Patient	Gen	Nucleotide Protein Change	Zygosis	dbSNP	Effect	Variant Classification	Disease (Dominance, OMIM#)
1	ANK1	NM_001142446.2:c.747 C>G p.Tyr249*	het	-	stop gain	-	Sferocytosis type 1; AD:18900
1	SPTB	NM_001024858.3:c.4891 C>T p.Arg1631Cys	het	rs372503030	nonsynonymous-SNV	VUS	Spectrin Beta Erythrocytic; 182870)
2	SPTB	NM_001024858.3:c.4 355C>T p.Ala1452Val	het	rs768609633	nonsynonymous-SNV	VUS	Spectrin Beta Eritrocytic; 182870

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Pediatric Hematology Abstract Categories

Immunodeficiencies / Neutrophil Diseases
OP 20

A NEXT-GENERATION SEQUENCING TEST FOR CONGENITAL NEUTROPENIA IN PEDIATRIC PATIENTS

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Objective: Congenital neutropenia (CN) is a rare inherited hematological disease and its phenotypic, histologic and molecular aspects are heterogeneous. Congenital neutropenia can manifest as isolated neutropenia or neutropenia with extra-hematopoietic abnormalities, immunodeficiency or metabolic diseases and results in recurrent, life-threatening bacterial infections. Mutations in more than 20 genes have been demonstrated to cause CN, some of which cause complex phenotypes. **Case report:** Usually caused by ELANE mutations although mutations in other genes like HAX-1, G6PC3, and GF11 have also been reported. Identifying the causative mutation aids in the diagnosis and ruling out other secondary causes of neutropenia. In this study we aimed to identify the molecular defects in CN patients by next generation sequencing (NGS). **Methodology:** Next generation sequencing test was performed on peripheral blood specimens of 17 patients diagnosed with congenital or chronic neutropenia and bone marrow failure and hematological malignancy ruled out from January 2021- June 2023. The genes in the NGS panel were; LAMTOR2, CLPB, HAX1, USB1, SBDS, JAGN1, TAZ, ELANE, G6PC3, WAS, CXCR4, GFI1, VPS45, VPS13B. **Results:** The median age at presentation of neutropenia was 28.9 months. Mean neutrophil count at diagnosis was 380± 259/mm3. Bone marrow aspiration was performed in ten patients and myeloid maturation arrest was observed five. Granulocyte colony stimulating factor was given for seven patients and all had a response. In the NGS panel TAZ mutation was detected in one patient compatible with Barth Syndrome and VPS13 double heterozygous mutation was detected in one patient compatible with Cohen Syndrome. **Conclusion:** Considering the heterogeneity of CN in terms of genotypes and phenotypes expanded next generation sequencing panel would be necessary. The early onset of the disease, clinical severity and associated high risk of malignant transformation in CN strongly suggests the need for early diagnosis and therapeutic intervention.

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Pediatric Hematology Abstract Categories

Leukemia
OP 21

BK-VIRUS INFECTIONS IN PEDIATRIC LEUKEMIA PATIENTS DURING LEUKEMIA TREATMENT

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Objective: Polyoma BK virus (BKV) infection/reactivation is an important underlying condition that provokes hemorrhagic cystitis (HC) in hematopoietic stem cell transplantation (HSCT) recipients. However, BKV associated infections can rarely occur in acute leukemia patients without receiving HSCT. Here we present 12 pediatric acute leukemia patients with BKV infection during leukemia treatment. **Methodology:** Between September 2019 and July 2023, in Ankara Bilkent City Hospital, BKV by quantitative polymerase chain reaction (PCR) detected in the urine of 12 pediatric leukemia patients who had not got HSCT but receiving intensive chemotherapy. The clinical characteristics of these infections were retrospectively evaluated. **Results:** Ten of the 12 patients had acute lymphoblastic leukemia (ALL). Seven of the 10 ALL cases were T cell ALL. Ten of the patients were male and 10 of the patients' age were 10 years and older. Eleven patients experienced HC and one patient had epididymitis. The copy number of BKV varied between 470 to 1.3 trillion /mL. Seven patients had got treatment ranging from hydration, ciprofloxacin to bladder irrigation. Except a refractory T cell ALL patient, all of the patients had clinical improvement. **Conclusion:** Although it is a major complication of HSCT and solid organ transplantation, BK virus infection can also occur in pediatric acute leukemia patients during treatment. As in HSCT recipients, male gender and older age seems as risk factors in leukemia patients. Due to complete loss of virus specific T lymphocytes, T cell ALL patients may be more prone BK virus activation.

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