

Newborn Screening ACT Sheet [Increased Arginine] Amino Aciduria/Urea Cycle Disorder

Differential Diagnosis: Argininemia

Condition Description: The urea cycle is the enzyme cycle whereby ammonia is converted to urea. In argininemia, defects in **arginase**, a urea cycle enzyme, may result in hyperammonemia.

YOU SHOULD TAKE THE FOLLOWING ACTIONS:

- Contact family to inform them of the newborn screening result and ascertain clinical status (poor feeding, vomiting, lethargy, tachypnea).
- Immediate consult with pediatric metabolic specialists.
- Evaluate the newborn (poor feeding, lethargy, hypotonia, tachypnea, seizures, and signs of liver disease). If any sign is present or infant is ill, immediately initiate emergency treatment for hyperammonemia in consultation with metabolic specialist.
- Transport to hospital for further treatment in consultation with metabolic specialist.
- Initiate timely confirmatory/diagnostic testing and management, as recommended by specialist.
- Provide family with basic information about hyperammonemia.
- Report findings to newborn screening program.

Diagnostic Evaluation: Specific diagnosis is made by plasma amino acid analysis revealing increased arginine and urine orotic acid analysis revealing increased orotic acid, respectively. Blood ammonia determination may also reveal hyperammonemia.

Clinical Considerations: Argininemia is usually asymptomatic in the neonate although it can present with a mild – moderate hyperammonemia once the baby receives dietary protein. Later signs include mental retardation, seizures and spastic diplegia if untreated. Rarely, argininemia may cause severe neonatal illness as seen in the other urea cycle disorders.

Additional Information:

[Emergency Protocols \(New England Consortium of Metabolic Programs\)](#)

[Gene Reviews](#)

[Genetics Home Reference](#)

Referral (local, state, regional and national):

[Testing](#)

[Clinical Services](#)

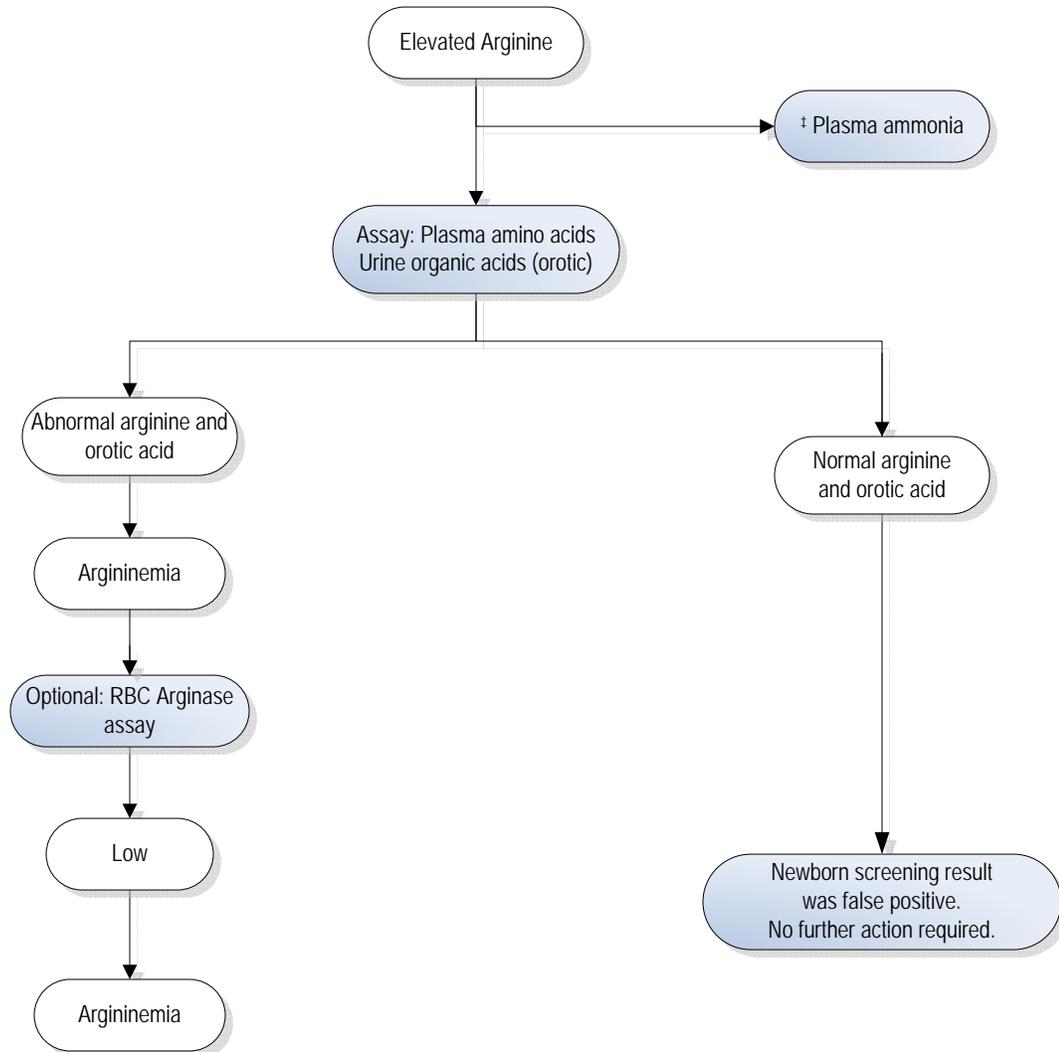
[Find Genetic Services](#)

Disclaimer: This guideline is designed primarily as an educational resource for clinicians to help them provide quality medical care. It should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed to obtaining the same results. Adherence to this guideline does not necessarily ensure a successful medical outcome. In determining the propriety of any specific procedure or test, the clinician should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. Clinicians are encouraged to document the reasons for the use of a particular procedure or test, whether or not it is in conformance with this guideline. Clinicians also are advised to take notice of the date this guideline was adopted, and to consider other medical and scientific information that become available after that date.





Arginine Elevated



Actions are shown in shaded boxes; results are in the unshaded boxes.

Abbreviations/Key

RBC = Red blood cell

‡ = When the positive predictive values of screening are sufficiently high and the risk to the baby is high, some initiate diagnostic studies that are available locally at the same time as confirmation of the screening result is done.

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