MAYO CLINIC LABORATORIES

Newborn Screen Follow-up for Elevated C5-OH

Newborn screen result: Elevated 3-hydroxyisovaleryl-/2-methyl-3-hydroxybutyryl-carnitine (C5-OH)

Perform:
Plasma ammonia, blood gas, glucose, electrolytes
Order:
- OAU / Organic Acids Screen, Random, Urine
- AGU20 / Acylglycines, Quantitative, Random Urine

AND 1 of the following:
- ACRN / Acylcarnitines, Quantitative, Plasma
- ACRNS / Acylcarnitines, Quantitative, Serum

- Consistent with 3-methylcrotonyl carboxylase deficiency
  - Consider CGPH / Custom Gene Panel, Hereditary, Next-Generation Sequencing, Varies (*MCCC1* and *MCCC2* Gene List ID: IEMCP-LFGJWQ)
- Consistent with 3-hydroxy-3-methylglutaryl-CoA lyase deficiency
  - Consider CGPH (*HMGCL* Gene List ID: IEMCP-MW6XC3)
- Consistent with beta-ketothiolase deficiency
  - Consider CGPH (*ACAT1* Gene List ID: IEMCP-T3JFHV)
- Consistent with 2-methyl-3-hydroxybutyric acidemia/2-methyl-3-hydroxybutyryl-CoA dehydrogenase (2M3HBA/MHBD) deficiency
  - Consider CGPH (*HSD17B10* Gene List ID: IEMCP-SJF9TL)
- Consistent with 3-methylglutaconic aciduria type 1
  - Order 3MGAP / 3-Methylglutaconic Aciduria Panel, Varies
- Suggestive of mitochondrially encoded ATP synthase membrane subunit 6 (MTATP6)-related mitochondrial disease
  - Order MITOP / Mitochondrial Full Genome Analysis, Next-Generation Sequencing (NGS), Varies
- Suggestive of multiple carboxylase deficiency (MCD)
  - Order BIOTS / Biotinidase, Serum OR Review biotinidase result on newborn screen
    - Consistent with biotinidase deficiency
      - Consider BTDZ / Biotinidase Deficiency, BTD Full Gene Analysis, Varies
    - Normal
      - Consider holocarboxylase synthetase deficiency
- Normal
  - False positive newborn screen
  - Rule out maternal inborn error of metabolism

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