



**EPILEPSY,
SEIZURES, & OTHER
NEUROMUSCULAR
CONDITIONS**

Dravet Syndrome	SCN1A
Early Infantile epileptic encephalopathy 7	KCNQ2
Early infantile epileptic encephalopathy 11 / Benign familial infantile seizures 3	SCN2A
Early infantile epileptic encephalopathy 13 / Benign familial infantile seizures 5	SCN8A
Ethylmalonic Encephalopathy	ETHE1
Familial Infantile Convulsions with Paroxysmal Choreoathetosis	PRRT2
Pyridoxine-Dependent Epilepsy	ALDH7A1
Pyridoxal 5'-Phosphate-Dependent Epilepsy	PNPO
Tuberous Sclerosis Complex (TSC)	TSC1, TSC2

**HEARING &
VISION LOSS**

Clouston syndrome	GJB6
Deafness, Autosomal Dominant, 12	TECTA
Deafness, Autosomal Recessive, 6 (DFNB6)	TMIE
Deafness, Autosomal Recessive, 8 (DFNB8)	TMPRSS3
Deafness, Autosomal Recessive, 28	TRIOBP
Deafness, Autosomal Recessive, 31 (DFNB31)	WHRN
Deafness, Autosomal Recessive, 79	TPRN
GJB2-Related Hearing Loss	GJB2
Hermansky-Pudlak syndrome	HPS4
Hermansky-Pudlak Syndrome, Type 1	HPS1
Jervell and Lange-Nielson Syndrome	KCNE1
Nonsyndromic Hearing Loss	CDH23
Ocular Albinism Type I	GPR143
Oculocutaneous Albinism Type IV	SLC45A2
Optic Atrophy Type 1	OPA1
Ornithine Aminotransferase Deficiency	OAT
Pendred Syndrome	SLC26A4
Sensorineural Hearing Loss	MYO15A
Shah-Waardenburg Syndrome	SOX10
Usher syndrome type 1C	USH1C
Usher Syndrome, Type ID	CDH23
Usher syndrome type 1G	USH1G
Usher Syndrome, Type IIA	USH2A
Usher syndrome type IID	WHRN
Waardenburg Syndrome	PAX3

**HEART
CONDITIONS**

Alagille Syndrome 1 / Tetralogy of Fallot	JAG1
Arrhythmogenic Right Ventricular Cardiomyopathy	DSC2
Arrhythmogenic Right Ventricular Cardiomyopathy	DSP
Arrhythmogenic right ventricular cardiomyopathy, type 5	TMEM43
Arrhythmogenic right ventricular dysplasia, type 11	DSC2
Barth Syndrome (3-Methylglutaconic Aciduria Type II)	TAZ
Brugada Syndrome	KCNQ1
Duchenne Muscular Dystrophy	DMD
Hereditary hemorrhagic telangiectasia type 1	ENG
Hereditary hemorrhagic telangiectasia type 2	ACVRL1
Heterotaxy	ZIC3
Hypercholesterolemia	LDLR
Hypertrophic, Dilated Cardiomyopathy	ACTC1, LMNA, MYH7, PRKAG2
Fabry Disease	GLA
Familial Mediterranean Fever	MEFV
Loeys-Dietz Syndrome, Type 1	TGFBR1
Loeys-Dietz Syndrome, Type 2	TGFBR2
Loeys-Dietz syndrome, Type 3	SMAD3
Loeys-Dietz Syndromes, & Familial Thoracic Aortic Aneurysms and Dissections	FBN1, MYH11
Long QT Syndrome Types 1, 2 & 3	KCNH2
Marfan Syndrome	FBN1
Romano-Ward Syndrome	SCN5A
Vascular Ehlers-Danlos Syndrome (VEDS)	COL3A1

**HEREDITARY
CHILDHOOD
CANCERS**

Congenital Insensitivity to Pain with Anhidrosis	NTRK1, RET
Hereditary Retinoblastoma	RB1
JAK3-Related Severe combined immunodeficiency	JAK3
Juvenile Polyposis Syndrome	BMPR1A, SMAD4
Multiple endocrine neoplasia Type 1	MEN1
Multiple Endocrine Neoplasia Type 2	RET
Neurofibromatosis Type 1	NF1
Neurofibromatosis Type 2	NF2
Nevoid Basal Cell Carcinoma Syndrome	PTCH1
Hereditary Paraganglioma- Pheochromocytoma Syndrome, Type 1	SDHD
Hereditary Paraganglioma- Pheochromocytoma Syndrome, Type 4	SDHB
Peutz-Jeghers Syndrome (PJS)	STK11
Von Hippel-Lindau Syndrome (VHL)	VHL
WT1-Related Wilms Tumor	WT1
Xeroderma Pigmentosum	ERCC2, ERCC5, XPA, XPC



**OVER 270 GENES
GENETIC INSIGHTS PANEL**

**IMMUNO-
DEFICIENCIES**

Agammaglobulinemia (X-linked)	BTK
Bare lymphocyte syndrome	RFX5
Bare lymphocyte syndrome I	RFXANK
Bare lymphocyte syndrome II	RFXAP
Chronic Granulomatous Disease	CYBA, CYBB, NCF2
Hyper-IgE Syndrome	CD40LG, DOCK8
Hyper IgM syndrome	CD40LG
Hyper-IgM Syndrome (X-linked)	MPL
Factor IX deficiency; Hemophilia B	F9
Severe Combined Immunodeficiency; Omenn Syndrome, Athabaskan-Type	ADA, ANK1, CD3E, CD3D, DCLRE1C, EPB42, IL2RG, IL7R, PTPRC RAG1, RAG2, SLC4A1, SPTB

**METABOLIC
DISORDERS**

3-beta-hydroxysteroid dehydrogenase deficiency	HSD3B2
3-hydroxy-3-methylglutaryl-CoA lyase deficiency; HMG-CoA Lyase Deficiency	HMGCL
3-hydroxyacyl-CoA dehydrogenase deficiency	HADH
3-Methylcrotonyl-CoA carboxylase 1 deficiency	MCCC1
3-Methylcrotonyl-CoA carboxylase 2 deficiency	MCCC2
3-phosphoglycerate dehydrogenase deficiency	PHGDH
6-Pyruvoyl-Tetrahydropterin Synthase Deficiencies	PTS
Abetalipoproteinemia	MTTP
Acrodermatitis enteropathica, zinc-deficiency type (AEZ)	SLC39A4
Acute Infantile Liver Failure	TRMU
Alport Syndrome	COL4A3, COL4A4, COL4A5
Arginase Deficiency (Argininemia)	ARG1
Arginine:glycine amidinotransferase deficiency	GATM
Argininosuccinic Aciduria	ASL
Ataxia with Isolated Vitamin E Deficiency	TTPA
Autosomal Dominant Polycystic kidney disease, Type 2	PKD2
Autosomal Recessive Polycystic Kidney Disease	PKHD1
Beta-Ketothiolase Deficiency (Alpha-Methylacetoacetic Aciduria)	ACAT1
BH4-deficient hyperphenylalaninemia B (Dopa-responsive dystonia)	GCH1
BH4-deficient hyperphenylalaninemia C (Dihydropteridine reductase deficiency)	QDPR
BH4-deficient hyperphenylalaninemia D (Tetrahydrobiopterin deficiency)	PCBD1
Biotinidase deficiency	BTD
Carbamoylphosphate Synthetase I Deficiency	CPS1
Carnitine Acylcarnitine Translocase Deficiency	SLC25A20
Carnitine Palmitoyltransferase I Deficiency	CPT1A
Carnitine Palmitoyltransferase II Deficiency	CPT2
Central hypothyroidism and testicular enlargement	IGSF1
Cerebral creatine deficiency syndromes	GAMT
Citrullinemia, Type 1	ASS1
Citrullinemia, Type 2; Citrin deficiency	SLC25A13
Combined Pituitary Hormone Deficiency 1	POU1F1
Combined Pituitary Hormone Deficiency 2	PROP1
Combined Pituitary Hormone Deficiency 3	LHX3
Congenital adrenal hyperplasia; 11-beta-hydroxylase deficiency	CYP11B1
Congenital Adrenal Hyperplasia; 17-Alpha-Hydroxylase Deficiency	CYP17A1
Congenital adrenal hypoplasia, X-Linked	NR0B1
Congenital Disorder of Glycosylation Type 1b	MPI
Congenital hyperinsulinism	HNF4A
Congenital hypothyroidism 1, nongoitrous / Nonautoimmune hyperthyroidism	TSHR
Congenital hypothyroidism 6, nongoitrous	THRA
Congenital hypothyroidism due to thyroid dysgenesis or hypoplasia	PAXB
Congenital nongoitrous Hypothyroidism 4	TSHB
Corticosterone Methyloxidase Deficiency	CYP11B2
Crigler-Najjar Syndrome, Types 1 & 2 (Gilbert Syndrome)	UGT1A1
Cystic Fibrosis	CFTR
Cystinosis	CTNS
Cytochrome P450 oxidoreductase deficiency	POR
Dihydroipoamide dehydrogenase deficiency	DLD
Familial hyperinsulinism (ABCC8-related); Congenital hyperinsulinism	ABCC8
Familial hyperinsulinism (KCNJ11-related); Congenital hyperinsulinism	KCNJ11
Fructose-1,6-bisphosphatase deficiency	FBP1



OVER 270 GENES
GENETIC INSIGHTS PANEL

**METABOLIC
DISORDERS
CONTINUED**

Galactokinase deficiency with cataracts	GALK1
Galactose epimerase deficiency; Galactosemia	GALE
Galactosemia	GALT
Generalized thyrotropin-releasing hormone resistance	TRHR
Generalized thyrotropin-releasing hormone resistance	TRHR
GLUT1 deficiency syndrome; Glucose transporter 1 deficiency syndrome and other SLC2A1-related disorders	SLC2A1
Glutaric Acidemia, Type I	GCDH
Glutaric Acidemia, Type IIa	ETFA
Glutaric acidemia, type IIb	ETFB
Glutaric Acidemia, Type IIc	ETFDH
Glutathione synthetase deficiency	GSS
Glycogen storage disease type Ib	SLC37A4
Glycogen storage disease type II (pompe disease)	GAA
Glycogen storage disease type IIIa	AGL
Glycogen storage disease type IXb	PHKB
Glycogen storage disease type VI	PYGL
Glycogen storage disease, type 0; Hypoglycemia with deficiency of glycogen synthetase	GYS2
Glycogen storage disease, type Ia; Von Gierke's disease	G6PC
Hereditary Fructose Intolerance	ALDOB
HMG-CoA Synthase 2 Deficiency	MLYCD, HMGCS2
Holocarboxylase Synthetase Deficiency	HLCS
Homocystinuria due to cystathionine (CBS-Related) beta-synthase deficiency	CBS
Homocystinuria-megaloblastic anemia, cobalamin G type	MTR
Homocystinuria, Cobalamin E Type	MTRR
Hyperinsulinism-hyperammonemia syndrome; Congenital hyperinsulinism with hyperammonemia;	GLUD1
Hyperornithinemia-hyperammonemia-homocitrullinuria syndrome	SLC25A15
Hyperornithinemia-Hyperammonemia-Homocitrullinuria Syndrome	SLC35A15
Hyperparathyroidism (Neonatal) Hypocalcemia (Autosomal Dominant)	CASR
Hypophosphatasia	ALPL
Hypophosphatemic rickets with hypercalciuria	SLC34A3
Isobutyryl-CoA Dehydrogenase Deficiency	ACAD8, ACAD10
Isovaleric Acidemia	IVD
Krabbe Disease	GALC
Leigh syndrome	SURF1
Liddle syndrome	SCNN1B
Lipoid Adrenal Hyperplasia, Congenital	STAR
Lipoprotein Lipase Deficiency (LPL)	LPL
Long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency; Mitochondrial trifunctional protein deficiency	HADHA
Lysinuric Protein Intolerance	SLC7A7
Lysosomal Acid Lipase Deficiency (Wolman Disease)	LIPA
Maple Syrup Urine Disease, Type Ia	BCKDHA
Maple Syrup Urine Disease, Type Ib	BCKDHB
Maple syrup urine disease, Type II	DBT
Medium chain acyl CoA dehydrogenase deficiency	ACADM
Menkes Syndrome	ATP7A
Metachromatic Leukodystrophy	ARSA
Methionine Adenosyl Transferase Deficiency	MAT1A
Methylmalonic Acidemia	MMAA, MMAB
Methylmalonic Acidemia; methylmalonyl CoA mutase Deficiency	MUT
Methylmalonic acidemia; Methylmalonyl-CoA epimerase deficiency	MCEE
Methylmalonic acidemia; Methylmalonyl-CoA epimerase deficiency	MCEE
Methylmalonic aciduria and homocystinuria	LMBRD1
Methylmalonic aciduria and homocystinuria	LMBRD1
Methylmalonic aciduria and homocystinuria, cobalamin C type	MMACHC
Methylmalonic aciduria and homocystinuria, cobalamin D type	MMADHC
Mitochondrial Trifunctional Protein Deficiency	HADHB
Mucopolysaccharidosis Type I (Hurler Syndrome)	IDUA
Mucopolysaccharidosis Type II (Hunter Syndrome)	IDS
Mucopolysaccharidosis Type IVA (Morquio A)	GALNS
Mucopolysaccharidosis Type IVb / GM1 Gangliosidosis	GLB1
Mucopolysaccharidosis Type VI (Maroteaux-Lamy Syndrome)	ARSB
Mucopolysaccharidosis Type VII (Sly Syndrome)	GUSB
N-acetyl Glutamate Synthase Deficiency	NAGS
Nephrogenic diabetes insipidus	AQP2
Nephrogenic diabetes insipidus (AVPR2-related)	AVPR2
Neurodegeneration due to Cerebral Folate Transport Deficiency	FOLR1
Niemann-Pick Disease Type C1	NPC1
Niemann-Pick Disease Types A&B	SMPD1
Ornithine transcarboxylase Deficiency (OTC)	OTC
Permanent Neonatal Diabetes Mellitus (INS-Related)	INS

**OVER 270 GENES
GENETIC INSIGHTS PANEL**

**METABOLIC
DISORDERS
CONTINUED**

Phenylketonuria (PKU); Phenylalanine Hydroxylase Deficiency	PAH
Primary Carnitine Deficiency (Systematic)	SLC22A5
Primary hyperoxaluria, Type 1	AGXT
Primary hyperoxaluria, Type 2	GRHPR
Primary Hyperoxaluria, Type 3	HOGA1
Propionic Acidemia	PCCA, PCCB
Pseudohypoaldosteronism	SCNN1A
Pyruvate Kinase Deficiency	PKLR
Sepiapterin Reductase Deficiency	SPR
Thyroid dyshormonogenesis, Type 1	SLCSA5
Thyroid dyshormonogenesis, Type 2A	TPO
Thyroid dyshormonogenesis, Type 3	TG
Thyroid dyshormonogenesis, Type 4	IYD
Thyroid dyshormonogenesis, Type 5	DUOXA2
Thyroid dyshormonogenesis, Type 6	DUOX2
Transcobalamin Deficiency	TCN2
Tyrosine Hydroxylase Deficiency, Segawa Syndrome	TH
Tyrosinemia, type I	FAH
Tyrosinemia, type II	TAT
Tyrosinemia, type III	HPD
Vitamin D-dependent rickets	VDR
Vitamin D-dependent rickets type I	CYP27B1
VLCAD deficiency; Very long chain acyl-CoA dehydrogenase deficiency	ACADVL

**OTHER
CONDITIONS**

Autosomal Dominant Polycystic kidney disease, Type 2	PKD2
Autosomal Recessive Polycystic Kidney Disease	PKHD1
Bare lymphocyte syndrome type II	CIITA
Biotinidase deficiency	BTD
Cerebrotendinous xanthomatosis	CYP27A1
Congenital bile acid synthesis defect type 1	HSD3B7
Congenital bile acid synthesis defect type 2	AKR1D1
Congenital neutropenia	ELANE, HAX1
Craniometaphyseal dysplasia	ANKH
Crisponi syndrome	CRLF1
Familial glucocorticoid deficiency	MC2R
Familial hemophagocytic lymphohistiocytosis	PRF1
Familial Mediterranean Fever Failure	MEFV
Hemolytic anemia (G6PD-related)	G6PD
Hermansky-Pudlak Syndrome 1	HPS1
Hermansky-Pudlak Syndrome 4	HPS4
Hypophosphatemic Rickets with Hypercalciuria	SLC34A3
Immune dysregulation, polyendocrinopathy, enteropathy	FOXP3
Malignant Hyperthermia Susceptibility	RYR1
Osteogenesis imperfecta (OI) type 2	COL1A1
Osteogenesis imperfecta (OI) type 3	COL1A2
Osteopetrosis	TCIRG1
Rotor syndrome	SLCO1B1
Smith-Lemli-Opitz syndrome	DHCR7
Thrombotic thrombocytopenic purpura	ADAMTS13
Thiopurine S-methyltransferase deficiency	TMPT
ZAP70-related severe combined immunodeficiency	ZAP70

Questions or Ready to Order
The Genetic Insights Panel?
**We're here to help.
Call 866-565-2237**