

CONGENITAL THROMBOTIC THROMBOCYTOPENIC PURPURA: HOW TO MANAGE? - A CASE FROM REAL WORLD

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Introduction. Congenital thrombotic thrombocytopenic purpura (cTTP) is a rare genetic condition also known as Upshaw-Schulman syndrome with an estimated prevalence of 0.5-2.5 cases per million. It is an autosomal recessive disorder caused by pathogenic homozygous or compound heterozygous mutations in the ADAMTS13 gene that result in ADAMTS13 activity <10%. Since its proven safety and efficacy, recombinant ADAMTS13 (rADAMTS13) is functional to treat acute episodes or prophylaxis in cTTP which can be triggered by infections, trauma, pregnancy and major surgery. In this regard, the aim of this paper is to discuss pre and postoperative management of patients with cTTP, throughout monitoring hemolysis indices and administer prophylaxis with rADAMTS13.

Case report. In this paper we discuss the case of a 46 year old female who presented at our center in March 2026 in anticipation of elective surgery of left hemicolectomy and hepatic excision of suspected sigma cancer metastasis. From childhood she suffered from cyclic thrombocytopenia with jaundice, treated by plasma transfusion. In 2016, because of multiple uterine fibroids, she underwent transverse laparotomic uterine myomectomy complicated by massive hemorrhage due to severe thrombocytopenia which required intensive care. Subsequently several episodes of thrombocytopenia non responding to steroids had followed and in 2017 Coombs-negative hemolytic anemia occurred in addition to severe thrombocytopenia, treated with steroids and plasmapheresis, until complete resolution. Diagnostic workup revealed a severe deficiency of ADAMTS13 activity in absence of specific anti-ADAMTS13 antibodies. Therefore she underwent a molecular test resulting in the presence of homozygous biallelic mutation c.4078-2A>G in the ADAMTS13 gene that confirmed cTTP diagnosis. From April 2019 she started prophylaxis with fresh-frozen plasma (FFP) once per month with increased level of platelets and from September 2025 she started treatment with rADAMTS13 at a prophylactic dose of 40 UI/kg body weight (corresponding to 2 vials each one containing 1500 UI) every two weeks without any disease recurrence. As a precautionary measure, one vial of medicine was administered the day before the surgery. Fortunately the surgery was performed without any complications, though we recommended keeping sufficient supplies of rADAMTS13 and FFP to deal with a possible relapse of cTTP. From the day of the surgery the patient was tested for blood count, hemolysis indices and ADAMTS13 activity, every day during the hospitalization and twice per week for the first month after the surgery. According to international guidelines, we decided to perform a vial of rADAMTS13 when ADAMTS13 activity was < 10%. It resulted in an administration of rADAMTS13 every two days for the first week after surgery and one administration per week for the first month after surgery.

Conclusions. cTTP is a very rare genetic condition: scientific literature does not set an optimal level of pre and postoperative ADAMTS13 activity, in fact clinical monitoring and therapeutic management are left up to physician's personal experience. This case report evidences that rADAMTS13 is safe and improves the patient's QoL, especially while undergoing surgery. So if available we suggest intensifying rADAMTS prophylaxis (possibly weekly) and a close monitoring of ADAMTS13 levels which can be lower during the postoperative period.