



# Serum Biotinidase (BTD)

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The information contained in this flyer is intended for healthcare professionals.

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## WHAT'S NEW

The Clinical laboratory is already performing a screening test for Biotinidase deficiency on dried blood spot specimens. Initiating quantification of serum biotinidase enzyme activity is a step forward for confirmation of Biotinidase deficiency.

## INTRODUCTION:

Biotinidase is responsible for the recycling of biotin (also known as Vitamin-H), by cleaving biocytin and biotinyl-peptides as a hydrolase, thereby liberating biotin for reutilization. Biotin is a cofactor required for the optimal activity of four biotin responsive carboxylase enzymes: propionic CoA Carboxylase and Methyl crotonyl CoA carboxylase involved in branched chain amino acid metabolism; acetyl CoA carboxylase involved in Krebs' cycle and pyruvate CoA Carboxylase involved in ketone metabolism.

Biotinidase deficiency is an autosomal recessive disorder caused by absent or markedly deficient activity of enzyme Biotinidase during the normal proteolytic turnover of holocarboxylase and other biotinylated protein, associated with secondary alterations in amino acid, carbohydrate, and fatty acid metabolism. The disorder is categorized into profound and partial Biotinidase deficiency. Patients with profound biotinidase deficiency have less than 10% of mean normal serum activity, while patients with the partial biotinidase deficiency variant have 10–30% of mean normal serum activity.

## INTENT OF USE:

This test is used for:

- Evaluation of patients with suspected biotinidase deficiency
- Patients with low enzyme activity on Dried blood spot Biotinidase testing

## IMPORTANT NOTE:

- Serum samples should be immediately frozen and sent to laboratory as early as possible.
- Serum samples may not be suitable for individuals who have undergone transfusion
- Moderate to gross hemolysis causes falsely elevated biotinidase values
- Lipemic samples must be ultracentrifuged before assaying
- Certain sulfa drugs may cause interference

## PRINCIPLE:

Colorimetry.  
Enzyme linked immunosorbent assay (ELISA)

## SPECIMEN TYPE:

5 cc clotted blood or 3-5 cc serum

## CHARGES:

Rs.5000.0

\*Revisions may apply

## SCHEDULE:

The test is performed on 2nd and 4th Monday every month and reported after two days.

For more information please call: 021 3486 1620  
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