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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Background. Apert syndrome (AS) is a dominant autosomal genetic disorder caused by heterogeneous mutation in FGFR2 genes on chromosome 10q26 and belongs to a group of disorders known as craniofacial congenital malformations. AS can promote the premature fusion of bones in the skull, hands, and feet. The incidence of infants born with Apert syndrome is approximately 1 in 50000 to 80000. In this study is emphasized the importance of clinical and genetic approaches in the research on the specific diagnosis in patients with Apert syndrome.
Case report. The clinical particularities of Apert syndrome are determined by craniosynostosis result from the premature fusion of the skull bones. The child present following clinical features: short anterioposterior diameter with high forehead and flat occiput, flat facies, shallow orbits, proptosis, hypertelorism, small nose, maxillary hypoplasia, a cleft palate, low set ears, and cutaneous syndactyly of the fingers and toes. The neuroimaging of the head revealed craniosynostosis of the skull bones. The diagnosis of Apert syndrome was confirmed by clinical manifestations and paraclinical investigations. The treatment of Apert syndrome is directed toward the specific symptoms that are apparent in each individual.
Conclusions. Clinical and genetic approaches during genetic counseling combined with a number of new methods of neonatal diagnosis in patients with Apert syndrome can reduce the frequency of chromosomal abnormalities and congenital malformations.
Key words: Apert syndrome, congenital malformations, craniosynostosis, syndactyly.

DEPARTMENT OF ORTHOPEDICS AND TRAUMATOLOGY

27. RECONSTRUCTION OF THE SCALP DEFECT WITH THE TRAPEZIUS MUSCLE FLAP (CLINICAL CASE)

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Background. Many patients with complex scalp and skull defects can benefit from scalp reconstruction using the trapezoid flap, reducing the morbidity of the donor site. The trapezius flap was first reported by Nakajima and Fujino in 1984. It was originally described as a myocutaneous or muscle flap, and it has also been used as a free flap. The blood that supply the trapezium muscle and the skin is mainly from the superficial and descending branches of the transverse cervical artery as well as the occipital artery.
Case report. This work presents the clinical case of a 65-year-old woman who was diagnosed with cornified pluristrative squoamos cancer with bone destruction and invasion to confluence sinus and left transverse venous sinus, who underwent the occipital extra-intracranial tumor removal. She addresses to our clinic with a massive defect of 10 by 18 cm. After making the operative planning, we decided to solve the case with distal trapezius muscle flap.
Conclusions. The decision of the reconstructive technique should be taken into account with regarding to its consequences on the affected anatomical structures, the personal pathological antecedents and pre-existing lesions at the level of the donor area.
Key words: reconstruction of scalp defect, trapezius muscle, clinical case

28. SURGICAL TREATMENT OF FRACTURES IN PATIENTS WITH POLYTRAUMA, CASE REPORT.