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Diagnosis of Atypical Hemolytic Uremic Syndrome in Post-Transplant End-Stage Kidney Disease Patient via Identification of CFH-22 Deletion: A Case Report

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Case Study: Atypical hemolytic uremic syndrome (aHUS), a rare form of thrombotic microangiopathy (TMA), is caused by dysregulation in the alternative pathway of the complement system and results in microvascular damage. We herein present a case of recurrent aHUS with a heterozygous deletion of complement factor H (*CFH*) exon 22 diagnosed after kidney transplantation (KT). A 45-year-old man with end-stage kidney disease of unknown etiology underwent KT. A month after transplantation, allograft dysfunction was observed, accompanied by Coomb's negative hemolytic anemia and thrombocytopenia. Peripheral blood smear revealed schistocytes (**Fig. 1A**). ADAMTS13 activity was 63.6% and shiga toxin was not detected. The allograft biopsy revealed focally proliferative glomerulonephritis with no pathological evidence of TMA, except the electron microscopy finding indicating an early stage of TMA (**Fig 1B, 1D and 1E**). Meanwhile, his past native kidney biopsy showed arteriolar thrombotic occlusion (**Fig. 1C**). He did not respond to plasmapheresis and began hemodialysis. Clinical exome sequencing was performed for a comprehensive analysis of multi-genes associated with aHUS. A variant of unknown significance, p.(His40Pro), was found in complement factor I (*CFI*), and none was found in *CFH* (**Fig. 2A**). In subsequent multiplex ligation-dependent probe amplification (MLPA) analysis of the *CFH* region, a pathogenic heterozygous deletion was observed in *CFH* exon 22 and its downstream (**Fig. 2B**). Finally, he was diagnosed with recurrent aHUS based on laboratory findings, underlying genetic variant, and past histopathology. After initiation of eculizumab, a humanized monoclonal antibody to C5, TMA did not recur and allograft function maintained stable without dialysis over 8 months (**Fig. 1A**). Post-transplant TMA is a rare but important cause of kidney allograft dysfunction. Active genetic testing should be conducted in patients suspected TMA to diagnose aHUS. Early treatment with eculizumab may prevent allograft failure and preserve allograft function in patients with aHUS.

Figure 1.