

(TYR) TYROSINEMIA - TYPE II & III

Amino Acid Disorder

Definition

Tyrosinemia is an inborn error of tyrosine metabolism. Tyrosinemia type II or oculocutaneous tyrosinemia is caused by a deficiency of the enzyme tyrosine aminotransferase. Tyrosinemia type III or 4-hydroxyphenylpyruvate dioxygenase deficiency occurs because of an enzyme deficiency of 4-hydroxyphenylpyruvate dioxygenase.

Incidence

Studies show that 1 of every 100,000 live births will have Tyrosinemia. Both sexes are affected equally.

Inheritance

Tyrosinemia Type II is an autosomal recessive genetic condition.

Characteristics

Type II: Clinical features involve only the skin, eyes and central nervous system. Clinical onset is variable. Skin and eye symptoms often present within the first year of life and include excessive tearing, photophobia, eye pain and redness, and skin lesions.

Type III: The clinical presentation of this form of tyrosinemia is not well known. Transient tyrosinemia: Elevated tyrosine levels in a healthy newborn with no liver, renal, and skin abnormalities. Risk factors include prematurity, high protein intake, and deficient intake of Vitamin C.

Transient tyrosinemia of the newborn: Most common disorder of amino acid metabolism in humans. Significant elevations of tyrosine in an asymptomatic infant that fall over time and may respond to ascorbic acid, together with the absence of liver, renal or cutaneous signs, distinguishes transient tyrosinemia from the other more serious forms of tyrosinemia.

Newborn Screening Methodology

The laboratory method used is tandem mass spectrometry for the amino acid tyrosine.

| RESULT | ACTIONS |
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| Within Normal Limits (<9.1 mg/dL or <336 μ M tyrosine) | Normal report sent to submitter. |
| Presumptive Positive (≥ 9.1 mg/dL or ≥ 336 μ M tyrosine) | The program follow-up staff will notify attending physician. |

Confirmation

Consultation with a Pediatric Metabolic Specialist or Geneticist should be made for confirmation and diagnosis, (319) 356-2674. Quantitative plasma tyrosine levels along with clinical history and physical evaluation determine the diagnosis.

Treatment and Outcome

Treatment includes a diet low in phenylalanine, methionine and tyrosine. The drug 2-(2-nitro-4-trifluoromethylbenzyl)-1,3-cyclohexanedione (NTBC) has been successful in the management of tyrosinemia. NTBC works by inhibiting the proximal tyrosine metabolic pathway.

Screening Practice Consideration

Some healthy newborns may have elevated tyrosine levels with no liver, renal and skin abnormalities. Risk factors for transient tyrosinemia include prematurity, high protein intake, and deficient intake of Vitamin C.

Other Sites of Reference

Dietary Specialties - Low Protein Foods

www.dietspec.com/

National Organization for Rare Disorders - Tyrosinemia

www.rarediseases.org/search/rdbdetail_abstract.html?disname=Tyrosinemia%2C%20Hereditary

Tyrosinemia Family Website

www.groups.msn.com/tyrosinemia

Support Groups

National Coalition for PKU & Allied Disorders

P.O. Box 1244

Mansfield, MA 02048

www.pku-allieddisorders.org/

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