**C5-OH Elevated**

Elevated C5-OH

- **Routine Labs:** Glucose, electrolytes, blood gas, ammonia

- **Assay:** Urine OA, Plasma AC

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**Plasma C5-OH: high**
- Urine OA: MCC

**Plasma C5-OH: high**
- Urine OA: BKT

**Plasma C5-OH: high**
- Urine OA: MGA

**Plasma C5-OH: high**
- Urine OA: MHBD

**Plasma C5-OH: high**
- Urine OA: MCD

**Plasma AC: normal**
- Urine OA: normal

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**3-MCC**
- **HMG-CoA Lyase deficiency**

**Beta-ketothiolase deficiency**
- **2M3HBA(MHBD) deficiency**

**3-methylglutaconic aciduria type 1**
- **Biotinidase deficiency**
- **Biotin deficiency**

- **Optional confirmatory testing:** DNA analysis and/or enzyme assay

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**Abbreviations/Key:**
- 2M3HBA = 2-methyl-3-hydroxybutyric acidemia
- AC = acylcarnitine
- BKT = Beta-ketothiolase
- HMG-CoA = 3-hydroxy-3-methylglutaryl-CoA
- IEM = inborn error of metabolism
- MCC = methylcrotonyl-CoA carboxylase
- MCD = multiple carboxylase deficiency
- MGA = 3-methylglutaconic aciduria
- MHBD = 2-methyl-3-hydroxybutyryl-CoA dehydrogenase
- OA = organic acid

* = Maternal MCC and holocarboxylase deficiency have been reported as having been identified in newborn screening.
‡ = When the positive predictive value of screening is sufficiently high and the risk to the newborn is high, some initiate diagnostic studies that are locally available at the same time as confirmation of the screening result is done.

**Disclaimer:** These standards and guidelines are designed primarily as an educational resource for physicians to help them provide quality clinical services. Adherence to these standards and guidelines does not necessarily ensure a successful medical outcome. These standards and guidelines should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed to obtaining the same results. In determining the propriety of any specific procedure or test, the healthcare provider should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. It may be prudent, however, to document in the patient’s record the rationale for any significant deviation from these standards and guidelines.

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