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ABSTRACT N. PO85

ACUTE RETINAL ISCHEMIA IN HEMOGLOBIN SC DISEASE: A CASE REPORT AND REVIEW OF THE LITERATURE.

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Background

Sickle cell disease (SCD) encompasses a group of hereditary hemoglobinopathies characterized by the presence of hemoglobin S (HbS), and includes homozygous sickle cell anemia (HbSS), sickle β^0-thalassemia (HbS/β^0), and heterozygous disorders, such as HbSC disease. The latter results from compound heterozygosity for HbS and hemoglobin C (HbC) mutations affecting the β-globin gene. Although HbSC disease generally presents with a milder clinical phenotype than HbSS, it shares the risk of vaso-occlusive complications in various organ systems, including the ocular district.

Case Report

We report the case of a 28-year-old Ghanaian male with known HbSC disease under hydroxyurea treatment, who presented to the Emergency Department with sudden-onset floaters and a central scotoma in the left eye upon awakening. He reported intense physical exertion and episodes of diarrhea the previous day. His medical history included childhood hospitalization for osteomyelitis and a previous ischemic ocular event in 2020, complicated by vitreous hemorrhage and retinal detachment in the right eye, leading to profound vision loss. On examination, he was alert, afebrile, with normal vital signs. Ophthalmologic evaluation revealed left posterior vitreous detachment with vitreous strands and an area of retinal whitening and edema consistent with suspected macular retinal artery occlusion. Laboratory tests showed hemoglobin 121 g/L, hematocrit 36.8%, platelets 116 x 10^9/L, normal white blood cell count and inflammatory markers, and mildly elevated LDH (231 U/L) with undetectable haptoglobin. Hemoglobin electrophoresis confirmed 51% HbS and 43% HbC. Emergency erythrocytapheresis (red cell exchange transfusion) was performed, hydroxyurea was discontinued, and antiplatelet therapy with aspirin was initiated. During hospitalization, visual symptoms remained stable with no further deterioration. Additional evaluations including echocardiography, ECG-Holter, carotid ultrasound, and brain MRI/MRA revealed only a chronic right optic nerve atrophy. A mild functional protein C deficiency was noted, with normal antigen levels. The patient was discharged on a chronic monthly erythrocytapheresis regimen.

Conclusions

HbSC disease can present significant clinical challenges, including vaso-occlusive episodes and proliferative retinopathy, whereas acute ischemic manifestations, such as retinal artery occlusion, are rare and lack evidence-based management guidelines. Compared to other SCD genotypes, HbSC disease remains understudied in terms of pathophysiology and treatment strategies. Unlike HbSS and HbS/β^0, where hemolysis plays central role, HbSC-related vaso-occlusion may be more strongly influenced by increased blood viscosity. Treatment strategies align with those for HbSS, including the use of hydroxyurea, although evidence for its efficacy in HbSC is less robust and still under investigation. Alternative approaches focus primarily on minimizing the risk of further ischemic events by addressing modifiable risk factors such as hydration status and smoking. Therapeutic phlebotomy has been proposed as an adjunctive treatment for patients with HbSC disease, particularly in those with frequent vaso-occlusive episodes and elevated hemoglobin levels. Finally, red cell exchange (erythrocytapheresis) has been explored as a therapeutic option, however, evidence remains limited, and its use is generally extrapolated from experience in HbSS patients.

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