

LISTEN FROM EAR TO EAR TIPS OF THE MONTH

January/February
2013

NEWBORN HEARING
SCREENING PROGRAM (NHSP)

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Biotinidase and Hearing Loss



What is Biotinidase Deficiency?

- Biotinidase deficiency is an inherited disorder in which the body is unable to reuse and recycle the vitamin biotin.
- Infants with Biotinidase deficiency do not have symptoms at birth but may develop critical symptoms within the first few weeks or months of life
- Children diagnosed with profound Biotinidase deficiency if left untreated have:
 - ◊ seizures, weak muscle tone (hypotonia), breathing problems, delayed development, hearing loss, loss of vision, problems with movement and balance (ataxia), skin rashes, hair loss (alopecia) and fungal infections (candidiasis).
- Children diagnosed with untreated partial biotinidase deficiency experience similar symptoms primarily during times of stress i.e. illness, infection and other times of stress.

The role of biotinidase in the auditory system as a cause of hearing loss is unknown therefore, it is recommended that babies with this deficiency have hearing monitored periodically.

Biotinidase deficiency occurs in approximately 1/60,000 births. Since the Oklahoma Newborn Screening program started screening for Biotinidase deficiency in November 2010, 27 infants have been identified with partial deficiency 2 with profound deficiency.

Detection of the deficiency through the Newborn Screening (NBS) does not depend on protein or lactose ingestion; therefore, may be identified on the first specimen unless the infant has been transfused.

The newborn screen sample collected when the baby is 24 hours of age or older is a reliable screening test for Biotinidase deficiency. If a sample is contaminated with alcohol this could lead to a false positive result for this disorder.

Treatment:

- Daily biotin supplements clear the skin rash and alopecia and improve neurological status in patients not diagnosed by screening.
- With early diagnosis and treatment, symptoms can be prevented.

For any questions, please contact Screening and Special Services at 1-800-766-2223

REMEMBER:

Folic Acid Awareness Week

January 6-12, 2013

- Folic acid can help prevent serious birth defects if taken in the first few weeks of pregnancy.
- Easiest source is a daily vitamin with **400 mcg** of folic acid.
- Eat plenty of fruits and green leafy vegetables.
- Some breads and breakfast cereals have 100% of the Daily Value of folic acid.

EVENT OF THE MONTH

January

is

Birth Defects Prevention
Month