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Letter to the Editor

Genetic Variation Impacts in Patients with Major Beta-Thalassemia

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Dear Editor-in-Chief

Thalassemia is the most common single gene disorder around the world (1, 2). This disease is a type of chronic, microcytic and inherited anemia associated with defect in hemoglobin synthesis and reduction in life span of red blood cells (3). Thalassemia is distributed in Mediterranean, Middle East, Arabian Peninsula, Turkey, Iran, India, Burma and Southeast Asia (1). There are two types of thalassemia including alpha and beta thalassemia (2).

In Iran, beta thalassemia is more common than alpha thalassemia (2). Quality and life span for patients with beta thalassemia has significant progress in recent decades. Nevertheless, a complication of disease such as iron over load is a major problem that cannot be ignored (4).Iron deposition in renal epithelial cells in renal tubes is the main cause of renal damage due to iron over load and hypoxia due to red blood cells hemolysis. Their short life span also is effective in this process because this hypoxia has the most influence in epithelial cells near the renal tubes that more sensitive to oxygen reduction (5).

Some of the renal complications in patients with beta thalassemia major are including increased renal plasma flow, failure of urine concentration, renal tubular acidosis, renal tubulopathy such as hypercalciuria, proteinuria, glocosuria, magnisiuria (4, 6, 7). Glomerular filtration rate (GFR) as an effective factor in evaluation of glomelolar function depends on height, age, weight, race and sex of patients. Some reports showed normal GFR, whereas some of them have reported abnormal GFR (8,9). Therefore, evaluation of renal function in patients with thalassemia is very important approach in different areas.

We evaluated renal function in patients with thalassemia. Like the other studies, our results showed renal dysfunction in patients with beta-thalassemia major. Beside the existence of renal dysfunction, the effect of genetic factors like polymorphisms, in development of renal dysfunction is important and maybe opened the way for diagnosis and prevention of disease. Interesting results on VDR (vitamin D receptor) polymorphism on population in foreign countries (10) encourage us to evaluate this polymorphism in our thalassemia patients. We evaluated routine markers of renal disease and results showed specific markers it is better to check this correlation. Further studies are needed for evaluation of other genetic factors and possible mechanisms involved.

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