

# Genetic Insights Panel

# 290+ GENES

ASSOCIATED WITH ACTIONABLE,  
CHILDHOOD-ONSET CONDITIONS

## EPILEPSY, SEIZURES, & OTHER NEUROMUSCULAR CONDITIONS

|                                                                                    |            |
|------------------------------------------------------------------------------------|------------|
| ATAXIA WITH ISOLATED VITAMIN E DEFICIENCY                                          | TTPA       |
| CACNAIS - RELATED DISORDERS                                                        | CACNAIS    |
| 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                  | HP54    |
| HERMANSKY-PUDLAK SYNDROME, TYPE 1          | HP51    |
| JERVELL AND LANGE-NIELSEN SYNDROME         | KCNIE1  |
| 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 |
| RPE65-RELATED RETINOPATHY                  | RPE65   |
| SENSORINEURAL HEARING LOSS                 | MYO15A  |
| SHAH-WAARDENBURG SYNDROME                  | SOX10   |
| USHER SYNDROME TYPE 1C                     | USH1G   |
| 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, DSP, PKP2           |
| ARRHYTHMOGENIC RIGHT VENTRICULAR CARDIOMYOPATHY, TYPE 5                                               | TMEM43                    |
| ARRHYTHMOGENIC RIGHT VENTRICULAR DYSPLASIA, TYPE 11                                                   | DSC2, DSG2                |
| BARTH SYNDROME (3-METHYLGUTACONIC ACIDURIA TYPE II)                                                   | TAZ                       |
| BRUGADA SYNDROME                                                                                      | KCNQ1                     |
| CATECHOLAMINERGIC POLYMORPHIC VENTRICULAR TACHYCARDIA                                                 | CASQ2                     |
| CATECHOLAMINERGIC POLYMORPHIC VENTRICULAR TACHYCARDIA; ARRHYTHMOGENIC RIGHT VENTRICULAR DYSPLASIA     | RYR2                      |
| DILATED CARDIOMYOPATHY                                                                                | BAG3, DES, RBM20, TNNI3   |
| DUCHENNE MUSCULAR DYSTROPHY                                                                           | DMD                       |
| FABRY DISEASE                                                                                         | GLA                       |
| FAMILIAL THORACIC AORTIC ANEURYSM; MULTISYSTEMIC SMOOTH MUSCLE DYSFUNCTION SYNDROME; MOYAMOYA DISEASE | ACTA2                     |
| FAMILIAR MEDITERRANEAN FEVER                                                                          | MEFV                      |
| HEREDITARY HEMORRHAGIC TELANGIECTASIA TYPE 1                                                          | ENG                       |
| HEREDITARY HEMORRHAGIC TELANGIECTASIA TYPE 2                                                          | ACVRL1                    |
| HETEROTAXY                                                                                            | ZIC3                      |
| HYPERCHOLESTEROLEMIA                                                                                  | LDLR                      |
| HYPERTROPHIC CARDIOMYOPATHY                                                                           | MYL2, MYL3, TNNI3         |
| HYPERTROPHIC CARDIOMYOPATHY; DILATED CARDIOMYOPATHY, LEFT VENTRICULAR NONCOMPACTION                   | MYBPC3, TPM1              |
| HYPERTROPHIC, DILATED CARDIOMYOPATHY                                                                  | ACTC1, LMNA, MYH7, PRKAG2 |
| LOEYS-DIETZ SYNDROME, TYPE 1                                                                          | TGFBRI                    |
| 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 14; VENTRICULAR TACHYCARDIA, CATECHOLAMINERGIC POLYMORPHIC, 4                        | CALM1                     |
| LONG QT SYNDROME 15                                                                                   | CALM2                     |
| LONG QT SYNDROME 16                                                                                   | CALM3                     |
| LONG QT SYNDROME TYPES 1, 2 & 3                                                                       | KCNH2                     |
| LONG QT SYNDROME; CATECHOLAMINERGIC POLYMORPHIC VENTRICULAR TACHYCARDIA                               | TRDN                      |
| MARFAN SYNDROME                                                                                       | FBN1                      |
| ROMANO-WARD SYNDROME                                                                                  | SCN5A                     |
| TNNT2 - RELATED DISORDERS                                                                             | TNNT2                     |
| TTN - RELATED CARDIOMYOPATHY                                                                          | TTN                       |
| VASCULAR EHLERS-DANLOS SYNDROME (VEDS)                                                                | COL3A1                    |

## HEREDITARY CHILDHOOD CANCERS

|                                                             |                        |
|-------------------------------------------------------------|------------------------|
| APC-ASSOCIATED POLYPOSIS CONDITIONS                         | APC                    |
| CONGENITAL INSENSITIVITY TO PAIN WITH ANHIDROSIS            | NTRK1, RET             |
| HEREDITARY RETINOBLASTOMA                                   | RB1                    |
| JAK3-RELATED SEVERE COMBINED IMMUNODEFICIENCY               | JAK3                   |
| JUVENILE POLYPOSIS SYNDROME                                 | BMPRIA, SMAD4          |
| LI-FRAUMENI SYNDROME                                        | TP53                   |
| MULTIPLE ENDOCRINE NEOPLASIA TYPE 1                         | BMPRIA, SMAD4, 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 |

## IMMUNODEFICIENCIES

|                                                                   |                                                                                    |
|-------------------------------------------------------------------|------------------------------------------------------------------------------------|
| AGAMMAGLOBULINEMIA (X-LINKED)                                     | BTK                                                                                |
| BARE LYMPHOCYTE SYNDROME                                          | RFK5                                                                               |
| 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-HYDROXYMETHYLGLUTARIC DEHYDROGENASE DEFICIENCY                                                   | HSD3B2                 |
| 3-HYDROXY-3-METHYLTETRAHYL-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                   |
| ALPORT SYNDROME                                                                                         | COL4A3, COL4A4, COL4A5 |
| ARGINASE DEFICIENCY (ARGININEMIA)                                                                       | ARG1                   |
| ARGININE:GLYCINE AMIDINOTRANSFERASE DEFICIENCY                                                          | GATM                   |
| ARGININOSUCCINIC ACIDURIA                                                                               | ASL                    |
| 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)                                        | GCHI                   |
| BH4-DEFICIENT HYPERPHENYLALANINEMIA C (DIHYDROPTERIN REDUCTASE DEFICIENCY)                              | QDPR                   |
| BH4-DEFICIENT HYPERPHENYLALANINEMIA D (TETRAHYDROBIPTERIN DEFICIENCY)                                   | PCBD1                  |
| BIOTINIDASE DEFICIENCY                                                                                  | BTID                   |
| 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                                                                 | PROPI                  |
| 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                                                                 | NROB1                  |
| 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                    |
| DIHYDROLIPOAMIDE DEHYDROGENASE DEFICIENCY                                                               | DLID                   |
| FAMILIAL HYPERCHOLESTEROLEMIA                                                                           | PCSK9                  |
| FAMILIAL HYPERINSULINISM (ABCC8-RELATED); CONGENITAL HYPERINSULINISM                                    | ABCC8                  |
| FAMILIAL HYPERINSULINISM (KCNJ11-RELATED); CONGENITAL HYPERINSULINISM                                   | KCNJ11                 |
| FAMILIAL HYPERCHOLESTEROLEMIA; HYPOBETALIPOPROTEINEMIA                                                  | APOB                   |
| FRUCTOSE-1,6-BISPHOSPHATASE DEFICIENCY                                                                  | FBP1                   |
| GALACTOKINASE DEFICIENCY WITH CATARACTS                                                                 | GALK1                  |
| GALACTOSE EPIMERASE DEFICIENCY; GALACTOSEMIA                                                            | GALE                   |
| GALACTOSEMIA                                                                                            | GALT                   |
| GENERALIZED THYROTROPIN-RELEASING HORMONE RESISTANCE                                                    | TRHR                   |
| GLUTI DEFICIENCY SYNDROME; GLUCOSE TRANSPORTER 1 DEFICIENCY SYNDROME AND OTHER SLC2A1-RELATED DISORDERS | SLC2A1                 |
| GLUTARIC ACIDEMIA, TYPE I                                                                               | GCDF                   |
| 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                   | GYSD                   |
| GLYCOGEN STORAGE DISEASE, TYPE IA; VON GIERKE'S DISEASE                                                 | G6PC                   |
| HEREDITARY FRUCTOSE INTOLERANCE                                                                         | ALDOB                  |
| HMG-COA SYNTHASE 2 DEFICIENCY                                                                           | MLYCD, HMGCS2          |
| HOMOCARBOXYLASE SYNTHETASE DEFICIENCY                                                                   | HLCS                   |
| HOMOCYSTINURIA DUE TO CYSTATHIONINE (CBS-RELATED) BETA-SYNTHASE DEFICIENCY                              | CBS                    |
| HOMOCYSTINURIA, COBALAMIN E TYPE                                                                        | MTRR                   |
| HOMOCYSTINURIA-MEGALOBlastic ANEMIA, COBALAMIN G TYPE                                                   | MTR                    |
| HYPERINSULINISM-HYPERAMMONEMIA SYNDROME; CONGENITAL HYPERINSULINISM WITH HYPER-AMMONEMIA;               | GLUD1                  |
| HYPERORNITHINEMIA-HYPERAMMONEMIA-HOMOCITRULLINURIA SYNDROME                                             | SLC25A15, SLC35A15     |
| HYPERPARATHYROIDISM (NEONATAL) HYPOCALCEMIA (AUTOSOMAL DOMINANT)                                        | ASPL                   |
| HYPOPHOSPHATASIA                                                                                        | CALR                   |
| HYPOPHOSPHATEMIC RICKETS WITH HYPERCALCIURIA                                                            | SLC34A3                |
| ISOBUTYRYL-COA DEHYDROGENASE DEFICIENCY                                                                 | ACAD8                  |
| 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                    |
| MATURITY-ONSET OF DIABETES OF THE YOUNG                                                                 | HNFA                   |
| MEDIUM CHAIN ACYL COA DEHYDROGENASE DEFICIENCY                                                          | ACADM                  |
| MENKES SYNDROME                                                                                         | ATP7A                  |
| METACHROMATIC LEUKODYSTROPHY                                                                            | ARSA                   |
| METHIONINE ADENOSYLTRANSFERASE DEFICIENCY                                                               | MAT1A                  |
| METHYLMALONIC ACIDEMIA                                                                                  | MMAA, MMAB             |
| METHYLMALONIC ACIDEMIA; METHYLMALONYL COA MUTASE DEFICIENCY                                             | MUT                    |
| METHYLMALONIC ACIDEMIA; METHYLMALONYL COA EPIMERASE DEFICIENCY                                          | MCEE                   |
| 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 (MARFANO-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                    |
| 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                  |
| PROIONIC 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                                                                       | DUOX2                  |
| THYROID DYSHORMONOGENESIS, TYPE 6                                                                       | DUOX2                  |
| TRANSNINE HYDROXYLASE DEFICIENCY, SEGAWA SYNDROME                                                       | TCN2                   |
| TYROSINEMIA, TYPE I                                                                                     | TH                     |
| TYROSINEMIA, TYPE II                                                                                    | FAH                    |
| TYROSINEMIA, TYPE III                                                                                   | TAT                    |
| VITAMIN D-DEPENDENT RICKETS                                                                             | HPD                    |
| VITAMIN D-DEPENDENT RICKETS                                                                             | VDR                    |
| VLCD DEFICIENCY; VERY LONG CHAIN ACYL-COA DEHYDROGENASE DEFICIENCY                                      | CYP27B1                |
| WILSON DISEASE                                                                                          | ACADVL                 |
|                                                                                                         | ATP7B                  |

## OTHER CONDITIONS

|                                                       |                            |
|-------------------------------------------------------|----------------------------|
| AUTOSOMAL DOMINANT POLYCYSTIC KIDNEY DISEASE, TYPE 2  | PKD2                       |
| AUTOSOMAL RECESSIVE POLYCYSTIC KIDNEY DISEASE         | PKHD1                      |
| BARE LYMPHOCYTE SYNDROME TYPE II                      | CIITA                      |
| BIOTINIDASE DEFICIENCY                                | BTID                       |
| 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                       |
| CRISPOLI SYNDROME                                     | CRF1                       |
| FAMILIAL GLUCOCORTICOID DEFICIENCY                    | MC2R                       |
| FAMILIAL HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS           | PRF1                       |
| FAMILIAR MEDITERRANEAN FEVER FAILURE                  | MEFV                       |
| HEMOLYTIC ANEMIA (G6PD-RELATED)                       | G6PD                       |
| HEREDITARY PARAGANGLIOMA-PHEOCHROMOCYTOMA SYNDROME    | MAX, SDHAF2, SDHC, TMEM127 |
| HERMANSKY-PUDLAK SYNDROME 1                           | HP51                       |
| HERMANSKY-PUDLAK SYNDROME 4                           | HP54                       |
| 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                     |
| PTEN HAMARTOMA TUMOR SYNDROME                         | PTEN                       |
| ROTOR SYNDROME                                        | SLC01B1                    |
| SMITH-LEMLI-OPITZ SYNDROME                            | DHCR7                      |
| THROMBOTIC THROMBOCYTOPENIC PURPURA                   | ADAMTS13                   |
| ZAP70-RELATED SEVERE COMBINED IMMUNODEFICIENCY        | ZAP70                      |