Hematologic studies reveal the characteristic changes in RBCs (e.g., microcytosis, hypochromia, anisocytosis, poikilocytosis, target cells, and basophilic stippling of various stages). Low Hgb and Hct levels are seen in severe anemia, although they are typically lower than the reduction in RBC count because of the proliferation of immature erythrocytes. Hgb electrophoresis confirms the diagnosis and is helpful in distinguishing the type of the thalassemia because it analyzes the quantity and kind of hemoglobin variants found in the blood.

Therapeutic Management

The objectives of supportive therapy are to maintain sufficient Hgb levels to prevent bone marrow expansion and bony deformities and to provide sufficient RBCs to support normal growth and normal physical activity. Transfusions are the foundation of medical management with the goal of maintaining the Hgb level above 9.5 g/dl, an aim that may require transfusions as often as every 3 to 5 weeks. The advantages of this therapy include (1) improved physical and psychological well-being because of the ability to participate in normal activities, (2) decreased cardiomegaly and hepatosplenomegaly, (3) fewer bone changes, (4) normal or near-normal growth and development until puberty, and (5) fewer infections.

One of the potential complications of frequent blood transfusions is iron overload (hemosiderosis). Because the body has no effective means of eliminating the excess iron, the mineral is deposited in body tissues. To minimize the development of hemosiderosis, oral iron chelators (deferasirox, deferiprone) have shown in short-term studies to be a safe equivalent to **deferoxamine (Desferal)**, a parenteral iron-chelating agent, and more tolerable by patients and families (Bakai and Pennell, 2014; Cappellini, Porter, El-Beshlawy, et al, 2010; Meerpohl, Schell, Rucker, et al, 2014; Vichinsky, Bernaudin, Forni, et al, 2011).

In some children with severe splenomegaly who require repeated transfusions, a splenectomy may be necessary to decrease the disabling effects of abdominal pressure and to increase the life span of supplemental RBCs. Over time, the spleen may accelerate the rate of RBC destruction and thus increase transfusion requirements. After a splenectomy, children generally require fewer transfusions,