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TWO LIBYAN SIBLINGS WITH BETA-KETOTHIOLASE DEFICIENCY: A CASE REPORT AND REVIEW OF LITERATURE

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Beta-ketothiolase (mitochondrial acetoacetyl-CoA thiolase, T2) deficiency is an autosomal recessive disorder characterized by impaired metabolism of ketones and isoleucine. In this study, we report on the first two siblings with T2 deficiency from Libya. Both siblings presented with ketoacidosis, but the severity and outcomes were quite distinctive. T2 deficiency in patient 1, the younger sister, manifested as recurrent severe episodes of ketoacidosis during the first year of life. She unfortunately experienced neurodevelopmental complications and died at 14 months old, after her 5th episode. In contrast, patient 2, the elder brother, experienced only one ketoacidotic episode at the age of 4 years. He recovered uneventfully and has continued to achieve age-appropriate development to date. Upon analysis, the siblings' blood acylcarnitine profiles had shown increased levels of C5:1 and C5-OH carnitine. ACAT1 mutational analysis revealed patient 2 is homozygotic for a novel mutation–c.674C > A (p.Ala225Glu); this mutation was then confirmed by familial analysis. Transient expression analysis of c.674C > A mutant T2 cDNA revealed neither potassium ion-activated acetoacetyl-CoA thiolase activity, which represents T2 activity, nor mutant T2 protein. Therefore, this mutation is truly pathogenic. Interestingly, the incidence of T2 deficiency may be high among the Arab population. This disease should be considered in the differential diagnosis for unexplained ketoacidosis in children. Patients with T2 deficiency could have a favorable outcome if diagnosed and treated early.

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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