Condition Description

Biotinidase deficiency, also called late-onset multiple carboxylase deficiency, is a disorder of biotin recycling (distinct from holocarboxylase synthase deficiency or early onset MCD). It is inherited in an autosomal recessive pattern with an estimated incidence of 1:60,000. Individuals with profound biotinidase deficiency (less than 10% of normal activity in serum) may exhibit seizures, hypotonia, alopecia, skin rash, hearing loss, developmental delay, keto-lactic acidosis and organic aciduria, if untreated. The biotinidase enzyme releases biotin from biotinylated peptides and biocytin. Deficiencies in biotinidase will result in the decreased levels of free biotin, impairing the activity of multiple biotin-dependent enzymes. Early diagnosis of the condition and supplementation with biotin may alleviate or prevent progress symptoms.

Click here for the GeneReviews summary for this condition.

Indications

The biotinidase assay is used to rule out the disorder in clinically symptomatic patients and to confirm the abnormal biotinidase results from newborn screening.

Methodology

Colorimetric enzyme assay.

Detection

Diagnosis of biotinidase deficiency is made by measurement of biotinidase enzyme activity. Diagnosis of profound biotinidase deficiency by this method is very sensitive but carriers and partial biotinidase deficiency may not be detected.

Reference Range

5 - 10 nmol/min/ml.

Specimen Requirements

Additional Specimen Collection/Handling Instructions Required for this Test
A control sample is required for this test.

Serum from an unrelated individual must accompany the specimen as a control for sample integrity. For control sample, separate serum immediately and freeze immediately (preferably at -80C).

Type: Serum

Specimen Requirements:

In a no additive (red top) or SST tube: 3-5 ml
Separate serum immediately and freeze immediately (preferably at -80C).

Specimen Collection and Shipping: Ship frozen sample on dry ice with overnight delivery.

Special Instructions

See above for instructions regarding control samples.

Related Tests

Organic acid analysis (OA) and plasma acylcarnitine (AR) analysis can be used to confirm the diagnoses of biotinidase deficiency.