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**GUIDELINES FOR THE CLINICAL MANAGEMENT OF A POSITIVE NEWBORN SCREEN FOR GLUTARIC ACIDURIA TYPE 1 (GA1)**

Glutaric Aciduria Type 1 (GA1) is a rare genetic metabolic disorder caused by a deficiency of the enzyme Glutaryl-CoA dehydrogenase, in which the body cannot metabolise natural protein properly and this leads to an accumulation of glutaric acid. Affected infants and children may present with neurological symptoms or encephalopathy. Hypotonia and macrocephaly may be found in young infants. If untreated, GA1 may lead to severe and chronic neurological symptoms.

**ACTION UPON RECEIPT OF POSITIVE NEWBORN SCREENING RESULT FOR GA1:**

Local Paediatric/Neonatal Unit:

* Examine the baby
* Measure weight, length and head circumference
* Review feeding regime and continue oral feeds
* Take the following samples and send to the metabolic lab, Temple Street Children’s University Hospital:
	+ Acyl-Carnitine profile (dried blood spot card)
	+ Plasma amino acids (Lithium Heparin sample)
	+ Urine for organic acids
* Some babies may need treatment with Carnitine (L-Carnitine 100mg TDS orally) at this point. This will be advised by the Director of Newborn Screening.
* Consider cranial ultrasound examination

**IF BABY IS UNWELL (OR BECOMES UNWELL PRIOR TO CONFIRMATION OF DIAGNOSIS):**
**Discuss immediately with Metabolic Team and Neonatal Team**

* Start IV Dextrose 10% + electrolytes at 150mls/kg/day
* Perform investigations as above + other tests as clinically indicated

**IF BABY IS WELL:**

**Discuss with and refer to metabolic team for follow up upon confirmation of diagnosis of GA1**

**CONTACT NUMBERS:**

Newborn Screening Lab: 00 353 1 8784277

Temple Street Hospital: 00 353 1 8784200

**OTHER RESOURCES**: National Newborn Bloodspot Screening Programme on [www.hse.ie](http://www.hse.ie) National Centre for Inherited Metabolic Disorders [www.metabolic.ie](http://www.metabolic.ie)

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