linea vita

Disease list targeted by LINEA VITA test

Nr. crt.	Disease	OMIM (disease)	Gene	OMIM (gene)
1	Acyl-CoA Dehydrogenase medium chain deficiency	201450	*ACADM*	607008
2	Acyl-CoA Dehydrogenase short chain deficiency	201470	*ACADS*	606885
3	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency	202010	*CYP11B1*	610613
4	Adrenoleukodystrophy	300100	*ABCD1*	300371
5	Albinism Type 1	203100	*TYR*	606933
6	Albinism Type 2	203200	*OCA2*	611409
7	Albinism Type 3	203290	*TYRP1*	115501
8	Albinism Type 4	606574	*SLC45A2*	606202
9	Alport syndrome type 2	203780	*COL4A3*	120070
10	Alport syndrome type 2	203780	*COL4A4*	120131
11	Alport syndrome X-linked	301050	*COL4A5*	303630
12	Alstrom syndrome	203800	*ALMS1*	606844
13	Argininemia	207800	*ARG1*	608313
14	Argininosuccinic aciduria	207900	*ASL*	608310
15	Aspartylglucosaminuria	208400	*AGA*	613228
16	Ataxia telangiectasia	208900	*ATM*	607585
17	Bardet-Biedl syndrome 1	209900	*BBS1*	209901
18	Bardet-Biedl syndrome 10	615987	*BBS10*	610148
19	Bardet-Biedl syndrome 12	615989	*BBS12*	610683
20	Bardet-Biedl syndrome 13	615990	*MKS1*	609883
21	Bardet-Biedl syndrome 2	615981	*BBS2*	606151
22	Bardet-Biedl syndrome 3	600151	*ARL6*	608845
23	Bardet-Biedl syndrome 4	615982	*BBS4*	600374
24	Bardet-Biedl syndrome 9	615986	*BBS9*	615986
25	Batten disease- ceroid lipofuscinosis neuronal 2	204500	*TPP1*	607998
26	Batten disease- ceroid lipofuscinosis neuronal 3	204200	*CLN3*	607042
27	Becker Muscular dystrophy	300376	*DMD*	300377
28	Biotinidase deficiency	253260	*BTD*	609019
29	Cardiomyopathy dilated	601287	*SGCD*	601411
30	Cardiomyopathy dilated	606685	*SGCD*	601411
31	Cardiomyopathy dilated type 1X	611615	*FKTN*	607440
32	Cardiomyopathy dilated type 3B	302045	*DMD*	300377
33	Carnitine deficiency	212140	*SLC22A5*	603377
34	Cerebrotendinous xanthomatosis	213700	*CYP27A1*	606530
35	Citrullinemia	215700	*ASS1*	603470
36	COACH syndrome	212065	*PMM2*	601785
37	Combined immunodeficiency, X-linked, moderate	312863	*IL2RG*	308380
38	Congenital disorder of glycosylation, type Ib	602579	*MPI*	154550
39	Congenital disorder of glycosylation, type Ic	603147	*ALG6*	604566
40	Crigler-Najjar syndrome type 1	218800	*UGT1A1*	191740
41	Cystic fibrosis	219700	*CFTR*	602421
42	D-bifunctional protein deficiency	261515	*HSD17B4*	601860
43	Deafness, autosomal recessive 1A	220290	*GJB2*	121011
44	Deafness, autosomal recessive 1A	220290	*GJB3*	603324
45	Deafness, autosomal recessive 1B	612645	*GJB6*	604418
46	Deafness, autosomal recessive 23	609533	*PCDH15*	605514
47	Deafness, autosomal recessive 4	600791	*SLC26A4*	605646
48	Deafness, digenic GJB2/GJB6	220290	*GJB6*	604418
49	Diabetes insipidus, nephrogenic	304800	*AVPR2*	300538
50	Duchenne muscular dystrophy	310200	*DMD*	300377
51	Ectodermal dysplasia X-linked	305100	*EDA*	300451
52	Ectodermal dysplasia X-linked	249100	*MEFV*	608107
53	Enhanced S-cone syndrome	268100	*NR2E3*	604485
54	Epidermolysis bullosa dystrophica	226600	*COL7A1*	120120
55	Epidermolysis bullosa simplex, type 1	601001	*KRT14*	148066
56	Epidermolysis bullosa simplex, type 1	601001	*KRT5*	148040
57	Epidermolysis bullosa, junctional	226650	*COL17A1*	113811
58	Epidermolysis bullosa, junctional, type Herlitz	226700	*LAMB3*	150310
59	Epidermolysis bullosa, junctional, type Herlitz	226650	*LAMC2*	150292
60	Epidermolysis bullosa, junctional, type non-Herlitz	226700	*LAMC2*	150292
61	Fabry disease	301500	*GLA*	300644
62	Fragile X syndrome	300624	*FMR1*	309550
63	Fructose intolerance, hereditary	229600	*ALDOB*	612724
64	Fundus flavimaculatus	248200	*ABCA4*	601691
65	Galactosemia	230400	*GALT*	606999
66	Gaucher disease perinatal lethal	608013	*GBA*	606463
67	Gaucher disease type I	230800	*GBA*	606463
68	Gaucher disease type II	230900	*GBA*	606463
69	Gaucher disease type III	231000	*GBA*	606463
70	Gaucher disease type IIIC	231005	*GBA*	606463
71	Glutaric acidemia type 1	231670	*GCDH*	608801
72	Glycogen storage disease type 1A	232200	*G6PC*	613742
73	Glycogen storage disease type 1B	232220	*SLC37A4*	602671
74	Glycogen storage disease type II (Pompe)	232300	*GAA*	606800
75	Heimler syndrome type 1	234580	*PEX1*	602136
76	Heimler syndrome type 2	616617	*PEX6*	601498
77	Hemochromatosis	235200	*HFE*	613609
78	Hemochromatosis	604250	*TFR2*	604720
79	Hemolytic anemia due to G6PD deficiency	300908	*G6PD*	305900
80	Hemophilia A	306700	*F8*	300841
81	Hemophilia B	306900	*F9*	300746
82	HMG-CoA lyase deficiency	246450	*HMGCL*	613898
83	Holocarboxylase synthetase deficiency	253270	*HLCS*	609018
84	Homocystinuria, B6-responsive and nonresponsive types	236200	*CBS*	613381
85	Hyperinsulinemic hypoglycemia, familial, type 1	256450	*ABCC8*	600509
86	Hyperinsulinemic hypoglycemia, familial, type 2	601820	*KCNJ11*	600937
87	Hyperphenylalaninemia, BH4-deficient, A	261640	*PTS*	612719
88	Hypophosphatasia, infantile	241500	*ALPL*	171760

89	**Ichthyosis, congenital, autosomal recessive 1**	242300	***TGM1***	190195
90	**Isovaleric acidemia**	243500	***IVD***	607036
91	**Joubert syndrome type 2**	608091	***TMEM216***	613277
92	**Joubert syndrome type 28**	617121	***MKS1***	609883
93	**Joubert syndrome type 7**	611560	***RPGRIP1L***	610937
94	**Joubert syndrome type 8**	612291	***ARL13B***	608922
95	**Krabbe disease**	245200	***GALC***	606890
96	**LCHAD deficiency**	609016	***HADHA***	600890
97	**Maple syrup urine disease, type 1a**	248600	***BCKDHA***	608348
98	**Maple syrup urine disease, type 1b**	248600	***BCKDHB***	248611
99	**Maple syrup urine disease, type II**	248600	***DBT***	248610
100	**Meckel syndrome type 1**	249000	***MKS1***	609883
101	**Menkes disease**	309400	***ATP7A***	300011
102	**Metachromatic leukodystrophy**	250100	***ARSA***	607574
103	**Methylmalonic aciduria (mut0)**	251000	***MMUT***	609058
104	**Methylmalonic aciduria and homocystinuria, type cblD**	277410	***MMADHC***	611935
105	**Methylmalonic aciduria, vitamin B12-responsive, type cblA**	251100	***MMAA***	607481
106	**Methylmalonic aciduria, vitamin B12-responsive, type cblB**	251110	***MMAB***	607568
107	**Mitochondrial trifunctional protein deficiency**	609015	***HADHA***	600890
108	**Mucolipidosis II alpha/beta**	252500	***GNPTAB***	607840
109	**Mucolipidosis III gamma**	252605	***GNPTG***	607838
110	**Mucolipidosis IV**	252650	***MCOLN1***	605248
111	**Mucopolysaccharidosis Ih/s**	607015	***IDUA***	252800
112	**Mucopolysaccharidosis II**	309900	***IDS***	300823
113	**Mucopolysaccharidosis type IIIA**	252900	***SGSH***	605270
114	**Mucopolysaccharidosis type IIIB**	252920	***NAGLU***	609701
115	**Mucopolysaccharidosis type IIIC**	252930	***HGSNAT***	610453
116	**Muscular dystrophy limb-girdle type 2C**	253700	***SGCG***	608896
117	**Muscular dystrophy, limb-girdle type 2D**	608099	***SGCA***	600119
118	**Muscular dystrophy, limb-girdle type 2E**	604286	***SGCB***	600900
119	**Muscular dystrophy-dystroglycanopathy, type A, 5**	613153	***FKRP***	606596
120	**Myotubular myopathy, X-linked**	310400	***MTM1***	300415
121	**Naxos/Carvajal syndrome**	601214	***JUP***	173325
122	**Nephrotic syndrome, type 2**	600995	***NPHS2***	604766
123	**Niemann-Pick disease type A**	257200	***SMPD1***	607608
124	**Niemann-Pick disease type C1**	257220	***NPC1***	607623
125	**Niemann-Pick disease type C2**	607625	***NPC2***	601015
126	**Nijmegen syndrome**	251260	***NBN***	602667
127	**Nonaka myopathy**	605820	***GNE***	603824
128	**Ornithine transcarbamylase deficiency**	311250	***OTC***	300461
129	**Phenylketonuria**	261600	***PAH***	612349
130	**Polycystic kidney and hepatic disease**	263200	***PKHD1***	606702
131	**Proprionicacidemia**	606054	***PCCA***	232000
132	**Proprionicacidemia**	606054	***PCCB***	232050
133	**Pyruvate carboxylase deficiency**	266150	***PC***	608786
134	**Retinal dystrophy, early-onset severe**	248200	***ABCA4***	601691
135	**Retinitis pigmentosa 19**	601718	***ABCA4***	601691
136	**Retinitis pigmentosa 37**	611131	***NR2E3***	604485
137	**Retinitis pigmentosa 39**	613809	***USH2A***	608400
138	**Retinitis pigmentosa 41**	612095	***PROM1***	604365
139	**Retinitis pigmentosa 59**	613861	***DHDDS***	608172
140	**Retinitis pigmentosa 61**	614180	***CLRN1***	606397
141	**Retinitis pigmentosa 73**	616544	***HGSNAT***	610453
142	**Retinitis pigmentosa 76**	617123	***POMGNT1***	606822
143	**Retinoschisis X-linked**	312700	***RS1***	300839
144	**Roberts-SC phocomelia syndrome**	268300	***ESCO2***	609353
145	**Sandhoff disease**	268800	***HEXB***	606873
146	**Segawa syndrome**	605407	***TH***	191290
147	**Severe combined immunodeficiency, X-linked**	300400	***IL2RG***	308380
148	**Sjogren-Larsson syndrome**	270200	***ALDH3A2***	609523
149	**Smith-Lemli-Opitz syndrome**	270400	***DHCR7***	602858
150	**Spastic ataxia, Charlevoix-Saguenay**	270550	***SACS***	604490
151	**Spinal muscular atrophy type 1**	253300	***SMN1***	600354
152	**Spinal muscular atrophy type 2**	253550	***SMN1***	600354
153	**Spinal muscular atrophy type 3**	253400	***SMN1***	600354
154	**Spinal muscular atrophy type 4**	271150	***SMN1***	600354
155	**STAR syndrome**	201710	***STAR***	600617
156	**Stargardt disease type 1**	248200	***ABCA4***	601691
157	**Tay-Sachs disease**	272800	***HEXA***	606869
158	**Thalassemia B; Sickle cell anemia**	603903	***HBB***	141900
159	**Thalassemia, beta**	613985	***HBB***	141900
160	**Tyrosinemia, type I**	276700	***FAH***	613871
161	**Usher syndrome type 1b,**	276900	***MYO7A***	276903
162	**Usher syndrome type 1c**	276904	***USH1C***	605242
163	**Usher syndrome type 1d**	601067	***CDH23***	605516
164	**Usher syndrome type 2a**	276901	***USH2A***	608400
165	**Usher syndrome type 3a**	276902	***CLRN1***	606397
166	**VLCAD deficiency**	201475	***ACADVL***	609575
167	**Wilson disease**	277900	***ATP7B***	606882
168	**Wolman disease**	278000	***LIPA***	613497
169	**Xeroderma pigmentosum, group A**	278700	***XPA***	611153
170	**Xeroderma pigmentosum, group C**	278720	***XPC***	613208