Emergency Management Protocol for Isovaleric Acidemia (C5)
(Primary Analyte C5 < 0.87 µmol/L Secondary Analyte C5/C2 < 0.05 µmol/L)
Newborn Screening Program of the Oklahoma State Department of Health

Evaluation & Initial Management Guidelines for High Risk Isovaleric Acidemia Screen Results

1. Contact the family by within **one hour** of notification. Inform family of newborn screening results and assess clinical status (poor feeding, vomiting, lethargy, tachypnea.)
2. **Immediate consult with the geneticist**– pager number listed below.
3. History and Physical Exam in pediatrician's office or at local Emergency Department if after hours, in consultation with the geneticist:

   May appear normal at birth

   Assess specifically for signs and symptoms of Metabolic Crises:

   - Poor Feeding
   - Hypoglycemia
   - Lethargy
   - Metabolic Ketoacidosis
   - Vomiting /Dehydration
   - Seizures
   - Hyperammonemia
   - "Sweaty Feet"Odor
   - Coma
   - Failure to thrive

4. **If symptomatic**, immediate phone consultation with a geneticist regarding treatment and emergency clinical management is required.
5. If not symptomatic, in consult with a geneticist for medical management, consider admission until follow-up with a geneticist and schedule diagnostic work-up within 24 hours.
6. Ensure that baby is awoken **every three hours** for feedings. If baby is **feeding poorly**, **lethargic** and/or **vomiting** seek **immediate attention**.

Description

This disorder is caused by an abnormal buildup of Isovaleric acid. People with this inherited organic acid disorder cannot process certain proteins properly which can lead to a buildup of particular organic acids. Abnormal levels of organic acids in the blood, urine and tissue can be toxic and lead to serious health problems.

Resources

- **ACMG Newborn Screening ACT Sheets:** https://www.ncbi.nlm.nih.gov/books/NBK55827/
- **Integris Pediatric Specialty Clinic, Inborn Error of Metabolism (IEM) Clinic**
  Geneticist pager: (405) 630-3794
- **OU Children's Physicians – Genetics Clinic**
  Page Operator: (405) 271-3636
- **Newborn Screening Follow-Up Program**
  (405) 271-6617 option 2 or (800) 766-2223; www.nsp.health.ok.gov