Malonic Acidemia
(updated 7/13/10)

Clinical Considerations

- Developmental delay
- Cardiomyopathy
- Seizures
- Short stature

Initial labs (diagnostic & baseline)

- ACP
- UOA
- Comp metabolic panel
- Serum CK
- Enzymatic studies of malonyl-CoA decarboxylase and/or molecular analysis of MLYCD gene

Diet considerations/ treatment

- Limited long chain fat diet with MCT oil supplementation (~30% total calories from fat with 50% MCT)
- Avoidance of fasting
- Carnitine

Monitoring

- Consider ACP / UOA

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

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

Clinic visit labs

- Carnitine
- CK
- ACP and UOA

Emergency management

- Immediate IV 10% dextrose
- IV carnitine, begin at 100 mg/kg/day
- Consider cardiac monitoring

Labs to obtain during illness

- Comp metabolic panel
- BNP
- CK

Other evaluations

- Cardiology (echo and EKG) at time of dx and consider yearly thereafter
- Early intervention
- Yearly developmental questionnaires (to be completed by parents)
- Developmental eval @ 3 & 6 yrs
- Neuropsych @ 9 yrs
- Psychiatric screening @ 18 yrs
- Metabolic dietitian (at least yearly)

Yearly labs

- Comp metabolic panel
- ACP
- Essential fatty acid profile @ 6 m then yearly thereafter
- BNP
- Consider nutritional labs

Malonic Acidemia
(updated 7/13/10)

Performance Measures

1. Age of diagnosis and diet initiation
2. Presence of illness at time of diagnosis including metabolic acidosis, hyperammonemia, hypoglycemia, cardiomyopathy, and seizures.
3. Initial lab studies
    a. Acylcarnitine profile /UOA
    b. Other abnormal laboratory findings (if obtained) CMP, ammonia, and BNP
    c. Carnitine
    d. Genotype
    e. Enzymatic studies
4. Frequency of clinic contacts and visits (track compliance with visits)
5. Growth parameters (ht, wt, OFC, BMI)
6. Total decompensations and hospitalizations (including infections)
    a. # of days for hospitalizations
    b. # of ER visits
    c. Track labs including CMP, ammonia, and BNP
7. Monitoring lab studies
    a. Plasma acylcarnitine profile
    b. Carnitine
    c. Fatty acid profile
    d. Comp metabolic panel
    e. Nutrition labs
8. Carnitine supplementation (Y/N, dosage)
9. Diet
    a. Frequency of Dietitian visits
        i. Frequency of dietary analysis (3 day diet records)
    b. Percent of MCT and LCFA in diet.
        i. Including if essential fatty acids are added
    c. Formula (Y/N)
    d. Medical foods (Y/N)
    e. Mode (oral, G-tube, bolus/drip, meds only/meds and diet)
10. Neuropsychological evaluation results
11. Developmental services (PT, OT, & speech)
12. School Performance
    a. Grade appropriate (Y/N)
    b. IEP (Y/N)
    c. Special services (Y/N)
13. Genetic Counseling (Y/N)

Outcome measures

1. Mortality
2. History and/or presence of cardiomyopathy
3. History and/or presence of neurological symptoms
4. History and/or presence of muscular involvement
5. Development
    a. IQ
    b. Level of functioning
6. Growth
    a. Final adult parameters