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 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 GSP3 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 4.5 cm. 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, allo-geneic 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 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.

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

1Sindić Iva Vukšić*, Tomislav Čaleta, Petra Čepina, Dorotea Ninković, Andrea Dasović Buljević, Boris Filipović-Grčić, Vesna Benjak, David Oezretić, Ana Petrović Gluščić, Ernest Bilč, Marko Radlo, Nada Sindić Dessard, Ruža Grizelj, University Hospital Zagreb, University of Zagreb, School of Medicine
2Department of Pediatrics, University Hospital Centre Zagreb, Zagreb, Croatia; 
3Department of Diagnostic and Interventional Radiology, University Hospital Centre Zagreb, Zagreb, Croatia; 
4University of Zagreb, School of Medicine

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