EGG DONOR PROFILE

INTERNAL DONOR CODE: **OVOK007161**	
GENERAL INFORMATION:	
DATE OF BIRTH: 10/03/1991	PLACE OF BIRTH: UKRAINE
CHILDREN: 2	MARITAL STATUS: SINGLE
ADOPTED: NO	RELIGION/BELIEF SYSTEM: OTHER

PHENOTYPICAL CHARACTERISTICS:		
B.T/RH: O+	RACE: CAUCASIAN	COMPLEXION: WHITE
EYE COLOUR: BLUE	HAIR COLOUR: BROWN	TYPE OF HAIR: STRAIGHT
HEIGHT: cm 160	WEIGHT: kg 58	BMI: 22.66

MOTHER:	**FATHER**:
RACE: CAUCASIAN COMPLEXION: WHITE HAIR COLOUR: DARK BLONDE EYE COLOUR : BLUE	RACE: CAUCASIAN COMPLEXION: WHITE HAIR COLOUR: LIGHT BROWN EYE COLOUR : GREEN

MATERNAL GRANDMOTHER:	**MATERNAL GRANDFATHER**:	**PATERNAL GRANDMOTHER**:	**PATERNAL GRANDFATHER**:
RACE: CAUCASIAN COMPLEXION: WHITE HAIR COLOUR: DARK BLONDE EYE COLOUR : BLUE	RACE: CAUCASIAN COMPLEXION: WHITE HAIR COLOUR: BROWN EYE COLOUR : BLUE-GREY	RACE: CAUCASIAN COMPLEXION: WHITE HAIR COLOUR: DARK BLONDE EYE COLOUR : BLUE-GREY	RACE: CAUCASIAN COMPLEXION: WHITE HAIR COLOUR: BROWN EYE COLOUR : HAZEL

GENETIC ANALYSIS PERFORMED:	
KARYOTYPE: 46XX	
CYSTIC FIBROSIS [1]: NEGATIVE	G6PD[1]: NON CARRIER
X-FRAGILE[1]: NORMAL	THALASSEMIA[1]: NEGATIVE
SPINAL MUSCULAR ATROPHY[1]: NON CARRIER	NONSYNDROMIC HEARING LOSS[1]: NON CARRIER
RECESSIVE MUTATIONS ANALYSIS/GENETIC COMPATIBILITY TEST[2]: (SEQ) HERES LITE CARRIER TEST	
OTHER STUDIES: -	

[1] No pathogenic mutations were identified in the test, performed on the donor, attached to this document. However, a negative result does not rule out the possibility of genetic predisposition, nor does it rule out pathogenic mutations in areas not evaluated in the test performed or in regions covered at too low a level to be reliably evaluated. In addition, it does not rule out mutations not analysed by this test.

[2]The receiving center has the possibility of genetic matching with the gamete receptor (which will be recommended to carry out the same carrier test), to minimize the risk of offspring affected by the genes analysed with the full panel of recessive genetic diseases studied.

EDUCATION & OCCUPATION:	
EDUCATION LEVEL:	ASSOCIATE DEGREE
ACADEMIC STRENGHTS:	MATHEMATICS
SPOKEN LANGUAGES:	UKRAINIAN
CURRENT OCCUPATION:	CATERING, PERSONAL, PROTECTIVE AND VENDOR SERVICE WORKERS

LIFE STYLE & INTERESTS:	
HOBBIES:	MUSIC, KNITTING, KNITTING WITH BEADS
ARTISTIC SKILLS:	DRAWING
MUSIC TALENT/INSTRUMENT:	NO
FAVORITE SINGER/GROUP:	ALINA GROSU
TYPE OF DIET:	NON VEGETARIAN
FAVORITE MEAL:	BORSCHT- BEET SOUP, RICE WITH MEAT
FAVORITE COLOR:	RED, GREEN
SPORT/EXERCISE:	NO
FAVORITE SPORTS:	RUNNING

OTHER:

IMPORTANT ELEMENTS TO ACHIEVE PERSONAL GOALS:

- [x] FAMILY
- [ ] WELFARE
- [ ] WILL
- [x] HEALTH
- [ ] JOB
- [ ] LOVE
- [ ] PETS
- [ ] PEACE OF MIND

REASONS TO BECOME EGG DONOR:

- [ ] HELP OTHER WOMEN
- [ ] COMPENSATION
- [x] MOTHER WHO WANTS OTHER WOMEN TO BE SO THANKS MY DONATION
- [ ] FAMILY/FRIENDS WITH FERTILITY PROBLEMS
- [ ] OTHER:_____

MESSAGE TO EGG RECIPIENT:

I WISH YOU HAVE HEALTHY CHILDREN AND PATIENCE

MEDICAL HISTORY:	
GENERAL GOODHEALTH	YES
LATERALITY	RIGHT-HANDED
ILLNES/DISABILITY	NO
ALLERGIES	NO
DEPRESSION/ANXIETY	NO
BIRTH DEFECTS	NO
HOSPITALIZATION/SURGERY	NO
CORRECTIVE LENSES/GLASSES	NO
NEARSIGHTED	NO
FARSIGHTED	NO
ABNORMAL HEARING	NO
TEETH CONDITION	FAIR
EXLUDED AS BLOOD DONOR:	NO

TATTOOS PAST 12 MONTHS	NO
BODY PIERCINGS PAST 12 MONTHS	NO
CHEMICAL EXPOSURE	NO
GAS EXPOSURE	NO
RADIATON EXPOSURE	NO
CURRENT MEDICATION	NO
ALCOHOL CONSUMPTION	NO
RELATIVES WITH ALCOHOLISM	NO
SMOKER	NO

REPRODUCTIVE/SEXUAL INFORMATION:	
AGE FIRST PERIOD	14
DAYS BETWEEN PERIODS	28
OTHER VAGINAL BLEEDING	NO
CURRENT SEXUAL PARTNER	-
SEXUAL PARTNERS PAST 6 MONTHS	1
CONTRACEPTIVE METHOD	CONDOM
CONFIRMED PREGANCIES	3
SPONTANEOUS MISCARRIAGES	NO
ELECTIVE MISCARRIAGES	1
ENDOMETRIOSIS	NO
SEXUAL PROBLEMS	NO

MEDICAL-GENETIC FAMILY HISTORY:		
HEART MEDICAL CONDITION:	**DONOR**	**FAMILY**
STROKE	NO	NO
HEART ATTACK	NO	NO
CONGENNITAL HEART DISEASE	NO	NO
HEART DISEASE	NO	NO
HIGH BLOOD PRESSURE	NO	NO
BLOOD MEDICAL CONDITION:	**DONOR**	**FAMILY**
ANEMIA	NO	NO
SICKLE-CELL ANEMIA	NO	NO
HEMOPHILIA/OTHER BLEEDING PROBLEM	NO	NO
LEUKEMIA	NO	NO
IMMUNE DEFICIENCY	NO	NO
POLYARTERITIS NODOSA	NO	NO
OTHER BLOOD DISORDER	NO	NO

RESPIRATORY (LUNGS) MEDICAL CONDITION:	DONOR	FAMILY
HAY FEVER	NO	NO
ASHMA	NO	NO
EMPHYSEMA	NO	NO
TUBERCULOSIS	NO	NO
LUNG CANCER	NO	NO
PNEUMONIA	NO	NO
CYSTIC FIBROSIS	NO	NO
ALPHA-1 ANTITRYPSIN DISORDER	NO	NO
OTHER LUNG DISEASE	NO	NO
GASTROINTESTINAL MEDICAL CONDITION:	**DONOR**	**FAMILY**
STOMACH ULCER/DUODENUM	NO	NO
GALLSTONES	NO	NO
HEPATITIS A (INFECTIOUS)	NO	NO
HEPATITIS B (SERUM)	NO	NO
OTHER LIVER DISEASE	NO	NO
ULCERATIVE COLITIS	NO	NO
PYLORIC STENOSIS	NO	NO
CROHN'S DISEASE	NO	NO
INTESTINAL CANCER	NO	NO
INFLAMMATORY	NO	NO
BOWEL DISEASE	NO	NO
RECTAL DISORDER	NO	NO
OTHER CANCER/DIGESTIVE SYSTEM PROBLEM	NO	NO
METABOLIC/ENDOCRINE MEDICAL CONDITION:	**DONOR**	**FAMILY**
DIABETES WITH INSULIN THERAPY	NO	NO
DIABETES WITHOUT INSULIN THERAPY	NO	NO
HYPOGLYCEMIA	NO	NO
THYROID CANCER	NO	NO
THYROID DISEASE	NO	NO
GOITER	NO	NO
ADRENAL DYSFUNCTION/DISORDER	NO	NO
HYPERACTIVITY	NO	NO
PKU/INHERITED METABOLISM DISORDER	NO	NO
URINARY MEDICAL CONDITION:	**DONOR**	**FAMILY**
PROGRESSIVE KIDNEY DISEASE	NO	NO
POLYCYSTIC KIDNEY DISEASE	NO	NO

OTHER DISEASE URINARY TRACT (URETHRA, BLADDER, URETER)	NO	NO
GENITAL/REPRODUCTIVE SYSTEM MEDICAL CONDITION:	**DONOR**	**FAMILY**
UTERINE FIBROIDS	NO	NO
OVARIAN CYSTS	NO	NO
CERVIX/OVARIES/UTERUS CANCER	NO	NO
MISCARRIAGE/STILLBORN	NO	NO
HERPES SIMPLEX VIRUS, GENITAL	NO	NO
NEUROLOGICAL MEDICAL CONDITION:	**DONOR**	**FAMILY**
MIGRAINES	NO	NO
MENTAL RETARDATION	NO	NO
SENILITY/MENTAL DETERIORATION BEFORE AGE 50	NO	NO
MULTIPLE SCLEROSIS	NO	NO
CEREBRAL PALSY	NO	NO
EPILEPSY/SEIZURES	NO	NO
NEURAL TUBE DEFECTS	NO	NO
SPINAL CORD DISORDERS	NO	NO
GAUCHER'S DISEASE	NO	NO
WILSON'S DISEASE	NO	NO
CREUTZFELDT-JAKOB DISEASE	NO	NO
HUNTINGTON'S DISEASE	NO	NO
TUBEROUS SCLEROSIS	NO	NO
NEUROFIBROMATOSIS	NO	NO
DEMENTIA/DEGENERATIVE DISORDER	NO	NO
ALZHEIMER'S	NO	NO
PARKINSON'S DISEASE	NO	NO
BRAIN TUMOR	NO	NO
MYASTENIA GRAVIS	NO	NO
DOWN'S SYNDROME	NO	NO
TRANSMISSIBLE SPONGIFORM ENCEPHALOPATHY	NO	NO
OTHER NERVOUS SYSTEM DISEASES	NO	NO
MENTAL HEALTH MEDICAL CONDITION:	**DONOR**	**FAMILY**
SCHIZOPHRENIA	NO	NO
MANIC DEPRESSIVE PSYCHOSIS	NO	NO
MUSCLES/BONES/JOINTS MEDICAL CONDITION:	**DONOR**	**FAMILY**
MUSCULAR DISTROPHY	NO	NO
OTHER CHRONIC MUSCLE DISEASE	NO	NO
LOSS OS MUSCLE COORDINATION	NO	NO

SPYNAL MUSCULAR ATROPHY	NO	NO
SYSTEMIC LUPUS	NO	NO
DEFORMITY OF SPINE	NO	NO
OSTEOPOROSIS	NO	NO
DWAFIRSM	NO	NO
HEREDITARY LOW BACK DISORDER	NO	NO
RHEUMATOID ARTHRITIS	NO	NO
REITER'S DISEASE	NO	NO
GOUT	NO	NO
CLUB FOOT	NO	NO
METABOLIC BONE DISEASE	NO	NO
SIGHT/SOUND/SMELL MEDICAL CONDITION:	**DONOR**	**FAMILY**
DEAFNESS BEFORE AGE 60	NO	NO
DEFORMITY OF THE EAR	NO	NO
CATARACTS BEFORE AGE 60	NO	NO
BLINDNESS IN BOTH EYES BEFORE AGE 60	NO	NO
COLOR BLINDNESS	NO	NO
GLAUCOMA	NO	NO
DEVIATED SEPTUM	NO	NO
OTHER SIGHT/SOUND/SMELL DISORDER	NO	NO
SKIN MEDICAL CONDITION:	**DONOR**	**FAMILY**
ACNÉ	NO	NO
ECZEMA	NO	NO
PSORIASIS	NO	NO
PIGMENTATION DISORDERS	NO	NO
ALBINISM	NO	NO
INFECTIOUS SKIN DISEASE	NO	NO
MORE THAN 5 PURPLE/COFFEE COLORED SPOTS ON SKIN	NO	NO
NUMEROUS LUMPS UNDER THE SKIN	NO	NO
OTHER SKIN DISORDER	NO	NO
OTHER MEDICAL CONDITIONS:	**DONOR**	**FAMILY**
ALCOHOLISM	NO	NO
DRUG ABUSE/MISUSE/ADDICTION	NO	NO
BREAST CANCER	NO	NO
ANY CANCER NOT MENTIONED ABOVE	NO	NO
CLEFT PALADE/LIP	NO	NO
SERIOUS BIRTH DEFECTS	NO	NO

INGUINAL HERNIA	NO	NO
EARLY DEATH (LESS THAN AGE 50)	NO	NO
SARCOIDOSIS	NO	NO
PREMATURE DEGENERATION OF ANY ORGAN SYSTEM	NO	NO
ANY OTHER CONDITION NOT MENTIONED ABOVE	NO	NO

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HERES

RESULTS REPORT
B.A.
7161 HERES-14742

CLINIC	DONOR INFORMATION
Center Name: FIV MARBELLA-KIEV	**Initials:** B.A.
Center Code: FG26.11	**Patient Code:** 7161
Phone: -	**D.O.B:** 03/10/1991
Clinician: Dr. Sevostianova	**Sex:** Female
Report Date: 24/11/2021	**Ethnicity:** Caucasian
Test information	**Sample information**
Test: HERES	**Code:** HERES-14742
Type of test: SEQ-F-Heres V1-Donor Panel F v2	**Barcode:** 23456789918219
Genes tested: 30	**Specimen type:** Blood
	Collection Date: 23/10/2021
	Reception Date: 03/11/2021

SUMMARY OF RESULTS: NO MUTATIONS HAVE BEEN IDENTIFIED

No pathological/potentially pathological variants were identified in the submitted specimen. A negative result does not rule out the possibility of a genetic predisposition nor does it rule out any pathogenic mutations in areas not assessed by this test or in regions that were covered at a level too low to reliably assess. Also, it does not rule out mutations that are of the sort not queried by this test. See Methods and Limitations for more information. Genetic counseling is recommended.

INTERPRETATION

Notes and Recommendations:

The test results indicate that this individual is a NON-CARRIER. Genetic counseling is strongly recommended to discuss reproductive risk and prenatal testing options.

- Testing for a 3 nucleotide (CGG) repeat sequence in the *FMR1* gene was performed to screen for your carrier status for Fragile X Syndrome. The repeat sizes detected were: 30 and 33 repeats. These results are within the normal range. Therefore, you are not considered a carrier for Fragile X Syndrome.
- Testing for copy number changes in the *SMN1* gene was performed to screen for your carrier status for Spinal Muscular Atrophy. 2 copies of the *SMN1* gene were detected. These results are within the normal range for non-carriers. See Limitations section for more information.
- This carrier screening test does not screen for all possible genetics conditions, nor for all possible mutations in every gene tested. Individuals with negative test results may still have up to a 3-4% risk to have a child with a birth defect due to genetic and/or environmental factors.

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ANALYZED and INFORMED GENES

HERES CarrierScreening test. 30 genes tested (99.41% of coding bases at >20x). For more specific information on genes and calculation of residual risk see ADDITIONAL TABLE.

ABCD1	*COL4A5*	*F8*	*GJB2*	*IDS*	*PDHA1*
AR	*CYBB*	*F9*	*GLA*	*IL2RG*	*PRPS1*
ATP7A	*DMD*	*FMR1*	*HBA1*	*MTM1*	*RS1*
CFTR	*EDA*	*G6PD*	*HBA2*	*OCRL*	*SMN1*
CHM	*EMD*	*GJB1*	*HBB*	*OTC*	*WAS*

METHODS

Genomic DNA was isolated from the submitted specimen indicated above (if cellular material was submitted). DNA was barcoded, and enriched for the coding exons of targeted genes using hybrid capture technology. Prepared DNA libraries were then sequenced using a Next Generation Sequencing technology. Following alignment, variants were detected in regions of at least 10x coverage. For this specimen, 99.45% and 99.41% of coding regions and splicing junctions of genes listed had been sequenced with coverage of at least 10x and 20x respectively or by Sanger sequencing. The remaining regions did not have 10x coverage, and were not evaluated. Variants were interpreted manually using locus specific databases, literature searches, and other molecular biological principles. All the variants with quality score less than 500 (roughly 40x of coverage for a heterozygous variant) will be confirmed by Sanger sequencing. Only variants classified as pathogenic, likely-pathogenic are reported. All genes listed were evaluated for large deletions and/or duplications. However, single exon deletions or duplications will not be detected in this assay, nor will copy number alterations in regions of genes with significant pseudogenes (see Gene Specific Limitations below). Putative deletions or duplications identified are confirmed by an orthogonal method (qPCR or MLPA). If included in the panel, *FMR1* repeat analysis is performed by repeat-primed PCR (rpPCR) and amplicon length analysis. Methylation studies are not performed. Variants are classified using the ACMG Guidelines for Sequence Variant Interpretation (PubMed: 27993330) unless otherwise specified.

GENERAL LIMITATIONS

These test results and variant interpretation are based on the proper identification of the submitted specimen, accuracy of any stated familial relationships, and use of the correct human reference sequences at the queried loci. In very rare instances, errors may result due to mix-up or comingling of specimens. Positive results do not imply that there are no other contributors, genetic or otherwise, to future pregnancies, and negative results do not rule out the genetic risk to a pregnancy. Official gene names change over time. Fulgent uses the most up to date gene names based on HUGO Gene Nomenclature Committee (https://www.genenames.org) recommendations. If the gene name on report does not match that of ordered gene, please contact the laboratory and details can be provided. Result interpretation is based on the available clinical and family history information for this individual, collected published information, and Alamut annotation available at the time of reporting. This assay is not designed or validated for the detection of low-level mosaicism or somatic mutations. This assay will not detect certain types of genomic aberrations such as translocations, inversions, or repeat expansions other than specified genes. DNA alterations in regulatory regions or deep intronic regions (greater than 20bp from an exon) may not be detected by this test. Unless otherwise indicated, no additional assays have been performed to evaluate genetic changes in this specimen. There are technical limitations on the ability of DNA sequencing to detect small insertions and deletions. Our laboratory uses a sensitive detection algorithm; however these types of alterations are not detected as reliably as single nucleotide variants. Rarely, due to systematic chemical, computational, or human error, DNA variants may be missed. Although next generation sequencing technologies and our bioinformatics analysis significantly reduce the confounding contribution of pseudogene sequences or other highly-homologous sequences, sometimes these may still interfere with the technical ability of the assay to identify pathogenic alterations in both sequencing and deletion/duplication analyses. Deletion/duplication analysis can identify alterations of genomic regions which include one whole gene (buccal swab specimens and whole blood specimens) and are two or more contiguous exons in size (whole blood specimens only); single exon deletions or duplications may occasionally be identified, but are not routinely detected by this test. When novel DNA duplications are identified, it is not possible to discern the genomic location or orientation of the duplicated segment; hence the effect of the duplication cannot be predicted. Where deletions are detected, it is not always possible to determine whether the predicted product will remain in-frame or not. Unless otherwise indicated, deletion/duplication analysis has not been performed in regions that have been sequenced by Sanger.

GENE SPECIFIC LIMITATIONS

AR
The current testing method does not assess trinucleotide repeat expansions in this gene.

CFTR
Analysis of the intron 8 polymorphic region (e.g. IVS8-5T allele) is only performed if the p.Arg117His (R117H) mutation is detected. Single exon deletion/duplication analysis is limited to deletions of previously reported exons: 1, 2, 3, 11, 19, 20, 21.

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DMD
Single exon deletion/duplication analysis is limited to exons with >1 patient reported in the UMD database (http://www.umd.be/DMD/W_DMD/index.html), accessed Dec 29,2020 and all out-of-frame exons after exon 3. This includes deletion of exon 1, and duplication of exon 2, and del/dup for exons 3,6~8,11,12,17~22,43~46,48,50~56,58~63,65~70,75,76 and 78. Single-exon detection is limited to blood samples.

F8
The current testing method does not include detection of intron 1/intron 22 inversions in the *F8* gene. This detection is available upon request.

FMR1
The exact size of alleles >200 CGG repeats cannot be determined; these alleles are pathogenic for X-Linked Fragile X Syndrome. Alleles with <10 repeats may fail to amplify; these alleles are benign. The repeat length for this gene may vary by +/- 1 repeat unit. Methylation is not analyzed. Small degrees of size mosaicism, including gonadal mosaicism, may not be detected.

HBA1
The phase of heterozygous alterations in the *HBA1* gene and the *HBA2* gene cannot be determined, but can be confirmed through parental testing.

HBA2
The phase of heterozygous alterations in the *HBA1* gene and the *HBA2* gene cannot be determined, but can be confirmed through parental testing.

SMN1
The current testing method detects sequencing variants in exon 7 and copy number variations in exons 7-8 of the *SMN1* gene (NM_022874.2). Sequencing and deletion/duplication analysis are not performed on any other region in this gene. About 5%-8% of the population have two copies of *SMN1* on a single chromosome and a deletion on the other chromosome, known as a [2+0] configuration (PubMed: 20301526). The current testing method cannot directly detect carriers with a [2+0] *SMN1* configuration, but can detect linkage between the silent carrier allele and certain population-specific single nucleotide changes. As a result, a negative result for carrier testing greatly reduces but does not eliminate the chance that a person is a carrier. The 3-copy *SMN1* state can be detected by this test and will be reported out if present.

SIGNATURE

Montserrat Palahi Bages M.Sc.
Col. 21913-C

Results based on Fulgent Genetics report FT-TS9516623AA

DISCLAIMER

This test was developed and its performance characteristics determined by Fulgent Genetics. It has not been cleared or approved by the FDA. The laboratory is regulated under CLIA as qualified to perform high-complexity testing. This test is used for clinical purposes. It should not be regarded as investigational or for research. Since genetic variation, as well as systematic and technical factors, can affect the accuracy of testing, the results of testing should always be interpreted in the context of clinical and familial data. For assistance with interpretation of these results, healthcare professionals may contact us directly at (+34-93-241-77-24) or at heres@fullgenomics.es. It is recommended that patients receive appropriate genetic counseling to explain the implications of the test result, including its residual risks, uncertainties and reproductive or medical options.

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HERES-SEQ

SUPPLEMENTAL TABLE

Gene	Inheritance	Condition	Ethnicity	Carrier frequency	Detection rate	Post test carrier probability*	Post-test probability of having an affected child**
ABCD1	XL	Adrenoleukodystrophy, X-linked	General	1/21000	99%	1/2099901	1/8399604
AR	XL	Androgen Insensitivity Syndrome: Complete	General	1/14286	98%	1/714251	1/2857004
ATP7A	XL	Menkes disease	General	1/50000	99%	1/4999901	1/19999604
CFTR	AR	Cystic fibrosis	General	1/32	99%	1/3101	1/396928
			African American	1/61	99%	1/6001	1/1464244
			African	1/61	99%	1/6001	1/1464244
			Ashkenazi Jewish	1/24	99%	1/2301	1/220896
			European	1/25	99%	1/2401	1/240100
			East Asian	1/94	99%	1/9301	1/3497176
			Latino	1/58	99%	1/5701	1/1322632
CHM	XL	Choroideremia	General	1/25000	95%	1/499981	1/1999924
COL4A5	XL	Alport syndrome, COL4A5-related	General	1/139	98%	1/6901	1/27604
CYBB	XL	Chronic granulomatous disease, X-linked	General	1/149254	99%	1/14925301	1/59701204
DMD	XL	Duchenne muscular dystrophy (+)	General	1/2350	93%	1/33558	1/134232
DMD	XL	DMD-Related Muscular Dystrophies	General	1/2350	93%	1/33558	1/315445200
EDA	XL	Hypohidrotic ectodermal dysplasia	General	1/14167	99%	1/1416601	1/5666404
EMD	XL	Emery-Dreifuss muscular dystrophy	General	1/81967	99%	1/8196601	1/32786404
F8	XL	Hemophilia A	General	1/3250	48%	1/6249	1/24996
F9	XL	Hemophilia B	General	1/15000	99%	1/1499901	1/5999604
FMR1	XL	Fragile X syndrome	General	1/259	99%	1/25801	1/103204
			Ashkenazi Jewish	1/115	99%	1/11401	1/45604
G6PD	XL	Glucose-6-Phosphate Dehydrogenase Deficiency	General	1/7	98%	1/301	1/1204
GJB1	XL	Charcot-Marie-Tooth disease, X-linked type 1	General	1/667	90%	1/6661	1/26644
GJB2	AR	Nonsyndromic hearing loss, GJB2-related	General	1/42	99%	1/4101	1/688968
			African	1/25	99%	1/2401	1/240100
			African American	1/25	99%	1/2401	1/240100
			Ashkenazi Jewish	1/21	99%	1/2001	1/168084
			European	1/33	99%	1/3201	1/422532
			Latino	1/100	99%	1/9901	1/3960400
			Middle-Eastern	1/83	99%	1/8201	1/2722732
			South Asian/Indian	1/148	99%	1/14701	1/8702992
GLA	XL	Fabry disease	General	1/25000	99%	1/2499901	1/9999604
HBA1	AR, DG	Alpha thalassemia	General	1/20	90%	1/191	1/15280
			African	1/3	90%	1/21	1/252
			African American	1/3	90%	1/21	1/252
			Ashkenazi Jewish	1/13	90%	1/121	1/6292
			East Asian	1/8	90%	1/71	1/2272
			Middle-Eastern	1/3	90%	1/21	1/252
			South Asian/Indian	1/5	90%	1/41	1/820
HBA2	AR, DG	Alpha thalassemia	General	1/20	90%	1/191	1/15280
			African	1/3	90%	1/21	1/252
			African American	1/3	90%	1/21	1/252
			Ashkenazi Jewish	1/13	90%	1/121	1/6292
			East Asian	1/8	90%	1/71	1/2272
			Middle-Eastern	1/3	90%	1/21	1/252
			South Asian/Indian	1/5	90%	1/41	1/820
HBB	AR	Beta thalassemia (+)	General	1/158	95%	1/3141	1/1985112
			African	1/10	95%	1/181	1/7240
			African American	1/10	95%	1/181	1/7240
			East Asian	1/50	95%	1/981	1/196200
			Latino	1/128	95%	1/2541	1/1300992
			Mediterranean	1/3	95%	1/41	1/492
			South Asian/Indian	1/25	95%	1/481	1/48100
HBB	AR	Sickle cell disease (+)	General	1/158	95%	1/3141	1/1985112
			African	1/10	95%	1/181	1/7240
			African American	1/10	95%	1/181	1/7240
			East Asian	1/50	95%	1/981	1/196200
			Latino	1/128	95%	1/2541	1/1300992
			Mediterranean	1/3	95%	1/41	1/492
			South Asian/Indian	1/25	95%	1/481	1/48100
HBB	AR	Hemoglobinopathy: HbC (+)	General	1/158	95%	1/3141	1/1985112
			African	1/10	95%	1/181	1/7240
			African American	1/10	95%	1/181	1/7240
			East Asian	1/50	95%	1/981	1/196200
			Latino	1/128	95%	1/2541	1/1300992
			Mediterranean	1/3	95%	1/41	1/492
			South Asian/Indian	1/25	95%	1/481	1/48100
HBB	AR	HBB-related disorders	General	1/158	95%	1/3141	1/1985112
			African	1/10	95%	1/181	1/7240
			African American	1/10	95%	1/181	1/7240
			East Asian	1/50	95%	1/981	1/196200
			Latino	1/128	95%	1/2541	1/1300992
			Mediterranean	1/3	95%	1/41	1/492
			South Asian/Indian	1/25	95%	1/481	1/48100
IDS	XL	Mucopolysaccharidosis type II (Hunter syndrome)	General	1/50000	91%	1/555545	1/2222180
IL2RG	XL	Severe combined immunodeficiency, X-linked	General	1/25000	99%	1/2499901	1/9999604

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SUPPLEMENTAL TABLE

Gene	Inheritance	Condition	Ethnicity	Carrier frequency	Detection rate	Post test carrier probability*	Post-test probability of having an affected child**
MTM1	XL	Myotubular myopathy, X-linked	General	1/25000	98%	1/1249951	1/4999804
OCRL	XL	Dent disease 2 (+)	General	<1/250000	95%	<1/4999981	<1/19999924
OCRL	XL	Lowe syndrome (+)	General	1/250000	95%	1/4999981	1/19999924
OCRL	XL	OCRL-Related Disorders	General	1/250000	95%	1/4999981	1/19999924
OTC	XL	Ornithine transcarbamylase deficiency	General	1/7000	90%	1/69991	1/279964
PDHA1	XL	Pyruvate dehydrogenase E1-alpha deficiency	General	<1/250000	98%	<1/12499951	<1/49999804
PRPS1	XL	Arts syndrome (+)	General	<1/250000	98%	<1/12499951	<1/49999804
PRPS1	XL	Charcot-Marie-Tooth disease, X-linked recessive, 5 (+)	General	<1/250000	98%	<1/12499951	<1/49999804
PRPS1	XL	Phosphoribosylpyrophosphate synthetase superactivity (+)	General	<1/250000	98%	<1/12499951	<1/49999804
PRPS1	XL	Rosenberg-Chutorian syndrome (+)	General	<1/250000	98%	<1/12499951	<1/49999804
PRPS1	XL	PRPS1-Related Disorders	General	<1/250000	98%	<1/12499951	<1/49999804
RS1	XL	Juvenile retinoschisis, X-linked	General	1/2500	96%	1/62476	1/249904
SMN1	AR	Spinal Muscular Atrophy	General African African American Ashkenazi Jewish European East Asian Latino	1/54 1/72 1/72 1/67 1/47 1/59 1/68	91% 71% 71% 91% 95% 93% 90%	1/590 1/246 1/246 1/734 1/921 1/830 1/671	1/127440 1/70848 1/70848 1/196712 1/173148 1/195880 1/182512
WAS	XL	Severe Congenital Neutropenia, WAS-related (+)	General	1/125000	99%	1/12499901	1/49999604
WAS	XL	Thrombocytopenia, X-linked (+)	General	1/125000	99%	1/12499901	1/49999604
WAS	XL	Wiskott-Aldrich syndrome (+)	General	1/125000	99%	1/12499901	1/49999604
WAS	XL	WAS-Related Disorders	General	1/125000	99%	1/12499901	1/49999604

*For genes that have tested negative

**For genes that have tested negative and reproductive couple not tested.

Abbrevations: AR, autosomal recessive, XL, X-Linked, DG, digenic