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Caso clínico

CONDUCTIVE HEARING LOSS IN BETA-THALASSEMIA

BETA-TALASSEMIA E HIPOACUSIA DE CONDUÇÃO

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Abstract

Introduction: Beta-thalassemia is an autossomic recessive disorder characterized by a reduced or absent synthesis of the hemoglobin subunit beta, resulting in a microcytic hypochromic anemia.

Methods: We describe 2 cases of young individuals with beta-thalassemia minor and intermedia presenting with a conductive hearing loss. Computed tomography scans showed a possible osteolysis of the incus long crus on the first case and no relevant abnormalities on the second case.

Discussion: Chronic anemia is known to increase ineffective erythropoiesis with resultant extramedullary erythropoiesis. The possible existence of bone marrow in the middle ear and severe bone changes due to anemia may be responsible for stiffness in the middle ear transmission system, leading to conductive hearing loss.

Keywords: beta-thalassemia, hearing loss, extramedullary erythropoiesis.

Resumo

Introdução: A beta-talassemia é uma doença autossómica recessiva caracterizada por uma anemia microcítica hipocrómica resultante da ausência ou dimimuição da síntese da subunidade beta da hemoglobina.

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Métodos: Neste estudo descrevemos 2 casos de crianças com beta-talassemia minor e intermédia com hipoacusia de condução. As imagens de tomografia computorizada dos ouvidos sugeriram a presença de osteólise na longa apófise da bigorna no primeiro caso e ausência de alterações significativas no segundo caso.

Discussão: A anemia crónica acompanha-se de eritropoiese extra-medular devido à ineficácia da eritropoiese. A possibilidade de existir medula óssea nos ossículos e as anomalias ósseas decorrentes da anemia poderão provocar uma rigidez da cadeia ossicular e consequente hipoacusia de condução.

Palavras-chave: beta-talassemia, hipoacusia, eritropoiese extra-medular

patients with thalassemia intermedia present later in life with moderate anemia and do not require regular transfusions. Thalassemia minor is clinically asymptomatic but some individuals may have moderate anemia[3].

It has been estimated that about 1.5% of the global population are carriers of beta-thalassemia, with a total annual incidence of symptomatic individuals of 1 in 100,000 throughout the world and 1 in 10,000 people in the European Union. Because thalassemia heterozygosity confers some immunity against malaria, there is a particularly high incidence of thalassemia (2.5%-25%) in the Mediterranean basin, the Middle East, the tropical and subtropical regions of Africa, the Asian subcontinent, and Southeast Asia, where milder forms of the disease are most commonly seen[4].

Introduction

Thalassemia syndromes are a group of inherited hemoglobinopathies that result from significantly reduced or absent synthesis of normal hemoglobin[1].

Beta-thalassemia results from mutations of the beta globin gene leading to various degrees of defective beta chain production, an imbalance in globin chain synthesis, ineffective erythropoiesis and anemia[1]. Transmission is autosomal recessive in most cases, although dominant mutations have also been reported. Diagnosis of thalassemia is based on hematologic and molecular genetic testing[2].

Three main forms have been described: thalassemia major, intermedia and minor. While patients with thalassemia major usually present within the first two years of life with severe anemia requiring regular red blood cell transfusions,

Case Presentation

We describe 2 young patients with beta-thalassemia and conductive hearing loss.

The first patient was a boy with the diagnosis of beta-thalassemia intermedia, with no need for blood transfusions. He had short stature, delayed psychological development and bilateral valgus flatfoot. At 6 years old he was diagnosed with bilateral otitis media with effusion and was submitted to adenotonsillectomy and bilateral myringotomy with Shepard's tubes placement. Despite these procedures, a conductive hearing loss was still observed during follow-up consultations (Figure 1). The Computed Tomography (CT) scan showed bilateral myringotomy tubes well positioned with no significant changes in the middle and inner ear bilaterally (Figure 2).

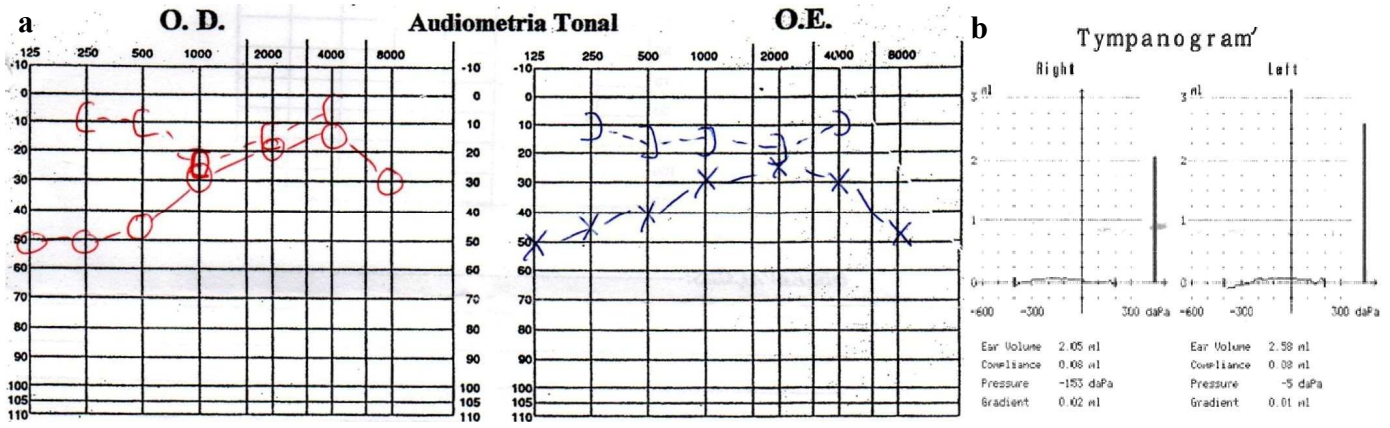

Figure 1: Audiogram showing bilateral moderate conductive hearing loss (a) and type b tympanogram bilaterally (b).

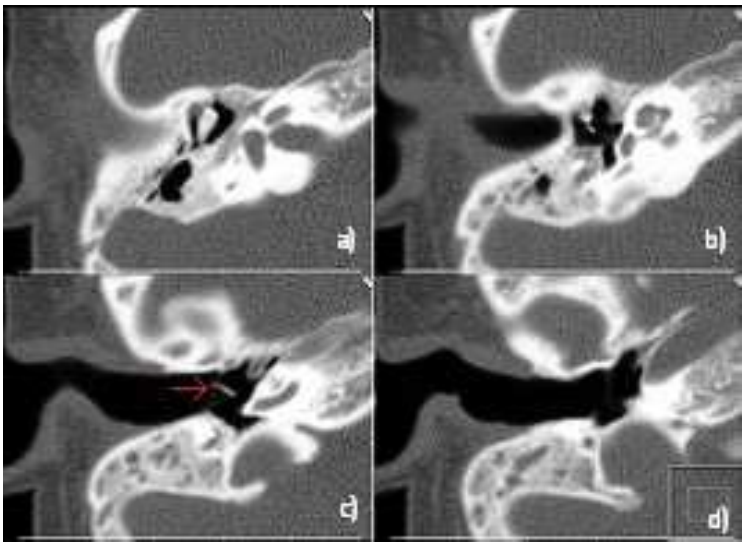

Figure 2: Axial CT scan images of the right ear showing myringotomy tube well positioned (arrow).

The second patient, a female carrier of beta-thalassemia minor, was first observed by an Otorhinolaryngologist at age 11 for recurrent otitis media. She presented with a left chronic stable otitis media and a right normal otoscopy. Despite the normal right tympanogram, the audiogram showed a bilateral conductive hearing loss (Figure 3).

The CT scan revealed no signs of active or sequelae of inflammatory processes on the middle ear and mastoid antrum bilaterally. There

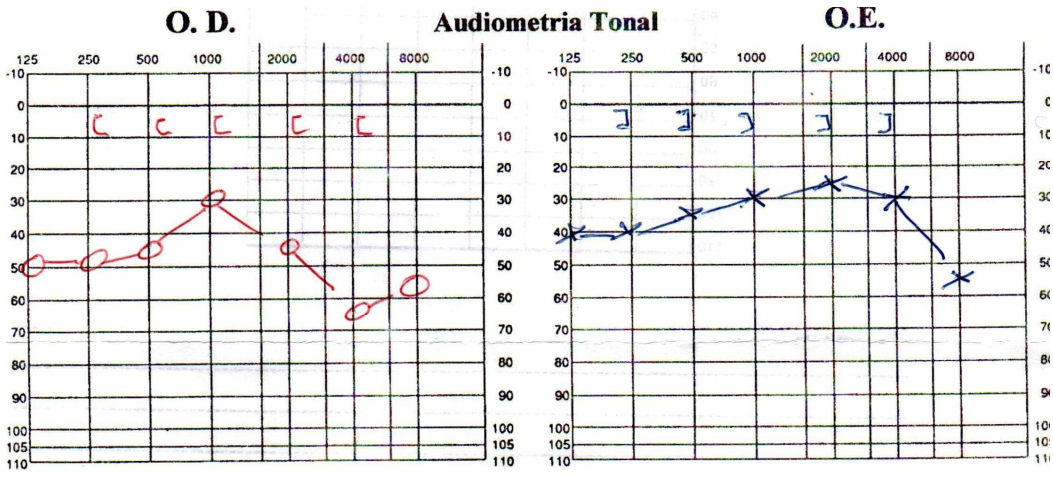

Figure 3: Audiogram showing bilateral moderate conductive hearing loss.

was a poor visualization of the right incus long process and mild signs of residual inflammatory changes on some right posterior mastoid cells. She was submitted to a bone anchored hearing aid implantation on the right side with good audiometric outcomes.

Discussion

Beta-thalassemia patients present with various degrees of microcytic hypochromic anemia and, in the most severe cases, regular red blood cells transfusion and lifelong iron chelation are often necessary.

Hearing impairment in beta-thalassemia patients was first described by De Virgilliis et al in 1979[5] and subsequently several studies strengthened the relationship between iron quelants and cochlear ototoxicity. However, little attention has been paid to the conductive hearing loss in patients without middle ear effusion.

In patients with chronic anemia, the increased erythropoietic drive results in hepatosplenomegaly and extramedullary erythropoiesis. In beta-thalassemia major patients, because of the intense bone marrow hyperplasia, expansion of facial bones occurs leading to increased prominence of the cheekbones with exposure of the upper teeth and production of frontal bossing. Pneumatization of sinuses is delayed and overgrowth of the maxilla produces severe malocclusion[6].

Transfusion therapy and anemia correction usually lead to suppression of erythropoiesis and inhibition of gastrointestinal iron absorption. In patients with mild or moderate anemia, like the ones described in this study, we verify an increased, although ineffective, erythropoiesis and bone marrow proliferation[2].

The possible existence of bone marrow in the middle ear and severe bone changes due to anemia may be responsible for stiffness in the middle ear transmission system[8]. Accordingly, Hazel et al. reported that less transfused patients had more severe bone changes and found evidence of bone marrow in the middle ear cleft[9]. These alterations may be responsible for the conductive hearing loss observed in our patients. The paper published by M. Önerci and colleagues favors this hypothesis as conductive hearing loss was found in beta-thalassemic patients without changes in tympanometric parameters[10].

However, more research on this topic is necessary in order to identify the real effects of bony alterations caused by beta-thalassemia in the middle ear ossicular chain. Nevertheless, attention must be paid to children with beta thalassemia and their auditory capacity, even in the mildest cases. Close observation and early diagnosis are key aspects on these patients, allowing for the establishment of a correct line of treatment and as soon as possible hearing rehabilitation.

Conflicts of Interest: No conflict of interest was declared by the authors.

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