SoonerStart Automatic Qualifying Syndromes and Conditions	
001	Abetalipoproteinemia 272.5
002	Acanthocytosis (see Abetalipoproteinemia) 272.5
003	Accutane, Fetal Effects of (see Fetal Retinoid Syndrome) 760.79
004	Acidemia, 2-Oxoglutaric 276.2
005	Acidemia, Glutaric I 277.8
006	Acidemia, Isovaleric 277.8
007	Acidemia, Methylmalonic 277.8
008	Acidemia, Propionic 277.8
009	Aciduria, 3-Methylglutaconic Type II 277.8
010	Aciduria, Argininosuccinic 270.6
011	Acoustic-Cervico-Oculo Syndrome (see Cervico-Oculo-Acoustic Syndrome) 759.89
012	Acrocephalopolysyndactyly Type II 759.89
013	Acrocephalosyndactyly Type I 755.55
014	Acrodysostosis 759.89
015	Acrofacial Dysostosis, Nager Type 756.0
016	Adams-Oliver Syndrome (see Limb and Scalp Defects, Adams-Oliver Type) 759.89
017	Adrenoleukodystrophy, Neonatal (see Cerebro-Hepato-Renal Syndrome) 759.89
018	Aglossia Congenita (see Hypoglossia-Hypodactylia) 759.89
019	Albinism, Ocular (includes Autosomal Recessive Type) 759.89
020	Albinism, Oculocutaneous, Brown Type (Type IV) 759.89
021	Albinism, Oculocutaneous, Tyrosinase Negative (Type IA) 759.89
022	Albinism, Oculocutaneous, Tyrosinase Positive (Type II) 759.89
023	Albinism, Oculocutaneous, Yellow Mutant (Type IB) 759.89
024	Albinism-Black Locks-Deafness 759.89
025	Albright Hereditary Osteodystrophy (see Parathyroid Hormone Resistance) 759.89
026	Alexander Disease 759.89
027	Alopecia - Mental Retardation 759.89
028	Alpers Disease 759.89
029	Alpha 1,4 - Glucosidase Deficiency (see Glycogenosis, Type IIA) 271.0
030	Alpha-L-Fucosidase Deficiency (see Fucosidosis) 271.8
031	Alport Syndrome (see Nephritis-Deafness, Hereditary Type) 759.89
032	Amaurosis (see Blindness) 369.00
033	Amaurosis Congenita of Leber, Types I and II (see Retina, Amaurosis Congenita,
	Leger Type) 362.74
034	Amelia (see Limb Reduction Defects) 755.31 (lower limb) 755.21 (upper limb)
035	Angelman Syndrome 759.89
036	Aniridia 743.45
037	Aicardi Syndrome 759.89
038	AIDS Infection (see Fetal Acquired Immune Deficiency Syndrome) 042
039	Alaninuria (see Pyruvate Dehydrogenase Deficiency) 759.89
040	Albers-Schonberg Disease (see Osteopetrosis, Malignant Recessive) 759.89

041	Anophthalmia, recessive Waardenburg type (Anophthalmia - Limb Anomalies) 759.89
042	Anophthalmia-Limb Anomalies 759.89
043	Anus-Hand-Ear Syndrome 759.89
044	Apert Snydrome (see Acrocephalosyndactyly (Type I) 755.55
045	Apolipoprotein B Deficiency (see Abetalipoproteinemia) 272.5
046	Aracerebroside Sulfatase Deficiency (see Metachromatic Leukodystrophies) 759.89
047	Arachnodactyly, Contractural Beals Type 759.82
048	Argininemia 759.89
049	Arthro-Ophthalmopathy, Hereditary, Progressive, Stickler Type 759.89
050	Arthrogryposis (Arthrogryposes Multiplex Congenita) 754.89
051	Arthrogryposis, Amyplasia Type 754.89
052	Arthrogryposis, Distal Types I and II 754.89
053	Arylsulfatase A Deficiency (see Metachromatic Leukodystophies) 759.89
054	Ataxia with Lactic Acidosis I (see Pyruvate Dehydrogenase Deficiency) 759.89
055	Ataxia with Lactic Acidosis II (see Pyruvate Carboxylase Deficiency with Lactic
	Acidemia) 759.89
056	Ataxia-Telangiectasia Syndrome 334.8
*	Auditory Neuropathy Spectrum Disorder (ANSD) (See #569)
057	Autism, Infantile 299.0
058	BADS Syndrome (see Albinism-Black Locks-Deafness) 759.89
059	Bardet-Biedl Syndrome 759.89
060	Bartter Syndrome 255.1
061	Batten Disease (see Neuronal, Ceroid-Lipofuscinoses) 330.1
062	Beals Syndrome (see Arachnodactyly, Contractural Beals Type) 759.82
063	Beals-Hecht Syndrome (see Arachnodactyly, Contractural Beals Type) 759.82
064	Behr Syndrome (see Optic Atrophy, Infantile Heredofamilial) 759.89
065	Bertrand Spongy Degeneration of the CNS (see Brain, Spongy Degeneration) 759.89
066	Beta-Galactosidase-I Deficiency (see G(MI)-Gangliosidosis, Type I) 330.1
067	Biedel-Bardet Syndrome (see Bardet-Biedel Syndrome) 759.89
068	Bing-Siebenmann Dysplasia (see Ear, Inner Dysplasias) 744.05
069	Biotinidase Deficiency 759.89
070	Blindness or Near Blindness 369.00 or 369.04
071	Boder-Sedgwick Syndrome (see Ataxia-Telangiectasis) 334.8
072	BOR Syndrome (see Brachio-Oto-Renal Dysplasia) 759.89
073	Borjeson-Forssman-Lehmann Syndrome 759.89
074	Bourneville Syndrome (see Tuberous Sclerosis) 759.5
075	Brain, Micropolygyria 742.2
076	Brain, Porencephaly 742.4
077	Brain, Schizencephaly 742.4
078	Brain, Spongy Degeneration 759.89
079	Branchio-Oculo-Facial Syndrome 759.89
080	BBB Syndrome (see Hypertelorism-Hypospadius Syndrome) 759.89
081	Branchio-Oto-Renal Dysplasia (when lip pits or clefts are present) 759.89

082	Burns Syndrome (see Ichthyosiform Erythrokeratoderma, Atypical with Deafness) 757.1
083	C Syndrome 759.89
084	Camptodactyly-Trismus Syndrome 759.89
085	Canavan Disease (see Brain, Spongy Degeneration) 759.89
086	Carbamoyl Phosphate Synthetase Deficiency 759.89
087	Cardio-Auditory Syndrome 759.89
088	Carpenter Syndrome (see Acrocephalopolysyndactyly II) 759.89
089	Cat Cry Syndrome (see Chromosome 5, Monosomy 5p) 758.3
090	Cat Eye Syndrome 758.5
091	Cataract, Autosomal Dominant Congenital 743.34
092	Cataract, Cortical and Nuclear 743.33
093	Cataract, Polar 743.31
094	Cataracts 366.9
095	Caudal Dysplasia (see Caudal Regression Syndrome) 759.89
096	Caudal Regression Syndrome (when paralysis is present) 759.89
097	Central Ray Defects (see Limb Reduction Defects) 755.21 (upper limb) 755.31 (lower
	limb)
098	Ceramidase Deficiency (see Lipogranulomatosis) 759.89
099	Cerebellar Parenchymal Disorder, Type IV (see Joubert Syndrome) 759.89
*	Cerebral Gigantism 759.89 (See #118)
*	Cerebral G(MI)-Gangliosidosis (see G(MI)-Gangliosidosis, Type I) 330.1 (See #119)
*	Cerebral Palsy 343.9 (See #120)
100	Cerebro-Costo-Mandibular Syndrome 759.89
101	Cerebro-Hepato-Renal Syndrome 759.89
102	Cerebro-Oculo-Facio-Skeletal Syndrome 759.89
103	Cerebroocular Dysgenesis (see Walker-Warburg Syndrome) 759.89
104	Cerebroside Liposis (see Gaucher Disease) 272.7
105	Cerebrosidosis (see Gaucher Disease) 272.7
106	Cervico-Oculo-Acoustic Syndrome 759.89
107	Cervicooculofacial Dysplasia (see Cervico-Oculo-Acoustic Syndrome) 759.89
108	Charge Association 759.89
109	Chemke Syndrome (see Walker-Warburg Syndrome) 759.89
110	Chicken Pox, Fetal Effects (see Fetal Effects from Varicella-Zoster) 760.2
111	CHILD Syndrome (see Limb Reduction-Ichthyosis) 755.2
112	Chondrodysplasia Calcificans Congenita (see Chondrodysplasia Punctata, X-Linked
	Dominant Type) 759.89
113	Chondrodysplasia Punctata, Rhizomelic Type 759.89
114	Chondrodysplasia Punctata, X-Linked Dominant Type 759.89
115	Chondrodystrophic Myotonia, Schwartz-Jampel Type 756.89
116	Christensen Krabbe Disease (see Alpers Disease) 759.89
117	Chromosome 1, Monosomy 1q 758.9
118	Cerebral Gigantism 759.89
119	Cerebral G(MI)-Gangliosidosis (see G(MI)-Gangliosidosis, Type I) 330.1

120	Cerebral Palsy 343.9
121	Chromosome 1, Monosomy 1q4 758.9
122	Chromosome 2, Monosomy of Medial 2q 758.9
123	Chromosome 2, Partial Trisomy 2p 758.9
124	Chromosome 2, Trisomy Distal 2q 758.9
125	Chromosome 3, Trisomy 3p2 758.9
126	Chromosome 3, Trisomy 3q2 758.9
127	Chromosome 4, Monosomy 4p 758.3
128	Chromosome 4, Monosomy Distal 4q 758.9
129	Chromosome 4, Trisomy 4p 758.9
130	Chromosome 4, Trisomy Distal 4q 758.9
131	Chromosome 5, Monosomy 5p 758.3
132	Chromosome 5, Trisomy 5q3 758.9
133	Chromosome 6, Monosomy Proximal 6q 758.9
134	Chromosome 6, Ring 6 758.9
135	Chromosome 6, Trisomy 6q2 758.9
136	Chromosome 7, Trisomy 7q2-3 758.9
137	Chromosome 8, Trisomy 8 758.5
138	Chromosome 8, Trisomy 8p 758.9
139	Chromosome 9, Partial Monsomy 9p 758.9
140	Chromosome 9, Trisomy 9 758.9
141	Chromosome 9, Trisomy 9p 758.9
142	Chromosome 9, Trisomy 9q3 758.9
143	Chromosome 10, Monosomy 10p 758.9
144	Chromosome 10, Monosomy 10q2 758.9
145	Chromosome 10, Trisomy 10q2 758.9
146	Chromosome 11, Monosomy 11q 758.9
147	Chromosome 11, Partial Monosomy 11p 758.9
148	Chromosome 11, Partial Trisomy 11q 758.9
149	Chromosome 11, Partial Trisomy 11p 758.9
150	Chromosome 12, Isochromosome 12p mocaicism (see Pallister-Killian Mosaic Syndrome)
	758.9
151	Chromosome 12, Monosomy 12p 758.9
152	Chromosome 12, Partial Trisomy 12p 758.9
153	Chromosome 12, Trisomy 12q2 758.9
154	Chromosome 13, Monosomy 13q 758.3
155	Chromosome 13, Monosomy 13q3 758.9
156	Chromosome 13, Trisomy 13 758.1
157	Chromosome 13, Trisomy 13q1 758.9
158	Chromosome 13, Trisomy Distal 13q 758.9
159	Chromosome 14, Partial Trisomy 14q 758.9
160	Chromosome 14, Ring 14 758.9
161	Chromosome 15, Partial Trisomy Distal 15q 758.9

162	Chromosome 15, Ring 15 758.9
163	Chromosome 15, Trisomy 15q1 758.9
164	Chromosome 16, Trisomy 16q 758.9
165	Chromosome 17, deletion or monosomy 17p13 (see Lissencephaly Syndrome) 742.2
166	Chromosome 17, Interstitial Deletion 17p 758.9
167	Chromosome 18, Monosomy 18p 758.3
168	Chromosome 18, Monosomy 18q or 18r (ring) 758.3
169	Chromosome 18, Ring 18 758.9
170	Chromosome 18, Tetrasomy 18p 758.9
171	Chromosome 18, Trisomy 18 758.2
172	Chromosome 18, Trisomy 18q2 758.9
173	Chromosome 20, Trisomy 20p 758.9
174	Chromosome 21, Trisomy 21 758.0
175	Chromosome 22, Monosomy 22q 758.9
176	Chromosome 22, Partial Trisomy 22 (see Cat Eye Syndrome) 758.5
177	Chromosome 22, Ring 22 758.9
178	Chromosome XXXX (see XXXX Syndrome) 758.81
179	Chromosome XXXXX (see XXXXX Syndrome) 758.81
180	Chromosomer XXY (see Klinefelter Syndrome) 758.7
181	Cleft Palate-Micrognathia-Glossoptosis 756.0
182	Club Hand (see Hand, Radial Club Hand) 754.89
183	Cockayne Syndrome 759.89
184	Coffin-Lowry Syndrome 759.89
185	Coffin-Siris Syndrome 759.89
186	COFS (see Cerebro-Oculo-Skeletal Syndrome) 759.89
187	Cohen Syndrome 759.89
*	Congenital Hypothyroidism 243 (See #564)
188	Corneal Dystrophy, Endothelial, Congenital Hereditary 743.43
189	Cornelia de Lange Syndrome (see De Lange Syndrome) 759.89
190	Corpus Callosum Agenesis 742.2
*	Cranio-Carpo-Tarsal Dysplasia, Whistling Face Type 759.89 (See #209)
*	Cri Du Chat Syndrome (see Chormosome 5, Monosomy 5p) 758.3 (See #210)
191	Cytochrome C Oxidase Deficiency (see Myopathy-Metabolic, Mitochondrial Cytochrome
	C Oxidase Deficiency) 756.89
192	De Lange Syndrome 759.89
193	De Morsier Syndrome (see Septo-Optic Dysplasia) 742.4
194	De Toni-Fanconi-Debre Syndrome (some cases) (see Myopathy-Metabolic,
	Mitochondrial Cytochrome C Oxidase Deficiency) 756.89
195	Deafness (see Hearing Loss) 389.9
196	Deafness, Congenital I or II (see Deafness (Sensorineural), Recessive Profound) 744.00
197	Deafness (Sensorineural), Recessive Profound 744.00
198	Deafness-Ear Pits 744.00
199	Deafness-Malformed Ears-Mental Retardation 759.89

200	Deafness-Pili Torti, Bjornstad Type 744.00
201	Dejerine-Sottas Disease 356.0
202	Dermal Hypoplasia, Focal 759.89
203	DeSanctis-Cacchione Syndrome (see Xeroderma Pigmentosum-Mental Retardation)
	759.89
204	Desbuquois Syndrome (see Larson Syndrome) 759.89
205	Diastrophic Dysplasia 756.4
206	DiGeorge Syndrome (see Immunodeficiency, Thymic Agenesis) 279.11
207	Diplegia, Congenital Facial 352.6
208	Donohue Syndrome (see Leprechauism) 259.8
209	Cranio-Carpo-Tarsal Dysplasia, Whistling Face Type 759.89
210	Cri Du Chat Syndrome (see Chormosome 5, Monosomy 5p) 758.3
211	Down Syndrome (see Chromosome 21, Trisomy 21) 758.0
212	Dubowitz Syndrome 759.89
213	Duchenne Muscular Dystrophy (see Muscular Dystrophy, Psuedohypertropic) 359.1
214	Dwarfism, Metatropic Type II (see Kneist Dysplasia) 759.89
215	Dwarfism, Seckle Type (see Seckle Syndrome) 759.89
216	Dysautonomia, Type 1, Riley-Day Type 742.8
217	Dysautonomia, Type II, Familial (see Neuropathy, Congenital Sensory with Anhidrosis)
	742.8
218	Ear, Inner Dysplasias 744.05
219	Ear, Microtia - Atresia 744.23
220	Ear, Ossicle and Middle Ear Malformations 744.04
221	Ectrodactyly 755.25
222	Ectrodactyly-Tibial Hemimelia (see Tibial Hypoplasia/Aplasia-Ectrodactyly) 755.25 &
	755.36
223	Edwards Syndrome (see Chromosome 18, Trisomy 18) 758.2
224	Ehlers-Danlos Syndrome 756.83
225	Encephalocele 742.0
226	Encephalopathy, Necrotizing 330.8
227	Epidermal Nevus Syndrome (see Nevus, Epidermal Nevus Syndrome) 759.89
228	Escobar Syndrome (see Pterygium Syndrome, Multiple) 759.89
229	Eye, Anophthalmia 743.00
230	Eye, Anterior Segment Dysgenesis 743.48
231	Facio-Oculo-Acoustic-Renal Syndrome 759.89
*	Eye, Microphthamia/Coloboma 743.10 & 743.46 (See #249)
*	Eye, Orbital Teratoma, Congenital 238.8 (See #250)
232	Failure to Thrive 783.4
233	Falciform Detachment, Congenital (see Retinal Fold) 743.56
234	Farber Disease (see Lipogranulomatosis) 759.89
235	Femoral Hypoplasia-Unusual Facies Syndrome 759.89
236	Fetal Acquired Immune Deficiency Syndrome 042
237	Fetal Alcohol Syndrome 760.71

238	Fetal Aminoglycoside Ototoxicity 744.00
239	Fetal Brain Disruption Sequence 771.1
240	Fetal Cytomegalovirus Syndrome 771.1
241	Fetal Effects from Maternal PKU 760.8
242	Fetal Effects from Varicella-Zoster 760.2
243	Fetal Herpes Simplex Infection 771.2
244	Fetal Retinoid Syndrome 760.79
245	Fetal Rubella Syndrome 771.0
246	Fetal Syphilis Syndrome 090.0
247	Fetal Toxoplasmosis Syndrome (clinically apparent cases) 771.2
248	Fetal Warfarin Syndrome 760.79
249	Eye, Microphthamia/Coloboma 743.10 & 743.46
250	Eye, Orbital Teratoma, Congenital 238.8
251	FG Syndrome, Opitz-Kaveggia Type 759.89
252	Fibromatosis, Juvenile Hyaline 759.89
253	Fibula, Congenital Absence of (Type II & III) 755.36
254	FOAR Syndrome (see Facio-Oculo-Acoustic-Renal Syndrome) 759.89
255	Fragile X Syndrome (see X-Linked Mental Retardation, Fragile X Syndrome) 758.81
256	Fraser Syndrome 759.89
257	Freeman-Sheldon Syndrome (see Cranio-Carpo-Tarsal Dysplasia, Whistling Face Type)
	759.89
258	Frontometaphyseal Dysplasia 759.89
259	Fucosidosis 271.8
260	G(MI)-Gangliosidosis, Type 1 330.1
261	G(MI)-Gangliosidosis Type 2 330.1
262	G(M2)-Gangliosidois with Hexosaminidase A and B Deficiency 330.1
263	G(M2)-Gangliosidosis with Hexosaminidase A Deficiency 330.1
*	Galactosemia 271.1 (See #566)
264	Galactosialidosis (early-infantile type and late-infantile form) 330.1
265	Ganglioside Neuroaminidase Deficiency (see Mucolipidosis IV) 272.7
266	Ganglioside Sialidase Deficiency (see Mucolipidosis IV) 272.7
267	Gangliosidosis, Generalized Juvenile Type (see G(M1)-Gangliosidosis, Type 2) 330.1
268	Gangliosidosis, Type 1 (see G(M1)- Gangliosidosis, Type 1) 330.1
269	Gaucher Disease (acute or infantile form) 272.7
*	Giedion-Langer Syndrome 759.89 (See #287)
270	Glutaric Aciduria Type 1 (see Acidemia, Glutaric Acidemia I) 277.8
271	Glutaryl-CoA Dehydrogenase Deficiency (see Acidemia, Glutaric Acidemia I) 277.8
272	Glycogen Storage Disease, Type IIA (see Glysogenois, Type IIA) 271.0
273	Glycogenosis, Type IA 271.0
274	Glycogenosis, Type IIA 271.0
275	Goldberg Syndrome (see Galactosialidosis) 330.1
276	Goldenhar Syndrome (see Oculo-Auriculo-Vertebral Anomaly) 759.89
277	Gollop-Wolfgang Syndrome (see Tibial Hypoplasia/Aplasia-Ectrodactyly) 759.89

278	Goltz-Gorlin Syndrome (see Dermal Hypoplasia, Focal) 759.89
279	Guerin-Stern Syndrome (see Arthrogryposis) 754.89
280	Hallermann-Streiff Syndrome (see Oculo-Mandibulo-Facial Syndrome) 756.0
281	Hallgren Syndrome (see Usher Syndrome) 759.89
282	Haltia-Santavuori Disease (infantile) (see Neuronal Ceroid-Lipofuscinoses) 330.1
283	Hand, Radial Club Hand 754.89
284	Handmann Disk Anomaly (see Optic Disk, Morning Glory Anomaly) 743.57
285	Hanhart Syndrome (see Hypoglossia-Hypodactylia) 759.89
286	Happy Puppet Syndrome (see Angelman Syndrome) 759.89
287	Giedion-Langer Syndrome (see Tricho-Rhino-Phalangeal Syndrome, Type II) 759.89
288	HARD Syndrome (see Walker-Warburg Syndrome) 759.89
289	Hearing Loss - Permanent Unilateral or Bilateral hearing loss of 25 dB or greater
290	Heart-Hand Syndrome 759.89
291	Hecht Syndrome (see Camptodactyly-Trismus Syndrome) 759.89
292	Hemifacial Microsomia (see Oculo-Auriculo-Vertebral Anomaly) 759.89
293	Hemimelia (see Limb Reduction Defects) 755.21 (upper limb) 755.31 (lower limb)
294	Hereditary Motor Sensory Neuropathy, Type III (see Dejerine-Sottas Disease) 356.0
295	Herpes Simplex Infection (see Fetal Herpes Simplex Infection) 771.2
296	HGPRT (Hypoxanthine Guanine Phosphoribosyl Transferase) Deficiency (see Lesch-
	Nyhan Syndrome) 277.2
297	HHH Syndrome (see Hyperornithinemia-Hyperammonemia-Homocitrullinuria) 270.6
298	Holoprosencephaly 742.2
299	Holt-Oram Syndrome (see Heart-Hand Syndrome) 759.89
300	Hunter Syndrome (see Mucopolysaccharidosis II) 277.5
301	Hurler Syndrome (see Mucopolysaccharidosis I-H) 277.5
302	Hurler-Pfakundler Syndrome (see Mucopolysaccharidosis I-H) 277.5
303	Hurler-Scheie Syndrome (see Mucopolysaccharidosis I-H) 277.5
304	Hydrocephalus 742.3
*	Hyperammonemia 270.6 (See #321)
*	Hyperglycinemia, Non-ketotic 270.7 (See #322)
305	Hyperornithinemia-Hyperammonemia-Homocitrullinuria 270.6
306	Hypertelorism-Hypospadius Syndrome 759.89
307	Hypoglossia-Hypodactylia 759.89
308	I-Cell Disease (see Mucolipidosis II) 272.7
309	Ichthyosiform Erythrokeratoderma, Atypical with Deafness 757.1
310	Immunodeficiency, Thymic Agenesis 279.11
311	Infantile Spasms 345.6
312	Isovaleric Acidemia (see Acidemia, Isovaleric) 277.8
313	Jacobsen Syndrome (see Chromosome 11, Monosomy 11q) 758.9
314	Jansky-Bielchowsky Disease (late infantile) (see Neuronal Ceroid-Lipoufscinoses) 330.1
315	Jervell Syndrome (see Cardio-Auditory Syndrome) 759.89
316	Johanson-Blizzard Syndrome 759.89
317	Joubert Syndrome 759.89

318	Kearns-Sayre Disease 759.89
319	Keratitis-Ichthyosis-Deafness (KID) Syndrome (see Ichthyosiform Erythrokeratoderma,
	Atypical with Deafness) 757.1
320	Killian Syndrome (see Pallister-Killian Mosaic Syndrome) 758.9
321	Hyperammonemia 270.6
322	Hyperglycinemia, Non-ketotic 270.7
323	Kinky Hair Disease (see Menkes Syndrome) 759.89
324	Klinefelter Syndrome 758.7
325	Klippel-Feil Anomaly 756.16
326	Kneist Dysplasia 759.89
327	Krabbe Disease (see Leukodystrophy, Globoid Cell Type) 330.0
328	Lacrimo-Auriculo-Dento-Digital Syndrome 759.89
329	LADD Syndrome (see Lacrimo-Auriculo-Dento-Digital Syndrome) 759.89
330	Lange-Nielson Syndrome (Cardio-Auditory Syndrome) 759.89
331	Larson Syndrome 759.89
332	Laurence-Moon Syndrome 759.89
333	Leigh Syndrome (see Myopathy-Metabolic, Mitochodrial Cytochrome C Oxidase
	Deficiency (some cases) or Encephalopathy, Necrotizing) 756.89 & 330.8
334	Lens and Pupil, Ectopic 743.46
335	Lens, Aphakia 743.35
336	Lens, Ectopic 743.37
337	Lens, Microspherophakia 743.36
338	Lenz Microphthalmia Syndrome 759.89
339	Leprechaunism 259.8
340	Leroy Disease (see Mucolipidosis II) 272.7
341	Lesch-Nyhan Syndrome 277.2
342	Leukodystrophy, Alexander Disease (see Alexander Disease) 759.89
*	Leukodystrophy, Globoid Cell Type 330.0 (See #361)
*	Levy-Hollister Syndrome 759.89 (See #362)
*	Limb and Scalp Defects, Adams-Oliver Type 759.89 (See #363)
343	Limb Reduction Defects 755.21 (upper limbs) 755.31 (lower limbs)
344	Limb Reduction-Ichthyosis 755.2
345	Linear Nevus Sebaceous Syndrome (see Nevus, Epidermal Nevus Syndrome) 759.89
346	Lipogranulomatosis 759.89
347	Lipomatosis of Pancreas, Congenital (see Shwachman Syndrome) 288.0
348	Lipomucopolysaccaridosis (see Mucolipidosis I) 272.7
349	Lissencephaly Syndrome 742.2
350	Lissencephaly Syndrome II (see Walker-Warburg Syndrome) 759.89
351	Liver Disease - Neuronal Degeneration of Childhood (see Alpers Disease) 759.89
352	Lobster claw deformity (see Ectrodactyly) 755.25
353	Loken-Senior Syndrome (see Renal Dysplasia-Retinal Aplasia, Loken-Senior Type)
	759.89
354	Louis-Barr Syndrome (see Ataxia-Telangiectasis) 334.8

*	Low Birth Weight (LBW) (See #567)
355	Lowe Syndrome (see Oculo-Cerebro-Renal Syndrome) 270.8
356	Mandibular Dysostosis, Treacher-Collins Type-Limb Anomalies (see Acrofacial
	Dysostosis, Nager Type) 756.0
357	Mandibulofacial Dysostosis 756.0
358	Mannosidosis (Type I and II) 271.8
359	Marden-Walker Syndrome 759.89
360	Marinesco-Garland Syndrome (see Marinesco-Sjogren Syndrome) 759.89
361	Leukodystrophy, Globoid Cell Type 330.0
362	Levy-Hollister Syndrome (see Lacrimo-Auriculo-Dento-Digital Syndrome) 759.89
363	Limb and Scalp Defects, Adams-Oliver Type 759.89
364	Marinesco-Sjogren Syndrome 759.89
365	Marshall-Smith Syndrome 759.89
366	Martin Bell X-Linked Mental Retardation (see Fragile X Syndrome) 758.81
367	Maumenee Corneal Dystrophy (see Corneal Dystrophy, Endothelial, Congenital
	Hereditary) 743.43
368	Maxillofacial Dysostosis 756.0
369	Melnick-Fraser Syndrome (see Brachio-Oto-Renal Dysplasia) 759.89
370	Meningomyelocele 741.9
371	Menkes Syndrome 759.89
372	Metachromatic Leukodystrophy - Late Infantile 759.89
373	Metatropic Dysplasia (Dwarfism) 756.4
374	Methemoglobinemia, NADH-Dependent Diaphorase Deficiency, Type II 289.7
375	Methylmalonic Acidemia (see Acidemia, Methymalonic) 277.8
376	Microcephaly (Two Standard Deviations Below Mean) 742.1
377	Micromelia (see Limb Reduction Defects) 755.21 (upper limb) 755.31 (lower limb)
378	Miller-Dieker Syndrome (see Lissencephaly Syndrome) 742.2
379	Mitochondrial Encephalomyopathy (see Kearns-Sayre Disease) 759.89
380	Moebius Syndrome (see Diplegia, Congenital Facial) 352.6
381	Mohr Syndrome (see Oro-Facio-Digital Syndrome, Mohr Type) 759.89
382	Molybdenum Co-Factor Deficiency 277.8
383	Mondini-Alexander Malformation of Inner Ear (see Ear, Inner Dysplasias) 744.05
384	Moravcsik-Marinesco-Sjogren Syndrome (see Marinesco-Sjorgen Syndrome) 759.89
385	Mucolipidosis III 272.7
386	Mucolipidosis IV 272.7
387	Mucopolysaccharidosis (MPS) F (see Fucosidosis) 271.8
388	Mucopolysaccharidosis (MPS) I-H 277.5
389	Mucopolysaccharidosis (MPS) I-S 277.5
390	Mucopolysaccharidosis (MPS) II 277.5
391	Mucopolusaccharidosis (MPS) III 277.5
392	Mucopolysaccharidosis (MPS) IV 277.5
393	Mucopolysaccharidosis (MPS) VII 277.5
394	Mucosulfatidosis (see Sulfatase Deficiency, Multiple) 277.8

395	Murray Syndrome or Murray Puretic Syndrome (see Fibromatosis, Juvenile Hyaline)
	759.89
396	Muscular Dystrophy, Pseudohypertrophic 359.1
397	Myasthenia Gravis, Familial Infantile (see Myasthenic Syndrome, Familial Infantile Type)
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398	Myasthenic Syndrome, Familial Infantile Type 358.0
399	Myelomeningocele (see Meningomyelocele) 741.9
400	Myopathy, Central Core Disease Type 756.89
401	Myopathy, Disproportionate Fiber Type I 756.89
402	Myopathy, Myotubular 756.89
403	Morquio Syndrome (see Mucopolysaccharidosis IV) 277.5
404	Mucolipidosis I 272.7
405	Myotonic Dystrophy, Congenital 756.89
406	Myopathy, Reducing Body 756.89
407	Myopathy-Metabolic, Mitochondrial Cytochrome C Oxidase Deficiency 756.89
408	Nager Acrofacial Dysostosis (see Acrofacial Dysostosis, Nager Type) 756.0
409	Nasal Hypoplasia-Peripheral Dysostosis-Mental Retardation (see Acrodysostosis) 759.89
410	Nephritis-Deafness (Sensorineural), Hereditary Type 759.89
411	Nettleship-Falls Ocular Albinism (see Albinism, Ocular) 759.89
412	Neuroaxonal Dystrophy, Infantile 759.89
413	Neuronal Ceroid-Lipofuscinoses (NCL) 330.1
414	Neuropathy, Congenital Sensory with Anhidrosis 742.8
415	Neuropathy, Giant Axonal 356.9
416	Nevus, Epidermal Nevus Syndrome 759.89
417	Niemann-Pick Disease, Group A (Acute Neuronopathic Form) 272.7
418	Norman-Roberts Syndrome (see Lissencephaly Syndrome) 742.2
419	Norrie Disease 743.8
420	Ocular Albinism (see Albinism, Ocular) 759.89
421	Oculo-Auriculo-Vertebral Anomaly 759.89
422	Oculo-Cerebro-Renal Syndrome 270.8
423	Oculo-Mandibulo-Facial Syndrome 756.0
424	Opitz Trigonocephaly Syndrome (see C Syndrome) 759.89
425	Optic Atrophy, Infantile Heredofamilial 759.89
426	Optic Disk, Morning Glory Anomaly 743.57
*	Optic Disk, Tilted 743.57 (See #446)
*	Optic Nerve Hypoplasia 743.57 (See #447)
427	Ornithine Transcarbamylase Deficiency 270.6
428	Oro-Facial-Digital Syndrome, Mohr Type 759.89
429	Oro-Palatal-Digital Syndrome, Varadi Type 759.89
430	Osteodystrophy-Mental Retardation, Ruvalcaba Type 759.89
431	Osteogenesis Imperfecta 756.51
432	Osteopetrosis, Malignant Recessive 759.89
433	Oto-Brachio-Renal Dysplasia (see Brachio-Oto-Renal Dysplasia) 759.89

434	Oto-Facio-Cervical Syndrome (see Brachio-Oto-Renal Dysplasia 759.89
435	Oto-Palato-Digital Syndrome I 759.89
436	Palatopharyngeal Incompetence 750.29
437	Pallister-Killian Mosaic Syndrome 758.9
438	Parathyroid Hormone Resistance 759.89
439	Patau Syndrome (see Chromsome 13, Trisomy 13) 758.1
440	Pelizaeus-Merzbacher Syndrome 759.89
441	Pena-Shokeir Syndrome I 759.89
442	Pena-Shokeir Syndrome II (see Cerebro-Oculo-Skeletal Syndrome 759.89
443	Peroxisome Deficiency (see Cerebro-Hepato-Renal Syndrome) 759.89
444	Perrault Syndrome 759.89
445	Peters Anomaly (see Eye, Anterior Segment Dysgenesis) 743.48
446	Optic Disk, Tilted 743.57
447	Optic Nerve Hypoplasia 743.57
*	Phenylketonuria PKU 270.1 (See #565)
448	Phocomelia (see Limb Reduction Defects) 755.21 (upper limb) 755.31 (lower limb)
449	Phytanic Acid Oxidase Deficiency, Infantile Type 356.3
450	Pierre Robin Sequence (see Cleft palate-Micrognathis-Glossoptosis) 756.0
451	Poland Syndrome (Anomaly) 756.8
452	Pompe Disease (see Glycogenosis, Type IIA) 271.0
453	Porencephaly (see Brain, Porencephaly) 742.4
454	Prader-Willi Syndrome 759.81
455	Proprionic Acidemia (see Acidemia, Proprionic) 277.8
456	Prune Belly Syndrome 756.71
457	Pseudo-Arysulfatase A Deficiency (see Metachromatic Leukodystrophies) 759.89
458	Psuedo-Hurler Disease (see G(MI)-Gangliosidosis, Type I) 330.1
459	Psuedopolydystrophy (see Mucolipidosis III) 272.7
460	Pterygium Syndrome, Multiple 759.89
461	Pterygium Syndrome, Popliteal 759.89
462	Pyruvate Carboxylase Deficiency with Lactic Acidemia 759.89
463	Pyruvate Dehydrogenase Deficiency 759.89
464	Radial Dysplasia (see Hand, Radial Club Hand) 754.89
465	REAR Syndrome (see Anus-Hand-Ear Syndrome) 759.89
466	Refsum Disease, Infantile Form (see Phytanic Acid Oxidase Deficiency, Infantile Type)
	356.3
467	Retina, Amaurosis Congenita, Leber Type 362.74
468	Retina, Congenital Detachment of (see Retinal Dysplasia) 743.56
469	Retinal Aplasia (Hereditary), Blindness, or Degeneration, Congenital (see Retina,
	Amaurosis Congenita, Leger Type) 362.74
470	Retinal Dysplasia 743.56
471	Retinal Fold 743.56
472	Retinal Septum, Congenital (see Retinal Fold) 743.56
473	Retinitis Pigmentosa, Congenital (see Retina, Amaurosis Congenita, Leber Type 362.74

474	Retinoblastoma 190.5
475	Retinoic Acid Syndrome (see Fetal Retinoid Syndrome) 760.79
476	Rett Syndrome 330.8
477	Rhizomelic Chondrodysplasia Punctata (see Chondrodysplasia Punctata, Rhizomelic Type)
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478	RHS Syndrome (see Smith-Lemli-Opitz Syndrome) 759.89
479	Richner-Hanhart Syndrome (see Tyrosinemia II, Oregon Type) 270.2
480	Rieger Syndrome 743.44
481	Riley-Day Syndrome (see Dysautonomia I, Riley-Day Type) 742.8
482	Roberts Syndrome 759.89
483	Robin Sequence (see Cleft Palate-Micrognathia-Glossoptosis) 756.0
484	Renal Dysplasia-Retinal Aplasia, Loken-Senior Type 759.89
485	Renal Tubular Acidosis-Sensorineural Deafness 759.89
486	Renal-Brachio-Oto Dysplasia (see Brachio-Oto-Renal Dysplasia) 759.89
487	Rubella Syndrome (see Fetal Rubella Syndrome) 771.0
488	Rubinstein-Taybi Broad Thumbs-Hallux Syndrome 759.89
489	Rud Syndrome (see Seizures-Ichthyosis-Mental Retardation) 759.89
490	Ruvalcaba Syndrome (see Osteodystrophy-Mental Retardation, Ruvalcaba Type 759.89
491	Sacral Agenesis/Regression (see Caudal Regression Syndrome) 759.89
492	Sacrococcygeal Dysgenesis Syndrome 756.19
493	Sandhoff Disease (see G(M2)-Gangliosidosis with Hexoaminidase A and B Deficiency)
	330.1
494	Sanfilippo Syndrome (see Mucopolysaccharidosis III) 277.5
495	Scheie Syndrome (see Mucopolysaccharidosis I-S) 277.5
496	Schwartz-Jampel Syndrome (see Chondrodystrophic Myotonia, Schwartz-Jampel Type)
	756.89
497	Sclerosing Poliodystrophy, Progressive (see Alpers Disease) 759.89
498	Seckel Syndrome 759.89
499	Seitelberger Disease (see Neuroaxonal Dystrophy, Infantile) 759.89
500	Seizures-Ichthyosis-Mental Retardation 759.89
501	Senter Syndrome (see Ichthyosiform Erythrokeratoderma, Atypical with Deafness) 757.1
502	Septo-Optic Dysplasia 742.4
503	Shprintzen Syndrome (see Velo-Cardio-Facial Syndrome) 759.89
504	Shwachman Syndrome 288.0
505	Shy-Magee Disease (see Myopathy, Central Core Disease Type) 756.89
*	Small for Gestational Age (SGA) (See #568)
506	Smith-Lemli-Opitz Syndrome 759.89
507	Smith-Magenis Syndrome (see Chromosome 17, Interstitial Deletion 17p) 758.9
508	Sotos Syndrome (see Cerebral Gigantism) 759.89
509	Spastic Ataxia, Charlevoix-Saguenay Type 759.89
510	Spherophakia-Brachymorphia Syndrome 759.89
511	Sphingomyelin Lipidosis (see Niemann-Pick Disease) 272.7
512	Spina Bifida Cystica with Paralysis (see Meningomyelocele) 741.9

513	Spinal Muscle Atrophy, Infantile Type I 335.11
514	Split Hand Deformity (see Ectrodactyly) 755.25
515	Split Hand Deformity-Mandibulofacial Dysostosis (see Acrofacial Dysostosis, Nager
	Type) 756.0
516	Spondyloepiphyseal Dysplasia Congenita 756.4
517	Spongy Degeneration of Brain (see Brain, Spongy Degeneration 759.89
518	Spongy Glioneuronal Dystrophy (see Alpers Disease) 759.89
519	Steinert Disease (see Myotonic Dystrophy) 756.89
520	Stephens Syndrome (see Kearns-Sayre Disease) 759.89
521	Stickler Syndrome (see Arthro-Ophthalmopathy, Hereditary, Progressive, Stickler Type)
	759.89
522	Sialidase Deficiency (see Mucolipidosis I) 272.7
523	Sjogren-Larsson Syndrome 757.1
524	Sly Syndrome (see Mucopolysaccaridosis VII) 277.5
525	Sulfatase Deficiency, Multiple 277.8
526	Sulfatide Lipidosis (see Metachromatic Leukodystrophies) 759.89
527	Sulfatidosis, Juvenile, Austin Type (see Sulfatase Deficiency, Multiple) 277.8
528	Sweaty Feet Syndrome (see Acidemia, Isovaleric) 277.8
529	TAR Syndrome (see Thrombocytopenia-Absent Radius) 759.89
530	Tay-Sachs Disease (see G(M2)-Gangliosidosis with Hexosaminidase A Disease) 330.1
531	Teschler-Nicola/Killian Syndrome (see Pallister-Killian Mosaic Syndrome) 758.9
532	Thrombocytopenia - Absent Radius Syndrome 759.89
533	Tibial Hypoplasia/Aplasia-Ectrodactyly 755.25 & 755.36
534	Townes-Brocks Syndrome (see Anus-Hand-Ear Syndrome) 759.89
535	Toxoplasmosis, Infantile (see Fetal Toxoplasmosis Syndrome) 771.2
536	Transsuccinylase (E2) Deficiency (see Acidemia, 2-Oxoglutaric) 276.2
537	Treacher Collins Syndrome (see Mandibulofacial Dysostosis) 756.0
538	Tricho-Rhino-Phalangeal Syndrome, Type II 759.89
539	Trichothiodystrophy 759.89
540	Tuberous Sclerosis 759.5
541	Tyrosinemia II, Oregon Type 270.2
542	Usher Syndrome 759.89
543	Vacterl Association (see Vater Association) 759.89
544	Van Bogaert Spongy Degeneration of the Brain (see Brain, Spongy Degeneration) 759.89
545	Varadi-Papp Syndrome (see Oro-Palatal-Digital Syndrome, Varadi Type) 759.89
*	Vater Association 759.89 (See #562)
*	Velo-Cardio-Facial Syndrome 759.89 (See #563)
546	Velopharyngeal Insufficiency (see Palatopharyngeal Incompetence) 750.29
547	Waardenburg Anophthalmia Syndrome (see Anophthalmia-Limb Anomalies) 759.89
548	Walker-Warburg Syndrome 759.89
549	Warburg Syndrome (see Walker-Warburg Syndrome) 759.89
550	Weaver Syndrome 759.89
551	Weill-Marchesani Syndrome (see Spherophakia-Brachymorphia Syndrome) 759.89

552	Werdnig-Hoffman Disease (see Spinal Muscular Atrophy, Infantile Type I) 335.11
553	Whistling Face Syndrome (see Cranio-Carpo-Tarsal Dysplasia, Whistling Face Type)
	759.89
554	Wildervanck Syndrome (see Cervico-Oculo-Acoustic Syndrome) 759.89
555	Williams Syndrome 759.89
556	Wolf-Hirschhorn Syndrome (see Chromosome 4, Monosomy 4p) 758.3
557	X-Linked Mental Retardation, Fragile X Syndrome 758.81
558	Xeroderma Pigmentosum-Mental Retardation 759.89
559	XXXX Syndrome 758.81
560	XXXXX Syndrome 758.81
561	Zellweger Syndrome (see Cerebro-Hepato-Renal Syndrome) 759.89
562	Vater Association 759.89
563	Velo-Cardio-Facial Syndrome 759.89
564	Congenital Hypothyroidism 243
565	Phenylketonuria PKU 270.1
566	Galactosemia 271.1
567	Low Birth Weight (LBW) BW < 1200 Grams (2 lbs, 10 oz)
568	Small for Gestational Age (SGA) GA 37-40 wks & BW < 2000 grams (4 lbs, 6 oz) GA 36 wks & BW < 1875 grams (4 lbs, 2 oz) GA 35 wks & BW < 1700 grams (3 lbs, 12 oz) GA 34 wks & BW < 1500 grams (3 lbs, 5 oz) GA 33 wks & BW < 1325 grams (2 lbs, 15 oz) * Qualifies children up to 1 year of age only
569	Auditory Neuropathy Spectrum Disorder (ANSD)
570	Hypoxic-Ischemic Encephalopathy, Moderate to Severe 768.70