Case report

# Pregnancy complicated with May-Hegglin anomaly

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## Abstract

The May-Hegglin Anomaly (MHA) is a rare autosomal dominant giant platelet disorder characterized by abnormally large platelets with defective leucocytes and thrombocytopenia with varying degrees of bleeding manifestation. Here we report successful pregnancy outcome in a primigravida with MHA. (Preterm Pre-labour Rupture of Membranes) PPROM mother, with abnormalities of the extremities due to ABS is discussed.

**Key words:** May-Hegglin anomaly, thrombocytopenia, pregnancy, multidisciplinary input, neonatal screening

## Background

The May-Hegglin Anomaly (MHA) is a rare autosomal dominant giant platelet disorder characterized by abnormally large platelets with defective leucocytes and thrombocytopenia. It belongs to a group of hereditary thrombocytopenia disorders with variable genetic expression of single gene defect. Sebastian syndrome, Fechtner syndrome and Epstein syndrome have similar gene mutation to MHA[1].

It was firstly described in 1909 by May and later by Hegglin in 1945. The responsible gene is mapped to Gene map 22q12-13 and mutation of this gene causes abnormal production of non-muscle myosin heavy chains which is asso ciated with macrothrombocytopenia and leucocyte inclusion bodies (Dohle-like bodies) due to defective megakaryocyte maturation and fragmentation. It is characterized by variable thrombocytopenia. Though there is varying degree of thrombocytopenia, leucocytes and platelet functions are conserved[2].

Bleeding is not a major manifestation in May Hegglin anomaly and platelet transfusion is needed in patients with severe thrombocytopenia[3].

## Case presentation

We report a case of 29-year -old primigravida with May-Hegglin anomaly following successful pregnancy and peripartum outcome.

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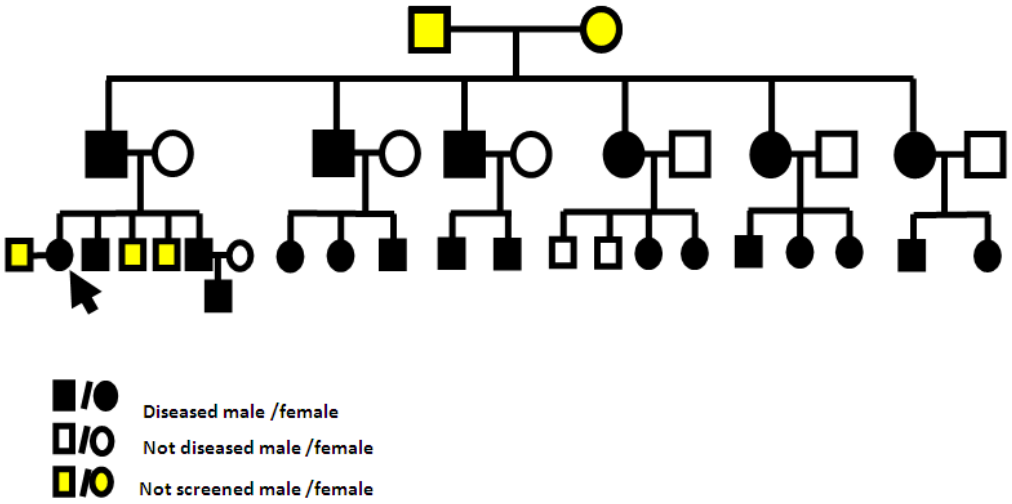


Figure 1. **Family pedigree. The arrow marks the index patient.**

At the age of 24 years, she was diagnosed with May-Hegglin anomaly in a family screening for platelet disorders as her paternal aunt developed menorrhagia with thrombocytopenia with abnormal leucocytes. Similarly, many of her siblings and relatives were found to have MHA following the screening program. (Figure 1 family pedigree). There is no history of abnormal bleeding or bruises in the past and her menstrual bleeding was minimal. Since the diagnosis, she was on regular haematology clinic follow up.

At the age of 29 years, following spontaneous conception from a non-consanguineous marriage, she was under specialist led antenatal care with haematologist input.

Her initial platelet count was $50\times10^{6}/mm^{3}$ and it fluctuated between $30\text{-}84\times10^{6}/mm^{3}$ throughout the pregnancy. Her blood picture showed macrothrombocytopenia with normal looking leucocytes and microcytic hypochromic red cells. Her coagulation tests such as APTT, PT, INR, serum ferritin level and fibrinogen were within the normal range. She did not have any bleeding manifestation during the antenatal period.

At term, after a failed induction of labour , she had a caesarean section under spinal anesthesia. She was transfused 6 units platelets prior to the caesarean section and prophylacticoxytocin infusion was given. A baby girl with a birth weight of 2810g was delivered. She had uneventful postpartum period with normal haematological values.

Genetic counselling was given to the mother and the baby awaits screening for MHA.

## Discussion

This patient was diagnosed with MHA 5 years ago and she was on regular haematology clinic follow-up. She did not have any bleeding manifestations despite of having thrombocytopenia.

Since first trimester in this pregnancy , she was cared by a multidisciplinary team consisted of obstetrician and haematologist with regular follow-ups. Foetal growth was monitored with regular growth scans.

Following a multidisciplinary team discussion, an induction of labour at term was considered as the patient was haematologically asymptomatic.

Although there were differing opinions on mode of delivery in pregnant patients with MHA, individual case-based delivery plans were made for individual cases. Traumatic instrumental delivery should be avoided to

minimize fetal intracranial haemorrhage and caesarean sections were done mainly for obstetric indications and on patients with bleeding manifestations[4].

In this patient, after a failed induction of labour , category 3 caesarean section was done [5]. Though the platelet count was 84 $\times 10^{6}/mm^{3}$, she was transfused 6 units platelets prior to the caesarean section. Prophylactic oxytocin infusion was started to minimize postpartum haemorrhage. She had minimal blood loss during delivery and postpartum period. Both mother and baby were discharged in healthy condition.

As MHA is an autosomal dominant disorder, half the infants are at the risk of having MHA and may be associated with intracranial haemorrhage during parturition. The need for screening in the neonate for MHA was discussed with the couple and they agreed for the neonatal screening test after delivery.

## Conclusions

This May-Hegglin anomaly case handling emphasizes the importance of multidisciplinary care pathway for complicated pregnant cases and the mode of delivery need to be decided upon individual clinical presentation and the platelet count. Proactive measures such as platelet transfusion and oxytocin infusion are effective in minimizing postpartum haemorrhage.

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