Maternal Carnitine Deficiency Care Plan
(Reviewed 7/13/10)

Clinical Considerations • Symptomatic heterozygotes • Cardiac arrhythmias • Follow patient if free carnitine is <15 umol/L	**Initial labs (diagnostic & baseline)** • Carnitine panel • Maternal carnitine panel • Consider maternal urine organic acids • Consider maternal acylcarnitine profile following maternal carnitine supplementation • Consider maternal OCTN2 sequencing or enzymatic studies/functional studies (20% of patients have 0 or only 1 mutation)
Treatment Considerations • Mother: carnitine supplementation • Baby: carnitine supplementation for 2-4 weeks. If levels are high, discontinue and recheck	**Monitoring labs** • Plasma carnitine panel yearly
Frequency of metabolic visits • At time of diagnosis then every 3 year thereafter	**Clinic Visit labs** • Carnitine panel
Other evaluations • No specialty visits for baby • Maternal cardiac evaluation to include holter monitor, EKG, and echo	**Yearly labs** • Carnitine panel

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Performance Indicators	Outcome Indicators
1. Age of diagnosis and initiation of carnitine 2. Initial labs a. Infant's NBS b. Infant's initial carnitine panel c. Maternal carnitine panel a. Maternal genotype b. Maternal enzymatic studies 3. Maternal cardiac status at time of diagnosis and follow-up	1. History and/or presence of maternal cardiomyopathy and cardiac arrhythmia 2. History and/or presence of maternal rhabdomyolysis and myoglobinuria 3. History and/or presence of maternal skeletal muscle weakness 4. History and/or presence of maternal liver dysfunction