

Congress Abstract

Differential Diagnostic Significance of a Comprehensive Laboratory Profile in Verifying Atypical Hemolytic Uremic Syndrome in Children in Kazakhstan

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ABSTRACT

Introduction and Aim: Atypical hemolytic uremic syndrome (aHUS) is an ultra-rare orphan disease that requires differential diagnosis from other thrombotic microangiopathies (TMAs). The aim of this study was to determine the diagnostic value of ADAMTS13 activity levels, anti-complement factor H (anti-CFH) antibody titers, and genetic screening in verifying aHUS subtypes among the pediatric population in Kazakhstan.

Materials and Methods: A retrospective analysis was conducted on 15 children with aHUS (8 girls and 7 boys) from 8 regions of Kazakhstan between 2019 and 2025. The mean age was 7.2 ± 3.8 years. ADAMTS13 activity was measured in 7 patients (46.6%), and anti-CFH titers were assessed in 10 patients (66.7%). In the remaining patients, these tests were not performed because they had received fresh frozen plasma transfusions prior to testing. Genetic testing was completed for 10 out of 15 children (66.7%); the remaining 5 patients (33.3%) died before testing could be carried out.

Results: In all patients tested ($n=7$, 100%), ADAMTS13 activity was $>10\%$, effectively ruling out thrombotic thrombocytopenic purpura (TTP). Autoimmune aHUS associated with anti-CFH antibodies was diagnosed in 4 out of 10 children (40%), of whom 3 were of Asian descent and 1 was Caucasian. Genetic testing in 10 children revealed pathogenic variants in complement system genes in 2 patients (20%): one child had a *CFHR3/CFHR1* deletion, and another had a *CFHR1/CFHR4* microdeletion; both exhibited high anti-CFH titers (up to 11,490 U/mL). Additionally, one child was found to have a clinically insignificant heterozygous autosomal recessive mutation in the *ADAMTS13* gene.

The comprehensive laboratory workup allowed for the stratification of aHUS subtypes and personalized therapy. Patients with idiopathic forms received complement-blocking therapy with eculizumab alone, while children with anti-CFH antibodies received combined treatment with eculizumab and immunosuppressive agents.

Conclusions: This is the first comprehensive diagnostic data report on pediatric aHUS in Kazakhstan. Genetically determined aHUS was identified in 20% of the cohort. Anti-CFH-associated aHUS appears to occur more frequently in the Asian population (reported as 50–60% in Indian cohorts by Khandelwal et al.) compared to Caucasians (5–25%). Our findings represent an intermediate frequency between European and Asian populations, which is consistent with the mixed ethnic composition of the Kazakhstani cohort.

Keywords: Atypical Hemolytic Uremic Syndrome; Children; ADAMTS13; Anti-Complement Factor H Antibodies; Genetic Diagnosis