Tyrosinemia (Type I) – Amino Acid Disorder

What are amino acid disorders?

The amino acid disorders are a class of inherited metabolic conditions that occur when certain amino acids either cannot be broken down or cannot be produced by the body, resulting in the toxic accumulation of some substances and the deficiency of other substances.

What is tyrosinemia?

In tyrosinemia, the amino acid tyrosine cannot be broken down properly, leading to a toxic accumulation of this amino acid and its metabolites in the body.

What is its incidence?

Tyrosinemia affects about 1 out every 100,000 babies born in BC. Although tyrosinemia occurs in all ethnic groups, it is more common in certain populations. Its incidence has been reported as high as 1 in 2,000 in the French Canadian population living in the Saguenay-Lac-St-Jean region of Quebec.

What causes the disease?

Mutations in the gene for fumarylacetoacetase (FAH) result in enzyme that is not working well or is deficient.

What are the clinical features of the disease?

Babies with tyrosinemia are normal at birth. Unless they are treated, however, damage to the liver and kidneys will begin to occur. Early symptoms can include failure to thrive, lethargy, vomiting and diarrhea, and irritability. The symptoms of liver damage can include an enlarged liver, jaundice, a tendency towards bruising, and swelling of the legs or abdomen. Kidney disease may lead to rickets, a bone disease. The nerves may also be affected. Some babies may have a rapid heart rate, breathing difficulties, and seizures. Occasionally, individuals with liver damage have a higher risk of developing liver cancer. Acute liver and kidney damage can lead to death.

How is the diagnosis confirmed?

The diagnosis is confirmed by measuring the levels of amino acids in the blood and organic acids in the urine. The finding of succinylacetone in the urine is diagnostic. Enzyme testing and genetic testing of the FAH gene may also be used to confirm the diagnosis. Diagnostic testing is arranged by specialists at BC Children's Hospital.

What is the treatment of the disease?

Children with tyrosinemia are treated with a medication called nitisinone (previously called NTBC). A diet low in tyrosine, phenylalanine, and methionine, and a special medical formula is often recommended. Dietary supplementation with vitamin D may be considered. In the long term, individuals with liver damage or liver cancer may require a liver transplant. Treatment is coordinated by specialists at BC Children's Hospital.

What is the outcome of treatment?

Treatment can prevent liver disease, kidney problems, and the neurological problems that can be associated with tyrosinemia.

The BC Newborn Screening Program is a collaboration of BC Women's Hospital & Health Centre, BC Children's Hospital and the BC Perinatal Health Program, all part of the Provincial Health Services Authority.

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Can a family have more than one child with tyrosinemia?

Tyrosinemia is inherited as an autosomal recessive disorder. The parents of a child who has this disorder are assumed to be carriers for the disease and have a 1 in 4 (25%) chance, in each pregnancy, of having another child with tyrosinemia. Prenatal testing can be done as early as 10-12 weeks of pregnancy. Genetic counselling to discuss the benefits of prenatal testing options in more detail is recommended.

Unaffected siblings of a child with tyrosinemia have a 2/3 chance of being carriers. Tyrosinemia carriers are healthy and do not have symptoms of the disease.

Resources

http://www.newbornscreening.info/Parents/aminoaciddisorders/Tyrosinemia.html
http://depts.washington.edu/tyros/pdfs/New_Parents_Guide_to_Tyrosinemia.pdf
http://www.geneclinics.org

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