Arkansas Department of Health
Newborn Screening Result: Absent activity of Biotinidase enzyme
Interpretation Sheet for Parents
Infant may have Biotinidase deficiency

You have just heard that your infant may have Biotinidase deficiency. Please understand that the newborn screening is just that – a screening test. Not all cases that are screened positive will be confirmed to have the diagnosis. The below information is meant to keep you informed while further testing is done to evaluate the newborn screening result.

Overview
Biotinidase deficiency is a rare condition that affects the way a person's body uses the vitamin biotin. Individuals with biotinidase deficiency cannot use the biotin that is normally found in foods and recycled in the body. Treatment is very effective for these individuals, and can prevent any symptoms from occurring.

What is biotinidase deficiency?
Biotinidase deficiency is an inherited disorder that prevents babies from using the vitamin biotin in a normal manner. Biotin is found in many foods and is essential for proper growth and development. An enzyme (a type of chemical in our body) called biotinidase helps separate biotin from the food we eat so that our body can then use it. In babies with biotinidase deficiency, the enzyme doesn't work well so the baby's body doesn't get enough biotin and cannot recycle the biotin that is already there.

Why is newborn screening done for biotinidase deficiency?
Newborn screening is done for biotinidase deficiency so that babies with this condition can be diagnosed quickly. If babies are diagnosed quickly, treatment can begin before any health problems occur.

Does a positive result of newborn screening mean that a baby has biotinidase deficiency?
Not always. Some babies who are screened "positive" are found not to have the disorder after further testing. This is because the screening test is not a diagnostic test. A screening test is designed to identify all infants with the disorder, but sometimes picks up infants who don't have the disorder. Therefore further testing needs to be done in order to determine if the baby has the disorder, or to rule it out as a "false positive."

How common is biotinidase deficiency?
This condition occurs in about 1 out of every 60,000 babies born.

What are the signs and symptoms of Biotinidase Deficiency?
Babies that have biotinidase deficiency will appear normal at birth. Within a few weeks or months after birth, symptoms will develop if the individual is untreated. The number of symptoms that a baby will develop, as well as how severe the symptoms will be, varies from baby to baby. Some common early signs include: seizures, low muscle tone (floppiness), hair loss, and skin rash.

Once treatment begins, most of the symptoms will disappear. If babies are diagnosed early and treatment is started before symptoms appear, they usually won't develop any symptoms.

How is biotinidase deficiency diagnosed?
After receiving a positive newborn screen, the most important thing parents can do is be sure that their baby goes in for a new specimen to be collected and tested as soon as possible. The test results should be available within 3 days.

How is biotinidase deficiency treated?
Children with biotinidase deficiency will need to take extra biotin every day. Your baby's doctor will start biotin supplements while waiting for the results of the diagnostic test. If the test confirms that your baby has biotinidase deficiency, he or she will have to take the supplement throughout their life. If the diagnostic test is normal, then the biotin supplements can be discontinued. There are no side effects associated with Biotin supplementation. The body will excrete excess Biotin in the urine.

Where can I get additional information?
http://www.arkansas.gov/newborn_screening/index.html