Citrulline Elevated

Elevated Citrulline

Assay*: Plasma amino acids Urine amino acids

Plasma ammonia

Citroline elevated

Increased citrulline

Increased citrulline Argininosuccinic acid +

Increased citrulline, lysine, alanine, proline.

Normal

Assay pyruvate carboxylase

Newborn screening result was false positive. No further action required.

Citrullinemia I

Citrullinemia II (Citrin deficiency)

Argininosuccinic aciduria

Pyruvate carboxylase deficiency

Affiliations/Key

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

* = Urine organic acids (orotic) may be informative.

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.