Paediatric Hematology and Oncology

285 POSTERIOR REVERSIBLE ENCEPHALOPATHY SYNDROME (PRES) – A RARE, SEVERE AND NEW COMPLICATION DURING THE TREATMENT OF ACUTE LEUKEMIA IN CHILDREN

R Jurac*, AM Ardelean, M Gafencu, C Petrescu, C Jinca, E Boeriu, S Arghirescu, G Doros. 'Victor Babes' University of Medicine and Pharmacy Timisoara, Romania, Ilrd Pediatric Clinic

10.1136/archdischild-2021-europaediatrics.285

Aim To present seven cases of Posterior Reversible Encephalopathy Syndrome (PRES), diagnosed between 2014-2019, as a complication of acute leukemia treatment in children. PRES was cited for the first time in 1996 by Hinkey and collaborators as a posterior leukoencephalopathy syndrome.

Methods These patients were admitted and treated for Acute lymphoblastic leukaemia during a 5 years period. From the total number of 162 patients with leukemia only 7 cases developed PRES. They were 3 to 12 yo, 5 males and 2 females. None of them had a preexisting neurologic disorder. PRES occurred during the induction chemotherapy. All patients underwent clinical evaluation, ECG, echocardiography and laboratory tests. At the onset of the neurological symptoms emergency head CT-scan was performed, followed by head MRI which confirmed the diagnosis.

Results Neurologic manifestation of PRES consisted of upper or lower limb muscle spasm, marked agitation, aggressive behavior, generalized seizures followed by coma, right upper limb hemiparesis and facial paresis. The first patient developed PRES as early as the 9-th day of treatment after receiving high dose Prednison that induced hypertension. In the rest of the children PRES occurred simultaneously with sepsis (due to severe bone marrow aplasia), SIADH (Syndrome of inappropriate antidiuretic hormone secretion) or uncontrolled high blood pressure due to corticotherapy. Blood pressure was controlled only with combined antihypertensive agents at maximal dose.

The PRES diagnose was consistently based on cerebral MRI neuroimages that showed typical lesions of vasogenic edema with hyperintense T2 and FLAIR signals with a characteristic location for PRES. Prompt anticonvulsant and antihypertensive therapy was started, and the patients made a full neurological recovery. The MRI was repeated with minimal ischemic lesions after 1 mo and almost normal after 3 mo. The treatment protocol for leukemia was interrupted from 12 to 43 days.

Conclusions PRES is a rare, severe and almost new described complication in children with leukemia. We have to recognize and to treat it promptly in order to save the patient. A multi-disciplinary team (pediatric oncologist, cardiologist, neurologist, radiologist and intensive care specialists) is necessary to manage such cases. A positive diagnosis was established after performing cerebral MRI. Severe acute hypertension was controlled only with a combination of for anti-hypertensive drugs administered at a maximum dose.

The question regarding the etiology of PRES is still present, because only patients with sepsis, SIADH and hypertension due to corticotherapy developed the syndrome from all leukemia patients.

286 MALIGNANT PERIVASCULAR EPITHELOID CELL TUMOR IN A TEN-YEAR-OLD GIRL

Jelena Roganovic*, Roganović Jelena, Jonjić Nives, Hadzisejdic Ita, Glavan Neda, Seili Bekafigo Irena. Clinical Hospital Centre Rijeka

10.1136/archdischild-2021-europaediatrics.286

Perivascular epitheloid cell tumors (PEComas) represent a group of mesenchymal neoplasms characterized by the presence of epitheloid cells of mixed myo-melanocytic differentiation with uncertain malignant potential. The tumors are very rare with a female preponderance and median age of 45 years. The most common location is retroperitoneum, but almost every site has been reported. PEComas have an unpredictable clinical behavior.

We present a 10-year old girl with a 3-week history of progressive swelling in the left infraclavicular region. At examination, a painless, well circumscribed, firm mass measuring 3 cm in diameter was palpable, with no other abnormal clinical findings. Laboratory tests were within normal limits. The ultrasound showed well defined, echogenic heterogeneous soft tissue mass with discrete vascularization. Fine needle cytology was suspicious of a malignant mesenchymal tumor. The girl was referred to the pediatric surgeon, and complete resection was done. Pathology finding established the diagnosis of PEComa with malignant morphology and peculiar immunophenotype of tumor cells (negative for melanocytic markers HMB-45 and Melan A, but positive for MITF). Two weeks later, two painless nodules, 5 and 10 mm in diameter, were noticed at the site of the excised tumor.

Subsequent thorough examination, including magnetic resonance imaging of the primary site, chest X-ray, chest and abdominal computed tomography, and positron emission tomography-computed tomography, was without evidence of disease. The girl underwent re-excision with clear resection margins. She was treated with adjuvant chemoradiotherapy for non-rhabdomyosarcoma pediatric soft tissue tumors. At 2-year regular follow-up, the girl is well and with no signs of recurrence.

To the best of our knowledge, this is the first reported case of malignant PEComa in a young patient. Our case highlights the challenges with regard to preoperative diagnosis and treatment. Due to the uncertain prognosis, close clinical surveillance accompanied by radiological imaging is mandatory.

287 FREQUENCY OF FACTOR II, FACTOR V LEIDEN AND MTHFR MUTATIONS IN CHILDREN WITH CANCER

Jelena Roganovic Roganovic*, Ana Djordjevic, Blazenka Grahovac, Lidija Bilic Zulle, Jelena Roganovic. Clinical Hospital Centre Rijeka

10.1136/archdischild-2021-europaediatrics.287

Thrombosis is an increasingly recognized complication of malignancy, occurring in up to 20% of patients with cancer. Cancer-associated thrombosis is linked with poor prognosis, being the second leading cause of death in cancer patients. The pathogenesis is complex, and includes multiple genetic and acquired factors. There is significantly less knowledge

about thrombosis in pediatric cancer population compared to adults. Besides, the role of inherited thrombophilia in all cancer patients with thrombosis is still unclear. The aim of this study was to examine the role of genetic thrombophilic defects in cancer-associated thrombosis in children.

The study included 47 children (13 girls and 34 boys) with newly diagnosed cancer, treated at the Division of Hematology and Oncology, Department of Pediatrics, Clinical Hospital Centre Rijeka, between January 2010 and December 2015. Thirty-six patients had hematological malignancies and 11 patients had solid tumors. The median age was 8.8 years (range 0.4 – 19.3 years). Molecular tests on samples of isolated DNA comprised genetic polymorphisms of Factor V (FV Leiden G1691A), prothrombin (FII G20210A) and the enzyme methylenetetrahydrofolate reductase (MTHFR C677T).

In the study group, 8.5% were heterozygous for Factor V Leiden, 6.4% were heterozygous for prothrombin G20210A mutation, 6.4% were homozygous for MTHFR C677T mutation, and 10.6% had combined thrombophilic defects. The prevalence of thrombophilic defects was in accordance with the prevalence in general population. There was no difference between boys and girls, and between patients with hematological malignancies and solid tumors. The rate of thrombosis was 8.5%. Three patients had upper extremity deep venous thrombosis and 1 had right atrial thrombosis. In half (2/4) of patients combined Factor V Leiden and MTHFR C677T heterozygosity were identified.

Our data suggest that inherited thrombophilia is not related to cancer pathogenesis in children. Inherited thrombophilia may play a role in the pathogenesis of cancer-associated thrombosis. A better understanding of the interactions between genetic and non-genetic causes will help to improve the prophylactic and treatment strategies for thrombosis pediatric oncology patients.

288

EXTRACORPOREAL MEMBRANE OXYGENATION AND ALLOGENIC HEMATOPOIETIC STEM CELL TRANSPLANTATION IN A CHILD WITH ACUTE MYELOID LEUKEMIA

Toni Matić*, Zrinko Šalek, Gordana Jakovljević, Izabela Kranjčec, Slobodan Galić, Filip Rubić, Miran Cvitković, Sandro Dessardo, Milivoj Novak, Maja Pavlović, Lucija Mucavac, Ana Petrović Gluščić, Ernest Bilić, Darko Anić, Dražen Belina. *Department of Pediatrics, UHC Zagreb*

10.1136/archdischild-2021-europaediatrics.288

ECMO is rarely used in children with acute leukemia due to possible complications.

We describe a 3.5-year-old boy with acute myeloid leukemia (AML) who required extracorporeal membrane oxygenation (ECMO) support due to severe acute respiratory distress syndrome (ARDS) and septic shock.

At the age of 2 years, he was diagnosed with T-cell lymphoblastic lymphoma and he received treatment according to EURO-LB 02 protocol. At the end of the maintenance therapy leukocytosis with anemia and thrombocytopenia occurred. Bone marrow aspiration showed suspicion of myeloid neoplasm.

Second bone marrow aspiration, which was sent to a foreign hematologic center, confirmed AML. Because of the previous lymphoma which started with eosinophilia, and unusual occurrence of AML early after, immature lymphoid/myeloid

neoplasm with eosinophilia was suspected. AML induction treatment was started, with a plan to proceed to allogeneic hematopoietic stem cell transplantation (HSCT) after chemotherapy, because of poor prognosis. The patient received AIE part of the AML-BFM 2012 protocol without serious complications, but on the 9th day of the HAM part of the protocol septic shock developed. The patient deteriorated and was transferred to the Pediatric Intensive Care Unit. A chest X-ray showed extensive infiltrative shading. *Pseudomonas aeruginosa* was isolated in blood culture. Further development of respiratory failure with ARDS and progression of the septic shock required intubation, mechanical ventilation and inotropes. However, conventional therapy was insufficient so central veno-arterial ECMO was indicated and performed. The ECMO was continued for

22 days. Three days after the ECMO discontinuation he was extubated and respiratory and hemodynamically stable. Bone marrow aspiration after the ECMO treatment showed complete remission so the decision to proceed to HSCT without further chemotherapy was made. Allogeneic HSCT from a matched unrelated donor with myeloablative conditioning was performed 40 days after ECMO discontinuation. Three weeks after HSCT acute skin GVHD grade II responded well to corticosteroid treatment together with cyclosporine. Four weeks after the HSCT steroid-resistant liver GVHD developed but responded well to mini extracorporeal photopheresis (ECP). Five months after HSCT patient is in good condition, with good engraftment, in complete remission, and without GVHD.

In our patient, ECMO support provided survival of septic shock and pneumonia with ARDS, and treatment with allogeneic HSCT provided survival of high-risk leukemia with poor prognosis.

289

HEREDITARY SPHEROCYTOSIS ASSOCIATED WITH GILBERT SYNDROME – A CASE REPORT

Ivona Butorac Ahel*, Kristina Baraba Dekanić, Goran Palčevski, Jelena Roganović. *Clinical Hospital Center Rijeka*

10.1136/archdischild-2021-europaediatrics.289

Gilbert syndrome (GS) and hereditary spherocytosis (HS) are hereditary diseases with high prevalence rates and similar symptoms. The calculated coexistence of these two diseases is 15-35/million births. Severe hyperbilirubinemia compared with the degree of hemolysis in patients with HS should raise suspicion of associated clinical conditions such as GS, glucose-6-phosphate-dehydrogenase deficiency or thalassemia.

We present the case of a 21-month-old girl with HS and GS. The diagnosis of HS was established by osmotic fragility test results in the age of 40 days.

During infancy the patient had several hemolytic episodes precipitated by upper respiratory tract infections, requiring transfusion therapy. Bilirubin levels were around 100 mmol/l. At the age of 21 months during viral infection, an unusually high unconjugated hyperbilirubinemia was noticed (total bilirubin/direct level 239/13 mmol/l) with mild anemia in the absence of severe hemolysis. As hyperbilirubinemia could not be explained by hemolysis, further evaluation was performed. Genotyping of the conjugation enzyme uridine diphosphate-glucuronosyltransferase confirmed GS. The girl has been followed-up regularly, with consistently higher values of total