Attachment A

Rare and Expensive Disease List as of December 27, 2010

ICD-9 Code	Disease	Age Group	Guidelines
042.	Symptomatic HIV disease/AIDS (pediatric)	0-20	(A) A child <18 mos. who is known to be HIV seropositive or born to an HIV-infected mother **and:** * Has positive results on two separate specimens (excluding cord blood) from any of the following HIV detection tests: --HIV culture (2 separate cultures) --HIV polymerase chain reaction (PCR) --HIV antigen (p24) N.B. Repeated testing in first 6 mos. of life; optimal timing is age 1 month and age 4-6 mos. **or** * Meets criteria for Acquired Immunodeficiency Syndrome (AIDS) diagnosis based on the 1987 AIDS surveillance case definition
V08	Asymptomatic HIV status (pediatric)	0-20	(B) A child >18 mos. born to an HIV-infected mother or any child infected by blood, blood products, or other known modes of transmission (e.g., sexual contact) who: * Is HIV-antibody positive by confirmatory Western blot or immunofluorescense assay (IFA) **or** * Meets any of the criteria in (A) above
795.71	Infant with inconclusive HIV result	0-12 months	(E) A child who does not meet the criteria above who: * Is HIV seropositive by ELISA and confirmatory Western blot or IFA and is 18 mos. or less in age at the time of the test **or** * Has unknown antibody status, but was born to a mother known to be infected with HIV
270.0	Disturbances of amino-acid transport Cystinosis Cystinuria Hartnup disease	0-20	Clinical history and physical exam; laboratory studies supporting diagnosis. Subspecialist consultation note may be required.
270.1	Phenylketonuria - PKU	0-20	Clinical history and physical exam; laboratory studies supporting diagnosis. Subspecialist consultation note may be required. Lab test: high plasma phenylalanine and normal/low tyrosine
270.2	Other disturbances of aromatic-acid metabolism	0-20	Clinical history and physical exam; laboratory studies supporting diagnosis. Subspecialist consultation note may be required.
270.3	Disturbances of branched-chain amino-acid metabolism	0-20	
270.4	Disturbances of sulphur-bearing amino-acid metabolism	0-20	
270.5	Disturbances of histidine metabolism Carnosinemia Histidinemia Hyperhistidinemia Imidazole aminoaciduria	0-20	Clinical history and physical exam; laboratory studies supporting diagnosis. Subspecialist consultation note may be required.

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ICD-9 Code	Disease	Age Group	Guidelines
270.6	Disorders of urea cycle metabolism	0-20	Clinical history and physical exam; laboratory studies supporting diagnosis. Subspecialist consultation note may be required.
270.7	Other disturbances of straight-chain amino-acid Glucoglycinuria Glycinemia (with methylmalonic acidemia) Hyperglycinemia Hyperlysinemia Pipecolic acidemia Saccharopinuria Other disturbances of metabolism of glycine, threonine, serine, glutamine, and lysine	0-20	Clinical history and physical exam; laboratory studies supporting diagnosis. Subspecialist consultation note may be required.
270.8	Other specified disorders of amino-acid metabolism Alaninemia Ethanolaminuria Glycoprolinuria Hydroxyprolinemia Hyperprolinemia Iminoacidopathy Prolinemia Prolinuria Sarcosinemia	0-20	Clinical history and physical exam; laboratory studies supporting diagnosis. Subspecialist consultation note may be required.
271.0	Glycogenosis	0-20	Clinical history and physical exam; laboratory studies supporting diagnosis. Sub specialist consultation note may be required.
271.1	Galactosemia	0-20	Clinical history and physical exam; laboratory studies supporting diagnosis. Sub specialist consultation note may be required.
271.2	Hereditary fructose intolerance	0-20	Clinical history and physical exam; laboratory studies supporting diagnosis. Sub specialist consultation note may be required.
272.7	Lipidoses	0-20	Clinical history and physical exam; laboratory studies supporting diagnosis. Sub specialist consultation note may be required.
277.00	Cystic fibrosis without ileus.	0-64	Clinical history and physical exam; laboratory studies supporting diagnosis. Sub specialist consultation note may be required.
277.01	Cystic fibrosis with ileus.	0-64	Clinical history and physical exam; laboratory studies supporting diagnosis. Sub specialist consultation note may be required.
277.02	Cystic fibrosis with pulmonary manifestations	0-64	Clinical history and physical exam; laboratory studies supporting diagnosis. Sub specialist consultation note may be required.
277.03	Cystic fibrosis with gastrointestinal manifestations	0-64	Clinical history and physical exam; laboratory studies supporting diagnosis. Sub specialist consultation note may be required.

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ICD-9 Code	Disease	Age Group	Guidelines
277.09	Cystic fibrosis with other manifestations	0-64	Clinical history and physical exam; laboratory studies supporting diagnosis. Sub specialist consultation note may be required.
277.2	Other disorders of purine and pyrimidine metabolism	0-64	Clinical history and physical exam; laboratory studies supporting diagnosis. Sub specialist consultation note may be required. Demonstration of deficient enzyme such as: alpha-L-Idurondase, Iduronosulfate sulfatase, Heparan sulfate sulfatase, N-Acetyl-alpha-D-glucosaminidase, Arylsulfatase B, Beta-Glucuronidase, Beta-Galactosidase, N-Aacetylhexosaminidase-6-SO4 sulfatase.
277.5	Mucopolysaccharidosis	0-64	
277.81	Primary Carnitine deficiency	0-64	Clinical history and physical exam; laboratory or imaging studies supporting diagnosis. Sub specialist consultation note may be required.
277.82	Carnitine deficiency due to inborn errors of metabolism	0-64	Clinical history and physical exam; laboratory or imaging studies supporting diagnosis. Sub specialist consultation note may be required.
277.89	Other specified disorders of metabolism	0-64	Clinical history and physical exam; laboratory or imaging studies supporting diagnosis. Sub specialist consultation note may be required.
284.01	Constitutional red blood cell asplasia	0-20	Clinical history and physical exam; laboratory studies supporting diagnosis. Sub specialist consultation note may be required.
284.09	Other constitutional aplastic anemia	0-20	
286.0	Congenital factor VIII disorder	0-64	
286.1	Congenital factor IX disorder	0-64	
286.2	Congenital factor XI deficiency	0-64	
286.3	Congenital deficiency of other clotting factors	0-64	
286.4	von Willebrand's disease	0-64	
330.0	Leukodystrophy	0-20	Clinical history and physical exam; laboratory or imaging studies supporting diagnosis. Subspecialist consultation note may be required.
330.1	Cerebral lipidoses	0-20	
330.2	Cerebral degenerations in generalized lipidoses	0-20	
330.3	Cerebral degeneration of childhood in other diseases classified	0-20	
330.8	Other specified cerebral degeneration in childhood	0-20	
330.9	Unspecified cerebral degeneration in childhood	0-20	
331.3	Communicating hydrocephalus	0-20	Clinical history and physical exam; imaging studies supporting diagnosis. Sub specialist consultation note may be required.
331.4	Obstructive hydrocephalus	0-20	
333.2	Myoclonus	0-5	Clinical history and physical exam. Sub specialist consultation note may be required.

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ICD-9 Code	Disease	Age Group	Guidelines
333.6	Idiopathic torsion dystonia	0-64	Clinical history and physical exam; laboratory or imaging studies supporting diagnosis. Sub specialist consultation note may be required.
333.7	Symptomatic torsion dystonia	0-64	
333.90	Unspecified extrapyramidal disease and abnormal movement disorder	0-20	Clinical history and physical exam; laboratory or imaging studies supporting diagnosis. Subspecialist consultation note may be required.
334.0	Friedreich's ataxia	0-20	Clinical history and physical exam. Neurology consultation note.
334.1	Hereditary spastic paraplegia	0-20	
334.2	Primary cerebellar degeneration	0-20	
334.3	Cerebellar ataxia NOS	0-20	
334.4	Cerebellar ataxia in other diseases	0-20	
334.8	Other spinocerebellar diseases NEC	0-20	
334.9	Spinocerebellar disease NOS	0-20	
335.0	Werdnig-Hoffmann disease	0-20	Clinical history and physical exam. Neurology consultation note.
335.10	Spinal muscular atrophy unspecified	0-20	
335.11	Kugelberg-Welander disease	0-20	
335.19	Spinal muscular atrophy NEC	0-20	
335.20	Amyotrophic lateral sclerosis	0-20	
335.21	Progressive muscular atrophy	0-20	
335.22	Progressive bulbar palsy	0-20	
335.23	Pseudobulbar palsy	0-20	
335.24	Primary lateral sclerosis	0-20	
335.29	Motor neuron disease NEC	0-20	
335.8	Anterior horn disease NEC	0-20	
335.9	Anterior horn disease NOS	0-20	
341.1	Schilder's disease	0-64	Clinical history and physical examination; supporting imaging studies and neurologic consultation note may be required.
343.0	Diplegic infantile cerebral palsy	0-20	Clinical history and physical exam. Neurology consultation note may be required.
343.2	Quadriplegic infantile cerebral palsy	0-64	Clinical history and physical examination; supporting imaging studies and neurologic consultation note may be required.
344.00	Quadriplegia, unspecified	0-64	
344.01	Quadriplegia, C1-C4, complete	0-64	
344.02	Quadriplegia, C1-C4, incomplete	0-64	
344.03	Quadriplegia, C5-C7, complete	0-64	

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ICD-9 Code	Disease	Age Group	Guidelines
344.04	Quadriplegia, C5-C7, incomplete	0-64	
344.09	Quadriplegia, Other	0-64	
359.0	Congenital hereditary muscular dystrophy	0-64	Clinical history and physical examination; supporting imaging studies and neurologic consultation note may be required.
359.1	Hereditary progressive muscular dystrophy	0-64	Clinical history and physical examination; supporting imaging studies and neurologic consultation note may be required.
359.21	Myotonic muscular dystrophy (Steinert's only)	0-64	Clinical history and physical examination; supporting imaging studies and neurologic consultation note may be required.
437.5	Moyamoya disease	0-64	Clinical history and physical examination; supporting imaging studies and neurologic consultation note may be required.
579.3	Short gut syndrome	0-20	Clinical history and imaging studies supporting diagnosis. Gastrointestinal sub-specialist consultation note may be required.
582.0	Chronic glomerulonephritis with lesion of proliferative glomerulonephritis	0-20	Clinical history, laboratory evidence of renal disease. Nephrology sub-specialist consultation note may be required.
582.1	Chronic glomerulonephritis with lesion of membranous glomerulonephritis	0-20	
582.2	Chronic glomerulonephritis with lesion of membranoproliferative glomerulonephritis	0-20	
582.4	Chronic glomerulonephritis with lesion of rapidly progressive glomerulonephritis	0-20	
582.81	Chronic glomerulonephritis in diseases classified elsewhere	0-20	
582.89	Other Chronic glomerulonephritis with lesion of exudative nephritis interstitial (diffuse) (focal) nephritis	0-20	
582.9	With unspecified pathological lesion in kidney Glomerulonephritis: NOS specified as chronic hemorrhagic specified as chronic Nephritis specified as chronic Nephropathy specified as chronic	0-20	
585.1	Chronic kidney disease, Stage I (diagnosed by a pediatric nephrologists)	0-20	
585.2	Chronic kidney disease, Stage II (mild) (diagnosed by a pediatric nephrologists)	0-20	
585.3	Chronic kidney disease, Stage III (moderate) (diagnosed by a pediatric nephrologists)	0-20	

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ICD-9 Code	Disease	Age Group	Guidelines
585.4	Chronic kidney disease, Stage IV (severe) (diagnosed by a pediatric nephrologists)	0-20	
585.5	Chronic kidney disease, Stage V (diagnosed by a pediatric nephrologists)	0-20	
585.6	End stage renal disease (diagnosed by a pediatric nephrologists)	0-20	
585.9	Chronic kidney disease, unspecified (diagnosed by a pediatric nephrologists)	0-20	
585.6, V45.11	Chronic kidney disease with dialysis	21-64	Clinical history, laboratory, evidence of renal disease. Nephrology sub-specialist consultation note may be required.
741.00	Spina bifida with hydrocephalus NOS	0-64	Clinical history and physical exam, imaging studies supporting diagnosis. Sub-specialist consultation may be required.
741.01	Spina bifida with hydrocephalus cervical region	0-64	
741.02	Spina bifida with hydrocephalus dorsal region	0-64	
741.03	Spina bifida with hydrocephalus lumbar region	0-64	
741.90	Spina bifida unspecified region	0-64	
741.91	Spina bifida cervical region	0-64	
741.92	Spina bifida dorsal region	0-64	
741.93	Spina bifida lumbar region	0-64	
742.0	Encephalocele Encephalocystocele Encephalomyelocele Hydroencephalocele Hydromeningocele, cranial Meningocele, cerebral Menigoencephalocele	0-20	Clinical history and physical examination, radiographic or other neuroimaging studies. Neurology or neurosurgery consultation note may be required.
742.1	Microcephalus Hydromicrocephaly Micrencephaly	0-20	Clinical history and physical examination, radiographic or other neuroimaging studies. Neurology or neurosurgery consultation note may be required.
742.3	Congenital hydrocephalus	0-20	
742.4	Other specified anomalies of brain	0-20	
742.51	Other specified anomalies of the spinal cord Diastematomyelia	0-64	
742.53	Other specified anomalies of the spinal cord Hydromyelia	0-64	

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ICD-9 Code	Disease	Age Group	Guidelines
742.59	Other specified anomalies of spinal cord Amyelia Congenital anomaly of spinal meninges Myelodysplasia Hypoplasia of spinal cord	0-64	
748.1	Nose anomaly - cleft or absent nose ONLY	0-5	Clinical history and physical examination. Radiographic or other imaging studies and specialist consultation note (ENT, plastic surgery) may be required.
748.2	Web of larynx	0-20	Clinical history and physical exam; laboratory or imaging studies supporting diagnosis. Sub-specialist consultation note may be required.
748.3	Laryngotracheal anomaly NEC-Atresia or agenesis of larynx, bronchus, trachea, only	0-20	
748.4	Congenital cystic lung	0-20	Clinical history and physical exam; laboratory or imaging studies supporting diagnosis. Sub-specialist consultation note may be required.
748.5	Agenesis, hypoplasia and dysplasia of lung	0-20	
749.00	Cleft palate NOS	0-20	Clinical history and physical examination. Supporting consultation note from ENT/plastic surgery may be required.
749.01	Unilateral cleft palate complete	0-20	
749.02	Unilateral cleft palate incomplete	0-20	
749.03	Bilateral cleft palate complete	0-20	
749.04	Bilateral cleft palate incomplete	0-20	
749.20	Cleft palate and cleft lip NOS	0-20	
749.21	Unilateral cleft palate with cleft lip complete	0-20	
749.22	Unilateral cleft palate with cleft lip incomplete	0-20	
749.23	Bilateral cleft palate with cleft lip complete	0-20	
749.24	Bilateral cleft palate with cleft lip incomplete	0-20	
749.25	Cleft palate with cleft lip NEC	0-20	
750.3	Congenital tracheoesophageal fistula, esophageal atresia and stenosis	0-3	Clinical history and physical exam; imaging studies supporting diagnosis. Sub-specialist consultation note may be required.
751.2	Atresia large intestine	0-5	Clinical history and physical exam; laboratory or imaging studies supporting diagnosis. Sub-specialist consultation note may be required.
751.3	Hirschsprung's disease	0-15	
751.61	Biliary atresia	0-20	
751.62	Congenital cystic liver disease	0-20	
751.7	Pancreas anomalies	0-5	
751.8	Other specified anomalies of digestive system NOS	0-10	

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ICD-9 Code	Disease	Age Group	Guidelines
753.0	Renal agenesis and dysgenesis, **bilateral only** Atrophy of kidney: congenital infantile Congenital absence of kidney(s) Hypoplasia of kidney(s)	0-20	Clinical history, physical examination, radiographic or other imaging studies. Sub-specialist consultation note may be required.
753.10	Cystic kidney disease, **bilateral only**	0-20	
753.12	Polycystic kidney, unspecified type, **bilateral only**	0-20	
753.13	Polycystic kidney, autosomal dominant, **bilateral only**	0-20	
753.14	Polycystic kidney, autosomal recessive, **bilateral only**	0-20	
753.15	Renal dysplasia, **bilateral only**	0-20	
753.16	Medullary cystic kidney, **bilateral only**	0-20	
753.17	Medullary sponge kidney, **bilateral only**	0-20	
753.5	Exstrophy of urinary bladder	0-20	
756.0	Musculoskeletal--skull and face bones Absence of skull bones Acrocephaly Congenital deformity of forehead Craniosynostosis Crouzon's disease Hypertelorism Imperfect fusion of skull Oxycephaly Platybasia Premature closure of cranial sutures Tower skull Trigonocephaly	0-20	Clinical history, physical examination, radiographic or other imaging studies supporting diagnosis. Sub-specialist consultation note may be required.
756.4	Chondrodystrophy	0-1	
756.50	Osteodystrophy NOS	0-1	
756.51	Osteogenesis imperfecta	0-20	Clinical history, physical exam; imaging studies supporting diagnosis. Sub-specialist consultation note may be required
756.52	Osteopetrosis	0-1	Clinical history, physical examination, imaging studies supporting diagnosis. Sub-specialist consultation note may be required.
756.53	Osteopoikilosis	0-1	
756.54	Polyostotic fibrous dysplasia of bone	0-1	
756.55	Chondroectodermal dysplasia	0-1	
756.56	Multiple epiphyseal dysplasia	0-1	

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ICD-9 Code	Disease	Age Group	Guidelines
756.59	Osteodystrophy NEC	0-1	
756.6	Anomalies of diaphragm	0-1	
756.70	Anomaly of abdominal wall	0-1	
756.71	Prune belly syndrome	0-1	
756.72	Omphalocele	0-1	
756.73	Gastrochisis	0-1	
756.79	Other congenital anomalies of abdominal wall	0-1	
759.7	Multiple congenital anomalies NOS	0-10	Clinical history, physical exam; laboratory or imaging studies supporting diagnosis. Sub-specialist consultation note may be required.
V46.1	Dependence on respirator	1-64	Clinical history and physical exam. Sub-specialist consultation note required.