

A Case Report :
Undiagnosed ADAMTS 13 deficiency – A rare cause of Thrombocytopenia in Pregnancy
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INTRODUCTION

ADAMTS 13 is a specific von Wille brand Factor (VWF), or cleaving metalloprotease found in plasma. Deficiency of this factor can be either congenital or acquired. Congenital deficiency is caused by reduction in synthesis of ADMTS 13 due to genetic defect. This type shows autosomal recessive inheritance. When ADMTS 13 activity is reduced thrombotic thrombocytopenic purpura (TTP) develops. If woman has deficiency of this factor, she develops thrombocytopenia during pregnant.

Thrombocytopenia during pregnancy may occur because of any reasons. However, when it occurs due to ADMTS 13 deficiency it results into thrombotic thrombocytopenic purpura (TTP), an uncommon life-threatening condition. Generally, TTP is characterised by microangiopathic haemolytic anaemia and consumptive coagulopathy (thrombocytopenia) which leads to disseminated intravascular (microvascular) thrombosis. Whenever this occurs there is development of variety of signs and symptoms as a result of organ ischaemia. Here is presented a case report where woman was diagnosed with severe thrombocytopenia during the pregnancy due to undiagnosed ADMTS 13 deficiency.

CASE HISTORY

A 24-year-old primigravida was booked for the consultant led care clinic in her early pregnancy. She did not have any significant medical or surgical history. However, her partner was diagnosed with phenylketonuria (PKU) but she declined for carrier testing. She was a known case of smoker. In her family, her both parents had history of hypertension, and mother had a history of preeclampsia toxemia (PET). Her booking and 28 weeks bloods results were normal. Her antenatal period was uneventful until she was 35 weeks pregnant.

At 35 weeks of gestation, she presented in Antenatal clinic with no fetal movement, headache, swellings of hands and feet. Her fetal growth scan was normal. Her blood pressure was 140/90 mm of Hg, and there was 2+ protein in her urine. PET bloods were sent awaiting results. She was admitted in antenatal ward for observation and further management plan. On examination, there were brisk knee jerk reflexes but clonus was absent. On CTG examination, baseline was raised up to 180 bpm, variability 1-3 bpm, and there was a prolonged bradycardia lasting for more than 4 minutes. It was diagnosed as abnormal antenatal CTG. Therefore, decision was taken to expedite the birth of baby as a category 1 Caesarean section under general anaesthesia. Her surgery was uneventful.

However, blood result showed severe thrombocytopenia, with haemoglobin level 56 g/L, platelets $21 \times 10^9/L$, creatinine $97 \mu\text{mol/L}$, warning AKI flag 1. Therefore, she was transferred to intensive care unit (ITU). On further investigations, lactate dehydrogenase was very high, 1446 unit/L . Next day repeat blood examination showed a little rise in haemoglobin, 67 g/L , creatinine $106 \mu\text{mol/L}$, and ALT 151 U/L . On day 3 blood examination, all blood results were improving but platelet was still $43 \times 10^9/L$. Therefore decision was taken to transfer her to the tertiary care unit for the plasma exchange. At tertiary care unit she was diagnosed as ADAMTS 13 deficiency, and she was managed further accordingly. After plasma exchange she recovered well. It was first time when she was diagnosed with ADAMTS 13 deficiency, on detail investigation.

DISCUSSION

During pregnancy, it is normal to fall in platelet count, particularly during the 3rd trimester. At term pregnancy the level of platelet is approximately 10% less than the pre-pregnancy level. If the starting platelet count is at the lower end of the normal range, or there is a more severe fall in platelet levels during the pregnancy thrombocytopenia occurs. It is important to learn about the underlying causes of thrombocytopenia during pregnancy, and management of the more severe cases is very important. Causes of thrombocytopenia in pregnancy include; gestational thrombocytopenia, immune thrombocytopenic purpura, hypertensive disorders or pregnancy such as preeclampsia and HELLP syndrome, microangiopathic, hereditary thrombocytopenia, viral infections or drug-induced.

TTP has been shown to be due to a severe deficiency of Von Willebrand's factor cleaving protein. Most of the time this is an acquired deficiency caused by an auto antibody or, rarely, a congenital deficiency caused by a genetic defect. TTP occurs in about 1 in 25000 pregnancies. However TTP due to ADAMTS 13 deficiency, accounts 2-4% of all TTP. TTP is often overlooked and missed diagnosed. This could be because of the rarity of the disease and a definitive method of diagnosis has not been established. The classical symptoms of TTP are thrombocytopenia, haemolytic anaemia, fluctuating neurological abnormalities, renal failure and fever. However the symptoms are not specific to TTP. Our patient presented with mild preeclampsia, severe thrombocytopenia, raised lactate dehydrogenase, ALT and bilirubin suggestive of HELLP syndrome. She was investigated for the rare causes of thrombocytopenia, and therefore she was diagnosed as a pre-existing ADAMTS 13 deficiency with thrombocytopenia in pregnancy.

CONCLUSION

In conclusion, pregnancy associated TTP is attributed to late onset TTP in many of the patients during pregnancy. It is important to find out the underlying cause of TTP during pregnancy and appropriate management of more severe cases. As such patients are high-risk cases this should be referred to Tertiary care unit especially those who require plasma therapy.

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