Method: This prospective and retrospective study was performed in the neonatal intensive care unit in Turkey. In order to identify SP-B iΔ4 gene polymorphism, we analysed genomic DNA by polymerase chain reaction. Preterm neonates with a gestational age below 34 completed weeks, who were treated in our unit were included the study. The neonates with congenital anomalies and the neonates who died before the postnatal 28th day were excluded. The study was approved by the Local Committee on Investigations in Human Subjects. Total of 131 preterms and 50 healthy term infants were investigated. Premature babies were divided into 4 with respect to the development of RDS and CLD.

Results: The frequency of SP-B iΔ4 gene variations did not differ between preterms (10.7%) and terms (10%). A total of 117 preterms with SP-B iΔ4 gene wild type (group A) and 14 preterms carrying the genetic variations (group B) did not differ in gestational age, gender distributionn and birth weight, RDS and CLD. In premature study supgroups, SP-B iΔ4 gene variations was 10.7% in control group (n:80), 15.8% in RDS group (n:19), 10% in RDS and CLD group (n:20) and 0% in CLD group (n:8). There wasn't any statistically significant difference between all supgroups.

Conclusions: In our premature babies, we coulnd't show any association between SP-B iΔ4 gene polymorphism and RDS / CLD.

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NEONATAL PRESENTATION OF EHLERS-DANLOS TYPE VII: DIAGNOSTIC CONSIDERATIONS

M. Klaassens[1], Y. Hilhorst-Hofstee[2], H. Staal[3], L. Speth[4], J. Schrander[1], G. Pals[5], A. De Paepe[6], F. Malfait[6], C. Schrander-Stumpel[7,8]

[1]Pediatrics, Maastricht University Medical Center, Maastricht, [2]Clinical Genetics, Leiden University Medical Center, Leiden, [3]Orthopedic Surgery, Maastricht University Medical Centert, Maastricht, [4]Pediatric Rehabilitation Medicine, Adelante Zorggroep, Valkenburg, [5]Clinical Genetics, VU Medical Center, Amsterdam, The Netherlands, [6]Center for Medical Genetics, Ghent University Medical Center, Ghent, Belgium, [7]Clinical Genetics, Maastricht University Medical Center, [8]Research Institute Growth and Development (GROW), Maastricht, The Netherlands

Ehlers-Danlos syndrome (EDS) is a clinically and genetically heterogeneous group of inherited connective-tissue disorders characterized by hypermobility, tissue fragility and skin abnormalities. Eleven subtypes have been characterized based on clinical features and molecular genetic abnormalities. EDS type VII (arthrochalasia type EDS) is characterized by bilateral congenital dislocation of the hips, severe generalized joint hypermobility with multiple dislocations, muscular hypotonia and discrete skin abnormalities. The diagnosis of type VII-EDS is of importance in the neonatal period because of consequences for mobility in later life. However, the differential diagnosis may be difficult because of overlap with other hypermobility syndromes. In addition, the significant hypotonia may direct the paediatrician towards neuromuscular disorders. In this report we describe three patients who, in retrospect, presented with the classical neonatal clinical features of EDS type VII. Before confirmation of the diagnosis, several differential diagnoses were considered. Diagnosis of EDS type VII was confirmed by mutation analysis showing a *de novo* mutation in COL1A2, resulting in skipping of exon 6 leading to the production of abnormal procollagen. This leads to defective collagen synthesis which is responsible for the clinical features. For physicians treating patients with EDS type VII achieving mobility for the patient is the greatest challenge. The prognosis regarding the achievement of independent walking is poor due to recurrent luxations of nearly all joints in severe cases. We summarize the literature and present some guidelines for the paediatrician.

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UGT1A1 GENE VARIANTS IN NEONATAL HYPERBILIRUBINEMIA

A. Kumar[1], P.K. Tiwari[1], A. Bhutada[1], R. Agarwal[1], R. Raman[2]

[1]Department of Pediatrics, [2]Department of Zoology, Banaras Hindu University, Varanasi, India

Background and aims: Polymorphisms of *UGT1A1* gene may contribute to neonatal hyperbilirubinemia (NNH). This study analyzes the role of seven variants of *UGT1A1* gene and certain clinical risk factors in NNH.

Methods: This was a prospective case control study which included 247 cases

(serum bilirubin ≥15 mg/dl) and 278 control (serum bilirubin< 15 mg/dl) newborns in the first 2 weeks of age from central India.Both term and preterm neonates were studied.Genomic DNA was subjected to PCR-RFLP,SSCP and DNA sequencing to find out *UGT1A1* gene variants.

Results: *UGT1A1* -3297 G >T variant was found in 44(17.8%) cases and 22(7.9%) controls (OR 2.5; 95% CI 1.46-4.34). CAT box insertion $(CAT)_{1\rightarrow}$

$(CAT)_2$ was seen in 5(2.1%) cases and 1(0.4) control (OR 5.7; 95% CI 0.6-49.3). TATA box variant $(TA)_6\rightarrow(TA)_7$ was detected in151 (61.2%) cases and 141 (50.7%) controls (OR1.42;95% CI 1.06-2.13). 211G>A variant was seen in 13 (5.3%) cases and 5(1.8%) controls (OR3.03;95% CI1.06-8.63). 1456 T>G variant was found in one case and none in controls while 686C>A and 1091 C>T variants were not detected in any case or control newborns. Compound variations of *UGT1A1* gene such as -3297 G >T and 211G>A, -3297G>T and $(TA)_6\rightarrow(TA)_{7,}$ and 211G>A and$(TA)_6\rightarrow$(TA)7 significantly increased the risk of hyperbilirubinemia. Logistic regression analysis showed prematurity, ABO incompatibility, sepsis, weight loss ≥10%, and -3297G>T, CAT box insertion and TATA box variants as significant risk factors for NNH.

Conclusion: Variants of *UGT1A1* gene, singly or in combination, contribute significantly to neonatal hyperbilirubinemia in Indian population.

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ORAL-FACIAL CLEFTS/THE INCIDENCE, CAUSES AND SYMPTOMS AT THE CHILDREN HOSPITALIZED IN CHILDREN'S WARD IN STIP

N. Velickova, M. Gacova

Faculty of Medical Science, University Goce Delcev, Stip, FYR Macedonia

Background: Oral-facial clefts are birth defects in which the tissues of the mouth or lip don´t form properly during fetal development. The causes of these birth defects are not well understood, but believe it may be a combination of genetic and environmental factors (drugs, illnesses, and the use of alcohol or tobacco while a woman is pregnant) - multifactorial inheritance.

Aim: To evaluate the incidence, causes and symptoms of oral facial clefts at the children´s ward in Stip.

Methods: After birth, cleft lip and palate are diagnosed by physical exam. Special exposure of their mothers during pregnancy indicates that environmental factors increase the risk of cleft lip and palate.

Results: During the past 10 year children with oral facial clefts are borns in series in the same or closer month in the year. We councluded that every year the number od children with oral-facial clefts growing and they were not associated with any syndrome Their parents doesn´t have a oral-facial clefts. Children with oral-facial clefts have special problems and complication like feeding difficulties, ear infections and hearing loss.

Conclusion: The obtain results suggest that environmental factors, such as drugs (several different anti-seizure drugs) and maternal smoking, are risk factors for appearance of oral-facial clefts. Cleft lips and palates not associated with a syndrome are caused by a combination of genetic and environmental factors. Other environmental factors that are suspected of playing a role include infections, maternal alcohol use and deficiency of the B vitamin folic acid.

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CYTOGENETIC PARAMETERS FOR CHILDREN LIVING IN REGIONS WITH RADIATION AND CHEMICAL TOXIC FACTORS

M. Appasova, S. Choy

Department of Paediatrics, Kazakh National Medical University, Almaty, Kazakhstan

By the purpose of the present research was the realization of a comparative estimation of cytogenetic parameters for children of Semipalatinsk and Aral locales. The cytogenetic method inspected 100 children living in locales, accumbent to Semipalatinsk test nuclear range (STNR), 100 children of Aral sea region and 50 children of group of matching. For the greater objectivity and the reliabilities as monitoring cytogenetic parameters were usage the data a CART. Cultivating of lymphocytes conducted pursuant to a standard technique in lab of an experimental mutagenesis of Institute of general genetics and cytology of a National Academy of sciences of Republic of Kazakhstan. The general frequency of aberrations in locales STNR (1,86) and Aral sea region (1,90) was approximately equal, but exceeded a similar parameter of children of group of matching (0,62) ($p < 0,001$ and $p <$