min and 5 minutes respectively. The patient developed respiratory distress syndrome in the first few hours. He had inter- and subcostal retractions, grunting, tachypnea (80 breaths per minute), nasal flaring and the pulse was 127 beats per minute with a SpO2 under 90% in room air and higher than 95% with oxygen supplementation. The treatment with Dexamethasone showed no improvement and an urgent Chest X ray was ordered which revealed a left pneumothorax with mediastinal shift to the opposite site. ABG revealed severe acidosis. (pH – 7.13, PCO2 – 70, PO2 – 46 mmHg). In view of impending respiratory failure and shock baby was intubated, the pneumothorax was drained. Hemoculture was positive with GBS. The antibiotic therapy (Ampicillin/Sulbactam and Amikacin) was started and the patient was carefully monitored.
Conclusions. In conclusion, although respiratory distress syndrome is rare in near term or term newborn, is usually secondary to a parenchimal pathology, being a common case of spontaneous pneumothorax in these infants. Early recognition and treatment is life saving. Usual manifestation is progressive respiratory difficulty starting soon after birth.
Key words: GBS infection, respiratory distress, near term infant, spontaneous pneumothorax

DEPARTMENT OF MOLECULAR BIOLOGY AND HUMAN GENETICS

25. DUCHENNE MUSCULAR DYSTROPHY AND LIMB-GIRDLE MUSCULAR DYSTROPHY: CLINICAL CASES

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Background : Muscular dystrophies (MD) represent a large group of genetic disorders that are manifested by progressive increase of muscle weakness. Duchenne muscular dystrophy (DMD) is an X-linked disorder and limb-girdle muscular dystrophies (LGMDs) include over thirty subtypes, that are classified in autosomal dominant (1A-1H) and recessive (2A-2W). Our aims was to highlight the clinical and genetic aspects in MD by reporting two clinical cases with the aim of improving the early diagnosis.
Case report. The study was performed on the basis of the literature review and presentation of two clinical cases: a 6-year-old boy with DMD and another 17 years old boy with LGMD. Patient G.V. was diagnosed with DMD at the age of 3 years. Electroneuromyography (ENMG) and genetic test (deletion of exons 45-52 in the dystrophin gene) confirmed the diagnosis. He has the following clinical signs: calf pseudohypertrophy, waddling gait, lordosis, elevated serum creatine kinase (CK) - 14 740 U/l, MB fraction – 833 U/l, lactate dehydrogenase (LDH) – 1934 U/l. Patient M.A. was diagnosed with LGMD at the age of 7 years through ENMG. He presents severe motor deficit, waddling gait, hypoplasia of the thigh muscles, permanent asthenia, total CK - 486 U/l, MB fraction - 36 U/l, LDH - 358 U/l. He has first-degree disability and cardiomyopathy.
Conclusions. The first signs of MD (DMD and LGMD) occur at early stages, but often are not recognized. Genetic counseling and prenatal diagnosis will significantly reduce morbidity and mortality, will contribute to the improving of the quality of life.
Key words: Muscular dystrophies (MD), Duchenne muscular dystrophy (DMD), limb-girdle muscular dystrophies (LGMDs).

26. THE CLINICAL-GENETIC PARTICULARITIES IN APERT SYNDROME

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