NEVADA RARE DISEASE ADVISORY COUNCIL

Rare Disease Dashboard: Current Conditions & Proposed CHD Additions

Overview

The Nevada Rare Disease Dashboard was originally established to track childhood cancer, lupus, and sickle cell disease. Through collaboration between the NV-RDAC and the State, the dashboard has been significantly expanded to include the comprehensive list of rare disease categories shown in this document.

This document provides a reference of current dashboard conditions and proposes the addition of **Congenital & Structural Heart Disorders** to further expand cardiovascular rare disease tracking.

Yellow highlighting indicates proposed new additions.

Proposed Addition: Congenital & Structural Heart Disorders

These high-impact congenital and structural heart disorders require coordinated care across multiple pediatric subspecialties and have significant implications for healthcare planning.

ICD-10	Condition	Est. Prevalence
Q23.4	Hypoplastic Left Heart Syndrome (HLHS)	2–3 per 10,000
Q22.4	Tricuspid Atresia	1 per 10,000
Q21.3	Tetralogy of Fallot (TOF)	3–4 per 10,000
Q20.3	Transposition of the Great Arteries (TGA)	2–3 per 10,000
Q26.2	Total Anomalous Pulmonary Venous Return (TAPVR)	0.5–1 per 10,000
Q20.0	Common Truncus (Truncus Arteriosus)	0.5–1 per 10,000
Q25.5	Pulmonary Atresia (all types)	1 per 10,000
Q22.5	Ebstein Anomaly of the Tricuspid Valve	0.5–1 per 10,000
Q89.3	Heterotaxy Syndrome (multisystem, multi-subspecialist care)	~2 per 10,000
Q24.8/Q24.9	Other / Unspecified Severe CHD (subset only)	Variable

Note on Rare Disease Definition: Under the federal Orphan Drug Act, a rare disease is defined as a condition affecting fewer than 200,000 people in the U.S., or approximately fewer than 6 per 10,000. All proposed conditions meet this threshold—most are well under 1 per 10,000, making them significantly rarer than the federal cutoff.

Current Dashboard Conditions (Reference)

The following conditions are currently tracked on the Nevada Rare Disease Dashboard.

Rare Genetic Disorders	
E84.9	Cystic Fibrosis
G12.0	Spinal Muscular Atrophy (SMA)
G71.0	Duchenne Muscular Dystrophy (DMD)
Q79.6	Ehlers-Danlos Syndrome (EDS)
Q87.4	Marfan Syndrome
Q99.2	Fragile X Syndrome
Q99.9	Chromosomal abnormality, unspecified

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

Metabolic & Mitochondrial Disorders	
E70.0	Phenylketonuria (PKU)
E71.0	Maple Syrup Urine Disease (MSUD)
G31.81	Mitochondrial Diseases (Leigh Syndrome, MELAS)
E75.22	Gaucher Disease
E75.21	Fabry Disease
E74.02	Pompe Disease
E83.30	Hypophosphatasia (HPP)
E88.9	Metabolic disorder, unspecified

Neurological & Neuromuscular Disorders	
G10	Huntington’s Disease
G12.21	Amyotrophic Lateral Sclerosis (ALS)
F84.2	Rett Syndrome
E75.02	Tay-Sachs Disease
Q93.51	Angelman Syndrome
G60.0	Charcot-Marie-Tooth Disease
E75.4	Batten Disease

Endocrine Disorders	
E25.0	Congenital Adrenal Hyperplasia (CAH)
E89.1	Congenital Hypothyroidism

Immune Deficiency Disorders	
D81.9	Severe Combined Immunodeficiency (SCID)

D71	Chronic Granulomatous Disease (CGD)
D83.9	Common Variable Immune Deficiency (CVID)
D84.9	Immunodeficiency, unspecified
P78.83	Neonatal immune system disorder

Rare Connective Tissue & Skeletal Disorders	
Q78.0	Osteogenesis Imperfecta (OI)
M61.19	Fibrodysplasia Ossificans Progressiva (FOP)

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

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

Cystic Fibrosis & Pulmonary Disorders	
E84.0	Cystic Fibrosis with Pulmonary Manifestations
E84.9	Cystic Fibrosis, Unspecified

Hearing & Vision Screening	
H90.5	Sensorineural Hearing Loss, Unspecified
H54.0	Blindness, Both Eyes

Childhood Cancers (Current Dashboard)

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

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

Central Nervous System (CNS) Tumors	
C74.90	Neuroblastoma
C71.6	Medulloblastoma
C71.0	Glioblastoma / Astrocytoma
C71.9	Ependymoma

Bone and Soft Tissue Sarcomas	
C41.9	Osteosarcoma / Ewing Sarcoma
C49.9	Rhabdomyosarcoma
C49.2	Synovial Sarcoma
C49.1	Fibrosarcoma

Renal and Liver Tumors	
C64.9	Wilms Tumor (Nephroblastoma)
C22.2	Hepatoblastoma

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

Other Rare Pediatric Cancers	
C74.9	Adrenocortical Carcinoma
C73	Thyroid Carcinoma
C96.6	Langerhans Cell Histiocytosis (LCH, malignant cases)
C76.0	NUT Carcinoma
C80.1	Malignant (primary) neoplasm, unspecified site
D49.9	Neoplasm of unspecified behavior, site unspecified

Summary

Current dashboard: ~70 conditions across rare genetic, hematologic, metabolic, neurological, endocrine, immune, connective tissue, lysosomal, peroxisomal, pulmonary, sensory, and childhood cancer categories.

Proposed addition: 10 high-impact Congenital & Structural Heart Disorders requiring coordinated multi-subspecialty care.

Requested Action

Council review and input on the proposed Congenital & Structural Heart Disorders list prior to formal recommendation to the State for dashboard inclusion.