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PRINT: ISSN 0972-3757 ONLINE: ISSN 2456-6330

Int J Hum Genet, 26(3): 144-151 (2026)

DOI: 10.31901/24566330.2026/25.02.938

CFH/CFHR1 Gene-Linked Mutations Causing Atypical Hemolytic Uremic Syndrome: A Case Report and Literature Review

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KEYWORDS Acute Kidney Injury. Complement Factor H. Hemoptysis. Monoclonal Gammopathy of Renal Significance. Plasma Exchange. Thrombotic Microangiopathy

ABSTRACT An unusual case of atypical hemolytic uremic syndrome (aHUS) featuring both diffuse alveolar haemorrhage (DAH) and renal-significant monoclonal gammopathy (MGRS) is reported. The researchers describe the clinical course, laboratory findings, genetic analysis, and treatment response of a 38-year-old male patient, supplemented by a review of reported cases with similar genetic variants. The patient exhibited haemolytic anaemia, thrombocytopenia, acute kidney injury, and haemoptysis. Whole exome sequencing identified heterozygous mutations in both CFH (c.3547T>A; p.Trp1183Arg) and CFHR1 (c.700G>C; p.Glu234Gln). The condition improved following plasma exchange and high-dose methylprednisolone, with no recurrence over six months. The researchers present an uncommon manifestation of atypical haemolytic uremic syndrome associated with concomitant CFH and CFHR1 mutations. In regions where eculizumab is unavailable, plasma exchange and steroid therapy remain effective.