

STROKE IN A YOUNG WOMAN

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Background. Stroke in young patients (<50 years old) represents a diagnosis and therapeutic challenge, given the multitude of etiologies and mimics of the disease. Pregnant women have a higher risk of stroke during the entire pregnancy, especially in the postpartum period.

Case report. We present the case of a young woman of 35 years old, admitted to the neuroemergencies department, with previous transient amaurosis, single episode of complex motor seizures the week before admission, moderate right sided hemiparesis, and temporal-spatial disorientation. Anamnesis reveals an emergency c-section 12 days before hospitalization at 36 weeks of pregnancy. Past medical history – unremarkable. Admission neurological state: awake, alert, disoriented in time, and space. Intact cranial nerves. Diminished strength in the right upper and lower limbs – 2/5 points; hypertonus and brisk deep tendon reflexes on the right; bilateral Babinski sign; mild hemihypoalgesia on the right side; temporal and spatial disorientation, cognitive decline (MMSE 15p). No meningeal signs. To exclude a possible posterior circulation ischemic stroke, a brain computed tomography was performed showing some diffuse occipital lobe lesions suggestive for encephalitis. Further investigation by 3T brain MRI showed diffuse, bilateral, white matter lesions of possible inflammatory or toxic-metabolic etiology. Posterior Reversible Encephalopathy Syndrome (PRES) diagnosis was established and targeted treatment performed. Two weeks later we noticed complete resolution of the motor deficit (patient walking alone without support), the patient was alert, oriented in time, space and herself, the cognitive function improved (MMSE 25 p) with home discharge. Normal follow-up MRI (1 month) was obtained.

Conclusions. Stroke should be excluded in post-partum women given the higher incidence in this group of population. PRES syndrome is a benign stroke-mimic that should be suspected in the appropriate clinical and imaging context for correct management of the pathology.

Key words: PRES syndrome, stroke mimics in youth, stroke in women.

24. SPONTANEOUS PNEUMOTHORAX AFTER A RESPIRATORY DISTRESS SYNDROME – A CASE REPORT

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Background. Respiratory distress syndrome (RDS) of the newborn is caused by pulmonary surfactant deficiency in the lungs of neonates which leads to alveolar collapse and noncompliant lungs. It can be primary or secondary, due to meconium aspiration or Group B Streptococcus (GBS) infection. RDS is usually diagnosed with a combination of clinical signs and/or symptoms (apnea, cyanosis, grunting, inspiratory stridor, nasal flaring, poor feeding, and tachypnea), chest radiographic findings, and arterial blood gas Results. In near term or term infants with great respiratory effort, RDS can be complicated with spontaneous pneumothorax.

Case report. A 2700 g male neonate was admitted to the neonatal intensive care unit (NICU) of Mures County Emergency Hospital with respiratory distress syndrome. Baby was vaginal born at a gestational age of 39/40 weeks at Ludus Emergency Hospital. Apgar score was 10/10 at the 1

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 parenchymal 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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