

FACT SHEET

Healthcare Provider

Tyrosinemia Type I

Description:

Tyrosinemia Type I is a rare autosomal recessive disorder due to fumaryl acetoacetase deficiency, an enzyme involved in the catabolism of tyrosine. Tyrosinemia Type I, the most severe form of tyrosinemia, results in the accumulation of tyrosine and its metabolites in the liver causing severe liver damage. In the acute infantile form, onset of symptoms occurs between 2 and 6 weeks of age with symptoms of failure to thrive and severe liver dysfunction.

Symptoms:

Patients with Tyrosinemia Type I may have acute liver disease, episodes of peripheral neuropathy, and chronic liver disease. Kidney function and peripheral nerves also are affected. Effects on the kidneys can range from mild tubular dysfunction to renal failure. Symptoms may include poor weight gain, fever, diarrhea, vomiting, enlarged liver and spleen, swelling of the legs, and increased tendency of bleeding.

Diagnosis:

Newborn screening—Tandem mass spectrometry identifies elevated Tyrosine.

Some cases of tyrosinemia may not be detected by newborn screening when specimens are collected in the first few days of life, as tyrosine levels may not be sufficiently elevated for detection by tandem mass spectrometry.

A second dried-blood-spot filter paper card may be requested by the Newborn Screening Laboratory if the initial screening result is above the normal range. Infants with presumptive positive screening (critical) results require prompt follow up. If this occurred, the clinician would be contacted by the Metabolic Treatment Center. When notified of these results, the clinician should immediately check on the clinical status of the baby and facilitate referral to the Metabolic Treatment Center. The Metabolic Treatment Center will provide consultation and assistance with diagnostic testing.

Monitoring:

When receiving a presumptive positive result, the clinician should immediately check on the clinical status of the baby and consult with the Metabolic Treatment Center. Long-term management, monitoring, and compliance with treatment recommendations are essential to the child's well being. Regular monitoring of laboratory values is also part of the treatment. Monitoring includes:

- Regular monitoring of plasma amino acid concentrations.
- Regular clinic appointments (this varies between clinics and depends on patient's status).

Treatment:

Early diagnosis and prompt treatment is essential for an improved prognosis. There are three strategies for tyrosinemia type I.

- Treatment consists of a special dietary formula restricted in tyrosine and phenylalanine.
- Medication treatment with Nitisinone (Orfadin®) has been successful to date and has improved the outcome in tyrosinemia type I.
- In some cases, liver transplantation has been an effective treatment.

Illness and Immunizations:

During illness, it is important to minimize tissue breakdown. This can be accomplished by encouraging a continuous intake of the specialized metabolic formula during these times. It is important to avoid dehydration. With severe illness, it may be necessary to introduce an intravenous line for fluids or a nasogastric tube for formula intake. There are no immunization contraindications because of Tyrosinemia.

Growth and Development:

- A child with appropriately managed blood tyrosine levels can look and act like other children of the same age.
- It is crucial to closely monitor all growth parameters on a regular basis.
- To support ideal growth for children with tyrosinemia, intakes of energy (calories), protein, carbohydrate, fat, vitamins, and minerals are carefully monitored.



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