

Supplemental table 1. Major IMD groups associated with hypoglycaemia. Most common Glycogen storage diseases, Fatty acid oxidation disorders and congenital disorders of glycosylation are included.

Disorder	Gene(s)	Protein	Inheritance
GSD0a	<i>GYS2</i>	Liver glycogen synthase	Autosomal recessive
GSD0b	<i>GYS1</i>	Muscle glycogen synthase	Autosomal recessive
GSDIa	<i>G6PC</i>	Glucose 6-phosphatase α	Autosomal recessive
GSDIb	<i>SLC37A4</i>	Glucose 6-phosphate transporter	Autosomal recessive
GSDII (Pompe disease)	<i>GAA</i>	lysosomal α -glucosidase	Autosomal recessive
GSDIII	<i>AGL</i>	Glycogen debrancher enzyme	Autosomal recessive
GSDIV	<i>GBE1</i>	Glycogen brancher enzyme	Autosomal recessive
GSDV	<i>PYGM</i>	Muscle glycogen phosphorylase	Autosomal recessive
GSDVI	<i>PYGL</i>	Liver glycogen phosphorylase	Autosomal recessive
GSDVII	<i>PFKM</i>	Muscle phosphofructokinase	Autosomal recessive
GSDIXa	<i>PHKA2</i>	Glycogen phosphorylase kinase $\alpha 2$	X-linked recessive
GSDIXb	<i>PHKB</i>	Glycogen phosphorylase kinase β	Autosomal recessive
GSDIXc	<i>PHKG2</i>	Glycogen phosphorylase kinase γ	Autosomal recessive
GSDIXd	<i>PHKA1</i>	Glycogen phosphorylase kinase $\alpha 1$	X-linked recessive
GSDX	<i>PGAM2</i>	Muscle phosphoglycerate mutase	Autosomal recessive
GSDXI (Fanconi-Bickel)	<i>SLC2A2</i>	GLUT2	Autosomal recessive
GSDXII	<i>ALDOA</i>	Aldolase A	Autosomal recessive
GSDXIII	<i>ENO3</i>	β -enolase	Autosomal recessive
GSDXIV	<i>PGM1</i>	Phosphoglucomutase	Autosomal recessive
GSDXV	<i>GYG1</i>	Muscle glycogenin deficiency	Autosomal recessive
Hereditary fructose intolerance	<i>ALDOB</i>	Aldolase B	Autosomal recessive
Classical galactosemia	<i>GALT</i>	Galactose-1p uridyltransferase	Autosomal recessive
Fructose 1,6-Bisphosphatase deficiency	<i>FBP1</i>	Fructose 1,6-Bisphosphatase	Autosomal recessive
Pyruvate carboxylase deficiency	<i>PC</i>	Pyruvate carboxylase	Autosomal recessive
Methylmalonic acidemia	<i>MMUT, MMA A, MMAB, MMA DHC, MCEE</i>	Methylmalonil-CoA mutase, Methylmalonil-CoA epimerase	Autosomal recessive

Pediatric hypoglycemia diagnostic flowchart

Propionic acidemia	<i>PCCA, PCCB</i>	Propionyl-CoA carboxylase	Autosomal recessive
Isovaleric acidemia	<i>IVD</i>	Isovaleryl-Coa dehydrogenase	Autosomal recessive
Carnitine transporter deficiency	<i>SLC22A5</i>	Carnitine transporter	Autosomal recessive
Carnitine palmitoyltransferase I (CPT I) deficiency	<i>CPT1</i>	Carnitine palmitoyltransferase I	Autosomal recessive
Carnitine acylcarnitine translocase (CACT) deficiency	<i>SLC25A20</i>	Carnitine acylcarnitine translocase	Autosomal recessive
Carnitine palmitoyltransferase II (CPT II) deficiency	<i>CPT2</i>	Carnitine palmitoyltransferase II	Autosomal recessive
Very-long-chain acyl-CoA dehydrogenase (VLCAD) deficiency	<i>ACADVL</i>	Very-long-chain acyl-CoA dehydrogenase	Autosomal recessive
Long-chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) and mitochondrial trifunctional protein (MTP) deficiency	<i>HADHA, HADHB</i>	Long-chain 3-hydroxyacyl-CoA dehydrogenase, Long-chain enoyl-CoA hydratase, Long-chain ketoacylCoA thiolase	Autosomal recessive
Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency	<i>ACADM</i>	Medium-chain acyl-CoA dehydrogenase	Autosomal recessive
Short-chain acyl-CoA dehydrogenase (SCAD) deficiency	<i>ACADS</i>	Short-chain acyl-CoA dehydrogenase	Autosomal recessive
3-Hydroxyacyl-CoA dehydrogenase (SCHAD) deficiency	<i>HADH</i>	3-hydroxyacyl-CoA dehydrogenase	Autosomal recessive
Multiple acyl-CoA dehydrogenase (MAD) deficiency	<i>ETF A, ETF B, ETF D H</i>	Electron transfer flavoproteins	Autosomal recessive
Riboflavin metabolism defects	<i>SLC52A1, SLC52A2, SLC52A3, FLAD1, SLC25A32</i>	RFVT1, RFVT2, RFVT3, FAD synthase, FAD transporter	Autosomal recessive
3-Hydroxy-3-Methylglutaryl-CoA (HMG-CoA) Synthase Deficiency	<i>HMGCS2</i>	3-Hydroxy-3-Methylglutaryl-CoA synthase	Autosomal recessive
HMG-CoA Lyase Deficiency	<i>HMGCL2</i>	3-Hydroxy-3-Methylglutaryl-CoA lyase	Autosomal recessive
Succinyl-CoA:3-oxoacid CoA transferase (SCOT) deficiency	<i>OXCT1</i>	Succinyl-CoA:3-oxoacid CoA transferase	Autosomal recessive
Mitochondrial acetoacetyl-CoA thiolase (T2) deficiency	<i>ACAT1</i>	Acetoacetyl-CoA thiolase	Autosomal recessive
Monocarboxylate transporter 1 (MCT1) deficiency	<i>SLC16A1</i>	Monocarboxylate transporter 1	Autosomal recessive
OXPPOS disorders	<i>Multiple genes</i>	Respiratory complexes subunits	Variable
Phosphomannose isomerase (MPI-CDG) deficiency	<i>PMI</i>	Phosphomannose isomerase	Autosomal recessive
Phosphomannomutase 2 (PMM2-CDG) deficiency	<i>PMM2</i>	Phosphomannomutase 2	Autosomal recessive