

KENTUCKY NEWBORN SCREENING PROGRAM

PARENT TEACHING SHEET

Kentucky Newborn Screening Program, 275 East Main Street, Frankfort, KY 40621, Phone (502) 564-3756, Fax (502) 564-1510

Citrullinemia (CIT)

CAUSE

CIT occurs when an enzyme called “argininosuccinic acid synthetase” (ASAS), is either missing or not working properly. This enzyme’s job is to help break down certain amino acids and to remove ammonia from the body. When the enzyme is not working, an amino acid called citrulline builds up in the blood. Ammonia and other harmful substances also build up. This causes brain damage and, if not treated, can cause death.

IF NOT TREATED

Normally, the body changes ammonia into a substance called “urea.” Urea is then safely removed from the body in the form of urine. If urea is not removed from the body, it begins to build up in the blood and causes brain damage. In the most common form of CIT, infants may seem healthy at birth but develop symptoms within the first few days of life. There is also a milder form of CIT in which symptoms start in late infancy or early childhood. Without treatment, most babies die within the first few weeks of life.

TREATMENT OPTIONS

Your child will need to be under the care of a metabolic specialist and dietician. Treatment is usually needed throughout life.

- Most children need to eat a diet made up of very low-protein foods, special medical foods, and sometimes a special formula. The dietician will develop a plan for you to follow.
- Most children with CIT are given arginine supplements. Arginine helps the body remove ammonia from the blood. Your child’s metabolic specialist will determine the best treatment available. Arginine is available by prescription only.
- Your child will require frequent blood tests to monitor amino acid and ammonia levels in the blood. Diet and medication may require adjustments based on these lab results.
- Contact your child’s doctor immediately at the start of any illness. Children with CIT may need to be treated in a hospital to prevent serious health problems.

IF TREATED

With prompt and lifelong treatment, children with CIT may be able to live healthy lives with typical growth and learning. Early treatment can lessen the risk for brain damage and mental delays by preventing high ammonia levels. Even with treatment, some children still have episodes of high ammonia. This can result in brain damage and can cause lifelong learning problems, mental delays, and spasticity (spasms of the muscles and tendons).



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