mitochondrial transfer RNAs. Autosomal recessive loss of function mutations of TRNT1 leads sideroblastic anemia, immunodeficiency, fevers and developmental delay at varying degrees. Here we present a 10-year-old girl with periodic fever, retinitis pigmentosa, B cell deficiency, seizures and transfusion free sideroblastic anemia due to compound heterozygote TRNT1 mutation.

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PEDIATRIC ONCOLOGY ABSTRACT CATEGORIES

LYMPHOMAS

OP 34

BURKITT LYMPHOMA PRESENTING WITH EYE AND KIDNEY INVOLVEMENT: CASE REPORT

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Case report: Burkitt lymphoma (BL) is an aggressive form of B-cell non-Hodgkin lymphoma. It may present with a variety of symptoms leading to possible misdiagnosis and delay in treatment. BL is fatal if left untreated, and early diagnosis and treatment can improve prognosis. In this case report, a 3.5-year-old male patient with no known disease had left eyelid swelling and hematuria, and orbital magnetic resonance imaging performed after his admission showed contrast enhancement in the bulbus oculi, and increased uptake in both kidneys (suvmax:9.5) in positron emission tomography. The patient's bone marrow aspiration was normal. There was no involvement in the evaluation of the central nervous system. As a result of kidney biopsy, he was diagnosed with high-grade B-cell lymphoproliferative disease (Ki-67 95-100%, diffuse positivity with CD79a and EBV). Burkitt lymphoma. The treatment of the patient was started in the NHL-BFM 2012 R4 arm. At the end of the treatment, the ocular findings regressed. Burkitt lymphoma may present with different clinical presentations. If appropriate and rapid imaging techniques are used, positive results on survival can be obtained. Our patient is being followed up alive and well.

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BRAIN TUMOURS

OP 35

NECESSITY FOR A CUSTOMIZED NGS PANEL FOR ACCURATE DIAGNOSIS AND TARGETED THERAPIES IN PEDIATRIC GLIAL TUMORS

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Objective: Pediatric glial tumors comprise wide range pathologies which may mimic histomorphological features of each other's but generally have very diverse disease course. WHO Classification of Tumors of Central Nervous System (2016 and 2021) points to the necessity of investigating several molecular alterations for integrated pathological diagnosis of childhood CNS tumors. This makes customized next-generation sequencing (NGS) a powerful tool for the diagnosis of childhood CNS tumors. **Methodology:** Acıbadem Molecular Pathology Brain Tumor NGS Panel was designed according to targeted deep RNA and DNA sequencing. RNA and DNA were isolated from paraffin blocks containing more than 50% tumor in 45 cases with childhood CNS tumors. Miniseq Sequencıng System, Illumına and Archer Analysıs Ver 6.0.3.2 platforms were used. Fusions (translocations), mutations, and DNA copy number changes in 81 genes were screened for the most common molecular alterations in CNS tumors. **Results:** Fourty-five childhood CNS tumors were evaluated with NGS results. Among these there were 19 pilocytic astrocytomas, 1 case of high grade astrocytoma with piloid features, 4 diffuse leptomeningeal glioneuronal tumors, 1 pleomorphic xanthoastrocytoma, 4 pediatric diffuse glial tumors, 1 infantile hemispheric astrocytoma, 1 astroblastoma, 12 diffuse midline glioma. Sixteen of these tumors were able to be diagnosed based on these molecular findings. Thirty-four cases received targeted therapies. **Conclusion:** The customized NGS panel, as a single molecular workflow is very helpful and supportive in diagnosis for CNS childhood tumors. Since the number of driver mutations are few in childhood tumors, detection of the driver molecular alteration is guiding the medical treatment startegy in terms of targeted regimens.

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OP 36

CONSTITUTIONAL MISSMATCH DEFECT REPAIR DISORDER (CMMRD) IN PEDIATRIC HIGH GRADE GLIOMA

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