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

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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