Mountain States Genetics Regional Collaborative Center

Tyrosinemia Type I Care Plan
(last updated 2/21/09)

Clinical Considerations

- Transients
  - May be shortened with vitamin C
- Will be missed on NBS
- Liver carcinoma risk (even on diet)

Initial labs (diagnostic & baseline)

- Quant plasma amino acids, urine amino acids, and urine succinylacetone
- Comp metabolic panel
- Liver- Serum alpha-fetoprotein, PT, PT, vitamin K dependent coagulation factors.
- Abdominal/renal ultrasound
- X-ray of wrist (>3 months)
- Fumarylacetoacetate hydrolyase enzyme assay OR Common mutation (Fr CAN)/ sequencing

Diet considerations/ treatment

- Restriction of Tyr and Phe (Met restriction only if elevated)
- NTBC
- Liver transplant
- Avoid fasting (prevent hepatic decompensations and neuro crises)

Monitoring

- Quant plasma AA or quant Phe/Tyr, AFP, urine succinylacetone
- Targeted Tyr level on NTBC <600µmol/L

Age	Frequency
0-6 months	Every month
6-12 months	Every 2 months
> 1 year	Every 3 months

Frequency of visits

Age	clinic visits
0-6 months	Every 2 months
6-12 months	Every 3 months
1-18 years	Every 6 months
>18 years	Every year

***Post transplant-clinic visit every 6-12 months to check renal function

Clinic visit labs

- See above

Emergency management

- IV dextrose to prevent hepatic decomp and neuro crises
- It is necessary to provide symptomatic treatment for hypertension, hyponatremia, hypokalemia, and hypophosphatemia

Labs to obtain during illness

- None

Other evaluations

- Ophthalmology- yearly
- Refer to Liver or Renal if clinically warranted.
- Yearly developmental questionnaires (to be completed by parents).
- Developmental eval @ 3 & 6 y
- Neuropsych at 9 y
- Abdominal ultrasound every 6 months with alternating CT scan of abdomen every 6 months
- Metabolic dietitian eval (at least yearly)

Yearly labs

- Comp metabolic panel
- Prealbumin
- Ferritin/transferritin
- Liver- Serum alpha-fetoprotein, PT, PT, and vitamin K dependent coagulation factors

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Performance Measures

1. Age of diagnosis and initiation of diet
2. Frequency of clinic visits and compliance with visits
3. Initial lab studies
    a. Phe, Tyr, and Met levels
    b. Frequency of levels
4. Monitoring lab studies
    a. Phe, Tyr, and Met levels OR
    b. Quant plasma amino acids
    c. Urine succinylacetone
    d. Liver (LFTs, serum alpha-fetoprotein, PT/PTT, and vit K dependent coag factors)
    e. Nutrition labs
5. NTBC
    a. Dosage
    b. Age of initiation
6. Liver ultrasound (track freq. and findings)
7. Ophthalmology evaluation
8. Neurospycholgical evaluation
9. Growth parameters (ht, wt, OFC, BMI)
10. Diet
    a. Frequency of Metabolic dietitian visits
    b. Frequency of dietary analysis (3 day diet records)
    c. Natural protein intake (tolerance)
    d. Formula (Y/N)
    e. Medical foods (Y/N)
    f. Mode
11. Transplant (Y/ N)
12. Neuropsychological evaluation results
13. Developmental services (PT, OT, & speech)
14. Genetic Counseling (Y/N)
15. School performance
    a. Grade appropriate (Y/N)
    b. Special services (Y/N)
    c. IEP (Y/N)
16. Genetic Counseling (Y/N)

Outcome measures

1. History and/or presence of liver failure, rickets, and hypertension
2. History and/or presence of liver carcinoma and adenomas
    a. Age of diagnosis
3. History and/or presence of corneal erosions/crystals and photophobia
4. Outcome of liver transplants
5. History of neurological crises including episodic behavioral issues and peripheral nerve deficiencies
6. Growth
    a. Final adult parameters
7. Development
    a. IQ
    b. Level of functioning
    c. Decline in IQ or level of function

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