

diagnosed with JMML have somatic, activating RAS mutations. Response to steroids/IVIG in our patient prompted RALD diagnosis, rather than JMML. Finally genetic analysis of the RAS mutation should be performed in cases that does not meet the defined diagnostic criteria of ALPS or JMML.

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LEUKEMIA/LYMPHOMA/HISTIOCYTE DISORDERS

OP 22

Bone mineral density and bone resorption in the acute leukemia during childhood

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Objective: Acute leukemia is the most common malignancy in children and has been reported to be associated with low bone mass. The urinary cross-links lysyl-pyridinoline (dipyridinoline [Dpd]) are established biochemical markers of osteoclastic bone resorption and collagen degradation. We believe that acute leukemia treatment; we wanted to investigate the effect on bone mineral density (BMD, g/cm²) and bone resorption. It has been asked to investigate whether this effect is continuing or not with the passing years.

Methodology: Our materials were 29 leukemia patients who completed their treatments. The patients were divided into two groups. Group I consisted of 19 patients (the ones in the 1.00 ± 0.15 th months after treatment) and Group II consisted of 10 patients (the ones in the 43.36 ± 18.39 th month). 52 healthy children formed BMD group and 20 children formed Dpd control group. The BMD and urine Dpd values of the healthy ones and the patients were measured.

Results: In 10 of total 29 cases (4.48) osteopeni and osteoporosis were determined. A meaningful difference could not be found in the average values of BMD between the groups. In the evaluation of all cases and the groups separately, any effect of the chemotherapy could not be found on BMD. It was found that age had a meaningful effect on BMD in the Group I ($p < 0.05$). The age and the time after the treatment affected BMD in a meaningful level in Group II ($p < 0.00001$, $p < 0.05$, respectively). BMD was increasing significantly with age and interval. The average BMD of 29 cases was 0.66 ± 0.17 g/cm², while of control group was 0.65 ± 0.16 g/cm². Average Dpd levels in urine were 32.92 ± 13.74 and 30.15 ± 13.48 nmol/mmol Cr in the patients and control group respectively. The average BMD and Dpd values of the patients were not different than of the control group. A meaningful negative relation was determined between BMD and Dpd values separately in both all cases and Group II. Dpd value in urine decreased with the increase in the value of BMD. As the age of diagnosis increased, BMD increased. When the age of diagnosis increased, Dpd was determined as decreased. In the evaluation of all cases and groups separately, bone resorption and BMD were not different

between the one taking radiotherapy (0.60 ± 0.15 g/cm² and 31.29 ± 18.09 nmol/mmol Cr) and the one not taking radiotherapy (0.67 ± 0.20 g/cm² and 37.29 ± 11.91 nmol/mmol Cr). In Group I, there was a meaningful difference ($p < 0.05$) between Bsds of the patients taking cranial radiotherapy (1.04 ± 0.74) and the ones not taking cranial radiotherapy (-0.19 ± 0.80) and taking extracranial radiotherapy (-1.36 ± 0.93). Cranial radiotherapy effected Bsds negatively in Group I while this effect could not be seen in Group II.

Conclusion: It was concluded that the childrens completing acute leukemia treatment could reach carry out the ideal height and weight with a sufficient and balanced nutrition program and maintain BMD values proper to their ages.

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LEUKEMIA/LYMPHOMA/HISTIOCYTE DISORDERS

OP 23

A girl with SAMD9L mutation presenting with pancytopenia, immunodeficiency and myelodysplasia

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Objective: Several monogenic causes of familial myelodysplastic syndrome (MDS) have recently been identified. Genetic studies disclosed heterozygous missense mutations in SAMD9L, a tumor suppressor gene located on chromosome arm 7q. Consistent with a gain-of-function effect, ectopic expression of the 2 identified SAMD9L mutants decreased cell proliferation relative to wild-type protein.

Case report: A one month old girl was referred to our hospital with bruising. She was followed-up at a local hospital with thrombocytopenia for three weeks. She had normal physical examination findings except petechiae on her extremities, trunk, and face. There was no bleeding diathesis and consanguineous marriage in her family history. Complete blood count showed hemoglobin of 7.2 g/dL, reticulocyte of 2.4%, leukocyte count of 3.1×10^9 /L, absolute neutrophil count of 0.3×10^9 /L, platelet count of 2×10^9 /L. Coagulation tests, liver and kidney functions were normal. Her viral serologies were negative for EBV, CMV, rubella, hepatitis and parovirus B19. However, vitamin B12 level was below normal limits, then cyanocobalamin treatment was started. Her mother's serum vitamin B12 level was normal. Immune thrombocytopenia was considered and intravenous immunoglobulin (IVIG) was given to her, and platelets raised to 87×10^9 /L, thereafter decreased to 14×10^9 /L within a few days. Bone marrow aspiration showed hypocellularity with dysplastic changes in myeloid lineage. Karyotype analysis revealed 46,XX der(20), and negative for monosomy 7. Her neurologic examination was normal except bulging of anterior fontanel, cranial ultrasonography was performed and it showed triventricular hydrocephalus and left cerebellar hypoplasia. A

ventriculo-peritoneal shunt was inserted to her. She had cellular and humoral immunodeficiency with decreased peripheral blood B and natural killer (NK) cell numbers (CD19+20 cell number of 1%) and low immunoglobulin levels. On the follow-up, she received monthly IVIG prophylaxis and platelet transfusions as needed. Genetic analysis disclosed that a heterozygous missense variant in SAMD9L (c.2627T>C). Bone marrow aspiration was planned to be 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 $64 \pm 4.7\%$, and the mean left ventricular shortening fraction (LVSF) was $34.2 \pm 3.8\%$ before vitamin D replacement, and LVEF was $65.1 \pm 5.2\%$ and LVSF $35 \pm 3.7\%$ after vitamin D replacement ($p > 0.05$). The mean left ventricle global longitudinal strain (LVGLS) was $19 \pm 2.7\%$ before replacement and $24 \pm 2.7\%$ after replacement ($p: 0.04$). The left ventricle global circumferential strain (LVGCS) was $20 \pm 2.8\%$ before replacement and $25 \pm 3.8\%$ 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

