

Keywords: Thrombotic Microangiopathy, Genetic Disease, Haemolytic anaemia, Kidney Injury, Thrombocytopenia.

Background: Atypical haemolytic uremic syndrome (aHUS) is a severe, rare genetic disease presenting as systemic thrombotic microangiopathy (TMA). Patients typically exhibit non-immune haemolytic anaemia, thrombocytopenia and organ dysfunction, most frequently renal, with high mortality. Prompt diagnosis and treatment of aHUS is challenging but crucial.

Objectives: The aim of the study is to identify potential aHUS patients within the frame of a screening program at Pauls Stradiņš Clinical University Hospital.

Methods: Over 12 months, the screening enrolled patients with TMA like signs, including anaemia (HGB <90 g/l), acute renal failure (creatinine >130 mmol/l) and thrombocytopenia (PLT <70 000 or >25% platelet count reduction within 48 hours). 10 patient were assigned to genetic testing to rule out aHUS. During the screening program, ADAMTS13 laboratory test was implemented to improve aHUS diagnosis. Demographics and clinical data were analysed using IBM SPSS.

Results (Preliminary): The screening included 132 patients, 39.4% female and 60.6% male, with a mean age of 64.4 years (SD ± 14.27). In-hospital mortality was 51.5%, with a median hospital stay of 17 days. The most common TMA-like pathologies were infections (28.5%), oncologic diseases (19.8%) and bleeding (9.3%). No aHUS patients were found during the screening period. Of the 10 genetic tests, one showed heterozygous ADAMTS13 gene.

Conclusions: In the implemented screening program, no patient with confirmed aHUS was detected, highlighting the challenge to diagnose aHUS within a wide range of similar differential diagnoses. TMA-like diseases are linked to high in-hospital mortality.