UREA CYCLE DEFECTS [UCD]

What is the Urea Cycle?

When we eat food, specifically proteins, our body breaks them down into smaller substances to be used efficiently for our body's function. Our body makes use of enzymes or chemical scissors to cut up proteins into building blocks called amino acids. We usually take in more protein than our body needs. The excess protein is then broken down into ammonia and organic acids. A high amount of ammonia in the body is dangerous and should be properly excreted. The urea cycle is the pathway for the ammonia to be processed and excreted by the body.

What is a Urea Cycle Defect?

A urea cycle defect occurs if there is a lack or absence of any enzyme within the cycle. While the presentation may be variable, it is one of the conditions that can be detected through newborn screening. Untreated children may have drowsiness, fast breathing, vomiting, irritability and they may die.

What is Citrullinemia type 1?

Citrullinemia type 1 is a condition that results from a lack of the enzyme argininosuccinate synthetase. Argininosuccinate synthetase is one of the enzymes, or chemical scissors, which are part of the urea cycle. The condition is called citrullinemia because citrulline (an amino acid) accumulates along with ammonia.

What is Argininosuccinic Aciduria?

Argininosuccinic Aciduria (or ASA) is a condition that results from a lack of the enzyme, or chemical scissors, argininosuccinate lyase. Argininosuccinate lyase is one of the enzymes which are part of the urea cycle. The condition is called argininosuccinate aciduria because argininosuccinate (an intermediate product of the cycle) accumulates along with ammonia.

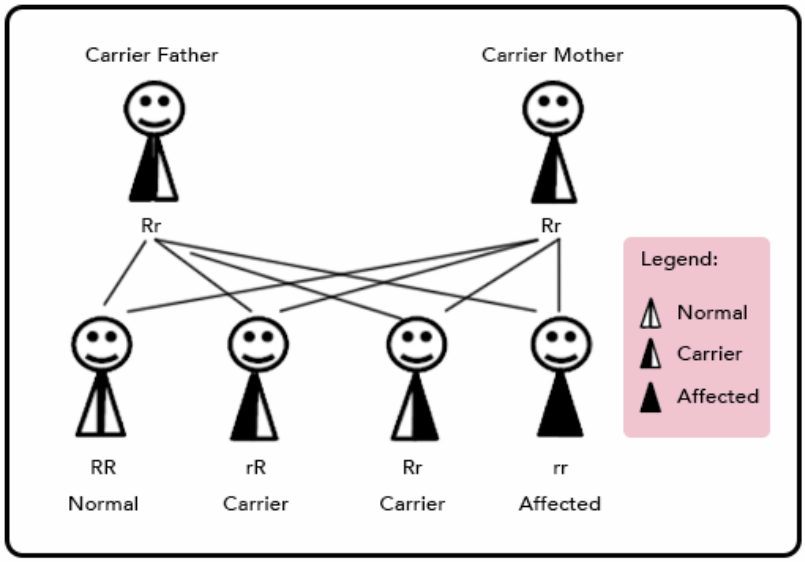
What Causes Urea Cycle Defects (Citrullinemia type 1 and Argininosuccinic Aciduria)?

Citrullinemia type 1 and Argininosuccinic aciduria are inherited conditions. The genes coding for the enzymes are contained in the genetic material that we inherit from our parents. Because one part of the genetic material comes from the father and the other from the mother, the gene comes in pairs. In order to work correctly, at least one of the pairs should be working.

Parents of children with this condition have one working and one non-working gene coding for the enzyme. They do not manifest the disease but can pass them on to their children. They are known as carriers.

Controlled Copy

UREA CYCLE DEFECTS [UCD]



If the child inherits the non-working gene from both parents, he or she will have the condition. Thus, in each pregnancy, there is a 25% chance that the child will have the disorder, 50% chance of being a carrier and 25% chance of having two working genes.

WHAT ARE THE SIGNS AND SYMPTOMS OF UCD?

Children with UCD may may have drowsiness, fast breathing, vomiting, convulsions, irritability and they may die.

WHAT IS THE TREATMENT OF UCD?

The main aim of treatment of urea cycle defects is to keep the ammonia levels in the blood low or normal. To achieve this, patients with UCD are advised to follow a low protein diet and to take a special milk formula. Sodium benzoate, a medication that helps in getting rid of excess ammonia, is also prescribed. Because children with UCD are found to have low levels of arginine (an essential amino acid), they are also given arginine supplementation.

Please consult with your attending physician prior to starting treatment.

WHAT SHOULD I DO WHEN MY BABY IS SICK? (WILL UNDERGO SURGERY OR HAS AN INFECTION I.E. RESPIRATORY OR GASTROINTESTINAL)?

When children with UCD are sick, ammonia may accumulate in the blood. Your child may present with drowsiness, vomiting, seizures or convulsions and irritability. If not treated properly and immediately, it might lead to serious brain damage. Once these signs and symptoms are present, please bring your child to the hospital for management and alert your pediatrician or metabolic physician.

Controlled Copy