Neonatal Biotinidase

Early Life

Neonatal Biotinidase Test

Neurocutaneous Disorder in a Newborn Could be Biotinidase Deficiency

Symptoms • Hypotonia • Conjunctivitis • Seizures • Ataxia • Eczematous skin rash • Alopecia • Developmental delay • Vision problems, such as optic atrophy • Respiratory problems, such as hyperventilation, laryngeal stridor and apnea

Screening for biotinidase deficiency

- Meets the major criteria for inclusion in New Born Screening (NBS) programs
- Universally adopted in screening programs of developed countries

Who Should be Tested ?

- Children with Neurocutaneous symptoms
- At risk families with previous history of undiagnosed neonatal deaths, sudden infant deaths or acute metabolic encephalopathy

Screening for Biotinidase Deficiency @ Metropolis

- Qualitative method for determination of biotinidase activity in dried blood spot specimen
- Partial deficient and deficient samples require re-testing and confirmation by serum by serum biotinidase activity estimation
- Normal Range:

Normal	: > 0.253 AV
Partial deficient	: < 0.253 to 0.033 AV
Deficient	: < 0.033 AV

(AV : absorbance value of normal control)

Other Neonatal Screen Panels @ Metropolis

1. Neonatal screen - I	2. Neonatal screen - II	3. Neonatal screen - III	4. Neonatal screen - Iv
TSH, G6PD, Phenyl Alanine	TSH, G6PD and 17OH progesterone Phenyl Alanine	TSH, G6PD and 17OH progesterone Phenyl Alanine, Cystic Fibrosis, Galactosemia, Biotinidase	Comprehensive panel covers 40 plus parameters

Barry W. Biotinidase Deficiency: "If you have to have an inherited metabolic disease, this is the one to have" Genet Med 2012: 14(6): 565-575

Neo

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