

Leukodystrophies	Lysosomal disorders	Haemoglobin disorders	Immunological	Neurological & muscular disorders	Vitamin, mineral & trace element disorders	Cholesterol disorders	Multisystem disorders	Carbohydrate disorders	Organic acid disorders	Amino Acid disorders	Fatty acid disorders	Remethylation Defects	Endocrine disorders
X-Linked Adrenoleukodystrophy	Fabry disease	Sickle cell disease	Severe combined immunodeficiencies	Spinal muscular atrophy	Biotinidase Deficiency	Familial Hypercholesterolaemia	Cystic fibrosis	Galactosaemia	3-hydroxy-3-methylglutaric aciduria	Argininosuccinic aciduria	Carnitine acylcarnitine translocase deficiency	MTHFR, MTR, MTRR deficiencies	X-Linked Adrenal Hypoplasia Congenita
Metachromatic Leukodystrophy	Cystinosis	Beta thalassaemia	T-Cell Related Lymphocyte Deficiencies	Duchenne muscular dystrophy	Wilson disease	Autosomal Recessive Hypercholesterolaemia	Hereditary Angioedema	Hereditary Fructose Intolerance	β-ketothiolase deficiency	Citrullinemia type I	Carnitine palmitoyltransferase I deficiency	Cbl D v1, Cbl G deficiencies	
3 other disorders	10 other disorders	*2 other disorders	Bare lymphocyte syndrome	2 other disorders	6 other disorders	2 other disorders			8 other disorders	8 other disorders	6 other disorders		