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GENETIC 280 GENES INSIGHTS PANEL

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
	Familiar 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

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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 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
Adrenoleukoystrophy, X-linked	ABCD1
Alpha-Thalassemia	HBA1, HBA2
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 hypothryoidism 1, nongoitrous / Nonautoimmune hyperthyroidism	TSHR
Congenital hypothryoidism 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
Dihydrolipoamide dehydrogenase deficiency	DLD
Familial hyperinsulinism (ABCC8-related); Congenital hyperinsulinism	ABCC8
Familial hyperinsulinism (KCNJ11-related); Congenital hyperinsulinism	KCNJ11
Fructose-1,6-bisphosphatase deficiency	FBP1

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METABOLIC DISORDERS

Galactokinase deficiency with cataracts	GALK1
Galactose epimerase deficiency; Galactosemia	GALE
Galactosemia	GALT
Gaucher disease	GBA
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 trancarboxylase Deficiency (OTC)	OTC
Permanent Neonatal Diabetes Mellitus (INS-Related)	INS

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METABOLIC DISORDERS

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	SLC5A5
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
Beta-thalassemia; Sickle-Cell Anemia	HBB
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
Disorder due cytochrome p450 CYP2D6 variant	CYP2D6
Essential hypertension	CYP3A5
Familial glucocorticoid deficiency	MC2R
Familial hemophagocytic lymphohistiocytosis	PRF1
Familiar 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 Insight Panel?
We're here to help.
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