Metabolic Laboratory
Laboratories Wing
Sheba Medical Center
Tel HaShomer, 52621,
Israel
Tel 03-530-2553
Fax 03-5305225

המעבדה המטבולית
אגף המעבדות
מרכז רפואי ע"ש שיבא
תל השומר, 52621, ישראל
טל. 03-5302553
פקס 03-5302552

BIOTINIDASE ACTIVITY DETERMINATION IN SERUM

Useful For
Preferred test for diagnosing biotinidase deficiency

Follow-up testing for certain organic acidurias

Methodology:
Spectrophotometry (SP)

Clinical Information
Biotinidase deficiency is an autosomal recessive disorder caused by mutations in the biotinidase gene (*BTD*). Age of onset and clinical phenotype vary among individuals depending on the amount of residual biotinidase activity. Profound biotinidase deficiency occurs in approximately 1 in 137,000 live births and partial biotinidase deficiency occurs in approximately 1 in 110,000 live births. Untreated profound biotinidase deficiency typically manifests within the first decade of life as seizures, ataxia, developmental delay, hypotonia, sensorineural hearing loss, vision problems, skin rash, and alopecia. Partial biotinidase deficiency is associated with a milder clinical presentation, which may include cutaneous symptoms without neurologic involvement. Certain organic acidurias, such as holocarboxylase synthase deficiency, isolated carboxylase synthase deficiency, and 3-methylcrotonylglycinuria, present similarly to biotinidase deficiency. Serum biotinidase levels can help rule out these disorders.
Treatment with biotin is successful in preventing the clinical features associated with biotinidase deficiency. In symptomatic patients, treatment will reverse many of the clinical features except developmental delay, vision, and hearing complications. As a result, biotinidase deficiency is included in most newborn screening programs. This enables early identification and treatment of presymptomatic patients.

Specimen Volume: 1 mL serum
Collection Instructions: Spin down immediately and separate the serum. Freeze the serum and send it frozen to the lab along with clinical anamnesis.

Turnaround time 26 working days

Ministary of Health code 82261

Specimen Minimum Volume 0.5 mL

Metabolic Laboratory
Laboratories Wing
Sheba Medical Center
Tel HaShomer, 52621,
Israel
Tel 03-530-2553
Fax 03-5305225

המעבדה המטבולית
אגף המעבדות
מרכז רפואי ע"ש שיבא
תל השומר, 52621, ישראל
טל. 03-5302553
פקס 03-5302552

Specimen Stability Information

Frozen (preferred) 21days
Refrigerated 5 days

Interpretation

Normal 4.4 – 9.0 nmol/ml/min
Partial Deficiency 0.7 – 2.1 nmol/ml/min
Profound Deficiency < 0.7 nmol/ml/min
Obligate heterozygote 2.2 – 5.2 nmol/ml/min

Values below 3.5 U/L are occasionally seen in specimens from unaffected patients.
Confirmation by molecular testing is useful.

Clinical Reference

1. Wolf B. Disorders of biotin metabolism. In: Scriver CR, Beaudet AL, Sly WS,Valle D, editors. The metabolic and molecular bases of inherited disease. New York, NY: McGraw-Hill, 2001:3935–3962.
2. Schubiger G, Caflisch U, Baumgartner R, Suormala T, Bachmann C. Biotinidase deficiency: clinical course and biochemical findings. J Inherit Metab Dis 1984; 7:129 –130.
3. Grier RE, Heard GS, Watkins P, Wolf B. Low biotinidase activities in the sera of patients with impaired liver function: evidence that the liver is the source of serum biotinidase. Clin Chim Acta 1990; 186:397– 400
4. Hymes J, Wolf B. Biotinidase and its role in biotin metabolism. Clin Chim Acta 1996; 255:1–11

Dr. Elena Dumin MD, PhD

Director, Metabolic Laboratory

Sheba Medical Center,

Tel Hashomer, 5265601 Israel

Elena.Dumin@sheba.health.gov.il

Office Tel: 972-3-5304786 Cell: 972-54-5337851

Site: www.sheba.co.il