Amplexa Genetics®

Analysis: Genes2Life

Test type: Carrier screening test

| # | Condition | Gene |
|---|---|---|
| 1 | 17-Beta-Hydroxysteroid Dehydrogenase Deficiency, Type III | *HSD17B3* |
| 2 | 21-Hydroxylase-Deficient Congenital Adrenal Hyperplasi | *CYP21A2* |
| | **A** | |
| 3 | Achondrogenesis, Type 1B | *SLC26A2* |
| 4 | Achromatopsia CNGB3-Related | *CNGB3* |
| 5 | Albinism, Oculocutaneous, Type I | *TYR* |
| 6 | Alkaptonuria | *HGD* |
| 7 | Alpers Syndrome | *POLG* |
| 8 | Alpha thalassemia | *HBA1* |
| 9 | Alpha thalassemia | *HBA2* |
| 10 | Amish Infantile Epilepsy Syndrome | *ST3GAL5* |
| 11 | Aspartylglycosaminuria | *AGA* |
| 12 | Ataxia Telangiectasia | *ATM* |
| | **B** | |
| 13 | Bardet-Biedl Syndrome 1 | *BBS1* |
| 14 | Bardet-Biedl Syndrome 13 / Meckel-Gruber Syndrome 1 / Joubert Syndrome 28 | *MKS1* |
| 15 | Bardet-Biedl Syndrome 2 | *BBS2* |
| 16 | Beta-Thalassemia | *HBB* |
| 17 | Biotinidase Deficiency | *BTD* |
| 18 | Bloom Syndrome | *BLM* |
| | **C** | |
| 19 | Canavan Disease | *ASPA* |
| 20 | Cartilage-Hair Hypoplasia | *RMRP* |
| 21 | Chronic Granulomatous Disease 4 | *CYBA* |
| 22 | Citrin Deficiency | *SLC25A13* |
| 23 | Classical homocystinuria | *CBS* |
| 24 | Congenital Adrenal Hyperplasia, 11-b hydroxylase | *CYP11B1* |
| 25 | Congenital Disorder of Glycosylation, Type 1a | *PMM2* |
| 26 | Cystic Fibrosis | *CFTR* |
| 27 | Cystinosis | *CTNS* |
| | **D** | |
| 28 | Dihydropyrimidine Dehydrogenase Deficiency | *DPYD* |
| | **E** | |
| 29 | Ellis-van Creveld Syndrome | *EVC2* |
| | **F** | |
| 30 | Factor XI Deficiency | *F11* |
| 31 | Familial Chloride Diarrhea | *SLC26A3* |
| 32 | Familial Dysautonomia | *ELP1* |
| 33 | Familial Hyperinsulinemic Hypoglycemia | *ABCC8* |

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|---|---|---|
| 34 | Familial Hyperinsulinism | *KCNJ11* |
| 35 | Familial Mediterranean Fever | *MEFV* |
| 36 | Fanconi Anemia, Group C | *FANCC* |
| 37 | Fanconi Anemia, Group G | *FANCG* |
| 38 | Fanconi Anemia, Troup A | *FANCA* |
| | **G** | |
| 39 | Galactokinase Deficiency, Type II | *GALK1* |
| 40 | Galactosemia | *GALT* |
| 41 | Gaucher Disease | *GBA* |
| 42 | Gitelman Syndrome | *SLC12A3* |
| 43 | Glutaric Acidemia, Type 1 | *GCDH* |
| 44 | Glycogen Storage Disease, Type 1A | *G6PC1* |
| 45 | Glycogen Storage Disease, Type 2 (Pompe Disease) | *GAA* |
| 46 | Glycogen Storage Disease, Type 3 | *AGL* |
| 47 | Glycogen Storage Disease, Type 4 | *GBE1* |
| | **H** | |
| 48 | Hereditary Fructose Intolerance | *ALDOB* |
| 49 | Hermansky-Pudlak Syndrome 1 | *HPS1* |
| 50 | Holocarboxylase Synthetase Deficiency | *HLCS* |
| 51 | Hydrolethalus Syndrome | *HYLS1* |
| 52 | Hypophosphatasia | *ALPL* |
| | **J** | |
| 53 | Joubert Syndrome 2 | *TMEM216* |
| | **L** | |
| 54 | Lethal Congenital Contracture Syndrome 1 | *GLE1* |
| 55 | Limb-Girdle Muscular Dystrophy, Type C3 | *POMGNT1* |
| | **M** | |
| 56 | Maple syrup urine Disease, Type 1A | *BCKDHA* |
| 57 | Maple syrup urine Disease, Type 1B | *BCKDHB* |
| 58 | Maple Syrup Urine Disease, Type III | *DLD* |
| 59 | Medium Chain Acyl-CoA Dehydrogenase Deficiency | *ACADM* |
| 60 | Metachromatic Leukodystrophy | *ARSA* |
| 61 | Mucolipidosis II/IIIA | *GNPTAB* |
| 62 | Mucolipidosis, Type IV | *MCOLN1* |
| 63 | Mucopolysaccharidosis, Type IVB | *GLB1* |
| 64 | Mulibrey Nanism | *TRIM37* |
| | **N** | |
| 65 | Nephrotic Syndrome, Type 1 | *NPHS1* |
| 66 | Neuronal ceroid lipofuscinosis, PPT1-Related | *PPT1* |

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|---|---|---|
| 67 | Neuronal ceroid lipofuscinosis, TPP1-Related | *TPP1* |
| 68 | Neuronal ceroid-lipofuscinosis, CLN5-Related | *CLN5* |
| 69 | Niemann-Pick Disease | *SMPD1* |
| 70 | Non-Syndromic Hearing Loss, GJB2-Related | *GJB2* |
| | **O** | |
| 71 | Osteopetrosis, Infantile Malignant | *TCIRG1* |
| | **P** | |
| 72 | Pendred Syndrome | *SLC26A4* |
| 73 | Phenylketonuria | *PAH* |
| 74 | Pituitary Hormone Deficiency 2 | *PROP1* |
| 75 | Polyglandular aAutoimmune Syndrome, Type 1 | *AIRE* |
| 76 | Primary congenital glaucoma | *CYP1B1* |
| | **R** | |
| 77 | Retinitis Pigmentosa 28 | *FAM161A* |
| | **S** | |
| 78 | Salla Disease | *SLC17A5* |
| 79 | Sandhoff Disease | *HEXB* |
| 80 | Smith-Lemli-Opitz Syndrome | *DHCR7* |
| 81 | Spinal Muscular Atrophy | *SMN1* |
| | **T** | |
| 82 | Tay-Sachs Disease | *HEXA* |
| 83 | Tyrosinemia, Type I | *FAH* |
| | **U** | |
| 84 | Usher Syndrome, Type 1B | *MYO7A* |
| 85 | Usher Syndrome, Type 1C | *USH1C* |
| 86 | Usher Syndrome, Type 1D | *CDH23* |
| 87 | Usher Syndrome, Type 1F | *PCDH15* |
| 88 | Usher Syndrome, Type 3 | *CLRN1* |
| | **V** | |
| 89 | Very-long Chain Acyl-CoA Dehydrogenase Deficiency | *ACADVL* |
| | **W** | |
| 90 | Walker-Warburg Syndrome | *FKTN* |
| 91 | Wilson Disease | *ATP7B* |
| 92 | Wolman Disease | *LIPA* |
| | **Z** | |
| 93 | Zellweger spectrum Disorders 4A | *PEX6* |