Sickle cell disease (SCD)

Sickle cell disease (SCD) is an autosomal recessive haemoglobinopathy, characterized by sickling of the red blood cells in response to a hypoxic stress and vaso-occlusive crises.

Sickle Cell Disease Associated Moyamoya Syndrome

Sickle Cell Disease Associated Moyamoya Syndrome

Complications

Sickle cell disease is a common haemoglobinopathy that significantly increases the risk of ischemic stroke. Because the ischemic stroke risk factors onset and mortality in non-sickle cell disease patients have been largely elucidated, Miller et al. aimed to analyze risk factors for ischemic stroke mortality in sickle cell disease patients, which remain largely unknown.

The National Inpatient Sample database (2016-2017) was used to develop a multivariable regression model for risk quantification of known ischemic stroke risk factors for in-hospital mortality in ischemic stroke patients with and without sickle cell disease.

Classical risk factors for ischemic stroke onset, including ischemic heart disease, carotid artery disease, lipidemias, hypertension, obesity, tobacco use, atrial fibrillation, personal or family history of stroke, congenital heart defects, congestive heart failure, cardiac valve disorder, peripheral vascular disease, and diabetes mellitus are associated with in-hospital mortality in non-sickle cell patients ($p < 0.05$). However, no significant association was found between these stroke risk factors and in-hospital mortality in sickle cell disease patients presenting with ischemic stroke ($p > 0.05$).

While the classical risk factors for stroke onset are associated with in-hospital mortality in non-sickle cell stroke patients, they are not associated with in-hospital mortality in sickle cell stroke patients [1].

Vascular injury, hypercoagulability and vaso-occlusion play a role in the pathophysiology of stroke in SCA. Transcranial Doppler ultrasound (TCD) has lowered the incidence of ischemic stroke from 11% to 1% as TCD identifies children who are at risk for stroke, providing opportunities for interventions to reduce this risk. Whereas blood exchange is indicated in acute stroke, chronic transfusions (either simple or exchange on a monthly basis) are used for primary as well as secondary stroke prevention in developed countries. Children with abnormally high TCD velocities (≥ 200 cm/s) are at high risk of stroke and might benefit from hydroxyurea or hydroxycarbamide (HU) after a period of a successful transition from chronic transfusions. Hematopoietic stem cell transplant presents a cure for SCA. Gene therapy is currently investigated and may be offered to patients with SCA who had a stroke or who are at high risk of stroke if proven efficacious and safe. However, gene therapy is not likely to be implemented in low-income countries due to cost. Alternatively, HU is utilized for primary and secondary stroke prevention in developing countries. Further expansion of TCD implementation should be a priority in those settings [2].

Children with Sickle Cell Disease (SCD) are at risk for developing multiple intracranial aneurysms, and a high index of suspicion must be maintained during the interpretation of routine magnetic resonance imaging or angiography of the brain [3].

Spontaneous (nontraumatic) acute epidural hematoma is a rare and poorly understood complication of sickle cell disease

Case reports

2015

A 19-year-old African American male with hemoglobin SC disease (HbSC) presented with generalized body aches and was managed for acute painful crisis. During his hospital stay he developed rapid deterioration of his mental status and computed topography revealed a spontaneous massive epidural hematoma with mass effect and midline shift with Kernohan notch phenomenon for which urgent craniotomy and evacuation was done.This is the first case of HbSC disease associated with catastrophic epidural hematoma progressing to transtentorial herniation and Kernohan's notch phenomena within few hours with rapid clinical deterioration. The etiopathogenesis and the rare presentation are discussed in detail in this case report [4]

1)
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