Sickle Cell Anaemia

It is an autosomal recessive genetic blood disorder in which RBC's become long, stiff, rod like structures or sickle shaped (RBCs are normally shaped like a disc).

Causes

It is caused by an abnormal type of Hb called Haemoglobin S. Hbs distorts the shape of RBCs, especially when exposed to low oxygen levels. The distorted RBC's are fragile and shaped like crescents or sickles.

Symptoms

Joint or bone pain, breathlessness, delayed growth and puberty, fatigue, fever, paleness, rapid heart rate, ulcers on the lower legs and yellowing of the eye and skin (jaundice).

Pathophysiology

When exposed to low oxygen tensions, polymerization of HbS take place in several stages, Initially small aggregates containing approx. 30 HbS molecules an formed which on further polymerization are organized into long tubular fibers. In the final step (crystallization), the fibers align themselves into parallel bundles seen under the electron microscope to run along the long axis of the sicked cells. The net effect of polymerization is to conver HbS from a freely flowing liquid to viscous gel that is responsible for the distortion and reduced plasticity of the red cells.

Factors, that affects that rate and degree of sickling, is the amount of HbS and its interaction with other Hb chains in the cell. The rate of HbS polymerization is also significantly affected by the haemoglobin concentration per cell, i.e. the MCHC. The higher the HbS concentration with in the cell the greater are the chances of contact and interaction between HbS molecules. Thus dehydration, which increases the MCHC, greatly fascilitates sickling may trigger occlusion of blood vessels.

Sickly of red cells is initially a reversible phenomenon, with oxygenation, HbS return to the depolymerized state. However, with repeated episodes of sickling membrane damage ensures.

Reference

Shakya M. K., Singh S, Singh P.K. “Pathophysiology an introduction to human diseases” published by R. Narain Publishers & Distributors, first edition 2019, page no 101-102.