

## 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 metabolic specialist** – pager and phone numbers 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 metabolic specialist.
  - May appear normal at birth
  - Assess specifically for signs and symptoms:
    - Lethargy
    - Hypotonia
    - Dermatitis
    - 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 metabolic specialist regarding treatment and emergency clinical management is required.
5. If not symptomatic, schedule diagnostic workup with metabolic specialist 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

**Mary Monks, RN Metabolic Nurse Specialist**  
**Phone (405) 271-8685 Pager (405) 559-1378**

**Newborn Screening Follow-up Program (405) 271-6617 or 1-800-766-2223**

[www.nsp.health.ok.gov](http://www.nsp.health.ok.gov)

### Metabolic Specialists

**Klaas Wierenga, MD**

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**Susan Palmer, MD**

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