



State of CT
Genetics Newborn Screening Program
Health Professional Fact Sheet

Biotinidase Deficiency

Biotinidase deficiency is a metabolic disorder that limits the liberation and recycling of the vitamin biotin. Deficiency of the enzyme biotinidase results in improper functioning of many carboxylases essential to the production of fats, carbohydrates, and protein metabolism. Screening in Connecticut was instituted in 1993.

Genetic Basis

- Autosomal recessive condition - Recurrence risk is 25%
- On the basis of family studies to date heterozygotes (carriers) can be differentiated from affected and normal individuals with 90-95% accuracy

Ct Prevalence 1:73,387

Clinical Features Of Untreated Disease

Severity

- Physical disabilities: seizures, skin rash/skin infection, alopecia, hypotonia, hearing loss, conjunctivitis, ataxia, breathing problems
- Developmental disabilities: mental retardation, developmental delay
- Mortality: metabolic acidosis and organic acidemia can cause coma and death

Symptomatic diagnosis

A symptomatic diagnosis is difficult, as the age of onset of symptoms may be anywhere between one week to ten years. Usually though, symptoms appear between three to six months of age.

Variants

Partial biotinidase deficiency is a milder form where activity of biotinidase is about 10-30% of the enzyme's normal activity. Symptoms usually do not develop except under periods of stress from infection or poor diet.

Clinical Outcome With Screening And Treatment

All symptomatic children improve with treatment of oral pharmacological biotin. In some treated children all symptoms resolve, whereas with others deficits are irreversible.

Screening For Biotinidase Deficiency

Laboratory tests

- Test for levels of biotinidase activity on filter paper blood specimen
- A qualitative colorimetric assay is used with color change indicating presence of the enzyme.

Abnormal Test Results

Biotinidase level \leq 20% may indicate biotinidase deficiency or false positive results

- The primary care provider is notified by the DPH, Genetic NBS Tracking Unit nurse consultant.
- A referral is made by the DPH, Genetic NBS Tracking Unit nurse consultant to one of the Genetic Regional Treatment Centers.

- The primary care provider will be advised by the DPH, Genetic NBS Tracking Unit nurse consultant to contact the Genetic Regional Treatment Center to make a prompt referral and arrange for confirmation testing and evaluation.



Keeping Connecticut Healthy

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