Carrier Screening Condition List (170+ panel)

No.	Diseases	Genes
1	Ichthyosis, congenital, autosomal recessive 4A	ABCA12
2	Ichthyosis, congenital, autosomal recessive 4B (harlequin)	ABCA12
3	Cholestasis, progressive familial intrahepatic 2	ABCB11
4	Cholestasis, progressive familial intrahepatic 3	ABCB4
5	Sitosterolemia 2	ABCG5
5	Sitosterolemia 1	ABCG8
6	Acyl-CoA dehydrogenase, medium chain, deficiency of	ACADM
7	VLCAD deficiency	ACADVL
8	Alpha-methylacetoacetic aciduria	ACAT1
9	Adenosine Deaminase Deficiency	ADA
10	Joubert Syndrome 3	AHI1
11	Sjogren-Larsson syndrome	ALDH3A2
12	Fructose intolerance, hereditary	ALDOB
13	Hypophosphatasia, infantile	ALPL
14	Hypophosphatasia, childhood	ALPL
15	Glycine encephalopathy1	GLDC
15	Glycine encephalopathy 2	AMT
16	Argininemia	ARG1
17	Metachromatic leukodystrophy	ARSA
18	Mucopolysaccharidosis type VI (Maroteaux-Lamy)	ARSB
19	Argininosuccinic aciduria	ASL
20	Canavan Disease	ASPA
21	Citrullinemia	ASS1
22	Wilson Disease	ATP7B
23	Maple syrup urine disease, type Ia	BCKDHA
24	Maple syrup urine disease, type Ib	BCKDHB
25	Biotinidase Deficiency	BTD
26	Albinism, oculocutaneous, type VII	LRMDA
27	Joubert Syndrome 17	CPLANE1
28	Muscular dystrophy, limb-girdle, autosomal recessive 1	CAPN3
29	Homocystinuria, B6-responsive and nonresponsive types	CBS
30	Joubert Syndrome 9	CC2D2A
31	Joubert Syndrome 5	CEP290
32	Meckel Syndrome 4	CEP290
33	Cystic Fibrosis	CFTR
34	Ceroid lipofuscinosis, neuronal, 3	CLN3
35	Ceroid lipofuscinosis, neuronal, 5	CLN5
36	Ceroid lipofuscinosis, neuronal, 6B (Kufs type)	CLN6

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37	Ceroid lipofuscinosis, neuronal, 6A	CLN6
38	Epidermolysis bullosa, junctional 4, intermediate	COL17A1
39	Alport syndrome 3B, autosomal recessive	COL4A3
39	Alport syndrome 2, autosomal recessive	COL4A4
40	Epidermolysis bullosa dystrophica, autosomal recessive	COL7A1
41	Carbamoylphosphate Synthetase I Deficiency	CPS1
42	Cystinosis, nephropathic	CTNS
43	Maple syrup urine disease, type II	DBT
44	Smith-Lemli-Opitz syndrome	DHCR7
45	Dihydrolipoamide dehydrogenase deficiency	DLD
46	Duchenne Muscular Dystrophy	DMD
47	Immunodeficiency-centromeric instability-facial anomalies syndrome 1	DNMT3B
48	Muscular dystrophy, limb-girdle, autosomal recessive 2	DYSF
49	Ectodermal dysplasia 1, hypohidrotic, X-linked	EDA
50	Glutaric acidemia IIA	ETFA
51	Glutaric acidemia IIB	ETFB
52	Glutaric acidemia IIC	ETFDH
53	Ellis-van Creveld Syndrome	EVC2
54	Hemophilia B	F9
55	Tyrosinemia, type I	FAH
56	Fanconi anemia, complementation group A	FANCA
57	Fanconi anemia, complementation group C	FANCC
58	Fanconi anemia, complementation group D2	FANCD2
59	Fanconi anemia, complementation group G	FANCG
60	Fanconi anemia, complementation group I	FANCI
61	Glycogen storage disease Ia	G6PC1
62	Glycogen storage disease II	GAA
63	Krabbe Disease	GALC
64	Mucopolysaccharidosis IVA	GALNS
65	Galactosemia	GALT
66	Glycogen storage disease IV	GBE1
67	Glutaricaciduria, type I	GCDH
68	Deafness, autosomal recessive 1A	GJB2
69	Fabry Disease	GLA
70	Mucopolysaccharidosis type IVB (Morquio)	GLB1
71	Mucopolysaccharidosis type IIID	GNS
72	Ocular albinism, type I, Nettleship-Falls type	GPR143
73	Hyperinsulinemic hypoglycemia, familial, 4	HADH
74	LCHAD deficiency	HADHA
75	Mitochondrial trifunctional protein deficiency 1	HADHA
75	Mitochondrial trifunctional protein deficiency 2	HADHB
76	Thalassemia, alpha-	HBA1

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76	Thalassemia, alpha-	HBA2
76	Thalassemia, alpha-	HBA1/HBA2
77	Thalassemia, beta	HBB
78	Sickle cell disease	HBB
79	Tay-Sachs Disease	HEXA
80	Mucopolysaccharidosis type IIIC (Sanfilippo C)	HGSNAT
81	Holocarboxylase synthetase deficiency	HLCS
82	HMG-CoA synthase-2 deficiency	HMGCS2
83	Hermansky-Pudlak Syndrome 1	HPS1
84	Hermansky-Pudlak Syndrome 3	HPS3
85	Mucopolysaccharidosis II	IDS
86	Mucopolysaccharidosis Ih	IDUA
87	Mucopolysaccharidosis Ih/s	IDUA
88	Mucopolysaccharidosis Is	IDUA
89	Severe combined immunodeficiency, X-linked	IL2RG
90	Isovaleric Acidemia	IVD
91	Hyperinsulinemic hypoglycemia, familial, 2	KCNJ11
92	LAMA3-Related Junctional Epidermolysis Bullosa	LAMA3
93	LAMB3-Related Junctional Epidermolysis Bullosa	LAMB3
94	LAMC2-Related Junctional Epidermolysis Bullosa	LAMC2
95	Osteoporosis-pseudoglioma syndrome	LRP5
96	Mannosidosis, alpha-, types I and II	MAN2B1
97	Methylmalonyl-CoA epimerase deficiency	MCEE
98	Ceroid lipofuscinosis, neuronal, 7	MFSD8
99	Megalencephalic Leukoencephalopathy with Subcortical Cysts 1	MLC1
100	Methylmalonic aciduria, vitamin B12-responsive, cblA type	MMAA
101	Methylmalonic aciduria, vitamin B12-responsive, cblB type	MMAB
102	Methylmalonic aciduria and homocystinuria, cblC type	MMACHC
103	Methylmalonic aciduria and homocystinuria, cblD type	MMADHC
104	Methylmalonic aciduria, mut(0) type	MMUT
105	Molybdenum Cofactor Deficiency A	MOCS1
106	Myopathy, centronuclear, X-linked	MTM1
107	Homocystinuria-megaloblastic anemia, cblG complementation type	MTR
108	Homocystinuria-megaloblastic anemia, cbl E type	MTRR
109	Mucopolysaccharidosis type IIIB (Sanfilippo B)	NAGLU
110	Niemann-Pick disease, type C1	NPC1
111	Niemann-pick disease, type C2	NPC2
112	Nephronophthisis 3	NPHP3
113	Nephrotic syndrome, type 1	NPHS1
114	Adrenal hypoplasia, congenital	NR0B1
115	Albinism, oculocutaneous, type II	OCA2

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116	Ornithine Transcarbamylase Deficiency	OTC
117	Phenylketonuria	PAH
118	Propionicacidemia	PCCA
118	Propionicacidemia	PCCB
119	Peroxisome biogenesis disorder 1A (Zellweger)	PEX1
120	Congenital disorder of glycosylation, type Ia	PMM2
121	Ceroid lipofuscinosis, neuronal, 1	PPT1
122	Hemophagocytic lymphohistiocytosis, familial, 2	PRF1
123	Hyperphenylalaninemia, BH4-deficient, A	PTS
124	Omenn syndrome	RAG1
124	Omenn syndrome	RAG2
125	Severe combined immunodeficiency, B cell-negative	RAG1
125	Severe combined immunodeficiency, B cell-negative	RAG2
126	Muscular dystrophy, limb-girdle, autosomal recessive 3	SGCA
127	Muscular dystrophy, limb-girdle, autosomal recessive 5	SGCG
128	Mucopolysaccharidosis type IIIA (Sanfilippo A)	SGSH
129	Carnitine deficiency, systemic primary	SLC22A5
130	Albinism, oculocutaneous, type VI	SLC24A5
131	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome	SLC25A15
132	Deafness, autosomal recessive 4, with enlarged vestibular aqueduct	SLC26A4
133	Glycogen storage disease Ib	SLC37A4
134	Glycogen storage disease Ic	SLC37A4
135	Albinism, oculocutaneous, type IV	SLC45A2
136	Spinal Muscular Atrophy	SMN1
137	Niemann-Pick disease, type A	SMPD1
138	Niemann-Pick disease, type B	SMPD1
139	Netherton syndrome	SPINK5
140	Hemophagocytic lymphohistiocytosis, familial, 4	STX11
141	Hemophagocytic lymphohistiocytosis, familial, 5, with or without microvillus inclusion disease	STXBP2
142	Osteopetrosis, autosomal recessive 1	TCIRG1
143	Ichthyosis, congenital, autosomal recessive 1	TGM1
144	Cholestasis, progressive familial intrahepatic 4	TJP2
145	Joubert Syndrome 2	TMEM216
146	Meckel Syndrome 2	TMEM216
147	Joubert Syndrome 6	TMEM67
148	Nephronophthisis 11	TMEM67
149	Meckel Syndrome 3	TMEM67
150	COACH syndrome 2	CC2D2A
150	COACH syndrome 1	TMEM67
151	Ceroid lipofuscinosis, neuronal, 2	TPP1

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152	Oculocutaneous Albinism Type 1	TYR
153	Albinism, oculocutaneous, type III	TYRP1
154	Hemophagocytic lymphohistiocytosis, familial, 3	UNC13D
155	Wolfram Syndrome 1	WFS1
156	Acyl-CoA dehydrogenase, short-chain, deficiency of	ACADS
157	Aspartylglucosaminuria	AGA
158	Ataxia-telangiectasia	ATM
159	Bloom syndrome	BLM
160	Hemolytic anemia, G6PD deficient (favism)	G6PD
161	Galactokinase deficiency with cataracts	GALK1
162	Dysautonomia, familial	ELP1
163	3-Methylcrotonyl-CoA carboxylase 1 deficiency	MCCC1
164	3-Methylcrotonyl-CoA carboxylase 2 deficiency	MCCC2
165	Mucolipidosis IV	MCOLN1
166	Familial Mediterranean fever, AR	MEFV
167	Nemaline myopathy 2, autosomal recessive	NEB
168	Polycystic kidney disease 4, with or without hepatic disease	PKHD1
169	Alpha- 1 antitrypsin deficiency	SERPINA1
170	Gitelman syndrome	SLC12A3
171	Spastic paraplegia 11, autosomal recessive	SPG11
172	Segawa syndrome, recessive	TH

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