

Donor 6056

Genetic Testing Summary

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

Last Updated: 4/21/20

Donor Reported Ancestry: Swedish, Scandinavian, French Canadian, Salvadoran Jewish Ancestry: No

Genetic Test*	Result	Comments/Donor's Residual Risk**
---------------	--------	----------------------------------

Chromosome analysis (karyotype)	Normal male karyotype	No evidence of clinically significant chromosome abnormalities
Hemoglobin evaluation	Normal hemoglobin fractionation and MCV/MCH results	Reduced risk to be a carrier for sickle cell anemia, beta thalassemia, alpha thalassemia trait (aa/ and a-/a-) and other hemoglobinopathies
Cystic Fibrosis (CF) carrier screening	Negative by gene sequencing in the CFTR gene	1/440
Spinal Muscular Atrophy (SMA) carrier screening	Negative for deletions of exon 7 in the SMN1 gene	1/4900
Expanded Genetic Disease Carrier Screening Panel attached- 283 diseases by gene sequencing	Negative for genes sequenced	

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

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





Patient Information

Name: Donor 6056

Client ID:

Date of Birth
Sema4

Indication: Carrier Testing

Specimen Information

Specimen Type: Blood
Date Collected: 09/09/2019
Date Received: 09/10/2019
Final Report: 09/24/2019

Referring Provider

Fairfax Cryobank, Inc.

Expanded Carrier Screen (283) Minus TSE

Number of genes tested: 283

SUMMARY OF RESULTS AND RECOMMENDATIONS

Negative

Negative for all genes tested

To view a full list of genes and diseases tested please see Table 1 in this report

Recommendations

- CGG repeat analysis of *FMR1* for fragile X syndrome is not performed on males as repeat expansion of premutation alleles is not expected in the male germline.
- Individuals of Asian, African, Hispanic and Mediterranean ancestry should also be screened for hemoglobinopathies by CBC and hemoglobin electrophoresis.
- Consideration of residual risk by ethnicity after a negative carrier screen is recommended for the other diseases on the panel, especially in the case of a positive family history for a specific disorder.

Test description

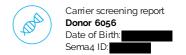
Minneenar

This patient was tested for a panel of diseases using a combination of sequencing, targeted genotyping and copy number analysis. Please note that negative results reduce but do not eliminate the possibility that this individual is a carrier for one or more of the disorders tested. Please see Table 1 for a list of genes and diseases tested, and **go.sema4.com/residualrisk** for specific detection rates and residual risk by ethnicity. With individuals of mixed ethnicity, it is recommended to use the highest residual risk estimate. Only variants determined to be pathogenic or likely pathogenic are reported in this carrier screening test.

Rebekah Zimmerman, Ph.D., FACMG, Laboratory Director

Laboratory Medical Consultant: George A. Diaz, M.D., Ph.D.





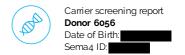
Genes and diseases tested

For specific detection rates and residual risk by ethnicity, please visit go.sema4.com/residualrisk

Table 1: List of genes and diseases tested with detailed results

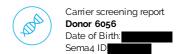
	Disease	Gene	Inheritance Pattern	Status	Detailed Summary
Θ	Negative				
	3-Beta-Hydroxysteroid Dehydrogenase Type II Deficiency	HSD3B2	AR	Reduced Risk	
	3-Methylcrotonyl-CoA Carboxylase Deficiency (<i>MCCC1</i> -Related)	MCCC1	AR	Reduced Risk	
	3-Methylcrotonyl-CoA Carboxylase Deficiency (<i>MCCC2</i> -Related)	MCCC2	AR	Reduced Risk	
	3-Methylglutaconic Aciduria, Type III	OPA3	AR	Reduced Risk	
	3-Phosphoglycerate Dehydrogenase Deficiency	PHGDH	AR	Reduced Risk	
	6-Pyruvoyl-Tetrahydropterin Synthase Deficiency	PTS	AR	Reduced Risk	
	Abetalipoproteinemia	MTTP	AR	Reduced Risk	
	Achromatopsia	CNGB3	AR	Reduced Risk	
	Acrodermatitis Enteropathica	SLC39A4	AR	Reduced Risk	
	Acute Infantile Liver Failure	TRMU	AR	Reduced Risk	
	Acyl-CoA Oxidase I Deficiency	ACOX1	AR	Reduced Risk	
	Adenosine Deaminase Deficiency	ADA	AR	Reduced Risk	
	Adrenoleukodystrophy, X-Linked	ABCD1	XL	Reduced Risk	
	Aicardi-Goutieres Syndrome (SAMHD1-Related)	SAMHD1	AR	Reduced Risk	
	Alpha-Mannosidosis	MAN2B1	AR	Reduced Risk	
	Alpha-Thalassemia	HBA1/HBA2	AR	Reduced Risk	HBA1 Copy Number: 2 HBA2 Copy Number: 2 No pathogenic copy number variants detected HBA1/HBA2 Sequencing: Negative
	Alpha-Thalassemia Mental Retardation Syndrome	ATRX	XL	Reduced Risk	
	Alport Syndrome (COL4A3-Related)	COL4A3	AR	Reduced Risk	-
	Alport Syndrome (COL4A4-Related)	COL4A4	AR	Reduced Risk	
	Alport Syndrome (COL4A5-Related)	COL4A5	XL	Reduced Risk	
	Alstrom Syndrome	ALMS1	AR	Reduced Risk	
	Andermann Syndrome	SLC12A6	AR	Reduced Risk	
	Argininosuccinic Aciduria	ASL	AR	Reduced Risk	
	Aromatase Deficiency	CYP19A1	AR	Reduced Risk	
	Arthrogryposis, Mental Retardation, and Seizures	SLC35A3	AR	Reduced Risk	





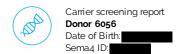
Asparagine Synthetase Deficiency	ASNS	AR	Reduced Risk
Aspartylglycosaminuria	AGA	AR	Reduced Risk
Ataxia With Isolated Vitamin E Deficiency	TTPA	AR	Reduced Risk
Ataxia-Telangiectasia	ATM	AR	Reduced Risk
Autosomal Recessive Spastic Ataxia of Charlevoix- Saguenay	SACS	AR	Reduced Risk
Bardet-Biedl Syndrome (BBS10-Related)	BBS10	AR	Reduced Risk
Bardet-Biedl Syndrome (<i>BBS12</i> -Related)	BBS12	AR	Reduced Risk
Bardet-Biedl Syndrome (<i>BBS1</i> -Related)	BBS1	AR	Reduced Risk
Bardet-Biedl Syndrome (BBS2-Related)	BBS2	AR	Reduced Risk
Bare Lymphocyte Syndrome, Type II	CIITA	AR	Reduced Risk
Bartter Syndrome, Type 4A	BSND	AR	Reduced Risk
Bernard-Soulier Syndrome, Type A1	GP1BA	AR	Reduced Risk
Bernard-Soulier Syndrome, Type C	GP9	AR	Reduced Risk
Beta-Globin-Related Hemoglobinopathies	HBB	AR	Reduced Risk
Beta-Ketothiolase Deficiency	ACAT1	AR	Reduced Risk
Bilateral Frontoparietal Polymicrogyria	GPR56	AR	Reduced Risk
Biotinidase Deficiency	BTD	AR	Reduced Risk
Bloom Syndrome	BLM	AR	Reduced Risk
Canavan Disease	ASPA	AR	Reduced Risk
Carbamoylphosphate Synthetase I Deficiency	CPS1	AR	Reduced Risk
Carnitine Palmitoyltransferase IA Deficiency	CPT1A	AR	Reduced Risk
Carnitine Palmitoyltransferase II Deficiency	CPT2	AR	Reduced Risk
Carpenter Syndrome	RAB23	AR	Reduced Risk
Cartilage-Hair Hypoplasia	RMRP	AR	Reduced Risk
Cerebral Creatine Deficiency Syndrome 1	SLC6A8	XL	Reduced Risk
Cerebral Creatine Deficiency Syndrome 2	GAMT	AR	Reduced Risk
Cerebrotendinous Xanthomatosis	CYP27A1	AR	Reduced Risk
Charcot-Marie-Tooth Disease, Type 4D	NDRG1	AR	Reduced Risk
Charcot-Marie-Tooth Disease, Type 5 / Arts Syndrome	PRPS1	XL	Reduced Risk
Charcot-Marie-Tooth Disease, X-Linked	GJB1	XL	Reduced Risk
Choreoacanthocytosis	VPS13A	AR	Reduced Risk
Choroideremia	СНМ	XL	Reduced Risk
Chronic Granulomatous Disease (CYBA-Related)	CYBA	AR	Reduced Risk
Chronic Granulomatous Disease (CYBB-Related)	CYBB	XL	Reduced Risk
Citrin Deficiency	SLC25A13	AR	Reduced Risk





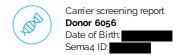
Citrullinemia, Type 1	ASS1	AR	Reduced Risk	
Cohen Syndrome	VPS13B	AR	Reduced Risk	
Combined Malonic and Methylmalonic Aciduria	ACSF3	AR	Reduced Risk	
Combined Oxidative Phosphorylation Deficiency 1	GFM1	AR	Reduced Risk	
Combined Oxidative Phosphorylation Deficiency 3	TSFM	AR	Reduced Risk	
Combined Pituitary Hormone Deficiency 2	PROP1	AR	Reduced Risk	
Combined Pituitary Hormone Deficiency 3	LHX3	AR	Reduced Risk	
Combined SAP Deficiency	PSAP	AR	Reduced Risk	
Congenital Adrenal Hyperplasia due to 17-Alpha- Hydroxylase Deficiency	CYP17A1	AR	Reduced Risk	
Congenital Adrenal Hyperplasia due to 21-Hydroxylase Deficiency	CYP21A2	AR	Reduced Risk	CYP21A2 copy number: 2 CYP21A2 sequencing: Negative
Congenital Amegakaryocytic Thrombocytopenia	MPL	AR	Reduced Risk	
Congenital Disorder of Glycosylation, Type la	PMM2	AR	Reduced Risk	
Congenital Disorder of Glycosylation, Type Ib	MPI	AR	Reduced Risk	
Congenital Disorder of Glycosylation, Type Ic	ALG6	AR	Reduced Risk	
Congenital Insensitivity to Pain with Anhidrosis	NTRK1	AR	Reduced Risk	
Congenital Myasthenic Syndrome (CHRNE-Related)	CHRNE	AR	Reduced Risk	
Congenital Myasthenic Syndrome (RAPSN-Related)	RAPSN	AR	Reduced Risk	
Congenital Neutropenia (<i>HAX1</i> -Related)	HAX1	AR	Reduced Risk	
Congenital Neutropenia (VPS45-Related)	VPS45	AR	Reduced Risk	
Corneal Dystrophy and Perceptive Deafness	SLC4A11	AR	Reduced Risk	
Corticosterone Methyloxidase Deficiency	CYP11B2	AR	Reduced Risk	
Cystic Fibrosis	CFTR	AR	Reduced Risk	
Cystinosis	CTNS	AR	Reduced Risk	
D-Bifunctional Protein Deficiency	HSD17B4	AR	Reduced Risk	
Deafness, Autosomal Recessive 77	LOXHD1	AR	Reduced Risk	
Duchenne Muscular Dystrophy / Becker Muscular Dystrophy	DMD	XL	Reduced Risk	
Dyskeratosis Congenita (RTEL1-Related)	RTEL1	AR	Reduced Risk	
Dystrophic Epidermolysis Bullosa	COL7A1	AR	Reduced Risk	
Ehlers-Danlos Syndrome, Type VIIC	ADAMTS2	AR	Reduced Risk	
Ellis-van Creveld Syndrome (EVC-Related)	EVC	AR	Reduced Risk	
Emery-Dreifuss Myopathy 1	EMD	XL	Reduced Risk	
Enhanced S-Cone Syndrome	NR2E3	AR	Reduced Risk	
Ethylmalonic Encephalopathy	ETHE1	AR	Reduced Risk	
Fabry Disease	GLA	XL	Reduced Risk	





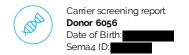
Factor IX Deficiency	F9	XL	Reduced Risk	
Factor XI Deficiency	F11	AR	Reduced Risk	
Familial Autosomal Recessive Hypercholesterolemia	LDLRAP1	AR	Reduced Risk	
Familial Dysautonomia	IKBKAP	AR	Reduced Risk	
Familial Hypercholesterolemia	LDLR	AR	Reduced Risk	
Familial Hyperinsulinism (ABCC8-Related)	ABCC8	AR	Reduced Risk	
Familial Hyperinsulinism (KCNJ:1-Related)	KCNJ11	AR	Reduced Risk	
Familial Mediterranean Fever	MEFV	AR	Reduced Risk	
Fanconi Anemia, Group A	FANCA	AR	Reduced Risk	
Fanconi Anemia, Group C	FANCC	AR	Reduced Risk	
Fanconi Anemia, Group G	FANCG	AR	Reduced Risk	
Fragile X Syndrome	FMR1	XL	Reduced Risk	FMR1 CGG repeat sizes: Not Performed FMR1 Sequencing: Negative Fragile X CGG triplet repeat expansion testing was not performed at this time, as the patient has either been previously tested or is a male.
Furnarase Deficiency	FH	AR	Reduced Risk	
GRACILE Syndrome and Other BCS1L-Related Disorders	BCS1L	AR	Reduced Risk	
Galactokinase Deficiency	GALK1	AR	Reduced Risk	
Galactosemia	GALT	AR	Reduced Risk	
Gaucher Disease	GBA	AR	Reduced Risk	
Gitelman Syndrome	SLC12A3	AR	Reduced Risk	
Glutaric Acidemia, Type I	GCDH	AR	Reduced Risk	
Glutaric Acidemia, Type IIa	ETFA	AR	Reduced Risk	
Glutaric Acidemia, Type IIc	ETFDH	AR	Reduced Risk	
Glycine Encephalopathy (AMT-Related)	AMT	AR	Reduced Risk	
Glycine Encephalopathy (GLDC-Related)	GLDC	AR	Reduced Risk	
Glycogen Storage Disease, Type II	GAA	AR	Reduced Risk	
Glycogen Storage Disease, Type III	AGL	AR	Reduced Risk	
Glycogen Storage Disease, Type IV / Adult Polyglucosan Body Disease	GBE1	AR	Reduced Risk	
Glycogen Storage Disease, Type la	G6PC	AR	Reduced Risk	
Glycogen Storage Disease, Type Ib	SLC37A4	AR	Reduced Risk	
Glycogen Storage Disease, Type V	PYGM	AR	Reduced Risk	
Glycogen Storage Disease, Type VII	PFKM	AR	Reduced Risk	
HMG-CoA Lyase Deficiency	HMGCL	AR	Reduced Risk	
Hemochromatosis, Type 2A	HFE2	AR	Reduced Risk	
Hemochromatosis, Type 3	TFR2	AR	Reduced Risk	





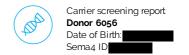
Hereditary Fructose Intolerance	ALDOB	AR	Reduced Risk
Hereditary Spastic Paraparesis 49	TECPR2	AR	Reduced Risk
Hermansky-Pudlak Syndrome, Type 1	HPS1	AR	Reduced Risk
Hermansky-Pudlak Syndrome, Type 3	HPS3	AR	Reduced Risk
Holocarboxylase Synthetase Deficiency	HLCS	AR	Reduced Risk
Homocystinuria (CBS-Related)	CBS	AR	Reduced Risk
Homocystinuria due to MTHFR Deficiency	MTHFR	AR	Reduced Risk
Homocystinuria, cblEType	MTRR	AR	Reduced Risk
Hydrolethalus Syndrome	HYLS1	AR	Reduced Risk
Hyperomithinemia-Hyperammonemia-Homocitrullinuria Syndrome	SLC25A15	AR	Reduced Risk
Hypohidrotic Ectodermal Dysplasia 1	EDA	XL	Reduced Risk
Hypophosphatasia	ALPL	AR	Reduced Risk
Inclusion Body Myopathy 2	GNE	AR	Reduced Risk
Infantile Cerebral and Cerebellar Atrophy	MED17	AR	Reduced Risk
Isovaleric Acidemia	IVD	AR	Reduced Risk
Joubert Syndrome 2	TMEM216	AR	Reduced Risk
Joubert Syndrome 7 / Meckel Syndrome 5 / COACH Syndrome	RPGRIP1L	AR	Reduced Risk
Junctional Epidermolysis Bullosa (<i>LAMA3</i> -Related)	LAMA3	AR	Reduced Risk
Junctional Epidermolysis Bullosa (<i>LAMB3</i> -Related)	LAMB3	AR	Reduced Risk
Junctional Epidermolysis Bullosa (<i>LAMC2</i> -Related)	LAMC2	AR	Reduced Risk
Krabbe Disease	GALC	AR	Reduced Risk
Lamellar Ichthyosis, Type 1	TGM1	AR	Reduced Risk
Leber Congenital Amaurosis 10 and Other CEP290- Related Ciliopathies	CEP290	AR	Reduced Risk
Leber Congenital Amaurosis 13	RDH12	AR	Reduced Risk
Leber Congenital Amaurosis 2 / Retinitis Pigmentosa 20	RPE65	AR	Reduced Risk
Leber Congenital Amaurosis 5	LCA5	AR	Reduced Risk
Leber Congenital Amaurosis 8 / Retinitis Pigmentosa 12 / Pigmented Paravenous Chorioretinal Atrophy	CRB1	AR	Reduced Risk
Leigh Syndrome, French-Canadian Type	LRPPRC	AR	Reduced Risk
Lethal Congenital Contracture Syndrome 1 / Lethal Arthrogryposis with Anterior Horn Cell Disease	GLE1	AR	Reduced Risk
Leukoencephalopathy with Vanishing White Matter	EIF2B5	AR	Reduced Risk
Limb-Girdle Muscular Dystrophy, Type 2A	CAPN3	AR	Reduced Risk
Limb-Girdle Muscular Dystrophy, Type 2B	DYSF	AR	Reduced Risk
Limb-Girdle Muscular Dystrophy, Type 2C	SGCG	AR	Reduced Risk





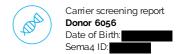
Limb-Girdle Muscular Dystrophy, Type 2D	SGCA	AR	Reduced Risk
Limb-Girdle Muscular Dystrophy, Type 2E	SGCB	AR	Reduced Risk
Limb-Girdle Muscular Dystrophy, Type 2I	FKRP	AR	Reduced Risk
Lipoamide Dehydrogenase Deficiency	DLD	AR	Reduced Risk
Lipoid Adrenal Hyperplasia	STAR	AR	Reduced Risk
Lipoprotein Lipase Deficiency	LPL	AR	Reduced Risk
Long-Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency	HADHA	AR	Reduced Risk
Lysinuric Protein Intolerance	SLC7A7	AR	Reduced Risk
Maple Syrup Urine Disease, Type 1a	BCKDHA	AR	Reduced Risk
Maple Syrup Urine Disease, Type 1b	BCKDHB	AR	Reduced Risk
Meckel 1 / Bardet-Biedl Syndrome 13	MKS1	AR	Reduced Risk
Medium Chain Acyl-CoA Dehydrogenase Deficiency	ACADM	AR	Reduced Risk
Megalencephalic Leukoencephalopathy with Subcortical Cysts	MLC1	AR	Reduced Risk
Menkes Disease	ATP7A	XL	Reduced Risk
Metachromatic Leukodystrophy	ARSA	AR	Reduced Risk
Methylmalonic Acidemia (<i>MMAA</i> -Related)	MMAA	AR	Reduced Risk
Methylmalonic Acidemia (<i>MMAB</i> -Related)	MMAB	AR	Reduced Risk
Methylmalonic Acidemia (<i>MUT</i> -Related)	MUT	AR	Reduced Risk
Methylmalonic Aciduria and Homocystinuria, Cobalamin C Type	ММАСНС	AR	Reduced Risk
Methylmalonic Aciduria and Homocystinuria, Cobalamin D Type	MMADHC	AR	Reduced Risk
Microphthalmia / Anophthalmia	VSX2	AR	Reduced Risk
Mitochondrial Complex I Deficiency (ACADg-Related)	ACAD9	AR	Reduced Risk
Mitochondrial Complex I Deficiency (NDUFAF5-Related)	NDUFAF5	AR	Reduced Risk
Mitochondrial Complex I Deficiency (NDUFS6-Related)	NDUFS6	AR	Reduced Risk
Mitochondrial DNA Depletion Syndrome 6 / Navajo Neurohepatopathy	MPV17	AR	Reduced Risk
Mitochondrial Myopathy and Sideroblastic Anemia 1	PUS1	AR	Reduced Risk
Mucolipidosis II / IIIA	GNPTAB	AR	Reduced Risk
Mucolipidosis III Gamma	GNPTG	AR	Reduced Risk
Mucolipidosis IV	MCOLN1	AR	Reduced Risk
Mucopolysaccharidosis Type I	IDUA	AR	Reduced Risk
Mucopolysaccharidosis Type II	IDS	XL	Reduced Risk
Mucopolysaccharidosis Type IIIA	SGSH	AR	Reduced Risk
Mucopolysaccharidosis Type IIIB	NAGLU	AR	Reduced Risk





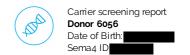
Mucopolysaccharidosis Type IIIC	HGSNAT	AR	Reduced Risk
Mucopolysaccharidosis Type IIID	GNS	AR	Reduced Risk
Mucopolysaccharidosis Type IVb / GM1 Gangliosidosis	GLB1	AR	Reduced Risk
Mucopolysaccharidosis type IX	HYAL1	AR	Reduced Risk
Mucopolysaccharidosis type VI	ARSB	AR	Reduced Risk
Multiple Sulfatase Deficiency	SUMF1	AR	Reduced Risk
Muscle-Eye-Brain Disease and Other <i>POMGNT1</i> -Related Congenital Muscular Dystrophy-Dystroglycanopathies	POMGNT1	AR	Reduced Risk
Myoneurogastrointestinal Encephalopathy	TYMP	AR	Reduced Risk
Myotubular Myopathy 1	MTM1	XL	Reduced Risk
N-Acetylglutamate Synthase Deficiency	NAGS	AR	Reduced Risk
Nemaline Myopathy 2	NEB	AR	Reduced Risk
Nephrogenic Diabetes Insipidus, Type II	AQP2	AR	Reduced Risk
Nephrotic Syndrome (<i>NPHS1</i> -Related) / Congenital Finnish Nephrosis	NPHS1	AR	Reduced Risk
Nephrotic Syndrome (<i>NPHS2</i> -Related) / Steroid- Resistant Nephrotic Syndrome	NPHS2	AR	Reduced Risk
Neuronal Ceroid-Lipofuscinosis (CLN3-Related)	CLN3	AR	Reduced Risk
Neuronal Ceroid-Lipofuscinosis (<i>CLN5</i> -Related)	CLN5	AR	Reduced Risk
Neuronal Ceroid-Lipofuscinosis (CLN6-Related)	CLN6	AR	Reduced Risk
Neuronal Ceroid-Lipofuscinosis (CLN8-Related)	CLN8	AR	Reduced Risk
Neuronal Ceroid-Lipofuscinosis (MFSD8-Related)	MFSD8	AR	Reduced Risk
Neuronal Ceroid-Lipofuscinosis (PPT1-Related)	PPT1	AR	Reduced Risk
Neuronal Ceroid-Lipofuscinosis (TPP1-Related)	TPP1	AR	Reduced Risk
Niemann-Pick Disease (SMPD1-Related)	SMPD1	AR	Reduced Risk
Niemann-Pick Disease, Type C (NPC1-Related)	NPC1	AR	Reduced Risk
Niemann-Pick Disease, Type C (NPC2-Related)	NPC2	AR	Reduced Risk
Nijmegen Breakage Syndrome	NBN	AR	Reduced Risk
Non-Syndromic Hearing Loss (<i>GJB2</i> -Related)	GJB2	AR	Reduced Risk
Odonto-Onycho-Dermal Dysplasia / Schopf-Schulz- Passarge Syndrome	WNT10A	AR	Reduced Risk
Omenn Syndrome (RAG2-Related)	RAG2	AR	Reduced Risk
Omenn Syndrome / Severe Combined Immunodeficiency, Athabaskan-Type	DCLRE1C	AR	Reduced Risk
Ornithine Aminotransferase Deficiency	OAT	AR	Reduced Risk
Ornithine Transcarbomylase Deficiency	ОТС	XL	Reduced Risk
Osteopetrosis 1	TCIRG1	AR	Reduced Risk
Pendred Syndrome	SLC26A4	AR	Reduced Risk





Primy position Primy production Primy Primy production Primy Primy production Primy Primy production Primy Primy Primy production Primy Primy production Primy Primy Primy production Primy				
Polyglandular Alzoimmune Syndrome Type 1 ARE AR Reduced Risk Portocerebellar Hypoplasia, Type 5 Portocerebellar Hypoplasia, Type 6 RARSS AR Reduced Risk Portocerebellar Hypoplasia, Type 6 RARSS AR Reduced Risk Primary Clillary Dysidnesia (DMANE-Related) Primary Hypoprosialuria, Type 1 AGRT AR Reduced Risk Primary Hypoprosialuria, Type 2 GRIPER AR Reduced Risk Primary Hypoprosialuria, Type 3 HOGAL AR Reduced Risk Primary Hypoprosialuria, Type 3 HOGAL AR Reduced Risk Progressive Corrobald-Corrobal Alzophy SEPSECS AR Reduced Risk Progressive Rimitial Intrinsipasaic Cholestania, Type 2 ASCRII AR Reduced Risk Proportion Ackiderina (PCCA-Related) PCCB Related) PCCB Related Pyrurate Dehydrogenase B-Alpha Deficiency PDHAL AR Reduced Risk Pyrurate Dehydrogenase B-Bata Deficiency PDHB AR Reduced Risk Related Risk R	Phenylalanine Hydroxylase Deficiency	PAH	AR	Reduced Risk
Portocere-bellar Hypoplasia, Type sA ViNI Portocere-bellar Hypoplasia, Type 6 PARIS2 AR Reduced Risk Primary Clainy Dyskrinosia (DNAM-Related) Primary Hypoprocalatia, Type 2 GR-HPR AR Reduced Risk Primary Hypoprocalatia, Type 3 Primary Hypoprocalatia, Type 3 Primary Hypoprocalatia, Type 3 Primary Hypoprocalatia, Type 3 Programsive Cerebral Arcophy SEPSECS AR Reduced Risk Propositive Cerebral Arcophy SEPSECS AR Reduced Risk Propositive Acidemia (PCCA-Related) Programsive Firmitial Intrahepatic Orbidatesis, Type 2 ASCRII Propositive Acidemia (PCCA-Related) Propositive Delhydrogeneose (B-Alpha Deficiency) PDHAI XI. Reduced Risk Protocod Risk Protocod Risk Protocod Risk Protocod Risk Related	Polycystic Kidney Disease, Autosomal Recessive	PKHD1	AR	Reduced Risk
Primary Carnitire Deficiency SLC2245 AR Reduced Risk Primary Cillary Dysthresia (DNAH5-Reisted) DNAH5 AR Reduced Risk Primary Hypercostuluta (Dybe 2 GR-SPR AR Reduced Risk Primary Hypercostuluta (Tybe 2 ARCSEL AR Reduced Risk Progressive Corebial Artrophy SEPSECS AR Reduced Risk Progressive Familial Intrahspatic Cholestasis, Type 2 ARCSEL AR Reduced Risk Prophoric Acidemia (PCCA-Reistad) PCCA AR Reduced Risk Prophoric Acidemia (PCCA-Reistad) PCCA AR Reduced Risk Prophoric Acidemia (PCCA-Reistad) P	Polyglandular Autoimmune Syndrome, Type 1	AIRE	AR	Reduced Risk
Primary Caratine Deficiency Primary Ciliary Dyskinesia (DNANS-Related) Primary Ciliary Dyskinesia (DNANS-Related) Primary Ciliary Dyskinesia (DNANS-Related) Primary Hyperrovaluria, Type 1 Primary Hyperrovaluria, Type 2 Primary Hyperrovaluria, Type 3 Progressive Cerebolio-Corebral Arrophy SEPSECS AR Reduced Risk Progressive Familial Intrahepatic Chrisestasis, Type 2 ARCS21 AR Reduced Risk Proportion Acidemia (PCOA-Related) Proportion Aci	Pontocerebellar Hypoplasia, Type 1A	VRK1	AR	Reduced Risk
Primary Ciliany Dyskinesia (DNA4-Related) Primary Ciliany Dyskinesia (DNA4-Related) Primary Ciliany Dyskinesia (DNA4-Related) Primary Ciliany Dyskinesia (DNA4-Related) Primary Hyperoxaturia, Type 1 AGXT AR Reduced Risk Primary Hyperoxaturia, Type 2 GRHPR AR Reduced Risk Primary Hyperoxaturia, Type 2 GRHPR AR Reduced Risk Progressive Cerebratic Condental Arrophy SEPSECS AR Reduced Risk Progressive Cerebratic Cholestasis, Type 2 ABCE21 AR Reduced Risk Propincia Addemia (PCCA-Related) PCCA AR Reduced Risk Propincia Addemia (PCCA-Related) PCCA AR Reduced Risk Pyrunate Dehydrogenase Ex-Alpha Deficiency PDHA1 XL Reduced Risk Pyrunate Dehydrogenase Ex-Baba Deficiency PDHA1 XL Reduced Risk Reduced Risk Resultitis Pigmentosa 25 EYS AR Reduced Risk Resultitis Pigmentosa 25 Resultitis Pigmentosa 25 Resultitis Pigmentosa 28 Resultitis Pigmentosa 28 Resultitis Pigmentosa 28 Resultitis Pigmentosa 29 Resultitis Pigmentosa 29 Resultitis Pigmentosa 30 Resultitis Pigmentosa 30 Resultitis Pigmentosa 40 Resultitis Pigmentosa 40 Resultitis Pigmentosa 50 Resu	Pontocerebellar Hypoplasia, Type 6	RARS2	AR	Reduced Risk
Primary Ciliary Dysidinesia (DNAta-Related) Primary Ciliary Dysidinesia (DNAta-Related) Primary Hyperoxaluria, Type 1 AGXT AR Reduced Risk Primary Hyperoxaluria, Type 2 CRAPR AR Reduced Risk Primary Hyperoxaluria, Type 3 HOGAL AR Reduced Risk Progressive Cerebello-Corebral Atrophy SEPSECS AR Reduced Risk Progressive Cerebello-Corebral Atrophy SEPSECS AR Reduced Risk Progressive Remillal intrahapatic Cholestass, Type 2 ABCRIL Propointe Acidemia (PCCA-Related) PCCA AR Reduced Risk Propointe Acidemia (PCCA-Related) PCCA AR Reduced Risk Pythylate Dehydrogenesse Ex-Alpha Deficiency PDHAL XI. Reduced Risk Pythylate Dehydrogenesse Ex-Beta Deficiency PDHAL XI. Reduced Risk Renal Tubular Acidesia and Deefness ATROVER Renal Tubular Acidesia and Deefness ATROVER Resimits Pigmentosa 25 Resimits Pigmentosa 26 Resimits Pigmentosa 28 Resimits Pigmentosa 29 DHODS AR Reduced Risk Sala Disease SLC37A6 AR Reduced Risk Sepawa Syndrome AR Reduced Risk Sepawa Syndrome ALDH3A2 AR Reduced Risk SMATCopy number > 3 c'19807-CQ Number > 3 c'1980	Primary Carnitine Deficiency	SLC22A5	AR	Reduced Risk
Primary Pyterroxaluria, Type 1 AGXT AR Reduced Risk Primary Hyperoxaluria, Type 2 GRHFR AR Reduced Risk Primary Hyperoxaluria, Type 3 HOGA1 AR Reduced Risk Primary Hyperoxaluria, Type 3 HOGA1 AR Reduced Risk Progressive Cerobello-Corebral Atrophy SEPSECS AR Reduced Risk Progressive Cerobello-Corebral Atrophy SEPSECS AR Reduced Risk Progressive Rimillal Intrahepatic Cholestadis, Type 2 ABCB11 AR Reduced Risk Propolonic Addismia (PCCA-Related) PCCA AR Reduced Risk Propolonic Addismia (PCCA-Related) PCCB AR Reduced Risk Propolonic Addismia (PCCA-Related) PCCB AR Reduced Risk Pythytopyteroses Et-Alpha Deficiency PDHA1 XL Reduced Risk Pythytopyteroses Et-Beta Deficiency PDHB AR Reduced Risk Pythytopyteroses Et-Beta Deficiency PDHB AR Reduced Risk Salla Disease SICTA6 AR Reduced Risk Salla Disease SICTA6 AR Reduced Risk Salla Disease SICTA6 AR Reduced Risk Solla Disease SICTA6 AR Reduced Risk Solla Disease ALDH3A2 AR Reduced Risk Solla Disease ALDH3A2 AR Reduced Risk Solla Disease SICTA6 AR Reduced Risk Solla Disease ALDH3A2 AR Reduced	Primary Ciliary Dyskinesia (<i>DNAH5</i> -Related)	DNAH5	AR	Reduced Risk
Primary Hyperoxaluria, Type 1 AGXT AR Reduced Risk Primary Hyperoxaluria, Type 2 GRHPR AR Reduced Risk Primary Hyperoxaluria, Type 3 HOGAL AR Reduced Risk Progressive Cerebello-Cerebral Atrophy SEPSECS AR Reduced Risk Progressive Familial Intrehepatic Cholestasis, Type 2 ABCBII AR Reduced Risk Proplenic Acidemia (PCCA-Related) PCCA AR Reduced Risk Proplenic Acidemia (PCCA-Related) PCCB AR Reduced Risk Pymodysostosis CTSK AR Reduced Risk Pymodysostosis CTSK AR Reduced Risk Pymodysostosis CTSK AR Reduced Risk Pymodysostosis AR Reduced Risk Pymodysostosis AR Reduced Risk Pymodysostosis AR Reduced Risk Reduced Risk Result Tubular Acidosis and Desfinercy PDHB AR Reduced Risk Result Tubular Acidosis and Desfines AR Reduced Risk Result Tubular Reduced Risk Result Tubular Result Risk Reduced Risk Result Tubular Risk Reduced Risk Reduced Risk Reduced Risk Reduced Risk Shall Disease SLCLTAS AR Reduced Risk Sandhord Disease AL DHJ AR Reduced Risk Segared Syndrome AL DHJ AR Reduced Risk Shall Copy number: 2 Spinal Muscular Atrophy AR Reduced Risk Shall Copy number: 2 Spinal Muscular Atrophy Shall AR Reduced Risk Shall Copy number: 3 C'3-80 Dis Rogative	Primary Ciliary Dyskinesia (DNAlz-Related)	DNAl1	AR	Reduced Risk
Primary Hyperoxaluria. Type 2 GRHPR AR Reduced Risk Primary Hyperoxaluria. Type 3 HOGA1 AR Reduced Risk Progressive Cerebello-Cerebral Atrophy SEPSECS AR Reduced Risk Progressive Familial Intrahepatic Cholestasis. Type 2 ABC811 AR Reduced Risk Proplinic Acidemia (PCCA-Related) PCCA AR Reduced Risk Proplinic Acidemia (PCCA-Related) PCCB AR Reduced Risk Pymodysostasis CTSK AR Reduced Risk Pymodysostasis CTSK AR Reduced Risk Pymodysostasis Pymodysostasis CTSK AR Reduced Risk Pymodysostasis Pymodysostasis AR Reduced Risk Reduced Risk Reduced Risk Reduced Risk Reduced Risk Real Tubular Acidosis and Deafness ATRBVIBB AR Reduced Risk Sanhor Disease BLOTAG AR Reduced Risk Sequenced	Primary Ciliary Dyskinesia (DNAI2-Related)	DNAI2	AR	Reduced Risk
Primary Hyperoxaluria, Type 3 Progressive Carebello-Cerebral Atrophy SEPSECS AR Reduced Risk Progressive Familial Intrahepatic Cholestasis, Type 2 ABC811 AR Reduced Risk Propinic Acidemia (PCCA-Related) PCCA AR Reduced Risk Propinic Acidemia (PCCA-Related) PCCB AR Reduced Risk Pyroxidysostosis CTSK AR Reduced Risk Pyroxidysostosis CTSK AR Reduced Risk Pyroxidysostosis CTSK AR Reduced Risk Pyroxide Dehydrogenase B-Alpha Deficiency PDHA1 XL Reduced Risk Pyroxide Dehydrogenase B-Beta Deficiency PDHB AR Reduced Risk Reduced Risk Restricts Pigmentosa 25 EYS AR Reduced Risk Restricts Pigmentosa 26 Retribits Pigmentosa 28 FMM16A Restricts Pigmentosa 28 Retribits Pigmentosa 28 Retribits Pigmentosa 38 Retribits Pigmentosa 49 Reduced Risk Reduced Risk Retribits Pigmentosa 59 AR Reduced Risk Reduced Risk Reduced Risk Sibinities Pigmentosa 59 AR Reduced Risk Reduced Risk Sandhoff Disease SLC17A5 AR Reduced Risk Sibinities Immunoosseus Dysplasia SMARCALL AR Reduced Risk Signer-Lansson Syndrome TH AR Reduced Risk Signer-Lansson Syndrome ALDH3A2 AR Reduced Risk SMARCALL Spinal Muscular Atrophy AR Reduced Risk SMARCALL Spinal Muscular Atrophy AR Reduced Risk SMARCALL Spinal Muscular Atrophy AR Reduced Risk SMARCALL AR Reduced Risk SMARCALL Spinal Muscular Atrophy AR Reduced Risk SMARCALL Spinal Muscular Atrophy	Primary Hyperoxaluria, Type 1	AGXT	AR	Reduced Risk
Progressive Cerebello-Cerebral Atrophy Progressive Familial Intrahepatic Cholestasis Type 2 ABCB11 AR Reduced Risk Propionic Acidemia (PCCA-Related) PCCA AR Reduced Risk Propionic Acidemia (PCCA-Related) PCCB AR Reduced Risk Propionic Acidemia (PCCA-Related) PCCB AR Reduced Risk Pycnodysostosis CTSK AR Reduced Risk Pyruvate Dehydrogenase Et-Alpha Deficiency PDH/II XL Reduced Risk Pyruvate Dehydrogenase Et-Beta Deficiency PDH/B AR Reduced Risk Reduced Risk Restricts Pigmentosa 25 EYS AR Reduced Risk Restricts Pigmentosa 28 Rethibts Pigmentosa 28 Rethibts Pigmentosa 28 Rethibts Pigmentosa 39 DHDDS AR Reduced Risk Restricts Pigmentosa 59 Rethibts Pigmento	Primary Hyperoxaluria, Type 2	GRHPR	AR	Reduced Risk
Progressive Familial Intrahepatic Cholestasis, Type 2 ABCB11 AR Reduced Risk Propionic Acidemia (PCCA-Related) PCCB AR Reduced Risk Propionic Acidemia (PCCB-Related) PCCB AR Reduced Risk Pythodysostosis CTSK AR Reduced Risk Pythodysostosis Pythate Dehydrogenese Et-Alpha Deficiency PDHA1 XL Reduced Risk Pythate Dehydrogenese Et-Beta Deficiency PDHB AR Reduced Risk Reduced Risk Readuced Risk Readuced Risk Restribitis Pigmentosa 25 EVS AR Reduced Risk Reduced Risk Restribitis Pigmentosa 25 Restribitis Pigmentosa 26 Restribitis Pigmentosa 28 Restribitis Pigmentosa 28 Restribitis Pigmentosa 39 DHDDS AR Reduced Risk Reduced Risk Restribitis Pigmentosa 39 DHDDS AR Reduced Risk Reduced Risk Restribitis Pigmentosa 39 Restribitis Pigmentosa 39 AR Reduced Risk Reduced Risk AR Reduced Risk Reduced Risk Reduced Risk Reduced Risk Reduced Risk Salla Disease SLC17A5 AR Reduced Risk Salla Disease SLC17A5 AR Reduced Risk Segawa Syndrome TH AR Reduced Risk Segawa Syndrome ALDH3A2 AR Reduced Risk SMMz copy number 2 SMMz copy number 2 SMNz copy number 3-3 C'3-807-0 Regative	Primary Hyperoxaluria, Type 3	HOGA1	AR	Reduced Risk
Proplonic Acidemia (PCCA-Related) Proplonic Acidemia (PCCB-Related) Proplonic Acidemia (PCCB-Related) Proplonic Acidemia (PCCB-Related) Pycnodysostosis CTSK AR Reduced Risk Pyunate Dehydrogenase Ex-Alpha Deficiency PDHA1 XL Reduced Risk Pyunate Dehydrogenase Ex-Beta Deficiency PDHB AR Reduced Risk Reduced Risk Reduced Risk Rethitis Pigmentosa 25 Rethitis Pigmentosa 26 Rethitis Pigmentosa 28 Rethitis Pigmentosa 28 Rethitis Pigmentosa 28 Rethitis Pigmentosa 29 Rethitis Pigmentosa 39 Reduced Risk Rethitis Pigmentosa 39 Reduced Risk Salla Disease SLC17A6 AR Reduced Risk Salla Disease SLC17A6 AR Reduced Risk Segawa Syndrome TH AR Reduced Risk Segawa Syndrome ALDH3A2 AR Reduced Risk SMNt copy number: 2 SMNt copy numb	Progressive Cerebello-Cerebral Atrophy	SEPSECS	AR	Reduced Risk
Propionic Acidemia (PCCB-Related) Pycnodysostosis CTSK AR Reduced Risk Pytrodysostosis CTSK AR Reduced Risk Pytrodysostosis Reduced Risk Roberts Syndrome ESCO2 AR Reduced Risk Sala Disease SLC17/6 AR Reduced Risk Sandhoff Disease BLC17/6 AR Reduced Risk Segawa Syndrome TH AR Reduced Risk Segawa Syndrome ALDH3A2 AR Reduced Risk Smitt-Lemif-Opitz Syndrome DHCR7 AR Reduced Risk SMN12 copy number: >-3 C:3+60T>Q Nogative	Progressive Familial Intrahepatic Cholestasis, Type 2	ABCB11	AR	Reduced Risk
Pycnodysostosis CTSK AR Reduced Risk Pyruvate Dehydrogenase Et-Alpha Deficiency PDHA1 XL Reduced Risk Pyruvate Dehydrogenase Et-Beta Deficiency PDHB AR Reduced Risk Renal Tubular Acidosis and Deafness ATP6VIB1 AR Reduced Risk Retinitis Pigmentosa 25 EVS AR Reduced Risk Retinitis Pigmentosa 26 CERKL AR Reduced Risk Retinitis Pigmentosa 28 FAM161A AR Reduced Risk Retinitis Pigmentosa 29 DHDDS AR Reduced Risk Retinitis Pigmentosa 59 DHDDS AR Reduced Risk Retinitis Pigmentosa 59 DHDDS AR Reduced Risk Retinitis Pigmentosa 59 Rhizometic Chondrodysplasia Punctata, Type 1 PEX7 AR Reduced Risk Reduced Risk Roberts Syndrome ESCO2 AR Reduced Risk Salla Disease SLC17A6 AR Reduced Risk Salla Disease BLC37A6 AR Reduced Risk Schimke Immunocsseous Dysplasia SMARCAL1 AR Reduced Risk Segawa Syndrome TH AR Reduced Risk Segawa Syndrome DHCR7 AR Reduced Risk SmNtz copy number: 2 SMNtz copy number: 2 SMNtz copy number: >-3 C:3+80T>Q Rogative	Propionic Acidemia (<i>PCCA</i> -Related)	PCCA	AR	Reduced Risk
Pyruvate Dehydrogenase Et-Alpha Deficiency PDHA1 XL Reduced Risk Pyruvate Dehydrogenase Et-Beta Deficiency PDHB AR Reduced Risk Renal Tubular Acidosis and Deafness ATP6VIBI AR Reduced Risk Retinitis Pigmentosa 25 EYS AR Reduced Risk Retinitis Pigmentosa 26 CERKL AR Reduced Risk Retinitis Pigmentosa 28 Retinitis Pigmentosa 28 Retinitis Pigmentosa 59 DHDDS AR Reduced Risk Retinitis Pigmentosa 59 Rhizomelic Chondrodysplasia Punctata, Type 1 PEX7 AR Reduced Risk Rhizomelic Chondrodysplasia Punctata, Type 3 AGPS AR Reduced Risk Roberts Syndrome ESCO2 AR Reduced Risk Salla Disease SLC17A5 AR Reduced Risk Sandhoff Disease HEXB AR Reduced Risk Schirrike Immunoosseous Dysplasia SMARCALLI AR Reduced Risk Segawa Syndrome TH AR Reduced Risk Segawa Syndrome ALDH3A2 AR Reduced Risk Smitt - Lemii-Opitz Syndrome DHCR7 AR Reduced Risk SMIt copy number: 2 SMIt copy number: 2 SMINt copy number: 2	Propionic Acidemia (<i>PCCB</i> -Related)	PCCB	AR	Reduced Risk
Pyruvate Dehydrogenase Et-Beta Deficiency PDHB AR Reduced Risk Renal Tubular Acidosis and Deafness ATP6VIBI AR Reduced Risk Retinitis Pigmentosa 25 EVS AR Reduced Risk Retinitis Pigmentosa 26 CERKL AR Reduced Risk Retinitis Pigmentosa 28 FAMI61A AR Reduced Risk Retinitis Pigmentosa 59 DHDDS AR Reduced Risk Retinitis Pigmentosa 59 Rhizomelic Chondrodysplasia Punctata, Type 1 PEX7 AR Reduced Risk Rhizomelic Chondrodysplasia Punctata, Type 3 AGPS AR Reduced Risk Roberts Syndrome ESC02 AR Reduced Risk Salla Disease SLC17A5 AR Reduced Risk Sandhoff Disease HEXB AR Reduced Risk Sandhoff Disease Schimke Immunoosseous Dysplasia SMARCAL1 AR Reduced Risk Segawa Syndrome TH AR Reduced Risk Signgen-Larsson Syndrome ALDH3A2 AR Reduced Risk Smitt-Lemil-Opitz Syndrome DHCR7 AR Reduced Risk SMNI copy number > 3 SMNI copy number > 3 C '3+80T>G Negative	Pycnodysostosis	CTSK	AR	Reduced Risk
Renal Tubular Acidosis and Deafness ATP6V1B1 AR Reduced Risk Retinitis Pigmentosa 25 EYS AR Reduced Risk Retinitis Pigmentosa 26 CERKL AR Reduced Risk Retinitis Pigmentosa 28 FAM161A AR Reduced Risk Retinitis Pigmentosa 59 DHDDS AR Reduced Risk Retinitis Pigmentosa 59 DHDDS AR Reduced Risk Retinitis Pigmentosa 59 Rhizomelic Chondrodysplasia Punctata, Type 1 PEX7 AR Reduced Risk Roberts Syndrome ESCO2 AR Reduced Risk Salla Disease SLC17A5 AR Reduced Risk Sandhoff Disease HEXB AR Reduced Risk Schimke Immunoosseous Dysplasia SMARCAL1 AR Reduced Risk Schimke Immunoosseous Dysplasia SMARCAL1 AR Reduced Risk Segawa Syndrome ALDH3A2 AR Reduced Risk Smith-Lemii-Opitz Syndrome DHCR7 AR Reduced Risk SMN1 copy number: >-3 c*3*B0T>G Negative	Pyruvate Dehydrogenase E1-Alpha Deficiency	PDHA1	XL	Reduced Risk
Retinitis Pigmentosa 25 EYS AR Reduced Risk Retinitis Pigmentosa 26 CERKL AR Reduced Risk Retinitis Pigmentosa 28 FAM161A AR Reduced Risk Retinitis Pigmentosa 59 DHDDS AR Reduced Risk Rhizomelic Chondrodysplasia Punctata, Type 1 PEX7 AR Reduced Risk Rhizomelic Chondrodysplasia Punctata, Type 3 AGPS AR Reduced Risk Roberts Syndrome ESCO2 AR Reduced Risk Salla Disease SLC17A5 AR Reduced Risk Sandhoff Disease HEXB AR Reduced Risk Schimke Immunoosseous Dysplasia SMARCAL1 AR Reduced Risk Segawa Syndrome TH AR Reduced Risk Sogren-Larsson Syndrome ALDH3A2 AR Reduced Risk Smith-Lemil-Opitz Syndrome DHCR7 AR Reduced Risk Spinal Muscular Atrophy SMN1 AR Reduced Risk Spinal Muscular Atrophy SMN1 AR Reduced Risk	Pyruvate Dehydrogenase E1-Beta Deficiency	PDHB	AR	Reduced Risk
Retinitis Pigmentosa 26 CERKL AR Reduced Risk Retinitis Pigmentosa 28 FAM161A AR Reduced Risk Retinitis Pigmentosa 59 DHDDS AR Reduced Risk Rhizomelic Chondrodysplasia Punctata, Type 1 PEX7 AR Reduced Risk Rhizomelic Chondrodysplasia Punctata, Type 3 AGPS AR Reduced Risk Roberts Syndrome ESCO2 AR Reduced Risk Salla Disease SLC17A5 AR Reduced Risk Sandhoff Disease HEXB AR Reduced Risk Schimke Immunoosseous Dysplasia SMARCAL1 AR Reduced Risk Segawa Syndrome TH AR Reduced Risk Sjogren-Larsson Syndrome ALDH3A2 AR Reduced Risk Smith-Lemil-Opitz Syndrome DHCR7 AR Reduced Risk Spinal Muscular Atrophy SMN1 AR Reduced Risk Smith-Lemil-Opitz Syndrome AR Reduced Risk Spinal Muscular Atrophy AR Reduced Risk	Renal Tubular Acidosis and Deafness	ATP6V1B1	AR	Reduced Risk
Retinitis Pigmentosa 28 Retinitis Pigmentosa 59 DHDDS AR Reduced Risk Rhizomelic Chondrodysplasia Punctata, Type 1 PEX7 AR Reduced Risk Rhizomelic Chondrodysplasia Punctata, Type 3 AGPS AR Reduced Risk Roberts Syndrome ESC02 AR Reduced Risk Salla Disease SLC17A5 AR Reduced Risk Sandhoff Disease HEXB AR Reduced Risk Schimke Immunoosseous Dysplasia SMARCAL1 AR Reduced Risk Segawa Syndrome TH AR Reduced Risk Siggren-Larsson Syndrome ALDH3A2 AR Reduced Risk Smith-Lemil-Opitz Syndrome DHCR7 AR Reduced Risk SMNI Copy number: 2 SMNI Copy number: >-3 C:3+80T>G Negative	Retinitis Pigmentosa 25	EYS	AR	Reduced Risk
Retinitis Pigmentosa 59 DHDDS AR Reduced Risk Rhizomelic Chondrodysplasia Punctata, Type 1 PEX7 AR Reduced Risk Rhizomelic Chondrodysplasia Punctata, Type 3 AGPS AR Reduced Risk Roberts Syndrome ESCO2 AR Reduced Risk Salla Disease SLC17A5 AR Reduced Risk Sandhoff Disease HEXB AR Reduced Risk Schimke Immunoosseous Dysplasia SMARCAL1 AR Reduced Risk Segawa Syndrome TH AR Reduced Risk Sjogren-Larsson Syndrome ALDH3A2 AR Reduced Risk Smith-Lemli-Opitz Syndrome DHCR7 AR Reduced Risk SMN1 Copy number: 2 SMN2 copy number: >-3 c. '3+80T>Q Negative	Retinitis Pigmentosa 26	CERKL	AR	Reduced Risk
Rhizomelic Chondrodysplasia Punctata, Type 1 Rhizomelic Chondrodysplasia Punctata, Type 3 AGPS AR Reduced Risk Roberts Syndrome ESCO2 AR Reduced Risk Salla Disease SLC17A5 AR Reduced Risk Sandhoff Disease HEXB AR Reduced Risk Schimke Immunoosseous Dysplasia SMARCAL1 AR Reduced Risk Segawa Syndrome TH AR Reduced Risk Sigogren-Larsson Syndrome ALDH3A2 AR Reduced Risk Smith-Lemii-Opitz Syndrome DHCR7 AR Reduced Risk SMN1 Copy number: 2 SMN2 copy number: >-3 c. '3+80T>Q Negative	Retinitis Pigmentosa 28	FAM161A	AR	Reduced Risk
Rhizomelic Chondrodysplasia Punctata, Type 3 AGPS AR Reduced Risk Roberts Syndrome ESCO2 AR Reduced Risk Salla Disease SLC17A5 AR Reduced Risk Sandhoff Disease HEXB AR Reduced Risk Schimke Immunoosseous Dysplasia SMARCAL1 AR Reduced Risk Segawa Syndrome TH AR Reduced Risk Sjogren-Larsson Syndrome ALDH3A2 AR Reduced Risk Smith-Lemli-Opitz Syndrome DHCR7 AR Reduced Risk Smith-Lemli-Opitz Syndrome DHCR7 AR Reduced Risk SMN12 Sylvar Reduced Risk SMN12 copy number: 2 SMN2 copy number: >-3 c. '3+80T>Q Negative	Retinitis Pigmentosa 59	DHDDS	AR	Reduced Risk
Roberts Syndrome ESCO2 AR Reduced Risk Salla Disease SLC17A5 AR Reduced Risk Sandhoff Disease HEXB AR Reduced Risk Schimke Immunoosseous Dysplasia SMARCAL1 AR Reduced Risk Segawa Syndrome TH AR Reduced Risk Sjogren-Larsson Syndrome ALDH3A2 AR Reduced Risk Smith-Lemli-Opitz Syndrome DHCR7 AR Reduced Risk Spinal Muscular Atrophy SMN1 AR Reduced Risk Spinal Muscular Atrophy SMN1 AR Reduced Risk	Rhizomelic Chondrodysplasia Punctata, Type 1	PEX7	AR	Reduced Risk
Salla Disease SLC17A5 AR Reduced Risk Sandhoff Disease HEXB AR Reduced Risk Schimke Immunoosseous Dysplasia SMARCAL1 AR Reduced Risk Segawa Syndrome TH AR Reduced Risk Sjogren-Larsson Syndrome ALDH3A2 AR Reduced Risk Smith-Lemli-Opitz Syndrome DHCR7 AR Reduced Risk Spinal Muscular Atrophy SMN1 AR Reduced Risk Spinal Muscular Atrophy SMN1 AR Reduced Risk SmN2 copy number: >-3 c. '3+80T>Q Negative	Rhizomelic Chondrodysplasia Punctata, Type 3	AGPS	AR	Reduced Risk
Sandhoff Disease ##EXB ##EAUced Risk ##EXB ##EXB ##EXB ##EXB ##EXB ##EXB ##EXB ##EAUced Risk ##EXB ##EXB ##EXB ##EXB ##EAUced Risk ##EAU	Roberts Syndrome	ESCO2	AR	Reduced Risk
Schimke Immunoosseous Dysplasia SMARCAL1 AR Reduced Risk Segawa Syndrome TH AR Reduced Risk Sjogren-Larsson Syndrome ALDH3A2 AR Reduced Risk Smith-Lemli-Opitz Syndrome DHCR7 AR Reduced Risk Spinal Muscular Atrophy SMN1 AR Reduced Risk SMN1 copy number: 2 SMN2 copy number: >=3 c. '3+80T>G: Negative	Salla Disease	SLC17A5	AR	Reduced Risk
Segawa Syndrome TH AR Reduced Risk Sjogren-Larsson Syndrome ALDH3A2 AR Reduced Risk Smith-Lemli-Opitz Syndrome DHCR7 AR Reduced Risk Spinal Muscular Atrophy SMN1 AR Reduced Risk SMN1 copy number: 2 SMN2 copy number: >= 3 c. '3+80T>G: Negative	Sandhoff Disease	HEXB	AR	Reduced Risk
Sjogren-Larsson Syndrome ALDH3A2 AR Reduced Risk Smith-Lemli-Opitz Syndrome DHCR7 AR Reduced Risk Spinal Muscular Atrophy SMN1 AR Reduced Risk SMN1 copy number: 2 SMN2 copy number: >=3 c. '3+80T>G: Negative	Schimke Immunoosseous Dysplasia	SMARCAL1	AR	Reduced Risk
Smith-Lemli-Opitz Syndrome DHCR7 AR Reduced Risk Spinal Muscular Atrophy SMN1 AR Reduced Risk SMN2 copy number: 2 Spinal Muscular Atrophy SMN1 AR Reduced Risk SMN2 copy number: >=3 c. '3+80T>G: Negative	Segawa Syndrome	TH	AR	Reduced Risk
Spinal Muscular Atrophy SMN1 AR Reduced Risk SMN2 copy number: 2 SMN1 copy number: 2 SMN2 copy number: >=3 c. '3+80T>Q: Negative	Sjogren-Larsson Syndrome	ALDH3A2	AR	Reduced Risk
Spinal Muscular Atrophy SMN1 AR Reduced Risk SMN2 copy number: >=3 c. *3+80T>G: Negative	Smith-Lemli-Opitz Syndrome	DHCR7	AR	Reduced Risk
	Spinal Muscular Atrophy	SMN1	AR	Reduced Risk SMN2 copy number: >=3
Spondylothoracic Dysostosis MESP2 AR Reduced Risk				





Steel Syndrome	COL27A1	AR	Reduced Risk
Stuve-Wiedemann Syndrome	LIFR	AR	Reduced Risk
Sulfate Transporter-Related Osteochondrodysplasia	SLC26A2	AR	Reduced Risk
Tay-Sachs Disease	HEXA	AR	Reduced Risk
Tyrosinemia, Type I	FAH	AR	Reduced Risk
Usher Syndrome, Type IB	MYO7A	AR	Reduced Risk
Usher Syndrome, Type IC	USH1C	AR	Reduced Risk
Usher Syndrome, Type ID	CDH23	AR	Reduced Risk
Usher Syndrome, Type IF	PCDH15	AR	Reduced Risk
Usher Syndrome, Type IIA	USH2A	AR	Reduced Risk
Usher Syndrome, Type III	CLRN1	AR	Reduced Risk
Very Long Chain Acyl-CoA Dehydrogenase Deficiency	ACADVL	AR	Reduced Risk
Walker-Warburg Syndrome and Other <i>FKTN</i> -Related Dystrophies	FKTN	AR	Reduced Risk
Wilson Disease	ATP7B	AR	Reduced Risk
Wolman Disease / Cholesteryl Ester Storage Disease	LIPA	AR	Reduced Risk
X-Linked Juvenile Retinoschisis	RS1	XL	Reduced Risk
X-Linked Severe Combined Immunodeficiency	IL2RG	XL	Reduced Risk
Zellweger Syndrome Spectrum (PEX10-Related)	PEX10	AR	Reduced Risk
Zellweger Syndrome Spectrum (PEX1-Related)	PEX1	AR	Reduced Risk
Zellweger Syndrome Spectrum (PEX2-Related)	PEX2	AR	Reduced Risk
Zellweger Syndrome Spectrum (PEX6-Related)	PEX6	AR	Reduced Risk

AR=Autosomal recessive; XL=X-linked

Test methods and comments

Genomic DNA isolated from this patient was analyzed by one or more of the following methodologies, as applicable:

Fragile X CGG Repeat Analysis (Analytical Detection Rate >99%)

PCR amplification using Asuragen, Inc. AmplideX® FMR1 PCR reagents followed by capillary electrophoresis for allele sizing was performed. Samples positive for FMR1 CGG repeats in the premutation and full mutation size range were further analyzed by Southern blot analysis to assess the size and methylation status of the FMR1 CGG repeat.

Genotyping (Analytical Detection Rate >99%)

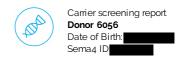
Multiplex PCR amplification and allele specific primer extension analyses using the MassARRAY® System were used to identify variants that are complex in nature or are present in low copy repeats. Rare sequence variants may interfere with assay performance.

Multiplex Ligation-Dependent Probe Amplification (MLPA) (Analytical Detection Rate >99%)

MLPA® probe sets and reagents from MRC-Holland were used for copy number analysis of specific targets versus known control samples. False positive or negative results may occur due to rare sequence variants in target regions detected by MLPA probes. Analytical sensitivity and specificity of the MLPA method are both 99%.

For alpha thalassemia, the copy numbers of the *HBA1* and *HBA2* genes were analyzed. Alpha-globin gene deletions, triplications, and the Constant Spring (CS) mutation are assessed. This test is expected to detect approximately 90% of all alpha-thalassemia mutations, varying by ethnicity. Carriers of alpha-thalassemia with three or more *HBA* copies on one chromosome, and one or no copies on the other chromosome,





may not be detected. With the exception of triplications, other benign alpha-globin gene polymorphisms will not be reported. Analyses of *HBA1* and *HBA2* are performed in association with long-range PCR of the coding regions followed by short-read sequencing.

For Duchenne muscular dystrophy, the copy numbers of all *DMD* exons were analyzed. Potentially pathogenic single exon deletions and duplications are confirmed by a second method. Analysis of *DMD* is performed in association with sequencing of the coding regions.

For congenital adrenal hyperplasia, the copy number of the *CYP21A2* gene was analyzed. This analysis can detect large deletions due to unequal meiotic crossing-over between *CYP21A2* and the pseudogene *CYP21A1P*. These 30-kb deletions make up approximately 20% of *CYP21A2* pathogenic alleles. This test may also identify certain point mutations in *CYP21A2* caused by gene conversion events between *CYP21A2* and *CYP21A1P*. Some carriers may not be identified by dosage sensitive methods as this testing cannot detect individuals with two copies (duplication) of the *CYP21A2* gene on one chromosome and loss of *CYP21A2* (deletion) on the other chromosome. Analysis of *CYP21A2* is performed in association with long-range PCR of the coding regions followed by short-read sequencing.

For spinal muscular atrophy (SMA), the copy numbers of the *SMN1* and *SMN2* genes were analyzed. The individual dosage of exons 7 and 8 as well as the combined dosage of exons 1, 4, 6 and 8 of *SMN1* and *SMN2* were assessed. Copy number gains and losses can be detected with this assay. Depending on ethnicity, 6 - 29 % of carriers will not be identified by dosage sensitive methods as this testing cannot detect individuals with two copies (duplication) of the *SMN1* gene on one chromosome and loss of *SMN1* (deletion) on the other chromosome (silent 2+0 carrier) or individuals that carry an intragenic mutation in *SMN1*. Please also note that 2% of individuals with SMA have an *SMN1* mutation that occurred *de novo*. Typically in these cases, only one parent is an SMA carrier.

The presence of the c.*3+80T>G (chr5:70,247,901T>G) variant allele in an individual with Ashkenazi Jewish or Asian ancestry is typically indicative of a duplication of *SMN1*. When present in an Ashkenazi Jewish or Asian individual with two copies of *SMN1*, c.*3+80T>G is likely indicative of a silent (2+0) carrier. In individuals with two copies of *SMN1* with African American, Hispanic or Caucasian ancestry, the presence or absence of c.*3+80T>G significantly increases or decreases, respectively, the likelihood of being a silent 2+0 silent carrier.

Pathogenic or likely pathogenic sequence variants in exon 7 may be detected during testing for the c.*3+80T>G variant allele; these will be reported if confirmed to be located in SMN1 using locus-specific Sanger primers

Pathogenic or likely pathogenic sequence variants in exon 7 may be detected during testing for the c.*3+80T>G variant allele; these will be reported if confirmed to be located in SMN1 using locus-specific Sanger primers.

MLPA for Gaucher disease (*GBA*), cystic fibrosis (*CFTR*), and non-syndromic hearing loss (*GJB2/GJB6*) will only be performed if indicated for confirmation of detected CNVs. If *GBA* analysis was performed, the copy numbers of exons 1, 3, 4, and 6 - 10 of the *GBA* gene (of 11 exons total) were analyzed. If *CFTR* analysis was performed, the copy numbers of all 27 *CFTR* exons were analyzed. If *GJB2/GJB6* analysis was performed, the copy number of the two *GJB2* exons were analyzed, as well as the presence or absence of the two upstream deletions of the *GJB2* regulatory region, del(*GJB6* -D13S1830) and del(*GJB6* -D13S1854).

Next Generation Sequencing (NGS) (Analytical Detection Rate >95%)

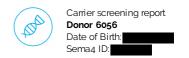
NGS was performed on a panel of genes for the purpose of identifying pathogenic or likely pathogenic variants.

Agilent SureSelectTMQXT technology was used with a custom capture library to target the exonic regions and intron/exon splice junctions of the relevant genes, as well as a number of UTR, intronic or promoter regions that contain previously reported mutations. Samples were pooled and sequenced on the Illumina HiSeq 2500 platform in the Rapid Run mode or the Illumina NovaSeq platform in the Xp workflow, using 100 bp paired-end reads. The sequencing data was analyzed using a custom bioinformatics algorithm designed and validated in house. The coding exons and splice junctions of the known protein-coding RefSeq genes were assessed for the average depth of coverage (minimum of 20X) and data quality threshold values. Most exons not meeting a minimum of >20X read depth across the exon are further analyzed by Sanger sequencing. Please note that several genomic regions present difficulties in mapping or obtaining read depth >20X. The exons contained within these regions are noted within Table 1 (as "Exceptions") and will not be reflexed to Sanger sequencing if the mapping quality or coverage is poor. Any variants identified during testing in these regions are confirmed by a second method and reported if determined to be pathogenic or likely pathogenic. However, as there is a possibility of false negative results within these regions, detection rates and residual risks for these genes have been calculated with the presumption that variants in these exons will not be detected, unless included in the MassARRAY® genotyping platform.

This test will detect variants within the exons and the intron-exon boundaries of the target regions. Variants outside these regions may not be detected, including, but not limited to, UTRs, promoters, and deep intronic areas, or regions that fall into the Exceptions mentioned above. This technology may not detect all small insertion/deletions and is not diagnostic for repeat expansions and structural genomic variation. In addition, a mutation(s) in a gene not included on the panel could be present in this patient.

Variant interpretation and classification was performed based on the American College of Medical Genetics Standards and Guidelines for the Interpretation of Sequence Variants (Richards et al. 2015). All potentially pathogenic variants may be confirmed by either a specific genotyping assay or Sanger sequencing, if indicated. Any benign variants, likely benign variants or variants of uncertain significance identified during this analysis will not be reported.





Copy Number Variant Analysis (Analytical Detection Rate >95%)

Large duplications and deletions were called from the relative read depths on an exon-by-exon basis using a custom exome hidden Markov model (XHMM) algorithm. Deletions or duplications determined to be pathogenic or likely pathogenic were confirmed by either a custom arrayCGH platform, quantitative PCR, or MLPA (depending on CNV size and gene content). While this algorithm is designed to pick up deletions and duplications of 2 or more exons in length, potentially pathogenic single-exon CNVs will be confirmed and reported, if detected.

Exon Array (Confirmation method) (Accuracy >99%)

The customized oligonucleotide microarray (Oxford Gene Technology) is a highly-targeted exon-focused array capable of detecting medically relevant microdeletions and microduplications at a much higher resolution than traditional aCGH methods. Each array matrix has approximately 180,000 60-mer oligonucleotide probes that cover the entire genome. This platform is designed based on human genome NCBI Build 37 (hg19) and the CGH probes are enriched to target the exonic regions of the genes in this panel.

Quantitative PCR (Confirmation method) (Accuracy >99%)

The relative quantification PCR is utilized on a Roche Universal Library Probe (UPL) system, which relates the PCR signal of the target region in one group to another. To test for genomic imbalances, both sample DNA and reference DNA is amplified with primer/probe sets that specific to the target region and a control region with known genomic copy number. Relative genomic copy numbers are calculated based on the standard $\Delta\Delta$ Ct formula.

Long-Range PCR (Analytical Detection Rate >99%)

Long-range PCR was performed to generate locus-specific amplicons for *CYP21A2*, *HBA1* and *HBA2* and *GBA*. The PCR products were then prepared for short-read NGS sequencing and sequenced. Sequenced reads were mapped back to the original genomic locus and run through the bioinformatics pipeline. If indicated, copy number from MLPA was correlated with the sequencing output to analyze the results. For *CYP21A2*, a certain percentage of healthy individuals carry a duplication of the *CYP21A2* gene, which has no clinical consequences. In cases where two copies of a gene are located on the same chromosome in tandem, only the second copy will be amplified and assessed for potentially pathogenic variants, due to size limitations of the PCR reaction. However, because these alleles contain at least two copies of the *CYP21A2* gene in tandem, it is expected that this patient has at least one functional gene in the tandem allele and this patient is therefore less likely to be a carrier. When an individual carries both a duplication allele and a pathogenic variant, or multiple pathogenic variants, the current analysis may not be able to determine the phase (cis/trans configuration) of the *CYP21A2* alleles identified. Family studies may be required in certain scenarios where phasing is required to determine the carrier status.

Residual Risk Calculations

Carrier frequencies and detection rates for each ethnicity were calculated through the combination of internal curations of >28,000 variants and genomic frequency data from >138,000 individuals across seven ethnic groups in the gnomAD database. Additional variants in HGMD and novel deleterious variants were also incorporated into the calculation. Residual risk values are calculated using a Bayesian analysis combining the *a priori* risk of being a pathogenic mutation carrier (carrier frequency) and the detection rate. They are provided only as a guide for assessing approximate risk given a negative result, and values will vary based on the exact ethnic background of an individual. This report does not represent medical advice but should be interpreted by a genetic counselor, medical geneticist or physician skilled in genetic result interpretation and the relevant medical literature.

Sanger Sequencing (Confirmation method) (Accuracy >99%)

Sanger sequencing, as indicated, was performed using BigDye Terminator chemistry with the ABI 3730 DNA analyzer with target specific amplicons. It also may be used to supplement specific guaranteed target regions that fail NGS sequencing due to poor quality or low depth of coverage (<20 reads) or as a confirmatory method for NGS positive results. False negative results may occur if rare variants interfere with amplification or annealing.

Please note these tests were developed and their performance characteristics were determined by Mount Sinai Genomics, Inc. They have not been cleared or approved by the FDA. These analyses generally provide highly accurate information regarding the patient's carrier or affected status. Despite this high level of accuracy, it should be kept in mind that there are many potential sources of diagnostic error, including misidentification of samples, polymorphisms, or other rare genetic variants that interfere with analysis. Families should understand that rare diagnostic errors may occur for these reasons.

SELECTED REFERENCES

Carrier Screening

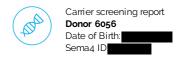
Grody W et al. ACMG position statement on prenatal/preconception expanded carrier screening. Genet Med. 2013 15:482-3.

Fragile X syndrome:

Chen L et al. An information-rich CGG repeat primed PCR that detects the full range of Fragile X expanded alleles and minimizes the need for Southern blot analysis. *J Mol Diag* 2010 12:589-600.

Spinal Muscular Atrophy:





Luo M et al. An Ashkenazi Jewish SMN1 haplotype specific to duplication alleles improves pan-ethnic carrier screening for spinal muscular atrophy. *Genet Med* . 2014 16:149-56.

Ashkenazi Jewish Disorders:

Scott SA et al. Experience with carrier screening and prenatal diagnosis for sixteen Ashkenazi Jewish Genetic Diseases. *Hum. Mutat.* 2010 31:1-11.

Duchenne Muscular Dystrophy:

Flanigan KM et al. Mutational spectrum of DMD mutations in dystrophinopathy patients: application of modern diagnostic techniques to a large cohort. *Hum Mutat* . 2009 30:1657-66.

Variant Classification:

Richards S et al. Standards and guidelines for the interpretation of sequence variants: a joint consensus recommendation of the American College of Medical Genetics and Genomics and the Association for Molecular Pathology. *Genet Med.* 2015 May;17(5):405-24 Additional disease-specific references available upon request.