Neonatal Biotinidase

What is biotin & why is it important ?

- Biotin is a **B vitamin** that is sometimes referred to as vitamin H or vitamin B7.
- It is one of the eight vitamins in the vitamin B-complex.
- The B vitamins, in general, help in promoting healthy nerves, skin, eyes, hair, liver and a healthy mouth.

Biotinidase test is universally adopted in new born screening programs of developed countries

Neonatal Biotinidase

- Biotinidase is an enzyme that is essential for the recycling of the vitamin biotin.
- Because the body needs free biotin to break down fats, proteins, and carbohydrates effectively, individuals with biotinidase deficiency are less able to process important nutrients.

What causes this disease ?

- Mutations in the gene for biotinidase, called BTD, produces deficient or defective enzyme.

Screening for Biotinidase Deficiency @ Metropolis

- Qualitative method for determination of biotinidase activity in dried blood spot specimen

- **Range :**

Normal : > 0.253 AV

Partial deficient : < 0.253 to 0.033 AV

Deficient : < 0.033 AV

*** AV : absorbance value of normal control*

Sample Report

Biotinidase neonatal

Test :	Biotinidase Activity
Specimen :	Neonatal Card
Results :	0.263
Normal Range :	Normal : > 0.253 AV (Absorbance value of normal control) Partial deficient : < 0.253 to > 0.033 AV Deficient : < 0.033 AV
Method:	Biorad Neonatal Biotinidase screening assay is a qualitative method for determination of biotinidase activity in dried blood spot specimen.
Note :	Partial deficient and deficient samples require re-testing and confirmation by serum biotinidase activity estimation.

Why screening for Biotinidase deficiency is important?

Individuals with biotinidase deficiency who are diagnosed before they have developed symptoms (e.g., by newborn screening or because a previous child in the family was diagnosed with the disorder) and who are treated with biotin appear to have normal development.