

Biotinidase Deficiency (Partial)

Clinic Visits:

- At time of diagnosis with follow-up at 1 year
- Every three years thereafter

Laboratory studies:

- Biotinidase enzyme assay or DNA analysis following positive newborn screen
- Urine organic acids or acylcarnitine profile prior to start of biotin
- Urine organic acids or acylcarnitine profile at clinic visits (every 3 years)

Other Evaluations:

- Yearly developmental questionnaires to be completed by parents
- Developmental screening at 3 and 6 years old

Topics:

- Biochemistry and genetics of biotinidase deficiency
- Treatment
 - Biotin 5 -10 mg

Please make sure your child has clinic visits and levels done on time. Call the metabolic clinic at _____ to make an appointment, schedule a blood draw, or with any other questions.

Thank You,
Your Metabolic Team

