Risk 351 - Inborn Errors of Metabolism

Risk description

An inborn error of metabolism has been diagnosed by a health care provider. This diagnosis can be self reported.

An inborn error of metabolism is a genetic condition that alters metabolism in the body. Examples include but are not limited to:

- Phenylketonuria (PKU)
- Maple Syrup Urine Disease (MSUD)
- Homocystinuria
- Tyrosinemia
- Galactosemia
- Glycogen Storage Disease
- Fructose Aldolase Deficiency
- Hyperlipoproteinuria
- Homocystinuria
- Medium and very long chain acyl-CoA dehydrogenase deficiency
- Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency
- Trifunctional protein deficiency
- Isovaleric acidemia
- Propionic Acidemia
- Beta-ketothiolase deficiency
- Fabry disease
- Gauchers disease
- Pompe disease
- Carnitine uptake effect
- Leber hereditary optic neuropathy
- Mitochondrial encephalomyopathy
- Lactic acidosis and stroke like episodes
- Mitochondrial neurogastrointestinal encephalopathy disease
- Myoclonic epilepsy
- Neuropathy or ataxia
- Retinitis pigmentosa
- Pyruvate carboxylase deficiency
- Zellweger Syndrome

◆ Glutaric Acidemia ◆ Multiple carboxylase deficiency ◆ Methylmalonic Acidemia	◆ Adrenoleukodystrophy ◆ Citrullinemia ◆ Argininosuccinic aciduria ◆ Carbamoyl phosphate synthetase 1 deficiency

Reason for risk

These inherited metabolic disorders are caused by a defect in the enzymes or co-factors that metabolize protein, carbohydrate or fat. Metabolic disorders affect nutrition status and often require special diets.

Category	All
Risk level	High
At risk if:	Health care provider diagnosed a metabolic disorder
Not at risk if:	Metabolic disorder has not been diagnosed by a health care provider
How is risk assigned?	Certifier selected from risk list in the data system.
Additional documentation	Document the specific type of metabolic disorder in the data system. Referral to the RD is required.