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EXPERIENCE OF A SINGLE CENTER IN NTBC USE IN MANAGEMENT OF HEREDITARY TYROSINEMIA TYPE I IN LIBYA

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Background: Hereditary tyrosinemia type I (HTI) is a metabolic disease caused by deficiency of fumarylacetoacetate hydrolase enzyme.

Objectives: This study reports beside its clinical and biochemical presentation, the outcome of NTBC [2- (2-nitro-4-trifloro-methylbenzoyl)-1, 3-cyclohexanedion] treatment of the disease and evaluates its biochemical markers in 16 pediatric Libyan patients.

Patients & Methods: The diagnosis was based on presence of high tyrosine levels in blood and succinylacetone in urine.

Results: The consanguinity rate was 81.2%, the median age at onset, at diagnosis and at starting treatment were 4.5, 8, and 9.5 months respectively. At presentation hepatomegaly, jaundice, rickets and high gamma glutamyl transferase (GGT) were observed in 87.5% of patients. All patients had extremely high alpha fetoprotein (AFP) and high alkaline phosphatase (ALP) levels. Fifteen patients were treated with NTBC, normalization of PT (Prothrombin time) was achieved in average of 14 days. The other biochemical parameters of liver function (transaminases, GGT, ALP, bilirubin and albumin) took longer to improve and several months to be normalized. Survival rate with NTBC was 86.6%. Patients who started treatment in a median of 3 months post onset observed a fast drop of AFP in 90.6% of patients (P=0.003). Abnormal liver function and rickets were the common presentations and GGT was an early cholestatic sensitive test. ALP was constantly high even in asymptomatic patients.

Conclusions: In HT1 a faster dropping of AFP is a marker of good prognosis

Biography

Hanna Alobaidy has completed her MB BCh from Faculty of Medicine, Alexandria University, Egypt in 1980 and Postdoctoral studies in Pediatrics from Karolinska Institute Stockholm Sweden, 1990. She is a Consultant of clinical pediatrics and inborn errors of metabolism. She is currently serving as Consultant Metabolist in outpatient clinic Alkhadra Hospital, Tripoli, Libya. She has committed to a career in Academic Medicine and has over 20 years of experience of undergraduate and postgraduate medical education in University of Tripoli Medical College. She has published papers in national and international peer-reviewed medical journals with over 25 citations. Her top clinical researches are: The 1st study and reference report about the "Pattern of metabolic disorders in Libya, long term experience in tyrosinemia type I with NTBC (17years) and Niemann-Pick C disease follow up".

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