



Patient name: DOB: Sex assigned at birth: Gender: Patient ID (MRN):	Male Man BFA0168	Sample type: Sample collection date: Sample accession date:	Blood 30-AUG-2022 06-SEP-2022	Report date: Invitae #: Clinical team:	22-SEP-2022 Valerie Shaikly
Reason for testing Gamete donor			t performed tae Carrier Screen		

Invitae primary panel (CF, SMA)

Add-on genes



RESULT: NEGATIVE

This carrier test evaluated 175 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 did not identify any genetic changes in the gene(s) analyzed that are currently recognized as clinically significant. This negative result reduces, but does not eliminate, the chance that this individual is a carrier for conditions caused by any of the genes tested. This individual may still be a carrier for a genetic condition that is not evaluated by this test.

Next steps

- Even for genes that have a negative test result, there is always a small risk that an individual could still be a carrier. This is called "residual risk." See the table below for residual risks, which presumes a negative family history of the conditions listed.
- 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.





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Results to note

FMR1

Normal triplet repeats observed: 30. CGG repeat ranges: normal (<45 CGG repeats), intermediate (45-54 CGG repeats), premutation (55-200 CGG repeats), full mutation (>200 CGG repeats).

SMN1

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

Pseudodeficiency allele(s)

- Benign change, c.742G>A (p.Asp248Asn), known to be a pseudodeficiency allele, 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.





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Residual risk

This table displays residual risks after a negative result for each of the genes and corresponding disorders. The values provided assume a negative family history and the absence of symptoms for each disorder. For genes associated with both dominant and recessive inheritance, the numbers in this table apply to the recessive condition(s) associated with the gene, unless otherwise noted. Residual risk values are provided for disorders when carrier frequency is greater than 1 in 500. For disorders with carrier frequency equal to, or less than, 1 in 500, residual risk is considered to be reduced substantially. When provided, residual risk values are inferred from published carrier frequencies, and estimated detection rates are based on testing technologies used at Invitae. Residual risks are provided only as a guide for assessing approximate risk given a negative result; values will vary based on the ethnic background of an individual. For individuals of mixed ethnicity, it is recommended to use the highest residual risk estimate. For any genes marked with an asterisk*, refer to the Limitations section below for detailed coverage information. In the case of a sample-specific limitation, "N/A" indicates that a residual risk value could not be calculated. AR = autosomal recessive, XL = X-linked, AD = autosomal dominant.

DISORDER (INHERITANCE)	GENE	ETHNICITY	CARRIER FREQUENCY BEFORE SCREENING	CARRIER RESIDUAL RISK AFTER NEGATIVE RESULT
3-hydroxy-3-methylglutaryl-CoA lyase deficiency (AR)	HMGCL	Pan-ethnic	≤1 in 500	Reduced
NM_000191.2	TIMOCE	Portuguese	1 in 160	1 in 15900
ABCC8-related conditions (AR)		Ashkenazi Jewish	1 in 52	1 in 5100
NM_000352.4		Finnish	1 in 100	1 in 9900
When the mother is a noncarrier, but the father is a carrier, there is a residual risk for focal disease (1 in 540 for the Ashkenazi Jewish population; undetermined in other ethnic groups)	ABCC8	Pan-ethnic	1 in 177	1 in 17600
Adenosine deaminase deficiency (AR) NM_00022.2	ADA	Pan-ethnic	1 in 224	1 in 2788
Alpha-mannosidosis (AR) NM_000528.3	MAN2B1	Pan-ethnic	1 in 354	1 in 35300
		African-American	1 in 30	1 in 291
Alpha-thalassemia (AR)	HBA1/	Asian	1 in 20	1 in 191
NM_000558.4, NM_000517.4	HBA2 *	Caucasian	≤1 in 500	Reduced
		Pan-ethnic	1 in 25	1 in 241
		Ashkenazi Jewish	1 in 192	1 in 19100
Alport syndrome (COL4A3-related) (AR) NM 000091.4	COL4A3	Caucasian	1 in 284	1 in 28300
NM_000091.4		Pan-ethnic	1 in 354	1 in 35300
Alport syndrome (COL4A4-related) (AR) NM_000092.4	COL4A4	Pan-ethnic	1 in 353	1 in 35200
Alport syndrome (COL4A5-related) (XL) NM_000495.4	COL4A5 *	Pan-ethnic	≤1 in 500	Reduced
Alström syndrome (AR) NM_015120.4	ALMS1	Pan-ethnic	≤1 in 500	Reduced
Arginase deficiency (AR) NM_000045.3	ARG1	Pan-ethnic	1 in 274	1 in 27300
Argininosuccinate lyase deficiency (AR) NM_000048.3	ASL	Pan-ethnic	1 in 133	1 in 1321
Aspartylglucosaminuria (AR)	AGA	Finnish	1 in 69	1 in 6800
NM_000027.3	AGA	Pan-ethnic	≤1 in 500	Reduced
Ataxia with vitamin E deficiency (AR)	ТТРА	Italian	1 in 274	1 in 2731
NM_000370.3	TIPA	Pan-ethnic	≤1 in 500	Reduced
ATM-related conditions (AR)	ATM	Pan-ethnic	1 in 100	1 in 9900
NM_000051.3		Sephardic Jewish	1 in 69	1 in 6800
ATP7A-related conditions (XL) NM_000052.6	ATP7A	Pan-ethnic	≤1 in 500	Reduced
		Finnish	1 in 79	1 in 7800
Autoimmune polyendocrinopathy with candidiasis and	AIRE	Pan-ethnic	1 in 150	1 in 14900
ectodermal dysplasia (AR) NM_000383.3	AIKE	Sardinian	1 in 60	1 in 5900
		Sephardic Jewish (Iranian)	1 in 48	1 in 4700
Autosomal recessive congenital ichthyosis		Norwegian	1 in 151	1 in 3000
(TGM1-related) (AR) NM_000359.2	TGM1	Pan-ethnic	1 in 224	1 in 4460





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DISORDER (INHERITANCE)	GENE	ETHNICITY	CARRIER FREQUENCY BEFORE SCREENING	CARRIER RESIDUAL RISK AFTER NEGATIVE RESULT		
Autosomal recessive spastic ataxia of Charlevoix- Saguenay (AR)	SACS	French Canadian (Saguenay-Lac-St- Jean)	1 in 21	1 in 2000		
NM_014363.5		Pan-ethnic	≤1 in 500	Reduced		
Bardet-Biedl syndrome (BBS10-related) (AR) NM_024685.3	BBS10	Pan-ethnic	1 in 354	1 in 35300		
Bardet-Biedl syndrome (BBS12-related) (AR) NM_152618.2	BBS12	Pan-ethnic	1 in 708	Reduced		
BBS1-related conditions (AR)	BBS1	Faroese	1 in 30	1 in 2900		
NM_024649.4	DDST	Pan-ethnic	1 in 330	1 in 32900		
BBS2-related conditions (AR)	BBS2	Ashkenazi Jewish	1 in 140	1 in 13900		
NM_031885.3	BB22	Pan-ethnic	1 in 560	Reduced		
		Caucasian	1 in 407	1 in 40600		
BCS1L-related conditions (AR) NM 004328.4	BCS1L	Finnish	1 in 108	1 in 2000 Reduced 1 in 35300 Reduced 1 in 2900 1 in 32900 1 in 13900 Reduced		
NW_001520.1		Pan-ethnic	≤1 in 500	Reduced		
Biopterin-deficient hyperphenylalaninemia (PTS-related)		Chinese	1 in 122	1 in 12100		
(AR) NM_000317.2	PTS	Pan-ethnic	1 in 433	1 in 43200		
Biotinidase deficiency (AR) NM_000060.3	BTD	Pan-ethnic	1 in 125	1 in 12400		
Bloom syndrome (AR)	BLM	Ashkenazi Jewish	1 in 100	1 in 9900		
NM_000057.3	BLIVI	Pan-ethnic	≤1 in 500	Reduced		
Canavan disease (AR)	ASPA	Ashkenazi Jewish	1 in 57	1 in 5600		
NM_000049.2	ASFA	Pan-ethnic	1 in 159	1 in 15800		
Carbamoyl phosphate synthetase I deficiency (AR) NM_001875.4	CPS1	Pan-ethnic	≤1 in 500	Reduced		

NM_000060.3	BTD	Pan-ethnic	1 in 125	1 in 12400
Bloom syndrome (AR)	BLM	Ashkenazi Jewish	1 in 100	1 in 9900
NM_000057.3	BLIVI	Pan-ethnic	≤1 in 500	Reduced
Canavan disease (AR)	4604	Ashkenazi Jewish	1 in 57	1 in 5600
VM_000049.2	ASPA	Pan-ethnic	1 in 159	1 in 15800
arbamoyl phosphate synthetase I deficiency (AR) IM_001875.4	CPS1	Pan-ethnic	≤1 in 500	Reduced
arnitine palmitoyltransferase I deficiency (AR)	CPT1A	Hutterite	1 in 16	1 in 1500
M_001876.3	CPITA	Pan-ethnic	≤1 in 500	Reduced
arnitine palmitoyltransferase II deficiency (AR)	CPT2	Ashkenazi Jewish	1 in 45	1 in 4400
M_000098.2	CFTZ	Pan-ethnic	1 in 182	1 in 18100
artilage-hair hypoplasia-anauxetic dysplasia spectrum		Amish	1 in 10	1 in 900
sorders (AR)	RMRP	Finnish	1 in 76	1 in 7500
IR_003051.3		Pan-ethnic	≤1 in 500	Reduced
erebrotendinous xanthomatosis (AR)	CYP27A1	Pan-ethnic	1 in 112	1 in 5550
NM_000784.3	CTP2/AI	Sephardic Jewish	1 in 76	1 in 3750
		African-American - classic CF	1 in 61	1 in 6000
		Ashkenazi Jewish - classic CF	1 in 29	1 in 2800
		Asian - classic CF	1 in 88	1 in 8700
EFTR-related conditions (AR)	CFTR	Caucasian - classic CF	1 in 28	1 in 2700
		Pan-ethnic - classic CF	1 in 45	1 in 4400
		Pan-ethnic - classic CF and CFTR- related disorders	1 in 9	1 in 800
itrullinemia type 1 (AR) IM_000050.4	ASS1	Pan-ethnic	1 in 120	1 in 2975
LN3-related conditions (AR) IM_001042432.1	CLN3	Pan-ethnic	1 in 230	1 in 22900
CLRN1-related conditions (AR)	CLRN1	Ashkenazi Jewish	1 in 120	1 in 11900
IM_174878.2	CLNII	Pan-ethnic	1 in 533	Reduced
obalamin C deficiency (AR) IM_015506.2	ММАСНС	Pan-ethnic	1 in 123	1 in 12200
Cockayne syndrome A (AR) NM_000082.3	ERCC8	Pan-ethnic	1 in 514	Reduced
ockayne syndrome B (AR) IM_000124.3	ERCC6	Pan-ethnic	1 in 377	1 in 37600
Cohen syndrome (AR)	VPS13B	Amish (Ohio)	1 in 12	1 in 1100
IM_017890.4	VPSIJD	Pan-ethnic	≤1 in 500	Reduced
Combined pituitary hormone deficiency (PROP1-related) (AR) NM_006261.4	PROP1	Pan-ethnic	1 in 45	1 in 2200





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Congenital adrenal hyperplasia due to 21-hydroxylase deficiency (AR) NM_000500.7	CYP21A2 *	Pan-ethnic	1 in 61	1 in 751
		Ashkenazi Jewish	1 in 61	1 in 6000
Congenital disorder of glycosylation type Ia (AR) NM 000303.2	PMM2	Caucasian	1 in 60	1 in 5900
NM_000303.2		Pan-ethnic	1 in 190	1 in 18900
Congenital disorder of glycosylation type Ib (AR) NM_002435.2	MPI	Pan-ethnic	≤1 in 500	Reduced
Congenital disorder of glycosylation type Ic (AR) NM_013339.3	ALG6 *	Pan-ethnic	≤1 in 500	Reduced
		Finnish	1 in 46	1 in 4500
Congenital nephrotic syndrome type 1 (AR) NM_004646.3	NPHS1	Old Order Mennonite	1 in 12	1 in 1100
		Pan-ethnic	≤1 in 500	Reduced
Congenital nephrotic syndrome type 2 (AR) NM_014625.3	NPHS2	Pan-ethnic	≤1 in 500	Reduced
CYP11B1-related conditions (AR)	CYP11B1	Pan-ethnic	1 in 194	1 in 19300
NM_000497.3	СТРПЫ	Sephardic Jewish (Moroccan)	1 in 40	1 in 3900
Cystinosis (AR)	CTNC	French Canadian (Saguenay-Lac-St- Jean)	1 in 39	1 in 3800
NM_004937.2	CTNS	Pan-ethnic	1 in 158	1 in 15700
		Sephardic Jewish (Moroccan)	1 in 100	1 in 9900
Dihydrolipoamide dehydrogenase deficiency (AR)	DLD	Ashkenazi Jewish	1 in 107	1 in 5300
NM_000108.4	DLD	Pan-ethnic	≤1 in 500	Reduced
DMD-related conditions (XL) NM_004006.2	DMD	Pan-ethnic	1 in 667	Reduced
DYSF-related conditions (AR)	DVCF	Pan-ethnic	1 in 311	1 in 31000
NM_003494.3	DYSF	Sephardic Jewish (Libyan)	1 in 10	1 in 900
Dyskeratosis congenita spectrum disorders		Ashkenazi Jewish	1 in 222	1 in 22100
(RTEL1-related) (AR) NM_001283009.1	RTEL1	Pan-ethnic	≤1 in 500	Reduced
Ellis-van Creveld syndrome (EVC-related) (AR)	EVC	Amish	1 in 8	1 in 700
NM_153717.2	LVC	Pan-ethnic	1 in 220	1 in 21900
EVC2-related conditions (AR) NM_147127.4	EVC2	Pan-ethnic	1 in 199	1 in 19800
Fabry disease (XL) NM_000169.2	GLA	Pan-ethnic	≤1 in 500	Reduced
Familial dysautonomia (AR)	ELP1	Ashkenazi Jewish	1 in 36	1 in 3500
NM_003640.3		Pan-ethnic	≤1 in 500	Reduced
		Armenian	1 in 8	1 in 71
Familial Mediterranean fever (AR)		Ashkenazi Jewish	1 in 13	1 in 121
NM_000243.2	MEFV	Pan-ethnic	1 in 64	1 in 631
		Sephardic Jewish	1 in 14	1 in 131
		Turkish	1 in 8	1 in 71
		Afrikaner	1 in 83	1 in 8200
Fanconi anemia type A (AR)	FANCA	Pan-ethnic	1 in 345	1 in 34400
NM_000135.2		Sephardic Jewish	1 in 133	1 in 13200
		Spanish Roma	1 in 64	1 in 6300
Fanconi anemia type C (AR)	FANCC	Ashkenazi Jewish	1 in 89	1 in 8800
NM_000136.2		Pan-ethnic	1 in 417	1 in 41600
FMR1-related conditions including fragile X syndrome		Ashkenazi Jewish	1 in 58	1 in 5700
	-	Asian	≤1 in 500	Reduced
(XL) NM_002024.5	FMR1 *	Caucasian	1 in 187	1 in 18600
INIM_002024.5		Hispanic	≤1 in 500	Reduced
		Pan-ethnic	1 in 259	1 in 25800
Galactokinase deficiency galactosemia (AR)	GALK1	Pan-ethnic	1 in 122	1 in 12100
NM_000154.1		Roma	1 in 47	1 in 4600
Galactosemia (GALT-related) (AR)	GALT	African-American	1 in 87	1 in 8600
NM_000155.3		Ashkenazi Jewish	1 in 156	1 in 15500





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DISORDER (INHERITANCE)	GENE	ETHNICITY	CARRIER FREQUENCY BEFORE SCREENING	CARRIER RESIDUAL RISK AFTER NEGATIVE RESULT
		Irish Traveller	1 in 11	1 in 1000
		Pan-ethnic	1 in 100	1 in 9900
GBA-related conditions including Gaucher disease (AR)	GBA *	Ashkenazi Jewish	1 in 15	1 in 234
NM_001005741.2	ODA "	Pan-ethnic	1 in 158	1 in 561
		Ashkenazi Jewish	1 in 13	1 in 1200
GJB2-related conditions (AR) NM_004004.5	GJB2	Pan-ethnic	1 in 50	1 in 4900
		Thai	1 in 9	1 in 800
		Pan-ethnic	1 in 158	1 in 15700
GLB1-related conditions (AR) NM_000404.2	GLB1	Roma	1 in 50	1 in 4900
		South Brazilian	1 in 58	1 in 5700
		Amish	1 in 9	1 in 800
Glutaric acidemia type I (AR) NM_000159.3	GCDH	Oji-Cree First Nations	1 in 9	1 in 800
NM_000139.5		Pan-ethnic	1 in 87	1 in 8600
Glycine encephalopathy (AMT-related) (AR)		Finnish	1 in 142	1 in 14100
NM_000481.3	AMT	Pan-ethnic	1 in 325	1 in 32400
Glycine encephalopathy (GLDC-related) (AR)		Caucasian	1 in 141	1 in 14000
NM_000170.2	GLDC	Pan-ethnic	1 in 165	1 in 16400
Glycogen storage disease type Ia (AR)		Ashkenazi Jewish	1 in 71	1 in 1400
NM_000151.3	G6PC	Pan-ethnic	1 in 177	1 in 3520
		African-American	1 in 60	1 in 5900
Glycogen storage disease type II (Pompe disease) (AR)		Ashkenazi Jewish	1 in 58	1 in 5700
NM 000152.3	GAA	Asian	1 in 112	1 in 11100
		Pan-ethnic	1 in 100	1 in 9900
		Faroese	1 in 28	1 in 540
Glycogen storage disease type III (AR)	AGL	Pan-ethnic	1 in 159	1 in 3160
NM_000642.2	AGE	Sephardic Jewish (Moroccan)	1 in 34	1 in 660
		Pan-ethnic	1 in 179	1 in 17800
GNE-related conditions (AR) NM_001128227.2	GNE	Sephardic Jewish (Iranian)	1 in 10	1 in 900
		Irish Traveller	1 in 15	1 in 1400
GNPTAB-related conditions (AR) NM_024312.4	GNPTAB	Pan-ethnic	1 in 200	1 in 19900
		Caucasian	1 in 250	1 in 24900
HADHA-related conditions (AR)	HADHA	Finnish	1 in 125	1 in 124900
NM_000182.4	HADHA	Pan-ethnic	1 in 350	
				1 in 34900
	_	African-American	1 in 8	1 in 700
	_	Asian	1 in 54	1 in 5300
HBB-related hemoglobinopathies (AR) NM_000518.4	НВВ	Caucasian	1 in 373	1 in 37200
NM_000318.4		Hispanic	1 in 17	1 in 1600
		Mediterranean	1 in 28	1 in 2700
		Pan-ethnic	1 in 49	1 in 4800
Hereditary fructose intolerance (AR)		African-American	1 in 226	1 in 22500
NM_000035.3	ALDOB	Middle Eastern	1 in 97	1 in 9600
		Pan-ethnic	1 in 122	1 in 12100
HGSNAT-related conditions (AR) NM_152419.2	HGSNAT	Pan-ethnic	≤1 in 500	Reduced
Holocarboxylase synthetase deficiency (AR)		Faroese	1 in 20	1 in 1900
Holocarboxylase synthetase deficiency (AR) NM_000411.6	HLCS	Japanese	1 in 158	1 in 15700
		Pan-ethnic	1 in 224	1 in 22300
Homocystinuria due to cystathionine beta-synthase		Norwegian	1 in 40	1 in 3900
deficiency (AR)	CBS	Pan-ethnic	1 in 224	1 in 22300
NM_000071.2		Qatari	1 in 21	1 in 2000
HSD17B4-related conditions (AR) NM_000414.3	HSD17B4	Pan-ethnic	1 in 158	1 in 15700
Hydrolethalus syndrome type 1 (AR)		Finnish	1 in 40	1 in 3900
NM_145014.2	HYLS1	Pan-ethnic	≤1 in 500	Reduced
Hypophosphatasia (AR)		Mennonite	1 in 25	1 in 480
NM_000478.5	ALPL	Pan-ethnic	1 in 150	1 in 2980





DOB:

Invig <th>DISORDER (INHERITANCE)</th> <th>GENE</th> <th>ETHNICITY</th> <th>CARRIER FREQUENCY BEFORE SCREENING</th> <th>CARRIER RESIDUAL RISK AFTER NEGATIVE RESULT</th>	DISORDER (INHERITANCE)	GENE	ETHNICITY	CARRIER FREQUENCY BEFORE SCREENING	CARRIER RESIDUAL RISK AFTER NEGATIVE RESULT
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(TMM 20173902 a) InstanceTMEM216Pan-ethnics1 in 500Reduced(MU 00175602 a) InstanceLAMC2Pan-ethnica1 in 500Reduced(KCN)11 - leated canditons (AR) MU 000523KCN)11Pan-ethnica1 in 500Reduced(KCN)11 - leated canditons (AR) MU 000153CALC - Pan-ethnicDiraza11 in 13011 in 1500(KCN)11 - leated canditons (AR)LAMA2Pan-ethnic11 in 13011 in 1500(LAMA2-calaed muccular dystrophy (AR)LAMA2Pan-ethnic11 in 310Reduced(LAMA3-related oxicular dystrophy (CAPN3-related) (AR)CAPN3Pan-ethnic11 in 317Reduced(LAMA3-related conditons (AR)LAMA3Pan-ethnic11 in 317Reduced(LAMA3-related conditons (AR)LAMA3CAPN3Caucasian11 in 13011 in 1300(Lambe girle muscular dystrophy (CAPN3-related) (AR)CAPN3Caucasian11 in 25011 in 2500(Lambe girle muscular dystrophy type 2C (AR)Caucasian11 in 25011 in 250011 in 2500(Lambe girle muscular dystrophy type 2D (AR)Caucasian11 in 25011 in 250011 in 2500(Lambe girle muscular dystrophy type 2D (AR)Caucasian11 in 25011 in 250011 in 2500(Lambe girle muscular dystrophy type 2D (AR)Caucasian11 in 25011 in 250011 in 2500(Lambe girle muscular dystrophy type 2D (AR)Caucasian11 in 2500Reduced(Lambe girle muscular dystrophy type 2D (AR)Caucasian11 in 250011 in 2500 <td></td> <td>MKS1</td> <td>Pan-ethnic</td> <td>1 in 260</td> <td>1 in 5180</td>		MKS1	Pan-ethnic	1 in 260	1 in 5180
NLL_0117390.2 intercal relation of All in SolReduced Reduced and in SolReduced Reduced and in SolReduced Reduced and in SolNLL_01525.3 NLL_00525.3ILAMC2 intercal relation NLL_00525.3Reduced intercal relation intercal relatio		THEMOLO	Ashkenazi Jewish	1 in 92	1 in 9100
NM_05522 NM_00233ChildDuicePar.ethnicLi in 300ReducedNM_00233 NM_002333CALC*DuceI in 6In in 500NM_002333CALC*Par.ethnic1 in 181 in 1700NM_00242 a.LMA2Par.ethnic1 in 871 in 660NM_00242 a.LMA3Par.ethnic1 in 370ReducedNM_00242 a.LMA3Par.ethnic1 in 370ReducedNM_00222 b.LMA3Par.ethnic1 in 3171 in 3160NM_00222 b.LMA3Par.ethnic1 in 3171 in 3160NM_00222 b.LMA3Par.ethnic1 in 3711 in 3180NM_00222 b.LMA3Par.ethnic1 in 1371 in 3180NM_00222 b.CAP13Par.ethnic1 in 1371 in 1390NM_00222 b.EXCLSGucacaian1 in 2501 in 2500NM_00232 b.SCCACaucaian1 in 2501 in 2500NM_00232 b.SCCACaucaian1 in 18001 in 2500NM_00232 b.SCCACaucaian1 in 1801 in 2500NM_00232 b.SCCACaucaian1 in 1801 in 1800NM_00232 b.SCCACaucaian1 in 1701 in 1800NM_00232 b.SCCACaucaian1 in 1701 in 1800NM_00232 b.SCCAPar.ethnic1 in 30ReducedNM_00232 b.SCCACaucaian1 in 1701 in 1800NM_00233 b.SCCAPar.ethnic1 in 30ReducedNM_00234 b.<		IMEMZI6	Pan-ethnic	≤1 in 500	Reduced
NML 000323 3RK (N) IIParteminRelucedRelucedNML 000333 3GAC 2DruceIin 6Iin 500NML 000333 3GAC 2ParethnicIin 6Iin 500NML 000424 3LMA2 veluced onutions (AR)LMA3ParethnicIin 317Iin 800NML 000227 4LMA3ParethnicIin 317ReducedNML 000227 4LMA3ParethnicIin 317Iin 31600NML 000228 2LMB3ParethnicIin 317ReducedNML 000228 4LMB3ParethnicIin 317ReducedNML 000228 4SCC 6CAPN3ParethnicIin 517ReducedNML 00023 2Reduced on thin 57ReducedIin 3200Iin 3200NML 00023 2SCC 6CaucasianIin 509Iin 2300NML 00023 2SCC 6CaucasianIin 250Iin 2500NML 00023 2SCC 7CaucasianIin 280Iin 2800NML 00023 2SCC 8CaucasianIin 1400Iin 1400NML 00023 2SCC 8CaucasianIin 140Iin 508NML 00032 3SCC 8CaucasianIin 140Iin 508NML 00032 3TARParethnicSI in 500ReducedNML 00032 3SCC 8CaucasianIin 140Iin 508NML 00032 3TARParethnicSI in 500ReducedNML 00032 4SCC 8CaucasianIin 140Iin 508NML 00032 5SCC 8CaucasianIin 140Iin 508NML 00032		LAMC2	Pan-ethnic	≤1 in 500	Reduced
NML 00033.3 (**)CALC*Pan ethnic1 in 1581 in 1570LAMA2-related conditions (AR)LAMA2Pan ethnic1 in 6701 in 680LAMA3-related conditions (AR)LAMA3Pan ethnic1 in 3100ReducedLAMA3-related conditions (AR)LAMB3Pan ethnic1 in 13171 in 31600LAMB3-related conditions (AR)LAMB3Pan ethnic1 in 1341 in 3300MA_300022.4LAMB3Pan ethnic1 in 1341 in 3300Lamb gielle muscular dystrophy (CAPN3-related) (AR)CAPN3Pan ethnic1 in 1571ReducedMA_300023.1ExecceI in 2501 in 2501 in 24900NM_00023.1ExecceCaucasian1 in 5201 in 2500NM_00023.2SCCGCaucasian1 in 1501 in 14900NM_00023.2SCCBCaucasian1 in 1501 in 14900NM_00023.2SCCBCaucasian1 in 1001 in 1600Limb-girdle muscular dystrophy type 2E (AR)SCCBCaucasian1 in 1001 in 1600NM_00023.4SCCBCaucasian1 in 1001 in 1600ReducedLimb-girdle muscular dystrophy type 2E (AR)SCCBCaucasian1 in 1101 in 1500NM_00023.4SCCBPan ethnic1 in 1501 in 1600NM_00023.5SCCBCaucasian1 in 1001 in 1600NM_00023.6SCCBPan ethnic1 in 1701 in 1500NM_00023.6SCCBSCCBNot the caucasian1 in 1001 in 100		KCNJ11	Pan-ethnic	≤1 in 500	Reduced
NML.00013-34Call Pan-ethnic1 in 1581 in 1570NML.00024.5.3LAMA2Pan-ethnic1 in 871 in 860NML.00025.6.4LAMA3Pan-ethnicsl in 500ReducedNML.00025.7LAMB3Pan-ethnicsl in 500ReducedLAMB3-related conditions (AR) NML.00025.8LAMB3Pan-ethnic1 in 3171 in 31600LAMB3-related conditions (AR) NML.00025.8LAMB3Pan-ethnic1 in 3741 in 3300LImb-girdle muscular dystrophy (CAPN3-related) (AR) NML.00025.2SCCEGaucasian1 in 771ReducedLImb-girdle muscular dystrophy type 2C (AR) NML.00025.2SCCEGaucasian1 in 3261 in 2500NML.00025.2SCCEGaucasian1 in 1501 in 14900NML.00025.2SCCECaucasian1 in 1501 in 14900NML.00025.2SCCECaucasian1 in 1501 in 14900NML.00025.2SCCECaucasian1 in 1501 in 14900NML.00025.4SCCEPan-ethnicsl in 500ReducedLimb-girdle muscular dystrophy type 2D (AR) NML.00025.4SCCECaucasian1 in 1501 in 14900NML.00025.4SCCEPan-ethnicsl in 500ReducedLipold congenital adrenal hyperplasia (AR) NML.00025.4SCCESCCESCCESCCESCCEPan-ethnic1 in 1501 in 1501 in 1501 in 50NML.00025.4SCEESCCEScenaria1 in 150ReducedLipold congenital adrenal hyper		GALC *	Druze	1 in 6	1 in 500
NML 00023.3 LMMA3 elited conditions (AR) LMMA3 elited conditions (AR)LMMA3 LMMA3Pan-ethnic Pan-ethnical in 500ReducedLMM3-related conditions (AR) LMM300028.7LMM3Pan-ethnic1 in 3171 in 31600LMM30-related conditions (AR) NML 000070.2CAN3Pan-ethnic1 in 3171 in 31800Limb-girdle muscular dystrophy (CPN3-related) (AB) NML 000070.2CAN3Pan-ethnic1 in 3741 in 3200Limb-girdle muscular dystrophy type 2C (AR) NML 000231.2SGCCCAucasian1 in 2501 in 2500Limb-girdle muscular dystrophy type 2D (AR) NML 0000231.2SGCACaucasian1 in 2501 in 2500Limb-girdle muscular dystrophy type 2D (AR) NML 0000232.4SGCACaucasian1 in 1001 in 1000Limb-girdle muscular dystrophy type 2E (AR) NML 000023.4SGCACaucasian1 in 4041 in 5038Limb-girdle muscular dystrophy type 2E (AR) NML 000023.4SGCACaucasian1 in 4041 in 5038Limb-girdle muscular dystrophy type 2E (AR) NML 00023.4SGCACaucasian1 in 4041 in 5038Lipoid congenital adrenal hyperplasia (AR) NML 00023.4SGCACaucasian1 in 1001 in 503Lipoid congenital adrenal hyperplasia (AR) NML 00023.4SGCACaucasian1 in 11001 in 503Lipoid congenital adrenal hyperplasia (AR) NML 00023.4SGCACaucasian1 in 14041 in 503Muscular dystrophy type 2E (AR) NML 00025.4SGCASGCACaucasian1 in 14041 in 503 <tr< td=""><td></td><td>GALC</td><td>Pan-ethnic</td><td>1 in 158</td><td>1 in 15700</td></tr<>		GALC	Pan-ethnic	1 in 158	1 in 15700
NML.00027.4LAWA3PanethnicRelicedLAMB3 related confilions (AR)LAMB3Panethnic1 in 3171 in 3160LAMB3 related confilions (AR)CAPN3Panethnic1 in 1341 in 1300NML00007.2CAPN3Panethnic1 in 1341 in 1300LImbgirdle muscular dystrophy (CAPN3-related) (AR)CAPN3Panethnic1 in 571ReducedNML00007.1Caucasian1 in 570ReducedNML00023.2SGCAMoroccan1 in 2801 in 2800NML00023.2SGCAFinnish1 in 1800ReducedNML00023.2SGCACaucasian1 in 4041 in 1930NML00023.2SGCAPanethnics1 in 500ReducedNML00023.2SGCAPanethnics1 in 500ReducedNML00023.4SGCAPanethnics1 in 500ReducedNML00023.4SGCACaucasian1 in 10401 in 503NML00023.5SGCAPanethnics1 in 500ReducedNML00023.5SGCAPanethnics1 in 500ReducedNML00023.5SGCAPanethnics1 in 500ReducedNML00023.5SGCAPanethnics1 in 1281 in 5930NML00023.5SGCAPanethnics1 in 500ReducedNML00023.5SGCAPanethnics1 in 500ReducedNML00023.6SGCAPanethnics1 in 500NGCANML00023.5SGCAPanethnics1 in 500NGCANML00023.6SGCA <td></td> <td>LAMA2</td> <td>Pan-ethnic</td> <td>1 in 87</td> <td>1 in 8600</td>		LAMA2	Pan-ethnic	1 in 87	1 in 8600
NML 000228.2 Lumb gird muscular dystrophy (CAPN3-related) (AR) NML 000070.2CAPN3 CAPN3 Pan-ethnicI in 131I in 13400Lumb-girdle muscular dystrophy type 2C (AR) NML 000231.2A SCCCBGaucasian1 in 574ReducedPan-ethnic-1 in 3741 in 37300Nat 2000Nat 2000Nat 2000Lumb-girdle muscular dystrophy type 2C (AR) NML 000231.2SCCCBMoroccan1 in 500ReducedRoma1 in 500Nat 2000Nat 2000Nat 2000Nat 2000Lumb-girdle muscular dystrophy type 2D (AR) NML 000232.4SCCCBCaucasian1 in 1001 in 1000Lumb-girdle muscular dystrophy type 2E (AR) NML 000232.4SCCCBCaucasian1 in 100ReducedLumb-girdle muscular dystrophy type 2E (AR) NML 000323.4STARPan-ethnic-1 in 500ReducedLumb-girdle muscular dystrophy type 2E (AR) NML 000324.4STARPan-ethnic-1 in 100ReducedLumb girdle muscular dystrophy type 2E (AR) NML 000325.3STARPan-ethnic-1 in 100ReducedLub do corgenital adrenal hyperplasia (AR) NML 00025.3BCKDHBPan-ethnic-1 in 1301 in 1540Maple syrup urine disease type 1A (AR) NML 00025.3BCKDHBPan-ethnic1 in 331 in 3540Maple syrup urine disease type 1A (AR) NML 000076.3BCKDHBPan-ethnic1 in 3461 in 3500Maple syrup urine disease type 1A (AR) NML 000076.3BCKDHBPan-ethnic1 in 1361 in 1360Maple syrup urine disease type 2 (AR) NML 000		LAMA3	Pan-ethnic	≤1 in 500	Reduced
NML.000070.2CAPHSPartemineIn 19.30In 19.300Imbgindle muscular dystrophy type 2C (AR) NM_000231.2 $A = A = A = A = A = A = A = A = A = A =$		LAMB3	Pan-ethnic	1 in 317	1 in 31600
$ \frac{1}{M_{0}00231.2} \frac{1}{M_{0}00231.2} \frac{1}{M_{0}00231.2} \frac{1}{M_{0}00231.2} \frac{1}{M_{0}00231.2} \frac{1}{M_{0}00231.2} \frac{1}{M_{0}00231.2} \frac{1}{M_{0}000231.2} \frac{1}{M_{0}00023.2} \frac{1}{M_{0$		CAPN3	Pan-ethnic	1 in 134	1 in 13300
$ \begin{array}{c} \mberline mascular dystrophy type 2C (AR) \\ ML 000231.2 \\ \mberline mascular dystrophy type 2D (AR) \\ ML 0000232.4 \\ \mberline mascular dystrophy type 2D (AR) \\ ML 0000232.4 \\ \mberline mascular dystrophy type 2D (AR) \\ ML 0000232.4 \\ \mberline mascular dystrophy type 2E (AR) \\ ML 0000232.4 \\ \mberline mascular dystrophy type 2E (AR) \\ ML 0000232.4 \\ \mberline mascular dystrophy type 2E (AR) \\ ML 0000232.4 \\ \mberline mascular dystrophy type 2E (AR) \\ ML 0000232.4 \\ \mberline mascular dystrophy type 2E (AR) \\ ML 0000232.4 \\ \mberline mascular dystrophy type 2E (AR) \\ ML 0000232.4 \\ \mberline mascular dystrophy type 2E (AR) \\ ML 0000349.2 \\ \mberline mascular dystrophy type 2E (AR) \\ ML 0000349.2 \\ \mberline mascular dystrophy type 2E (AR) \\ ML 0000349.2 \\ \mberline mascular dystrophy type 2E (AR) \\ ML 0000349.2 \\ \mberline mascular dystrophy type 2E (AR) \\ ML 0000349.2 \\ \mberline mascular dystrophy type 2E (AR) \\ ML 0000349.2 \\ \mberline mascular dystrophy type 2E (AR) \\ ML 0000349.2 \\ \mberline mascular dystrophy type 2E (AR) \\ ML 0000349.2 \\ \mberline mascular dystrophy type 2E (AR) \\ ML 000035.3 \\ \mberline mascular dystrophy type 2E (AR) \\ ML 0000703 \\ \mberline mascular dystrophy type 2E (AR) \\ \mberline mascular dystrophy type 2E (AR) \\ \mberline mascular dystrophy type 2 (AR) \\ \mberline disease type 1B (AR) \\ \mberline disease type 1B (AR) \\ \mberline disease type 2 (AR) \\ \$			Caucasian	1 in 571	Reduced
$\begin{split} \text{NM}_000231.2 \text{Letr} \text{A} \text{A} \text{Letr} \text{A} \text{Letr} \text{A} \text{Letr} \text{A} \text{A} \text{Letr} \text{A} \text{Letr} \text{A} \text{Letr} \text{A} \text{A} \text{Letr} \text{A} \text{A} \text{Letr} \text{A} \text{Letr} \text{A} \text{A} \text{Letr} \text{A} \text{A} \text{Letr} \text{A} \text{A} \text{Letr} \text{A} \text{A} \text{A} \text{Letr} \text{A} \text{A} \text{A} \text{Letr} \text{A} \text{A} \text{A} \text{Letr} \text{A} \text{A} \text{A} \text{A} \text{A} \text{A} \text{A} A$			Japanese	1 in 374	1 in 37300
		SGCG	Moroccan	1 in 250	1 in 24900
$\begin{split} \begin{tabular}{ c c c c } \hline \begin{tabular}{ c c c c } \hline \begin{tabular}{ c c c c c c } \hline \begin{tabular}{ c c c c c c c } \hline \begin{tabular}{ c c c c c c c c c c c c c c c c c c c$	NM_000231.2		Pan-ethnic	≤1 in 500	Reduced
$\begin{split} \begin{tabular}{ c c c c c c } \begin{tabular}{ c c c c } \begin{tabular}{ c c c c c } \end{tabular} \\ \begin{tabular}{ c c c c c c c c c c c c c c c c c c c$			Roma	1 in 59	1 in 5800
NM_000023.2SCAFind is of the iso of the is			Caucasian	1 in 286	1 in 28500
IndexPan-thnicIn 500ReducedLimb-girdle muscular dystrophy type 2E (AR) MU_000232.4SCGBCaucasian1 in 4041 in 5008Lippoid congenital adrenal hyperplasia (AR) NM_000239.2STARKorean1 in 1701 in 16900Lippoid congenital adrenal hyperplasia (AR) NM_000239.2STARKorean1 in 1701 in 1800Lysosomal acid lipase deficiency (AR) NM_000235.3LIPACaucasian1 in 1201 in 5907Maple syrup urine disease type 1A (AR) NM_000735.3BCKDHA Pan-ethnic1 in 3731 in 3700Maple syrup urine disease type 1B (AR) NM_000736.3BCKDHA Pan-ethnic1 in 3731 in 3700Maple syrup urine disease type 2 (AR) NM_0001918.3BCKDHA PAn-ethnic1 in 3600ReducedMaple syrup urine disease type 2 (AR) NM_0001918.3DBTPan-ethnic1 in 6601 in 3500Medium-chain acyl-CoA dehydrogenase deficiency (AR) NM_00016.5ACADM MCIPan-ethnic1 in 6601 in 3900Medium-chain acyl-CoA dehydrogenase deficiency (AR) NM_00016.5ACADM MCIPan-ethnic1 in 6001 in 3900Medium-chain acyl-CoA dehydrogenase deficiency (AR) NM_00087.5MACAPan-ethnic1 in 4001 in 3900Medium-chain acyl-CoA dehydrogenase (Start action acidemia (MMAA-related) (AR) NM_00349.2MACAPan-ethnic1 in 6001 in 3900Medium-chain acidemia (MMAA-related) (AR) NM_00349.5MMAAPan-ethnic1 in 4001 in 3900Methylmalonic acidemia (MMAA-related) (AR)		SGCA	Finnish	1 in 150	1 in 14900
NM_000232.4SCCBPan-ethnic $\leq 1 \ln 500$ ReducedLipoid congenital adrenal hyperplasia (AR) NM_000349.2STARKorean1 ln 1701 in 16900Lysosomal acid lipase deficiency (AR) NM_000255.3LIPAPan-ethnic $\leq 1 \ln 500$ ReducedLysosomal acid lipase deficiency (AR) NM_000255.3LIPARennethnic1 in 331 in 534Maple syrup urine disease type 1A (AR) NM_000709.3BCKDHAMennonite1 in 3731 in 37200Maple syrup urine disease type 1B (AR) NM_000709.3BCKDHAAshkenazi flwish1 in 971 in 9600Maple syrup urine disease type 2 (AR) NM_01918.3DBTPan-ethnic1 in 3461 in 34500Maple syrup urine disease type 2 (AR) NM_0001918.3DBTPan-ethnic1 in 600ReducedMedium-chain acyl-CoA dehydrogenase deficiency (AR) NM_00016.5ACADMNorthern European1 in 401 in 3900Medium-chain acyl-CoA dehydrogenase deficiency NM_0015166.3ACADMNavajo1 in 401 in 3900Metachromatic leukodystrophy (ARSA-related) (AR) NM_000487.5ARSAPan-ethnic1 in 1001 in 1900Methylmalonic acidemia (MMAA-related) (AR) NM_00247.5MMAAPan-ethnic1 in 3161 in 10500Methylmalonic acidemia (MMAA-related) (AR) NM_002353.3MMABPan-ethnic1 in 3161 in 10500Methylmalonic acidemia (MMAA-related) (AR) NM_002345.5MMAAPan-ethnic1 in 2041 in 5075Methylmalonic acidemia (MMAA-related) (AR) NM_002345.3MUT<	NNI_000023.2		Pan-ethnic	≤1 in 500	Reduced
NM_000232.4SNCMPan-ethnic $\leq 1 \text{ in 500}$ ReducedLipoid congenital adrenal hyperplasia (AR) NM_000349.2STARKorean1 in 1701 in 1990Lipoid congenital adrenal hyperplasia (AR) NM_000349.2STARRan-ethnic1 in 1701 in 1890Lipoid congenital adrenal hyperplasia (AR) NM_000253.3LIPAPan-ethnic1 in 1301 in 1850Maple syrup urine disease type 1A (AR) NM_000253.3BCKDH8Mennonite1 in 331 in 37200Maple syrup urine disease type 1B (AR) NM_183050.2BCKDH8Ashkenazi Jewish1 in 971 in 9600Maple syrup urine disease type 2 (AR) NM_0001918.3DBTPan-ethnic1 in 3461 in 34500Maple syrup urine disease type 2 (AR) NM_0001918.3DBTPan-ethnic1 in 6601 in 6500Medium-chain acyl-CoA dehydrogenase deficiency (AR) NM_00016.5ACADMNorthern European1 in 6601 in 3900Medium-chain acyl-CoA dehydrogenase deficiency (AR) NM_00016.5ACADMPan-ethnic1 in 6601 in 3900Medium-chain acyl-CoA dehydrogenase deficiency (AR) NM_00016.5ACADMPan-ethnic1 in 6601 in 3900Medium-chain acyl-CoA dehydrogenase deficiency (AR) NM_000487.5ACADMPan-ethnic1 in 4001 in 3900Methylmalonic acidemia (MMAA-related) (AR) NM_002847.5ARSAPan-ethnic1 in 4001 in 3900Methylmalonic acidemia (MMAA-related) (AR) NM_002847.5MMABPan-ethnic1 in 3161 in 10500Methylmalonic acidemia (MMAA-related	Limb-girdle muscular dystrophy type 2E (AR)	CCCD	Caucasian	1 in 404	1 in 5038
$\frac{Pan-ethnic}{Pan-ethnic}$		SUCR	Pan-ethnic	≤1 in 500	Reduced
NML00039.2Image: section of the section	Lipoid congenital adrenal hyperplasia (AR)	CTAD	Korean	1 in 170	1 in 16900
$ \frac{\mbox{Lysosomal acid lipase deficiency (AR)}{\mbox{M}_000235.3} & LIPA \\ \begin{tabular}{ llllllllllllllllllllllllllllllllllll$	NM_000349.2	STAR	Pan-ethnic	≤1 in 500	Reduced
NM_000235.3LIPAParterinicIn 1939In 1939In 1959Maple syrup urine disease type 1A (AR) NM_000709.3 $BCKDHA$ Mennonite1 in 331 in 534Maple syrup urine disease type 1B (AR) NM_0183050.2 $BCKDHB$ $Ashkenazi Jewish$ 1 in 971 in 9600Maple syrup urine disease type 2 (AR) NM_001918.3 $BCKDHB$ $Ashkenazi Jewish$ 1 in 3461 in 34500Maple syrup urine disease type 2 (AR) NM_001918.3 BT Pan-ethnic1 in 3461 in 34500Medium-chain acyl-CoA dehydrogenase deficiency (AR) NM_00016.5 $ACADM$ Northern European1 in 661 in 6500Megalencephalic leukoencephalopathy with subcortical cysts 1 (AR) NM_0015166.3 $MLC1$ Sephardic Jewish (Libyan)1 in 401 in 3900Metachromatic leukodystrophy (ARSA-related) (AR) NM_00255.3 $ARSA$ Pan-ethnic1 in 401 in 780Methylmalonic acidemia (MMAA-related) (AR) NM_002553.3MMAAPan-ethnic1 in 3161 in 10500Methylmalonic acidemia (MUT-related) (AR) NM_002553.3MMABPan-ethnic1 in 4561 in 22750Mitochondrial complex IV deficiency / Leigh syndrome, French Canadian type (AR)MITPan-ethnic1 in 2341 in 2300Mitochondrial complex IV deficiency / Leigh syndrome, French Canadian type (AR)MITPan-ethnic1 in 2341 in 5075			Caucasian	1 in 112	1 in 1850
Sephadic jewish (Iranian)1 in 331 in 534Maple syrup urine disease type 1A (AR) M_000709.3BCKDHAPan-ethnic1 in 101 in 900Maple syrup urine disease type 1B (AR) N_183050.2BCKDHBPan-ethnic1 in 3731 in 37200Maple syrup urine disease type 2 (AR) N_0001918.3DBTPan-ethnic1 in 34601 in 34500Maple syrup urine disease type 2 (AR) N_00016.5DBTPan-ethnic1 in 4001 in 3900Medium-chain acyl-CoA dehydrogenase deficiency (AR) N_000016.5ACADMNorthern European1 in 6601 in 6500Medium-chain acyl-CoA dehydrogenase deficiency (AR) N_000016.5MCC1Sephardic Jewish (Libyan)1 in 4001 in 3900Medium-chain acyl-CoA dehydrogenase deficiency (AR) N_000016.5MC1Sephardic Jewish (Libyan)1 in 4001 in 3900Medium-chain acyl-CoA dehydrogenase deficiency (AR) N_00016.5MC1Sephardic Jewish (Libyan)1 in 4001 in 3900Medium-chain acyl-CoA dehydrogenase deficiency (AR) N_00016.5MAC1Sephardic Jewish (Libyan)1 in 4001 in 3900Medium-chain acyl-CoA dehydrogenase deficiency (AR) N_0015166.3MMAAPan-ethnic1 in 4001 in 3900Methylmalonic acidemia (MMAA-related) (AR) N_0017250.2MMAAPan-ethnic1 in 4001 in 900Methylmalonic acidemia (MMAB-related) (AR) N_002553.3MMAAPan-ethnic1 in 4561 in 22750Methylmalonic acidemia (MUAF-related) (AR) N_002553.3MUTPan-ethnic1 in 231 in 2200Mitt		LIPA	Pan-ethnic	1 in 359	1 in 5967
$\frac{BCKDHA}{M_000709.3} \frac{BCKDHA}{M_00709.3} \frac{BCKDHA}{M_00709.3} \frac{BCKDHA}{M_00709.3} \frac{BCKDHA}{M_00709.3} \frac{BCKDHA}{M_00709.3} \frac{BCKDHA}{M_000709.3} \frac{BCKDHA}{M_000709.3} \frac{BCKDHA}{M_000709.3} \frac{BCKDHA}{M_000709.3} \frac{BCKDHA}{M_000709.3} \frac{BCKDHA}{M_000709.3} \frac{ASKDHA}{M_000709.3} \frac{ASKDHA}{M_000709.3} \frac{ASKDHA}{M_000709.3} \frac{ASKDHA}{M_000709.3} \frac{BCKDHA}{M_000709.3} \frac{ASKDHA}{M_000709.3} \frac{BCKDHA}{M_000709.3} \frac{ASKDHA}{M_000709.3} \frac{ASKDHA}{M_000709.3} \frac{ASKDHA}{M_000709.3} \frac{ASKDHA}{M_000709.3} \frac{ASCDHA}{M_000709.3} \frac{ASCDHA}{M_00700070.3} \frac{ASCDHA}{M_00700000000$	NNI_000233.5		Sephardic Jewish (Iranian)	1 in 33	1 in 534
NM_000709.3Art of the formation of the synch	Maple syrup urine disease type 1A (AR)	DOKDULA	Mennonite	1 in 10	1 in 900
$\frac{\text{BCKDHB}}{\text{Maple synup urine disease type 10 km/s}} = \frac{\text{BCKDHB}}{\text{Pan-ethnic}} = \frac{1 \text{ in 346}}{1 \text{ in 34500}} = \frac{1 \text{ in 3460}}{1 \text{ in 34500}}$ $\frac{\text{Maple synup urine disease type 2 (AR)}{\text{Maple synup urine disease type 2 (AR)}} = \frac{\text{DBT}}{\text{ACADM}} = \frac{\text{Pan-ethnic}}{\text{Pan-ethnic}} = \frac{1 \text{ in 40}}{1 \text{ in 66}} = \frac{1 \text{ in 3900}}{1 \text{ in 66}}$ $\frac{\text{Meduced}}{1 \text{ in 6600}} = \frac{1 \text{ in 6600}}{1 \text{ in 65000}} = \frac{1 \text{ in 6600}}{1 \text{ in 65000}} = \frac{1 \text{ in 6600}}{1 \text{ in 65000}}$ $\frac{\text{Megalencephalic leukoencephalopathy with subcortical cysts 1 (AR)}{\text{NM_00015166.3}} = \frac{\text{MLC1}}{\text{MLC1}} = \frac{\text{Navajo}}{1 \text{ in 40}} = \frac{1 \text{ in 40}}{1 \text{ in 78000}} = \frac{1 \text{ in 7800}}{1 \text{ in 78000}}$ $\frac{\text{Metachromatic leukodystrophy (ARSA-related) (AR)}{\text{NM_000487.5}} = \frac{\text{MMAA}}{1 \text{ in 78000}} = \frac{1 \text{ in 40}}{1 \text{ in 780000}} = \frac{1 \text{ in 660000}}{1 \text{ in 780000}}$ $\frac{\text{Methylmalonic acidemia (MMAA-related) (AR)}}{\text{NM_000487.5}} = \frac{\text{MMAA}}{1 \text{ in 780000}} = \frac{1 \text{ in 40}}{1 \text{ in 780000}} = 1 \text{ in 660000000000000000000000000000000000$	NM_000709.3	вскрна	Pan-ethnic	1 in 373	1 in 37200
NM_183050.2EXCMBPan-ethnic1 in 3461 in 34500Maple syrup urine disease type 2 (AR) NM_001918.3DBTPan-ethnics1 in 500ReducedMedium-chain acyl-CoA dehydrogenase deficiency (AR) NM_000016.5ACADMNorthern European1 in 401 in 3900Megalencephalic leukoencephalopathy with subcortical cysts 1 (AR) NM_015166.3ACADMNorthern European1 in 401 in 3900Metachromatic leukodystrophy (ARSA-related) (AR) NM_000487.5MLC1Sephardic Jewish (Libyan)1 in 401 in 780Methylmalonic acidemia (MMAA-related) (AR) NM_052845.3MMAAPan-ethnic1 in 3161 in 10500Methylmalonic acidemia (MUT-related) (AR) NM_00255.3MMABPan-ethnic1 in 4561 in 22750Mitochondrial complex IV deficiency / Leigh syndrome, French Canadian type (AR)MUTPan-ethnic1 in 2041 in 5075Mitochondrial complex IV deficiency / Leigh syndrome, French Canadian type (AR)LRPPRCFrench Canadian (Saguenay-Lac-St- Jean)1 in 231 in 2200	Maple syrup urine disease type 1B (AR)	DCKDUB	Ashkenazi Jewish	1 in 97	1 in 9600
NM_001918.3Match and barDB1Pan-etnnicS1 in 500ReducedMedium-chain acyl-CoA dehydrogenase deficiency (AR) NM_000016.5ACADMNorthern European1 in 401 in 3900Megalencephalic leukoencephalopathy with subcortical cysts 1 (AR) NM_015166.3MLC1Pan-ethnics1 in 500ReducedMetachromatic leukodystrophy (ARSA-related) (AR) NM_000487.5MLC1Sephardic Jewish (Libyan)1 in 401 in 780Methylmalonic acidemia (MMAA-related) (AR) NM_052845.3MMAAPan-ethnic1 in 3161 in 1000Methylmalonic acidemia (MUT-related) (AR) NM_000255.3MMABPan-ethnic1 in 4561 in 22750Mitochondrial complex IV deficiency / Leigh syndrome, French Canadian type (AR)MLTFrench Canadian (Saguenay-Lac-St- Jean)1 in 231 in 2200		вскрнв	Pan-ethnic	1 in 346	1 in 34500
NM_000016.5ACADMPan-ethnic1 in 661 in 6500Megalencephalic leukoencephalopathy with subcortical cysts 1 (AR) NM_015166.3MLC1Pan-ethnic≤1 in 500ReducedMLC1Sephardic Jewish (Libyan)1 in 401 in 3900Metachromatic leukodystrophy (ARSA-related) (AR) NM_000487.5ARSANavajo1 in 401 in 780Metachromatic leukodystrophy (ARSA-related) (AR) NM_172250.2MMAAPan-ethnic1 in 1001 in 1980Methylmalonic acidemia (MMAA-related) (AR) NM_172250.2MMAAPan-ethnic1 in 3161 in 10500Methylmalonic acidemia (MMAB-related) (AR) NM_052845.3MMABPan-ethnic1 in 4561 in 22750Methylmalonic acidemia (MUT-related) (AR) NM_002255.3MUTPan-ethnic1 in 2041 in 5075Mitochondrial complex IV deficiency / Leigh syndrome, French Canadian type (AR)LRPPRCFrench Canadian (Saguenay-Lac-St- Jean)1 in 231 in 2200		DBT	Pan-ethnic	≤1 in 500	Reduced
NM_000016.5In 66In 6500Megalencephalic leukoencephalopathy with subcortical cysts 1 (AR) NM_015166.3MLC1Pan-ethnic≤1 in 500ReducedMetachromatic leukodystrophy (ARSA-related) (AR) NM_000487.5MASANavajo1 in 401 in 780Metachromatic leukodystrophy (ARSA-related) (AR) NM_000487.5ARSAPan-ethnic1 in 1001 in 1980Methylmalonic acidemia (MMAA-related) (AR) NM_172250.2MMAAPan-ethnic1 in 3161 in 200Methylmalonic acidemia (MMAB-related) (AR) NM_052845.3MMABPan-ethnic1 in 4561 in 22750Methylmalonic acidemia (MUT-related) (AR) NM_000255.3MUTPan-ethnic1 in 2041 in 5075Mitochondrial complex IV deficiency / Leigh syndrome, French Canadian type (AR)LRPPRCFrench Canadian (Saguenay-Lac-St- Jean)1 in 231 in 2200			Northern European	1 in 40	1 in 3900
cysts 1 (AR) NM_015166.3MLC1Sephardic Jewish (Libyan)1 in 401 in 3900Metachromatic leukodystrophy (ARSA-related) (AR) NM_000487.5ARSANavajo1 in 401 in 780Metachromatic leukodystrophy (ARSA-related) (AR) NM_000487.5ARSAPan-ethnic1 in 1001 in 1980Methylmalonic acidemia (MMAA-related) (AR) NM_172250.2MMAAPan-ethnic1 in 3161 in 10500Methylmalonic acidemia (MMAB-related) (AR) NM_052845.3MMABPan-ethnic1 in 4561 in 22750Methylmalonic acidemia (MUT-related) (AR) NM_00255.3MUTPan-ethnic1 in 2041 in 5075Mitochondrial complex IV deficiency / Leigh syndrome, French Canadian type (AR)LRPPRCFrench Canadian (Saguenay-Lac-St- Jean)1 in 231 in 2200	NM_000016.5		Pan-ethnic	1 in 66	1 in 6500
MM_015166.3Constraint of the sephardic Jewish (Libyan)1 in 401 in 3900Metachromatic leukodystrophy (ARSA-related) (AR) NM_000487.5ARSANavajo1 in 401 in 780Methylmalonic acidemia (MMAA-related) (AR) NM_172250.2MMAAPan-ethnic1 in 1001 in 1980Methylmalonic acidemia (MMAB-related) (AR) NM_052845.3MMABPan-ethnic1 in 3161 in 2050Methylmalonic acidemia (MUT-related) (AR) NM_00255.3MMABPan-ethnic1 in 4561 in 22750Mitochondrial complex IV deficiency / Leigh syndrome, French Canadian type (AR)LRPPRCFrench Canadian (Saguenay-Lac-St- Jean)1 in 231 in 2200			Pan-ethnic	≤1 in 500	Reduced
Metachromatic leukodystrophy (ARSA-related) (AR) ARSA Pan-ethnic 1 in 100 1 in 1980 Methylmalonic acidemia (MMAA-related) (AR) MMAA Sephardic Jewish 1 in 46 1 in 900 Methylmalonic acidemia (MMAA-related) (AR) MMAA Pan-ethnic 1 in 316 1 in 10500 Methylmalonic acidemia (MMAB-related) (AR) MMAB Pan-ethnic 1 in 456 1 in 22750 Methylmalonic acidemia (MUT-related) (AR) MUT Pan-ethnic 1 in 204 1 in 5075 Mitochondrial complex IV deficiency / Leigh syndrome, French Canadian type (AR) LRPPRC French Canadian (Saguenay-Lac-St-Jean) 1 in 23 1 in 2200		MLC1	Sephardic Jewish (Libyan)	1 in 40	1 in 3900
NM_000487.5ARSAPan-etrnicIn 100I in 100I in 1980Methylmalonic acidemia (MMAA-related) (AR)MMAAPan-ethnic1 in 3161 in 10500Methylmalonic acidemia (MMAB-related) (AR)MMABPan-ethnic1 in 4561 in 22750Methylmalonic acidemia (MUT-related) (AR)MUTPan-ethnic1 in 2041 in 5075Mitochondrial complex IV deficiency / Leigh syndrome, French Canadian type (AR)LRPPRCFrench Canadian (Saguenay-Lac-St- Jean)1 in 231 in 2200			,		
Methylmalonic acidemia (MMAA-related) (AR)MMAAPan-ethnic1 in 461 in 900Methylmalonic acidemia (MMAA-related) (AR)MMAAPan-ethnic1 in 3161 in 10500Methylmalonic acidemia (MMAB-related) (AR)MMABPan-ethnic1 in 4561 in 22750Methylmalonic acidemia (MUT-related) (AR)MUTPan-ethnic1 in 2041 in 5075Mitochondrial complex IV deficiency / Leigh syndrome, French Canadian type (AR)LRPPRCFrench Canadian (Saguenay-Lac-St- Jean)1 in 231 in 2200		ARSA	Pan-ethnic	1 in 100	1 in 1980
NM_172250.2MMAAPan-ethnicI in 316I in 10500Methylmalonic acidemia (MMAB-related) (AR)MMABPan-ethnic1 in 4561 in 22750Methylmalonic acidemia (MUT-related) (AR)MUTPan-ethnic1 in 2041 in 5075Methylmalonic acidemia (MUT-related) (AR)MUTPan-ethnic1 in 2041 in 5075Mitochondrial complex IV deficiency / Leigh syndrome, French Canadian type (AR)LRPPRCFrench Canadian (Saguenay-Lac-St- Jean)1 in 231 in 2200			Sephardic Jewish	1 in 46	1 in 900
NM_052845.3MMABPan-ethnicI in 436I in 22750Methylmalonic acidemia (MUT-related) (AR) NM_000255.3MUTPan-ethnic1 in 2041 in 5075Mitochondrial complex IV deficiency / Leigh syndrome, French Canadian type (AR)LRPPRCFrench Canadian (Saguenay-Lac-St- Jean)1 in 231 in 2200		MMAA	Pan-ethnic	1 in 316	1 in 10500
NM_000255.3 MOT Pan-etrinic I in 204 I in 5075 Mitochondrial complex IV deficiency / Leigh syndrome, French Canadian type (AR) LRPPRC French Canadian (Saguenay-Lac-St- Jean) 1 in 23 1 in 2200		ММАВ	Pan-ethnic	1 in 456	1 in 22750
French Canadian type (AR) LRPPRC Jean)		MUT	Pan-ethnic	1 in 204	1 in 5075
		LRPPRC		1 in 23	1 in 2200
	NM_133259.3		Pan-ethnic	≤1 in 500	Reduced





Patient name: Invitae #: DOB:

DISORDER (INHERITANCE)	GENE	ETHNICITY	CARRIER FREQUENCY BEFORE SCREENING	CARRIER RESIDUAL RISK AFTER NEGATIVE RESULT
Mucolipidosis type III gamma (AR) NM_032520.4	GNPTG	Pan-ethnic	≤1 in 500	Reduced
Mucolipidosis type IV (AR)	MCOLN1	Ashkenazi Jewish	1 in 100	1 in 9900
NM_020533.2	MCOLINI	Pan-ethnic	≤1 in 500	Reduced
Mucopolysaccharidosis type I (AR) NM_000203.4	IDUA	Pan-ethnic	1 in 148	1 in 4900
Mucopolysaccharidosis type II (XL) NM_000202.6	IDS *	Pan-ethnic	≤1 in 500	Reduced
Mucopolysaccharidosis type IIIA (AR)		Northern European	1 in 173	1 in 17200
NM_000199.3	SGSH	Pan-ethnic	1 in 215	1 in 21400
		Taiwanese	≤1 in 500	Reduced
Mucopolysaccharidosis type IIIB (AR) NM_000263.3	NAGLU	Pan-ethnic	1 in 224	1 in 22300
Muscular dystrophy-dystroglycanopathy (FKRP-related)	FKDD	Norwegian	1 in 116	1 in 11500
(AR) NM_024301.4	FKRP	Pan-ethnic	1 in 158	1 in 15700
Muscular dystrophy-dystroglycanopathy (FKTN-related)		Ashkenazi Jewish	1 in 80	1 in 7900
(AR) NM 001079802.1	FKTN	Japanese	1 in 188	1 in 18700
		Pan-ethnic	≤1 in 500	Reduced
MYO7A-related conditions (AR) NM_000260.3	MYO7A	Pan-ethnic	1 in 200	1 in 3980
Nemaline myopathy 2 (AR)	NEB *	Ashkenazi Jewish	1 in 108	1 in 10700
NM_001271208.1		Pan-ethnic	1 in 158	1 in 3140
Neuronal ceroid lipofuscinosis type 1 (AR)	PPT1	Finnish	1 in 70	1 in 3450
NM_000310.3		Pan-ethnic	1 in 199	1 in 9900
Neuronal ceroid lipofuscinosis type 2 (AR)	TPP1	Newfoundland	1 in 53	1 in 1734
NM_000391.3		Pan-ethnic	1 in 250	1 in 8300
Neuronal ceroid lipofuscinosis type 5 (AR)	CLN5	Finnish	1 in 115	1 in 11400
NM_006493.2		Pan-ethnic	≤1 in 500	Reduced
Neuronal ceroid lipofuscinosis type 6 (AR) NM_017882.2	CLN6	Pan-ethnic	≤1 in 500	Reduced
Neuronal ceroid lipofuscinosis type 8 (AR)	CLN8	Finnish	1 in 135	1 in 13400
NM_018941.3		Pan-ethnic	≤1 in 500	Reduced
Niemann-Pick disease type C (NPC1-related) (AR) NM_000271.4	NPC1	Pan-ethnic	1 in 183	1 in 18200
Niemann-Pick disease type C (NPC2-related) (AR) NM_006432.3	NPC2	Pan-ethnic	1 in 871	Reduced
Niemann-Pick disease types A and B (AR)	SMPD1	Ashkenazi Jewish	1 in 90	1 in 1780
NM_000543.4	51111 01	Pan-ethnic	1 in 250	1 in 4980
Nijmegen breakage syndrome (AR)	NBN *	Eastern European	1 in 155	1 in 15400
NM_002485.4		Pan-ethnic	≤1 in 500	Reduced
OPA3-related conditions (AR)	OPA3	Pan-ethnic	≤1 in 500	Reduced
NM_025136.3		Sephardic Jewish (Iraqi)	1 in 10	1 in 900
Ornithine transcarbamylase deficiency (XL) NM_000531.5	OTC	Pan-ethnic	≤1 in 500	Reduced
Osteopetrosis (TCIRG1-related) (AR)		Ashkenazi Jewish	1 in 350	1 in 34900
NM_006019.3	TCIRG1	Chuvash	1 in 30	1 in 2900
		Pan-ethnic	1 in 317	1 in 31600
PCDH15-related conditions (AR)	PCDH15	Ashkenazi Jewish	1 in 78	1 in 7700
NM_033056.3		Pan-ethnic	1 in 400	1 in 39900
PEX7-related conditions (AR) NM_000288.3	PEX7	Pan-ethnic	1 in 157	1 in 15600
		African-American	1 in 111	1 in 11000
		Ashkenazi Jewish	1 in 225	1 in 22400
Phenylalanine hydroxylase deficiency (AR)	PAH	East Asian	1 in 50	1 in 1225
NM_000277.1		Finnish	1 in 225	1 in 22400
		Irish	1 in 33	1 in 3200
		Japanese	1 in 200	1 in 19900





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CARRIER FREQUENCY BEFORE SCREENING	CARRIER RESIDUAL RISK AFTER NEGATIVE RESULT

DISORDER (INHERITANCE)	GENE	ETHNICITY	BEFORE SCREENING	
		Pan-ethnic	1 in 58	1 in 5700
		Turkish	1 in 26	1 in 2500
Polycystic kidney disease (PKHD1-related) (AR) NM_138694.3	PKHD1	Pan-ethnic	1 in 70	1 in 6900
POMGNT1-related conditions (AR)	DOMONITI	Finnish	1 in 111	1 in 11000
NM_017739.3	POMGNT1	Pan-ethnic	≤1 in 500	1 in 6900 1 in 11000 Reduced 1 in 800 1 in 9900 1 in 7000 1 in 13400 Reduced 1 in 35300 1 in 2475 1 in 5575 1 in 5575 1 in 1900 1 in 43700 1 in 43700 1 in 180 1 in 1900 1 in 1900 1 in 1900 1 in 22300 1 in 1900 1 in 2490 1 in 1400 1 in 17900 1 in 24980 Reduced 1 in 24900 1 in 24900 1 in 24900 1 in 2200 Reduced 1 in 7300 1 in 7300 1 in 7300 1 in 7900 1 in 7900
		Faroese	1 in 9	1 in 800
Primary carnitine deficiency (AR) NM_003060.3	SLC22A5	Japanese	1 in 100	
		Pan-ethnic	1 in 71	1 in 7000
Primary hyperoxaluria type 1 (AR) NM_000030.2	AGXT	Pan-ethnic	1 in 135	1 in 13400
Primary hyperoxaluria type 2 (AR) NM_012203.1	GRHPR	Pan-ethnic	≤1 in 500	Reduced
Primary hyperoxaluria type 3 (AR) NM_138413.3	HOGA1	Pan-ethnic	1 in 354	1 in 35300
Propionic acidemia (PCCA-related) (AR)	PCCA	Arab	1 in 100	
NM_000282.3		Pan-ethnic	1 in 224	
Propionic acidemia (PCCB-related) (AR)		Arab	1 in 100	
NM_000532.4	PCCB	Greenlandic Inuit	1 in 20	
		Pan-ethnic	1 in 224	1 in 22300
Pycnodysostosis (AR) NM_000396.3	СТЅК	Pan-ethnic	1 in 438	
Pyruvate carboxylase deficiency (AR)	PC	Algonquian Indian	1 in 10	
VM_000920.3		Pan-ethnic	1 in 250	1 in 4980
Roberts syndrome (AR) NM_001017420.2	ESCO2	Pan-ethnic	≤1 in 500	Reduced
Sandhoff disease (AR)	HEXB	Metis (Saskatchewan)	1 in 15	
VM_000521.3		Pan-ethnic	1 in 180	
ialic acid storage diseases (AR)	SLC17A5	Finnish	1 in 100	
VM_012434.4		Pan-ethnic	≤1 in 500	
ijögren-Larsson syndrome (AR) IM_000382.2	ALDH3A2	Pan-ethnic Swedish	≤1 in 500 1 in 250	
SLC12A6-related conditions (AR) NM_133647.1	SLC12A6	French Canadian (Saguenay-Lac-St- Jean)	1 in 23	1 in 2200
NN_155047.1		Pan-ethnic	≤1 in 500	Reduced
LC26A2-related conditions (AR)	SLC26A2	Finnish	1 in 75	1 in 1480
VM_000112.3	SECZUAZ	Pan-ethnic	1 in 158	1 in 3140
LC26A4-related conditions (AR)	SLC26A4	Asian	1 in 74	
JM_000441.1	02020711	Pan-ethnic	1 in 80	1 in 7900
SLC37A4-related conditions (AR) NM_001164277.1	SLC37A4	Pan-ethnic	1 in 354	1 in 7060
		African-American	1 in 339	1 in 33800
		Ashkenazi Jewish	1 in 41	1 in 4000
mith-Lemli-Opitz syndrome (AR)		Hispanic	1 in 135	1 in 13400
VM_001360.2	DHCR7	Northern European	1 in 50	1 in 4900
		Pan-ethnic	1 in 71	1 in 7000
		Sephardic Jewish	1 in 68	1 in 6700
pastic paraplegia type 15 (AR)	ZFYVE26	Southern European	1 in 83	1 in 8200
NM_015346.3	ZFIVEZO	Pan-ethnic	≤1 in 500	Reduced
Spinal muscular atrophy (AR) NM_000344.3		African-American	1 in 59	1 in 342
		Ashkenazi Jewish	1 in 62	1 in 1017
	SMN1 *	Asian	1 in 50	1 in 701
Carrier residual risks listed are for 2 copy SMN1 results. Carrier residual risk for >2 copies are 5- to 10-fold lower.		Caucasian	1 in 45	1 in 880
		Hispanic	1 in 48	1 in 784
		Pan-ethnic	1 in 49	1 in 800
Spondylocostal dysostosis (MESP2-related) (AR)	MESP2	Pan-ethnic	1 in 224	1 in 22300
NM_001039958.1		Puerto Rican	1 in 55	1 in 5400





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DISORDER (INHERITANCE)	GENE	ETHNICITY	CARRIER FREQUENCY BEFORE SCREENING	CARRIER RESIDUAL RISK AFTER NEGATIVE RESULT
		Ashkenazi Jewish	1 in 27	1 in 2600
		Asian	1 in 126	AFTER NEGATIVE RESULT
Tou Sacha diagona (AD)		Caucasian	1 in 182	1 in 18100
Tay-Sachs disease (AR) NM_000520.4	HEXA	French Canadian	1 in 27	1 in 2600
		Irish	1 in 41	1 in 4000
		Pan-ethnic	1 in 250	1 in 24900
		Sephardic Jewish	1 in 125	1 in 12400
Tyrosine hydroxylase deficiency (AR)	тн	Caucasian	1 in 224	1 in 22300
NM_199292.2		Pan-ethnic	≤1 in 500	Reduced
		Ashkenazi Jewish	1 in 143	1 in 2840
Tyrosinemia type I (AR)		French Canadian	1 in 66	1 in 1300
NM_000137.2	FAH *	Jean)	1 in 300	
		Pan-ethnic	1 in 125	1 in 2480
Tyrosinemia type II (AR) NM_000353.2	TAT	Pan-ethnic	1 in 250	1 in 24900
		French Canadian/Acadian	1 in 227	1 in 22600
USH1C-related conditions (AR) NM_005709.3	USH1C *	Pan-ethnic	1 in 353	1 in 3521
NW_003703.3		Sephardic Jewish	1 in 125	1 in 1241
		Caucasian	1 in 70	1 in 6900
USH2A-related conditions (AR) NM_206933.2	USH2A	Pan-ethnic	1 in 112	1 in 11100
NM_200933.2		Sephardic Jewish	1 in 36	1 in 3500
Very long-chain acyl-CoA dehydrogenase deficiency (AR) NM_000018.3	ACADVL	Pan-ethnic	1 in 100	1 in 9900
		Ashkenazi Jewish	1 in 67	1 in 3300
		Canary Islander	1 in 25	1 in 1200
Wilson disease (AR) NM_000053.3	ATP7B	· · · · · · · · · · · · · · · · · · ·	1 in 4450	
NM_000033.3		1 in 50	1 in 2450	
		Sephardic Jewish	1 in 65	1 in 3200
X-linked adrenoleukodystrophy (XL)	10001	Pan-ethnic	1 in 16800	Reduced
NM_000033.3	ABCD1	Sephardic Jewish	≤1 in 500	Reduced
X-linked juvenile retinoschisis (XL) NM_000330.3	RS1	Pan-ethnic	≤1 in 500	Reduced
X-linked myotubular myopathy (XL) NM_000252.2	MTM1	Pan-ethnic	≤1 in 500	Reduced
X-linked severe combined immunodeficiency (XL) NM_000206.2	IL2RG	Pan-ethnic	≤1 in 500	Reduced
Xeroderma pigmentosum complementation group A		Japanese	1 in 100	1 in 9900
(AR) NM_000380.3	XPA	Pan-ethnic	1 in 1667	Reduced
Xeroderma pigmentosum complementation group C	VDC	Pan-ethnic	1 in 763	Reduced
(AR) NM_004628.4	XPC	Tunisian	1 in 50	1 in 4900
Zellweger spectrum disorder (PEX1-related) (AR) NM_000466.2	PEX1	Pan-ethnic	1 in 144	1 in 14300
Zellweger spectrum disorder (PEX2-related) (AR)	PEX2	Ashkenazi Jewish	1 in 227	1 in 22600
NM_000318.2	1 2/12	Pan-ethnic	≤1 in 500	Reduced
Zallwager spectrum disorder (DEX6 related) (AD)		French Canadian	1 in 55	1 in 5400
Zellweger spectrum disorder (PEX6-related) (AR) NM_000287.3	PEX6	Pan-ethnic	1 in 294	1 in 29300
		Sephardic Jewish	1 in 18	1 in 1700
Zellweger spectrum disorder (PEX10-related) (AR) NM_153818.1	PEX10	Pan-ethnic	1 in 606	Reduced
Zellweger spectrum disorder (PEX12-related) (AR) NM_000286.2	PEX12	Pan-ethnic	1 in 409	1 in 40800



) INVITAE CARRIER SCREEN RESULTS

Patient name:

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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 below. Enrichment and analysis focus on the coding sequence of the indicated transcripts, 10bp of flanking intronic sequence, and other specific genomic regions demonstrated to be causative of disease at the time of assay design. Promoters, untranslated regions, and other non-coding regions are not otherwise interrogated. Exonic deletions and duplications are called using an in-house algorithm that determines copy number at each target by comparing the read depth for each target in the proband sequence with both mean read-depth and read-depth distribution, obtained from a set of clinical samples. Markers across the X and Y chromosomes are analyzed for quality control purposes and may detect deviations from the expected sex chromosome complement. Such deviations may be included in the report in accordance with internal guidelines. Invitae utilizes a classification methodology to identify next-generation sequencing (NGS)-detected variants that require orthogonal confirmation (Lincoln, et al. J Mol Diagn. 2019 Mar;21(2):318-329.). Confirmation of the presence and location of reportable variants is performed based on stringent criteria established by Invitae (1400 16th Street, San Francisco, CA 94103, #05D2040778), as needed, using one of several validated orthogonal approaches (PubMed ID 30610921). The following analyses are performed if relevant to the requisition. For GBA the reference genome has been modified to mask the sites of polymorphic paralog sequence variants (PSVs) in both the gene and pseudogene. For CYP21A2 and GBA, if one or more reportable variants, gene conversion, or fusion event is identified via our NGS pipeline (see Limitations), these variants are confirmed by PacBio sequencing of an amplicon generated by long-range PCR and subsequent shortrange PCR. In some cases, it may not be possible to disambiguate between the gene and pseudogene. For HBA1/2, the reference genome has been modified to force some sequencing reads derived from HBA1 to align to HBA2, and variant calling algorithms are modified to support an expectation of 4 alleles in these regions. HBA1/2 copy number calling is performed by a custom hypothesis testing algorithm which generates diplotype calls. If sequence data for a sample does not support a unique high confidence match from among hypotheses tested, that sample is flagged for manual review. Copy number variation is only reported for coding sequence of HBA1 and HBA2 and the HS-40 region. This assay does not distinguish among the $-\alpha$ 3.7 subtypes, and all $-\alpha$ 3.7 variants are called as HBA1 deletions. This assay may not detect overlapping copy gain and copy loss events when the breakpoints of those events are similar. For FMR1, triplet repeats are detected by PCR with fluorescently labeled primers followed by capillary electrophoresis. Reference ranges: Normal: <45 CGG repeats, intermediate: 45-54 CGG repeats, premutation: 55-200 CGG repeats, full mutation: >200 CGG repeats. For alleles with 55-90 triplet repeats, the region surrounding the FMR1 repeat is amplified by PCR. The PCR amplicons are then processed through PacBio SMRTBell library prep and sequenced using PacBio long read technology. The number of AGG interruptions within the 55-90 triplet repeat is read directly from the resulting DNA sequences. Technical component of confirmatory sequencing is performed by Invitae Corporation (1400 16th Street, San Francisco, CA 94103, #05D2040778).
- The following transcripts were used in this analysis. 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: ABCC8 (NM_000352.4), ABCD1 (NM_000033.3), ACADM (NM_000016.5), ACADVL (NM_000018.3), ADA (NM_000022.2), AGA (NM_000027.3), AGL (NM_000642.2), AGXT (NM_000030.2), AIRE (NM_000383.3), ALDH3A2 (NM_000382.2), ALDOB (NM_000035.3), ALG6 (NM_013339.3), ALMS1 (NM_015120.4), ALPL (NM_000478.5), AMT (NM_000481.3), ARG1 (NM_000045.3), ARSA (NM_000487.5), ASL (NM_000048.3), ASPA (NM_000049.2), ASS1 (NM_000050.4), ATM (NM_000051.3), ATP7A (NM_000052.6), ATP7B (NM_000053.3), BBS1 (NM_024649.4), BBS10 (NM_024685.3), BBS12 (NM_152618.2), BBS2 (NM_031885.3), BCKDHA (NM_000709.3), BCKDHB (NM_183050.2), BCS1L (NM_004328.4), BLM (NM_000057.3), BTD (NM_000060.3), CAPN3 (NM_000070.2), CBS (NM_000071.2), CFTR (NM_000492.3), CLN3 (NM_001042432.1), CLN5 (NM_006493.2), CLN6 (NM_017882.2), CLN8 (NM_018941.3), CLRN1 (NM_174878.2), COL4A3 (NM_000091.4), COL4A4 (NM_000092.4), COL4A5 (NM_000495.4), CPS1 (NM_001875.4), CPT1A (NM_001876.3), CPT2 (NM_00098.2), CTNS (NM_004937.2), CTSK (NM_000396.3), CYP11B1 (NM_000497.3), CYP21A2 (NM_000500.7), CYP27A1 (NM_000784.3), DBT (NM_001918.3), DHCR7 (NM_001360.2), DLD (NM_000108.4), DMD (NM_004006.2), DYSF (NM_003494.3), ELP1 (NM_003640.3), ERCC6 (NM_000124.3), ERCC8 (NM_000082.3), ESCO2 (NM_001017420.2), EVC (NM_153717.2), EVC2 (NM_147127.4), FAH (NM_000137.2), FANCA (NM_000135.2), FANCC (NM_000136.2), FKRP (NM_024301.4), FKTN (NM_001079802.1), FMR1 (NM_002024.5), G6PC (NM_000151.3), GAA (NM_000152.3), GALC (NM_000153.3), GALK1 (NM_000154.1), GALT (NM_000155.3), GBA (NM_001005741.2), GCDH (NM_000159.3), GJB2 (NM_004004.5), GLA (NM_000169.2), GLB1 (NM_000404.2), GLDC (NM_000170.2), GNE (NM_001128227.2), GNPTAB (NM_024312.4), GNPTG (NM_032520.4), GRHPR (NM_012203.1), HADHA (NM_000182.4), HBA1 (NM_000558.4), HBA2 (NM_000517.4), HBB (NM_000518.4), HEXA (NM_000520.4), HEXB (NM_000521.3), HGSNAT (NM_152419.2), HLCS (NM_000411.6), HMGCL (NM_000191.2), HOGA1 (NM_138413.3), HSD17B4 (NM_000414.3), HYLS1 (NM_145014.2), IDS (NM_000202.6), IDUA (NM_000203.4), IL2RG (NM_000206.2), IVD (NM_002225.3), KCNJ11 (NM_000525.3), LAMA2 (NM_000426.3), LAMA3 (NM_000227.4), LAMB3 (NM_000228.2), LAMC2 (NM_005562.2), LIPA (NM_000235.3), LRPPRC (NM_133259.3), MAN2B1 (NM_000528.3), MCOLN1 (NM_020533.2), MEFV (NM_000243.2), MESP2 (NM_001039958.1), MKS1 (NM_017777.3), MLC1 (NM_015166.3), MMAA (NM_172250.2), MMAB (NM_052845.3), MMACHC (NM_015506.2), MPI (NM_002435.2), MTM1 (NM_000252.2), MUT (NM_000255.3), MYO7A (NM_000260.3),





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NAGLU (NM_000263.3), NBN (NM_002485.4), NEB (NM_001271208.1), NPC1 (NM_000271.4), NPC2 (NM_006432.3), NPHS1 (NM_004646.3), NPHS2 (NM_014625.3), OPA3 (NM_025136.3), OTC (NM_000531.5), PAH (NM_000277.1), PC (NM_000920.3), PCCA (NM_000282.3), PCCB (NM_000532.4), PCDH15 (NM_033056.3), PEX1 (NM_000466.2), PEX10 (NM_153818.1), PEX12 (NM_000286.2), PEX2 (NM_000318.2), PEX6 (NM_000287.3), PEX7 (NM_000288.3), PKHD1 (NM_138694.3), PMM2 (NM_000303.2), POMGNT1 (NM_017739.3), PPT1 (NM_000310.3), PROP1 (NM_006261.4), PTS (NM_000317.2), RMRP (NR_003051.3), RS1 (NM_000330.3), RTEL1 (NM_001283009.1), SACS (NM_014363.5), SGCA (NM_00023.2), SGCB (NM_000232.4), SGCG (NM_000231.2), SGSH (NM_000199.3), SLC12A6 (NM_133647.1), SLC17A5 (NM_012434.4), SLC22A5 (NM_003060.3), SLC26A2 (NM_000112.3), SLC26A4 (NM_000441.1), SLC37A4 (NM_001164277.1), SMN1 (NM_000344.3), SMPD1 (NM_000543.4), STAR (NM_000349.2), TAT (NM_000353.2), TCIRG1 (NM_006019.3), TGM1 (NM_000359.2), TH (NM_199292.2), TMEM216 (NM_001173990.2), TPP1 (NM_000391.3), TTPA (NM_000370.3), USH1C (NM_005709.3), USH2A (NM_206933.2), VPS13B (NM_017890.4), XPA (NM_000380.3), XPC (NM_004628.4), ZFYVE26 (NM_015346.3).

- 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) 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.</p>
- FMR1: Sizing accuracy is expected to be +/-1 for CGG repeat alleles less than or equal to 90 repeat units and +/-3 for CGG repeat alleles greater than 90 repeat units. If the two CGG repeats listed are the same, this may indicate that both alleles are the same size or that one allele is too small to be detected by this analysis. The number of AGG interruptions is only determined for females with triplet repeat sizes of 55-90. 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.Phe2521le), 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



INVITAE CARRIER SCREEN RESULTS

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(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". IDS: Detection of complex rearrangements not offered (PMID: 7633410, 20301451). 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. COL4A5: Deletion/duplication analysis is not offered for exons 11-12. 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. NBN: Deletion/duplication analysis is not offered for exons 15-16. USH1C: Deletion/duplication analysis is not offered for exons 5-6. 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. FAH: Deletion/duplication analysis is not offered for exon 14. GALC: Deletion/duplication analysis is not offered for exon 6. 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. HBA2: Sequencing analysis is not offered for exons 1-2. ALG6: Deletion/duplication analysis is not offered for exons 11-12.

This report has been reviewed and approved by:

megh

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