13. Tyrosinemia Type I: Clinical, Biochemical and Molecular profile of Indian patients.

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Objective: - Determining clinical, biochemical and molecular profile of Indian children with HT1.

Subjects and method: In this paper we report experience during the period 2000 – 2006. 22 patients clinically suspected to have HT1, on the basis of hepatomegaly, jaundice and cirrhosis were studied. Of these 5 patients were confirmed to have HT1.

Results: - 2 babies presenting before 3 months expired soon after diagnosis and no NTBC could be started. The remaining 3 children received NTBC and Tyrosine restricted diet and are clinically and biochemically better. At the time of presentation mean biochemical parameters were as follows – Pl. SA – 24.05, Pl Tyrosine 700, Pl. Phe, 198, Pl. Methionine 250.5, Urine SA 335.25 and AFP 153950. All 3 – surviving children have shown reduction in AFP, SA, Tyr and Phe levels. Of the two expired children one showed IVS8 – 1g-> c/IVS8-1 g -> c and other one has Q64H /Q64H mutation. Of the 3 – surviving children 2 are Q64H homozygote and in 1 no kown mutation could be identified. However one child homozygous for Q64H presented at 1 – month age expired within next month. Other two children showed late onset and good response to NTBC. One child showed irritability and hyperactivity probably NTBC induced, as dose reduction (from 1 mgm/kg to 0.75 mgm / kg) led to disappearance of these symptoms. This child is also receiving a special diet Tyr. 2 from ComidaMed (Germany).