# Diseases list targeted by Linea Vita

| Nr. Crt. | Disease | OMIM Disease | Gene | OMIM Gene |
|---|---|---|---|---|
| 1 | 3-Methylcrotonyl-CoA carboxylase 1 deficiency | 210200 | *MCCC1* | 609010 |
| 2 | 3-Methylcrotonyl-CoA carboxylase 2 deficiency | 210210 | *MCCC2* | 609014 |
| 3 | Achondrogenesis, type IA | 200600 | *TRIP11* | 604505 |
| 4 | Achondrogenesis, type IB | 600972 | *SLC26A2* | 606718 |
| 5 | Achromatopsia, type 2 | 216900 | *CNGA3* | 600053 |
| 6 | Achromatopsia, type 3 | 262300 | *CNGB3* | 605080 |
| 7 | Acyl-CoA Dehydrogenase medium chain deficiency | 201450 | *ACADM* | 607008 |
| 8 | Acyl-CoA Dehydrogenase short chain deficiency | 201470 | *ACADS* | 606885 |
| 9 | Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency | 202010 | *CYP11B1* | 610613 |
| 10 | Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete | 613743 | *CYP11A1* | 118485 |
| 11 | Adrenal hypoplasia, congenital | 300200 | *NROB1* | 300473 |
| 12 | Adrenoleukodystrophy | 300100 | *ABCD1* | 300371 |
| 13 | Albinism, oculocutaneous, type IA | 203100 | *TYR* | 606933 |
| 14 | Albinism, oculocutaneous, type IB | 606952 | *TYR* | 606933 |
| 15 | Albinism, oculocutaneous, type II and brown | 203200 | *OCA2* | 611409 |
| 16 | Albinism, oculocutaneous, type IV | 606574 | *SLC45A2* | 606202 |
| 17 | Alpha-methylacetoacetic aciduria | 203750 | *ACAT1* | 607809 |
| 18 | Alport syndrome X-linked | 301050 | *COL4A5* | 303630 |
| 19 | Argininemia | 207800 | *ARG1* | 608313 |
| 20 | Argininosuccinic aciduria | 207900 | *ASL* | 608310 |
| 21 | Aspartylglucosaminuria | 208400 | *AGA* | 613228 |
| 22 | Ataxia telangiectasia | 208900 | *ATM* | 607585 |
| 23 | Atelosteogenesis, type II/De la Chapelle dysplasia | 256050 | *SLC26A2* | 606718 |
| 24 | Bardet-Biedl syndrome, type 1 | 209900 | *BBS1* | 209901 |
| 25 | Bardet-Biedl syndrome, type 10 | 615987 | *BBS10* | 610148 |
| 26 | Bardet-Biedl syndrome, type 12 | 615989 | *BBS12* | 610683 |
| 27 | Bardet-Biedl syndrome, type 13 | 615990 | *MKS1* | 609883 |
| 28 | Bardet-Biedl syndrome, type 14 | 615991 | *CEP290/BBS14* | 610142 |
| 29 | Bardet-Biedl syndrome, type 2 | 615981 | *BBS2* | 606151 |
| 30 | Bardet-Biedl syndrome, type 6 | 605231 | *BBS6/MKKS* | 604896 |
| 31 | Batten disease - Neuronal ceroid lipofuscinosis, type 2 | 204500 | *TPP1* | 607998 |
| 32 | Batten disease - Neuronal ceroid lipofuscinosis, type 3 | 204200 | *CLN3* | 607042 |
| 33 | Becker muscular dystrophy | 300376 | *DMD* | 300377 |
| 34 | Beta-thalassemia | 613985 | *HBB* | 141900 |
| 35 | Biotinidase deficiency | 253260 | *BTD* | 609019 |
| 36 | Bloom syndrome | 210900 | *RECQL3* | 604610 |
| 37 | Canavan disease | 271900 | *ASPA* | 608034 |
| 38 | Carnitine deficiency | 212140 | *SLC22A5* | 603377 |
| 39 | Cerebral creatine deficiency syndrome, type 1 | 300352 | *SLC6A8* | 300036 |
| 40 | Cerebral creatine deficiency syndrome, type 2 | 612736 | *GAMT* | 601240 |
| 41 | Cerebrotendinous xanthomatosis | 213700 | *CYP27A1* | 606530 |
| 42 | Citrullinemia | 215700 | *ASS1* | 603470 |
| 43 | COACH syndrome, type 1 | 216360 | *TMEM67* | 609884 |
| 44 | COACH syndrome, type 2 | 619111 | *CC2D2A* | 612013 |
| 45 | Combined immunodeficiency, X-linked, moderate | 312863 | *IL2RG* | 308380 |
| 46 | Cone-rod dystrophy, type 13 | 608194 | *RPGRIP1* | 605446 |
| 47 | Cone-rod dystrophy, type 3 | 604116 | *ABCA4* | 601691 |
| 48 | Congenital disorder of glycosylation, type Ia | 212065 | *PMM2* | 601785 |
| 49 | Congenital disorder of glycosylation, type Ib | 602579 | *MPI* | 154550 |

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| 50 | Congenital disorder of glycosylation, type Ic | 603147 | *ALG6* | 604566 |
| 51 | CPT II deficiency, infantile | 600649 | *CPT2* | 600650 |
| 52 | CPT II deficiency, lethal neonatal | 608836 | *CPT2* | 600650 |
| 53 | CPT II deficiency, myopathic, stress-induced | 255110 | *CPT2* | 600650 |
| 54 | CRASH syndrome/MASA syndrome | 303350 | *L1CAM* | 304100 |
| 55 | Cystic fibrosis | 219700 | *CFTR* | 602421 |
| 56 | D-bifunctional protein deficiency | 261515 | *HSD17B4* | 601860 |
| 57 | Deafness, autosomal recessive, type 12 | 601386 | *CDH23* | 605516 |
| 58 | Deafness, autosomal recessive, type 18A | 602092 | *USH1C* | 605242 |
| 59 | Deafness, autosomal recessive, type 1A | 220290 | *GJB2* | 121011 |
| 60 | Deafness, autosomal recessive, type 1A | 220290 | *GJB3* | 603324 |
| 61 | Deafness, autosomal recessive, type 1B | 612645 | *GJB6* | 604418 |
| 62 | Deafness, autosomal recessive, type 2 | 600060 | *MYO7A* | 276903 |
| 63 | Deafness, autosomal recessive, type 23 | 609533 | *PCDH15* | 605514 |
| 64 | Deafness, autosomal recessive, type 4 | 600791 | *SLC26A4* | 605646 |
| 65 | Deafness, digenic GJB2/GJB6 | 220290 | *GJB6* | 604418 |
| 66 | Diabetes mellitus, permanent neonatal, type 3 | 618857 | *ABCC8* | 600509 |
| 67 | Diastrophic dysplasia | 222600 | *SLC26A2* | 606718 |
| 68 | Duchenne muscular dystrophy | 310200 | *DMD* | 300377 |
| 69 | Ectodermal dysplasia, type 10B, hypohidrotic/hair/tooth type, autosomal recessive | 224900 | *EDAR* | 604095 |
| 70 | Ectodermal dysplasia, X-linked | 305100 | *EDA* | 300451 |
| 71 | Epidermolysis bullosa dystrophica | 226600 | *COL7A1* | 120120 |
| 72 | Epidermolysis bullosa simplex, type 1D | 601001 | *KRT14* | 148066 |
| 73 | Epidermolysis bullosa simplex, type 2D | 619599 | *KRT5* | 148040 |
| 74 | Epidermolysis bullosa, junctional, type Herlitz | 226700 | *LAMB3, LAMC2* | 150310, 226700 |
| 75 | Epidermolysis bullosa, junctional, type non-Herlitz | 226650 | *LAMB3, LAMC2, COL7A1* | 150310, 226700, 113811 |
| 76 | Epiphyseal dysplasia, multiple, type 4 | 226900 | *SLC26A2* | 606718 |
| 77 | Fabry disease | 301500 | *GLA* | 300644 |
| 78 | Familial hyperinsulinemic hypoglycemia, type 1 | 256450 | *ABCC8* | 600509 |
| 79 | Familial Mediterranean fever, autosomal recessive | 249100 | *MEFV* | 608107 |
| 80 | Fragile X syndrome | 300624 | *FMR1* | 309550 |
| 81 | Fructose intolerance, hereditary | 229600 | *ALDOB* | 612724 |
| 82 | Galactosemia | 230400 | *GALT* | 606999 |
| 83 | Gaucher disease perinatal lethal | 608013 | *GBA* | 606463 |
| 84 | Gaucher disease, type I | 230800 | *GBA* | 606463 |
| 85 | Gaucher disease, type II | 230900 | *GBA* | 606463 |
| 86 | Gaucher disease, type III | 231000 | *GBA* | 606463 |
| 87 | Gaucher disease, type IIIC | 231005 | *GBA* | 606463 |
| 88 | Glutaric acidemia, type 1 | 231670 | *GCDH* | 608801 |
| 89 | Glycogen storage disease, type 1A | 232200 | *G6PC* | 613742 |
| 90 | Glycogen storage disease, type 1B | 232220 | *SLC37A4* | 602671 |
| 91 | Glycogen storage disease, type 1C | 232240 | *SLC37A4* | 602671 |
| 92 | Glycogen storage disease, type II (Pompe) | 232300 | *GAA* | 606800 |
| 93 | Glycogen storage disease, type IV | 232500 | *GBE1* | 607839 |
| 94 | GM1-gangliosidosis, type I | 230500 | *GLB1* | 611458 |
| 95 | GM1-gangliosidosis, type II | 230600 | *GLB1* | 611458 |
| 96 | GM1-gangliosidosis, type III | 230650 | *GLB1* | 611458 |
| 97 | Heimler syndrome, type 1 | 234580 | *PEX1* | 602136 |
| 98 | Heimler syndrome, type 2 | 616617 | *PEX6* | 601498 |
| 99 | Hemochromatosis | 235200 | *HFE* | 613609 |
| 100 | Hemolytic anemia due to G6PD deficiency | 300908 | *G6PD* | 305900 |
| 101 | Hemophilia A | 306700 | *F8* | 300841 |
| 102 | Hemophilia B | 306900 | *F9* | 300746 |
| 103 | Holocarboxylase synthetase deficiency | 253270 | *HLCS* | 609018 |
| 104 | Homocystinuria, B6-responsive and nonresponsive types | 236200 | *CBS* | 613381 |
| 105 | Hydrocephalus due to aqueductal stenosis<br>Hydrocephalus with congenital idiopathic intestinal pseudoobstruction<br>Hydrocephalus with Hirschsprung disease | 307000 | *L1CAM* | 304100 |

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| 106 | Hyperphenylalaninemia, BH4-deficient, type A | 261640 | *PTS* | 612719 |
| 107 | Hypophosphatasia, infantile | 241500 | *ALPL* | 171760 |
| 108 | Ichthyosis, congenital, autosomal recessive, type 1 | 242300 | *TGM1* | 190195 |
| 109 | Isovaleric acidemia | 243500 | *IVD* | 607036 |
| 110 | Joubert syndrome, type 17 | 614615 | *CPLANE1* | 614571 |
| 111 | Joubert syndrome, type 2 | 608091 | *TMEM216* | 613277 |
| 112 | Joubert syndrome, type 28 | 617121 | *MKS1* | 609883 |
| 113 | Joubert syndrome, type 3 | 608629 | *AHI1* | 608894 |
| 114 | Joubert syndrome, type 5 | 610188 | *CEP290/bbs14* | 610142 |
| 115 | Joubert syndrome, type 6 | 610688 | *TMEM67* | 609884 |
| 116 | Joubert syndrome, type 9 | 612285 | *CC2D2A* | 612013 |
| 117 | LCHAD deficiency | 609016 | *HADHA* | 600890 |
| 118 | Leber congenital amaurosis, type 1 | 204000 | *GUCY2D* | 600179 |
| 119 | Leber congenital amaurosis, type 10 | 611755 | *CEP290/bbs14* | 610142 |
| 120 | Leber congenital amaurosis, type 13 | 612712 | *RDH12* | 608830 |
| 121 | Leber congenital amaurosis, type 2 | 204100 | *RPE65* | 180069 |
| 122 | Leber congenital amaurosis, type 6 | 613826 | *RPGRIP1* | 605446 |
| 123 | Leber congenital amaurosis, type 8 | 613835 | *CRB1* | 604210 |
| 124 | Maple syrup urine disease, type 1a | 248600 | *BCKDHA* | 608348 |
| 125 | Maple syrup urine disease, type 1b | 248600 | *BCKDHB* | 248611 |
| 126 | Maple syrup urine disease, type II | 248600 | *DBT* | 248610 |
| 127 | Meckel syndrome, type 1 | 249000 | *MKS1* | 609883 |
| 128 | Meckel syndrome, type 2 | 603194 | *TMEM216* | 613277 |
| 129 | Meckel syndrome, type 3 | 607361 | *TMEM67* | 609884 |
| 130 | Meckel syndrome, type 4 | 611134 | *CEP290/bbs14* | 610142 |
| 131 | Meckel syndrome, type 6 | 612284 | *CC2D2A* | 612013 |
| 132 | Menkes disease | 309400 | *ATP7A* | 300011 |
| 133 | Metachromatic leukodystrophy | 250100 | *ARSA* | 607574 |
| 134 | Methylmalonic aciduria (mut0) | 251000 | *MMUT* | 609058 |
| 135 | Methylmalonic aciduria and homocystinuria, type cblC | 277400 | *MMACHC* | 609831 |
| 136 | Methylmalonic aciduria, vitamin B12-responsive, type cblA | 251100 | *MMAA* | 607481 |
| 137 | Methylmalonic aciduria, vitamin B12-responsive, type cblB | 251110 | *MMAB* | 607568 |
| 138 | Mitochondrial DNA depletion syndrome, type 4A (Alpers type) | 203700 | *POLG* | 174763 |
| 139 | Mitochondrial DNA depletion syndrome, type 4B (MNGIE type) | 613662 | *POLG* | 174763 |
| 140 | Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE) | 607459 | *POLG* | 174763 |
| 141 | Mitochondrial trifunctional protein deficiency | 609015 | *HADHA* | 600890 |
| 142 | Mucolipidosis, type II alpha/beta | 252500 | *GNPTAB* | 607840 |
| 143 | Mucolipidosis, type III alpha/beta | 252600 | *GNPTAB* | 607840 |
| 144 | Mucolipidosis, type IV | 252650 | *MCOLN1* | 605248 |
| 145 | Mucopolysaccharidosis, type Ih | 607014 | *IDUA* | 252800 |
| 146 | Mucopolysaccharidosis, type Ih/s | 607015 | *IDUA* | 252800 |
| 147 | Mucopolysaccharidosis, type II | 309900 | *IDS* | 300823 |
| 148 | Mucopolysaccharidosis, type IIIA | 252900 | *SGSH* | 605270 |
| 149 | Mucopolysaccharidosis, type IIIB | 252920 | *NAGLU* | 609701 |
| 150 | Mucopolysaccharidosis, type Is | 607016 | *IDUA* | 252800 |
| 151 | Mucopolysaccharidosis, type IVB (Morquio) | 253010 | *GLB1* | 611458 |
| 152 | Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3 | 613157 | *POMGNT1* | 606822 |
| 153 | Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4 | 611588 | *FKTN* | 607440 |
| 154 | Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5 | 607155 | *FKRP* | 606596 |
| 155 | Muscular dystrophy-dystroglycanopathy, type A, 3 | 253280 | *POMGNT1* | 606822 |
| 156 | Muscular dystrophy-dystroglycanopathy, type A, 4 | 253800 | *FKTN* | 607440 |
| 157 | Muscular dystrophy-dystroglycanopathy, type A, 5 | 613153 | *FKRP* | 606596 |
| 158 | Muscular dystrophy-dystroglycanopathy, type B, 3 | 613151 | *POMGNT1* | 606822 |
| 159 | Muscular dystrophy-dystroglycanopathy, type B, 4 | 613152 | *FKTN* | 607440 |
| 160 | Muscular dystrophy-dystroglycanopathy, type B, 5 | 606612 | *FKRP* | 606596 |
| 161 | Myotonia congenita, recessive | 255700 | *CLCN1* | 118425 |
| 162 | Myotubular myopathy, X-linked | 310400 | *MTM1* | 300415 |
| 163 | Naxos/Carvajal syndrome | 601214 | *JUP* | 173325 |
| 164 | Niemann-Pick disease, type A | 257200 | *SMPD1* | 607608 |
| 165 | Niemann-Pick disease, type B | 607616 | *SMPD1* | 607608 |
| 166 | Niemann-Pick disease, type C1 + type D | 257220 | *NPC1* | 607623 |

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| 167 | Niemann-Pick disease, type C2 | 607625 | *NPC2* | 601015 |
| 168 | Nijmegen syndrome | 251260 | *NBN* | 602667 |
| 169 | Nonaka myopathy | 605820 | *GNE* | 603824 |
| 170 | Odontochondrodysplasia, type 1 | 184260 | *TRIP11* | 604505 |
| 171 | Ornithine transcarbamylase deficiency | 311250 | *OTC* | 300461 |
| 172 | Orofaciodigital syndrome, type VI | 277170 | *CPLANE1* | 614571 |
| 173 | Partial agenesis of corpus callosum | 304100 | *L1CAM* | 304100 |
| 174 | Pendred syndrome | 274600 | *SLC26A4* | 605646 |
| 175 | Peroxisome biogenesis disorder, type 1A (Zellweger) | 214100 | *PEX1* | 602136 |
| 176 | Peroxisome biogenesis disorder, type 1B (NALD/IRD) | 601539 | *PEX1* | 602136 |
| 177 | Peroxisome biogenesis disorder, type 4A (Zellweger) | 614862 | *PEX6* | 601498 |
| 178 | Peroxisome biogenesis disorder, type 4B | 614863 | *PEX6* | 601498 |
| 179 | Phenylketonuria | 261600 | *PAH* | 612349 |
| 180 | Polycystic kidney disease 4, with or without hepatic disease | 263200 | *PKHD1* | 606702 |
| 181 | Progressive external ophthalmoplegia, autosomal recessive, type 1 | 258450 | *POLG* | 174763 |
| 182 | Proprionicacidemia | 606054 | *PCCA, PCCB* | 232000, 232050 |
| 183 | Pyruvate carboxylase deficiency | 266150 | *PC* | 608786 |
| 184 | Retinitis pigmentosa, type 12 | 600105 | *CRB1* | 604210 |
| 185 | Retinitis pigmentosa, type 19 | 601718 | *ABCA4* | 601691 |
| 186 | Retinitis pigmentosa, type 2 | 312600 | *RP2* | 300757 |
| 187 | Retinitis pigmentosa, type 20 | 613794 | *RPE65* | 180069 |
| 188 | Retinitis pigmentosa, type 25 | 602772 | *EYS* | 612424 |
| 189 | Retinitis pigmentosa, type 39 | 613809 | *USH2A* | 608400 |
| 190 | Retinitis pigmentosa, type 43 | 613810 | *PDE6A* | 180071 |
| 191 | Retinitis pigmentosa, type 61 | 614180 | *CLRN1* | 606397 |
| 192 | Retinitis pigmentosa, type 74 | 616562 | *BBS2* | 606151 |
| 193 | Retinitis pigmentosa, type 76 | 617123 | *POMGNT1* | 606822 |
| 194 | Retinoschisis, X-linked | 312700 | *RS1* | 300839 |
| 195 | Sandhoff disease | 268800 | *HEXB* | 606873 |
| 196 | Senior-Loken syndrome, type 6 | 610189 | *CEP290/bbs14* | 610142 |
| 197 | Severe combined immunodeficiency, X-linked | 300400 | *IL2RG* | 308380 |
| 198 | Sickle cell anemia | 603903 | *HBB* | 141900 |
| 199 | Smith-Lemli-Opitz syndrome | 270400 | *DHCR7* | 602858 |
| 200 | Spinal muscular atrophy, type 1 | 253300 | *SMN1* | 600354 |
| 201 | Spinal muscular atrophy, type 2 | 253550 | *SMN1* | 600354 |
| 202 | Spinal muscular atrophy, type 3 | 253400 | *SMN1* | 600354 |
| 203 | Spinal muscular atrophy, type 4 | 271150 | *SMN1* | 600354 |
| 204 | Stargardt disease, type 1<br>Retinal dystrophy, early-onset severe<br>Fundus flavimaculatus | 248200 | *ABCA4* | 601691 |
| 205 | Tay-Sachs disease | 272800 | *HEXA* | 606869 |
| 206 | Trimethylaminuria | 602079 | *FMO3* | 136132 |
| 207 | Tyrosinemia, type I | 276700 | *FAH* | 613871 |
| 208 | Usher syndrome, type 1b | 276900 | *MYO7A* | 276903 |
| 209 | Usher syndrome, type 1C | 276904 | *USH1C* | 605242 |
| 210 | Usher syndrome, type 1d | 601067 | *CDH23, PCDH15* | 605516, 605514 |
| 211 | Usher syndrome, type 1F | 602083 | *PCDH15* | 605514 |
| 212 | Usher syndrome, type 2a | 276901 | *USH2A* | 608400 |
| 213 | Usher syndrome, type 2C | 605472 | *ADGRV1* | 602851 |
| 214 | Usher syndrome, type 3A | 276902 | *CLRN1* | 606397 |
| 215 | Vitamin D-dependent rickets, type I | 264700 | *CYP27B1* | 264700 |
| 216 | VLCAD deficiency | 201475 | *ACADVL* | 609575 |
| 217 | Wilson disease | 277900 | *ATP7B* | 606882 |