Emergency Management Protocol for Biotinidase

Newborn Screening Program of the Oklahoma State Department of Health

Evaluation & Initial Management Guidelines for High Risk Biotinidase screening results

1. Contact the family within **one hour** of notification. Inform family of newborn screen results and assess clinical status.
2. **Immediate consultation with the geneticist** – pager number listed below.
3. History and Physical Exam **on same day of notification** either in the pediatrician's office or in the local Emergency department if after hours, in consultation with a geneticist.
   - May appear normal at birth
   - Assess specifically for signs and symptoms:
     - Lethargy
     - Hypotonia
     - Dematitis
     - Alopecia
     - Seizures
     - Ataxia
     - Ketoacidosis
     - Vomiting/Diarrhea
     - Mild Hyperammonemia
     - Vision Problems/Conjunctivitis
     - Hearing loss
     - Breathing problems such as hyper- ventilation, stridor or apnea
     - Developmental Delay (childhood)
4. **If symptomatic**, **immediate** phone consultation with a geneticist regarding treatment and emergency clinical management is required.
5. If not symptomatic, schedule diagnostic workup with a geneticist within 24-48 hours.

Description

This disorder is caused by a deficiency of the enzyme biotinidase. People with this inherited genetic disorder cannot cleave biocytin to produce biotin and lysine producing a biotin deficiency. This deficiency can lead to characteristic features of this disorder such as alopecia and seizures.

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