

PARENT FACT SHEET

DISORDER

Argininosuccinic Acidemia (ASA)

CAUSE

ASA occurs when an enzyme called Argininosuccinic acid lyase is either missing or not working properly. This enzyme's job is to help remove ammonia from the body. When the ASA lyase enzyme is not working, ammonia and other harmful substances build up in the blood.

IF NOT TREATED

Normally, the body changes ammonia into a substance called "urea." Urea is then safely removed from the body in 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 ASA, infants are generally healthy at birth but usually develop symptoms within a few days of life. Without treatment, many babies die within the first few weeks of life. There is also a second form of ASA in which very mild symptoms start in late infancy or early childhood.

TREATMENT OPTIONS

Your child will need to be under the care of a metabolic specialist and dietician. Treatment is needed throughout the child's 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 ASA 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 ASA may need to be treated in a hospital to prevent serious health problems.

IF TREATED

With prompt and lifelong treatment, children with ASA 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).