

Disease Name:**TYROSINEMIA, TYPE II**

TYROSINE AMINOTRANSFERASE DEFICIENCY (TAT), OREGON TYPE TYROSINEMIA, RICHNER-HANHART SYNDROME, OCULOCUTANEOUS TYROSINEMIA

Classification:

Inborn Error of Amino Acid Metabolism

Genetic Information:

Inheritance: Autosomal Recessive
Population Incidence: >1:250,000
Ethnic Incidence: Not known
Gene & Location: 16q22.1-q22.3; tyrosine aminotransferase gene
Common Mutation: Not known, 12 mutations known
OMIM # 276600

Disease Information:

Symptom Onset: Usually in early infancy but may present at any age.
Symptoms: Photophobia, lacrimation and intense pain in eyes caused by herpetic-like corneal lesions, usually present in both eyes. Painful hyperkeratotic lesions also appear on the palms of the hands and soles of the feet. These are caused by deposition of tyrosine crystals in the tissues. Neurologic development is highly variable ranging from normal to severely retarded.
Physical Findings: No dysmorphisms
Treatment: Standard tyrosine/phenylalanine restricted diet
Natural History without treatment: Untreated serious eye damage results, including corneal scarring, visual impairment, nystagmus and glaucoma. Skin lesions become permanent and make it difficult and painful to walk.
Natural History with treatment: Eye symptoms are relieved within a week and skin lesions in a few months. These will not reoccur as long as tyrosine levels are kept low. Neurologic problems should be avoided, but the number of patients treated before symptoms occur has been small.

Metabolic Information:

Missing Enzyme & Location: Tyrosine Aminotransferase; Liver
MS/MS profile: Elevated tyrosine (>1200 umol/l). Measurement of tyrosine by MS/MS is nonspecific for tyrosinemias and patients may be missed.
Prenatal testing: Assay of tyrosine aminotransferase in Chorionic villus or amniocytes.

Miscellaneous Information: Diagnosis is confirmed by elevated tyrosine or its metabolites in urine and plasma. Assay of tyrosine aminotransferase is possible through a liver biopsy but is rarely done.

Prepared for the NW Regional Newborn Screening Program by Judi Tuerck, RN MS

References:

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