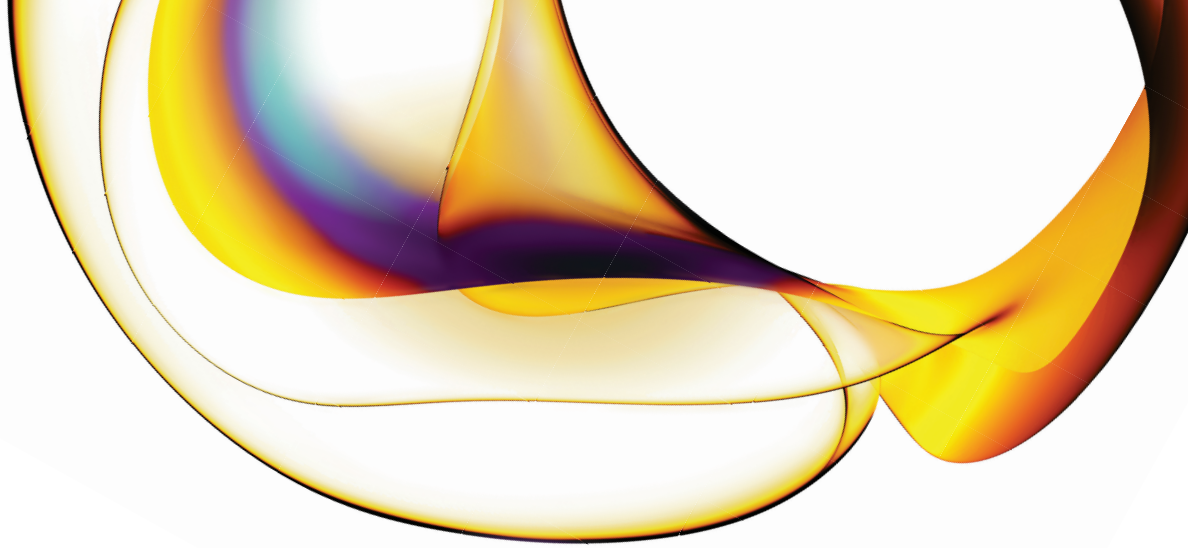

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Whole Exome and Whole Genome Sequencing

Secondary Findings | Carrier Status

In genetics, a carrier refers to an individual who carries a gene mutation that can be passed down to their offspring but is not necessarily affected by it. Carrier status tests can detect genetic changes that can cause hereditary disorders in future generations.

Disorders screened include

Gene	Associated Disorders
AAAS	Achalasia-Addisonianism-Alacrima Syndrome
ABCB11	Progressive familial intrahepatic cholestasis
ABCC6	Pseudoxanthoma Elasticum
ABCC8	Familial hyperinsulinism
ACAD9	Mitochondrial complex I deficiency
ACADL	Long chain acyl-CoA dehydrogenase deficiency
ACADM	Medium chain acyl-CoA dehydrogenase deficiency
ACADVL	Very long chain acyl-CoA dehydrogenase deficiency
ACAT1	Beta-ketothiolase Deficiency
ACOX1	Peroxisomal acyl-CoA Oxidase Deficiency
ACSF3	Combined Malonic and Methylmalonic Aciduria

Gene	Associated Disorders
ADA	Adenosine Deaminase Deficiency
ADAMTS2	Ehlers-Danlos Syndrome, Dermatosparaxis Type
ADGRG1	Bilateral Frontoparietal Polymicrogyria
AGA	Aspartylglucosaminuria
AGL	Glycogen Storage Disease, Type III
AGPS	Rhizomelic Chondrodysplasia Punctata, Type 3
AGXT	Primary Hyperoxaluria, Type 1
AIRE	Polyglandular Autoimmune Syndrome, Type 1
ALDH3A2	Sjogren-Larsson Syndrome
ALDH7A1	Pyridoxine-Dependent Epilepsy
ALG6	Congenital Disorder of Glycosylation, Type Ic
ALMS1	Alstrom Syndrome

Gene	Associated Disorders
ALPL	Hypophosphatasia
AMT	Glycine Encephalopathy AMT-Related
AP3B1	Hermansky-Pudlak Syndrome 2
APOB	Hypobetalipoproteinemia
AQP2	Nephrogenic Diabetes Insipidus
AR	Androgen Insensitivity Syndrome
ARG1	Argininemia
ARSA	Metachromatic Leukodystrophy, ARSA-Related
ARSB	Mucopolysaccharidosis Type VI
ASL	Argininosuccinic Aciduria
ASNS	Asparagine Synthetase Deficiency
ASPA	Canavan Disease
ASPM	Primary Microcephaly 5
ASS1	Citrullinemia, Type 1
ATM	Ataxia-Telangiectasia
ATP6V1B1	Renal Tubular Acidosis with Deafness
ATP7A	Menkes Syndrome
ATP7B	Wilson Disease
ATRX	Alpha-Thalassemia Intellectual Disability Syndrome
BBS1	Bardet-Biedl Syndrome 1
BBS10	Bardet-Biedl Syndrome 10
BBS12	Bardet-Biedl Syndrome 12
BBS4	Bardet-Biedl Syndrome 4
BCHE	Pseudocholinesterase Deficiency
BCKDHA	Maple Syrup Urine Disease, Type Ia
BCKDHB	Maple Syrup Urine Disease, Type Ib
BCS1L	GRACILE syndrome
BLM	Bloom Syndrome
BSND	Bartter Syndrome
BTD	Biotinidase Deficiency
BTK	Agammaglobulinemia, X-Linked

Gene	Associated Disorders
CAPN3	Limb-Girdle Muscular Dystrophy, Type 2A
CASQ2	Ventricular tachycardia, catecholaminergic polymorphic, 2
CBS	Homocystinuria due to Cystathionine Beta-Synthase Deficiency
CDH23	Usher Syndrome, Type 1D
CEP290	Joubert Syndrome 5
CERKL	Retinitis Pigmentosa 26
CFTR	Cystic Fibrosis
CHM	Choroideremia
CHRNE	Congenital Myasthenic Syndrome 4
CIITA	Bare Lymphocyte Syndrome Type II
CLCN1	Myotonia congenita, recessive
CLN3	Neuronal Ceroid Lipofuscinosis 3
CLN5	Neuronal Ceroid Lipofuscinosis 5
CLN6	Neuronal Ceroid Lipofuscinosis 6
CLN8	Neuronal Ceroid Lipofuscinosis 8
CLRN1	Usher Syndrome, Type 3
CNGB3	Achromatopsia 3
COL27A1	Steel Syndrome
COL4A3	Alport Syndrome, COL4A3-Related
COL4A4	Alport Syndrome, COL4A4-Related
COL4A5	Alport Syndrome, X-Linked
COL7A1	Dystrophic Epidermolysis Bullosa
CPS1	Carbamoyl Phosphate Synthetase I Deficiency
CPT1A	Carnitine Palmitoyltransferase Ia Deficiency
CPT2	Carnitine Palmitoyltransferase II Deficiency
CRB1	Leber Congenital Amaurosis 8
CRPPA	Muscular dystrophy-dystroglycanopathy, type A, 7; C, 7
CTNS	Cystinosis

Gene	Associated Disorders
CTSK	Pycnodysostosis
CYBA	Chronic Granulomatous Disease
CYBB	Chronic Granulomatous Disease, X-Linked
CYP17A1	17-Alpha-Hydroxylase Deficient Congenital Adrenal Hyperplasia
CYP19A1	Aromatase Deficiency
CYP1B1	Primary Congenital Glaucoma 3
CYP27A1	Cerebrotendinous Xanthomatosis
DBT	Maple Syrup Urine Disease, Type II
DCLRE1C	Omenn Syndrome, DCLRE1C-Related
DES	Myopathy, myofibrillar, 1
DHCR7	Smith-Lemli-Opitz Syndrome
DHDDS	Retinitis Pigmentosa 59
DKC1	Dyskeratosis Congenita, X-Linked
DLD	Dihydrolipoamide Dehydrogenase Deficiency
DMD	Duchenne/Becker Muscular Dystrophy
DNAH5	Primary Ciliary Dyskinesia 3
DNAI1	Primary Ciliary Dyskinesia 37
DNAI2	Primary Ciliary Dyskinesia 9
DOK7	Congenital Myasthenic Syndrome 10
DPYD	Dihydropyrimidine Dehydrogenase Deficiency
DSP	Cardiomyopathy, dilated, with woolly hair and keratoderma; Epidermolysis bullosa, lethal acantholytic; Skin fragility-woolly hair syndrome
DYSF	Limb-Girdle Muscular Dystrophy, Type 2B
EDA	Hypohidrotic Ectodermal Dysplasia I, X-Linked
EDAR	Hypohidrotic Ectodermal Dysplasia 10B
EIF2B5	Leukoencephalopathy with Vanishing White Matter

Gene	Associated Disorders
ELP1	Familial dysautonomia; Medulloblastoma
EMD	Emery-Dreifuss Muscular Dystrophy 1, X-Linked
ERCC6	Cockayne Syndrome, Type B
ERCC8	Cockayne Syndrome, Type A
ESCO2	Roberts Syndrome
ETFA	Glutaric Acidemia IIA
ETFDH	Glutaric Acidemia IIC
ETHE1	Ethylmalonic Encephalopathy
EVC	Ellis-van Creveld Syndrome, EVC-Related
EVC2	Ellis-van Creveld Syndrome, EVC2-Related
EYS	Retinitis Pigmentosa 25
F11	Factor XI Deficiency
F8	Hemophilia A
F9	Hemophilia B
FAH	Tyrosinemia, Type I
FAM161A	Retinitis Pigmentosa 28
FANCA	Fanconi Anemia, Group A
FANCC	Fanconi Anemia, Group C
FANCG	Fanconi Anemia, Group G
FH	Fumarase Deficiency
FKRP	Limb-Girdle Muscular Dystrophy, Type 2I
FKTN	Walker-Warburg Syndrome FKTN-Related
G6PC1	Glycogen Storage Disease, Type IA
G6PD	Glucose-6-Phosphate Dehydrogenase Deficiency
GAA	Glycogen Storage Disease, Type II
GALC	Krabbe Disease
GALK1	Galactokinase Deficiency
GALT	Galactosemia

Gene	Associated Disorders
GAMT	Guanidinoacetate Methyltransferase Deficiency
GAN	Giant Axonal Neuropathy 1
GBE1	Glycogen Storage Disease IV
GCDH	Glutaric Acidemia I
GFM1	Combined Oxidative Phosphorylation Deficiency 1
GJB1	Charcot-Marie-Tooth Disease with Deafness, X-Linked
GJB2	Non-Syndromic Hearing Loss, GJB2-Related
GJB6	Non-Syndromic Hearing Loss, GJB6-Related
GLA	Fabry Disesae
GLB1	Mucopolysaccharidosis Type IVB
GLDC	Glycine Encephalopathym, GLDC-Related
GLE1	Congenital Arthrogryposis with Anterior Horn Cell Disesae
GLUL	Congenital Glutamine Deficency
GNE	Inclusion Body Myopathy 2
GNPTAB	Mucolipidosis II
GNPTG	Mucolipidosis III
GNS	Mucopolysaccharidosis, Type IIID
GP1BA	Bernard-Soulier Syndrome, Type A1
GP9	Bernard-Soulier Syndrome, Type C
GRHPR	Primary Hyperoxaluria Type II
GUCY2D	Leber Congenital Amaurosis 1
HADH	3-Hydroxyacyl-CoA Dehydrogenase Deficiency
HADHA	Long-Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency
HAX1	Severe Congenital Neutropenia, 3
HBB	Beta-Hemoglobinopathies
HEXA	Tay-Sachs Disease
HEXB	Sandhoff Disease

Gene	Associated Disorders
HFE	Hemochromatosis Type 2A
HGSNAT	Mucopolysaccharidosis Type IIIC
HJV	Hemochromatosis, type 2A
HLCS	Holocarboxylase Synthetase Deficiency
HMGCL	HMG-CoA Lyase Deficiency
HOGA1	Primary Hyperoxaluria, Type III
HPRT1	Lesch-Nyham Syndrome
HPS1	Hermansky-Pudlak Syndrome 1
HPS3	Hermansky-Pudlak Syndrome 3
HSD17B3	17-Beta Hydroxysteroid Dehydrogenase III
HSD17B4	D-Bifunctional Protein Deficiency
HSD3B2	3-Beta-Hydroxysteroid Dehydrogenase 2 Deficient CongenitalAdrenal Hyperplasia
HYAL1	Mucopolysaccharidosis Type IX
HYLS1	Hydrolethalus Syndrome
IDS	Mucopolysaccharidosis Type II
IDUA	Mucopolysaccharidosis Type I (Hurler Syndrome)
IL2RG	Severe Combined Immunodeficiency, X-Linked
IVD	Isovaleric Acidemia
KCNJ11	Congenital Hyperinsulinism
KCNQ1	Jervell and Lange-Nielsen syndrome
L1CAM	Hydrocephalus, L1CAM-Related
LAMA2	LAMA-2-Related Muscular Dystrophy
LAMA3	Herlitz Junctional Epidermolysis Bullosa, LAMA3-Related
LAMB3	Herlitz Junctional Epidermolysis Bullosa, LAMB3-Related
LAMC2	Herlitz Junctional Epidermolysis Bullosa, LAMC2-Related
LARGE1	Walker-Warburg Syndrome LARGE1-Related

Gene	Associated Disorders
LCA5	Leber Congenital Amaurosis 5
LDLR	Familial Hypercholesterolemia, LDLR-Related
LDLRAP1	Familial Hypercholesterolemia, LDLRAP1-Related
LHX3	Combined Pituitary Hormone Deficiency 3
LIFR	Stuve-Wiedemann Syndrome
LIPA	Wolman Disease
LIPH	Hypotrichosis 7
LMNA	Charcot-Marie-Tooth disease, type 2B1; Emery-Dreifuss muscular dystrophy 3; Mandibuloacral dysplasia
LOXHD1	Deafness, Autosomal Recessive 77
LPL	Lipoprotein Lipase Deficiency
LRPPRC	Leigh Syndrome, French Canadian Type
LZTR1	Noonan syndrome 2
MAN2B1	Alpha-Mannosidosis
MCCC1	3-Methylcrotonyl-CoA Carboxylase 1 Deficiency
MCCC2	3-Methylcrotonyl-CoA Carboxylase 2 Deficiency
MCOLN1	Mucolipidosis IV
MED17	Postnatal Progressive Microcephaly with Seizures and Brian Atrophy
MEFV	Familial Mediterranean Fever
MESP2	Spondylothoracic Dysostosis
MFSD8	Neuronal Ceroid Lipofuscinosis 7
MKKS	Bardet-Biedle Syndrome 6
MKS1	Meckel-Gruber Syndrome 1
MLC1	Megalencephalic Leukoencephalopathy with Subcortical Cysts 1
MMAA	Methylmalonic Aciduria, cb1A Type
MMAB	Methylmalonic Aciduria, cb1B Type
MMACHC	Methylmalonic Aciduria and Homocystinuria, cb1C Type
MMADHC	Methylmalonic Aciduria and Homocystinuria, cb1D Type
MMUT	Methylmalonic Aciduria, Type mut(0)
MPI	Congenital Disorder of Glycosylation, Type Ib
MPL	Congenital Amegakaryocytic Thrombocytopenia
MPV17	Charcot-Marie-Tooth Disease, Axonal, Type 2EE
MTHFR	Homocystinuria due to MTHFR Deficiency
MTM1	Myotubular Myopathy, X-Linked
MTRR	Homocystinuria Type cblE
MTTP	Abetalipoproteinemia
MUTYH	Adenomas, multiple colorectal
MYO7A	Usher Syndrome, Type 1B
NAGLU	Mucopolysaccharidosis, Type IIIB
NAGS	N-Acetylglutamate Synthase Deficiency
NBN	Nijmegen Breakage Syndrome
NDRG1	Charcot-Marie-Tooth Disease, Type 4D
NDUFAF5	Mitochondrial Complex I Deficiency, Nuclear Type 16
NDUFS6	Mitochondrial Complex I Deficiency, Nuclear Type 9
NEB	Nemaline Myopathy 2
NPC1	Niemann-Pick Disease, Type C1/D
NPC2	Niemann-Pick Disease, Type C2
NPHP1	Joubert syndrome 4; Nephronophthisis 1, juvenile; Senior-Loken syndrome-1
NPHS1	Nephrotic Syndrome, Type 1
NPHS2	Nephrotic Syndrome, Type 2

Gene	Associated Disorders
NR0B1	X-Linked Congenital Adrenal Hypoplasia
NR2E3	Enhanced S-Cone Syndrome
NTRK1	Familial Dysautonomia, Type II
OAT	Ornithine Aminotransferase Deficiency
OPA3	3-Methylglutaconic Aciduria, Type III
OTC	Ornithine Carbamoyltransferase Deficiency
PAH	Phenylketonuria
PC	Pyruvate Carboxylase Deficiency
PCCA	Propionic Acidemia, PCCA-Related
PCCB	Propionic Acidemia, PCCB-Related
PCDH15	Usher Syndrome, Type 1F
PDHA1	Pyruvate Dehydrogenase E1-Alpha Deficiency
PDHB	Pyruvate Dehydrogenase E1-Beta Deficiency
PEX1	Zellweger Syndrome Disorders, PEX1-Related
PEX10	Zellweger Syndrome Disorders, PEX10-Related
PEX12	Zellweger Syndrome Disorders, PEX12-Related
PEX2	Zellweger Syndrome Disorders, PEX2-Related
PEX6	Zellweger Syndrome Disorders, PEX6-Related
PEX7	Rhizomelic Chondrodysplasia Punctata, Type 1
PFKM	Glycogen Storage Disease VII
PHGDH	Neu-Laxova Syndrome
PKHD1	Autosomal Recessive Polycystic Kidney Disease
PLP1	Spastic Paraplegia 2, X-Linked
PMM2	Congenital Disorder of Glycosylation, Type IA
POLG	POLG-Related Disorders

Gene	Associated Disorders
POMGNT1	Muscle-Eye-Brain Disease, POMGNT1-Related
PPT1	Neuronal Ceroid Lipofuscinosis 1
PROP1	Combined Pituitary Hormone Deficiency 2
PRPS1	Arts Syndrome
PSAP	Metachromatic Leukodystrophy, PSAP-Related
PTS	6-Pyruvoyl-Tetrahydropterin Synthase (PTPS) Deficiency
PUS1	Mitochondrial Myopathy and Sideroblastic Anemia
PYGM	Glycogen Storage Disease V
RAB23	Carpenter Syndrome
RAG1	Severe Combined Immunodeficiency/ Omenn Syndrome, RAG1-Related
RAG2	Severe Combined Immunodeficiency/ Omenn Syndrome, RAG2-Related
RAPSN	Congenital Myasthenic Syndrome 11
RARS2	Pontocerebellar Hypoplasia, Type 6
RDH12	Leber Congenital Amaurosis 13
RMRP	Cartilage-Hair Hypoplasia
RPE65	Leber Congenital Amaurosis 2
RPGRIP1L	Joubert Syndrome 7
RS1	X-Linked Juvenile Retinoschisis 1
RTEL1	Dyskeratosis Congenita
RYR1	Central core disease; Minicore myopathy with external ophthalmoplegia; Neuromuscular disease, congenital, with uniform type 1 fiber
SACS	Autosomal Recessive Spastic Ataxia of Charlevoix-Sa- Guenay
SAMHD1	Aicardi-Goutières Syndrome 5
SBDS	Shwachman-Diamond Syndrome 1
SEPSECS	Pontocerebellar Hypoplasia, Type 2D

Gene	Associated Disorders
SGCA	Muscular dystrophy, limb-girdle, autosomal recessive 3
SGCB	Muscular dystrophy, limb-girdle, autosomal recessive 4
SGCD	Muscular dystrophy, limb-girdle, autosomal recessive 6
SGCG	Muscular dystrophy, limb-girdle, autosomal recessive 5
SGSH	Mucopolysaccharidosis, Type IIIA
SLC12A3	Gitelman Syndrome
SLC12A6	Andermann Syndrome
SLC17A5	Sall Disease
SLC22A5	Carnitine Deficiency
SLC25A13	Citrullinemia, Type 2
SLC25A15	Hyperornithinemia-Hyperammonemia-Homocitrullinuria Syndrome
SLC26A2	Achondrogenesis, Type IB
SLC26A4	Pendred Syndrome
SLC35A3	Arthrogryposis, Mental Retardation and Seizure (AMRS)
SLC37A4	Glycogen Storage Disease, Type IB
SLC39A4	Acrodermatitis Enteropathica
SLC3A1	Cystinuria
SLC4A11	Corneal Dystrophy and Perceptive Deafness
SLC6A8	Cerebral Creatine Deficiency Syndrome 1
SLC7A7	Lysinuric Protein Intolerance
SMARCAL1	Schimke Immunoosseous Dysplasia
SMPD1	Niemann-Pick Disease, Type A/B
STAR	Lipoid Congenital Adrenal Hyperplasia
STS	Ichthyosis, X-linked
SUMF1	Multiple Sulfatase Deficiency
TAT	Tyrosinemia, Type II
TCIRG1	Infantile Malignant Osteopertosis 1

Gene	Associated Disorders
TECPR2	Spastic Paraplegia 49
TFR2	Hemochromatosis Type 3
TGM1	Lamellar Ichthyosis 1
TH	Segawa Syndrome
TMEM216	Joubert Syndrome 2
TMEM67	Joubert Syndrome 6/ Meckel Syndrome 3
TPP1	Neuronal Ceroid Lipofuscinosis 2
TRDN	Cardiac arrhythmia syndrome, with or without skeletal muscle weakness
TRMU	Liver Failure, Infantile, Transient (LFIT)
TSEN54	Pontocerebellar Hypoplasia, Type 2A/4
TSFM	Combined Oxidative Phosphorylation Deficiency 3
TTC8	Bardet-Biedl Syndrome 8
TTPA	Ataxia with Vitamin E Deficiency
TYMP	Myoneurogastrointestinal Encephalopathy Syndrome (MNGIE)
UBR1	Johanson-Blizzard Syndrome
USH1C	Usher Syndrome, Type 1C
USH2A	Usher Syndrome, Type 2A
VPS13A	Choreoacanthocytosis
VPS13B	Cohen Syndrome
VPS45	Congenital Neutropenia 5
VRK1	Pontocerebellar Hypoplasia Type 1A
VSX2	Microphthalmia, VSX2-Related
VWF	von Willebrand disease, types 2A, 2B, 2M, 2N and 3
WNT10A	Odontoonychodermal Dysplasia
XPA	Xeroderma Pigmentosum Group A
XPC	Xeroderma Pigmentosum Group C
ZFYVE26	Spastic Paraplegia 15

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