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PROPAEDEUTIC APPROACH TO SICKLE CELL DISEASE: PATHOPHYSIOLOGY, DIAGNOSIS, TREATMENT, AND PREVENTION

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Relevance. Sickle cell disease (SCD) ranks among the most prevalent genetic disorders globally, particularly affecting sub-Saharan Africa, where approximately 80% of affected newborns are born. Despite progress in diagnostics and treatment, access to early diagnosis and comprehensive care is uneven. In Ghana, the prevalence is 2% among newborns, whereas European countries like Belarus have seen an increase due to migration. This study combines traditional clinical skills with modern hematology to enhance recognition and management in various resource settings.

Aim: to analyze the pathophysiological mechanisms, diagnostic methods, treatment strategies, and preventive measures of sickle cell disease (SCD) using propaedeutic of internal medicine and to compare the disease burden and management in Ghana, sub-Saharan Africa, and Europe, particularly in Belarus.

Materials and methods. A systematic literature review was conducted using peer-reviewed articles, clinical guidelines, and epidemiological reports. Sources were analyzed for pathophysiology, diagnostics, clinical features, treatment, and public health strategies, with comparative analysis between high-burden (Ghana, sub-Saharan Africa) and low-burden (Europe, Belarus) settings.

Results and their discussion. Sickle Cell Disease (SCD) arises from a single nucleotide change in the β-globin gene, causing hemoglobin S to polymerize, leading to sickling of red blood cells, chronic hemolysis, and damage to multiple organs. Symptoms include vaso-occlusive crises, acute chest syndrome, strokes, and increased susceptibility to infections.

Diagnostic capabilities differ significantly. In high-income countries, universal newborn screening using high-performance liquid chromatography allows for early intervention, resulting in over 94% survival into adulthood. In sub-Saharan Africa, most countries lack universal screening; Ghana only has pilot programs, and up to 90% of children with SCD may die undiagnosed within five years. New point-of-care tests and smartphone-based microscopy provide affordable options for resource-limited settings.

Treatment methods include hydroxyurea, which decreases crises by increasing fetal hemoglobin, chronic transfusions, and hematopoietic stem cell transplantation, the sole curative option. Gene therapies are developing but are largely unavailable in low-resource areas. Preventive strategies, such as penicillin prophylaxis and vaccinations, are effective but are not consistently applied due to inadequate diagnostic infrastructure.

A comparison shows significant disparities: SCD is rare in Belarus and often linked to migration, while it poses a major public health issue in Ghana and surrounding regions. The propaedeutic approach—focusing on history taking, physical examinations, and bedside observations—is essential for early detection, especially in areas lacking advanced diagnostic tools.

Conclusions. Sickle cell disease illustrates the connection between molecular pathology, clinical examination, and global health disparities. Early diagnosis via newborn screening and preventive measures significantly lowers childhood mortality, yet these methods are often underused in high-burden areas. It is essential to expand access to hydroxyurea, enhance diagnostic capabilities, and incorporate new treatments. The tradition of direct clinical examination is crucial for internal medicine and should work alongside modern technologies to provide equitable care in all resource settings.