

Hereditary Tyrosinemia Type 1 Vesna Ambarkova*

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Description

Hereditary Tyrosinemia Type 1 is a rare genetic disorder in which newborns do not have the ability to break down the amino acid tyrosine. As a result of this deficiency, toxic substances accumulate in the blood and can cause liver failure, kidney dysfunction and neurological problems [1].

There are two different forms of the disease-acute and chronic. The acute form is more common. In the world, Hereditary Tyrosinemia Type 1 affects 1 in 100,000 children. The risk of giving birth to a child with this disease, in fact to inherit both defective genes from the parents is 25%. This is an autosomal recessive disease [2].

Children with Tyrosinemia Type 1 show symptoms such as inability to gain weight. Diarrhea, vomiting, enlarged liver, accumulation of fluid in the peritoneal cavity, renal failure, softening of the bones, and occurring of liver tumors. The acute form usually appears in the first months of life. The child has low weight, fever, diarrhea, blood in the stool and vomiting. The liver is enlarged and the skin along with the whites in the eyes turned yellow. Bleeding from the nose is also common. The spleen may also be enlarged, and the legs swelled. Without treatment many problems continue to arise and make life difficult. In children with a chronic form of Tyrosinemia Type 1, the symptoms develop gradually. The child may have an enlarged liver and abdomen, skeletal changes, liver and kidney failure. Symptoms such as abdominal pain, peripheral nerve damage, and high blood pressure may also occur. If left untreated, the child will develop a more severe form of liver failure and will develop liver tumors.

When this disease is suspected, tests and analyzes of the amino acids, succinylacetone and alpha-fetoprotein are performed. Typical biochemical tests show increased succinylacetone concentration in blood and urine, increased plasma tyrosine, methionine and phenylalanine concentration, increased urinary concentration of tyrosine metabolites and beta-ALA [3]. Today this disease is treated with diet, medication and liver transplantation. Prior to 1991, when the first drug was developed, the disease was treated only with liver transplantation. Diet and special protein replacement therapy are an important part of lifelong treatment. Combination treatment with nitisinone and a diet low in tyrosine are commonly used, as this allows for normal growth, improved liver function, prevention of cirrhosis, and contribute to skeletal improvement. The first thing you have to do for easier management of Tyrosinemia Type 1 is of course educate yourself as much as possible to know all the symptoms

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and complications that may occur and to know how to react to help. Stick to a diet low in phenylalanine and tyrosine. Vitamin D as a dietary supplement can help improve the condition. Regular blood and urine tests are needed to control the levels of the amino acids succinylacetone and alpha-fetoprotein. Liver and kidney function should also be checked. Be aware of the fact that a liver transplant may be needed to prevent liver tumors. That is why liver scans are done so that in case of tumors, they can be detected early and can be removed. The parents of children with this disease should consult a gastroenterologist and nephrologist.

Nitisinone[2-(2-nitro-4-trifluoromethylbenzoyl)-1,3-cyclohexanedione; NTBC] treatment should be started as soon as the patient is diagnosed to prevent the primary manifestations of the disease [4]. To prevent secondary complications, such as carnitine deficiency, osteoporosis, and rickets, treatment should be started to help alleviate such a condition as vitamin D supplementation. Despite the dramatic improvements in survival, outcomes and quality of life after NTBC treatment, Hereditary Tyrosinemia Type 1 as a chronic disorder leaves several long-term consequences, including a persistent risk of hepatocellular carcinoma [5-7]. Visits to the doctor and regular check-ups are important for controlling the disease and any consequences that may occur. In order to start treatment as early as possible after early diagnosis and treatment of particular importance is testing of the children of parents with Tyrosinemia immediately after birth. There are no data to suggest problems during pregnancy from the use of nitisinone treatment. But the developing fetus is at risk for tyrosine changes.

With treatment, 90% of patients have a normal life with normal growth, stabilization of liver function, prevention of cirrhosis,

correction of skeletal damage and rickets. With early treatment, the incidence of liver cancer is reduced.

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