levels of transaminases, creatinkinase and lactic dehydrogenases. Genetic studies were performed in 8 of the patients, 6 presenting deletions and point mutations 2.

Conclusions: Progressive muscular dystrophy is a relative frequent disorder in children, which can be recognized early; if the high levels of transaminases are found, without other signs of hepatic disease, is necessary to determine the creatinkinase level. Genetic studies are required to specify the type of mutation in the initiation of therapies to slow evolution.

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AUTOMATED ECO-GENETIC SYSTEM PROGNOSIS OF BRONCHIAL ASTHMA IN GEORGIAN POPULATION

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Background: According to the actual direction of nowadays allergology more attention is paid to the prevention in early age period significantly decreases morbidity of allergic diseases and improve quality of life. Hereditary predisposition plays the most important role in the developing of disease, but it is not everything recognizable, it may be not demonstrated clinical symptoms in parents and other close relatives of affected individual.

Aim: To elaborate the new automated assessment cost-effective system which would allow early diagnostic and prevention BA by using "formalized cards". This system is based on the primary and secondary prognosis of BA.

Methods: HLA- markers (Terrasaki microlymphocytotoxic tests) typing was performed with 2nd class (DR, DQ) and 1st class (A, B, C) antigens locus's; Erythrocyte markers: ABO, Rhesus and MNS systems, Clinical-genealogic method (Falconer D., 1965): study of genealogical tree of investigated families; Genetic - statistic analysis "Informative Criteria" of different signs: ontogeny, phenotypic, environmental factors (by Kulback, Goubler E.B, 1990). Method of multi-dimensional phenotyping: agglomerative- hierarchical cluster analyse of chosen systems of signs. Analyse was performed in the interactive order.

Results: Multi-dimensional phenotyping shows phenotype classes: Classes of basic predisposition,

including up to 80% of individuals with manifested asthma; Classes of relative predisposition, including less than 80% of individuals with manifested asthma; Phenotype classes including a few patients; Phenotype classes not predisposed to BA, basically including healthy persons.

Conclusion: Elaborated computing table is enough simple and opportune for comprehensive use. It is proposed, for primary prognostic of BA. This method gives

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CYTOGENETIC RESULTS OF 153 PATIENTS WITH MENTAL RETARDATION IN MIDDLE ANATOLIA IN TURKEY

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Mental retardation (MR), that is a major social, educational, and health problem, occurs with the prevalence of 1-3% of general population. The etiologies of MR are complex the cause remains unknown in about 50% of cases and include teratogenic and environmental factors, perinatal asphyxia and genetic causes. Cytogenetic studies have been reported to determine the contribution of chromosomal abnormalities in parents with mental retardation from various other countries. As far as we know, no enough studies have been done in the middle Anatolia in Turkey. The aim of the present study was to evaluate the frequency and nature of chromosomal aberrations in Turkish patients that contribute to mental retardation.

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POLYMORPHISM OF SURFACTANT PROTEIN B GENES AND THE RISK OF RESPIRATORY DISTRESS SYNDROME AND CHRONIC LUNG DISEASE IN PRETERM NEONATES

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Objectives: To determine whether SP-B iΔ4 gene polymorphism is associated with respiratory distress syndrome (RDS) and chronic lung disease (CLD) etiology.

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 distribution and birth weight, RDS and CLD. In premature study subgroups, 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 subgroups.

Conclusions: In our premature babies, we couldn'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

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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

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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