

Clinical Management of Patients With Thalassemia Syndromes

Marie Martin, RN, and Drucilla Haines, RN, MSN, CPNP



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Background: Thalassemia is a chronic inherited blood disorder that reduces hemoglobin production, causing chronic hemolytic anemia. Patients often are diagnosed via newborn screening programs. Patients diagnosed with the most severe form of thalassemia often require chronic red blood cell transfusions to control their anemia. The side effect of chronic transfusions is cumulative iron overload for which chelation therapy is required. The incidence of thalassemia is low; therefore, care is best delivered at specialized treatment centers that offer multidisciplinary coordination.

Objectives: This article reviews the diagnosis, management, and curative options for thalassemia.

Methods: This review follows a hypothetical patient with thalassemia and his family through the major stages of the disease: diagnosis, treatment, long-term monitoring, and continued support from childhood through adulthood.

Findings: Increasing knowledge about thalassemia and its management among healthcare providers can improve patient outcomes and quality of life.

Marie Martin, RN, is a retired thalassemia nurse coordinator at Children's Hospital of Philadelphia in Pennsylvania; and Drucilla Haines, RN, MSN, CPNP, is a pediatric nurse practitioner in the Hematology/Oncology Clinic at the Children's Hospital and Research Center Oakland in California. The authors take full responsibility for the content of the article. Writing and editorial support was provided by Michele Jacob, PhD, and Tara Beers Gibson, PhD, at Evidence Scientific Solutions and Susan DePetris, PhD, at Phase Five Communications through support from Novartis Pharmaceuticals Corporation. The content of this article has been reviewed by independent peer reviewers to ensure that it is balanced, objective, and free from commercial bias. No financial relationships relevant to the content of this article have been disclosed by the independent peer reviewers or editorial staff. Mention of specific products and opinions related to those products do not indicate or imply endorsement by the *Clinical Journal of Oncology Nursing* or the Oncology Nursing Society. Martin can be reached at cmboormart@verizon.net, with copy to editor at CJONEditor@ons.org. (Submitted January 2015. Revision submitted August 2015. Accepted for publication August 6, 2015.)

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Thalassemia occurs in regions of the world where malaria is prevalent and can affect 5%–30% of the population (Rund & Rachmilewitz, 2005). A recent survey conducted by the Cooley's Anemia Foundation estimates 1,000 individuals are affected by thalassemia in the United States. However, disease awareness remains low among nonspecialist healthcare providers (HCPs) who diagnose and treat these patients. Thalassemia is an inherited chronic autosomal recessive blood disorder resulting from impaired production of the alpha or beta subunit of hemoglobin (Muncie & Campbell, 2009). Symptoms vary depending on the amount and type of hemoglobin synthesized (Galanello, 2012; Muncie & Campbell, 2009), but chronic hemolytic anemia is common to all thalassemias (Cappellini, Poggiali, Taher, & Musallam, 2012; Children's Hospital and Research Center Oakland, 2012; Eldor & Rachmilewitz, 2002; Rund & Rachmilewitz, 2005).

Two forms of thalassemia exist: major and intermedia (see Table 1). Thalassemia major (TM), the most severe form, is characterized by profound reduction in hemoglobin production,

requiring lifelong peripheral red blood cell (RBC) transfusion therapy (Children's Hospital and Research Center Oakland, 2012; Muncie & Campbell, 2009). Thalassemia intermedia (TI) is less severe than TM and usually is transfusion independent; however, transfusions may be intermittently required and may become chronic later in life (Children's Hospital and Research Center Oakland, 2012; Galanello, 2012; Taher, Musallam, Karimi, & Cappellini, 2012). Some patients with TI remain asymptomatic until adulthood, whereas TM is typically diagnosed within the first few years of life (Taher, Isma'eel, & Cappellini, 2006). Complications such as extramedullary hematopoiesis, leg ulcers, gallstones, and thrombophilia are commonly associated with TI but rarely with TM (Taher et al., 2006).

Phenotypes range from mild to severe in both alpha and beta thalassemia. Alpha thalassemia is caused by deletions in the alpha-globin genes ($\alpha 1$ and $\alpha 2$) (Muncie & Campbell, 2009). Three deletions cause alpha TI, of which the most common form is hemoglobin H disease (Children's Hospital and Research Center Oakland, 2012; Vichinsky, 2012). Four deletions cause