1.24 SICKLE CELL DISEASE

Introduction

Sickle cell disease is the most common autosomal recessive disease in African American individuals, occurring in 1 in 625 live births to African American couples. While it is most common in African Americans, sickle cell disease also occurs in individuals of Hispanic, Arabic, Native American, and Caucasian heritage. Sickle cell disease results from a single base-pair substitution of thymine for adenine resulting in valine instead of glutamine in the sixth position of the Beta-globin molecule. Sickle cell disease results when this substitution occurs in a homozygous state. Other forms of sickle cell disease with variable severity can also occur when the heterozygote state is combined with a second variant Beta-globin chain such as hemoglobin C or Beta-thalassemia. Clinical manifestations result from polymerization of the abnormal hemoglobin and "sickling" of the red cells. The clinical manifestations most important to pediatric hospitalists include recurrent and chronic pain from dactylitis and vaso-occlusive crisis, acute chest syndrome, increased susceptibility to infections, aplastic crisis, splenic sequestration, stroke, acute hepatobiliary complications, and priapism. Pediatric hospitalists commonly encounter patients with known or suspected sickle cell disease and care for the various complications associated with the disease.

Knowledge

Pediatric hospitalists should be able to:

- Explain the impact of newborn screening on preventive care.
- Review the genetics and pathophysiology underlying the variants of sickle cell disease and their complications.
- Compare and contrast common sickle crisis presentations by age group.
- Describe the signs and symptoms of dactylitis, vaso-occlusive crisis, sepsis, acute chest syndrome, aplastic crisis, splenic sequestration, stroke, acute hepatobiliary complications, and priapism.
- Describe indications for hospital admission and escalation to intensive care.
- Identify the goals of inpatient therapy, attending to both acute and chronic needs.
- Distinguish patients with sickle cell disease presenting with fever who require inpatient management from those who can be safely managed in the outpatient setting.
- Recognize the unique considerations requiring expert consultation for patients with sickle cell disease undergoing surgical procedures.
- Summarize the roles of members of a comprehensive clinical care team, such as patients, the family/caregivers, subspecialty physicians, social worker, pharmacist, physical therapist, discharge planner, psychologist, and others.
- Discuss the therapeutic options available for complications of sickle cell disease, including the rationale for choosing a specific management plan.
- Discuss chronic complications of sickle cell disease, such as chronic pain, neurologic deficits with learning disabilities, hyposthenuria, delayed growth and development, psychosocial issues (including low self-esteem, anxiety, and depression), and others.
- Discuss the evidence-based guidelines for infection prevention, including vaccines and penicillin prophylaxis.
- Discuss medications for prevention and treatment of complications of sickle cell disease, including the use of hydroxyurea and L-glutamine.
- Explain the approach toward acute and chronic pain management, including the use of patient-controlled analgesia and timely transition to oral pain medications.
- Cite reasons for transfer to a referral center in cases requiring pediatric-specific services not available at the local facility.

Skills

Pediatric hospitalists should be able to:

- Diagnose sickle cell disease and its complications by performing an accurate history and physical examination, identifying cardinal features of the disease presentation and synthesizing information into unified assessment.
- Order appropriate laboratory and radiographic testing based on history and physical examination findings.
- Create a comprehensive evaluation and management plan, including the use of antimicrobial therapy, intravenous fluid hydration, pain management, transfusion therapy, and initiation of cardiovascular and pulmonary supportive care measures when appropriate.
- Identify patients with worsening status and respond with appropriate actions.
- Consult subspecialists in a timely manner when appropriate.
- Create a comprehensive discharge plan that includes clear home instructions, appropriate medications, and follow-up recommendations.

Attitudes

Pediatric hospitalists should be able to:

- Acknowledge the importance of effective communication and education with patients and the family/caregivers regarding the disease process, proposed therapies, expectations of inpatient therapy, and transition of care to the outpatient arena.
- Realize the psychosocial impact on the patient and family members/care givers.
- Recognize the value of collaboration with subspecialists and the primary care provider, to ensure coordinated longitudinal care for children with sickle cell disease.

Systems Organization and Improvement

In order to improve efficiency and quality within their organizations, pediatric hospitalists should:

- Collaborate with a multidisciplinary team consisting of subspecialty physicians, social workers, pharmacists, physical therapists, discharge planners, and psychologists to improve quality of care, increase patient satisfaction, and facilitate timely discharge from the acute care setting.
- Identify existing limitations for optimal care within the cur-

rent hospital setting and work with hospital administration and community partners to develop and sustain appropriate referral systems and coordinated transfers of care.
- Lead, coordinate, or participate in the development of coordinated discharge plans and programs in the local community.

References

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2. Wang CJ, Kavanagh PL, Little AA, et al. Quality-of-care indicators for children with sickle cell disease. *Pediatrics*. 2011;128:484-493. https://doi.org/10.1542/peds.2010-1791.