myeloma (4). Parenchymal involvement may be in the form of mass compression or leptomeningeal involvement due to plasmacytomas (5). The most common cranial nerve involvements in the literature are the oculomotor nerve, abducens nerve and hypoglossal nerve (2). Involvement of these cranial nerves is most commonly due to plasmacytomas originating from the skull base and sinuses (6). When other cases of multiple myeloma with hypoglossal nerve involvement were scanned in the literature, it was seen that there was plasmacytoma or leptomeningeal involvement (7,8). In our case, it is a very rare condition in terms of the absence of a mass lesion, plasmacytoma and leptomeningeal involvement in the brain. When the case was re-examined after the literature scan, it was seen that there was a prominent paranasal sinus wall thickening on the right side that developed during the period when the patient's complaints began, without any mass lesion or leptomeningeal involvement. However, in a case report, a patient with a soft tissue mass in the right paranasal sinus had significant plasmacytomas at the skull base, and clinically, there was oculomotor, facial and hypoglossus nerve involvement (9). This situation suggests that involvement in the paranasal region may require differentiation from classical infection conditions in patients at risk and close monitoring in terms of intracranial events. Plasmacytomas of the skull base occur as an extension of plasmacytoma originating from the clivus, petrous part of the temporal bone or from the submucosa of the sinonasal and nasopharyngeal (extramedullary plasmacytoma) region. Extramedullary plasmacytomas are most commonly seen in the nasal and paranasal sinuses, nasopharynx, tonsils and larynx (10). Due to the rarity of extramedullary plasmacytomas, data on treatment and prognosis are limited. However, studies have shown the effectiveness of radiotherapy. There are publications showing that 30-50 gy radiation most effectively reduces tumor sizes caused by multiple myeloma (11)..In our case, radiotherapy and appropriate chemotherapy were initiated. Cranial nerve involvement is very rare in cases of relapsed refractory disease, and radiotherapy and combined chemotherapy are among the treatment options.

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Adult Hematology Abstract Categories

Platelet Diseases

PP 13

A RARE DIAGNOSIS IN ADULTS: HEREDITARY THROMBOTIC THROMBOCYTOPENIC PURPURA

Süleyman ARSLAN*, İlhami Berber, İrfan Kuku, Emin Kaya, Mehmet Ali Erkurt, Ahmet Sarıcı, Mehmet Özcan

İNÖNÜ University, Türkiye

Introduction: Thrombotic thrombocytopenic purpura (TTP) is a thrombotic microangiopathy (TMA) resulting from severely reduced activity of ADAMTS-13 (a disintegrin and metalloproteinase with a thrombospondin type 1 motif, member 13), a metalloproteinase responsible for cleaving von Willebrand factor (vWF). It is characterized by disseminated platelet-rich microvascular thrombi leading to organ ischemia, neurological abnormalities, renal dysfunction, thrombocytopenia, and microangiopathic hemolytic anemia (MAHA). Hereditary TTP (hTTP; also referred to as congenital TTP [cTTP] or Upshaw–Schulman syndrome) arises from pathogenic variants in the *ADAMTS-13* gene and follows an autosomal recessive inheritance pattern. Although extremely rare, it can be life-threatening. Patients with hTTP require special attention during certain life stages such as the neonatal period and pregnancy . In contrast to immune-mediated TTP (iTTP), which typically presents with a dramatic and acute onset, hTTP may manifest with a more insidious clinical picture including lethargy, impaired concentration, abdominal pain, and headache . Severe renal failure, although uncommon in iTTP, may occur in hTTP patients due to lifelong ADAMTS-13 deficiency, which can cause progressive accumulation of thrombi within the renal vasculature Hematologic examination may reveal pallor, purpura, and jaundice as signs of hemolysis, while laboratory findings typically show thrombocytopenia, unconjugated hyperbilirubinemia, elevated LDH levels, and decreased haptoglobin . Peripheral blood smear is often diagnostic, demonstrating schistocytes, nucleated red blood cells, and polychromatic red cells, consistent with intravascular hemolysis . Here, we describe the case of an 18-year-old patient with congenital TTP who was initially misdiagnosed with myelodysplastic syndrome (MDS) at an early age and received intermittent transfusions due to cytopenias. The aim of this case report is to raise clinical awareness regarding this rare and potentially fatal subtype of TTP, which can be rapidly and effectively treated if recognized early. In addition, it underscores the importance of reassessing patients at each presentation, even when a pre-existing diagnosis is available, and highlights the critical diagnostic value of peripheral blood smear, as the presence of schistocytes is pathognomonic for this condition. **Case report:** We present the case of an 18-year-old female patient, with no family history of hematologic disorders, who has been followed since childhood for anemia and thrombocytopenia. At the age of 18, she was diagnosed with congenital thrombotic thrombocytopenic purpura (TTP) and treatment was initiated. Her hematologic evaluation began in 2009, at the age of three, due to anemia and thrombocytopenia, during which she received platelet and red blood cell transfusions. Following a bone marrow examination, she was diagnosed with myelodysplastic syndrome (MDS). In 2018, she was also diagnosed with chronic kidney disease secondary to vesicoureteral reflux by the pediatric nephrology department, Türkiye. In 2024, at the age of 18, she presented to the emergency department with complaints of fatigue and dizziness. Laboratory tests revealed: WBC 5.36×10^9/L, Hgb 8 g/dL, Plt 8×10^9/L, INR 0.98, fibrinogen 211 mg/dL, creatinine 5.8 mg/dL, AST 21 U/L, ALT 15 U/L, uric acid 8.6 mg/dL, LDH 622 U/L, total bilirubin 2.5 mg/dL, direct bilirubin 0.35 mg/dL, and both direct and indirect Coombs tests were negative. The patient was admitted to the hematology clinic for further evaluation, Türkiye. Physical examination was notable only for mild pallor; no lymphadenopathy or organomegaly was detected. Peripheral

blood smear performed in our clinic demonstrated approximately 7–8% schistocytes, polychromatic erythrocytes, and thrombocytopenia, consistent primarily with thrombotic microangiopathy Review of the patient's previous laboratory records revealed episodes of anemia and thrombocytopenia, accompanied by elevated LDH, indirect bilirubin, and reticulocyte counts during these periods. ADAMTS-13 antigen, activity, and inhibitor levels were subsequently evaluated. The patient received red blood cell transfusions and 3 units of fresh frozen plasma. Laboratory results showed ADAMTS-13 activity of 23.61% (reference range: 40–130%), ADAMTS-13 antigen 0.06 IU/mL (reference range: 0.19–0.81), and ADAMTS-13 inhibitor 3.36 U/mL, with the inhibitor interpreted as negative (<12 U/mL), borderline (12–15 U/mL), or positive (>15 U/mL). Given the severely reduced ADAMTS-13 antigen and negative inhibitor, the patient was diagnosed with congenital TTP and initiated on biweekly therapeutic plasma infusion (10 mL/kg). Two weeks later, follow-up blood tests showed a platelet count of 219×10^9/L, and peripheral smear findings had completely normalized (Figure 3). At the subsequent follow-up, ADAMTS-13 tests were repeated, revealing activity <0.20%, antigen <0.01 IU/mL, and inhibitor 2.96 U/mL, consistent once again with congenital TTP. The patient continues to be followed and managed in our clinic. **DISCUSSION:** The aim of this case report is to raise awareness among clinicians about this rare syndrome, which, if accurately diagnosed, can be effectively treated, whereas misdiagnosis may lead to fatal outcomes. In neonates and children, clinicians may suspect congenital TTP in the presence of jaundice, hemolytic anemia, and thrombocytopenia. This rare syndrome was first described in 1960 by Schulman in an eight-year-old girl who experienced recurrent thrombocytopenia episodes responsive to plasma infusions [7]. In 1978, Upshaw reported a similar case in a 29-year-old patient with recurrent thrombocytopenia attacks associated with microangiopathic hemolytic anemia (MAHA), also responsive to plasma infusions, documenting multiple MAHA episodes often triggered by acute infections or stressors such as pregnancy or surgery [8].Classically, TTP is characterized by a pentad of fever, microangiopathic hemolytic anemia, thrombocytopenia, and variable renal and neurological dysfunction (observed in 20–30% of patients).However, this full presentation is often absent inmost patients [9,10].The current standard treatment for congenital TTP involves prophylactic or on-demand infusions of fresh frozen plasma (FFP) or plasma-derived factor VIII–vWF concentrates containing ADAMTS-13 for replacement therapy [11,12]. Until recently, no drug had been specifically approved for routine prophylaxis in patients with congenital TTP. Recombinant ADAMTS-13 received FDA approval in November 2023 for prophylactic or on-demand ADAMTS-13 replacement therapy in both adults and children with congenital TTP [13].Scully et al. [14] recently reported a phase 3 study comparing recombinant ADAMTS-13 with standard therapy for prophylaxis in congenital TTP patients. The study demonstrated that recombinant ADAMTS-13 is an effective prophylactic treatment approach for these patients. No safety concerns were reported, and no neutralizing antibodies against ADAMTS-13 were detected. Our patient had previously been diagnosed with MDS by apediatric hematology clinic and monitored with intermittent blood transfusions.MDS in children (≤18 years) is exceedingly rare, with an incidence of 1–4 cases per million [15].Therefore, we approached the initial diagnosis cautiously. During an MAHA episode, detailed investigations led to the diagnosis of congenital TTP. The patient responded well to fresh frozen plasma therapy. Currently, she continues biweekly FFP infusions at 10 mL/kg in our center, has not experienced further microangiopathic episodes, and remains under close follow-up.

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PP 14

A CASE OF X-LINKED THROMBOCYTOPENIA CONFUSED WITH IMMUNE THROMBOCYTOPENIA

Bengü MACİT, Arzu AKYAY, Yurday ÖNCÜL

INONU UNIVERSITY TURGUT OZAL MEDICAL CENTER

Wiskott-Aldrich Syndrome (WAS) is an X-linked disorder characterized by severe thrombocytopenia, eczema, humoral and cellular immunodeficiency, and an increased susceptibility to lymphoid malignancies. The milder form is X-linked thrombocytopenia (XLT), characterized by persistent thrombocytopenia with minimal or no signs of eczema or immunodeficiency. An eight-year-old male patient was admitted to the hospital at six months of age with complaints of coughing and wheezing. Bone marrow aspiration was performed after thrombocytopenia was detected in a complete blood count, and the bone marrow was interpreted as technically hypocellular. Immun thrombocytopenia was suspected, and follow-up was recommended. Since he had no bleeding, he did not return to our clinic. When the patient was scheduled circumcision at the age of seven, his platelet count was 30,000/mm3, so he was referred to our clinic from anesthesia. In his medical history, he had severe eczema as a baby, which later improved, and he has no history of bleeding. In his family history, his uncles also had low platelet counts, and three of his uncles underwent splenectomy for this reason, after which their platelet counts returned to normal. On physical examination, the patient had no signs of dermatitis, petechiae, purpura, or ecchymosis. The liver and spleen were palpable at 2 cm. Laboratory tests: Hgb: 12.9 g/dl, Htc: 36.7%, white blood cells: 12,360/mm3, platelets: 30,000/mm3, MPV: 9.1 fl (normal), and platelet size appeared normal in the peripheral smear. Sedimentation was normal, and immunoglobulin values were normal for age. Based on these findings, a preliminary diagnosis of XLT was considered, and WAS genetics were sent. In the WAS gene, a c.223G>A (p.V75M) hemizygous mutation was detected. The patient was diagnosed with XLT based on clinical findings and this mutation in the WAS gene. Eltrombopag treatment was initiated but was ineffective, so a splenectomy was performed.Subsequently, the platelet count reached 323,000/mm3. No decrease was observed during follow-up. This situation can cause confusion with immune thrombocytopenia (ITP) and delay the diagnosis of XLT. Since XLT patients present with a milder clinical picture than