GENESYS Diagnostics INC.

CAP ACCREDITED
COLLEGE of AMERICAN PATHOLOGISTS

Clinical Laboratory Improvement Amendments

GENETIC CARRIER SCREENING

Our carrier screening panel covers 145 different genes, which correspond to various disorders. This test covers all the ACMG tier 1, tier 2, and tier 3 recommended disorders*, and covers additional disorders with a frequency of 1:200.

* ACMG tiered screening details: Tier 1 (CF/SMA), Tier 2 carrier frequency ≥1:100 (includes Tier 1), Tier 3 ≥1:200 carrier frequency (includes Tier 2 and x-linked conditions).

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Condition	Gene	Tier
Neuromuscular/Muscular		
Spinal Muscular Atrophy	SMN1/SMN2	Tier 1
Spinocerebellar ataxia 10	ANO10	Tier 2
Myasthenic syndrome, congenital, 4A/4B	CHRNE	Tier 2
Niemann–Pick disease, type A/B	SMPD1	Tier 2
Ehlers–Danlos-like syndrome due to tenascin-X deficiency	TNXB	Tier 2
Adrenoleukodystrophy (ALD)	ABCD1	Tier 3
Basal ganglia disease, biotin-responsive	SLC19A3	Tier 3
Congenital myotonia, autosomal recessive form	CLCN1	Tier 3
Familial dysautonomia	ELP1 (aka IKBKAP)	Tier 3
Metachromatic leukodystrophy	ARSA	Tier 3
Muscular dystrophy–dystroglycanopathy, type A/B, 5	FKRP	Tier 3
Pontocerebellar hypoplasia type 6	RARS2	Tier 3
Friedreich ataxia	FXN	Tier 3
Nemaline myopathy 2	NEB	Tier 3
Spastic paraplegia 2, X-linked (SPG2)	PLP1	Tier 3
Ataxia-Telangiectasia	ATM	Extra
Limb-Girdle Muscular Dystrophy Type 2A	CAPN3	Extra
Duchenne Muscular Dystrophy, Becker Muscular Dystrophy	DMD	Extra
Limb-Girdle Muscular Dystrophy Type 2	DYSF	Extra
Inclusion Body Myopathy 2	GNE	Extra
Endocrinology		
Diabetes mellitus, permanent neonatal 3	ABCC8	Tier 2
Congenital adrenal hyperplasia due to 21-hydroxylase deficiency	CYP21A2	Tier 2
Autoimmune Poly endocrinopathy syndrome type I	AIRE	Tier 3
Adrenal insufficiency, congenital, with 46, XY sex reversal, partial or complete	CYP11A1	Tier 3
Adrenal hypoplasia, congenital (AHC)	NR0B1	Tier 3
Neurodevelopmental		
Joubert syndrome 3	AHI1	Tier 2
Canavan disease	ASPA	Tier 2
Joubert syndrome 5	CEP290	Tier 2
Tay–Sachs disease	HEXA	Tier 2
Schindler disease, type 1 or type 3	NAGA	Tier 2
Smith–Lemli–Opitz syndrome	DHCR7	Tier 2
Mental retardation, X-linked, associated with fragile site FRAXE	AFF2	Tier 3
Aspartylglucosaminuria	AGA	Tier 3
Joubert syndrome 9	CC2D2A	Tier 3
Fragile X syndrome (FXS)	FMR1	Tier 3
Joubert syndrome 2	TMEM216	Tier 3
Dermatology		
Recessive dystrophic epidermolysis bullosa	COL7A1	Tier 2
Xeroderma pigmentosum	XPC	Tier 2
Bloom syndrome	BLM	Tier 3
Hermansky Pudlak S. 1	HPS1	Tier 3
Hermansky Pudlak S. 3	HPS3	Tier 3
Cardiology		
Carnitine palmitoyltransferase II deficiency, infantile/lethal neonatal	CPT2	Tier 2
Walker–Warburg congenital muscular dystrophy, Dilated Cardiomyopathy	FKTN	Tier 2
Alstrom Syndrome	ALMS1	Extra
Carnitine Deficiency, Primary	SLC22A5	Extra
Hypercholesterolemia, familial, 1	LDLR	Extra
Ear, Nose, and Throat (ENT)		
Nonsyndromic hearing loss	GJB2	Tier 2
Pendred syndrome	SLC26A4	Tier 2
Usher Syndrome Type 1D, Autosomal Recessive Deafness	CDH23	Extra
Deafness, autosomal recessive 77	LOXHD1	Extra
Nephrology		
Finnish congenital nephrotic syndrome	NPHS1	Tier 2
Hyperoxaluria, primary type I	AGXT	Tier 3
Autosomal recessive polycystic kidney disease	PKHD1	Tier 3
Nephrotic Syndrome Type 2	NPHS2	Extra

Condition	Gene	Tier
Nephrology Continued		
Gitelman syndrome	SLC12A3	Extra
Hematology		
Fanconi anemia, complementation group C	FANCC	Tier 2
Sickle cell anemia, b-thalassemia, HbC Disease, HbD Disease, HbO disease	HBB	Tier 2
Atransferrinemia	TF	Tier 3
Hemophilia A (HEMA)	F8	Tier 3
Hemophilia B (HEMB)	F9	Tier 3
α-Thalassemia	HBA1	Tier 3
α-Thalassemia	HBA2	Tier 3
Fanconi anemia, complementation group A	FANCA	Extra
Fanconi anemia, complementation group G	FANCG	Extra
Factor XI Deficiency	F11	Extra
Hemochromatosis	HFE	Extra
Ophthalmology		
Oculocutaneous albinism brown and type II	OCA2	Tier 2
Oculocutaneous albinism type 1A/1B	TYR	Tier 2
Usher syndrome, type 2A	USH2A	Tier 2
Congenital disorder of glycosylation type 1, Retinitis pigmentosa 59	DHDDS	Tier 3
Usher syndrome 3a	CLRN1	Tier 3
Achromatopsia 3	CNGB3	Tier 3
Retinitis pigmentosa 3 (RP3; RP)	RPGR	Tier 3
Retinoschisis 1, X-linked, juvenile (RS1)	RS1	Tier 3
Usher syndrome, type 1F	PCDH15	Tier 3
Glaucoma, Primary Congenital	CYP1B1	Extra
Retinitis pigmentosa 25	EYS	Extra
Usher syndrome, type 1B	MYO7A	Extra
Metabolic Disorders		
Glycogen storage disease type IA	G6PC	Tier 2
Glycogen storage disease, type II (Pompe disease)	GAA	Tier 2
Glycogen storage disease, type IV	GBE1	Tier 2
Methylmalonic aciduria–methyl malonyl–CoA mutase deficiency	MMUT	Tier 2
Medium-chain acyl-coenzyme A dehydrogenase deficiency	ACADM	Tier 2
Maple Syrup Urine Disease Type 1B	BCKDHB	Tier 2
Phenylketonuria	PAH	Tier 2
Carbohydrate-deficient glycoprotein syndrome type Ia	PMM2	Tier 2
Argininosuccinate aciduria	ASL	Tier 3
Very long chain acyl-CoA dehydrogenase deficiency	ACADVL	Tier 3
α-Methylacetoacetic aciduria	ACAT1	Tier 3
Biotinidase deficiency	BTD	Tier 3
Homocystinuria, B6 responsive and nonresponsive	CBS	Tier 3
Cerebro tendinous xanthomatosis	CYP27A1	Tier 3
Dihydrolipoamide dehydrogenase deficiency	DLD	Tier 3
Tyrosinemia type I	FAH	Tier 3
Trimethylaminuria	FMO3	Tier 3
Galactosemia	GALT	Tier 3
Gaucher disease, type I/II	GBA	Tier 3
Fabry disease	GLA	Tier 3
Mucolipidosis type II/III alpha/beta	GNPTAB	Tier 3
Mucopolysaccharidosis, Ih (Hurler S)	IDUA	Tier 3
Mucolipidosis type IV	MCOLN1	Tier 3
Methylmalonic aciduria with homocystinuria cblC type	MMACHC	Tier 3
Hyper-IgD syndrome, Mevalonic aciduria	MVK	Tier 3
Ornithine transcarbamylase deficiency	OTC	Tier 3
3-methylcrotonyl CoA carboxylase 2 deficiency	MCCC2	Tier 3
Mitochondrial DNA depletion syndrome 4A/4B	POLG	Tier 3
Mitochondrial complex IV deficiency, nuclear type 2	SCO2	Tier 3
Glycogen storage disease Ib/Ic	SLC37A4	Tier 3
Cerebral creatine deficiency syndrome 1 (CCDS1)	SLC6A8	Tier 3
Short Chain Acyl-CoA Dehydrogenase Deficiency	ACADS	Extra
Combined Malonic and Methylmalonic Aciduria	ACSF3	Extra

Condition	Gene	Tier
Metabolic Disorders Continued		
Krabbe Disease	GALC	Extra
Hyperoxaluria, primary, type III	HOGA1	Extra
Niemann-Pick Disease Type C1	NPC1	Extra
Niemann-Pick Disease Type C2	NPC2	Extra
Peroxisome biogenesis disorder 4A/4B (Zellweger Syndrome)	PEX6	Extra
Glycogen Storage Disease Type V (McArdle)	PYGM	Extra
Maple Syrup Urine Disease Type 2	DBT	ACOG
Pulmonary		
Cystic Fibrosis	CFTR	Tier 1
Surfactant metabolism dysfunction, pulmonary 3	ABCA3	Tier 3
Ciliary dyskinesia	DNAH5	Extra
Alpha-1 Antitrypsin Deficiency	SERPINA1	Extra
Non-specific or multiple specialties. Including but not limited to: Neuromuscular, Endocrinology, Neurodevelopmental, Dermatology, Cardiology, ENT, Nephrology, Hematology, Ophthalmology, and Metabolic Disorders.		
Wilson disease	ATP7B	Tier 2
Short-rib thoracic dysplasia 3 with or without polydactyly	DYNC2H1	Tier 2
Cerebrooculofacioskeletal syndrome 2, Trichothiodystrophy 1, photosensitive	ERCC2	Tier 2
Fraser syndrome	GRIP1	Tier 2
Hypophosphatasia, adult, childhood and infantile	ALPL	Tier 3
Developmental and epileptic encephalopathy 1 (DEE1)	ARX	Tier 3
Bardet–Biedl syndrome 1	BBS1	Tier 3
Bardet–Biedl syndrome 2	BBS2	Tier 3
Congenital hydrocephalus 1	CCDC88C	Tier 3
Vitamin D–dependent rickets, type 1	CYP27B1	Tier 3
Chondroectodermal dysplasia	EVC2	Tier 3
Hydrocephalus due to congenital stenosis of aqueduct of Sylvius (HSAS)	L1CAM	Tier 3
Donnai–Barrow syndrome	LRP2	Tier 3
Primary microcephaly 1, recessive	MCPH1	Tier 3
Opitz GBBB syndrome, type I (GBBB1)	MID1	Tier 3
Megalencephalic leukoencephalopathy with subcortical cysts	MLC1	Tier 3
Hemophagocytic lymphohistiocytosis, familial, 2	PRF1	Tier 3
Aicardi Goutieres syndrome 2	RNASEH2B	Tier 3
Epiphyseal dysplasia, multiple, 4; Achondrogenesis Ib	SLC26A2	Tier 3
Familial Mediterranean Fever	MEFV	Extra
Anauxetic dysplasia 1, Cartilage-hair hypoplasia	RMRP	Extra