



Metabolic Disorders

We specialize in the diagnosis and treatment of inborn errors of metabolism, including organic acidemias, urea cycle defects, fatty acid oxidation defects, glycogen storage disorders, lysosomal storage disorders and mitochondrial diseases among others. Many, but not all of these conditions can be detected through newborn screening programs.

A consultation is recommended when routine work-up does not lead to a clear diagnosis due to the fact that symptoms or signs of inherited metabolic disorders are frequently not specific.

Symptoms and Signs

- Failure to thrive
- Developmental delay
- Intermittent ataxia
- Intractable seizures
- Hypotonia/muscle weakness
- Chronic or cyclic vomiting
- Cardiomyopathy
- Cataracts/corneal opacities
- Family history of sudden infant death
- Hepatosplenomegaly (HSM)
- Progressive joint contractures/kyphosis
- Metabolic acidosis with high anion GAP
- Persistent ketosis
- Hypoglycemia
- Hyperammonemia
- MRI suggestive of Leigh's disease

Services offered include:

- Expert care with board-certified specialists in metabolic diseases
- Access to expert laboratory at CHOC Children's
- Team approach with a genetic counselor, case manager, metabolic dietitian, social worker and nurse practitioner

Orange

CHOC Clinic Building
1201 W. La Veta Ave.
Orange, CA 92868

Corona Health Center

854 Magnolia Ave.
Suite 101
Corona, CA 92879

**To refer patients, please call
888-770-2462 (1-888-770-CHOC)**

Fax: 855-246-2329 (1-855-CHOC-FAX)
www.choc.org/specialists