What is the differential diagnosis?
Argininemia (arginase deficiency, hyperargininemia)

What are the characteristics of argininemia?
- Disorders of arginine metabolism are included in a larger group of disorders, known as urea cycle disorders.
- Argininemia is an autosomal recessive inborn error of metabolism caused by a defect in the final step in the urea cycle.
- Most infants are born to parents who are both unknowingly asymptomatic carriers and have NO known history of a urea cycle disorder in their family.
- The incidence of all urea cycle disorders is estimated to be about 1:8,000 live births. The true incidence of argininemia is not known, but has been estimated between 1:350,000 and 1:1,000,000.
- Argininemia is usually asymptomatic in the neonatal period, although it can present with mild to moderate hyperammonemia. Untreated, argininemia usually progresses to severe spasticity, loss of ambulation, severe cognitive and intellectual disabilities and seizures.
- Lifelong treatment includes a special diet, and special care during times of illness or stress.

What is the screening methodology for argininemia?
1. An amino acid profile by Tandem Mass Spectrometry (MS/MS) is performed on each filter paper.
2. Arginine is the primary analyte.

What is an in-range (normal) screen result for arginine?
Arginine less than 100 μmol/L is NOT consistent with argininemia.
See Table 1.

What is an out-of-range (abnormal) screen for arginine?
Arginine ≥ 100 μmol/L requires further testing.

What screen results will require a repeat filter paper?
Arginine 100 – 199 μmol/L requires a repeat filter paper. Consultation with a Metabolic Specialist will be left to provider discretion.

What screen results will require diagnostic testing?
Arginine ≥ 200 μmol/L will require immediate action. The follow-up program will provide detailed guidance on required actions and a Follow-Up Management Protocol will be provided.

What are the follow-up needs?
The follow-up program will provide detailed guidance on needed actions. The following metabolic specialists have approved all recommendations:

Metabolic Specialist in Oklahoma
Klaas Wierenga, M.D. (405) 271-8685  Michelle Polan, M.D. (405) 271-8685
Susan Palmer, M.D. (405) 271-8685

What is my role in screening?
If you are listed as the infant’s planned health care provider on the filter paper requisition, you are required by the Newborn Screening Program Regulations to initiate follow-up activities.