List of recommended conditions to ask the state to include in the rare disease dashboard- Request made on 2/21/2025

Rare Genetic Disorders

- Cystic Fibrosis – E84.9
- Spinal Muscular Atrophy (SMA) – G12.0
- Duchenne Muscular Dystrophy (DMD) – G71.0
- Ehlers-Danlos Syndrome (EDS) – Q79.6
- Marfan Syndrome – Q87.4
- Fragile X Syndrome – Q99.2
- ICD-10 Code: Q99.9 – *Chromosomal abnormality, unspecified*

Hematologic (Blood) Disorders

- Sickle Cell Disease (SCD) – D57.1
- Hemophilia A – D66
- Hemophilia B – D67
- Von Willebrand Disease – D68.0
- Thalassemia – D56.9
- Aplastic Anemia – D61.9
- Paroxysmal Nocturnal Hemoglobinuria (PNH) – D59.5

Metabolic & Mitochondrial Disorders

- Phenylketonuria (PKU) – E70.0
- Maple Syrup Urine Disease (MSUD) – E71.0
- Mitochondrial Diseases (e.g., Leigh Syndrome, MELAS) – G31.81
- Gaucher Disease – E75.22
- Fabry Disease – E75.21
- Pompe Disease – E74.02
- Hypophosphatasia (HPP) – E83.30
- ICD-10 Code: E88.9 – *Metabolic disorder, unspecified*

Neurological & Neuromuscular Disorders

- Huntington's Disease – G10
- Amyotrophic Lateral Sclerosis (ALS) – G12.21
- Rett Syndrome – F84.2
- Tay-Sachs Disease – E75.02
- Angelman Syndrome – Q93.51

- Charcot-Marie-Tooth Disease – G60.0
- Batten Disease – E75.4

Endocrine Disorders

- Congenital Adrenal Hyperplasia (CAH) – E25.0
- Congenital Hypothyroidism – E89.1

Immune Deficiency Disorders

- Severe Combined Immunodeficiency (SCID) – D81.9
- Chronic Granulomatous Disease (CGD) – D71
- Common Variable Immune Deficiency (CVID) – D83.9
- **ICD-10 Code: D84.9** – *Immunodeficiency, unspecified*
- ***P78.83*** – *Neonatal immune system disorder*

Rare Connective Tissue & Skeletal Disorders

- Osteogenesis Imperfecta (OI) – Q78.0
- Fibrodysplasia Ossificans Progressiva (FOP) – M61.19

Lysosomal Storage Disorders

- GM1 Gangliosidosis – E75.0
- GM2 Gangliosidosis (Including Tay-Sachs Disease) – E75.1
- Neuronal Ceroid Lipofuscinoses (Batten Disease) – E75.4

Peroxisomal Disorders

- X-linked Adrenoleukodystrophy (ALD) – E71.52
- Other Peroxisomal Disorders – E71.59

Cystic Fibrosis & Pulmonary Disorders

- Cystic Fibrosis with Pulmonary Manifestations – E84.0
- Cystic Fibrosis, Unspecified – E84.9

Hearing & Vision Screening

- Sensorineural Hearing Loss, Unspecified – H90.5
- Blindness, Both Eyes – H54.0

Comprehensive List of Childhood Cancers
Leukemias

- Acute Lymphoblastic Leukemia (ALL) – C91.0
- Acute Myeloid Leukemia (AML) – C92.0
- Juvenile Myelomonocytic Leukemia (JMML) – C93.30

Lymphomas

- Hodgkin Lymphoma – C81.9
- Non-Hodgkin Lymphoma – C85.9

Central Nervous System (CNS) Tumors

- Neuroblastoma – C74.90
- Medulloblastoma – C71.6
- Glioblastoma – C71.0
- Astrocytoma – C71.0
- Ependymoma – C71.9

Bone and Soft Tissue Sarcomas

- Osteosarcoma – C41.9
- Ewing Sarcoma – C41.9
- Rhabdomyosarcoma – C49.9
- Synovial Sarcoma – C49.2
- Fibrosarcoma – C49.1

Renal and Liver Tumors

- Wilms Tumor (Nephroblastoma) – C64.9
- Hepatoblastoma – C22.2

Retinoblastoma and Germ Cell Tumors

- Retinoblastoma – C69.2
- Germ Cell Tumors (Malignant) – C62.9
- Ovarian Dysgerminoma – C56.9
- Testicular Yolk Sac Tumor – C62.9

Other Rare Pediatric Cancers

- Adrenocortical Carcinoma – C74.9
- Thyroid Carcinoma – C73
- Langerhans Cell Histiocytosis (LCH, malignant cases) – C96.6
- NUT Carcinoma – C76.0

- **ICD-10 Code: C80.1** – *Malignant (primary) neoplasm, unspecified site*
- **Alternative Code: D49.9** – *Neoplasm of unspecified behavior, site unspecified*