INTRODUCTION

Thalassemia was first described in 1927 by Cooley et al. Thalassemia is a hereditary anemia caused by mutations affecting hemoglobin synthesis which manifest clinically with splenomegaly and bone abnormalities. Severe thalassemia, also referred to as Cooley’s anemia, is characterized by anemia, hepatosplenomegaly, bone deformities, growth abnormalities, and hemosiderosis.

Beta thalassemia, which consists of thalassemia major and minor, results from a decreased or absent production of the beta polypeptide chain found in normal adult hemoglobin. Hemoglobin A, which accounts for 95% of hemoglobin in adults and children over 1 year old, is a tetrad of two pairs of alpha and beta polypeptide chains. Thalassemia minor, or the heterozygous trait, is generally mild and often asymptomatic. There is an intermediate form described as well in which patients demonstrate anemia but are able to maintain hemoglobin levels in the 6 – 7 g/dl range without transfusion. These patients generally demonstrate minimal growth abnormalities. Thalassemia major, which refers to the severe homozygous form of thalassemia, carries an absolute requirement for transfusion due to anemia that could lead to death in early childhood if undiagnosed. An estimated 800 to 1000 individuals in the United States have homozygous beta thalassemia. Changes in the skeletal system result from a combination of extramedullary hematopoiesis and marrow hyperplasia in response to chronic anemia. The skeletal response to these insults manifest as expansion of the medulla, resorption of cancellous bone, and thinning of cortical bone. The purpose of this presentation is to alert the practicing otolaryngologist to the varied head and neck manifestations of thalassemia and their potential impact on diagnosis and management.

DISCUSSION

Specific changes involving the skull and facial bones are related to marrow overgrowth causing widening of the diploic space and expansile boney changes. The widening of the diploic space is responsible for the “hair-on-end” appearance frequently demonstrated on plain radiograph of the skull. Expansile changes of the frontal, temporal, and facial bones impedes pneumatization of the paranasal sinuses, however the ethmoid sinuses are characteristically uninvolved due to the lack of red marrow in their walls. Overgrowth of the maxillary bone can lead to lateral displacement of the orbits and ventral displacement of the central maxillary incisors causing the characteristic "rodent faces."

Multiple otolaryngologic disease manifestations as a result of phenotypical changes associated with beta thalassemia have been described. This patient population has also been demonstrated to have a high rate of nasal obstruction, sinonasal polyposis, and sinusitis. Dehiscence of the lamina papyracea is common and should be considered during any sinonasal surgery. Hearing impairment results as a conductive loss both due to bony overgrowth and extramedullary hematopoiesis in the middle ear as well as the external auditory canal. Eustachian tube dysfunction likewise occurs due to bony expansion in the skull base. Aggressive treatment with desferrioxamine leads to high frequency sensorineural hearing loss in a significant percentage of patients that frequently improves after the dose is lowered or the medication is discontinued. A significant increase in temporomandibular joint dysfunction and pain has been demonstrated. In addition, extramedullary hematopoiesis has been described within the maxillary sinus, and should be considered in this patient population when presenting with a sinus mass or opacification.

CASE PRESENTATION

A 24 year old male with beta thalassemia presented to our clinic with complaints of 2 months of thick green rhinorrhea, nasal congestion, and facial pressure. He had been treated on multiple courses of antibiotics under the care of his primary physician prior to referral to our clinic. He endorsed a history of multiple episodes of acute sinusitis in the past. Computed tomogram (CT) of the sinuses demonstrated relatively poor pneumatization of the maxillary and frontal sinuses with scant inflammatory changes and extensive expansile changes of the skull base and facial bones as well as bilateral lamina papyracea dehiscence. Prior to presenting to our clinic the patient had been managed with multiple 7 day courses of antibiotics. We initiated him on a steroid nasal spray and a 2 week course of appropriate antibiotic therapy. At 2 month follow up he endorsed improvement in his nasal airflow, nasal drainage, and facial pain.

CONCLUSIONS

In conclusion, beta thalassemia is a systemic disease that can have significant manifestations in the head and neck. While this is relatively uncommon, the otolaryngologist should be aware of the disease process and its effects on sinonasal and skull base anatomy and function.