MALAYSIAN RARE DISEASE LIST

The list was last update in March 2023 and will be updated from time to time by The National Rare Disease Committee, Ministry of Health Malaysia

Source : Medical Development Division, Ministry of Health Malaysia

For futher information please contact : **03-8883 1161 & 1166**

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Source: Medical Development Division, Ministry of Health, Malaysia

1. RARE INHERITED METABOLIC DISEASES

No	Disease name	ICD 10 code
1.1	Urea cycle disorders	E72.2
1.2	Glutaric aciduria type 1	E72.3
1.3	Propionic aciduria	E71.1
1.4	Methylmalonic aciduria	E71.1
1.5	Isovaleric aciduria	E71.1
1.6	3-Hydroxy-3-methylglutaric aciduria (HMG-CoA lyase deficiency)	E71.1
1.7	Maple syrup urine disease	E71.0
1.8	Phenylalanine hydroxylase deficiency (Classical phenylketonuria	E71.0
1.9	Tyrosinaemia type I	E70.2
1.10	Alkaptonuria	E70.2
1.11	Cystathionine beta-synthase deficiency (classical homocystinuria)	E72.1
1.12	Isolated sulfite oxidase deficiency	E72.1
1.13	Phosphoglycerate dehydrogenase deficiency (serine deficiency disorders)	E72.8
1.14	Nonketotic hyperglycinaemia	E72.5
1.15	Ornithine aminotransferase deficiency (Gyrate atrophy of retina and choroidae)	E72.4
1.16	Lysinuric protein intolerance	E72.3
1.17	Cystinuria	E72.0
1.18	Classical galactosaemia	E74.2
1.19	Hereditary fructose intolerance	E74.1
1.20	Transaldolase deficiency	E74.9
1.21	Glycerol kinase deficiency	E74.9
1.22	Glucose transporter I deficiency	E74.9
1.23	Glucose-galactose malabsorption	E74.3
1.24	Fructose-1,6-bisphosphatase deficiency	E74.1
1.25	Pyruvate carboxylase deficiency	E74.4
1.26	Glycogen storage disease type 1a	E74.0

No	Disease name	ICD 10 code
1.27	Glycogen storage disease type 1b	E74.0
1.28	Glycogen storage disease type III	E74.0
1.29	Glycogen storage disease type IV	E74.0
1.30	Carnitine transporter deficiency	E71.3
1.31	Carnitine palmitoyltransferase I (CPTI) deficiency	E71.3
1.32	Carnitine acylcarnitine translocase deficiency	E71.3
1.33	Carnitine palmitoyltransferase II (CPTII) deficiency	E71.3
1.34	Very long - chain acyl CoA dehydrogenase deficiency	E71.3
1.35	Medium - chain acyl CoA dehydrogenase deficiency	E71.3
1.36	Mitochondrial trifunctional protein deficiency	E71.3
1.37	Multiple acyl-CoA dehydrogenase deficiency	E71.3
1.38	Succinyl-CoA:3-Oxoacid-CoA transferase (SCOT) deficiency	E79.8
1.39	Malonyl CoA decarboxylase deficiency	E79.8
1.40	Pyruvate dehydrogenase complex deficiency	E74.4
1.41	Kearns-Sayre syndrome	E88.8
1.42	Chronic Progressive External Ophthalmoplegia (CPEO)	E88.8
1.43	Mitochondrial Encephalopathy Lactic Acidosis and Stroke-like Episodes (MELAS)	E88.8
1.44	Myoclonic Epilepsy Associated with Ragged Red Fibres (MERRF)	E88.8
1.45	Neuropathy Ataxia and Retinitis Pigmentosa (NARP)	E88.8
1.46	Leber's Hereditary Optic Neuropathy (LHON)	E88.8
1.47	Leigh Syndrome	E88.8
1.48	Mitochondrial DNA Depletion Syndromes	E88.8
1.49	Primary mitochondrial disorders	E88.8
1.50	Primary Coenzyme Q10 deficiency	E88.8
1.51	Ethylmalonic encephalopathy (ETHE1)	E88.8
1.52	Creatine biosynthesis defect	E88.8
1.53	Adenylosuccinate lyase deficiency	E79.8
1.54	Adenosine deaminase deficiency	E79.8
1.55	Purine nucleoside phosphorylase [PNP] deficiency	D81.5
1.56	Deoxyguanosine kinase deficiency	E79.8
1.57	Lesch-Nyhan syndrome	E79.1
1.58	Thymidine phosphorylase deficiency [Mitochondrial neurogastrointestinal encephalomyopathy (MNGIE)]	E79.8
1.59	Thymidine kinase 2 deficiency	E79.8

No	Disease name	ICD 10 code
1.60	Aicardi-Goutières syndrome (AGS)	E88.8
1.61	Smith - Lemli - Opitz syndrome	Q87.1
1.62	X-linked dominant chondrodysplasia punctata 2 (Conradi-Hunermann syndrome)	Q77.3
1.63	Cerebrotendinous xanthomatosis	E75.5
1.64	Porphyria	E80.0
1.65	Congenital disorders of glycosylation	E74.4
1.66	MPS I, Hurler, Scheie disease	E76.0
1.67	MPS II, Hunter disease	E76.1
1.68	MPS IIIA, Sanfilippo A disease	E76.2
1.69	MPS IIIB, Sanfilippo B disease	E76.2
1.70	MPS IIIC, Sanfilippo C disease	E76.2
1.71	MPS IIID, Sanfilippo D disease	E76.2
1.72	MPS IVA, Morquio A disease	E76.2
1.73	MPS VI, Maroteaux-Lamy diseae	E76.2
1.74	MPS VII, Sly disease	E76.2
1.75	Fucosidosis	E77.1
1.76	Sialidosis	E77.1
1.77	GM1 - gangliosidosis	E75.1
1.78	GM2 - gangliosidosis	E75.0
1.79	Gaucher disease	E75.2
1.80	Krabbe disease	E75.2
1.81	Metachromatic leukodystrophy	E75.2
1.82	Fabry disease	E75.2
1.83	Niemann-Pick disease type A or B	E75.2
1.84	Niemann-Pick disease type C	E75.2
1.85	CLN1, Santavuori-Haltia disease	E75.4
1.86	CLN2, Jansky-Bielschowsky disease	E75.4
1.87	CLN3, Batten Spielmeyer-Vogt disease	E75.4
1.88	Neuronal ceroid lipofuscinoses type 6, CLN 6	E75.4
1.89	Neuronal ceroid lipofuscinoses type 7, CLN 7	E75.4
1.90	Cystinosis	E72.0
1.91	Mucolipidosis II, I-cell disease	E77.0
1.92	Mucolipidosis III, Pseudo-Hurler polydystrophy	E77.0

No	Disease name	ICD 10 code
1.93	Multiple sulphatase deficiency	E76.2
1.94	Wolman/cholesterol ester storage disease	E75.5
1.95	Pompe disease, GSD type II	E74.0
1.96	Galactosialidosis	E77.1
1.97	Pycnodysostosis	E88.8
1.98	Hermansky-Pudlak Syndrome	E70.3
1.99	Zellweger spectrum disorder	Q87.8
1.100	Rhizomelic chondrodysplasia punctata	Q77.3
1.101	X-linked adrenoleukodystrophy	E71.3
1.102	Peroxisomal fatty acid oxidation defects	E88.8
1.103	Refsum disease	G60.1
1.104	Primary hyperoxaluria type I	E88.8
1.105	Tyrosine hydroxylase deficiency	E70.8
1.106	Aromatic L-amino acid decarboxylase deficiency	E72.8
1.107	Succinic semialdehyde dehydrogenase deficiency	E72.2
1.108	GABA transaminase deficiency	E72.8
1.109	Guanosine 5 triphosphate cyclohydrolase I deficiency	E70.1
1.110	6-Pyruvoyl-tetrahydropterin synthase deficiency	E74.4
1.111	Sepiapterin reductase deficiency	E70.1
1.112	Methylenetetrahydrofolate reductase deficiency	E71.1
1.113	Disorder of cobalamin absorption due to intrinsic factor deficiency	D51.1
1.114	Defect in adenosylcobalamin synthesis-cbl A	E71.1
1.115	Defect in adenosylcobalamin synthesis-cbl B	E71.1
1.116	Disorders of intracellular cobalamin metabolism	E72.8
1.117	Biotinidase deficiency	D81.8
1.118	Holocarboxylase synthetase deficiency	D81.8
1.119	Pyridoxine-dependent seizures (antiquitin deficiency)	D88.8
1.120	Pyridoxal 5'-phosphate deficiency	E53.1
1.121	Molybdenum cofactor deficiency	E79.8
1.122	Menkes syndrome	E83.0
1.123	Fanconi-Biekel disease (GLUT-2 deficiency)	E74.0

2. RARE NEUROLOGICAL & NEUROMUSCULAR DISEASES

No	Disease name	ICD 10 code
2.1	Pelizaeus-Merzbacher disease	E75.2
2.2	Rett syndrome	F84.2
2.3	Huntington's disease	G10.0
2.4	Spinocerebellar ataxia	G11.0
2.5	Friedreich's ataxia	G11.1
2.6	Ataxia telangiectasia	G11.3
2.7	Hereditary spastic paraplegia	G11.4
2.8	Spinal muscular atrophy	G12.0
2.9	Amyotrophic lateral sclerosis (ALS)	G12.2
2.10	Multiple sclerosis	G35.0
2.11	Alexander disease	G37.8
2.12	Charcot-Marie-Tooth disease	G60.0
2.13	Congenital insensitivity to pain with anhidrosis (CIPA)	G60.9
2.14	Myotonia dystrophica	G71.1
2.15	Hypokalaemic periodic paralysis	G72.3
2.16	Joubert syndrome	Q04.9
2.17	Tuberous sclerosis	Q85.1
2.18	Neurofibromatosis type I	Q85.5
2.19	Neurofibromatosis type II	Q85.5
2.20	Acute necrotizing encephalopathy of childhood	G31.8
2.21	Alternating hemiplegia of childhood	G98
2.22	Ataxia with vitamin E deficiency	G11.1
2.23	Autoimmune encephalitis	G04.81
2.24	Autosomal dominant nocturnal frontal lobe epilepsy	G40.0
2.25	Dopa-responsive dystonia	G24.1
2.26	Bickerstaff's brainstem encephalitis	G61.0
2.27	Canavan disease	E75.2
2.28	Childhood absence epilepsy	G40.3
2.29	Childhood ataxia with diffuse central nervous system hypomyelination	E75.2
2.30	Chronic inflammatory demyelinating polyneuropathy	G61.8
2.31	Complex regional pain syndrome	G56.4
2.32	Congenital central hypoventilation syndrome	G47.3

No	Disease name	ICD 10 code
2.33	Dravet syndrome	G40.4
2.34	Early infantile epileptic encephalopathy	G40.3
2.35	Early myoclonic encephalopathy	G40.4
2.36	Epilepsy of infancy with migrating focal seizures	G40.8
2.37	Epilepsy with myoclonic absences	G40.4
2.38	Epilepsy with myoclonic-atonic seizures	G40.4
2.39	Epileptic encephalopathy with continuous spike-and-wave during slow sleep	F80.3
2.40	Episodic ataxia	G11.8
2.41	Febrile infection-related epilepsy syndrome	G40.5
2.42	Folinic acid-responsive seizures	G40.3
2.43	Hereditary sensory and autonomic neuropathy	G60.8
2.44	Hyperekplexia	G25.8
2.45	Hypomyelination with atrophy of basal ganglia and cerebellum	E75.2
2.46	Hypothalamic hamartomas with gelastic seizures	G40.5
2.47	Idiopathic acute transverse myelitis	G37.3
2.48	Idiopathic torsion dystonia	G24.1
2.49	Infantile spasms	G40.4
2.50	Juvenile absence epilepsy	G40.3
2.51	Juvenile myoclonic epilepsy	G40.3
2.52	Landau-Kleffner syndrome	F80.3
2.53	Lennox-Gastaut syndrome	G40.4
2.54	Limbic encephalitis with NMDA receptor antibodies	G13.1
2.55	Neurodegeneration with brain iron accumulation	G23.0
2.56	Neuromyelitis optica	G36.0
2.57	Ohtahara syndrome	G40.8
2.58	Opsoclonus-myoclonus-ataxia syndrome	G25.3
2.59	Primary dystonia	G24.1
2.60	Progressive myoclonic epilepsy	G40.3
2.61	Rasmussen syndrome	G04.8
2.62	Rapid-onset childhood obesity hypothalamic dysfunction hypoventilation- autonomic dysregulation (ROHHAD) syndrome	E66.2 E23.3 G90.8
2.63	Subacute sclerosing panencephalitis	A81.1
2.64	Subependymal giant cell astrocytoma	D43.2

No	Disease name	ICD 10 code
2.65	Benign essential blepharospasm	G24.5
2.66	Early onset generalized dystonia	G24.9
2.67	Focal dystonia	G24.9
2.68	Rippling muscle disease	G71.8
2.69	Narcolepsy	G47.4
2.70	Paraneoplastic neuromyopathy and neuropathy	G13.0
2.71	Myasthenia gravis	G70.0
2.72	Bethlem myopathy	G71.0
2.73	Central core disease	G71.2
2.74	Centronuclear myopathy	G71.2
2.75	Congenital muscular dystrophy	G71.2
2.76	Desmin-related myofibrillar myopathy	G71.8
2.77	Multiminicore myopathy	G71.2
2.78	Myotonia congenita	G71.1
2.79	Myotonic dystrophy	G71.1
2.80	Duchenne muscular dystrophy	G71.0
2.81	Nemaline Rod Myopathy	G71.2
2.82	Schwartz Jampel syndrome	G78.8
2.83	Facioscapulohumeral Muscular Dystrophy	G71.1
2.84	Myotubular Myopathy	G71.2
2.85	Becker Muscular Dystrophy	G71.0
2.86	Limb-girdle Muscular Dystrophy	G71.0
2.87	Congenital myasthenic syndrome	G70.2
2.88	Leukodystrophy	E75.2

3. RARE SKIN DISEASES

No	Disease name	ICD 10 code
3.1	Epidermolysis bullosa	Q81.0
3.2	Inherited ichthyosis	Q80.0
3.3	Ectodermal dysplasias	Q82.4
3.4	Dyskeratosis congenita	Q82.8
3.5	Incontinentia pigmenti	Q82.3
3.6	Netherton syndrome	Q82.8

No	Disease name	ICD 10 code
3.7	Xeroderma pigmentosum	Q82.1
3.8	Congenital generalized lipodystrophy	E88.1
3.9	Acrodermatitis enteropathica	E83.2
3.10	Albinism	E70.3
3.11	PTEN hamartoma tumour syndrome	E71.440
3.12	Klippel-Trenaunay syndrome	Q87.2
3.13	Keratitis-Ichthyosis-Deafness (KID) syndrome	Q80
3.14	Congenital hemidysplasia with icthyosiform erytroderma and limb defects (CHILD syndrome)	Q77.3
3.15	Gorlin syndorome	Q85
3.16	Complex vascular malformation	D18.0
3.17	Autoimmune blistering disease • Bullous pemphigoid • Pemphigus vulgaris • Pemphigus foliaceous • Linear IgA bullous dermatosis • Epidermolysis bullosa acquisita • Dermatitis herpetiformis • Paraneoplastic pemphigus • Cicatricial pemphigoid	 L12.0 L10.0 L10.2 L13.8 L12.3 L13.0 L10 H13.3
3.18	H syndrome	Q82.9
3.19	Generalized pustular psoriasis	L40.1
3.20	Hailey-hailey disease/Benign familial pemphigus	L11.8
3.21	Darier's disease	L11.8
3.22	Pyoderma gangrenosum	L08.0
3.23	Porokeratosis	Q82.8

4. RARE ENDOCRINE DISEASES

No	Disease name	ICD 10 code
4.1	Pseudohypoparathyroidism	E20.1
4.2	hypophosphatemic rickets	E83.3
4.3	Laron syndrome (Laron Dwarfism)	E34.3
4.4	Bardet-Biedl syndrome	Q87.8
4.5	Congenital hyperinsulinism	E16.1
4.6	Congenital adrenal hypoplasia	Q89.1

No	Disease name	ICD 10 code
4.7	Congenital adrenal hyperplasia	E25.0
4.8	Kallmann syndrome	E23.0
4.9	Adrenocortical carcinoma	C74.9
4.10	Craniopharyngioma	D44.4
4.11	Disorders of sex development	Q56.4
4.12	Neonatal diabetes	P70.2
4.13	Pallister-Hall syndrome	D33.0
4.14	Cushing disease/syndrome	E24.0
4.15	Acromegaly/gigantism	E22.0
4.16	Neuroendocrine neoplasm	C7A.1
4.17	Pheochromocytoma	C74.1
4.18	Paraganglioma	D44.7
4.19	Familial endocrine tumour syndrome (multiple endocrine neoplasia)	E31.2
4.20	Central diabetes insipidus	E23.2
4.21	Nephrogenic diabetes insipidus	N25.1
4.22	Primary adrenocortical insufficiency	E27.1
4.23	Polyostotic fibrous dysplasia (McCune-Albright syndrome)	Q78.1
4.24	Alstrom syndrome	E34.8
4.25	Primary pituitary hypophysitis	E23.6
4.26	ROHHAD Rapid-onset Obesity with Hypothalamic dysfunction Hypoventilation Autonomic Dysregulation	E66.2 E23.3 G90.8
4.27	Vitamin D dependent rickets type 2	E83.3
4.28	Pseudohypoaldosteronism type 1	N25.8
4.29	Wolfram syndrome (DIDMOAD)	E34.8
4.30	Familial hypercholesterolemia (severe homozygous/autosomal recessive)	E78.01
4.31	Congenital generalised lipodystrophy (Berardinelli-Seip syndrome)	E88.1
4.32	Donohue syndrome (Leprechaunism)	E34.8

5. RARE DISEASES AFFECTING BONE, CARTILAGE AND CONNECTIVE TISSUE

No	Disease name	ICD 10 code
5.1	Hypophosphatasia	E83.39
5.2	Achondroplasia	Q77.4
5.3	Osteogenesis imperfecta	Q78.0
5.4	Ehlers-Danlos syndrome	Q79.6
5.5	Osteochondrodysplasias (Primary bone dysplasias)	Q78
5.6	Osteopetrosis	Q78.2
5.7	Syndromic craniosynostosis	Q75.0

6. RARE RHEUMATOLOGICAL DISEASES

No	Disease name	ICD 10 code
6.1	Acquired purpura fulminans	D65
6.2	Kawasaki disease	M30.3
6.3	Pigmented villonodular synovitis	M12.2
6.4	Osteonecrosis	M87
6.5	Polymyalgia Rheumatica	M35.3
6.6	Felty Syndrome	M05.0
6.7	Dermatomyositis	M33.0
6.8	Polymyositis	M33.2
6.9	Inflammatory Inclusion body myositis	M60.8
6.10	Primary Sjogren syndrome	M35.0
6.11	Acquired thrombotic thrombocytopenic purpura	M31.1
6.12	Juvenile Idiopathic Arthritis	M08.0
6.13	Idiopathic juvenile osteoporosis	M81.5
6.14	SAPHO syndrome	M86.3
6.15	Adult-onset Stills disease	M06.1
6.16	Diffuse Systemic sclerosis	M34.0
6.17	Systemic polyarteritis nodosa	M30.0
6.18	Autoimmune necrotising myositis	G72.4
6.19	Paediatric systemic lupus erythematosus	M32.0
6.20	Pulmonary arterial hypertension associated with connective tissue disease	I27.2
6.21	Pyogenic arthritis-pyoderma gangrenosum-acne syndrome	M04.8

No	Disease name	ICD 10 code
6.22	Pauci immune glomerulonephritis	N05.7
6.23	Pediatric Castleman disease	D36.0
6.24	PFAPA (Periodic fever - aphthous stomatitis-pharyngitis - adenopathy) syndrome	E85.0
6.25	Henoch Schonlein Purpura	D69.0
6.26	Takayasu arteritis	M31.4
6.27	Beh et disease	M35.2
6.28	Hyperimmunoglobulin D with periodic fever	E85.0
6.29	Eosinophilic granulomatosis with polyangiitis*	M30.1
6.30	Microscopic polyangiitis	M31.7
6.31	Primary vasculitis of central nervous system	I67.7
6.32	Mixed connective tissue disease	M35.1
6.33	NLRP-12 associated hereditary periodic fever syndrome	E85.0
6.34	Catastrophic antiphospholipid syndrome	D68.6
6.35	Blau syndrome	M04.8
6.36	Sarcoidosis	D86.0
6.37	Chronic nonbacterial osteomyelitis (Chronic recurrent multifocal osteomyelitis)	M86.3
6.38	Majeed syndrome	M04.8
6.39	Granulomatosis with polyangiitis	M31.3
6.40	IgG4 related disease	M35.0 D89.89
6.41	Giant cell arteritis	M31.5
6.42	Ankylosing spondylitis	M45.0-9
6.43	Rheumatoud arthritis	M05
6.44	Systemic lupus erythematosus	M32
6.45	Raynaud's syndrome	I 73
6.46	Psoriatic arthritis	L40.5

7. RARE HEMATOLOGICAL DISEASES

No	Disease name	ICD 10 code
7.1	Atypical haemolytic uremic syndrome (aHUS)	D58.8
7.2	Paroxysmal nocturnal haemoglobinuria (PNH)	D59.5
7.3	Haemophilia with inhibitor	D66

No	Disease name	ICD 10 code
7.4	Primary thrombophilia	D68.5
7.5	Glanzmann thrombasthenia	D69.1
7.6	Erdheim-Chester disease (non-LCH)	D76.3
7.7	Multicentric Castleman Disease	D36.0
7.8	Wiskot-Aldrich syndrome	D82.0
7.9	Acquired haemophilia	D68.4
7.10	Autosomal recessive thrombotic thrombocytopaenic purpura	M31.1
7.11	Acquired thrombotic thrombocytopaenic purpura	M31.1
7.12	Langerhans cell histiocytosis (LCH)	C96.0
7.13	Haemophagocytic lymphohistiocytosis (HLH)	D76.1
7.14	Severe aplastic anaemia (SAA)	D61.9
7.15	POEMS syndrome	D47.7
7.16	AL amyloidosis	E85.9
7.17	Cold haemagglutinin disease (CHAD)	D59.1
7.18	Diamond Blackfan Anemia (DBA)	D61.01

8. RARE IMMUNOLOGICAL DISEASES

No	Disease name	ICD 10 code
8.1	Functional Disorders of Polymorphonuclear Neutrophils	D71
8.2	Immunodeficiency with Predominantly Antibody Defects	D80
8.3	Combined Immunodeficiencies	D81
8.4	Immunodeficiency associated with other major defects	D82
8.5	Common Variable Immunodeficiency	D83
8.6	Other Immunodeficiencies	D84
8.7	Other Disorders Involving the Immune Mechanism, not elsewhere classified	D89

9. RARE PULMONARY DISORDERS

No	Disease name	ICD 10 code
9.1	Cystic fibrosis	E84.0
9.2	Primary ciliary dyskinesia	Q34.8
9.3	Idiopathic pulmonary hemosiderosis	E83.1

No	Disease name	ICD 10 code
9.4	Congenital aerodigestive disease	J39.8
9.5	Primary interstitial lung disease	J84.9

10. RARE CARDIAC DISEASE

No	Disease name	ICD 10 code
10.1	Brugada syndrome	I49.8
10.2	Primary cardiomyopathy	I42.9

11. RARE RENAL DISEASES

No	Disease name	ICD 10 code
11.1	Autosomal recessive polycystic kidney disease	Q61.1
11.2	Autosomal dominant polycystic kidney disease	Q61.2
11.3	Alport Syndrome	Q87.8
11.4	Anti-glomerular basement membrane disease (Goodpasture)	N08.8
11.5	Atypical Haemolytic Uraemic Syndrome	D58
11.6	Autosomal recessive distal renal tubular acidosis	N25.8
11.7	Autosomal recessive proximal renal tubular acidosis	N25.8
11.8	Autosomal recessive polycystic kidney disease	Q61.1
11.9	(ARPKD) Batters syndrome	
11.10	Bardet-Biedl syndrome	Q87.8
11.11	Bartter syndrome	E26.8
11.12	BK-Virus Nephropathy	B97.89
11.13	Congenital nephropatic syndrome Finnish type	N04.8
11.14	Cystinosis	E72.0
11.15	Dense deposit disease (DDD)	N04.6
11.16	Denys-Drash Syndrome	N04.1
11.17	Dent Disease	N25.8
11.18	Exstrophy of bladder	Q64.1
11.19	Fabry disease	E75.2
11.20	Fanconi Anaemia	D61.0
11.21	Fanconi syndrome	E72.0
11.22	Familial hypomagnesaemia with hypercalciuria and nephrocalciuria	E83.4

No	Disease name	ICD 10 code
11.23	Focal segmental glomerulosclerosis	N04.1
11.24	Frasier syndrome	N04.1
11.25	Giant vessel / Giant cell arteritis	M31.6
11.26	Gitelman syndrome	N15.8
11.27	Hypophosphatemic rickets (X-linked)	E83.3
11.28	IgA vasculitis (Henoch Schonlein)	D69.0
11.29	Large vessel vasculitis	D69.0
11.30	Lesch Nyhan Syndrome	E79.1
11.31	Liddle syndrome	I15.1
11.32	Lowe's syndrome	E72.0
11.34	Medium vessel vasculitis	D69.0
11.35	Membranous nephropathy	N04.2
11.36	Membranoproliferative glomerunephritis	N00.5
11.37	Microscopic polyangiitis	M31.7
11.38	Minimal change nephropathy	N04.0
11.39	Multicystic dysplastic kidneys (bilateral)	Q61.4
11.40	Nail patella syndrome	Q87.2
11.41	Nephropathic cystinosis	E72.0
11.42	Nephrogenic diabetes insipidus	N25.1
11.43	Nephrogenic syndrome of inappropriate antidiuresis	E22.2
11.44	Nephronophthisis	Q61.5
11.45	Primary hyperoxaluria	E74.8
11.46	Prune belly syndrome	Q79.4
11.47	Pure red cell aplasia	D60.0
11.48	Renal agenesis	Q60.0 - Q60.2
11.49	Renal coloboma syndrome	Q60.4
11.50	Shiga Toxin associated hemolytic uremic syndrome	D58.8
11.51	Small vessel vasculitis (ANCA associated)	I77.6
11.52	Steroid resistant nephrotic syndrome	N04.9
11.53	Steroid sensitive nephrotic syndrome	N04.9
11.54	Takayasu arteritis	M31.4
11.55	Tuberous sclerosis	Q85.1
11.56	WAGR (Wilm' tumour, Anirida, genitourinary anomalies and mental retardation	Q87.8

12. RARE GASTROINTESTINAL AND HEPATIC DISEASES

No	Disease name	ICD 10 code
12.1	Progressive familial intrahepatic cholestasis	K76.8
12.2	α1-Antitrypsin deficiency	E88.01
12.3	Congenital bile acid synthesis defect	E78.70
12.4	Wilson's disease	E83.0
12.5	Hereditary haemochromatosis	E83.1

13. RARE MALFORMATIONS, DEVELOPMENTAL ANOMALIES AND GENETIC SYNDROME

No	Disease name	ICD 10 code
13.1	Alagille syndrome	Q44.7
13.2	Beckwith Wiedemann syndrome	Q87.3
13.3	Marfan syndrome	Q87.4
13.4	Russel silver syndrome	Q87.1
13.5	Mowat-Wilson syndrome	Q43.1
13.6	Noonan syndrome	Q87.1
13.7	Cornelia de Lange syndrome	Q87.1
13.8	Kabuki syndrome	Q87.0
13.9	Sotos syndrome	Q87.3
13.10	Treacher collins syndrome	Q75.4
13.11	Turner syndrome	Q96.0
13.12	Fragile X syndrome	Q99.2
13.13	Cardiofaciocutaneous syndrome	Q87.8
13.14	Angelman syndrome	Q93.5
13.15	DiGeorge syndrome (22q11.2 deletion syndrome)	D82.1
13.16	Prader-Willi syndrome	Q87.1
13.17	WAGR syndrome (Wilms' tumor-Aniridia-Genitourinary anomalies-mental retardation)	Q87.8
13.18	Miller-Dieker syndrome	Q04.3
13.19	Rubinstein-Taybi syndrome	Q87.2
13.20	Williams syndrome	Q93.8
13.21	Von Hippel-Lindau disease	Q85.8
13.21	Cockayne syndrome	Q87.1

No	Disease name	ICD 10 code
13.23	Hutchinson-Gilford progeria syndrome	E34.8
13.24	Barth syndrome	E71.1
13.25	Costello syndrome	Q87.8
13.26	CHARGE syndrome	Q87.8
13.27	Coffin-Siris syndrome	Q87.1
13.28	Renal coloboma syndrome	Q60.4
13.29	Rare chromosomal abnormality with multiple malformations, physical and learning disabilities	Q99.9
13.30	Mayer-Rokitansky-Küster-Hauser syndrome	Q51.8

14. RARE INFECTIONS

No	Disease name	ICD 10 code
14.1	Herpes simplex viral encephalitis	B00.4
14.2	Extrapulmonary tuberculosis	A18.89

15. RARE CANCER

No	Disease name	ICD 10 code
15.1	Dermatofibrosarcoma protuberans	C49.9
15.2	Advanced melanoma	C43.9
15.3	Giant cell tumour of bone	D48.0
15.4	Malignant mesothelioma	C45.0
15.5	Iodine refractory thyroid cancer	C73
15.6	Oligodendroglioma	C71.9
15.7	Medulloblastoma	C71.6
	ADRENAL GLAND	
15.8	Adrenal Gland Carcinoma	C74.9
15.9	Adrenal cortical carcinoma	C74.0
15.10	Ganglioneuroblastoma	C74.9
15.11	Giant cell sarcoma (except of bone)	C74.9
15.12	Large cell neuroendocrine carcinoma	C74.9
15.13	Liposarcoma	C74.9
15.14	Lymphoma	C74.9

No	Disease name	ICD 10 code
15.15	Neuroblastoma	C74.9
15.16	Pheochromocytoma	C74.9
15.17	Primitive neuroectodermal tumor	C74.9
	ANUS	
15.18	Carcinoma of the anus	C21.0
15.19	Melanoma of the anus	C21.0
15.20	Lymphoma	C21.0
15.21	Neuroendocrine carcinoma	C21.0
15.22	Sarcoma	C21.0
15.23	Plasmacytoma	C21.0
	BILE DUCT	
15.24	Carcinoid tumor of the bile duct	C24.0
15.25	Klatskin tumor	C24.0
15.26	Lymphoma	C24.0
	BLADDER	
15.27	Sarcoma of the bladder	C67.9
15.28	Lymphoma	C67.9
	BONE	
15.29	All bone tumours	C41.9
	BRAIN	
15.30	Oligodendroglioma	C71.9
15.31	Ependymoma	C71.9
15.32	Lymphoma (any other terms)	C71.9
15.33	Medulloblastoma	C71.9
15.34	Intracranial Germ Cell Tumor	C71.9
15.35	Plasmacytoma	C71.9
	BREAST	
15.36	Lymphoma	C50.9
15.37	Carcinoid tumor	C50.9
15.38	Sarcoma	C50.9
15.39	Phylloides tumor	C50.9
	BRONCHUS / LUNG	
15.40	Adenoid cystic carcinoma	C34.9
15.41	Carcinoid tumor	C34.9

No	Disease name	ICD 10 code
15.42	Lymphoma	C34.9
15.43	Giant cell tumor	C34.9
15.44	Mesothelioma	C34.9

16. RARE EYE DISEASES

No	Disease name	ICD 10 code
16.1	Retinitis pigmentosa	H35.5
16.2	Familial exudative vitreoretinopathy	H35.0
16.3	Norrie disease	H35.5

17. RARE CARDIAC DISEASES

No	Disease name	ICD 10 code	ICD 11 code
17.1	Transthyretin Amyloid Cardiomyopathy (ATTR-CM)	E85.4 I42.9	BC43.20 5D00.20

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