Newborn Screening Follow-up for Isolated C4 Acylcarnitine Elevations
(also applies to any plasma or serum C4 acylcarnitine elevation)*

Isolated elevation of C4 acylcarnitine

Perform:
- C4U / C4 Acylcarnitines, Quantitative, Urine
- OAU / Organic Acids Screen, Urine
- ACYLG / Acylglycines, Quantitative, Urine
AND 1 of the following:
- ACRN / Acylcarnitines, Quantitative, Plasma
- ACRNS / Acylcarnitines, Quantitative, Serum

**Routine labs performed locally: Glucose, electrolytes, blood gases, ammonia**

- **Plasma or serum C4–Normal**
  - **Urine C4–Normal**
  - **Urine isobutyrylglycine–Normal**

  **Diagnostic for isobutyryl-CoA dehydrogenase (IBDH) deficiency**

  Consider confirmatory testing for ACAD8 (IBDH) gene

- **Plasma or serum C4–High**
  - **Urine C4–High**
  - **Urine isobutyrylglycine–High or Normal**

  **Diagnostic for isobutyryl-CoA dehydrogenase (IBDH) deficiency**

  Consider confirmatory testing for ACAD8 (IBDH) gene

- **Plasma or serum C4–High or Normal**
  - **Urine C4–High or Normal**
  - **Urine ethylmalonic acid–High**

  **Diagnostic for short chain acyl-CoA dehydrogenase (SCAD) deficiency**

  Call for clinical consultation to determine the necessity for molecular testing: SCADZ / Short-Chain Acyl-CoA Dehydrogenase (SCAD) Deficiency, Full Gene Analysis

- **Plasma or serum C4–High**
  - **Urine C4–High**
  - **Urine ethylmalonic acid and isovalerylglycine–High**

  **Diagnostic for ethylmalonic encephalopathy**

  Consider confirmatory testing for ETHE1 gene

- **Plasma or serum C4–Normal**
  - **Urine C4–Normal**
  - **Urine isobutyrylglycine–Normal**

  **STOP**

* Interpretive report provided for all tests in this algorithm