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Atypical Presentation of Upshaw Schulman Syndrome: A Case Report

Goutham Ravipati, BS
Thomas Jefferson University, goutham.ravipati@jefferson.edu

David Strayer, MD, PhD
Thomas Jefferson University, david.strayer@jefferson.edu

Douglas Drelich, MD
Thomas Jefferson University, douglas.drelich@jefferson.edu

Atypical Presentation of Upshaw Schulman Syndrome: A Case Report

Goutham Ravipati BS, David S. Strayer MD, PhD, Douglass A. Drelich MD

Thomas Jefferson University, Department of Medicine, Division of Hematology

Thrombotic thrombocytopenic purpura (TTP) is a rare coagulation disorder with a typical clinical presentation of low platelets and excessive clotting. Mortality for this disorder may be high if untreated and therefore necessitates a high clinical suspicion. Here we describe a 46-year-old African American woman with a past medical history of multiple cerebrovascular accidents presenting to hematology after a suspected diagnosis of TTP. Presumptive diagnosis of acquired TTP called for treatment with IVIg, but a diagnosis of congenital TTP (Upshaw Schulman syndrome) was made after testing showed a lack of ADAMTS13 antibodies. Treatment with fresh frozen plasma (FFP) showed increase in ADAMTS13 levels which further supported the diagnosis of Upshaw Schulman syndrome. Genomic analysis of ADAMTS13 showed normal sequencing and was not consistent with the diagnosis of Upshaw Schulman syndrome. No prior literature provides an explanation for these findings. This new presentation of a well-classified disease suggests a potential subcategory of TTP that may traditionally be misdiagnosed. Potential explanations for this disease presentation may include IgA-mediated TTP, epigenetic changes of ADAMTS13 expression, or limitations of current testing modalities.