Biotinidase Deficiency (BTD)

Biotinidase deficiency (BTD) is caused by the lack of an enzyme called biotinidase. Without treatment, this disorder can lead to:

- Seizures.
- Developmental delay.
- Eczema.
- Hearing loss.

CAUSES

This is an inherited problem. It means parents pass this condition down to their children. It is caused when a male and female carrier of this gene have children. In a pregnancy between two carriers, there is a 25% chance (1 in 4) that the child will be born with the disease. There is a 50% chance (1 in 2) the child will be a carrier for the gene defect. One in every four children of this couple will be normal.

SYMPTOMS

Infants with biotinidase deficiency appear normal at birth. But they develop symptoms after the first weeks or months of life. Problems include:

- Looseness of the muscles (*hypotonia*).
- Convulsions (*seizures*).
- Loss of hair (*alopecia*).
- Hearing loss.
- Metabolic acidosis that can result in coma and death.
- Lack of coordination of movements (*ataxia*).
- Developmental delays.
- Cradle cap (*seborrheic dermatitis*).
- Loss of vision (*optic nerve atrophy*).

TREATMENT

Biotinidase deficiency is treated with biotin. In infants diagnosed through screening, treatment will clear the skin rash and alopecia. It will also improve the status of the nervous system. It is necessary that treatment be managed by your caregiver. Your caregiver will make sure the biotin is in a safe and usable form and also monitor the deficiency.

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