	
MCHC		**31.9 L**	32.0-36.0 g/dL	
RDW	12.5		11.0-15.0 %	
PLATELET COUNT	224		140-400 Thous/uL	
MPV	11.0		7.5-12.5 fl	

PATIENT INFORMATION
7240,DONOR

REPORT STATUS **Final**

Nichols Institute, Chantilly

ORDERING PHYSICIAN [redacted]

DOB: [redacted] Age: [redacted]
SEX: M
ID: 7240[redacted]

COLLECTED: 08/29/2023 00:00
REPORTED: 09/07/2023 19:23

Test Name	In Range	Out of Range	Reference Range	Lab
CBC (includes Differential and Platelets) (Continued)				
Absolute Neutrophils	3713		1500-7800 cells/uL	
Absolute Lymphocytes	1452		850-3900 cells/uL	
Absolute Monocytes	292		200-950 cells/uL	
Absolute Eosinophils	22		15-500 cells/uL	
Absolute Basophils	22		0-200 cells/uL	
Neutrophils	67.5		%	
Lymphocytes	26.4		%	
Monocytes	5.30		%	
Eosinophils	0.40		%	
Basophils	0.40		%	
Nucleated RBC	0.00		0 /100 WBC	
Comment(s)	Review of peripheral smear confirms automated results.			
[redacted]	[redacted]		[redacted]	[redacted]
[redacted]	[redacted]		[redacted] [redacted]	[redacted]
[redacted]	[redacted]		[redacted]	[redacted]
Chromosome Analysis, Blood				AMD
Chromosome Analysis, Blood				
Chromosome Analysis, Blood				

Order ID: [redacted]

Specimen Type: Blood

Clinical Indication: Gamete donor

RESULT:
NORMAL MALE KARYOTYPE

INTERPRETATION:
Chromosome analysis revealed normal G-band patterns within the limits of standard cytogenetic analysis.

Please expect the results of any other concurrent study in a separate report.

NOMENCLATURE:
46,XY

ASSAY INFORMATION:
Method: G-Band (Digital Analysis: MetaSystems/Ikaros)
Cells Counted: 20
Band Level: 550
Cells Analyzed: 5
Cells Karyotyped: 5

This test does not address genetic disorders that cannot be detected by standard cytogenetic methods or rare events such as low level

Test Name	In Range	Out of Range	Reference Range	Lab

Chromosome Analysis, Blood (Continued)
Chromosome Analysis, Blood (Continued)

mosaicism or subtle rearrangements.

Steven A. Schonberg, Ph.D., FACMG, Technical Director, Cytogenetics and Genomics, 703-802-7156

Electronic Signature: 9/7/2023 6:39 PM

For additional information, please refer to
http://education.questdiagnostics.com/faq/chromsblood
(This link is being provided for informational/
educational purposes only).

Performing Laboratory Information:

AMD Quest Diagnostics Nichols Institute 14225 Newbrook Drive Chantilly VA 20151 Laboratory Director: Patrick W Mason, MD PhD

Fairfax Cryobank
The Trusted Choice for Donor Sperm

Staff Impressions

DONOR NUMBER:
7240

PHYSICAL APPEARANCE:

Donor 7240 stands at 5’ 8” with a slim build. He has light fair skin, a narrow face, bright blue eyes, and strawberry blond hair. He has smaller pink lips, and a little button nose. He usually likes to have some facial hair.

PERSONALITY:

Donor 7240 is a logical and prompt person. He describes himself as respectful, open-minded, and a great problem solver. In his free time, you can find him playing or studying the game of curling as this is his top hobby. He also enjoys video gaming with friends, downhill skiing, or spending time at the family cabin. He is happy in his career as an IT manager but has big plans to climb the ladder in his field.

09-24-2025 10:03:22

Fairfax Cryobank
The Trusted Choice for Donor Sperm

Donor Personality Type

Donor # 7240 Personality Type: ISTJ

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 INSPECTOR

I | S | T | J

At first glance, ISTJs are intimidating. They appear serious, formal, and proper. They also love traditions and old-school values that uphold patience, hard work, honor, and social and cultural responsibility. They are reserved, calm, quiet, and upright. These traits result from the combination of I, S, T, and J, a personality type that is often misunderstood.

ISTJs are bright, logical, and wise individuals who are very direct – for them, truth and facts are the most important. With their love of facts, they tend to accumulate a lot of information in their memory. Their focus on concrete facts and data makes them excellent analysts in many different environments. ISTJs are respected for their exceptional loyalty to their duty. Their accuracy, patience and ability to concentrate make them ideal employees in many professions.

Common traits:

- Physically and mentally organized
- Excellent panning skills and highly intellectual
- Calm and clear-headed during tense situations
- Serious and committed in relationships
- Believes in traditions and strives to follow them
- Well-respected in the community
- Love to memorize details and facts

Fairfax Cryobank

The Trusted Choice for Donor Sperm

The Lifetime Series of Donor 7240

6 Months

Age 6

From Our Staff

"Donor 7240 is a logical and prompt person. He describes himself as respectful, open-minded, and a great problem solver. He is happy in his career as an IT manager but has big plans to climb the ladder in his field."

Age 10

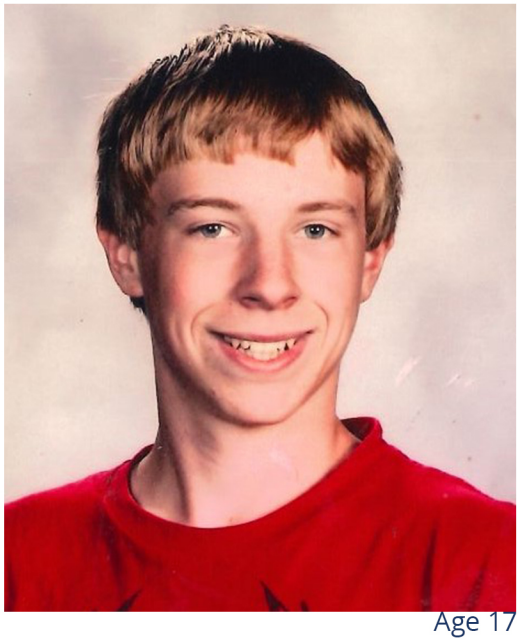
Fairfax Cryobank

The Trusted Choice for Donor Sperm

The Lifetime Series of Donor 7240

Age 14

Age 15



Age 17

Adult