SNF array for copy number variations (CNV’s) showed a unique 550 kb duplication involving SOX3, RP1-177G6, and CDR1 genes, and the microRNA MIR320D2. This CNV was absent in 13,839 controls.

**Conclusions** A SRY negative 46, XX male with renal hypoplasia was found to have an exceedingly rare duplication involving the SOX-3 gene, proving its role in sex determination and suggesting its involvement in kidney development.

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**Abstracts**

**PETERS PLUS SYNDROME**

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**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 5 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 newborns had skeletal system abnormalities (Short limbs, Short, broad hands, clindacodyly). Congenital heart malformations were present in 2 cases and renal anomalies 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.

**CHROMOSOMAL ABNORMALITIES IN A TERTIARY NEONATAL INTENSIVE CARE UNIT**

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**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 13,839 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 (85.9%), cranio-facial abnormalities (44.6%) and genito-urinary anomalies (16.9%) were most common concomitant 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 comparing with other populational studies. Trisomy 21 was most common chromosomal abnormality. Newborns with malformations in more than two organ system should be investigated chromosomally as well.

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

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**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.

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

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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
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 antifibrinolytics may be used with success.

747 ETHIOLOGICAL FACTORS AND PREVALENCE OF ZINC DEFICIENCY IN CHILDREN WITH IRON DEFICIENCY ANEMIA
doi:10.1136/archdischild-2012-302724.0747
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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 suplement used in pregnancy, 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 suplements 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.