

INBORN ERROR OF METABOLISM

(351)

PARTICIPANT TYPE	ALL
HIGH RISK	YES

RISK DESCRIPTION:

Inherited metabolic disorders caused by a defect in the enzymes or their co-factors that metabolize protein, carbohydrate or fat. Generally refers to gene mutations or gene deletions that alter metabolism in the body, including but not limited to:

<p>Amino Acid Disorders</p> <ul style="list-style-type: none"> • Phenylketonuria (includes clinically significant hyperphenylalaninemia variants) • Maple syrup urine disease • Homocystinuria • Tyrosinemia 	<p>Urea Cycle Disorders</p> <ul style="list-style-type: none"> • Citrullinemia • Argininosuccinic aciduria • Carbamoyl phosphate synthetase I deficiency
<p>Organic Acid Metabolism Disorders</p> <ul style="list-style-type: none"> • Isovaleric academia • 3-Methylcrotonyl-CoA carboxylase deficiency • Glutaric acidemia type 1 • Glutaric acidemia type II • 3-hydroxy-3-methylglutaryl-coenzyme A lyase deficiency • Multiple carboxylase deficiency • Methylmalonic acidemia • Propionic acidemia • Beta-ketothiolase deficiency 	<p>Carbohydrate Disorders</p> <ul style="list-style-type: none"> • Galactosemia • Glycogen storage disease type I • Glycogen storage disease type II (see also Pompe disease) • Glycogen storage disease type III • Glycogen storage disease type IV (Andersen Disease) • Glycogen storage disease type V • Glycogen storage disease type VI • Hereditary Fructose Intolerance
<p>Fatty Acid Oxidation Disorders</p> <ul style="list-style-type: none"> • Medium-chain acyl-CoA dehydrogenase deficiency • Long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency • Trifunctional protein deficiency type I • Trifunctional protein deficiency type II • Carnitine uptake defect • Very long-chain acyl-CoA dehydrogenase deficiency 	<p>Mitochondrial Disorders</p> <ul style="list-style-type: none"> • Leber hereditary optic neuropathy • Mitochondrial encephalomyopathy, lactic acidosis, and stroke-like episodes • Mitochondrial neurogastrointestinal encephalopathy disease • Myoclonic epilepsy with ragged-red fibers • Neuropathy, ataxia, and retinitis pigmentosa • Pyruvate carboxylase deficiency
<p>Lysosomal Storage Diseases</p> <ul style="list-style-type: none"> • Fabry disease • Gauchers disease • Pompe disease 	<p>Peroxisomal Disorders</p> <ul style="list-style-type: none"> • Zellweger Syndrome Spectrum • Adrenoleukodystrophy (x-ALD)

RISK DESCRIPTION (CON'T):

Note: IEM not listed within this write-up may be found under: <http://rarediseases.info.nih.gov/GARD>. Please keep in mind these additional resources are not meant for medical advice nor to suggest treatment.

Presence of inborn error(s) of metabolism diagnosed by a physician as self-reported by applicant, participant, or caregiver; or as reported or documented by a physician, or someone working under physician's orders

Self-reporting of a diagnosis by a medical professional should not be confused with self-diagnosis, where a person simply claims to have or to have had a medical condition without any reference to professional diagnosis. A self-reported medical diagnosis should prompt the CPA to validate the presence of the condition by asking more pointed questions related to that diagnosis.

ASK ABOUT:

- Attitude and knowledge about condition and treatment plans including diet and medications
- Barriers to following treatment plan (e.g., health beliefs, religious or cultural practices, finances, access to follow-up health care)
- Growth pattern and weight goal
- Food-medication interactions
- Supplements including vitamins, minerals, herbal products and targeted nutrition therapy products
- Feeding difficulties and strategies for dealing with them

NUTRITION COUNSELING/EDUCATION TOPICS:

- Appropriate dietary management, which may include the use of special formulas and medical foods, can minimize the medical risk to individuals with inborn errors of metabolism.
- Identify the WIC foods that are consistent with the treatment plan. Review and provide WIC-approved medical foods or formulas as prescribed by the primary care provider.

POSSIBLE REFERRALS:

- If the participant requires in-depth nutritional intervention beyond the scope of WIC services, refer to primary care provider or a dietitian with expertise in this area of practice.
- If the participant is taking any non-prescribed vitamin or mineral supplements, herbal supplements, or targeted nutrition therapy products, advise discussing these with the primary care provider.
- If the participant does not have an ongoing source of health care, refer to primary care providers in the community or the local public health department.
- Refer infants and children to Children's Special Health Services program (<http://www.ndhealth.gov/cshs/>).
- Refer infants and children to the Right Track Program for early intervention services (<http://www.nd.gov/dhs/services/disabilities/earlyintervention/parent-info/right-track.html>).