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.

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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/ μ L, 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.

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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