Newborn Screen Follow-up for Guanidinoacetate Methyltransferase Deficiency (GAMT)

Elevated guanidinoacetate

- CRDPU / Creatine Disorders Panel, Urine
- Guanidinoacetate, creatine, creatinine in plasma

Elevated guanidinoacetate in urine and plasma

- GAMT deficiency

Refer to Genetics Specialist

optional

GAMT gene sequencing**

Normal guanidinoacetate

- Not GAMT deficiency*

* If clinical suspicion is high, consider brain MR spectroscopy or GAMT enzyme activity
** GAMT deletion/duplication testing should be considered if sequencing is not informative