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

Isolated elevation of C4 acylcarnitine

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

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

- 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

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* Interpretive report provided for all tests in this algorithm