Mountain States Genetics
Regional Collaborative Center

Isovaleric Acidemia Care Plan
(last updated 2/21/09)

Clinical Considerations

- Stabilize neonate
- Thrombocytopenia, neutropenia,and pancytopenia
- Pancreatitis
- Mild variant
- Hypocalciumemia in acutely ill patients

Initial labs (diagnostic & baseline)

- UOA and SAA
- ACP
- Consider urine acylglycines
- Comp metabolic panel, ammonia, CBC, and urine ketones
- Consider mutational analysis and/or Isovaleryl-CoA dehydrogenase enzyme assay

Diet considerations/ treatment

- Leu-restricted diet
- Essential AA formula for IVA
- Avoid fasting
- Carnitine- 50-100 mg/kg/day
- Glycine- 250 mg/kg/day

Monitoring

- Quant plasma AA for acute form
- Monitoring for milder form not needed

Age	Frequency
0-6 months	Every 2 weeks
6-12 months	Monthly
1-6 years	Every 3 months
6-18 years	Every 6 months
>18 years	Yearly

Frequency of visits

Acute form

Age	Frequency
0-6 m	Every 2 months
6-12 m	Every 3 months
>12 m	Every 6 months

Milder form

Age	Frequency
0-4 y	Every 6 months
4-18y	Every year
>18y	Every 3 years

Clinic visit labs

- See above

Emergency management

- Immediate IV 10% dextrose + IV lipids
- Add IV carnitine 100 mg/kg/day
- Oral glycine if tolerated

Labs to obtain during illness

- Comp metabolic panel and phosphate
- Urinalysis for ketones
- CBC
- Amylase and lipase
- Consider ammonia
- Consider SAA and UOA

Other evaluations

- Referral to neurology if clinically warranted. Consider brain MRI.
- Bone health
  - DEXA-spine @ 9 & 18y
- Yearly developmental questionnaires (to be completed by parents)
- Developmental eval @ 3 & 6 y
- Neuropsych @ 9 y
- Metabolic dietitian (at least yearly)

Yearly labs

- Comp metabolic panel and phosphate
- Amylase and lipase
- Carnitine
- Consider UOA
- Prealbumin / albumin
- Plasma Ferritin, transferrin, or iron studies
- Consider CBC, hemoglobin, and hematocrit
- Folate and Vitamin B12
  - If noncompliant with formula
  - Consider urine MMA
- Consider other nutritional testing depending on formula (Zinc, selenium, vit D, essential fatty acids, and lipid profile).
- *** for milder form, carnitine only

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Performance Measures

1. Age of diagnosis (both positive NBS and confirmatory ACP and UOA)
2. Presence of illness at time of diagnosis including poor feedings, lethargy, vomiting, metabolic acidosis, ammonia, ketonuria, lactic acidemia, hypothermia, hepatomegaly, raised intracranial pressure, use of dialysis (type), or other neurological feature and a foul "sweaty feet" odor.
3. Type of IVA (acute presentation, milder presentation, asymptomatic, FTT)
4. Frequency of clinic contacts and visits (track compliance with visits)
5. Growth parameters (ht, wt, OFC,BMI)
6. Initial lab studies
    a. NBS results
    b. Acylcarnitine profile
    c. Urine organic acids
    d. Quant plasma amino aicds
    e. Urine acylglycines
    f. CMP
    g. Ammonia
    h. CBC
    i. Urine ketones
    j. Molecular studies
    k. Enzymatic studies
7. Monitoring lab studies
    a. Quant plasma amino acids
    b. CMP
    c. Amylase and Lipase
    d. Serum carnitine
    e. Nutrition labs
8. Total decompensations and hospitalizations
    a. Track ICU admissions
    b. # of days for hospitalizations
    c. # of ER visits
    d. Track labs including CMP, phosphate, CBC ketones, amylase/lipase, ammonia, SAA, & UOA
9. Number of pancreatitis episodes
10. Dosage of carnitine and glycine
11. Diet
    a. Frequency of Metabolic dietitian visits
    b. Frequency of dietary analysis (3 day diet records)
    c. Natural protein intake (tolerance)
    d. Formula (Y or N)
    e. Medical foods (Y or N)
    f. Mode (oral, G-tube, bolus/drip, meds only/meds and diet)
12. DEXA results and number of fractures
13. Neuropsychology evaluation results
14. Developmental services (PT, OT, & speech)
15. School Performance
    a. Grade appropriate (Y/N)
    b. IEP (Y/N)
    c. Special services (Y/N)
17. Genetic Counseling (Y/N)

Outcome measures

1. Mortality
2. Development
    a. IQ
    b. Level of functioning
3. History and/or presence of hematologic findings
4. History and/or presence of neurological symptoms and abnormal MRI findings
5. History and/or presence of pancreatitis
6. History and/or presence of osteopenia
7. Growth
    a. Final adult parameters

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