Biotinidase Deficiency

Biotinidase deficiency is an autosomal recessive inherited disorder that results in the inability to process the vitamin biotin in the normal way. Early symptoms of biotinidase deficiency generally appear in infancy or early childhood and may include seizures, skin rash, hair loss, hypotonia, hearing loss, developmental delay and metabolic acidosis which can result in coma and death. The number of symptoms that a child develops and the severity of the disorder vary from child to child, even within the same family.

Prevalence: 1: 72,000 - 1: 126,000

Analyte Measured: Biotinidase enzyme

Reporting Ranges: Enzyme absent or reduced

Feeding Effect: None

Timing Effect: No effect

Confirmation: Repeat newborn screen. Colorimetric assay for biotinidase.

Treatment: Referral is made to a Metabolic Specialist. Treatment includes daily Biotin supplements.

Comments: With early diagnosis and treatment, all the symptoms of biotinidase deficiency can be prevented. Treatment with biotin supplements is relatively inexpensive and has been well tolerated to date. If left untreated biotinidase deficiency can cause coma leading to death. There is research pending to support the link with sudden infant death syndrome.