

Conditions Associated with SIDS		NEXTGEN	NEXTGEN PLUS
Condition	Treatment		
Long QT Syndrome (& very rarely Short QT Syndrome)	Medication and avoiding QT prolonging medications	yes	yes
Brugada Syndrome	Avoiding certain drugs can prevent cardiac arrest, implantable defibrillator	yes	yes
Familial WPW (& very rarely hypertrophic cardiomyopathy)	Medication, ablation therapy, some medication absolutely contraindicated		yes
Life Threatening Conditions			
Condition	Treatment		
Tyrosinaemia Type I	Nitisinone and low protein diet >90% survival	yes	yes
Ornithine Transcarbamylase (OTC) Deficiency	Nitisinone and low protein diet & liver transplant	yes	yes
X-linked Severe Combined Immunodeficiency (SCID)	Bone marrow transplant < 3 months 94% survival	yes	yes
Adenosine Deaminase Deficiency	Bone marrow transplant < 3 months 94% survival	yes	yes

Interleukin 7 Receptor Alpha Deficiency (IL7-SCID)	Bone marrow transplant < 3 months 94% survival	yes	yes
Hereditary Fructose Intolerance	Fructose free diet		yes
X-Linked Adrenal Hypoplasia Congenita	Medication		yes
Familial Hemophagocytic Lymphohistiocytosis	Bone marrow transplant		yes
Citrullinaemia Type I	Diet and liver transplant		yes
LCHAD deficiency (Long-chain 3 hydroxyacyl-CoA dehydrogenase deficiency)	Dietary		yes
Systemic Primary Carnitine Deficiency	Vitamin supplementation		yes
MCAD deficiency (Medium-Chain Acyl-Coenzyme A Dehydrogenase Deficiency)	Dietary		yes
Conditions Causing Intellectual Disability			
Condition	Treatment		
X-Linked Adrenoleukodystrophy (X-ALD)	Stem cell transplant in boys with early stage cerebral disease improves survival and steroid replacement therapy can be lifesaving.	Yes	yes

Biotinidase Deficiency	Biotin can prevent complications	Yes	yes
Mucopolysaccharidosis type I (MPS I)	Bone marrow transplant can improve survival when done before complications arise	yes	yes
Arginase Deficiency	Medication and low protein diet	Yes	yes
Mucopolysaccharidosis type II (MPS II)	Enzyme replacement therapy effective for some types (not neurological features)	yes	yes
Krabbe Disease	Stem cell transplant improves lifespan and functional abilities	yes	yes
Galactosaemia Type I	Lactose free diet can prevent complications	Yes	yes
Glycine Encephalopathy (Non-Ketotic Hyperglycinemia)	Medication		yes
Acid Sphingomyelinase Deficiency (Niemann-Pick Disease Type A & B)	Supportive not curative treatment, avoiding contact sport that could cause trauma to enlarged liver/ spleen		yes
Arylsulfatase A deficiency (Metachromatic Leukodystrophy)	Bone marrow transplant may be effective if done before symptoms appear		yes

Hexosaminidase A Deficiency (Tay-Sachs disease)	Supportive not curative		yes
Hexosaminidase A and B Deficiency (Sandhoff Disease)	Supportive not curative		yes
Mucopolysaccharidosis type IIIA (Sanfilippo Syndrome type A)	Supportive not curative		yes
Mucopolysaccharidosis type IIIB (Sanfilippo Syndrome type B)	Supportive not curative		yes
GM1 gangliosidosis (beta-galactosidase-1 deficiency)	Experimental not curative		yes
Neuronal Ceroid Lipofuscinosis: CLN1, 2 & 3 (Batten Disease)	Supportive, avoid certain medications		yes
Dicarboxylic Aminoaciduria	Low protein diet, no evidence		yes
Phenylketonuria (PKU)	Dietary		yes
Methylmalonic Acidaemia with Homocystinuria	Diet and vitamin supplementation		yes
Methylmalonic Acidaemia	Diet and medication		yes

SCN1A- seizure disorders	Parental education & avoid specific anti epileptics, triggers & swimming alone etc		yes
Conditions Causing Heart Disease			
Condition	Treatment		
Familial Hypercholesterolaemia (FH)	Low fat diet, medication	Yes	yes
Fabry Disease	Enzyme replacement therapy (uncertain benefit for cardiac and renal and neurologic disease which occurs later)	yes	yes
Gaucher Disease	Enzyme replacement therapy effective for some subtypes (not neurological features)	yes	yes
Pompe Disease	Enzyme replacement therapy prolongs survival when started before 6 months	yes	yes
Autosomal Recessive Polycystic Kidney Disease	Kidney or liver transplant can prolong life		yes
Mucopolysaccharidosis type VI (Maroteaux Lamy Syndrome)	Enzyme replacement therapy can improve quality of life		yes
Cystinosis	Cystine-depleting agents		yes
Conditnios causing Liver or Lung disease			

Condition	Treatment		
Niemann-Pick Disease Type C1	Medication (Miglustat) may prevent developmental problems		yes
Type 1 Hereditary Haemochromatosis	Venesection, avoid alcohol		yes
Cystic Fibrosis	Medication, lung transplant		yes
Problems with Growth and Development			
Condition	Treatment		
Wilson Disease	Medication	Yes	yes
Hartnup Disease	High protein diet, avoidance of certain medications & sunlight	Yes	yes
Mucopolipidosis II (Inclusion Cell Disease)	Supportive not curative		yes
Mucopolipidosis III alpha/beta (pseudo-Hurler polydystrophy)	Supportive not curative		yes
Mucopolysaccharidosis type IVA (Morquio Syndrome Type A)	Enzyme replacement therapy can improve quality of life		yes

Mucopolysaccharidosis Type IVB (Morquio Syndrome type B)	Experimental not curative		yes
Holocarboxylase Synthetase Deficiency (Infantile Multiple Carboxylase Deficiency)	Vitamin supplementation		yes
Isovaleric Acidaemia	Diet and vitamin supplementation		yes
Glutaric Acidaemia Type I	Diet and vitamin supplementation		yes
Rare forms of Diabetes and Cataracts			
Condition	Treatment		
Galactosaemia Type II	Galactose free diet can prevent cataracts	Yes	yes
MODY Type 2 (Maturity Onset Diabetes in the Young Type 2)	Diet, oral hypoglycaemic agents		yes
Permanent Neonatal Diabetes Mellitus	Insulin		yes

Glucokinase-Congenital Hyperinsulinism
(GCK-CHI)

Diet, diazoxide drugs

yes