Gene and phenotype list: Genes4Life Plus

#	Phenotype	Gene(s)
	Syndromes with intellectual disability	
	Multiple congenital abnormalities with intellectual disability	
1	Achalasia-addisonianism-alacrimia syndrome	AAAS
2	Al Kaissi syndrome	CDK10
3	Athabaskan brainstem dysgenesis syndrome	HOXA1
4	Arthrogryposis, intellectual disability, and seizure disorder	SLC35A3
5	3MC syndrome	COLEC11, MASP1
6	Bardet-Biedl syndrome	ARL6, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, LZTFL1, MKKS, MKS1, SDCCAG8, TTC8
7	Basel-Vanagait-Smirin-Yosef syndrome	MED25
8	Behr syndrome	OPA1
9	Boucher-Neuhauser syndrome	PNPLA6
10	Bosley-Salih-Alorainy syndrome	HOXA1
11	Brunner syndrome	MAOA
12	Goldberg-Shprintzen megacolon syndrome	KIFBP
13	Borjeson-Forssman-Lehmann syndrome	PHF6
14	Bloom syndrome	BLM
15	Partington syndrome	ARX
16	Pitt-Hopkins-like syndrome	CNTNAP2
17	Polyhydramnios, megalencephaly, and symptomatic epilepsy	STRADA
18	PERCHING syndrome	KLHL7
19	Shaheen syndrome	COG6
20	Growth retardation, intellectual developmental disorder, hypotonia, and hepatopathy	IARS1
21	Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia (CAGSSS)	IARS2
22	Carey-Fineman-Ziter syndrome	MYMK
23	Cerebellofaciodental syndrome	BRF1
24	Craniofacial dysmorphism, skeletal anomalies, and intellectual disability syndrome	TMCO1
25	CHIME syndrome	PIGL
26	COACH syndrome	CC2D2A, RPGRIP1L, TMEM67
27	Cockayne syndrome	ERCC4, ERCC5, ERCC6, ERCC8
28	Cohen syndrome	VPS13B
29	Cerebrooculofacioskeletal syndrome (COFS)	ERCC2, ERCC6
30	Coffin-Lowry syndrome	RPS6KA3
31	Cowchock syndrome	AIFM1
32	De Sanctis-Cacchione syndrome	ERCC6

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33	Developmental delay with short stature, dysmorphic features, and sparse hair	DPH1
34	Donnai-Barrow syndrome	LRP2
35	DOOR syndrome	TBC1D24
36	XFE progeroid syndrome	ERCC4
37	Desmosterolosis	DHCR24
38	Dyggve-Melchior-Clausen disease	DYM
39	Elsahy-Waters syndrome	CDH11
40	Fragile X syndrome	FMR1
41	Frontometaphyseal dysplasia	FLNA
42	Galloway-Mowat syndrome	WDR73, OSGEP
43	Gillespie syndrome	ITPR1
44	Griscelli syndrome	RAB27A
45	HSAN2D syndrome	SCN9A
46	Hypoparathyroidism-retardation-dysmorphism syndrome	TBCE
47	Hypotonia, infantile, with psychomotor retardation and characteristic facies	TBCK, UNC80, NALCN
48	Jawad syndrome	RBBP8
49	Jensen syndrome	TIMM8A
50	Johanson-Blizzard syndrome	UBR1
51	IFAP syndrome with or without BRESHECK syndrome	MBTPS2
52	Immunoskeletal dysplasia with neurodevelopmental abnormalities	EXTL3
53	Infantile liver failure syndrome	LARS1
54	Intellectual developmental disorder with dysmorphic facies, seizures, and distal limb anomalies	OTUD6B
55	Intellectual developmental disorder with cardiac arrhythmia	GNB5
56	Lujan-Fryns syndrome	MED12
57	Kohlschutter-tonz syndrome	ROGDI
58	Ohdo syndrome	MED12
59	Opitz-Kaveggia syndrome	MED12
60	Opitz GBBB syndrome	MID1
61	Oliver-McFarlane syndrome	PNPLA6
62	Mosaic variegated aneuploidy syndrome	BUB1B
63	MEHMO syndrome	EIF2S3
64	Muscular dystrophy, congenital, with cataracts and intellectual disability	INPP5K
65	Nijmegen breakage syndrome	NBN, RAD50
66	Nance-Horan syndrome	NHS
67	Neurodevelopmental disorder with brain anomalies and additional features	PLAA, PRUNE1, VARS1, WDR45B
68	Multiple congenital anomalies-hypotonia-seizures syndrome	PIGA, PIGN, PIGT
69	Renpenning syndrome	PQBP1
70	Salt and pepper developmental regression syndrome	ST3GAL5
71	Seckel syndrome	ATR, CENPJ, CEP152, RBBP8

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72	SESAME syndrome	KCNJ10
73	Smith-Lemli-Opitz syndrome	DHCR7
74	Spastic paraplegia and psychomotor retardation with or without seizures	HACE1
75	LIG4 syndrome	LIG4
76	Wieacker-Wolff syndrome	ZC4H2
77	Alacrima, achalasia, and intellectual disability syndrome	GMPPA
78	Chudley-McCullough syndrome	GPSM2
79	Growth retardation, developmental delay, coarse facies, and early death	FTO
80	Martsolf syndrome	RAB3GAP2
81	Pierson syndrome	LAMB2
82	Hemorrhagic destruction of the brain with subependymal calcification and cataracts	JAM3
83	Hennekam lymphangiectasia-lymphedema syndrome	CCBE1, FAT4
84	Perlman syndrome	DIS3L2
85	Temtamy preaxial brachydactyly syndrome	CHSY1
86	Filippi syndrome	CKAP2L
87	Fraser syndrome	FRAS1, FREM2
88	Orofaciodigital syndrome	CPLANE1, C2CD3, DDX59, SERPINH1, TMEM107, TCTN3
89	Roberts syndrome	ESCO2
90	SC phocomelia syndrome	ESCO2
91	Warburg micro syndrome	RAB18, RAB3GAP1, RAB3GAP2
92	Woodhouse-Sakati syndrome	DCAF17
93	Van Maldergem syndrome	DCHS1, FAT4
94	Warsaw breakage syndrome	DDX11
95	You-Hoover-Fong syndrome	TELO2
	Syndromic microcephaly	
96	Microcephaly, epilepsy, and diabetes syndrome	IER3IP1
97	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy	QARS1
98	Microcephaly-capillary malformation syndrome	STAMBP
99	Microcephaly, short stature, and impaired glucose metabolism	TRMT10A
100	Microcephaly, short-stature and endocrine dysfunction	XRCC4
101	Microcephaly, short stature, and limb abnormalities	DONSON
102	Microcephaly and chorioretinopathy	TUBGCP4, TUBGCP6
103	Microcephaly, seizures, spasticity, and brain calcification	PCDH12
	X-linked syndromic intellectual disability	
104	Turner type	HUWE1
105	Claes-Jensen type	KDM5C
106	Christianson type	SLC9A6
107	Siderius type	PHF8
108	Type 14	UPF3B

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109	CK syndrome	NSDHL
110	Snyder-Robinson type	SMS
111	Nascimento type	UBE2A
112	Raymond type	ZDHHC9
113	Intellectual disability, truncal obesity, retinal dystrophy, and micropenis	INPP5E
114	Intellectual disability, X-linked, with cerebellar hypoplasia and distinctive facial appearance	OPHN1
	Syndromic brain malformations	
115	MASA syndrome	L1CAM
116	CRASH syndrome	L1CAM
117	Agenesis of the corpus callosum with peripheral neuropathy (Andermann syndrome)	SLC12A6
118	Acrocallosal syndrome	KIF7
119	Proud syndrome	ARX
120	Temtamy syndrome	C12orf57
121	Cerebroretinal microangiopathy with calcifications and cysts	CTC1
122	Vici syndrome	EPG5
123	Proliferative vasculopathy and hydraencephaly-hydrocephaly syndrome	FLVCR2
124	Neurodevelopmental disorder and structural brain anomalies with or without seizures and spasticity	PTPN23 *Not screened in WA, QLD and SA
	Syndromic skin conditions with intellectual disability	
125	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome	SNAP29
126	Adams-Oliver syndrome	DOCK6, EOGT
	Syndromic vision conditions with intellectual disability	
127	Peter's plus syndrome	B3GLCT
128	Congenital cataracts, hearing loss, and neurodegeneration	SLC33A1
129	Knobloch syndrome	COL18A1
130	Lowe syndrome	OCRL
131	Kaufman oculocerebrofacial syndrome	UBE3B
132	Kahrizi syndrome	SRD5A3
133	Optic atrophy with or without ataxia, intellectual disability, and seizures	RTN4IP1
134	Norrie disease	NDP
	Syndromic growth conditions with intellectual disability	
135	Simpson-Golabi-Behmel syndrome	OFD1, GPC3
	Severe, lethal, neonatal syndromes	
136	Meckel syndrome	CC2D2A, CEP290, MKS1, NPHP3, RPGRIP1L, TMEM216, TMEM231, TMEM67
137	Alkuraya-Kucinskas syndrome	KIAA1109
138	Bowen-Conradi syndrome	EMG1
139	Fetal akinesia deformation sequence	RAPSN
140	Lethal congenital contracture syndrome	CNTNAP1, GLE1, GLDN
141	Ventriculomegaly with cystic kidney disease	CRB2

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142	Hydrolethalus syndrome	HYLS1, KIF7
143	TARP syndrome	RBM10
144	Rigidity and multifocal seizure syndrome, lethal neonatal	BRAT1
	Syndromes without intellectual disability	
	Multiple pterygium syndrome	
145	Lethal type	CHRNA1, RIPK4
146	Escobar syndrome	CHRNG
	Multiple congenital abnormalities	
147	Burn-McKeown syndrome	TXNL4A
148	Bifid nose with or without anorectal and renal anomalies	FREM1
149	Crisponi syndrome	CRLF1, CLCF1
150	McKusick-Kaufman syndrome	MKKS
151	Shwachman-Diamond syndrome	SBDS
152	Split-hand foot malformation	WNT10B
153	Werner syndrome	WRN
154	VACTERL association X-linked	ZIC3
155	Lipodystrophy, congenital generalized	BSCL2, CAVIN1
156	Wolfram syndrome	CISD2, WFS1
157	Urofacial syndrome	HPSE2, LRIG2
	Syndromic skin and skeletal conditions	
158	Rothmund-Thomson syndrome	RECQL4
159	Alstrom syndrome	ALMS1
160	GAPO syndrome	ANTXR1
161	HELIX syndrome	CLDN10
162	Haim-Munk syndrome	CTSC
163	Laryngoonychocutaneous syndrome	LAMA3
164	Miller syndrome	DHODH
165	Macrocephaly, alopecia, cutis laxa, and scoliosis	RIN2
166	Mandibuloacral dysplasia with type B lipodystrophy	ZMPSTE24
167	Dyskeratosis congenita	DKC1, RTEL1, WRAP53
168	Papillon-Lefevre syndrome	CTSC
169	Spondyloocular syndrome	XYLT2
170	Treacher-Collins syndrome	POLR1C
171	Schimke immunoosseous dysplasia	SMARCAL1
	Syndromic vision and hearing conditions	
172	Usher syndrome	ADGRV1, CDH23, CLRN1, MYO7A, PCDH15, USH1C, USH1G, USH2A, WHRN
173	Retinitis pigmentosa with skeletal anomalies	CWC27
174	Jalili syndrome	CNNM4
	Syndromic vision and renal conditions	
175	Senior-Loken syndrome	CEP290, NPHP1, NPHP4, SDCCAG8, IQCB1, WDR19
	Mitochondrial conditions	

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	Conditions affecting multiple body systems	
176	Combined oxidative phosphorylation deficiency	AARS2, C12orf65, CARS2, FARS2, ELAC2, GFM1, GTPBP3, MTFMT, MTO1, NARS2, RMND1, TSFM, TUFM, VARS2, TRIT1, EARS2
	Leigh and Leigh-like syndrome	
177	Mitochondrial complex I deficiency	ACAD9, FOXRED1, NUBPL, NDUFA1, NDUFAF2, NDUFAF5, NDUFAF6, NDUFA10, NDUFA11, NDUFS6, NDUFS4, NDUFS2, NDUFS7, NDUFS8, NDUFS1, NDUFV1, NDUFV2
178	Leigh syndrome due to cytochrome c oxidase deficiency	COX15
179	Leigh syndrome, French Canadian type	LRPPRC
	Other mitochondrial conditions	
180	Mitochondrial complex II deficiency	SDHAF1
181	Mitochondrial complex III deficiency	BCS1L, LYRM7, TTC19, UQCRQ
182	Mitochondrial complex IV deficiency	COX10, COA8, COX20, SURF1, PET100
183	Mitochondrial complex V deficiency	TMEM70
184	Mitochondrial DNA depletion syndrome	DGUOK, FBXL4, MGME1, MPV17, RRM2B, SUCLA2, SUCLG1, TK2, TWNK, TYMP
185	Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE)	TWNK
186	Multiple mitochondrial dysfunctions syndrome	BOLA3, IBA57, ISCA2, NFU1
187	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2	COX15, SCO2
188	Sideroblastic anaemia with B-cell immunodeficiency, periodic fevers, and developmental delay	TRNT1
189	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation	DARS2
190	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis (HUPRA syndrome)	SARS2
191	HSD10 disease	HSD17B10
192	Mohr-Tranebjaerg syndrome	TIMM8A
193	Mitochondrial neurodevelopmental disorder, with abnormal movements and lactic acidosis	WARS2
194	Myopathy, lactic acidosis, and sideroblastic anaemia	PUS1, LARS2, YARS2
195	Myopathy, mitochondrial, and ataxia	MSTO1 *Not screened in WA, QLD and SA
196	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency	ECHS1
	Lysosomal storage disorders	
	Mannosidosis	
197	Alpha	MAN2B1
198	Beta	MANBA
	Mucopolysaccharidosis	
199	Mucopolysaccharidosis	GALNS, GNS, GUSB, IDS, IDUA
200	Type VI (Maroteaux-Lamy)	ARSB
201	Type IVB (Morquio)	GLB1
202	Type IIIA (Sanfilippo A)	SGSH
203	Type IIIB (Sanfilippo B)	NAGLU
204	Type IIIC (Sanfilippo C)	HGSNAT
	Cystinosis	

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205	Atypical nephropathic	CTNS
206	Nephropathic	CTNS
207	Late-onset juvenile or adolescent nephropathic	CTNS
208	Ocular non-nephropathic	CTNS
	Other lysosomal storage disorders	
209	Galactosialidosis	CTSA
210	Yunis-Varon syndrome	FIG4
211	Fucosidosis	FUCA1
212	Farber lipogranulomatosis	ASAH1
213	Glycogen storage disease (Pompe)	GAA
214	Geleophysic dysplasia	ADAMTSL2
215	Krabbe disease	GALC, PSAP
216	Fabry disease	GLA
217	GM1-gangliosidosis	GLB1
218	GM2-gangliosidosis	HEXA, GM2A
219	Metachromatic leukodystrophy	ARSA, PSAP
220	Mucolipidosis	GNPTAB, GNPTG, MCOLN1
221	Polyglucosan body myopathy 1 with or without immunodeficiency	RBCK1
222	Tay-Sachs disease	HEXA
223	Sandhoff disease	HEXB
224	Chediak-Higashi syndrome	LYST
225	Aspartylglucosaminuria	AGA
226	Schindler disease	NAGA
227	Sialidosis	NEU1
228	Combined SAP deficiency	PSAP
229	Marinesco-Sjogren syndrome	SIL1
230	Sialic acid storage disorder	SLC17A5
231	Niemann-Pick disease	NPC1, NPC2, SMPD1
	Metabolic conditions	
	Peroxisome biogenesis disorders	
232	Including Zellweger syndrome, neonatal adrenoleukodystrophy and infantile Refsum disease	PEX1, PEX10, PEX11B, PEX12, PEX13, PEX16, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7
	Organic acidemias	
233	Argininosuccinic aciduria	ASL
234	3-methylglutaconic aciduria	AUH, CLPB, DNAJC19, HTRA2, OPA3, SERAC1
235	D-2-hydroxyglutaric aciduria	D2HGDH
236	Glutaricaciduria	GCDH
237	D-glyceric aciduria	GLYCTK
238	L-2-hydroxyglutaric aciduria	L2HGDH
239	Methylmalonic aciduria	MMADHC, MMUT
240	Methylmalonic aciduria and homocystinuria	LMBRD1, MMACHC, MMADHC
241	Alpha-methylacetoacetic aciduria	ACAT1

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242	Methylmalonic aciduria, vitamin B12-responsive	MMAA, MMAB
243	Mevalonic aciduria	MVK
244	Combined D-2- and L-2-hydroxyglutaric aciduria	SLC25A1
245	Isovaleric acidemia	IVD
246	Glutaric acidemia	ETFA, ETFB, ETFDH
	Other metabolic conditions	
247	Adenylosuccinase deficiency	ADSL
248	Arts syndrome	PRPS1
249	Chanarin-Dorfman syndrome	ABHD5
250	Galactosemia	GALT*
		Not screened in WA, QLD and SA
251	Glycogen storage disease	AGL, G6PC, GYS2, GBE1, LDHA, PFKM, SLC37A4
252	GABA-transaminase deficiency	ABAT
253	Fanconi-Bickel syndrome	SLC2A2
254	Hyperinsulinemic hypoglycemia	ABCC8, HADH, KCNJ11
255	Hyperoxaluria	AGXT
256	Hypermanganesemia with dystonia	SLC39A14
257	Succinic semialdehyde dehydrogenase deficiency	ALDH5A1
258	Fructose intolerance	ALDOB
259	Congenital disorders of glycosylation	ALG1, ALG11, ALG12, ALG3, ALG6, ALG8, ALG9, CCDC115, COG6, COG7, DOLK, DPAGT1, MGAT2, MPI, PGM1, PMM2, RFT1, SLC39A8, SSR4, SRD5A3, TMEM165
260	Congenital disorder of deglycosylation	NGLY1
261	Glycine encephalopathy	AMT, GLDC
262	Glycosylphosphatidylinositol biosynthesis defect	GPAA1
263	Argininemia	ARG1
264	Asparagine synthetase deficiency	ASNS
265	Canavan disease	ASPA
266	Citrullinemia	ASS1, SLC25A13
267	Chylomicron retention disease	SAR1B
268	Menkes disease and occipital horn syndrome	ATP7A
269	Maple syrup urine disease	BCKDHA, BCKDHB, DBT
270	Branched-chain ketoacid dehydrogenase kinase deficiency	BCKDK
271	GRACILE syndrome	BCS1L
272	Homocystinuria	MMADHC, MTHFR, MTR, MTRR
273	Lysinuric protein intolerance	SLC7A7
274	Proteinuria	CLCN5
275	Prolidase deficiency	PEPD
276	Hypomagnesemia	CLDN19, SLC30A10, TRPM6
277	Coenzyme Q10 deficiency	COQ2, COQ4, COQ6, COQ8A
278	Carbamoylphosphate synthetase I deficiency	CPS1
279	CPT 2 deficiency	CPT1A, CPT2

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280	Methemoglobinemia	CYB5R3
281	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration	TANGO2
282	Lipid storage myopathy due to flavin adenine dinucleotide synthetase deficiency	FLAD1
283	Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency	ACADM *Not screened in WA, QLD and SA
284	Peroxisomal acyl-CoA oxidase deficiency	ACOX1
285	17-alpha-hydroxylase deficiency	CYP17A1
286	17,20-lyase deficiency	CYP17A1
287	Cerebrotendinous xanthomatosis	CYP27A1
288	Aromatic L-amino acid decarboxylase deficiency	DDC
289	Dihydrolipoamide dehydrogenase deficiency	DLD
290	Wolcott-Rallison syndrome	EIF2AK3
291	Hypophosphatemic rickets	ENPP1
292	Hyperphosphatasia with intellectual disability syndrome	PIGV, PIGO, PGAP2, PGAP3
293	Ethylmalonic encephalopathy	ETHE1
294	Tyrosinemia	FAH, HPD
295	Fructose-1,6-bisphosphatase deficiency	FBP1
296	Fumarase deficiency	FH
297	Glutamate formiminotransferase deficiency	FTCD
298	Cerebral creatine deficiency syndrome	GAMT, GATM, SLC6A8
299	Gaucher disease	GBA, PSAP
300	Glycerol kinase deficiency	GK
301	Molybdenum cofactor deficiency	GPHN, MOCS1, MOCS2
302	Glutathione synthetase deficiency	GSS
303	3-hydroxyacyl-CoA dehydrogenase deficiency	HADH
304	LCHAD deficiency	HADHA
305	Trifunctional protein deficiency	HADHA, HADHB
306	Hemochromatosis	HAMP, HJV
307	3-hydroxyisobutryl-CoA hydrolase deficiency	HIBCH
308	Holocarboxylase synthetase deficiency	HLCS
309	HMG-CoA lyase deficiency	HMGCL
310	HMG-CoA synthase-2 deficiency	HMGCS2
311	Lesch-Nyhan syndrome	HPRT1
312	D-bifunctional protein deficiency	HSD17B4
313	Leprechaunism	INSR
314	Norum disease	LCAT
315	Lactate dehydrogenase-B deficiency	LDHB
316	Familial hypercholesterolemia	LDLR, LDLRAP1
317	Pyruvate dehydrogenase lipoic acid synthetase deficiency	LIAS
318	Cholesteryl ester storage disease	LIPA
319	Wolman disease	LIPA

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320	Lipoyltransferase 1 deficiency	LIPT1
321	Lipoprotein lipase deficiency	LPL
322	Malonyl-CoA decarboxylase deficiency	MLYCD
323	Abetalipoproteinemia	MTTP
324	N-acetylglutamate synthase deficiency	NAGS
325	N-terminal acetyltransferase deficiency	NAA10
326	Ornithine transcarbamylase deficiency	OTC
327	Phenylketonuria (PKU)	PAH
328	Pyruvate carboxylase deficiency	PC
329	Hyperphenylalaninemia	PTS, QDPR, DNAJC12
330	Propionicacidemia	PCCA, PCCB
331	Proprotein convertase 1 deficiency	PCSK1
332	Pyruvate dehydrogenase deficiency	PDHA1, PDHB, PDP1
333	Phosphoglycerate kinase 1 deficiency	PGK1
334	Phosphoglycerate dehydrogenase deficiency	PHGDH
335	Refsum disease	PHYH
336	Pyruvate kinase deficiency	PKLR
337	Plasminogen deficiency	PLG
338	Dysplasminogenemia	PLG
339	Pyridoxamine 5'-phosphate oxidase deficiency	PNPO
340	Phosphoribosylpyrophosphate synthetase superactivity	PRPS1
341	Phosphoserine phosphatase deficiency	PSPH
342	Neu-Laxova syndrome	PHGDH, PSAT1
343	Riboflavin transport deficiency syndrome	SLC52A2, SLC52A3
344	Tumoral calcinosis, normophosphatemic	SAMD9
345	Lathosterolosis	SC5D
346	Emphysema-cirrhosis, due to AAT deficiency	SERPINA1
347	Hemorrhagic diathesis due to antithrombin Pittsburgh	SERPINA1
348	Monocarboxylate transporter 1 deficiency	SLC16A1
349	Thiamine metabolism dysfunction syndrome	SLC19A2, SLC19A3, SLC25A19, TPK1
350	Carnitine deficiency	SLC22A5
351	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome	SLC25A15
352	Acrodermatitis enteropathica	SLC39A4
353	Multiple sulfatase deficiency	SUMF1
354	Salla disease	SLC17A5
355	Sjogren-Larsson syndrome	ALDH3A2
356	Sulfite oxidase deficiency	SUOX
357	Transaldolase deficiency	TALDO1
358	Barth syndrome	TAZ
359	Adrenocorticotropic hormone deficiency	TBX19
360	Transcobalamin II deficiency	TCN2

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361	Hemolytic anaemia due to triosephosphate isomerase deficiency	TPI1
362	Crigler-Najjar syndrome	UGT1A1
363	Orotic aciduria	UMPS
364	VLCAD deficiency	ACADVL
365	Wilson disease	ATP7B
	Endocrine conditions	
	Congenital adrenal hyperplasia*	
366	Severe salt wasting type	CYP11A1, CYP11B2, NR0B1, POU1F1, PROP1, HSD3B2
367	Lipoid type	STAR
	**Excludes 21-hydroxylase deficiency, as the CYP21A2 gene is not screened for technical reasons*	
	Diabetes mellitus	
368	Neonatal, with congenital hypothyroidism	GLIS3
369	Insulin-resistant, with acanthosis nigricans	INSR
	Other endocrine conditions	
370	Disordered steroidogenesis due to cytochrome P450 oxidoreductase	POR
371	Glucocorticoid deficiency	MC2R, MRAP, NNT
372	Growth hormone deficiency with pituitary anomalies	HESX1
373	Hyperparathyroidism, neonatal severe	CASR
374	Hypothryoidism, congenital	TSHB
375	Insulin-like growth factor resistance	IGF1R
376	Laron syndrome	GHR
377	Obesity, morbid, due to leptin deficiency	LEP
378	Pituitary hormone deficiency	HESX1, LHX3
379	Proopiomelanocortin (POMC) deficiency	POMC
380	Rabson-Mendenhall syndrome	INSR
	Neurological conditions	
	White matter disorders	
381	Adrenoleukodystrophy	ABCD1
382	Aicardi-Goutieres syndrome	ADAR, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, TREX1
383	Leukodystrophy, hypomyelinating	AIMP1, FAM126A, GJC2, HSPD1, POLR3A, POLR3B, PYCR2, RARS1, UFM1, VPS11
384	Leukoencephalopathy with ataxia	CLCN2
385	Leukoencephalopathy with vanishing white matter	EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5
386	Leukoencephalopathy, cystic, without megalencephaly	RNASET2
387	Megalencephalic leukoencephalopathy with subcortical cysts	HEPACAM, MLC1
388	Hypomyelination with brainstem and spinal cord involvement and leg spasticity (HBSL)	DARS1
389	Pelizaeus-Merzbacher disease	PLP1
	Congenital brain malformations	
390	Pontocerebellar hypoplasia	AMPD2, CLP1, EXOSC3, EXOSC8, RARS2, SEPSECS, TBC1D23, TOE1, TSEN2, TSEN54, VPS53, VRK1
391	Lissencephaly	ARX, KATNB1, LAMB1, NDE1, DCX, TMTC3

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392	Joubert syndrome	AHI1, ARL13B, CC2D2A, CEP290, CEP41, CPLANE1, CSPP1, INPP5E, KIF7, NPHP1, OFD1, RPGRIP1L, TCTN2, TCTN3, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67
393	Polymicrogyria	ADGRG1, RTTN
394	Septooptic dysplasia	HESX1
395	Band heterotopia	DCX, EML1
396	Band-like calcification with simplified gyration and polymicrogyria	OCLN
397	Cerebellar hypoplasia and intellectual disability with or without quadrupedal locomotion	VLDLR
398	Periventricular heterotopia with microcephaly	ARFGEF2
399	Poretti-Boltshauser syndrome	LAMA1
400	Cortical malformations, occipital	LAMC3
	Microcephaly	
401	Isolated	ASPM, CDK5RAP2, CENPJ, CEP152, CIT, KIF14, KNL1, MCPH1, MFSD2A, MED17, PNKP, SLC25A19, STIL, WDR62, ZNF335
	Hydrocephalus	
402	Non-syndromic hydrocephalus	L1CAM, CCDC88C, MPDZ
403	Hydrocephalus with congenital idiopathic intestinal pseudoobstruction	L1CAM
404	Hydrocephalus due to aqueductal stenosis	L1CAM
405	Hydrocephalus with Hirschsprung disease	L1CAM
	Neurodegenerative conditions	
406	Neuronal ceroid lipofuscinoses	CLN3, CLN5, CLN6, CLN8, CTSD, CTSF, MFSD8, PPT1, TPP1
407	Parkinson disease, juvenile-onset	DNAJC6, FBXO7, PLA2G6, ATP13A2
408	Encephalopathy, progressive	BSCL2, TBCD, NAXE
409	Moyamoya disease	GUCY1A1
410	Neurodegeneration with brain iron accumulation	C19orf12, PANK2, PLA2G6
411	Neurodegeneration due to cerebral folate transport deficiency	FOLR1
412	Neurodegeneration with ataxia, dystonia, and gaze palsy, childhood-onset	SQSTM1
413	Neurodegeneration, stress-induced, with variable ataxia and seizures	ADPRS*
414		***Not screened in WA, QLD and SA**
415	Infantile or childhood-onset striatonigral degeneration	NUP62, VAC14*
416		***Not screened in WA, QLD and SA**
417	PEHO syndrome	ZNHIT3
418	Infantile cerebellar-retinal degeneration	ACO2
419	Infantile neuroaxonal dystrophy 1	PLA2G6
420	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly	SLC1A4
421	Troyer syndrome	SPART
	Ataxias	
422	Ataxia-telangiectasia	ATM, MRE11
423	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia	APTX
424	Ataxia, cerebellar, Cayman type	ATCAY

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425	Ataxia, posterior column, with retinitis pigmentosa	FLVCR1
426	Ataxia-oculomotor apraxia 4	PNKP
427	Ataxia with isolated vitamin E deficiency	TTPA
428	Cerebellar ataxia, cognitive disability, and disequilibrium (CAMRQ)	WDR81, ATP8A2
429	Spastic ataxia	KIF1C, MARS2, NKX6-2, SACS
430	Spinocerebellar ataxia	GRM1, PMPCA, SETX, SNX14, STUB1, SCYL1, TPP1, WWOX
	Movement disorders	
431	Choreoacanthocytosis	VPS13A
432	Dystonia	COL6A3, PRKRA*
		***Not screened in WA, QLD and SA**
433	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency	SPR
434	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia	GCH1
435	Parkinsonism-dystonia, infantile	SLC6A3
436	Segawa syndrome	TH
	Epilepsy	
437	Epilepsy, pyridoxine-dependent	ALDH7A1
438	Epileptic encephalopathy, early infantile	AP3B2, ARV1, ARX, ARHGEF9, DENND5A, FRRS1L, MECP2, SLC13A5, SLC12A5, SLC25A22, TBC1D24, UBA5, WWOX
439	Epilepsy, progressive myoclonic	CSTB, EPM2A, GOSR2, KCTD7, NHLRC1, PRICKLE1, SCARB2, TBC1D24
440	Hyperekplexia	ATAD1, SLC6A5
441	Epilepsy, early-onset, vitamin B6-dependent	PLPBP
442	Epilepsy, X-linked, with variable learning disabilities and behaviour disorders	SYN1
443	Epilepsy, hearing loss, and intellectual disability syndrome	SPATA5
444	Cortical dysplasia-focal epilepsy syndrome	CNTNAP2
445	Amish infantile epilepsy syndrome	ST3GAL5
	Intellectual disability	
446	Non-syndromic intellectual disability, X-linked	AP1S2, ARX, ATRX, BRWD3, CASK, CLCN4, CUL4B, DLG3, FTSJ1, GDI1, HCFC1, IL1RAPL1, IQSEC2, MECP2, NEXMIF, NLGN4X, PAK3, RAB39B, RLIM, SLC16A2, SYP, THOC2, TSPAN7, USP9X, ZNF711
447	Non-syndromic intellectual disability, autosomal recessive	ADAT3, CC2D1A, ELP2, GPT2, HERC2, KPTN, LINS1, MAN1B1, MBOAT7, MED23, METTL23, NSUN2, PGAP1, PIGG, TRAPPC9, TTI2, TUSC3
448	Intellectual developmental disorder with microcephaly and short stature	PUS7 *Not screened in WA, QLD and SA
	Other neurological conditions	
449	Sensorineural hearing loss, premature ovarian failure (females), variable intellectual disability, spasticity, ataxia	CLPP
	Cutaneous conditions	
	Ichthyosis	
450	Ichthyosis, congenital, autosomal recessive	ABCA12, ALOX12B, ALOXE3, CERS3, CYP4F22, NIPAL4, TGM1
451	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis	CLDN1
452	Epidermolytic hyperkeratosis	KRT10

	Cutis laxa	
453	Cutis laxa, autosomal recessive	ALDH18A1, ATP6V0A2, EFEMP2, FBLN5, LTBP4, PYCR1
	Ectodermal dysplasia	
454	Ectodermal dysplasia, ectrodactyly and macular dystrophy	CDH3
455	Ectodermal dysplasia	EDA, EDAR, IKBKG, KRT85
	Cutaneous conditions affecting the nervous system	
456	Xeroderma pigmentosum	ERCC2, ERCC4, ERCC5, XPA, XPC
	Other cutaneous conditions	
457	Kindler syndrome	FERMT1
458	Epidermolysis bullosa	COL7A1, COL17A1, DSP, ITGA6, ITGB4, KRT14, KRT5, LAMA3, LAMB3, LAMC2, PLEC
459	Hyaline fibromatosis syndrome	ANTXR2
460	Porokeratosis 3, disseminated superficial actinic	MVK
461	Keratosis linearis with ichthyosis congenital and sclerosing keratoderma	POMP
462	Netherton syndrome	SPINK5
463	Poikilderma with neutropenia	USB1
464	Restrictive dermopathy, lethal	LMNA, ZMPSTE24
465	Triochthiodystrophy	ERCC2, GTF2H5, MPLKIP
466	Transient bullous of the newborn	COL7A1
	Respiratory conditions	
	Surfactant conditions	
467	Surfactant metabolism dysfunction, pulmonary	ABCA3, SFTPB
	Ciliary dyskinesia	
468	Ciliary dyskinesia, primary	OCAD2*, CCDC103, CCDC114, CCDC39, CCDC40, CCNO, DNAAF1, DNAAF3, DNAAF4, DNAAF5, DNAAF6^, GAS8, HYDIN, LRRC6, RSPH1, RSPH4A, RSPH9, SPAG1, ZMYND10
		Formerly known as ARMC4
		^Formerly known as PIH1D3
469	Ciliary dyskinesia, primary, with or without situs inversus	DNAH11, DNAH5, DNAI1, DNAI2
	Other respiratory conditions	
470	Cystic fibrosis	CFTR
471	Pulmonary veno-occlusive disease	EIF2AK4
472	Interstitial lung and liver disease	MARS1
	Immunological conditions	
	Chronic granulomatous disease	
473	Deficiency of NCF-1	NCF1
474	Deficiency of NCF-2	NCF2
475	Deficiency of CYBA	CYBA
476	X-linked	CYBB
477	Combined cellular and humoral immune defects with granulomas	RAG1, RAG2
	Complement deficiencies	
478	C1q	C1QA, C1QB, C1QC

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479	C3	C3
480	C5	C5
481	C6	C6
482	C7	C7
483	C8	C8B
484	Factor D	CFD
485	Factor H	CFH
486	Factor I	CFI
	Immunodeficiencies	
487	Immunodeficiency	ATP6AP1, CARD11, CD3D, CTPS1, DOCK2, ICOS, IKBKB, IL12RB1, IL17RA LAT, LRBA, MALT1, ORAI1, PGM3, RORC, STIM1, TYK2
488	Mycobacteriosis	CYBB, IFNGR1, IFNGR2, STAT1
489	Purine nucleoside phosphorylase deficiency	PNP
490	Hyper-IgM	CD40, CD40LG
491	Hyper-IgD syndrome	MVK
492	Hyper-IgE recurrent infection syndrome	DOCK8
493	Centromeric instability-facial anomalies syndrome	DNMT3B, ZBTB24
494	Combined immunodeficiency, moderate	IL2RG
495	Combined immunodeficiency and megaloblastic anaemia with or without hyperhomocysteinemia	MTHFD1
	Neutropenia	
496	Severe, congenital	G6PC3, HAX1, JAGN1, VPS45, WAS
	Severe combined immunodeficiencies	
497	Severe combined immunodeficiency	IL2RG
498	Adenosine deaminase deficiency	ADA
499	With microcephaly, growth retardation, and sensitivity to ionizing radiation	NHEJ1
500	Athabascan type	DCLRE1C
501	B cell-negative	RAG1, RAG2
502	T-cell negative, B-cell/natural killer cell-positive type	IL7R, JAK3
503	Reticular dysgenesis	AK2
	Other immunological conditions	
504	Agammaglobulinemia	BTK, IGHM
505	Autoimmune disease, multisystem, with facial dysmorphism	ITCH
506	Autoinflammation, lipodystrophy, and dermatosis syndrome	PSMB8
507	Bone marrow failure syndrome	ERCC6L2, DNAJC21
508	Bare lymphocyte syndrome	CIITA, RFXAP, TAP1
509	Candidiasis, familial	CARD9
510	Histiocytosis-lymphadenopathy plus syndrome	SLC29A3
511	Hemophagocytic lymphohistiocytosis	PRF1, STX11, STXBP2, UNC13D
512	Hepatic veno-occlusive disease with immunodeficiency	SP110
513	Interleukin 1 receptor antagonist deficiency	IL1RN

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514	Immunodysregulation, polyendocrinopathy, and enteropathy	FOXP3
515	Leukocyte adhesion deficiency	FERMT3, ITGB2
516	Lymphoproliferative syndrome	CD27, ITK, SH2D1A, XIAP
517	MHC class II deficiency, complementation group B	RFXANK
518	Natural killer cell and glucocorticoid deficiency with DNA repair defect	MCM4
519	Platelet abnormalities with eosinophilia and immune-mediated inflammatory disease	ARPC1B
520	Properdin deficiency	CFP
521	Pyogenic bacterial infections, recurrent, due to MYD88 deficiency	MYD88
522	Selective T-cell defect	ZAP70
523	T-cell immunodeficiency, congenital alopecia, and nail dystrophy	FOXN1
524	Darsun syndrome	G6PC3
525	Majeed syndrome	LPIN2
526	Omenn syndrome	DCLRE1C, RAG1, RAG2
527	Wiskott-Aldrich syndrome	WAS
	Gastrointestinal conditions	
	Severe congenital diarrhea	
528	With tufting enteropathy, congenital	EPCAM
529	Secretory chloride, congenital	SLC26A3
530	Secretory sodium, congenital,	SPINT2, SLC9A3
531	Protein-losing enteropathy type	DGAT1
	Hepatic conditions	
532	Cholestasis, progressive familial intrahepatic	ABCB11, ABCB4, ATP8B1, TJP2
533	Hepatic lipase deficiency	LIPC
534	Porphyria	ALAD, UROS
535	Liver failure, transient infantile	TRMU
536	Hypercholanaemia	TJP2
	Other gastrointestinal conditions	
537	Microvillus inclusion disease	MYO5B
538	Bile acid synthesis defect, congenital	AKR1D1, CYP7B1, HSD3B7
539	Congenital short bowel syndrome	CLMP, FLNA
540	Complement hyperactivation, angiopathic thrombosis, and protein-losing enteropathy	CD55
541	Meconium ileus	GUCY2C
542	Mitchell-Riley syndrome	RFX6
543	Chronic atrial and intestinal dysrhythmia	SGO1
544	Inflammatory bowel disease, congenital, severe	IL10RA, IL10RB*
		Not screened in WA, QLD and SA
545	Trichohepatoenteric syndrome	SKIV2L, TTC37
546	Folate malabsorption, hereditary	SLC46A1
547	Gastrointestinal defects and immunodeficiency syndrome	TTC7A
548	Hyperbilirubinemia, familial transient neonatal	UGT1A1

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	Haematological conditions	
	Anaemia	
549	Sideroblastic, with ataxia	ABCB7
550	Anaemia, sideroblastic, pyridoxine-refractory	SLC25A38
551	Dyserythropoietic anaemia	SEC23B
552	Haemolytic anaemia due to hexokinase deficiency	HK1
553	Fanconi anaemia	ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, UBE2T
	Clotting conditions	
554	Hypoprothrombinemia	F2
555	Factor V deficiency	F5
556	Factor VII deficiency	F7
557	Haemophilia A	F8
558	Haemophilia B	F9
559	Afibrinogenemia	FGA, FGB, FGG
560	Dysfibrinogenemia	FGA, FGB, FGG
561	Hypodysfibrinogenemia	FGA, FGB, FGG
562	Hypofibrinogenemia	FGA, FGB, FGG
563	Combined factor V and VIII deficiency	LMAN1, MCFD2
564	Thrombotic thrombocytopenic purpura	ADAMTS13
565	Thrombocytopenia, congenital amegakaryocytic	MPL
566	Thrombophilia	PROC, PROS1
567	von Willebrand disease	VWF
568	Thrombocytopenia, X-linked	WAS
	Other haematological conditions	
569	Vitamin K-dependent clotting factors, combined deficiency of	VKORC1
570	Beta thalassemia	HBB
571	Sickle cell disease	HBB
572	Atransferrinemia	TF
	Cardiovascular conditions	
	Arrhythmias	
573	Ventricular tachycardia, catecholaminergic polymorphic	CASQ2
574	Jervell and Lange-Nielsen syndrome	KCNQ1
575	Ventricular tachycardia, catecholaminergic polymorphic with or without muscle weakness	TRDN
	Cardiomyopathies	
576	Cardiomyopathy, dilated, with woolly hair and keratoderma (Naxos disease)	DSP, JUP
577	Dilated cardiomyopathy	FKTN
	Structural cardiovascular conditions	
578	Arterial calcification of infancy	ENPP1
579	Cardiac valvular dysplasia, X-linked	FLNA
580	Right atrial isomerism	GDF1

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581	Hypoplastic left heart syndrome	GJA1
582	Arterial tortuosity syndrome	SLC2A10
583	Heterotaxy, visceral	ZIC3, MMP21
584	Congenital heart defects	ZIC3
	Other cardiovascular conditions	
585	Sudden cardiac failure, infantile	PPA2
	Renal conditions	
	Syndromic renal conditions	
586	Alport syndrome	COL4A3, COL4A4, COL4A5
587	Dent disease	OCRL, CLCN5
588	Renal tubular acidosis with other abnormalities	ATP6V1B1, SLC4A4, SLC4A1
589	Bartter syndrome	BSND, CLCNKB, KCNJ1, SLC12A1
590	Renal-hepatic-pancreatic dysplasia	NPHP3, NEK8
591	Polycystic kidney and hepatic disease	PKHD1
592	Nephrotic syndrome	COQ8B, DGKE, LAMB2, NPHS1, NPHS2, NUP107, NUP93, PLCE1, SGPL1
	Tubular conditions	
593	Renal tubular dysgenesis	ACE, AGT, REN
594	Renal tubular acidosis	ATP6V0A4
	Other renal conditions	
595	Focal segmental glomerulosclerosis	CRB2
596	Pseudohypoaldosteronism	SCNN1A, SCNN1B
597	Nephronophthisis and related conditions	ANKS6, DCDC2, INVS, MAPKBP1, NPHP1, NPHP3, NPHP4, TMEM67, TTC21B, WDR19
598	Nephrogenic diabetes insipidus	AQP2
	Neuromuscular conditions	
	Atrophy	
600	Spinal muscular atrophy with progressive myoclonic epilepsy	ASAH1
601	Spinal muscular atrophy	SMN1, UBA1
602	Spinal muscular atrophy with congenital bone fractures	ASCC1
	Arthrogryposis	
603	Arthrogryposis, distal	ECEL1, PIEZO2
604	Arthrogryposis lethal with anterior horn cell disease	GLE1
605	Arthrogryposis, renal dysfunction, and cholestasis	VIPAS39, VPS33B
606	Arthrogryposis multiplex congenita	LGI4
	Dystrophy	
607	Limb-girdle muscular dystrophy	CAPN3, DYSF, PLEC, SGCA, SGCB, SGCD, SGCG, TCAP, TRAPPC11, TRIM32, TTN
608	Muscular dystrophy-dystroglycanopathy	B3GALNT2, CRPPA, FKRP, FKTN, GMPPB, LARGE1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RXYLT1
609	Muscular dystrophy, congenital	CHKB, LAMA2
610	Ullrich congenital muscular dystrophy	COL6A1, COL6A2, COL6A3
611	Duchenne muscular dystrophy	DMD

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		In VIC and NSW, most DMD carriers are unable to be detected due to limitations in testing technology
612	Becker muscular dystrophy	DMD
		As above
613	Emery-Dreifuss muscular dystrophy	EMD, FHL1, LMNA
614	Muscular dystrophy, rigid spine	SELENON
	Myopathy	
615	Myopathy, congenital	ACTA1
616	Nemaline myopathy	ACTA1, CFL2, KLHL40, KLHL41, LMOD3, NEB, TNNT1, TPM3
617	Myopathy, centronuclear, autosomal recessive	BIN1, SPEG*
		Not screened in WA, QLD and SA
618	Distal myopathy	DYSF
619	Myopathy with extrapyramidal signs	MICU1
620	Myopathy, X-linked	FHL1
621	Myopathy, X-linked, with excessive autophagy	VMA21
622	Inclusion body myopathy	GNE
623	Myopathy, areflexia, respiratory distress, and dysphagia, early-onset	MEGF10
624	Myotubular myopathy, X-linked	MTM1
625	Minicore myopathy	RYR1
626	Myopathy, myofibrillar	KY, PYROXD1
627	Central core disease	RYR1
628	Myopathy, early-onset, with fatal cardiomyopathy	TTN
629	CAP myopathy	TPM3
	Myasthenia	
630	Myasthenic syndrome	AGRN, ALG2, CHAT, CHRNA1, CHRND, CHRNE, COLQ, DOK7, DPAGT1, GFPT1, IGHMBP2, MUSK, RAPSN, SLC5A7
	Neuropathy	
631	Charcot-Marie-Tooth disease	FGD4, FIG4, GDAP1, LMNA, MFN2, MPZ, MTMR2, NDRG1, PRPS1, PRX, SBF2, SH3TC2
632	Dysautonomia, familial	ELP1
633	Insensitivity to pain, congenital	SCN9A, NTRK1
634	Neuromyotonia and axonal neuropathy	HINT1
635	Neuropathy, hereditary motor and sensory	HK1, IGHMBP2, KIF1A, SLC25A46
636	Neuropathy, hereditary sensory and autonomic	NGF, PRDM12, RETREG1, WNK1
637	Giant axonal neuropathy	GAN
	Rhabdomyolysis	
638	Myoglobinuria, acute recurrent	LPIN1
	Spasticity	
640	Spastic paralysis, infantile onset ascending	ALS2
641	Juvenile primary lateral sclerosis	ALS2

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642	Spastic paraplegia	AP4M1, AP4B1, AP4S1, ATP13A2, ALDH18A1, B4GALNT1, CYP2U1, CYP7B1, DDHD2, DSTYK, FA2H, FARS2, GBA2, GJC2, KIF1A, NT5C2, PLP1, PNPLA6, SPG11, VPS37A, ZFYVE26
	Connective tissue conditions	
	Ehlers-Danlos syndrome (EDS)	
643	Ehlers-Danlos syndrome, progeroid type	ADAMTS2, B3GALT6, B4GALT7, PLOD1
644	Ehlers-Danlos syndrome, musculocontractural type	CHST14
645	Ehlers-Danlos syndrome with progressive kyphoscoliosis, myopathy, and hearing loss	FKBP14
	Vascular conditions	
646	Polyarteritis nodosa, childhood-onset	ADA2
647	Meester-Loeys syndrome	BGN
	Ocular conditions	
	Albinism	
648	Hermansky-Pudlak syndrome	HPS1, HPS3, HPS4, HPS5, HPS6
649	Oculocutaneous albinism	GPR143, LRMDA, OCA2, SLC24A5, SLC45A2, TYR, TYRP1
	Dystrophies	
650	Retinal dystrophy, early-onset severe	LRAT, RCBTB1, CFAP410
651	Macular dystrophy with central cone involvement	MFSD8
652	Cone-rod dystrophy	AIPL1, C8orf37, CEP78, CNGB3, KCNV2, PDE6C, RPGRIP1, SEMA4A
	Microphthalmia	
653	Isolated	ALDH1A3, RAX, VSX2
654	With coloboma	STRA6, VSX2
655	Syndromic	STRA6, RARB
	Other ocular conditions	
656	Achromatopsia	ATF6, CNGA3, CNGB3, GNAT2
657	Aphakia	FOXE3
658	Congenital cataracts	AGK, FYCO1, NHS, TDRD7
659	Cone-rod synaptic disorder, congenital non-progressive	CABP4
660	Choroideremia	CHM
661	Congenital stationary night blindness	GPR179, NYX
662	Persistent hyperplastic primary vitreous	ATOH7
663	Macular degeneration (congenital)	CNGB3, RPGR
664	Leber congenital amaurosis	AIPL1, CEP290, CRB1, GUCY2D, LCA5, LRAT, NMNAT1, RD3, RDH12, RPE65, RPGRIP1, SPATA7, TULP1
665	Glaucoma (congenital)	CYP1B1
666	Peters anomaly	CYP1B1
667	Retinal arterial macroaneurysm with supravalvular pulmonic stenosis	IGFBP7
668	Retinitis pigmentosa	AGBL5, AIPL1, C8orf37, CRB1, DHDDS, IFT172, LRAT, MERTK, REEP6, RP2, SEMA4A, SPATA7, TULP1, USH2A
669	Progressive external ophthalmoplegia	POLG
670	Brittle cornea syndrome	PRDM5

671	Corneal opacification and other ocular anomalies	PXDN
672	Gaze palsy, horizontal, with progressive scoliosis	ROBO3
673	Foveal hypoplasia, with or without optic nerve misrouting and/or anterior segment dysgenesis	SLC38A8
674	Optic atrophy	TMEM126A
	Skeletal conditions	
	Dysplasias	
675	Spondyloepiphyseal dysplasia with other abnormalities	CHST3, CCN6
676	Anauxetic dysplasia	POP1, RMRP
677	Spondyloepimetaphyseal dysplasia	B3GALT6, NANS
678	Desbuquois dysplasia	CANT1, XYLT1
679	Schneckenbecken dysplasia	SLC35D1
680	Short-rib thoracic dysplasia with or without polydactyly	CEP120, DYNC2H1, DYNC2I1*, DYNC2I2^, DYNC2LI1, KIAA0586, TTC21B, WDR35, IFT140, IFT172, IFT80, NEK1 *Formerly known as WDR60 ^Formerly known as WDR34
681	Spondylometaepiphyseal dysplasia, short limb-hand type	DDR2
682	Spondylo-megaepiphyseal-metaphyseal dysplasia	NKX3-2
683	Chondrodysplasia, Grebe type	GDF5
684	Oculodentodigital dysplasia	GJA1
685	Smith-McCort dysplasia	DYM, RAB33B
686	Omodysplasia	GPC6
687	Dyssegmental dysplasia, Silverman-Handmaker type	HSPG2
688	Cranioectodermal dysplasia	IFT122
689	Opsismodysplasia	INPPL1
690	Otospondylomegaepiphyseal dysplasia	COL11A2
691	Greenberg skeletal dysplasia	LBR
692	Cleft lip/palate-ectodermal dysplasia syndrome	NECTIN1
693	Spondylometaphyseal dysplasia with additional abnormalities	PCYT1A, CFAP410
694	Chondrodysplasia, Blomstrand type	PTH1R
695	Metaphyseal dysplasia without hypotrichosis	RMRP
696	Craniolenticulosutural dysplasia	SEC23A
697	Langer mesomelic dysplasia	SHOX
698	De la Chapelle dysplasia	SLC26A2
699	Diastrophic dysplasia	SLC26A2
700	Craniofrontonasal dysplasia	EFNB1
701	Chondrodysplasia punctata, rhizomelic	AGPS, GNPAT, PEX7
702	Mandibuloacral dysplasia	LMNA
	Acromesomelic dysplasia	
703	Hunter-Thompson type	GDF5
704	Maroteaux type	NPR2
705	Demirhan type	BMPR1B
	Arthropathies	

706	Arthropathy, progressive pseudorheumatoid	CCN6
707	Cranioosteoarthropathy	HPGD
708	Hypertrophic osteoarthropathy	HPGD
709	Multicentric osteolysis, nodulosis, and arthropathy	MMP2
710	Camptodactyly-arthropathy-coxa vara-pericarditis syndrome	PRG4
	Short stature and dwarfism	
711	Multiple joint dislocations, short stature, craniofacial dysmorphism, and congenital heart defects	B3GAT3
712	Amelogenesis imperfecta and short stature	LTBP3
713	Microcephalic osteodysplastic primordial dwarfism	PCNT, RNU4ATAC
714	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis	POC1A
715	Short stature, optic nerve atrophy, and Pelger-Huet anomaly	NBAS
716	Mulibrey nanism	TRIM37
	Other skeletal conditions	
717	3-M syndrome	CCDC8, OBSL1, CUL7
718	Antley-Bixler syndrome	POR
719	Hypophosphatasia, infantile	ALPL
720	Diaphanospondylodysostosis	BMPER
721	Meier-Gorlin syndrome	CDT1, CDC45, ORC1, ORC6
722	Osteopetrosis, infantile	CA2, CLCN7, OSTM1, TCIRG1, TNFRSF11A, TNFSF11
723	Fibrochondrogenesis	COL11A1, COL11A2
724	Osteogenesis imperfecta, recessive type	CRTAP, FKBP10, P3H1, PPIB*, SERPINF1, WNT1
		Not screened in WA, QLD and SA
725	Pycnodysostosis	CTSK
726	Spondylocostal dysostosis	DLL3, HES7, MESP2
727	Ellis-van Creveld syndrome	EVC, EVC2
728	Raine syndrome	FAM20C
729	Bruck syndrome	FKBP10, PLOD2
730	Spondylocarpotarsal synostosis syndrome	FLNB
731	Brachydactyly	GDF5
732	Geroderma osteodysplasticum	GORAB
733	Craniosynostosis	IL11RA
734	Alazami syndrome	LARP7
735	Schwartz-Jampel syndrome	HSPG2
736	Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome	LIFR
737	Acheiropody	LMBR1
738	Cenani-Lenz syndactyly syndrome	LRP4
739	Sclerosteosis	LRP4, SOST
740	Osteoporosis-pseudoglioma syndrome	LRP5
741	Orofacial cleft	NECTIN1
742	Brachyolmia 4 with mild epiphyseal and metaphyseal change	PAPSS2
743	Carpenter syndrome	RAB23, MEGF8

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744	Baller-Gerold syndrome	RECQL4
745	RAPADILINO syndrome	RECQL4
746	Cartilage-hair hypoplasia	RMRP
747	Robinow syndrome	ROR2
748	Van den Ende-Gupta syndrome	SCARF2
749	Frank-ter Haar syndrome	SH3PXD2B
750	Achondrogenesis	SLC26A2, TRIP11
751	Atelosteogenesis	SLC26A2
752	Van Buchem disease	SOST
753	Kenny-Caffey syndrome	TBCE
754	Paget disease of bone	TNFRSF11B
755	Ulna and fibula, absence of, with severe limb deficiency	WNT7A
756	Fuhrmann syndrome	WNT7A
757	CODAS syndrome	LONP1
758	Keutel syndrome	MGP
759	Steel syndrome	COL27A1

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