**Table S1. List of disorders and genes tested in the study**

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| --- | --- | --- | --- | --- |
|  | Disorder | Inherit-ance | Phenotype OMIM no. | Gene |
| 1 | Adrenoleukodystrophy | XL | 300100 | ABCD1 |
| 2 | Argininosuccinic aciduria | AR | 207900 | ASL |
| 3 | Ataxia telengiectasia | AR | 208900 | ATM |
| 4 |  Albinism type I  | AR | 203100,  | TYR |
| 5 | Albinism type II  | AR | 203200 | OCA2 |
| 6 | Albinism type III | AR | 203290 | TYRP1 |
| 7 | Alkaptonuria | AR | 203500 | HGD |
| 8 | Alpha 1 antitrypsin deficiency | AR | 613490 | SERPINA1 |
| 9 | Biotinidase deficiency | AR | 253260 | BTD |
| 10 | Beta Ketothiolase deficiency | AR | 203750 | ACAT1 |
| 11 | Congenital disorder of glycosylation type 1A | AR | 212065 | PMM2 |
| 12 |  Canavan disease | AR | 271900 | ASPA |
| 13 | Citrullinemia | AR | 215700 | ASS1 |
| 14 |  Cystic fibrosis | AR | 219700 | CFTR |
| 15 | Carnitine palmitoyl transferase deficiency type 1 | AR | 255120 | CPT1A |
| 16 | Congenital adrenal hyperplasia | AR | 201910 | CYP21A2 |
| 17 | Limb girdle muscle dystrophy AD 4/AR1 | AD/AR | 618129/253600 | CAPN3 |
| 18 | Deafness AD 3A/AR 1A | AD/AR | 601544/220290 | GJB2 |
| 19 | Deafness AR 18A/ Usher syndrome type 1C | AR | 602092/276904 | USH1C |
| 20 | Deafness AD 36/AR 7 | AD/AR | 606705/600974 | TMC1 |
| 21 | Deafness AR 8/10 | AR | 601072 | TMPRSS3 |
| 22 | Deafness AR 12 | AR | 601067 | CDH23 |
| 23 | Epidermolysis bullosa dystrophica | AD/AR | 131750/226600 | COL7A1 |
| 24 | Epidermolysis bullosa junctional | AR | 226700 | LAMC2 |
| 25 | Epidermolysis bullosa junctional/ Amelogenesis imperfecta | AR/AD | 226650/104530 | LAMB3 |
| 26 | Epidermolysis bullosa junctional/ epithelial recurrent erosion dystrophy | AR/AD | 226730, 122400 | COL17A1 |
| 27 | Epidermolysis Bullosa Junctional | AR/AD | 226650/131800 | ITGB4 |
| 28 | Fanconi anemia, complementation group C | AR | 227645 | FANCC |
| 29 | Fabry disease | XL | 301500 | GLA |
| 30 | Familial hypercholesterolemia | AD, AR | 143890 | LDLR |
| 31 | Familial hyperinsulinemic hypoglycaemia 1 | AD, AR | 256450 | ABCC8 |
| 32 | Glutaric aciduria type 1 | AR | 231670 | GCDH |
| 33 | Galactosemia | AR | 230400 | GALT |
| 34 | Gaucher disease | AR | 230800 | GBA |
| 35 | GM1 gangliosidosis | AR | 230500 | GLB1 |
| 36 | GM2 gangliosidosis | AR | 272800/268800 | HEXA, HEXB |
| 37 | Glycogen storage disease type 1A, 1B and 3  | AR | 232200 | G6PC |
| 38 | Glycogen storage disease type 1B | AR | 232220 | SLC37A4 |
| 39 | Glycogen storage disease type 3 | AR | 232400 | AGL |
| 40 | Homocystinuria | AR | 236200 | CBS |
| 41 | Hereditary fructose intolerance | AR | 229600 | ALDOB |
| 42 | Heme oxygenase-1 deficiency | AR | 614034 | HMOX1 |
| 43 | Hemophilia A (factor 8 deficiency)  | XL | 306700  | F8 |
| 44 | Hemophilia B (factor 9 deficiency) | XL | 306900 | F9 |
| 45 | Neurodegeneration with brain iron accumulation 1/HARP syndrome | AR | 234200/607236 | PANK2 |
| 46 | Ichthyosis, congenital, AR 1 | AR | 242300 | TGM1 |
| 47 | Ichthyosis, congenital, AR 4A, 4B (harlequin) | AR | 601277/242500 | ABCA12 |
| 48 | Krabbe disease | AR | 245200 | GALC |
| 49 | Meckel Gruber syndrome type 3 | AR | 607361 | TMEM67 |
| 50 | Medium chain acyl CoA dehydrogenase deficiency | AR | 201450 | ACADM |
| 51 | Metachromatic leukodystrophy | AR | 250100 | ARSA |
| 52 | Mucopolysaccharidosis type I | AR | 607014 | IDUA |
| 53 | Mucopolysaccharidosis type II  | XL | 309900, | IDS |
| 54 | Mucopolysaccharidosis type IVA | AR | 253000 | GALNS |
| 55 | Mucopolysaccharidosis type VI  | AR | 253200 | ARSB |
| 56 | Maple syrup urine disease type Ia | AR | 248600 | BCKDHA |
| 57 | Maple syrup urine disease type Ib | AR | 248600 | BCKDHB |
| 58 | Maple syrup urine disease type II | AR | 248600 | DBT |
| 59 | Methyl malonicaciduria mut A | AR | 251100 | MMAA |
| 60 | Methyl malonicaciduria mut B | AR | 251110 | MMAB |
| 61 | Methyl malonicaciduria mut C | AR | 277400 | MMACHC |
| 62 | Megalencephalic leukoencephalopathy with subcortical cysts | AR | 604004 | MLC1 |
| 63 | Nonketotic hyperglycinemia  | AR | 605899 | GLDC |
| 64 | Neuronal ceroid lipofuscinosis type 1 | AR | 256730 | PPT1 |
| 65 | Neuronal ceroid lipofuscinosis type 2 | AR | 204500 | TPP1 |
| 66 | Neuronal ceroid lipofuscinosis type 3 | AR | 204200 | CLN3 |
| 67 | Niemann Pick type A and B | AR | 257200/ 607616 | SMPD1 |
| 68 | Niemann Pick type C1 | AR | 257220 | NPC1 |
| 69 | Niemann Pick type C2 | AR | 607625 | NPC2 |
| 70 | Osteopetrosis AR 1 | AR | 259700 | TCIRG1 |
| 71 | Ornithine transcarbamylase deficiency | XL | 311250 | OTC |
| 72 | Phenylketonuria | AR | 261600 | PAH |
| 73 | Propionicacidemia | AR | 606054 | PCCA |
| 74 | Propionicacidemia | AR | 606054 | PCCB |
| 75 | Polycystic kidney disease 4 | AR | 263200 | PKHD1 |
| 76 | Glycogen storage disease type II (Pompe disease) | AR | 232300 | GAA |
| 77 | Pendred syndrome | AR | 274600 | SLC26A4 |
| 78 | Primary hyperoxaluria type 1 | AR | 259900 | AGXT |
| 79 | Progressive familial intrahepatic cholestasis type 1 | AR | 211600 | ATP8B1 |
| 80 | Progressive familial intrahepatic cholestasis type 2 | AR | 601847 | ABCB11 |
| 81 | Progressive familial intrahepatic cholestasis type 3 | AR | 602347 | ABCB4 |
| 82 | Smith Lemli Opitz syndrome | AR | 270400 | DHCR7 |
| 83 | Severe combined immunodeficiency  | XL | 300400 | IL2RG |
| 84 | Severe combined immunodeficiency  | AR | 102700 | ADA |
| 85 | Tyrosinemia type 1 | AR | 276700 | FAH |
| 86 | Very long chain acyl CoA dehydrogenase deficiency | AR | 201475 | ACADVL |
| 87 | Wilson disease | AR | 277900 | ATP7B |
| 88 | Zellweger syndrome | AR | 214100 | PEX1 |