

Table S3 - Inclusion Rules for Confirmed Lethal and High Severity Classification.

Rule ID	Severity Level	Clinical Category	Keywords / Patterns (Complete)
C1	Confirmed Lethal	Absolute Lethality	lethal, fatal, thanatophoric, hydrops fetalis
C2	Confirmed Lethal	Infantile-Lethal Syndrome	achondrogenesis, zellweger syndrome, walker-warburg, meckel syndrome, meckel-gruber, neu-laxova, pena-shokeir, fowler syndrome, alpers-huttenlocher, alpers syndrome, menkes disease, krabbe disease, tay-sachs, sandhoff disease, canavan disease, wolman disease, niemann-pick disease, type a, herlitz, restrictive dermopathy, hydroletharus syndrome, gracile syndrome, rhabdoid tumor, leigh syndrome, leigh-like, atypical teratoid rhabdoid tumor, glycine encephalopathy, nonketotic hyperglycinemia, alveolar capillary dysplasia, pontocerebellar hypoplasia, type 2, infantile neuroaxonal dystrophy, inad, ethylmalonic encephalopathy, arc syndrome
C3	Confirmed Lethal	Lethal Subtype	(Osteogenesis imperfecta + type II/perinatal), (Hypophosphatasia + infantile/perinatal), (Spinal muscular atrophy / sma + type 0/I/Werdnig-Hoffmann), (Pompe disease / Glycogen storage disease, type II + infantile), (Marfan syndrome + neonatal), (Osteopetrosis + infantile/malignant), (Epidermolysis bullosa + pyloric atresia), (Urea cycle + neonatal)
C4	Confirmed Lethal	Lethal Clinical Pattern	SEVERITY + EARLY ONSET + CRITICAL SYSTEMS
C5	Confirmed Lethal	Catastrophic Pattern	CRITICAL SYSTEMS + CATASTROPHIC FINDINGS
H1	High Severity	Potentially Treatable but Lethal	severe combined immunodeficiency, scid, hemophagocytic lymphohistiocytosis, fhl, urea cycle, maple syrup urine disease, msud, propionic acidemia, methylmalonic acidemia, glutaric aciduria type 1, cystic fibrosis, duchenne muscular dystrophy
H2	High Severity	High Risk of Sudden Death	arrhythmogenic, long qt syndrome, brugada syndrome, catecholaminergic polymorphic ventricular tachycardia, cpvt, pulmonary arterial hypertension, marfan syndrome, loeys-dietz syndrome, ehlers-danlos syndrome, vascular type, aortic dissection
H3	High Severity	Variable / Later Onset Lethality	niemann-pick disease, type c, gaucher disease, type 3, ataxia-telangiectasia, fanconi anemia, dyskeratosis congenita, adams-oliver syndrome, diaphragmatic hernia, hypoplastic left heart syndrome, renal agenesis, renal aplasia
H4	High Severity	High-Risk Metabolic Crisis	mcad, vlcd, lchad deficiency, mitochondrial trifunctional protein deficiency, salt-wasting
H5	High Severity	Progressive Critical System Disease	(progressive) + CRITICAL_SYSTEMS
H6	High Severity	Heritable Malignancy	cancer, carcinoma, sarcoma, lethal_pheno_confirmed_v11.txt, lymphoma, blastoma, glioma, melanoma, malignant

Note: These rules are applied sequentially after a phenotype has passed all exclusion checks. All keywords are listed completely.