AMINO ACID DISORDERS

Tyrosinemia Type II

What is Tyrosinemia Type II?

Tyrosinemia Type II is also known as oculocutaneous tyrosinemia or Richner-Hanhart syndrome. The deficient enzyme is tyrosine aminotransferase.[1]

CLINICAL MANIFESTATIONS

The most important manifestation are those involving the eye. which can lead to corneal scarringand permanent visual impairment.[1] Patients report lacrimation, photophobia and eye pain.[2] Cutaneous lesions are painful keratoses which occur particularly on peripheral pressure-bearing areas of the palms andsoles.[1•2•3]

PATHOPHYSIOLOGY

Tyrosine amino transferase normally converts tyrosine to p-hydroxyphenylpyruvic acid which is the rate-limiting step in the metabolism of tyrosine. The increased concentration of tyrosine and its metabolites is postulated to inhibit many transport functionand enzymatic activities.[2]

Inheritance: autosomal recessive [1.2 .3]

CONFIRMATORY TESTING

Confirmation can be done through plasma amino acid levels (increased tyrosine) and urine metabolic screening (increased succinylacetone).[2] Further confirmatory testing may be required after referral to a metabolic specialist.

Overview of Disease Management

The treatment consists of the institution of a diet low in tyrosine and phenylalanine through protein restriction and supple- mentation of a special milk formula.[1•2•3] Initiation of management should be done in consultation with an attending physician/ metabolic specialist.

Prognosis

If left untreated, visual imparement and mental retardation may occur.[1•2•3]

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WHAT TO DO

If unwell and cannot tolerate oral intake:

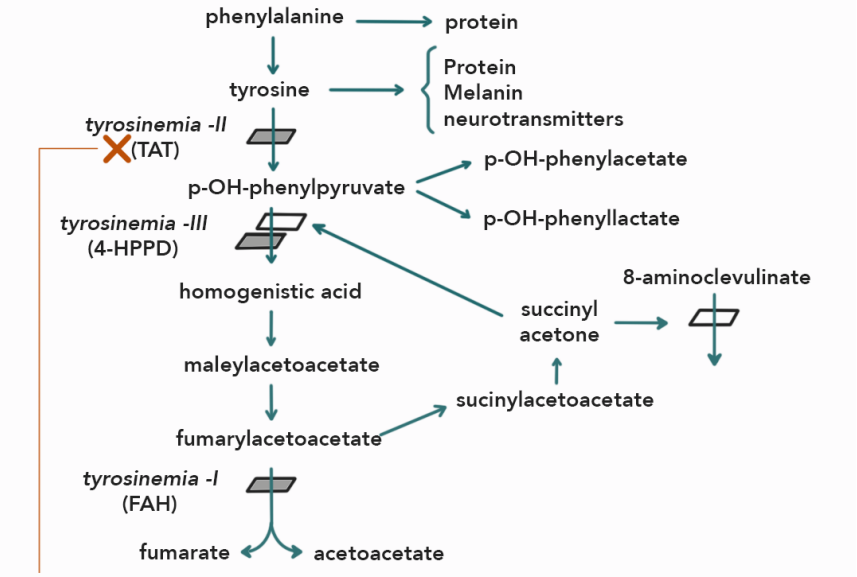
- Nothing per orem
- Ensure patient's airway is secure
- Insert IV access. Collect samples for blood glucose, plasma amino acids, liver function tests and coagulation studies. May request for investigations (i.e., CBC, blood gas, etc) as needed
- May give fluid boluses if patient requires.
- Start D12.55 0.3NaCl at full maintenance. Assess patient clinically, if there is need to increase fluid, may do so up to 1.2 to 1.5x the maintenance
- Monitor input and output strictly (q6 hours).

If unwell and can tolerate oral intake:

- Insert oro- or nasogatric tube and start continuous feeding with TYR milk formula or protein free formula at maintenance rate
- Insert IV access. Collect samples for blood glucose, plasma amino acids, liver function tests and coagulation studies. May request for investigations (i.e., CBC, blood gas, etc) as needed
- Start D12.5% 0.3NaCl at 5-10 cc/hr.
- Monitor input and output strictly (q6 hours)

**Children should not be protein restricted for longer than necessary (24-48 hours)*
**Inform the metabolic doctor on call for further guidance regarding on-going management*
**If the patient is well, coordinate with a metabolic specialist regarding further management*

Tyrosinemia Type II patient's breakdown of protein:



In Tyrosinemia Type II, the deficiency of tyrosine aminotransferace (TAT) causes buildup of tyrosine

[1] *Nyhan WL, Barshop BA and Ozand P. Chapter 20: Oculocutaneous tyrosinemia/tyrosine aminotransferase deficiency. Atlas of Metabolic Diseases 2nd ed. Great Britain:Oxford University Press, 2005 pp 164-170.*
[2] *Javadi MA, Mirhdehghan SA, Bagheri A, Einollahi Band Dowlati Y. Two Cases of Tyrosinemia Type II and its Rare Occurence in Two Brothers. Medical Journal of Islamic Republic of Iran 1996:10(2):169-173*
[3] *Janakiraman I., Sathiyasekeran M., Deenadayalan M., Ganesh Rand Mehesh U. Richner-Hanhart Syndrome. Indian J Pediatr 2006; 73(2):161-162*