Electronic Medical Record (EMR) Summary

Patient ID: PID18636310

Name: Anil Varma

Age: 45, Sex: Male

Visit ID: VISIT13871526

Date: 2025-05-17 14:30

Clinical Reasoning Summary

Definition & Key Concerns

This infant presents with severe hemolytic anemia, hepatosplenomegaly, and failure to thrive. These findings suggest a possible underlying hemoglobinopathy or other congenital hemolytic anemia. Hemoglobinopathies are a group of disorders, either inherited or acquired, that affect the structure, function, or production of hemoglobin.

Differential Diagnosis

1. **Hereditary Spherocytosis**: This is a common cause of hemolytic anemia in children. The presence of jaundice and splenomegaly supports this diagnosis.
2. **Sickle Cell Anemia**: This is a common hemoglobinopathy that can present with severe anemia, jaundice, and splenomegaly. However, symptoms usually do not present until 6 months of age.
3. **Thalassemia**: Beta-thalassemia major can present in infancy with severe anemia, jaundice, and hepatosplenomegaly.
4. **Congenital Dyserythropoietic Anemia**: This is a rare condition that can present with similar symptoms, but is less likely given its rarity.
5. **Glucose-6-Phosphate Dehydrogenase (G6PD) Deficiency**: This condition can cause hemolytic anemia, but usually only in response to certain triggers (e.g., certain medications, infections).

Can?t-Miss Diagnosis

Hereditary Spherocytosis, Sickle Cell Anemia, and Thalassemia are all serious conditions that can have significant morbidity and mortality if not diagnosed and managed appropriately.

Suggested Investigations

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A hemoglobin electrophoresis should be performed to identify abnormal hemoglobin variants that could be causing the patient's symptoms. Additional tests that may be helpful include a reticulocyte count, peripheral blood smear, direct antiglobulin test (DAT), and osmotic fragility test. Genetic testing may also be considered.

Management Plan

Management will depend on the specific diagnosis. For hereditary spherocytosis, initial management may include folic acid supplementation and possibly a splenectomy if symptoms are severe. For sickle cell anemia and thalassemia, management typically involves regular blood transfusions and iron chelation therapy to prevent iron overload. All patients with these conditions should be referred to a hematologist for further management.

Reference Insight

According to UpToDate, hemoglobin electrophoresis is the gold standard for diagnosing hemoglobinopathies such as sickle cell anemia and thalassemia. For hereditary spherocytosis, the diagnosis is typically made based on clinical findings, a positive family history, and laboratory studies showing hemolytic anemia with spherocytes on peripheral blood smear (UpToDate, 2023).

Rare Disease Alerts

Thalassemia Major (matched 3 symptoms)

Prescription

None provided