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THIAMINE–RESPONSIVE MEGALOBLASTIC ANEMIA, SENSORINEURAL DEAFNESS AND DIABETES MELLITUS

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Abstract:

Abstract- The syndrome of diabetes mellitus, sensorineural deafness and megaloblastic anemia dose not result from thiamine deficiency. The previous reported patients had no sign of beriberi, had normal nutrition, and had no evidence of malabsorption. The features of this syndrome with apparent inheritance of autosomal recessive trait may define this puzzling syndrome as a true thiamine dependency state. The first Iranian patient was described by Vossough et al. in 1995. We found nine new cases with diagnostic criteria of thiamine responsive megaloblastic anemia during eight years of our study. In two patients, presentation of diabetes and anemia was concomitant. All of them were deaf with sensorineural hearing loss which was detected in infancy up to two years of age. The presence of congenital valvular heart disease was eliminated by normal echocardiography, but cardiomyopathy was discovered in two. Nonspecific amino-aciduria was discovered in three but urinary screening tests for hereditary orotic aciduria were negative. Ox-Phos biochemistry of muscle mitochondria which demonstrates severe defect in complexes I, III, IV in diabetes mellitus associated with deafness, were done but was unremarkable in our patients. Urinary methylmalonic acid and methyl malonyl carnitine by GS/MS and TMS was done in our patients and showed abnormal results in six patients. Thiamine gene, SLC 19A2, was detected in four patients.

Keywords:

Refractory anemia ، thiamine responsive

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