The goal is to report our center experience treating a 12-year old child with unresectable optochiasmatic pilocytic astrocytoma using BRAF V600E inhibitor monotherapy.

Low grade gliomas (LGGs) are the most frequent pediatric tumors of the central nervous system. The outcome of the treated children is satisfactory, albeit at the expense of the poor quality of life due to neurologic impairment linked to the tumor localization and multimodal oncologic treatment, including chemotherapy application. Cytostatic agents are used with the aim of achieving better disease control and increasing survival.

In the modern era of pediatric oncology detecting BRAF V600E mutation in histopathological samples has become a standard procedure which enables BRAF inhibitor therapy commencement. LGGs are stable monogenetic tumors characterized by activated MAPK pathway during all the phases of the illness making these drugs an attractive therapeutic option in relapsed disease.

Taking into account tumor localization, in our patient, only partial surgical reduction of the tumor mass was feasible and followed by chemotherapy administration. Unfortunately disease relapse ensued requiring new neurosurgical procedure, again without the possibility of achieving a complete resection. After BRAF V600E mutation has been found in resected material, a compassionate use of dabrafenib has been initiated.

In our patient oral BRAF inhibitor monotherapy of incompletely resected low grade glioma was easy to implement, with good medication tolerance, resulting in satisfactory disease control and good quality of life.

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ACUTE LYMPHOBLASTIC LEUKEMIA IN CHILDREN: 5-YEAR EXPERIENCE OF A SINGLE NATIONAL PEDIATRIC ONCOLOGY CENTRE

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10.1136/archdischild-2021-europaediatrics.306

Study aimed to analyze epidemiological and clinical characteristics, hematologic, immunophenotypic and cytogenetic parameters of paediatric acute lymphoblastic leukemia cases, aged 1 year and above. Acute toxicities and treatment responsiveness were also studied.

Medical records of all children treated for acute lymphoblastic leukemia at the Department of Oncology and Haematology in Children's Hospital Zagreb from beginning of January 2015 to the end of December 2019 were reviewed.

Infants aged less than a year were excluded.

: In five-year period 28 children were treated for acute lymphoblastic leukemia, of whom 2 infants. Most cases of ALL were diagnosed in 2018 (N=10), and the least in the previous year (N=2). Out of 26 patients 16 were female (61.54%), average age 5.25 years. Median number of leukocytes at the time of diagnosis was $35.36 \times 10^9/L$, hemoglobin level 83.96 g/L and platelet count $120.42 \times 10^9/L$. The most represented immunophenotype was „common’

(73.07%), T phenotype present in only one child. 26.92% of blood marrow samples were hyperdiploid by cytogenetic testing and in two samples hypodiploidy was verified. One child had bcr/abl positive (Ph+) disease.

All 26 children were treated according to ALL IC-BFM 2009 protocol, 12 (71.06%) belonging to intermediate risk group, while standard and high-risk groups were evenly

represented (N=7). 25 children achieved early complete remission (by the end of induction), 3 were eligible for allogeneic stem cell transplant (1 Ph+, 1 resistant, 1 relapse), that was performed accordingly. While all children were faced with febrile neutropenia at some point of the treatment, 7 (26.92%) were treated for microbiologically proven sepsis. Four patients were diagnosed with clinically-radiologically verified pulmonary fungal disease. Acute neurotoxicity was observed in 7 children (seizures, ischemia, PRES). Thromboembolic events were registered in 4 (15.38%) patients. Two children (7.69%) died during intensive chemotherapy; 2-year old boy in induction therapy due to septic shock, pancreatitis and multiorgan failure; 1.5-year old girl due to the same reasons at the beginning of re-induction. No further deaths were observed; all 24 children are alive and in remission.

Acute lymphoblastic leukemia is the most common childhood malignancy, occurring in early age, predominantly of „common’ immunophenotype and most frequently classified as intermediate risk. Serious adverse events during intensive chemotherapy are frequent but with adequate management usually successfully resolved. Survival rates are excellent and have reached more than 90% in developed countries. Our results are completely comparable with literature data.

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HEPATIC SINUSOIDAL OBSTRUCTION SYNDROME IN CHILDREN TREATED FOR SOLID MALIGNANT TUMORS – DIAGNOSIS AND TREATMENT

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10.1136/archdischild-2021-europaediatrics.307

The aim of this report is to describe our center's experience regarding diagnosis and treatment of hepatic sinusoidal obstruction syndrome (SOS) in children treated for malignant solid tumor.

retrospective analysis of hospital records in 2014-2021 period was conducted. Data on patients treated for malignant solid tumor who developed hepatic SOS were extracted and additionally analyzed, with the focus on the type and staging of the tumor, oncologic treatment undertaken, SOS diagnostic criteria and therapy.

a total of 5 patients (1 boy, 4 girls), aged between 19 months and 8 years, developed hepatic SOS. Two patients suffered from nephroblastoma and one from nephroblastomatosis. Two patients (Ewing sarcoma, high risk neuroblastoma) developed SOS as a complication of autologous hematopoietic stem cell transplantation, during which both received abundant supportive care.

All patients had hepatomegaly, ascites, weight gain (>5%) and thrombocytopenia. Hyperbilirubinemia was registered in only one patient.

Clinical criteria were supported by radiological investigations, mainly abdominal ultrasound with Doppler. In one patient liver biopsy was performed due to the suspicion of metastatic liver disease.

Risk factors identified among our patients were cytostatic agents (actinomycin-D, busulfan), previous radiation therapy and supportive care (total parenteral nutrition, antifungal

medications such as liposomal amphotericin B, intravenous immunoglobulin therapy).

Pharmacological treatment included various diuretics, low-molecular-weight heparin, correction of coagulation abnormalities, thrombocyte transfusions, albumin supplementation, ursodeoxycholic acid, acetylcysteine and defibrotide. Fluid balance was carefully monitored. The only patient who received defibrotide had faster clinical recovery and normalization of laboratory parameters. In three patients abdominal drainage and in two patients thoracic drainage had to be performed.

hepatic sinusoidal obstruction syndrome is a severe complication of oncologic treatment with possible lethal outcome requiring rapid diagnosis and intensive treatment.

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ADMINISTRATION OF TARGETED THERAPY IN CHILDREN WITH NTRK REARRANGED MESENCHYMAL NEOPLASMS

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10.1136/archdischild-2021-europaediatrics.308

The aim of this report is to present 2 patients diagnosed with neoplasm of mesenchymal origin, in whom NTRK inhibitor entrectinib was successfully administered.

The first patient presented at the age of 9 months as recurrent chalazion.

During subsequent clinical and radiological follow-up a diagnosis of right upper eyelid tumor was made. Taking into account tumor localization, surgical procedure of a maximum possible tumor reduction was performed, unfortunately with positive resection margins. Histopathological analysis confirmed the diagnosis of „NTRK-rearranged spindle cell neoplasm“ according to the new 2020 WHO classification of soft tissue tumors. The neoplasm was characterized by increased cellularity and relatively low mitotic activity. The possibility of an aggressive clinical course could not be excluded. Molecular analysis (FusionPlex Sarcoma Kit SAR6) verified an LMNA-NTRK fusion and oral entrectinib therapy was initiated. The patient has tolerated the medication very well and is without the radiological signs of eventual local relapse (magnetic resonance imaging).

The second patient is a 12-year-old boy presenting with a solid expansive lesion of the lesser pelvis extending towards the groin and anterior abdominal wall (locoregional disease). Tumor tissue sampling confirmed malignant peripheral nerve sheath tumor. Neoadjuvant systemic chemotherapy according to the EpSSG-NRSTS protocol and radiotherapy accomplished only partial reduction of the tumor mass prompting next generation sequencing on formalin-fixed paraffin-embedded tissue blocks (FoundationOne®Heme) that confirmed LMNA-NTRK1 fusion. Oral entrectinib has been initiated leading to tumor shrinkage that enabled complete surgical resection. The adverse effects registered in this case are one forearm fracture and increased appetite accompanied by significant weight gain that are diminished after medication dose reduction.

In conclusion, implementation of the targeted therapy has enabled avoidance of mutilating surgery and adverse effects of conventional cytostatic agents in the first case. Excellent control of malignant mesenchymal tumor has been achieved in the second case.

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FANCONI ANEMIA: A CASE REPORT

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10.1136/archdischild-2021-europaediatrics.309

Background Fanconi anemia is a rare genetic inheritance disease. Although the transmission in most cases is autosomal recessive, in some instances, it can be X-linked. Congenital malformations are the first clinical presentation that could affect many organic systems like the skeletal, skin, cardiac, pulmonary, central nervous system, gastrointestinal and urogenital. Limb asymmetry, thenar hypoplasia, microcephaly and low stature and weight are very frequent. Patients may also present ear malformations with or without congenital deafness and intellectual disabilities. The most serious problem is early severe bone marrow failure which leads to pancytopenia and, some years later, to the progression of acute myeloid leukemia or myelodysplastic syndrome. Moreover, the presence of solid tumors (head and neck or anogenital) is more prevalent.

Case Report A 9 year old male child was born with polymalformations such as imperforate anus, micropenis, inverted nipples, low implanted ears, sacrococcygeal sinus, thenar and radius hypoplasia. His birth occurred under a supervised gestation of 42 weeks by caesarean because of fetal bradycardia. The Apgar index was 5/8/9 and reanimation was performed. The patient was hospitalized in intensive care and anus correction surgery was performed.

The child's parents were healthy but consanguineous (cousins). The boy is the couple's third child. The first one died at 7 years old with acute myeloid leukemia and had a previous diagnosis of Fanconi anemia. The second one had sudden death at 3 months old. Because of this, karyotype and evaluation of chromosomal breakage induced by diepoxybutane (DEB) was tested and the last one confirmed Fanconi anemia diagnosis. Until now, he developed pancytopenia and bone marrow failure without progression to leukemia. Later, he had a younger brother without Fanconi anemia. It should be noted that the child in question has a 17 year old male cousin also diagnosed with Fanconi anemia. The cousin has low stature, intellectual disability and performed Hematopoietic stem cell transplantation at 7 years old.

Currently, the child takes danazol and receives periodic transfusions. He has multidisciplinary follow-up and was proposed to Hematopoietic stem cell transplantation.

Conclusion Fanconi anemia is responsible for many malformations and inevitable bone marrow failure. The presence of an inheritance pattern increases the level of suspicion when other relatives have the disease.

After diagnosis, these patients need close multidisciplinary follow-up to improve the expectancy and quality of life. The prevention or early detection of neoplasia is the main objective for these patients, as it leads to poor prognosis.

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A GIRL WITH PRE-B ACUTE LYMPHOBLASTIC LEUKEMIA ON MAINTENANCE THERAPY WITH HEREDITARY COMPLEMENT-MEDIATED THROMBOTIC MICROANGIOPATHY (AHUS)

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10.1136/archdischild-2021-europaediatrics.310