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Case Report

May-Hegglin Anomaly and the Devastating Consequences of Acute Cerebral Hemispheric Infarction

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Abstract

May-hegglin anomaly (MHA) is a rare genetic disorder characterized by thrombocytopenia, giant platelets, and leukocyte inclusion bodies. Bleeding diatheses are known to occur in patients with the condition; however, episodes of thrombotic events have been rare. We present a case of acute cerebral infarction of the left hemisphere in a 29-year-old patient with the May-Hegglin anomaly, ultimately resulting in herniation and brain death. To our knowledge, this is only the second case of an acute ischemic stroke in a patient with MHA, and the first case to occur in a young person.

Keywords: Macrothrombocytopenia; May-Hegglin anomaly; MYH9; Cerebral infarction

Introduction

Giant platelets, thrombocytopenia, and Döhle-like body neutrophil inclusions characterize the May-hegglin anomaly (MHA) [1]. MHA is inherited in an autosomal dominant fashion [1] and caused by a mutation in the MYH9 gene responsible for encoding the non-muscle myosin heavy chain A [2]. Bleeding diatheses can range from negligible to severe hemorrhage and are treated with platelet transfusion if necessary [1]. Bleeding episodes can correlate with the degree of thrombocytopenia, however, thrombotic episodes have been rare in MHA and only described in case reports. We present the second known case report of acute cerebral ischemia in a patient with the May-Hegglin anomaly, which resulted in a devastating hemispheric stroke and eventually death.

Typically, thrombocytopenia is considered to place patients at lower risk of arterial thrombosis [1,3]. Our case demonstrates a unique presentation of a thrombotic event in a young patient with extreme thrombocytopenia. While thrombotic events can occur in cases of thrombocytopenia, they usually occur in cases of profound systemic illness or medication side effect. A review of the limited literature on the May-Hegglin anomaly suggests that normal platelet aggregation mechanisms can be present despite low platelet counts, indicating that this condition may behave differently than other thrombocytopenias.

Case Presentation

A 29 year-old female presented to the emergency department with acute onset of right upper extremity muscle weakness, paresthesia, and aphasia of five-hour duration, suggestive of an acute left-sided cerebrovascular accident. Her past medical history was significant for the May-Hegglin anomaly, monitored by a local hematologist. The patient had a medical history of hypertension, tobacco abuse, hearing loss, and advanced chronic kidney disease of an unknown etiology. Initial computed tomography (CT) of the brain revealed no evidence of intracerebral hemorrhage. Magnetic resonance imaging (MRI) of the brain demonstrated an acute ischemic stroke involving the left frontal and parietal lobes (Figure 1).

The patient was admitted to the neurocritical care unit for intensive stroke monitoring and therapies. Due to her low platelet count (2,000 cells/microL), she did not receive antiplatelet or anticoagulant therapy. Echocardiogram with bubble study was negative for intracardiac shunt, thrombus, or valvular vegetations; continuous telemetry revealed no cardiac arrhythmias. Carotid investigation was negative for hemodynamically significant stenosis. A battery of blood tests to search for hypercoagulable states, including Protein C, Protein S, Antithrombin III, Prothrombin gene mutation, Factor V Leiden mutation, and the lupus anticoagulant were unremarkable. The patient was observed in the neurocritical care unit for several days with mild neurologic improvement and then transferred to the general medical ward, where she became acutely obtunded and unresponsive within the next 24 hours. An emergent head CT and brain MRI demonstrated a large acute ischemic stroke involving the entire left cerebral hemisphere with left to right midline shift and effacement of basilar cisterns (Figure 2). Her condition deteriorated; she demonstrated decorticate and decerebrate posturing, absent pupillary reflex, and autonomic dysfunction. A repeat head CT

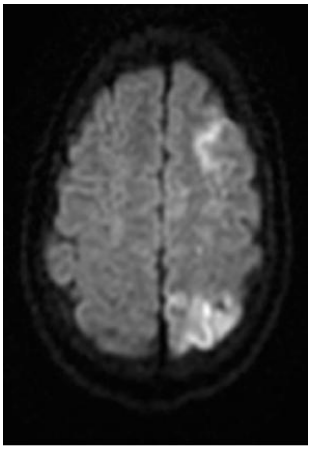

Figure 1: Initial Diffusion-weighted imaging (DWI) MRI of the brain displaying acute ischemia in the left frontal and parietal lobes with no mass effect.

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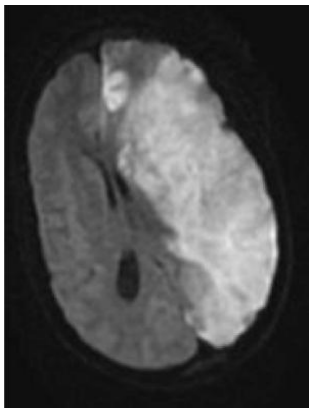

Figure 2: Final DWI MRI of the brain, displaying acute ischemia of the entire left cerebral hemisphere with left to right midline shift.

confirmed uncal herniation. Finally, a four-vessel cerebral angiogram demonstrated no intracranial blood flow supporting the clinical picture of brain death. The patient was provided comfort care and subsequently expired.

Discussion

Understanding of the May-Hegglin anomaly continues to evolve, but it may not be as predictable as other causes of thrombocytopenia. While bleeding events can occur as with thrombocytopenias of any cause, our case supports the fact that arterial thrombotic events, while rare, can occur in MHA as well. Only one other documented case of acute ischemic stroke in MHA has been reported of a 78-year-old female in Japan whose platelet count was 10,000/microL [2].

Other case reports have described coronary artery thromboses in patients with platelet counts less than 50,000 cells/microL [3, 4]. McDunn et al. discovered a normal platelet mass in their patient with MHA who experienced reocclusion of the coronary arteries after angioplasty [4]. In a case of acute myocardial infarction described by Goto et al., platelet aggregation and binding to von Willebrand factor (vWF) occurred even when the patient's platelets were less than 10,000 cells/microL [3]. These studies suggest that platelet enlargement occurs as a compensatory response to the degree of thrombocytopenia, and in fact, arterial thrombosis may be directly related to the degree of macrothrombocytopenia [3,4]. The longstanding belief that antiplatelet agents are contraindicated in cases of thrombocytopenia may not be as applicable in patients with the May-Hegglin anomaly. In fact, Fayyad et al. addressed this issue in the case of an obstetrical patient with MHA who experienced a fetal loss at 38 weeks during a second pregnancy. On pathological examination, areas of infarction were found throughout the placenta, which likely caused the death of the fetus. During the patient's third pregnancy, her obstetrical team discussed a trial of aspirin to prevent arterial thrombosis, even though it had not been done before. The patient was counseled on the risks of the antiplatelet agent but agreed to the therapy and delivered a healthy child with no postpartum complications [5].

Conclusion

Knowledge of bleeding and clotting risk is limited in patients with the May-Hegglin anomaly. Our case indicates that further research is needed. However, patients with MHA can have normal platelet aggregation and platelet mass despite the low number of platelets. Given the clinical circumstances, physicians may consider utilizing antiplatelet therapy in selected patients with the May-Hegglin anomaly to treat and even prevent instances of arterial ischemia.

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