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Abetalipoproteinemia: Case series and literature review

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Abetalipoproteinemia (ABL, OMIM 200100) is a very rare metabolic disease with reported prevalence of less than one case per 100,000. It is an autosomal recessive disease resulting from mutations in the gene encoding microsomal triglyceride transfer protein (MTP). Affected patients presented with a wide range of clinical symptoms during infancy. Typical manifestations are failure to thrive, low level of cholesterol and fat malabsorption. Other features like fatty liver, acanthocytosis and anemia usually present. Low fat diet and fat soluble vitamins are the main stay of therapy. Here is a retrospective review of three patients admitted to Salmaniya Medical Complex (SMC) with ABL. We presented the clinical presentations, diagnosis, response to medical therapy and outcome of these three infants along with a literature review about ABL.

Biography

Hasan M Isa has completed his MBBch from Alexandria University and then joined for Arab Board Residency Program in Pediatrics (CABP) in 2003 followed by a Fellowship Program in Pediatric Gastroenterology, Hepatology and Nutrition at The Children's Hospital at Westmead, Sydney, Australia. Since then, he has been serving as a Pediatric Consultant and Gastroenterologist at SMC. He is an Assistant Professor at Arabian Gulf University and has published multiple papers in reputed journals.

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