Case Report | Internal Medicine: Hematology

Acute vision loss as an initial manifestation of thrombotic thrombocytopenic purpura in pregnancy: a case report

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Background: Thrombotic thrombocytopenic purpura (TTP) is a medical emergency manifested by a pentad of thrombocytopenia, microangiopathic haemolytic anaemia (MAHA), renal impairment, fever, and neurological symptoms such as confusion and seizure. TTP is precipitated by conditions such as vasculitis, pregnancy, cancer, drugs-induced, solid organ or stem cell transplantation, and infections with a background deficiency of a disintegrin and metalloproteinase with a thrombospondin type 1 motif member 13 (ADAMTS13). TTP in pregnancy is not uncommon, however, acute vision loss is a rare presenting sign of TTP.

Case Description: A 17-year-old primigravida woman of Murut ethnicity presented with an acute vision loss at 32 weeks of gestation. She had no known medical illness with uneventful antenatal care. Her physical examination revealed bilateral visual loss with reduced visual acuity to finger counting over right eye and light perception over left eye, correlated with bilateral exudative retinal detachments in a fundoscopic examination. Her blood investigations showed MAHA and thrombocytopenia with a reduced ADAMTS13 activity at 10% reported later. Unfortunately, her pregnancy was complicated with intrauterine death. She was treated with plasma exchange and high dose intravenous corticosteroids with a complete resolution of symptoms.

Conclusions: TTP is one of the complications in pregnancy which requires urgent management. It can masquerade as various manifestations including vision loss and require good clinical acumen to ensure timely diagnosis and management.

Keywords: Vision loss; retinal detachment; pregnancy; thrombotic thrombocytopenic purpura (TTP); case report

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Introduction

Background

Pregnancy is a natural biological process where a woman undergoes significant anatomical and physiological changes in every organ system in order to accommodate the development of foetus (1). Most pregnant women can tolerate well with the physiological changes with uneventful pregnancies. However, the physiological changes during pregnancy may unmask or precipitate quiescent medical conditions such as insulin resistance and glucose intolerance, hypertensive disorders, as well as autoimmune and haematological disorders (2). Thrombotic thrombocytopenic purpura (TTP) is a life-threatening haematological disorder associated with the deficiency of a disintegrin and metalloproteinase with a thrombospondin type 1 motif, member 13 (ADAMTS13), a protease which cleaves the large von Willebrand factor (VWF) multimers secreted by endothelial cells. The VWF multimers are highly adhesive and trigger unchecked platelet aggregations when not cleaved, leading to microthrombi formation and mechanical haemolysis (3).

Rationale and knowledge gap

Pregnancy is a known risk factor for acute events of TTP which results in thrombotic events affecting all organs and rarely vision. Postulated theories behind the association of TTP and pregnancy are the physiological changes during pregnancy with haemostasis, hypercoagulability state and increased VWF concentration (4). Ocular changes during pregnancy involving the eyelids, conjunctiva, cornea, lens, retina, optic nerve, and orbit are usually benign and self-limiting which resolve after pregnancy and rarely result in permanent visual disturbance. Nevertheless, visual impairment in pregnancy requires thorough investigations to look for systemic diseases with ocular complications such as hypertensive disorders, diabetic retinopathy, occlusive vascular disorders, and intracranial pathology (5). It is important to consider TTP as a differential diagnosis for acute vision loss in pregnancy as it impacts the outcome with early suspicion and management.

Objective

We report a rare case of acute vision loss without the classical pentad signs as an initial manifestation of TTP in pregnancy with the aim to emphasize the importance of prompt recognition and management which gives a better clinical outcome. We present this case in accordance with the CARE reporting checklist (available at https://amj.amegroups.com/article/view/10.21037/amj-23-81/rc).

Highlight box

Key findings

- A young pregnant woman presented with acute vision loss as the first sign of thrombotic thrombocytopenic purpura (TTP) without fever and renal impairment as parts of the classical pentad. With plasma exchange and high dose intravenous corticosteroids, the ocular symptoms resolved however the pregnancy was complicated with intrauterine death.

What is known and what is new?

- TTP is known to manifest with the classical pentad of thrombocytopenia, microangiopathic haemolytic anaemia, renal impairment, fever, and neurological symptoms.
- TTP can masquerade as an alternative manifestation such as acute vision loss without the classical pentad.

What is the implication, and what should change now?

- High clinical suspicion for TTP in a correct clinical setting without the classical pentad is crucial for timely diagnosis and management to ensure a better outcome.

Case presentation

A 17-year-old primigravida woman of Murut ethnicity presented with a five-day duration of blurring vision and reduced foetal movements at 32nd week of gestation on June 20th, 2022. She did not have any history of trauma particularly to her head and eyes, and there was no fever, headache or other neurological symptoms. She had no known medical illness and her antenatal care prior to this event was unremarkable. She did not take any medications or history of surgery prior to this. Her family history was unremarkable particularly no history of autoimmune and haematological disorders.

She was orientated with full Glasgow Coma Scale upon presentation. Her physical examination revealed a reduced visual acuity of bilateral eyes limited to finger counting over right eye and light perception over left eye. A fundoscopic examination confirmed the presence of bilateral exudative retinal detachments. Other systemic examinations were unremarkable with a blood pressure of 120/70 mmHg and heart rate of 88 beats per minute.

Her blood investigations showed haemolytic anaemia with thrombocytopenia with a haemoglobin level of

9.8 g/dL, platelet count of 27×10^3/μL, reticulocyte count of 6.4%, serum bilirubin of 24 μmol/L, and serum lactate dehydrogenase of 1,043 U/L. A peripheral blood film confirmed the features of microangiopathic haemolytic anaemia (MAHA) with the presence of schistocytes. Other blood investigations were otherwise unremarkable particularly no renal and liver impairments and no coagulopathy. Unfortunately, an obstetric assessment with ultrasonography confirmed an intrauterine death.

Differentials of pre-eclampsia and eclampsia were made after assessment by obstetrics team and she was given intravenous magnesium sulphate. Given the benefit of doubt whereby vision loss maybe reversible if immunotherapy given in case of TTP, she was treated with immunotherapy prior to the result of ADAMTS13 activity after discussion with haematologist. She was prescribed with daily therapeutic plasma exchange (TPE) with 40 mL/kg of fresh frozen plasma (FFP) for 7 days and intravenous methylprednisolone 500 mg daily for three days followed by a tapering dose of oral prednisolone. She delivered a stillborn foetus on day 3 of hospitalization and achieved normalization of platelet count to 161×10^3/μL and marked visual acuity improvement of 6/12 bilateral eyes on day 9 of hospitalization. A reduced ADAMTS13 activity of 10% was reported two weeks later and a negative inhibitor assay confirmed the diagnosis of hereditary TTP. A follow-up three months later with haematology team reported a complete recovery of visual acuity and blood parameters particularly with a normal platelet count of 223×10^3/μL. She was also given an appointment for future pre-pregnancy counselling and planning with maternal foetal medicine team.

All procedures performed in this study were in accordance with the ethical standards of the institutional and/or national research committee(s) and with the Helsinki Declaration (as revised in 2013). Written informed consent was obtained from the patient's legal guardian for publication of this case report. A copy of the written consent is available for review by the editorial office of this journal.

Discussion

Key findings

Our case was diagnosed with TTP based on vision loss which is not the typical neurological symptoms in the presence of MAHA and thrombocytopenia. However, there is absence of fever and renal impairment which are parts of the classical pentad signs of TTP.

Strengths and limitations

Differentials diagnosis for our case include pre-eclampsia, eclampsia, and Haemolysis, Elevated Liver enzymes, Low Platelet count (HELLP) syndrome which the treatment is mainly supportive with early delivery of foetus. Intracranial tumour results in vision loss should be considered as well, however, acute onset of the symptom, presence of MAHA and thrombocytopenia, and absence of other focal neurological symptoms made it less likely. Our limitation to get the result of ADAMTS13 activity on time should not delay the diagnosis of TTP as early treatment often provides a better clinical outcome.

Comparison with similar researches

Ocular involvements were observed in 20% cases of TTP which consist of retinal detachment, choroidal bleeding, macular infarction, Purtscher-like retinopathy, and occlusions of central retinal artery and vein (6). Serous retinal detachment was first described as a complication of TTP in a post-mortem examination in 1970 (7). Only a few similar cases like ours were reported in the past with vision loss as the initial presentation of TTP with complete visual recovery after treatment (6,8). The case reported by Kovács *et al.* was first misdiagnosed as HELLP syndrome with successful delivery of newborn, however, developed bilateral visual impairment 10 days later, which took up to one and a half year later to have complete visual recovery (6). Sampo *et al.* reported a woman presented with similar features like our case where vision loss is the main symptom in the presence of MAHA and thrombocytopenia without fever and renal impairment, however, diagnosis was made easier with the history of TTP diagnosed 8 years ago which allowed a prompt management and achieved a complete visual recovery (8).

Explanation of findings

TTP is classically described in the literature with a pentad of thrombocytopenia, MAHA, acute kidney injury, fever and neurological symptoms ranging from subtle headache to confusion, seizure, stroke, and coma. However, one-third of patients with TTP were reported to have no neurological symptoms and a complete classical pentad was only observed in less than 5% of patients (9). This can be possibly explained by a timely detection of TTP before the development of neurological symptoms and prompt treatment to prevent the progression of disease into the classical pentad.

Implications and actions needed

The diagnosis of TTP is confirmed with reduced ADAMTS13 activity. It is recommended to start treatment with TPE as soon as possible when the diagnosis of TTP is suspected without the result of ADAMTS13 activity as earlier treatment is correlated with a better prognosis and outcome (10). The mainstay treatment of TTP is TPE with FFP and high-dose intravenous corticosteroids which has proven its efficacy in various cases of TTP including drug-induced (11). However, in refractory cases, intravenous immunoglobulin and second-line immunosuppressive agents such as cyclophosphamide, cyclosporine A, and azathioprine can be used (12).

Conclusions

TTP is one of the complications in pregnancy which requires urgent management. It can masquerade as various manifestations including vision loss. The classical pentad of TTP is not present in majority of cases. High suspicion for TTP in any clinical presentation with MAHA and thrombocytopenia is crucial to ensure timely diagnosis and management for a better outcome.

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Footnote

Reporting Checklist: The authors have completed the CARE reporting checklist. Available at https://amj.amegroups.com/article/view/10.21037/amj-23-81/rc

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Conflicts of Interest: All authors have completed the ICMJE uniform disclosure form (available at https://amj.amegroups.com/article/view/10.21037/amj-23-81/coif). The authors have no conflicts of interest to declare.

Ethical Statement: The authors are accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved. All procedures performed in this study were in accordance with the ethical standards of the institutional and/or national research committee(s) and with the Helsinki Declaration (as revised in 2013). Written informed consent was obtained from the patient's legal guardian for publication of this case report. A copy of the written consent is available for review by the editorial office of this journal.

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