Partial Biotinidase Deficiency Care Plan
(last updated 2/21/09)

Clinical Considerations
- Compliance

Initial labs (diagnostic & baseline)
- Biotinidase enzyme assay
- Consider mutational analysis
- Consider UOA /ACP before initiation of biotin treatment

Treatment Considerations
- Biotin 5-10 mg (free/D-biotin form)

Monitoring labs
- Consider UOA or ACP at clinic visit

Frequency of metabolic visits

Age	Frequency
0-12 months	Every year
>1 year	Every 3 years

Clinic Visit labs
- Consider UOA or ACP yearly

Other evaluations
- Yearly developmental questions (to be completed by parents)
- Developmental screening at 3 & 6 yrs

Yearly labs
- None

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Performance Indicators	Outcome Indicators

1. Age of biotin initiation
2. Dosage of biotin
3. Initial lab studies
  a. Biotinidase activity
  b. Molecular results
4. Monitoring lab studies
  a. ACP or UOA results
5. Frequency of clinic contact and visits (track compliance with visits)
6. Growth parameters (ht, wt, OFC, BMI)
7. Developmental services (PT, OT, & speech)
8. School performance
  a. Grade appropriate (Y/N)
  b. Special services (Y/N)
  c. IEP (Y/N)
9. Genetic Counseling (Y/N)
	1. Development (IQ)
	2. Presence of hearing loss
	3. History and/or presence of ophthalmologic problems
	4. History and/or presence of seizures
	5. History and/or presence of rashes
	6. Growth
	  a. Final adult parameters

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 mutational 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 age 3 years

Topics:

- ☑ Biochemistry and genetics of Biotinidase Deficiency
- ☑ Treatment
    - o Biotin 5 -10 mg

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

Thank You,
Your Metabolic Team