PP-054 A CASE OF PRIMARY IMMUNODEFICIENCY WITH IMMUNE DYSREGULATION: CD25 DEFICIENCY

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Aim We present a case of CD25 deficiency in a boy from an Uzbek family, confirmed by an identified mutation in the IL-2RA gene.
Material and Method DNA research using paired-end sequencing technology.
Results CD25 deficiency is a rare primary immunodeficiency with immune dysregulation, according to the International Union of Immunological Societies (IUIS) classification updated in 2022, it is classified as class 4, defects of regulatory T cells. In our patient the disease manifested itself in the neonatal period in the form of severe chronic diarrhea, susceptibility to viral, bacterial, fungal infections, autoimmune manifestations, malabsorption syndrome, malnutrition, and skin lesions. The patient received massive antibacterial therapy, antifungal therapy, intravenous immunoglobulin, and parenteral nutrition; despite treatment, the patient continued to have manifestations of a systemic inflammatory response and dysfunction of the gastrointestinal tract. No pronounced changes in the immunogram were detected. When studying DNA using clinical sequencing at the age of 5 months, a mutation c.473_485delCTCTACACAGAGG was identified in a homozygous variant in the region chr10:6021575ACCTCTGTGT AGAG>A, in the IL2RA gene. After clarifying the diagnosis, the patient was prescribed sirolimus with a positive effect. The first hematopoietic stem cell transplant was performed at 10 months, and the patient is currently preparing for a second hematopoietic stem cell transplant.
Conclusions CD25 deficiency is a rare primary immunodeficiency disorder with immune dysregulation, should be suspected in patients based on characteristic clinical manifestations. The most common and early symptom of CD25 deficiency is severe, chronic diarrhea. Skin lesions (eczema) and susceptibility to cytomegalovirus infection are also early symptoms of CD25 deficiency. Early diagnosis of CD25 deficiency can improve the prognosis and quality of life of patients. DNA testing using clinical sequencing can confirm the diagnosis of CD25 deficiency. Presentation and integration of data on CD25 deficiency is necessary to better understand the manifestations and natural history of this pathology.

PP-055 PETER'S ANOMALY – A RARE CAUSE OF VISUAL IMPAIRMENT IN PAEDIATRICS

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Aim Presenting case report of Peter's anomaly.
Material and Method Eight-month-old female infant who at birth was noted to have dysmorphic features (depressed nasal bridge, mid facial hypoplasia, low set ears and hypertelorism) and eyes malformations in form of bilateral corneal opacities, left eye proptosis, left megalocornea and bilateral cataracts. There was mild generalised hypotonia otherwise normal systemic examination. Genetic tests were done and given her eye features she was referred to the Ophthalmologist. Her microarray showed loss of approx. 2.4 Mb in short arm of chromosome 6 at band 6p25.3-p25.2 between base pairs 163083 and 2527433. This loss includes FOXC1(OMIM 601090) gene which has a strong association with abnormal eye development and Peters anomaly. Her other tests including TORCH screening, homocysteine levels and urine for CMV were all reported normal. She had bilateral corneal grafts and lens extraction. Currently she can perceive light but there are on-going concerns about her vision. Her growth and development to date are appropriate. Peters Anomaly (PA) is a rare form of developmental malformation involving the anterior segment of the eye as well as other body organs. Anterior chamber dysgenesis is characterized by corneal opacity and various other anomalies which can result in amblyopia of varying degree or even blindness. It can be unilateral or bilateral, isolated, or associated with systemic malformations. In one study bilateral eye involvement was associated with a higher rate of systemic malformations (71.8%) when compared to unilateral involvement (36.8%). Early recognition and treatment may help to prevent complications in these patients.
Results Case report.
Conclusions Case report.

PP-056 EVALUATION OF MICRONUTRIENTS IN PATIENTS WITH PHENYLKETONURIA

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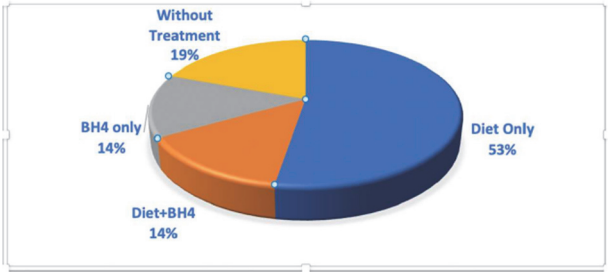
Aim The aim of this study was to contribute to the management of metabolic diseases by evaluating the development and micronutrient levels of patients diagnosed with hyperphenylalaninemia and phenylketonuria who received treatment and follow-up.
Material and Method This is a retrospective descriptive study conducted with the approval of the Istanbul Dr. Sadi Konuk Training Research Hospital Ethics Committee. The study was conducted at the İstanbul Dr. Sadi Konuk Training and Research Hospital between 01.11.2022 and 01.05.2023. Medical records of patients who were diagnosed with phenylketonuria and received outpatient or inpatient treatment at the Child Metabolism Clinic of Istanbul Dr. Sadi Konuk Training and Research Hospital between 01.01.2015 and 31.09.2022 were retrospectively reviewed. The clinical findings, anthropometric measurements, laboratory findings and treatment methods of the patients were evaluated. The study recorded the patients gender, date of application, date of birth, age at the



Abstract PP-056 Figure 1 Treatment status.

Abstract PP-056 Table 1 Weight, height, and head circumference SDS of the treatment groups

	Diet treatment	Diet+BH4 treatment	BH4 treatment	Without treatment
Height SDS	<2: 2	<2: 1	<2: 1	<2: 0
	-2/+2: 28	-2/+2: 7	-2/+2: 7	-2/+2: 11
	>2: 0	>2: 0	>2: 0	>2: 0
Weight SDS	<2: 11	<2: 2	<2: 1	<2: 0
	-2/+2: 17	-2/+2: 5	-2/+2: 7	-2/+2: 11
	>2: 2	>2: 1	>2: 0	>2: 0
Head circumference	<2: 1	<2: 0	<2: 2	<2: 2
SDS	-2/+2: 29	-2/+2: 8	-2/+2: 8	-2/+2: 11
	>2: 0	>2: 0	>2: 0	>2: 0

time of application, reason for application, phenylalanine levels at the time of application, phenylalanine levels during follow-up, treatment received, treatment compliance, consanguinity, and sibling history, physical examination findings, anthropometric measurements and laboratory values.

Results There was no significant difference in growth, development, or micronutrient levels between the diagnostic and treatment groups (figure 1, table 1). This result is associated with adherence to established standards in clinical and blood level monitoring through follow-up and treatment. Early diagnosis and the selection of an appropriate treatment protocol, along with frequent patient monitoring, are essential for ensuring appropriate development compared to peers.

Conclusions The fact that there is no difference in the growth, development and micronutrient levels of the patients according to the diagnosis type and treatment protocols in the follow-up of hyperphenylalaninemia is a result of the success of the follow-up. In particular, monitoring blood PA levels, diet and micronutrient deficiencies in patients receiving diet therapy enabled them to have similar physical and neurocognitive development as their healthy peers.

PP-057 THE STATE OF VITAMIN D SUPPLY AND THE INFLUENCE OF VDR GENE POLYMORPHISMS ON THE COURSE OF CYSTIC FIBROSIS, ASTHMA, JUVENILE IDIOPATHIC ARTHRITIS IN CHILDREN OF THE RUSSIAN FEDERATION

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Aim To assess the frequency of low vitamin D deficiency and conduct an association search for genetic variants (c.1206T>C, c.152T>C, c.1174+283G>A) of the VDR gene with clinical manifestations, calcidiol levels and response to therapy in cystic fibrosis (CF), bronchial asthma (BA), juvenile idiopathic arthritis (JIA).

Material and Method 283 patients with cystic fibrosis (CF), 160 with BA, 150 with JIA and 333 healthy children in the control group were examined, and calcidiol content was determined. Testing of polymorphic variants of the VDR gene (c.1206T>C, c.1175–9G>T, c.152T>C, c.1174+283G>A) was carried out using PCR and RFLP analysis.

Results A year-round high incidence of low vitamin D supply was revealed among CF in 50.7%, asthma in 89.6%, and JIA in 74.0%. The occurrence of meconium ileus, decreased lung function, chronic Ps. aeruginosa infection, a chronic lung infection caused by non-fermenting gram-negative bacteria, is higher among carriers of the TT c.152T>C FokI VDR gene genotype in cystic fibrosis. Liver cirrhosis is more often (OR=4.300; p=0.051) realized in carriers of the AA genotype BsmlI (c.1174+283G>A) of the VDR gene. The occurrence of manifestations of the 'atopic march' increases many times when carrying the genotype TT c.1206T>C(A>G) TaqI (OR=13.000; p=0.046), genotypes AA and GA BsmlI (c.1174+283G>A) (OR= 18.000; p=0.017). Calcidiol deficiency against the background of asthma is 2.7 times (p = 0.003) more often recorded among carriers of the TT and CT genotypes c.1206T>C (A>G) TaqI of the VDR gene. The risk of systemic onset of JIA, polyarticular variant, high degree of activity, uveitis, high frequency of biological therapy (p <0.05) are carriers of the TT genotype c.1206T>C(A>G) TaqI, TT genotype c.152T>C FokI, genotype AA polymorphism BsmlI (c.1174 +283G>A) of the VDR gene.

Conclusions The high frequency of low vitamin D supply, the contribution of VDR gene polymorphisms during the studied diseases and vitamin D supply are shown.

PP-058 POSTCHEMOTHERAPY COMPLICATIONS IN A CHILD WITH ACUTE LYMPHOBLASTIC LEUKEMIA

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Aim We present the case of an 11-year-old patient, diagnosed in September 2023 with B-Cell acute lymphoblastic leukemia, to whom induction chemotherapy was initiated according to the ALL IC-BFM 2002 protocol, with complete remission on day 33, undetectable minimal residual disease.

Material and Method He is readmitted in October 2023: normal clinical examination; biological: leukopenia, neutropenia, secondary grade 1 anemia, thrombocytopenia.

Results It presents fever, chills, with the repetition of fever. The general condition remains affected, pale, plantar edema, vesicular breathing sounds harsh, high fever persists. Antibiotic treatment, fluid and electrolytic infusion, red blood cell and platelet transfusions are instituted. It presents hypoproteinemia, hypokalemia, blood culture - staphylococcus aureus. Echocardiogram: FEVS 50%, mitral and tricuspid regurgitation secondary to pulmonary hypertension, heart failure class III NYHA. He receives cardiological treatment. In evolution, the patient presents with acute staphylococcal pneumonia (MRSA culture).