عنوان مقاله:

موتاسیون جدید در ژن SLC19A2 در آنمی مگالوبلابتیک پاسخ دهنده به تیامین

محل انتشار:

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خلاصه مقاله:

Introduction: The Thiamine Transporter gene SLC19A2 is the only gene known to be associated with TRMA. This syndrome is a trial clinical characterized by megaloblastic anemia, nonautoimmune diabetes mellitus and sensory-neural hearing loss. Methods: Described here are three children from consanguineous Iranian families with thiamine – responsive megaloblastic anemia (TRMA) or Rogers syndrome. Case one and two were siblings of healthy first-cousin parents and case three from a healthy second-cousin couple. These cases presented with hyperglycemia, anemia, and hearing loss. Thiamine reversed the anemia and there was a satisfactory response for the hyperglycemia as well. Results: In all three patients, direct sequencing revealed a homozygous mutation c.38 G> A (P.E.128K) resulting in the substitution of glutamic acid to lysine at position 128 in exon 2 of the SLC19A2 gene on chromosome 1q23.3. This novel mutation was confirmed by the PCR RFLP assay of more than 100 control alleles. Conclusion: TRMA or Rogers syndrome should be considered for patients with diabetes (DM) and other symptoms, including hearing loss and anemia. Early diagnosis can assist families in planning future pregnancies. The .administration of thiamine ameliorates the megaloblastic anemic condition and produces a better response in DM

کلمات کلیدی:

Rogers syndrome, Megaloblastic Anemia