Mountain States Genetics
Regional Collaborative Center

Galactosemia Care Plan
(last updated 2/21/09)

Clinical Considerations

- Cataracts
- Verbal dyspraxia
- Osteopenia
- Duartes
- Ovarian failure
- Ataxia, tremors

Initial labs (diagnostic & baseline)

- Gal-1-Phosphate
- Galactose-1-phosphate uridyl transferase enzyme
- Electrophoresis or mutation analysis
- Bilirubin (direct/total), transaminases, PT

Diet Considerations

- Galactose/lactose restricted
- Calcium supplementation
  - With Vit D

Monitoring labs

- Gal-1-P levels
- Consider urinary galactitol

Age	Frequency
0-6 years	every 6 months
>6 years	every year

Frequency of metabolic visits

Age	Frequency
0-6 years	every 6 months
>6 years	yearly

Clinic Visit labs

- Gal-1-P levels (see above)
- Consider urinary galactitol

Other evaluations

- Ophthalmology-time of diagnosis
  - If cataracts, recheck 6 mo
  - If noncompliant, repeat
  - Not warranted if compliant
- Speech eval (2, 5, 9 years)
- Spine DEXA (9 & 18 yrs then every 5 yrs)
- Yearly developmental questions (to be completed by parents)
- Developmental eval (3 and 6 yrs)
- Neuropsych (9 & 18 yrs)
- Females –endocrinology or GYN if labs abnl or at time of pubertal development (physical exam) of if there is a lack of pubertal development by age 14 years
- Metabolic dietitian eval (at least yearly)

Yearly labs

- Females: follicle stimulating hormone, luteinizing hormone, and estradiol (yearly starting at age 9yrs)
- Consider progesterone (yearly starting at age 9yrs)
- Consider calcium, phosphate, vitamin D and 25-OH vitamin D

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Performance Indicators

1. Age of diet initiation
2. Age at which Gal-1-P within treatment range (metabolic control)
3. Initial lab studies
    a. Gal-1-P levels
    b. Galactitol levels
    c. Molecular results
4. Monitoring lab studies
    a. Gal-1-P levels
    b. Galactitol levels
    c. Other labs: calcium, phosphate, vitamin D and 25-OH vitamin D
    d. Females: follicle stimulating hormone, luteinizing hormone, estradiol, and progesterone
5. Frequency of clinic contacts and visits (track compliance with visits)
6. Growth parameters (ht, wt, OFC, BMI)
7. Spontaneous puberty (Y/N)
8. Reported calcium and Vitamin D intake
9. DEXA results and number of fractures
10. Diet
    a. Frequency of Dietitian visits (phone and clinic visits)
    b. Frequency of dietary analysis (3 day diet records)
11. Neuropsychological evaluation results
12. Developmental services (PT, OT, & speech)
13. School performance
    a. Grade appropriate (Y/N)
    b. Special services (Y/N)
    c. IEP (Y/N)
14. Genetic Counseling (Y/N)

Outcome Indicators

1. History and/or presence of cataracts (age of diagnosis)
2. History and/or presence of verbal dyspraxia or other speech problems
3. History and/or presence of osteopenia and fractures
4. History and/or presence of neurological presentation (tremors, ataxia)
5. Development of premature ovarian failure
6. Development
    a. IQ
    b. Level of functioning
    c. Decline in IQ or level of function as an adult
7. Growth
    a. Final adult parameters

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