PP-086 NEUROTROPIC AUTOANTIBODIES AND CNS PATHOLOGY IN LOW-BIRTH-WEIGHT NEWBORNS

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Aim Perinatal central nervous system (CNS) damage encompasses a spectrum of diverse brain injuries occurring during pregnancy, childbirth, and the early days of a child's life. The identification of autoantibodies to organs and tissues represents a contemporary approach for the preclinical diagnosis of functional and organic changes. This study aimed to discern the predisposition to CNS pathology in low-birth-weight newborns through immunochemical screening.

Material and Method A total of 76 newborns were examined, with Group 1 comprising 33 infants whose weight aligned with gestational age, and Group 2 consisting of 31 infants with low birth weight. A comparison group was formed by healthy full-term newborns. The study assessed the levels of 12 types of IgG autoantibodies targeting brain cell antigenic components and receptors. The determination of autoantibodies was accomplished using the solid-phase IFA method with the 'ELI-Neuro-12-Test' test system developed by Immunculus in Russia.

Results Analysis of the results revealed pathologically elevated levels of neurotropic autoantibodies in low-birth-weight newborns. Specifically, autoantibodies to NF-200, GFAR, S100B, MBP (Myelin Basic Protein) and all studied receptor types showed heightened levels, indicating potential disruptions in the CNS. The MBP autoantibody levels were notably increased, pointing towards disturbances in axon myelination in conducting pathways. Furthermore, autoantibodies targeting cholinergic receptors demonstrated significant elevation, placing them in the 'red zone' for both Group 1 and 2, with a significant difference from full-term healthy infants. Elevated autoantibodies to serotonin, opioid, and beta-endorphin receptors suggested potential receptor damage, serving as precursors to cognitive and emotional-volitional sphere disturbances, impaired learning abilities, and a lowered pain perception threshold.

Conclusions Nhis study highlights a substantial degree of nerve tissue damage in low-birth-weight newborns. These findings hold valuable implications for the development of individualized rehabilitation programs for these vulnerable infants, emphasizing the importance of early intervention and targeted care.

PP-087 IMPLEMENTATION OF ADVANCED PAEDIATRIC LIFE SUPPORT COUSE IN BOSNA AND HERZEGOVINA

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Aim The establishment of Intensive Care at the Pediatric Clinic in Sarajevo was followed by the implementation of the following self-educational programs: Advanced Pediatric Life Support (APLS) Course, Neonatal Resuscitation Program (NRP) and PCEP-Perinatal Continuing Education Program (PCEP).

Material and Method The APLS course provides the knowledge and skills needed to identify, effectively treat, and stabilize children in life-threatening emergencies, using a structured, sequential approach

Results The APLS course has been practiced around the world and since 2000 it has also been implemented in Bosnia and Herzegovina. The advanced resuscitation and care of life-threatening children was successfully attended by 506 general practitioners, pediatricians and emergency physicians

Conclusions The set goals are that all those working with children and young people should have the appropriate knowledge and skills in order to meet their specific needs.

PP-088 NEONATAL ACUTE LEUKEMIA PRESENTING WITH SKIN INVOLVEMENT

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Aim Neonatal leukemia is a rare disease with an incidence rate of 1–5 per 1000000 live births. Leukemia cutis occurs in 25–30% of infants with leukemia. Neonatal leukemia cutis presents within the first four weeks of life. It often has a 'blueberry muffin baby' appearance of magenta-colored nodules affecting almost any skin area. Here, a case of neonatal leukemia with blueberry muffin was presented due to its rarity.

Material and Method We report a 20-day-old male baby with a blueberry muffin appearance and decreased activity. He was born at 39 weeks gestation by cesarean section as APGAR of 8–10 and a weight of 4020 g from a 25-year-old gravida 1



Abstract PP-088 Figure 1 Blueberry muffin.

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mother. The patient had no additional features in his medical and family history. Physical examination revealed a weight of 4020 g (50–90p), a height of 52 cm (50–90p), a head circumference of 37 cm (90p), blueberry muffin rash (figure 1) on the trunk and proximal extremities, hepatosplenomegaly. Laboratory studies revealed a white blood cell count of 1048x109/L, hemoglobin of 8.5 g/dL, and platelet count of 49.6x109/L. Diffuse blastic cells were observed in the microscopic examination of the peripheral blood smear.

Results Erythrocyte and platelet transfusions were performed. The patient underwent leukapheresis once, and the white blood cell count was reduced to 325x109/L. The B-acute lymphoblastic leukemia was detected in the patient according to diagnostic tests. The 11q23 translocation was detected positive in the genetic tests. The patient was started on a chemotherapy protocol. The patient, who did not respond adequately to chemotherapy and developed septic shock and respiratory distress during follow-up, died at the age of 55 days.

Conclusions Congenital leukemia is a disease that is rare in the first month of life and has a poor prognosis. Skin lesions can often be the initial symptom. Skin findings in infancy should be evaluated from this perspective.

PP-089 ATYPICAL HARLEQUIN COLOR CHANGE BETWEEN UPPER AND LOWER HALVES BODY CASE REPORT

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Aim

Material and Method HCC of the newborn, known also as Intractable vascular autonomic dysregulation (Harlequin phenomenon), is a transient unilateral erythema that is seen in up to 10% of healthy newborns and was first described by Neligan and Strang in 1952. HCC is macular and blanchable displaying a sharp midline cutoff. It is often gravity dependent. HCC typically presents between 2ndand 5th day of life, but some infants may present with episodes up to the third week of life. We herein report a case of full term male who had Harlequin Color Change between upper and lower halves of his body.

Results 39+1w singleton pregnancy boy born by spontaneous vaginal delivery without any antenatal, intranatal issues had a bluish lower half of the body and pale upper half which lasted 4 hours and resolved spontaneously without any other symptoms. In 3rdweek of life he presented to the hospital with virus infection, bluish discoloration was noted around the umbilicus and abdomen distension. ECG, ECHO, CSF analysis and abdominal ultrasound were all normal. Patient was discharged home and presented later for troponin level follow-up which was normal. The differential diagnosis includes port-wine stain, nascent hemangioma of infancy, cutis marmorata, cutis marmorata telangiectatica congenita, CHILD syndrome, Acrocyanosis, Drug reaction.

Conclusions The exact etiology of HCC is unknown, but the evidence points to sympathetic autonomic dysfunction in the control of the capillary tone, most likely secondary to hypothalamic immaturity in the newborns. Most cases are idiopathic in healthy newborns. HCC has also been noted in the setting of prematurity, low birth weight, hypoxia, infusion of prostaglandin E1(PGE1), neonatal meningitis. Learning points: Harlequin phenomenon occurs in healthy newborn. Recognition and education of this skin disorder are important as HCC is a surprising and alarming occurrence for parents, so reassurance about the benign nature of the condition, and to avoid additional diagnostic procedures and unnecessary treatments. Beyond the group age the search for an underlying cause should be vigorously pursued.

PP-090 BRAIN-LUNG-THYROID SYNDROME: NEONATAL PHENOTYPE OF DE-NOVO NKX2.1-RELATED DISORDER

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Aim We present a rare, genetically heterogenous, de-novo NKX2.1-related disorder in a neonate

Material and Method Brain-lung-thyroid syndrome, is a rare genetic syndrome.1 It is caused by autosomal dominant mutations in the NKX2-1 gene (previously TTF-1), on chromosome 14q13, which encodes for a particular transcription factor, critical for the development of the brain, lung and thyroid.2 It is a genetically heterogeneous condition, with over 30 different causative mutations identi ed.3 The syndrome is also referred to recently as NKX2.1-related disorders, due to the varied manifestations noted in heterozygous mutations, including benign hereditary chorea[BHC] which is a hallmark feature, while some can develop choreoathetosis, tremors, motor & vocal tics as well.4 Classic triad of the syndrome includes Congenital hypothyroidism, Benign hereditary chorea and Respiratory distress syndrome in infancy. Approximately 50% of the patients have a complete triad involved, while about 30% are affected by neurological and hypothyroidism phenotype and about 13% may only present with neurological phenotype. The overall prognosis varies depending on the presenting phenotype and severity of symptoms with respiratory involvement having the most impact on survival in some patients.5 We present a term neonate admitted in the neonatal intensive care unit for respiratory distress with incidental finding of macroglossia leading to diagnosis of congenital hypothyroidism with hypertonia at birth, with childhood Interstitial lung disease [figure 1]; eventually diagnosed as heterozygous for the pathogenic deletion of the entire coding region of NKX2-1 gene including the promoter region, de-novo mutation.



Abstract PP-090 Figure 1 CT Thorax: Ground glass appearance suggestive of childhood interstitial lung disease

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