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.

https://doi.org/10.1016/j.htct.2021.10.1000

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.

https://doi.org/10.1016/j.htct.2021.10.1001

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.

https://doi.org/10.1016/j.htct.2021.10.1002

OP 36

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

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Objective: Pediatric high grade gliomas(HGG) have dismal prognosis with median survival of 9-15 months after standard radiochemptherapy. Recent molecular investigations revealed a mismatch repair defect called Constitutional Mismatch Repair Deficiency (CMMRD), which induce pediatric HGG. In CMMRD, there are mutations at least one of the mismatch repair(MMR) genes in both tumoral and non-tumoral DNA. Patients generally have cafe au lait spots resembling the ones in NF-1. **Methodology:** Forty-four pediatric high-grade glioma cases operated in our clinic between 2015-2021 were included in the study. PMS2, MLH1, MSH6, MSH2 immunohistochemical antibodies were applied to the sections prepared from paraffin blocks with tumors of these 44 cases. Next generation Sequencing (NGS) Custom Panel for Brain Tumors was performed with DNA and RNA obtained from neoplastic tissue of 2 cases and germline NGS analysis was performed with DNA obtained from peripheral blood in 1 case. **Results:** MMR protein expression loss was detected in 11 (25%) cases. In 5 (45%) of these 11 cases, MMR protein loss was detected in both neoplastic and non-neoplastic tissue, and these cases were considered as CMMRD. NGS performed in 2 of these 5 cases revealed a hypermutant profile. At least one MMR protein loss was found only in the neoplastic tissue in 6 (55%) of 11 cases, and PMS2 deficiency was the most common. In 1 of these 6 cases, MSH6 deficiency was shown as germline by NGS. **Conclusion:** CMMRD and MMRD, are disorders with close relationship with pediatric high grade gliomas. Since CMMRD cases also may have cafe au lait spots, they should not be misdiagnosed as NF 1. Temozolomide induce more aggressive tumors in CMMRD ve MMRD, therefore its use is not suggested in those cases. Preliminary literature data advocate use of immunotherapy instead. All pediatric HGG cases should be evaluated for CMMRD and MMRD with molecular investigations to understand their biology.

https://doi.org/10.1016/j.htct.2021.10.1003

OP 37

IS METHYLATION STATUS SUBGROUPING REALLY A STRONG PROGNOSTIC FACTOR IN PEDIATRIC POSTERIOR FOSSA EPENDYMOMA?

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Objective: The effective treatment of posterior fossa ependymomas is surgery followed by radio-chemotherapy. Our aim is to evaluate the effects of sex, age, methylation subgrouping, extent of resection, radiation treatment (RT), MIB-1 index, grade, ATRX and H3K27M mutations on prognosis in pediatric patients with posterior fossa ependymoma (PFE). **Methodology:** This is a retrospective study. Forty-two children with PFE who had surgery in our institution between 1996 and 2018 were included. Formalin-fixed paraffin-embedded tumor samples were evaluated for H3K27me3 immunostaining, MIB-1 index, WHO grades, ATRX and H3K27M mutations.Samples with global H3K27me3 reduction were grouped as posterior fossa ependymoma group A (PFA), whereas tumor samples with H3K27me3 nuclear immunopositivity were grouped as posterior fossa ependymoma group B (PFB). **Results:** Mean age of patients was 4.4 years (range 0.71-14.51). Thirty-one patients (73.8%) were PFA, whereas 11 patients (26.2%) were PFB. WHO grades of PFAs were statistically higher in comparison to WHO grades of PFBs. There are no significant differences between PFAs and PFBs in terms of resection rates, disease recurrence and survival parameters.Patients with total surgical excisions had significantly better PFS and OS rates. **Conclusion:** Extent of surgical excision is the most important prognostic indicator in PFEs. Prognostic effect of methylation subgrouping may be minimized with more aggressive surgical strategy in PFAs.

https://doi.org/10.1016/j.htct.2021.10.1004

NEUROBLASTOMA

OP 38

NEUROBLASTOMA IN A CASE OF CONGENITAL ADRENAL HYPERPLASIA

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Case report: The majority of neuroblastomas are sporadic and not correlated with any specific constitutional germline chromosomal abnormality, inherited predisposition, or associated congenital anomalies. We report here a 1.5-year-old girl with a diagnosis of 21 hydroxylase deficiency and neuroblastoma. Neuroblastoma in a known case of congenital adrenal hyperplasia has rarely been reported. Based on our literature review, this is the fifth case report of congenital adrenal hyperplasia and neuroblastoma.

https://doi.org/10.1016/j.htct.2021.10.1005

BONE TUMOURS

OP 39

CAN SERUM KL-6 LEVEL BE USED AS A MARKER IN LUNG METASTASIS OF BONE SARCOMAS?

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