

## **Citrullinemia (CIT)** (Sit-ru-li-NE-mia)

### **What is CIT?**

Citrullinemia (CIT) is a metabolic disorder. This means the body has a chemical imbalance. CIT is a condition in which the body is not able to process protein properly. People with this condition lack a certain enzyme. The enzyme is called argininosuccinic acid synthetase (AS). The lack of this enzyme results in a build-up of chemicals in the blood. These chemicals include citrulline and ammonia. CIT is inherited. This means it is present at birth.

### **What type of problems occurs with CIT?**

A baby who has this condition may refuse to eat. The baby may develop a lack of energy (lethargy) and vomiting. If untreated, this condition could lead to seizures and coma. Early diagnosis and treatment will help prevent these problems.

### **What is the chance my baby will have CIT?**

This condition occurs in less than 1 in every 100,000 births. Babies born with this condition have a changed gene from each parent. A person who has one changed gene is called a carrier. A person who is a carrier does not have symptoms. If both parents are carriers, either parent can pass on the changed gene to their baby. If both parents pass on the changed gene, the baby will have the condition. If both parents are carriers, for each pregnancy:

- There is a 25% chance that the baby will be born with this condition.
- There is a 50% chance that the baby will be a carrier for this condition.
- There is a 25% chance that the baby will not be born with this condition and will not be a carrier.

### **What is the treatment of CIT?**

The treatment of this condition usually consists of a special diet. Nutritional supplement are sometimes needed. Your baby's metabolic doctor will help you make sure that your baby gets the right diet and medical care. Sometimes a liver transplant is needed.

### **Where in Virginia can I take my baby for care?**

Please speak to your baby's pediatrician about obtaining a referral to a pediatric metabolic specialist in your area. If you want to know more about this condition, please contact Virginia Newborn Screening Services, Virginia Department of Health. The Web site is <http://www.vahealth.org/gns>.



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