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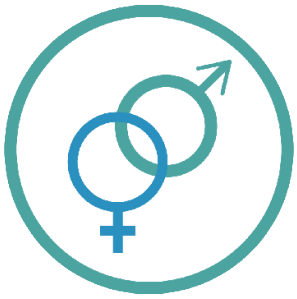

Genomic Inform™ Test

Carrier screening gene list

Genomic Inform™ provides comprehensive testing of more than 300 autosomal recessive and X-linked conditions, including all American College of Medical Genetics and Genomics (ACMG), American College of Obstetricians and Gynecologists (ACOG) and Jewish Genetic Disease Consortium recommended disorders.

Autosomal recessive

DISORDER	GENE
11-beta-hydroxylase-deficient congenital adrenal hyperplasia	CYP11B1
17-alpha-hydroxylase-deficient congenital adrenal hyperplasia	CYP17A1
17-beta-hydroxysteroid dehydrogenase deficiency, type III	HSD17B3
2-Methylbutyryl-CoA dehydrogenase deficiency	ACADSB
3-beta-hydroxysteroid dehydrogenase type II deficiency (Congenital adrenal hyperplasia)	HSD3B2
3-hydroxy-3-methylglutarayl-CoA (HMG-CoA) lyase deficiency	HMGCL
3-hydroxyacyl-CoA dehydrogenase deficiency	HADH
3-methylcrotonyl-CoA carboxylase deficiency	MCC1, MCC2
3-methylglutaconic aciduria type III (Costeff optic atrophy)[§]	OPA3
6-pyruvoyl-tetrahydropterin synthase deficiency	PTS
Abetalipoproteinemia[§]	MTTP
ACAD9 deficiency	ACAD9
Achalasia-addisonianism-alacrima syndrome	AAAS
Achromatopsia	ATF6, CNGA3, CNGB3, GNAT2, PDE6C
Acrodermatitis enteropathica	SLC39A4
Acute infantile liver failure[§]	TRMU
Adenosine deaminase 2 deficiency	ADA2
Adenosine deaminase deficiency	ADA
Adult polyglucosan body disease	GBE1
Aicardi-Goutieres syndrome	ADAR, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, TREX1
Alexander disease	GFAP
Alkaptonuria	HGD
Alpha-1 antitrypsin deficiency	SERPINA1
Alpha-mannosidosis	MAN2B1
Alpha-thalassemia[†‡§]	HBA1/HBA2
Alport Syndrome[§]	COL4A3, COL4A4
Alström syndrome	ALMS1

DISORDER	GENE
Amish infantile epilepsy syndrome	ST3GAL5
Andermann syndrome	SLC12A6
Arginase deficiency	ARG1
Argininosuccinic aciduria	ASL
Aromatase deficiency	CYP19A1
Arthrogryposis, mental retardation, and seizures[§]	SLC35A3
Asparagine synthetase deficiency[§]	ASNS
Aspartylglucosaminuria	AGA
Ataxia with vitamin E deficiency	TTPA
Ataxia-telangiectasia[§]	ATM
Ataxia-telangiectasia-like disorder 1	MRE11A
Atypical Gaucher disease	PSAP
Atypical Krabbe disease	PSAP
Autoimmune polyendocrinopathy with candidiasis and ectodermal dysplasia§	AIRE
Autosomal recessive deafness 77[§]	LOXHD1
Autosomal recessive spastic ataxia of Charlevoix-Saguenay (ARSACS)	SACS
Bardet-Biedl syndrome[§]	ARL6, BBS1, BBS2, BBS4, BBS5, BBS7, BBS9, BBS10, BBS12, MKKS, MKS1, TTC8
Bare lymphocyte syndrome type II	CIITA
Bartter syndrome type I	SLC12A1
Bartter syndrome type II	KCNJ1
Bartter syndrome type III	CLCNKB
Bartter syndrome type IV	BSND, CLCNKA, CLCNKB
BCS1L-related disorders (Mitochondrial complex IIIdeficiency, Bjornstad syndrome, Leigh syndrome)	BCS1L
Bernard-Soulier syndrome type A1	GP1BA
Bernard-Soulier syndrome type B1	GP1BB
Bernard-Soulier syndrome type C	GP9
Beta-globin-related hemoglobinopathies[†‡§] (Beta-thalassemia, Sickle cell disease)	HBB
Beta-ketothiolase deficiency	ACAT1
Beta-mannosidosis	MANBA
Bilateral frontoparietal polymicrogyria	ADGRG1

DISORDER	GENE
Biotinidase deficiency	BTD
Bloom syndrome†‡§	BLM
Canavan disease†‡§	ASPA
Carbamoylphosphate synthetase I deficiency	CPS1
Carnitine palmitoyltransferase I deficiency	CPT1A
Carnitine palmitoyltransferase II deficiency§	CPT2
Carpenter syndrome	RAB23
Cartilage-hair hypoplasia-anauxetic dysplasia spectrum disorders	RMRP
Cerebral creatine deficiency syndrome 1	SLC6A8
Cerebrotendinous xanthomatosis§	CYP27A1
Charcot-Marie-Tooth disease	NDRG1
Chediak-Higashi syndrome	LYST
Chorea-acanthocytosis§	VPS13A
Choroidal dystrophy, central areolar 1	GUCY2D
Choroideremia	CHM
Chronic granulomatous disease§	CYBA, NCF1, NCF2
Citrin deficiency	SLC25A13
Citrullinemia type 1	ASS1
COACH syndrome	RPGRIP1L
Cockayne syndrome type A	ERCC8
Cockayne syndrome type B	ERCC6
Cohen syndrome	VPS13B
Combined malonic and methylmalonic aciduria	ACSF3
Combined oxidative phosphorylation deficiency	GFM1, TSFM
Combined pituitary hormone deficiency	LHX3, PROP1
Congenital adrenal hyperplasia due to 21-alpha-hydroxylase deficiency	CYP21A2
Congenital amegakaryocytic thrombocytopenia§	MPL
Congenital disorder of glycosylation§	ALG6, MPI, PMM2
Congenital Finnish nephrosis	NPHS1
Congenital insensitivity to pain with anhidrosis§	NTRK1
Congenital myasthenic syndrome§	CHAT, CHRNE, COLQ, DOK7, GFPT1, RAPSN
Congenital neutropenia	HAX1, VPS45
Corneal dystrophy and perceptive deafness	SLC4A11
Corticosterone methyloxidase deficiency§	CYP11B2
Crigler-Najjar syndrome	UGT1A1
Cystic fibrosis†‡§	CFTR
Cystinosis	CTNS
D-bifunctional protein deficiency	HSD17B4
Desbuquois dysplasia type I	CANT1
DHDDS-related disorders (including Congenital disorder of glycoslylation)	DHDDS
Dihydropyrimidine dehydrogenase deficiency§	DPYD, DLD
DMD-related dystrophinopathy (Duchenne / Becker muscular dystrophy, Dilated cardiomyopathy)	DMD
Dysferlinopathy	DYSF
Dyskeratosis congenita	RTEL1, TERT
Dystrophic epidermolysis bullosa	COL7A1

DISORDER	GENE
Early onset myopathy with fatal cardiomyopathy (Salih myopathy)	TTN
Ehlers-Danlos syndrome§	ADAMTS2, FKBP14, PLOD1, TNXB
Ellis-van Creveld syndrome	EVC, EVC2
Enhanced S-cone syndrome§	NR2E3
Epiphyseal dysplasia, multiple, 7	CANT1
Ethylmalonic encephalopathy	ETHE1
Factor V deficiency	F5
Factor XI deficiency§	F11
Familial dysautonomia†‡§	IKBKAP
Familial hypercholesterolemia§	LDLR, LDLRAP1
Familial hyperinsulinemic hypoglycemia	HADH, KCNJ11
Familial hyperinsulinism†§	ABCC8, KCNJ11
Familial mediterranean fever§	MEFV
Fanconi anemia†‡§	FANCA, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM
Fetal akinesia deformation sequence	DOK7
Fumarate hydratase deficiency	FH
Galactokinase deficiency galactosemia	GALK1
Galactosemia§	GALT
Gaucher disease†‡§	GBA
Geroderma osteodysplastica	GORAB
Gilbert syndrome	UGT1A1
Gitelman syndrome	SLC12A3
Glutaric acidemia type I	GCDH
Glutaric acidemia type II	ETFA, ETFB, ETFDH
Glycine encephalopathy	AMT, GLDC
Glycogen storage disease IXb	PHKB
Glycogen storage disease IXc	PHKG2
Glycogen storage disease type Ia‡§	G6PC
Glycogen storage disease type Ib	SLC37A4
Glycogen storage disease type II (Pompe disease)§	GAA
Glycogen storage disease type IV§	GBE1
Glycogen storage disease type V	PYGM
Glycogen storage disease type III	AGL
Glycogen storage disease type VII§	PFKM
Glycogen storage disease VI	PYGL
GRACILE syndrome	BCS1L
Guanidinoacetate methyltransferase deficiency (Cerebral creatine deficiency syndrome 2)	GAMT
Hemochromatosis type 1	HFE
Hemochromatosis type 2A	HFE2
Hemochromatosis type 3	TFR2
Hereditary fructose intolerance	ALDOB
Hermansky-Pudlak syndrome§	AP3B1, HPS1, HPS3, HPS4, HPS5, HPS6
Holocarboxylase synthetase deficiency	HLCS
Homocystinuria§	CBS, MTHFR, MTRR
Hydrolethalus syndrome type 1	HYLS1
Hyperornithinemia-hyperammonemia-homocitrullinuria (HHH) syndrome	SLC25A15
Hyperphospatemic tumoral calcinosis, familial	GALNT3
Hypohidrotic ectodermal dysplasia	EDA, EDAR

DISORDER	GENE
Hypophosphatasia	ALPL
Hypoprothrombinemia	F2
Inclusion body myopathy 2[§]	GNE
Infantile cerebral and cerebellar atrophy[§]	MED17
Isovaleric acidemia	IVD
Johanson-Blizzard syndrome	UBR1
Joubert syndrome[‡§]	AHI1, CEP290, CC2D2A, CPLANE1, CSPP1, INPP5E, KIAA0586, MKS1, RPGRIP1L, TMEM67, TMEM216, TMEM231
Junctional epidermolysis bullosa	LAMA3, LAMB3, LAMC2
Krabbe disease	GALC
Kufor-Rakeb syndrome	ATP13A2
LAMA2-related muscular dystrophy	LAMA2
Lamellar ichthyosis type 1	TGM1
Leber congenital amaurosis[§]	CEP290, CRB1, GUCY2D, LCA5, RDH12, RPE65
Leigh syndrome, French Canadian type	LRPPRC
Lethal arthrogryposis with anterior horn cell disease	GLE1
Lethal congenital contracture syndrome 1	GLE1
Leukoencephalopathy with vanishing white matter	EIF2B5
Leydig cell hypoplasia	LHCGR
Limb-girdle muscular dystrophy	CAPN3, DYSF, POMT1, POMT2, SGCA, SGCB, SGCG, TTN
Lipoid congenital adrenal hyperplasia	STAR
Lipoprotein lipase deficiency	LPL
Long chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency	HADHA
Lysinuric protein intolerance	SLC7A7
Lysosomal acid lipase deficiency[§] (includes Wolman disease and Cholesterol ester storage disease)	LIPA
Major histocompatibility complex class II deficiency	CIITA
Maple syrup urine disease[‡§]	BCKDHA, BCKHB, DBT
Meckel syndrome	CC2D2A, MKS1, RPGRIP1L, TMEM67, TMEM216
Medium chain acyl-CoA dehydrogenase (MCAD) deficiency	ACADM
MEDNIK syndrome	AP1S1
Megalencephalic leukoencephalopathy with subcortical cysts type 1[§]	MLC1
Megalencephalic leukoencephalopathy with subcortical cysts types 2A & 2B[§]	HEPACAM
Metachromatic leukodystrophy[§]	ARSA
Metachromatic leukodystrophy due to SAP-b deficiency	PSAP
Methylmalonic acidemia	ACSF, MMAA, MMAB, MUT
Methylmalonic acidemia with homocystinuria, cobalamin C type	MMACHC
Methylmalonic acidemia with homocystinuria, cobalamin D type	MMADHC
Microphthalmia / Clinical anophthalmia[§]	VSX2
Mitochondrial complex 1 deficiency[§]	NDUFAF6
Mitochondrial complex I deficiency / Leigh syndrome	ACAD9, NDUFAF5, NDUFS6
Mitochondrial complex IV deficiency	PET100
Mitochondrial DNA depletion syndrome	MPV17, POLG

DISORDER	GENE
Mitochondrial myopathy and sideroblastic anemia[§]	PUS1
Mitochondrial recessive ataxia syndrome	POLG
Mucolipidosis type II/III	GNPTAB
Mucolipidosis type IV[†‡§]	MCOLN1
Mucolipidosis type III	GNPTG
Mucopolysaccharidosis type I (includes Hurler, Hurler-Scheie, and Scheie syndromes)	IDUA
Mucopolysaccharidosis type IX	HYAL1
Mucopolysaccharidosis type VI (Maroteaux-Lamy syndrome)	ARSB
Mucopolysaccharidosis type IIIA (Sanfilippo A syndrome)	SGSH
Mucopolysaccharidosis type IIIB	NAGLU
Mucopolysaccharidosis type IIIC (Sanfilippo syndrome)	HGSNAT
Mucopolysaccharidosis type IIID (Sanfilippo syndrome)	GNS
Mucopolysaccharidosis type IVB (Morquio B syndrome) / GM1 gangliosidosis	GLB1
Mulibrey nanism	TRIM37
Multiple sulfatase deficiency[§]	SUMF1
Muscle-Eye-Brain disease and other POMGNT1-related congenital muscular dystrophy-dystroglycanopathies	POMGNT1
Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies)	LARGE1, POMT1, POMT2
Muscular dystrophy-dystroglycanopathy (congenital with mental retardation)	LARGE1, MKKS, POMT1, POMT2
Muscular dystrophy-dystroglycanopathy (congenital with mental retardation) type 6B	LHCGR
Myoneurogastrointestinal encephalopathy[§]	TYMP
N-Acetylglutamate synthase deficiency	NAGS
Nemaline myopathy 2	NEB
Nephrogenic diabetes insipidus	AQP2
Neu-Laxova syndrome	PHGDH
Neuronal ceroid lipofuscinosis	CLN3, CLN5, CLN6, CLN8, CTSD, CTSF, DNAJC5, GRN. KCTD7, MFSD8, PPT1, TTP1
Niemann-Pick disease type A/B[†‡§]	SMPD1
Niemann-Pick disease type C[†]	NPC1, NPC2
Nijmegen breakage syndrome	NBN
Non-syndromic hearing loss[§]	GJB2, GJB6
Normophosphatemic familial tumoral calcinosis	SAMD9
Omenn syndrome[§]	DCLRE1C, RAG1, RAG2
Ornithine aminotransferase deficiency (Gyrate atrophy)[§]	OAT
Osteopetrosis[§]	TCIRG1
Pendred syndrome	SLC26A4
Peroxisomal acyl-CoA oxidase deficiency	ACOX1
Phenylalanine hydroxylase deficiency (including Phenylketonuria (PKU))[§]	PAH
Phosphoglycerate dehydrogenase deficiency[§]	PHGDH
Polycystic kidney disease (PKHD1-related)[§]	PKHD1
Polymicrogyria	ADGRG1
Pontocerebellar hypoplasia[§]	EXOC3, RARS2, SEPSECS, TSEN2, TSEN34, TSEN54, VRK1, VPS53

DISORDER	GENE
Primary carnitine deficiency	SLC22A5
Primary ciliary dyskinesia§	DNAH5, DNAI1, DNAI2
Primary congenital glaucoma 3A	CYP1B1
Primary hyperoxaluria§	AGXT, GRHPR, HOGA1
Progressive cerebello-cerebral atrophy§	SEPSECS
Progressive external ophthalmoplegia with mitochondrial deletions autosomal recessive type 1	POLG
Progressive familial intrahepatic cholestasis type 2	ABCB11
Prolidase deficiency	PEPD
Propionic acidemia	PCCA, PCCB
Pseudocholinesterase deficiency	BCHE
Pseudoxanthoma elasticum	ABCC6, GGCX
Pycnodysostosis	CTSK
Pyridoxine-dependent epilepsy	ALDH7A1
Pyruvate carboxylase deficiency	PC
Pyruvate dehydrogenase deficiency	DLAT, PDHB
Refsum disease	PEX7
Renal tubular acidosis with deafness§	ATP6V1B1
Retinitis pigmentosa§	CERKL, DHDDS, EYS, FAM161A, HGSNAT, NR2E3, TTC8
Rhizomelic chondrodysplasia punctata	AGPS, GNPAT, PEX7
Roberts syndrome	ESCO2
Salla disease	SLC17A5
Sandhoff disease	HEXB
Schimke immuno-osseous dysplasia	SMARCAL1
Severe combined immunodeficiency	DCLRE1C, RAG1
Short chain Acyl-CoA dehydrogenase deficiency	ACADS
Shwachman-Diamond syndrome	SBDS
Sialic acid storage disorders	SLC17A5
Sjögren-Larsson syndrome	ALDH3A2
SLC26A2-related disorders (including Diatrophic dysplasia, Atelosteogenesis type 2, Achondrogenesis type 1B)	SLC26A2
Smith-Lemli-Opitz syndrome§	DHCR7
Spastic paraplegia§	ATP13A2, TECPR2, ZFYVE26
Spinal muscular atrophy†‡§	SMN1
Spinocerebellar ataxia type 7	TPP1
Spondylocostal dysostosis	DLL3, LFNG
Spondylothoracic dysostosis	DLL3, LFNG, MESP2
Steel syndrome	COL27A1
Steroid-resistant nephrotic syndrome	NPHS2
Stüve-Wiedemann syndrome	LIFR
Tay-Sachs disease (Hexosaminidase A deficiency)†‡§	HEXA
Tyrosine hydroxylase deficiency (Segawa syndrome)	TH
Tyrosinemia type I§	FAH
Tyrosinemia type II	TAT
Tyrosinemia type III	HPD
Usher syndrome‡§	ADGRV1, CDH23, CLRN1, MYO7A, PCDH15, USH1C, USH1G, USH2A, WHRN
Very long-chain acyl-CoA dehydrogenase deficiency	ACADVL
Vitamin D-dependent rickets type I	CYP27B1
Walker-Warburg syndrome§	FKRP, FKTN, IPSD
Wilson disease§	ATP7B
WNT10A-related disorders (including Odonto-onycho-dermal dysplasia and Schopf-Schulz-Passarge syndrome)	WNT10A
Woolly hair / Hypotrichosis	LIPH
Xeroderma pigmentosum	ERCC2, XPA, XPC
Zellweger spectrum disorder§	PEX1, PEX2, PEX3, PEX6, PEX10, PEX12, PEX14, PEX16, PEX26

X-linked

DISORDER	GENE
Alpha-thalassemia X-linked intellectual disability syndrome	ATRX
Alport Syndrome§	COL4A5
Androgen insensitivity syndrome	AR
Arts syndrome	PRPS1
Charcot-Marie-Tooth disease	GJB1, PRPS1
Chronic granulomatous disease	CYBB
Dyskeratosis congenita	DKC1
Emery-Dreifuss muscular dystrophy	EMD
Fabry disease	GLA
Factor IX deficiency (Hemophilia B)	F9
Factor VIII deficiency (Hemophelia A)	F8
Fragile X syndrome†‡§	FMR1
Glucose-6-phosphate dehydrogenase deficiency	G6PD
Glycogen storage disease IXa	PHKA2
Hemolytic anemia due to G6PD deficiency	G6PD
HPRT-related gout	HPRT1
L1 syndrome / MASA syndrome / CRASH syndrome	L1CAM
Lesch-Nyhan syndrome	HPRT1
Menkes disease / ATP7A-related disorders (including Occipital horn syndrome and Distal hereditary motor neuropathy)	ATP7A
Mucopolysaccharidosis type II (Hunter syndrome)	IDS
Myotubular myopathy 1	MTM1
Ornithine transcarbamylase deficiency	OTC
Pyruvate dehydrogenase deficiency	PDHA1
X-lined Fanconi anemia	FANCB
X-linked adrenoleukodystrophy§	ABCD1
X-linked ichthyosis	STS
X-linked juvenile retinoschisis	RS1
X-linked severe combined immunodeficiency (X-SCID)	IL2RG

† Recommended by American College of Medical Genetics and Genomics (ACMG)
‡ Recommended by American College of Obstetricians and Gynecologists (ACOG)
§ Recommended by Jewish Genetic Disease Consortium