ventrculo-peritoneal shunt was inserted to her. She had cellular and humoral immunodeficiency with decreased peripheral blood B and natural killer (NK) cell numbers (C19+20 cell number of 1%) and low immunoglobulin levels. On the follow-up, she received monthly IVIG prophlylaxis and platelet transfusions as needed. Genetic analysis disclosed that a heterozygous missense variant in SAMD9L (c.2627T > C). Bone marrow aspiration was planned to done in every 3 months on the follow-up. Platelet count and hemoglobin levels gradually increased over the time, but monosomy 7 was positive at the age of 2 in the 52% of the cells. She underwent hematopoietic stem cell transplantation (HSCT) from a matched unrelated donor with myeloablative conditioning regimen.

Methodology: We herein report a girl presenting with pancytopenia and immunodeficiency which was revealed SAMD9L mutation.

Results: SAMD9L, the gene is located in a region of chromosome 7 that is commonly deleted in myeloid malignancies. In mice, Samd9l deficiency causes development of MDS with age, suggesting that SAMD9L is a tumor suppressor. Heterozygous SAMD9L missense mutations may cause of familial MDS like Ataxia-pancytopenia syndrome which is associated with neurological findings (ataxia and nystagmus), cytopenias and predisposition to myeloid leukemia involving −7/del(7q). In addition, SAMD9L may regulate differentiation of diverse immune cell lineages like B and NK cells, however cellular basis of neurological manifestations in the carriers remains unclear.

Conclusion: In conclusion, SAMD9L mutation screening should be considered in all pediatric patients with MDS, AML, or JMML with chromosome 7 aberrations, even in the absence of neurological symptoms or a family history of myeloid malignancies.

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RED BLOOD CELL DISORDERS

OP 24

The effects of vitamin D deficiency on myocardial deformation and functions in patients with β-thalassemia

A. Koca Yozgat[1,*], E. Azak[2], D. Kaçar[1], M. Işık[1], O. Arman Bilir[1], I. Çetin[2], N. Ozbek[1], N. Yaralı[1]

[1] *Ankara City Hospital, Pediatric Hematology, Ankara, Turkey*
[2] *Ankara City Hospital, Pediatric Cardiology, Ankara, Turkey*

Objective: β-Thalassemia major (TM) is an inherited hemoglobin disorder resulting in chronic hemolytic anemia, and regular lifelong transfusion therapy remains the mainstay in the treatment of patients. Cardiac involvement is the leading cause of death in patients with β-TM. The association between vitamin D deficiency and left ventricular systolic and diastolic dysfunction has been previously demonstrated in the literature. Speckle-tracking echocardiography (STE) is feasible and valid for the evaluation of cardiac function via an assessment of the longitudinal deformation of the myocardium through the cardiac cycle. Our study aims to evaluate the effect of vitamin D deficiency on myocardial deformation and functions in children with thalassemia major by STE.

Methodology: In this prospective study, 33 patients with β-TM, receiving regular blood transfusions, and undergoing iron chelation therapy were enrolled in April 2018-January 2020. Vitamin D and ferritin levels, cardiac magnetic resonance (MR) T2* value, conventional echocardiography, and speckle tracking were evaluated. LV regional circumferential, and longitudinal strain values were measured. Vitamin D levels considered <20 ng/ml, 20–30 ng/ml, >30 ng/ml as deficient, insufficient, and sufficient, respectively. Myocardial functions of patients with vitamin D deficiency or insufficiency were evaluated by STE before and after vitamin D replacement.

Results: The mean age of patients was 15.4 ± 3.09 years; the male/female ratio was 18/15, and mean ferritin levels were 2017 ± 1573 ng/ml. Vitamin D level deficiency was detected in 30 (90%) and insufficient in 3 (10%) of our patients. Cardiac T2* value was normal in 21 patients and 12 patients had iron accumulation on cardiac T2* MR. The mean of left ventricular ejection fraction (LVEF) was 64 ± 4.7%, and the mean left ventricular shortening fraction (LVSF) was 34.2 ± 3.8% before vitamin D replacement, and LVEF was 65.1 ± 5.2% and LVSF 35 ± 3.7% after vitamin D replacement ($p > 0.05$). The mean left ventricle global longitudinal strain (LVGLS) was 19 ± 2.7% before replacement and 24 ± 2.7% after replacement (p: 0.04). The left ventricle global circumferential strain (LVGCS) was 20 ± 2.8% before replacement and 25 ± 3.8% after replacement (p: 0.03). While there was no significant difference in right ventricular functions before and after vitamin D replacement, but a statistically significant increase was observed in parameters showing left ventricular diastolic functions after replacement. There was a significant improvement in the global longitudinal strain of left ventricular after vitamin D replacement.

Conclusion: Vitamin D deficiency is frequently observed in patients with β-TM. It is reported that vitamin D deficiency causes decreased contractility and leads to an increase in cardiac iron involvement accordingly cardiomyopathy in these patients. Speckle tracking echocardiography could be used as a feasible method for evaluating subclinical myocardial dysfunction in patients with β-TM. In patients with β-TM, diastolic functions are primarily affected in the case of cardiac toxicity. In our study, we observed that our patients' diastolic functions had improved after vitamin D replacement therapy.

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OP 25

The molecular spectrum of patients with hereditary spherocytosis: a single center experience

C. Coskun*, S. Unal, F. Gumruk

Hacettepe University, Department of Pediatric Hematology, Ankara, Turkey

Objective: Hereditary spherocytosis (HS) is a hemolytic anemia with variably severity, caused by defects in the