Tyrosinemia, type 1 (TYR I)

CAUSE
People with tyrosinemia 1 have problems breaking down an amino acid called tyrosine from the food they eat. In order for the body to use protein from the food we eat, it is broken down into smaller parts called amino acids. Special enzymes then make changes to the amino acids so the body can use them. Tyrosinemia 1 occurs when an enzyme called fumarylacetoacetase (FAH) is either missing or not working properly.

IF NOT TREATED
The symptoms can vary from person to person. Children with TYR 1 usually smell like cabbage. Babies usually will have symptoms within the first few months of life. Without prompt and careful treatment, babies with severe liver and kidney problems usually die.

TREATMENT OPTIONS
• Your doctor will work with a metabolic specialist and dietician to care for your child. Lifelong treatment is usually needed to prevent liver and kidney problems.
• A medication called nitisinone (Orfadin®) is used to prevent liver and kidney damage. Your child should take this medication as soon as possible. Nitisinone will increase the level of tyrosine in your child’s blood.
• Also a low tyrosine and low phenylalanine diet is very important. Your baby will have to avoid cow’s milk, regular formula, meats, eggs and cheeses. A special formula will be ordered for your baby so they will get those nutrients and protein they will need to keep their levels within a safe range.
• Your child will have regular visits to the doctor as well as regular blood tests to see if there should be any changes to the medication. Contact your child’s doctor immediately at the start of any illness.
• Liver transplant may be an option to prevent liver cancer.

IF TREATED
Children who are treated early usually live healthy lives with typical growth and development. When treatment is started early, severe kidney and neurologic symptoms can be prevented. If treatment is not started right away, liver and kidney damage may occur. Delays in growth and development may also be present.