PETERS PLUS SYNDROME

Background Peters Plus syndrome is an autosomal recessive and rare disorder characterized by a variety of anterior eye chamber defects, of which the Peters anomaly occurs most frequently. Other major symptoms are a disproportionate short stature, developmental delay, characteristic craniofacial features, and cleft lip and/or palate.

Observations We report 4 cases of peters plus syndrome who were admitted in the department of pediatric of sfax. The family history revealed consanguineous parents in 3 cases, ocular abnormalities in 2 cases and the death of sibling in one case. Multiples abnormalities were noted in all cases just after birth like bilateral corneal opacities and facial dysmorphism. 3 new borns had Skeletal system abnormalities (Short limbs, Short, broad hands, clinodactyly). Congenital heart malformations were present in 2 cases and renal abnormalities were noted in one case. During the evolution, 3 patients presents a failure to thrive and developmental delay. One patient was died because of pneumonia.

Conclusion Peters plus syndrome is inherited in an autosomal recessive manner. The diagnosis is based on clinical findings and genetic analysis; prenatal diagnosis for pregnancies at increased risk are possible if the disease-causing mutations in the family are known.

LYSOSOMAL STORAGE DISORDERS IN NON-IMMUNOLOGICAL HYDROPS FETALIS - MORE COMMON THAN ASSUMED?

Background Although non immunological hydrops fetalis (NIHF) is a very rare disorder, the disturbance accounts for a disproportionate share (3%) of overall mortality in the perinatal period. Lysosomal storage disorders (LSD) are only exceptionally considered to be the cause of NIHF. The reported incidence is about 1%. On the other hand, in about 18% of all cases, NIHF is classified as idiopathic.

Patients and methods We report four cases of transient NIHF due to LSD and reviewed the literature for LSD associated with NIHF.

Results At present, 12 different LSD are described to be associated with NIHF. The majority of reported patients already had a family history of NIHF which had not been investigated. A diagnostic approach to the fetus with NIHF due to suspected LSD is suggested.

Conclusions Extensive and thorough investigation of the etiology of NIHF is obligatory. In particular, LSD should be considered in idiopathic NIHF. Enzymatic studies in chorionic villous samples or amniotic cultured cells, once the most common conditions associated with NIHF have been ruled out, should be performed. We assume that the incidence of LSD in NIHF is significantly higher than the estimated 1% reported in previous studies. This is important for genetic counseling, as there is at first, a high risk of recurrence and, secondly, the availability of enzyme replacement therapy for an increasing number of LSD.

CHROMOSOMAL ABNORMALITIES IN A TERTIARY NEONATAL INTENSIVE CARE UNIT

Background and Aim Congenital malformations and chromosomal abnormalities are important problems for the neonatal morbidity and mortality especially in well developed countries. In this study we investigated the prevalence and distribution of the chromosomal abnormalities in our NICU.

Method Chromosomal abnormalities identified between 2008 and 2012 were retrospectively analyzed. Also, demographic features and concomitant congenital malformations were also collected. Cytogenetic analysis was performed on peripheral blood of newborns by standard chromosomal analysis methods. According to our hospital protocols, newborns with more than one major malformation or with 1 major plus 2 minor abnormalities were scheduled for chromosomal abnormalities.

Results During the study period, 431 chromosomal analyses (3.2%) were performed among the 5170 hospitalized newborns. 78.4% of the cases had normal chromosomal karyotype. Consanguinity rate was 27.9%. Major chromosomal abnormality rate was 1.8%.

Frequency of chromosomal abnormalities were as follows; Trisomy 21 13.5% (n=58), trisomy 18 2.3% (n=10), trisomy 13 1.2% (n=5), 45 XO 0.5% (n=2) and other chromosomal abnormalities were 4.2% (n=18). Congenital heart abnormalities (55.9%), craniofacial abnormalities (44.6%) and genito-urinary anomalies (16.9%) were most common comcomitant malformations. Cardiovascular abnormalities were most common malformations in newborns with trisomy 21.

Conclusion Frequency and distribution of the chromosomal abnormalities in our NICU were similar compering with other popultational studies. Trisomy 21 was most common chromosomal abnormality. Newborns with malformations in more than two organ system should be investigated chromosomally as well.

CHOLESTEROL ESTER TRANSFER PROTEIN GENE POLYMORPHISM AND SELECTED LIPIDS PARAMETERS IN CHILDREN FROM FAMILIES WITH HISTORY OF CVS DISEASES

The human population especially predisposed to early development of atherosclerosis are children from families with history of cardiovascular system diseases. The aim of this study was to examine lipids parameters associated with cardiovascular diseases and polymorphisms of G279A located in the Cholesterol Ester Transfer Protein (CETP) gene.

Material/methods The study covered 30 children aged 5–6 years from families with history of cardiovascular system diseases. The children were examined physically, and nutritional status assessed. In all of the children examined, the blood concentrations of
triglycerides, total cholesterol, high density lipoprotein (HDL) cholesterol, apolipoproteins (A1 and B), lipoprotein(a) and polymorphisms the G279A locus of the CETP gene, were determined.

Results In children with genotype B1B1, a significantly lower % cholesterol concentration in the HDL fraction was found than in those with genotype B1B2 and B2B2. Children with genotype B1B1 were significantly shorter in stature than children with genotypes B1B2 and B2B2. Other biochemical parameters, such as levels of apolipoproteins (A1 and B) and lipoprotein(a) were not significantly different between these genetic polymorphisms.

Conclusion In the future, proper early prevention based on the modification of risk factors, periodic lipid profile control, and the assay of markers of early atherosclerosis changes can reduce morbidity and mortality due to cardiovascular system diseases among children.

DENVER II DEVELOPMENTAL TEST PERFORMANCE OF THE CHILDREN WITH IRON-DEFICIENCY ANEMIA BEFORE AND AFTER THE TREATMENT
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M Inalhan, T Ozlem, A Ozlem, C Mehmet, Y Feyza, Y Aşıkay. Clinics of Paediatrics, Zeynep Kamil Obstetrics, Gynecology and Paediatric Training and Research Hospital, Istanbul, Turkey

Objective It is known that non-treatment of iron deficiency anemia in early childhood results mental and cognitive retardation. In our study we aimed to examine the Denver-II developmental test performance of the children with iron-deficiency or iron-deficiency anemia before and after 3 months of the treatment.

Materials and Methods 53 children aged between 1 and 6 years whom diagnosed as iron deficient or iron deficiency anemia were studied. Denver-II test were applied to the cases before and after 3 months of the treatment. The Denver Test results were classified as normal, suspicious and abnormal.

Results Suspicious Denver-II test results were found significantly higher in iron deficient anemic infants compared with iron deficient children (p<0.01). The initial measurement level of ferritin and tranferrin saturation were found significantly low in children with suspicious test results compared with children with normal test results.

Before iron therapy, the rate of normal Denver test results in iron deficient anemic children was 42.5% and 81.5% in iron deficient children. After treatment the rate is 100% for iron deficient anemic children.

Conclusion If iron deficiency could be treated before it becomes chronic or serious, motor, cognitive and behavioural development deficits can be prevented.

GLAZMANN THROMBASTHENIA: SINGLE CENTER EXPERIENCE
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B Kutlubay, N Ozdemir, G Tuysuz, H Apak, T Celkan. Pediatric Hematology-Oncology Dept., Istanbul University, Cerrahpasa Medical Faculty, Istanbul, Turkey

Aim Glazmann thrombasthenia is a rare autosomal recessive disease characterized by defect in platelet aggregation. Here we report the management of children with Glazmann thrombasthenia followed at Cerrahpasa Medical Faculty Pediatric Hematology Department.

Methods Nineteen patients’ (42% girls, 58% boys; median age: 10 months) files were retrospectively reviewed.

Results The median age of the start of bleeding symptoms was 9 months (2 weeks-24 months). All patients presented with easy bruising and mucosal bleeding. Fourteen patients’ parents were consanguineous. In 15 patients, flow cytometry was performed. According to this, 7 had type I, 6 had type II and 2 had type III disease. Nine patients were treated by thrombocyte transfusion, tranexamic acid, recombinant active factor VII and fibrin glue as a single or combined therapy in invasive procedures; none of them had a major bleeding complication.

Conclusion Bleeding control of invasive procedures may be challenging in children with Glazmann thrombasthenia; local treatments, DDAVP, steroid and antifibrinolitics may be used with success.

ETHIOLOGICAL FACTORS AND PREVALENCE OF ZINC DEFICIENCY IN CHILDREN WITH IRON DEFICIENCY ANEMIA
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M Inalhan, Y Feyza, T Ozlem, A Ozlem, C Mehmet, O şuera. Clinics of Paediatrics, Zeynep Kamil Obstetrics, Gynecology and Paediatrics Training and Research Hospital, Istanbul, Turkey

Introduction and aim: Iron deficiency is the most common nutritional deficiency in developing and developed countries. In the developing countries iron and zinc deficiency are seen together. In our study we researched the prevalence of zinc deficiency and etiological factors in iron deficiency anemia.

Materials and method A study group consisted of 40 children, aged between 1–14 years, diagnosed with iron deficiency anemia who submitted to our clinic with different complaints between January and August 2010. Control group consisted of 40 healthy children. Age, sex, birth weights, history of mother’s prophylactic iron supplement usage, breastfeeding duration time, initial supplement feeding age, meat, fruits-vegetables consumption, pica history, the blood count variables, serum iron levels, iron binding capacity, ferritin and serum zinc levels were recorded.

Findings There was no significant difference between test and control groups in duration of breast feeding time and initial time for supplementary feeding (p>0.05). Positive correlation was found between breast feeding time and iron levels and transferrin saturation indices (p<0.01). Fruits and vegetables consumption were found significantly low in test group.

Number of hospital admissions were also found to be significantly high in the test group.

The serum zinc levels of the children who have had prophylactic iron supplements were significantly high compared with the children who have had no prophylaxis (p<0.05).

Conclusions As a result of this research, factors namely breast feeding duration time, prophylactic use of iron supplements and fruit and vegetables consumptions have a large role to prevent children from the iron deficiency.

DETECTION OF INTRON 22 GENE INVERSION (COMMON MUTATION) IN SEVERE HEMOPHILIA A IN KHOUSESTAN PROVINCE
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A Ghasemi, B Keikhai, K Zandian, H Galedani. Pediatric Hematology-Oncology, Mashhad University of Medical Sciences, Mashhad, Iran

Introduction Hemophilia A, is the most common severe coagulation disorder with an incidence of 1 in 5000 males. The most common genetic alteration is a gene inversion, which occurs in approximately 48% of patients with severe hemophilia A.

The goal of the study was to assess the presence of the intron22 inversion in patients with severe hemophilia A from Khozestan province.

Materials Twenty-five patients with severe hemophilia A and 15 mothers of these patients participated in the study patients were referred from the thalassemia service of Shafa Hospital to detect the