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

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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

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

components of red cell membranes. It is characterized by anemia, jaundice, splenomegaly and cholelithiasis. The clinical manifestations vary widely, ranging from nearly asymptomatic to transfusion-dependent or severe life-threatening anemia. It is difficult to identify atypical cases with classical approaches. The known HS gene mutations are SPTA1 gene, SPTB gene, ANK1 gene, SLC4A1 gene and EPB42 gene. In this report, the next-generation sequencing (NGS) was used to analyze our patients with HS and we identified mutations responsible for HS.

Methodology: Patients who were diagnosed with hereditary spherocytosis with osmotic fragility testing between 2007–2019; ten were further tested for molecular background. Diagnosed in our center were analyzed retrospectively. Either NGS or ANK1 Sanger testing were used.

Results: The 10 cases of HS comprised 8 males and 2 females. The age of patients ranged from 5 months to 17 years. Hemolytic anemia, jaundice and splenomegaly were the most common findings in our cases. Gallstones were detected in four patients (40%). The family history was positive in 5 (50%) patients. Splenectomy and cholecystectomy was performed in two cases and three cases, respectively. The results corfirmed ANK1 gene mutation in 50%; SPTB gene mutation in 20%, EBP42 gene mutation in 10%; SPTA1 gene mutation in 10%. The clinical features of the patients are summarized in the Table 1. Table 1. Patient Age Sex Age of diagnosis Family history Splenomegaly Gallstone Splenectomy/Cholecystectomy Mutated gene 1 1 Female 1 year Yes + – –/– SPTB 2 10 Female 10 years Yes + – +/– ANK1 3 12 Female 7 years Yes + + +/+ ANK1 4 12 Female 2 years Yes – + –/+ ANK1 5 10 Male 6 years No – + –/– ABCG8 6 2 Female 5 months No + – –/– ANK1 7 13 Female 15 years Yes + – –/– ANK1 8 8 Female 7 years No + + –/+ EBP42 9 2.5 Male 2 years No + – –/– SPTB 10 19 Female 17 years No + – –/– SPTA1.

Conclusion: Consistent with the literature, the most common gene mutated was ANK1. Collectively, our results suggest that mutation analyses will complement other conventional tests for accurate diagnosis of HS, especially in those who are under transfusion programme and are followed with a diagnosis of unspecified hemolytic anemia.

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STEM CELL TRANSPLANTATION

OP 26

Eltrombopag for thrombocytopenia following pediatric allogeneic hematopoietic stem cell transplantation

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Objective: Failure of platelet recovery is a complication occurring after allogeneic hematopoietic stem cell transplantation (HSCT). Poor graft function, relapse, viral infections, drug toxicity, immune processes may lead to decreased platelets production. Treatment options are limited for thrombocytopenia caused by poor platelet production. While the use of Eltrombopag (ELT) was retrospectively investigated in adult patients, data regarding the potential benefit of these agent for pediatric posttransplant thrombocytopenia are lacking. We report three pediatric patients who received ELT for thrombocytopenia occurring after HSCT.

Case report: The median patients' age at HSCT was 13.3 years (10–18). All patients had HSCT from a sibling donor with the bone marrow stem cell source. All patients were treated with a myeloablative conditioning regimen. Patients engrafted at a median time of 19 days (10–24) for neutrophils and 49 days (44–49) for platelets. Bone marrow aspirates showed a decrease number of megakaryocytes, and all patients had been ineffectively treated with high-dose intravenous gamma globulin and with steroids before ELT initiation.

Methodology: ELT was started at a median time after HSCT of 57 days (42–90), the starting dose being 25 mg/day, and the maximum administered dose was 75 mg/day. ELT was continued for a median period of 64 days (28–286). All patients reached sustained platelets count >50,000/μL after a median time from starting ELT of 197 days (87–210). The median platelet count at last evaluation was 115,000/μL (range 66,000–125,000/μL). ELT was well tolerated, and no patient have developed important side effect.

Results: Our cases became transfusion independent after a median time from starting ELT of 197 days. In the pediatric post-HSCT setting, only few previously published case reports described the successful use of ELT as a treatment for thrombocytopenia. Li et al. reported three children transplanted for nonmalignant disease treated for both primary and secondary failure of platelet engraftment. Treatment was effective in two patients, but not in one patient transplanted for Gaucher disease. In Masettis' study, the 60-day cumulative incidence of platelet recovery >50,000/μL after ELT treatment was 75%. Similarly, Tanaka et al. described 12 adults treated for primary and secondary post-HSCT thrombocytopenia who reached platelet count>50,000/μL in 60% and 71% of cases respectively.

Conclusion: Our study supports the safety and efficacy of ELT for treatment of prolonged thrombocytopenia after allogeneic HSCT in children. Future prospective studies are needed to confirm these findings.

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