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Superior Repositioning of the Maxilla in Thalassemia-Induced Facial Deformity: Report of 3 Cases and a Review of the Literature

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Management of thalassemia-induced skeletal deformity is a major concern in thalassemic patients. Various surgical approaches to correct the facial deformity, methods of fixation, and perioperative management for patients with thalassemia intermedia (the milder clinical form of the disease with a longer life expectancy) have been advocated; however, treatment remains controversial. We present a single-stage treatment modality and report 3 cases treated after correction of the hematologic condition.

Thalassemic syndromes are genetically determined disorders of hemoglobin synthesis with decreased production of either alpha or beta polypeptide chains of hemoglobin molecules, resulting from markedly decreased amounts of globin messenger RNA. The imbalance may result from various genetic lesions. As a group, the thalassemias are the most common single gene disorder.[1]

Thalassemia was first reported in the literature in 1925, when Cooley and Lee[2] described patients with severe anemia early in life associated with bone changes and splenomegaly. Thalassemia is attributed to a partial autosomal dominant gene; the homozygous state is termed thalassemia major, and the heterozygous state is designated thalassemia minor.[1] Thalassemia intermedia describes a disorder less severe than the major form but more severe than the minor form; approximately 10% of patients with the homozygous trait have the clinical syndrome of intermediate severity.[1] Thalassemia major, the homozygous state, is a serious condition that usually becomes apparent within the first year of life. In the untreated patient, hemolysis and profound anemia lead to marrow hypertrophy and hyperplasia. The anemia may be severe enough so as to cause death within 5 years of diagnosis, although regular blood transfusions can prolong life.[1,3]

Radiographically, the skeletal response to marrow proliferation involves expansion of the medulla, thinning of cortical bone, and resorption of cancellous bone, resulting in a generalized loss of bone density. In severely affected patients, a widening of the diploic space (medulla) with a thinning of the cortices occurs, frequently with complete obliteration of the outer cortex.[4] These bony spicules may be seen radiographically and have a classic "hair-on-end" appearance.[5,6] Patients with thalassemia intermedia are an interesting subset, constituting 10% of cases of thalassemia major.[7] They have a milder clinical form of the disease and a longer life expectancy; however, in many cases, this appears to be at the expense of extensive marrow hypertrophy and associated skeletal deformities.[1]

A follow-up study of 88 patients with thalassemia found a tendency toward a Class I malocclusion with anterior maxillary dentoalveolar protrusion.[8] Logothetis et al[9] studied 138 patients with thalassemia major and found that patient age, duration of clinical symptoms, degree of anemia, timing of splenectomy, and duration of transfusion therapy were important in determining craniofacial deformities. The older the patient, the more likely he or she is to have a facial deformity. Karagiorga-Lagna et al[10] studied 13 children with thalassemia major undergoing transfusion therapy and found that the skel-

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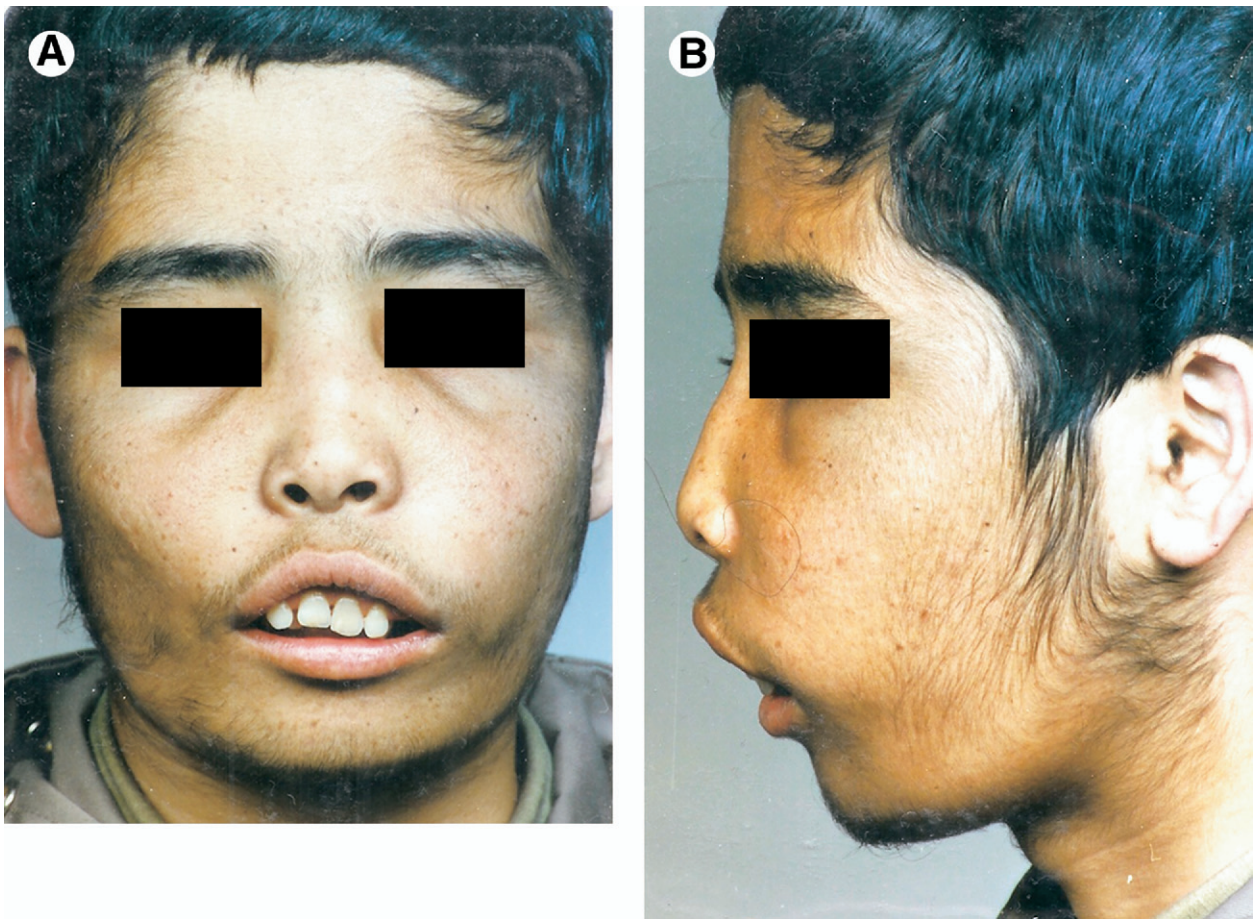

FIGURE 1. Typical and characteristic thalassemia-induced facial skeletal deformity (preoperative view).

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etal manifestations of thalassemia became normal with this transfusion therapy regimen.

The standard therapy for a patient with thalassemia is hypertransfusion of blood to maintain a hemoglobin level of 10 to 14 g/dL. Splenectomy is done in patients who develop hypersplenism.[1,3] The most serious complication of hypertransfusion is the development of iron overload, leading to sidrosis, especially of the myocardium, and secondary heart failure. Since 1977, long-term chelation therapy has been used to eliminate excess iron. This treatment, involving a slow subcutaneous injection of 1.5 to 2 g of deferoxamine 5 or 6 times per week, creates a negative iron balance, meaning that deferoxamine-induced iron excretion exceeds iron delivery by transfusion.[11]

In 1969, Jurkiewicz et al[12] reported 2 patients with major thalassemia who did not receive transfusion therapy and exhibited maxillary hyperplasia with distorted occlusion. Anterior segmental osteotomy with anterior shaving of the paranasal area was done for 1 of these patients; this patient required 2 transfusions during the surgery due to severe hemorrhage. The other patient was treated with a 2-stage technique, with anterior segmental osteotomy performed in the first stage and vertical reduction of the anterior segment done in the second stage.

In 1987, Weel et al[13] presented a 2-step technique for the treatment of a thalassemic African-American boy. The first step involved recontouring of the lateral maxilla; the second step, a Le Fort I osteotomy that allowed vertical reduction and posterior repositioning of the maxilla. In 1997, Drew et al[7] reported 3 patients with thalassemia intermedia treated with the Le Fort I osteotomy. The first patient underwent lateral shaving of the maxilla before the osteotomy. The hematologic situation remained uncorrected and caused hemorrhage during the operation and relapse of the skeletal deformity after 3 years.

Here we report 1 case of major thalassemia and 2 cases of thalassemia intermedia. In the patients with thalassemia intermedia, the hematologic condition was corrected, and facial growth was controlled during childhood. Orthodontic alignment of