PP 29

A RARE CASE OF PANCREATOBLASTOMA IN A PEDIATRIC PATIENT

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Case Report: Pancreatoblastoma (PB) is a rare malignant neoplasm. PB is frequently detected in children under 10 years of age. Symptoms are nonspecific. When diagnosed, most tumors are enlarged (> 5 cm). Abdominal pain is often the first complaint (44%). Alpha-fetoprotein (AFP) levels are high. Provides long-term survival with surgical resection. It has been reported that the prognosis is poor if metastases are detected. Here we present a seven-year-old female PB case. She applied with the complaint of abdominal pain. On physical examination, a mass was palpated in the epigastric region. The changed laboratory findings were an increased serum AFP level of 171.1 micrograms/L (normal range 0-9 micrograms/L). Abdominal computed tomography (CT) examination revealed a solitary mass of approximately 6 × 4 cm in the tail of the pancreas. Multiple mass lesions were observed in the liver. These lesions were evaluated as compatible with metastasis. She was diagnosed with PB histopathologically after Tru-cut biopsy. Pathologically increased Fluorodeoxyglucose (FDG) uptake (SUVmax: 9.99) was observed in the mass lesion around the right upper quadrant gastric corpus in F-18-Fluorodeoxyglucose Positron Emission Tomography/Computed Tomography (^{18}F-FDG PET/CT). Malignant hypermetabolic metastatic multiple hypodense mass lesions (SUVmax:7.7) were seen in the liver. OPEC chemotherapy was given. In the evaluation performed after 5 cycles of chemotherapy, a decrease in FDG uptake ^{18}F-FDG PET/CT was detected. The patient was evaluated as responsive to treatment. This case report may contribute to the literature with its rarity and treatment approach.

https://doi.org/10.1016/j.htct.2024.11.057

PP 30

RPL5 NOVEL MUTATION IN A PATIENT WITH DIAMOND BLACKFAN ANEMIA

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Case Report: Diamond-Blackfan anemia (DBA) is a rare inherited disorder characterized by macrocytic anemia, congenital malformations, and growth retardation, typically presenting in the first year of life. RPL5 encodes a component of the 60S ribosomal subunit, and mutations in this gene are associated with DBA, which is usually inherited in an autosomal dominant. Our case is presented after identifying a novel mutation that lacks the typical phenotypic features associated with the condition. A 17-month-old female patient was sent to our hospital because she was pale and had anemia. During the physical examination, the patient exhibited growth and developmental retardation (height in the 3rd to 10th percentile, weight in the 3rd percentile) and a pointed nose; however, no organomegaly or congenital malformations were detected. Laboratory results showed a hemoglobin level of 4.9 g/dL, an MCV of 90 fL, and a corrected reticulocyte count of 0.8%. HbF level in hemoglobin electrophoresis was 3.5%. Bone marrow examination revealed severe hypoplasia in the erythroid series. Genetic examination using next-generation sequencing detected a novel mutation in the RPL5 gene c. 10G>C (p. Val4Leu) (Heterozygous)). Although RPL5 mutations show more severe phenotypic features in DBA, the new mutation detected in our patient caused anemia and developmental and growth retardation without congenital malformation. This genetic change has not been previously reported in the literature as a novel mutation. However, according to the American College of Medical Genetics and Genomics (ACMG) criteria, this variant has been classified as a "variant of uncertain significance". Given that no additional mutations were identified in the whole exome sequencing (WES) analysis conducted on our patient, the hematological and bone marrow findings were consistent with a diagnosis of DBA. **Methodology:** A blood transfusion was administered to the patient, and steroid treatment was started. Our patient responded to steroid treatment during follow-up. WES analysis was also requested for our patient's mother, father, and sibling. Based on the results, donor screening for bone marrow transplantation will be initiated. Once the results are available, the phenotype-genotype relationship can be interpreted more accurately.

https://doi.org/10.1016/j.htct.2024.11.058

PP 31

HYPEREOSINOPHILIC SYNDROME

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Case Report: A 58-year-old female patient was referred to the hematology clinic in July for examination after leukocytosis was detected in her tests. She has a known history of CKD, Type 2 DM, HT, hyperlipidemia. The patient's general condition is good, and she had a complaint of numbness in her hands. First 1.5 months ago, numbness in her right hand up to the wrist began,especially severe in the first 3 fingers The same complaints started in her left hand 1 month ago. Physical examination findings were within normal limits. On sensory examination, there was hypoesthesia in the first 3 fingers of both hands, especially on the right. In the blood tests performed at the time of admission, leukocytes were 34.440μL, neutrophils 8.840μL, eosinophils 20.540μL, absolute lymphocyte count 3.890μL, monocytes 1.080 μL, hemoglobin 12 g/dL, platelets 348.000 μL, creatinine 1.07 mg/d/L, CRP: 56.6 mg/dL, sedimentation - 10 mm/h were measured. The patient has had borderline eosinophilia (1510μL) since 2022. Flow cytometry was performed on peripheral blood. 11% lymphoid series and 89% myeloid series cells were seen. No abnormality was observed in the lymphoid series. A slight regression in maturation was seen in myeloid series cells and