Biotinidase Deficiency
(Profound)

Clinic Visits:

- ☑ 0-12 months – every 6 months
- ☑ > 1 year- once a year

Laboratory studies:

- ☑ Biotinidase enzyme assay or DNA analysis following abnormal newborn screen
- ☑ Urine organic acids or acylcarnitine profile prior to start of biotin
- ☑ Yearly urine organic acids or acylcarnitine profile

Other Evaluations:

- ☑ Audiology evaluation after diagnosis with follow-up every 3 years
- ☑ Ophthalmology evaluation during first year of life and at 6 years old
- ☑ Yearly developmental questionnaires to be completed by parents
- ☑ Developmental evaluation at 3 and 6 years old
- ☑ Neuropsychological evaluation at 9 years old

Topics:

- ☑ Biochemistry and genetics of biotinidase deficiency
- ☑ Treatment
    - o Biotin 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