Multiple intestinal atresia (MIA) is a congenital malformation disorder in which atresia occurs at multiple levels throughout the gastrointestinal tract, most commonly small bowel. Frequently it presents as an isolated anomaly, but also as a part of a complex inherited disorder caused by homozygous or compound heterozygous mutation in tetratricopeptide repeat domain 7A (TTC7A) gene. These patients usually have associated mild or severe combined immunodeficiency.

Herein we report an infant with MIA and associated combined immunodeficiency (CID). The female child was born at term by vaginal delivery to a 26-year-old G5P3 mother. The parents are healthy, non-consanguineous couple of Albanian origin. Pregnancy was complicated by polyhydramnios. Fetal ultrasound at 19 weeks of gestation revealed dilated bowel loops. Postpartal abdominal X-ray showed signs of gastric atresia.

Surgery revealed widespread atresias, extending from stomach to cecum, and atrophic microcolon. Multiple small bowel resections with end-to-end anastomosis, and ileostomy were done. Histopathological examination showed pseudostratification of the epithelium, increased enterocyte apoptosis, inflammatory eosinophilic infiltrate in lamina propria, decreased small bowel villi, and disrupted apical-basal polarity. At two weeks of age GI obstruction recurred, and she underwent jejunal resection with end ileostomy. The residual small bowel length measured 45cm. Immunological investigation revealed CID: T cell lymphopenia affecting all subsets with lower B cell, natural killer cell count and impaired mitogen response. She experienced several septic episodes. At two months, allogeneic bone marrow transplantation was successfully performed. MIA-CID was suspected and genetic testing confirmed homozygous pathogenic variant in the TTC7A gene (c.315_318del/c.315_318del) causing premature translational stop signal that result with absent protein production. She is now 4 months old, has failure to thrive and parenteral nutrition-induced cholestasis and continues to be dependent on parenteral nutrition for a short gut.

MIA-CID is a rare hereditary disease with about 50 reported patients in literature. Abundant expression of TTC7A gene in thymus and colon and its critical role in intestinal and immune homeostasis can explain this severe phenotype and almost invariably poor prognosis. Clinical course may be more favourable if the bone marrow transplantation is done in the first three months of life. Therefore, immunological and genetic testing should be performed in every neonate born with MIA.

123

KAPOSIFORM HEMANGIOENDOTHELIOMA WITH KASABACH-MERRITT PHENOMENON IN A NEONATE: SUCCESSFUL TREATMENT WITH SIROLIMUS

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Kaposiform hemangioendothelioma (KHE) is extremely rare, life-threatening vascular tumor with estimated incidence of 0.071 cases per

100.000 children. It is notably associated with Kasabach-Merritt phenomenon (KMP), a condition characterised by

profound thrombocytopenia, hypofibrinogenemia, and elevated markers of coagulation activation (D-dimers or fibrin degradation products). Mortality is highly associated with degree of coagulopathy. Patients diagnosed prenatally appear to have increased disease severity. Optimal therapy for KHE is not known. Oral steroids and vincristine for patients with inoperable tumors is most commonly reported, but is associated with limited response and significant side effects.

Sirolimus (rapamycin), a mammalian target of rapamycin (mTOR) inhibitor, is currently being tested in a prospective phase II clinical trial.

Here we present a full-term newborn prenatally diagnosed with giant vascular tumor, affecting the lateral neck. He was delivered by elective C- section and initially presented with a firm, violaceous neck tumor 14x10 cm.

Following the birth, he rapidly evolved to severe KMP. Despite endovascular embolisation performed immediately after birth, his coagulopathy worsened to life threatening hemorrhage (platelets 8,000/μL, fibrinogen 0.8 g/L, D-dimer >10 mg/L, PT-INR 1.36, aPTT 26 s, Hb 80 g/L, Hct 23%), necessitating aggressive blood products replacement to maintain hemostasis.

MRI and laboratory investigations strongly suggested the diagnosis of KHE/KMP. Biopsy was not attempted because of the potential risk of hemorrhage. He was vitally endangered with rapidly enlarging tumor size compromising airway patency, as well as the worsening coagulopathy triggered by platelet entrapment. As there were no response to corticosteroid and propranolol therapy, we elected to start sirolimus (0.8 mg/m² per dose twice daily) along with tapering parenteral corticosteroids. This therapeutic approach led to remarkable resolution of consumptive coagulopathy with stepwise regression of tumor size. After 14 months of follow-up he remains on sirolimus without toxicity, the tumor is barely visible, and MRI showed remarkable reduction in the tumor size.

Currently, no standard treatment guidelines exist for KHE/KMP because of their rare nature and lack of prospective trials. Our experience adds to a growing body of evidence suggesting that sirolimus in the treatment of high-risk KHE/KMP patients might be an efficacious and safe treatment option.

124

VEIN OF GALEN ANEURYSMAL MALFORMATION: CASE SERIES OF 14 NEONATES IN CROATIAN NATIONAL REFERRAL CENTRE FOR NEONATE AND PEDIATRIC INTENSIVE CARE

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Vein of Galen aneurysmal malformation (VGAM) is a complex congenital cerebro-vascular malformation. Despite some controversy, the overall prognosis of VGAM has improved over time.

We present a series of VGAM patients with hyper-dynamic heart failure during the neonatal period treated in our Department in the past 20 years.

All cases diagnosed with chorioidal VGAM in a single tertiary centre between 2001 and 2020 were ascertained from

medical records. All infants underwent postnatal cardiac ECHO, electroencephalogram and brain MRI, along with calculation of Bicetre severity score at admission. Embolization procedures were analysed based on timing and presence of peri- and post-procedural complications. Long-term survivors had standard neurocognitive assessment.

For the purpose of outcome analysis we divided patients into two period cohorts: 2001-2010 and 2011-2020.

Overall fourteen out-born neonates were diagnosed with chorioidal VGAM. Bicetre neonatal score were similar in both chronological sub-groups. First cohort comprised six neonates. Prenatal diagnosis of VGAM was obtained in one patient. Four newborns died owing to refractory heart failure without neuroembolization procedure. Two survivors underwent the intervention at fourth and eleventh day, respectively. Post-procedural analysis revealed minor intraventricular bleeding and neonatal seizures in one patient that eventually has been lost to follow up. Other survivor developed ischaemic lesions and hydrocephalus. Ventriculo-peritoneal drainage was performed.

Severe neurodevelopmental delay and visual impairment were observed on follow-up. Second cohort comprised eight neonates, all prenatally diagnosed. Three patients died: at the age of five (intra-procedural rupture of aneurism), fifteen (multi-organ failure), and 205 days (pulmonary hypertension and congestive heart failure). All infants undergone the first embolization within the first ten days of life. Satisfactory immediate outcome including control of cardiac failure was achieved in six patients. Post-procedural analysis revealed minor intraventricular bleeding (n=2), ventriculomegaly (n=2), presence of ischaemic lesions (n=2), neonatal seizures (n=2) and abnormal encephalogram (n=2). Normal neurodevelopment was observed in four, and mild neuro-developmental delay was observed in one patient on follow-up.

Given a small sample size it is difficult to quantify the difference between the two groups in a statistically relevant manner. However, the observed difference in survival and long-term outcome between the cohorts are most probably related to an overall improvement of prenatal diagnosis, intensive care management and embolization techniques. Early neuroembolization may decrease the risk of refractory cardiac failure and improve the long-term neurocognitive outcome.

125

IS THERE A GOOD CORRELATION OF MAGNETIC RESONANCE IMAGING AND CRANIAL ULTRASOUND IN BRAIN IMAGING IN PRETERM INFANTS AT TERM AGE?

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The aim of this study was to compare MRI findings in pre-term infants at term age with cranial ultrasounds as predictors in the assessment of neurodevelopment outcomes and need for early habilitation procedures.

The study included 64 premature infants gestational age under 32 weeks who were admitted in NICU at University Hospital Centre Zagreb between years 2013. and 2016. and underwent brain MRI examination. 53,12% examinees were boys and 48,8% were girls. Mean gestational age was 29,0 (+/-3,2) weeks, and mean birth weight was 1336 (+/- 466) g. Reanimation at birth was necessary in 50% of examinees. Some method of mechanical ventilation (invasive or

noninvasive) was used in 70% of examinees, in duration of approximately 22 (1-150) days. Cranial ultrasound scans were performed at the age of one day, four days and seven days and then once a week and at term age. Brain MRI examinations were performed at gestational age of 32 weeks and at term age. A statistical analysis of the correlation of ultrasound findings and brain MRI examinations at term age was performed. Cranial ultrasound findings were classified into four groups by presence of IVH or leukomalacia: (0-normal finding; 1-mild hyperechogenicity or IVH grade I/II; 2-IVH with ventricular dilatation or presence of periventricular cysts; 3-ventriculomegaly or hydrocephalus and severe periventricular leukomalacia) and in two groups by presence of talamic lesions (present or not present).

MRI findings were classified as normal, mildly abnormal (mild gliosis); moderately abnormal (ventricular dilatation with gliosis, talamic lesions) and severely abnormal group (severe gliosis/PVL and hydrocephalus).

Group of examinees with Apgar score < 5 after 1st minute had moderately to severely abnormal MRI findings. Mechanically ventilated premature infants showed statistically significantly higher ($p < 0,001$) correlation with severe MRI abnormalities.

Comparison of ultrasound and MRI findings didn't show good correlation (Kendall Tau= 0,715; $p < 0,05$). Hi-square test showed ultrasound scans statistically significantly overestimate the number of premature infants with lesions (presence of IVH or leukomalacia). The number of talamic lesion was also overestimated by ultrasound scans (Hi-square test 8,824; $p = 0,004$).

But, correlation between methods considering moderately to severely abnormal MRI findings was better (Hi-square test 13,53; $p = 0,003$).

Magnetic resonance imaging is superior method comparing to brain ultrasound, but despite that brain ultrasound is still unavoidable as standardized routine screening method. Together they form the gold standard in the assessment of neurodevelopment outcomes and need for early habilitation procedures in premature infants.

126

A BROKEN NEC: CASE STUDY OF IDIOPATHIC SUPERIOR MESENTERIC VEIN THROMBOSIS IN A PREMATURE NEONATE

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Introduction Neonates are the pediatric population at highest risk for development of venous thromboembolism.¹ Intravascular catheters are a major risk factor.

Other risks include prematurity, neonatal asphyxia and sepsis.² Neonatal superior mesenteric venous thrombosis has not previously been described in medical literature.

Case A preterm female infant was born by emergency Caesarean section at 28 weeks' gestational age, weighing 760g. The indication was severe symmetrical intrauterine growth restriction and absent end diastolic flow.

Her mother was an Irish 23-year-old primigravida, with a background of severe depression and smoking. The neonate required continuous positive airway pressure (CPAP) and artificial surfactant. She required an umbilical venous catheter