doi: 10.5505/abantmedj.2015.96967

Abant Medical Journal

Olgu Sunumu / *Case Report*

Volume Cilt 4 *Issue* Sayı 2 *Year* Yıl 2015

Otalgia as the first presenting complaint in a patient with beta thalassemia: A Case report

İlk prezentasyon şikayeti otalji olan beta talasemi: olgu sunumu

Talih Özdaş[1], Kürşat Murat Özcan[2], Fatih Özdoğan[3], Gökhan Celbek[4]

[1] Yenimahalle Devlet Hastanesi, KBB Kliniği, Ankara
[2] Numune Eğitim ve Araştırma Hastanesi, 2. KBB Kliniği, Ankara
[3] Derince Eğitim ve Araştırma Hastanesi, KBB Kliniği, Kocaeli
[4] Yenimahalle Devlet Hastanesi, İç Hastalıkları Kliniği, Ankara

Özet

Beta talasemi gen mutasyonlarının sebep olduğu hematolojik bir hastalıktır ve Türkiye'yi de içeren tüm Akdeniz ülkelerinde önemli bir halk sağlığı problemi teşkil etmektedir. Talasemi intermedia talasemi major ve talasemi minor arasındaki bir ara formdur. Temporal kemik tutulumu talasemi intermediada oldukça nadirdir. Literatürde otalji ile birlikte olan ve talasemi minor tanısı alan vaka bulunmamaktadır. Bizim hastamız 10 aydır sol kulak ağrısı ve işitme kaybı çekmektedir. Bu vakada, yapılan tetkikler sonucunda talasemi intermedia tanısı alan ve manyetik rezonans görüntülemede temporal kemik tutulumu olan 44 yaşında bir erkek hasta sunulmaktadır.

Anahtar Kelimeler: Talasemi intermedia, otalji, temporal kemik.

Abstract

Beta thalassemia is a hematological disease caused by gene mutations and represents an important public health problem in all Mediterranean countries, including Turkey. Thalassemia intermedia is an intermediate form between thalassemia major and thalassemia minor. Temporal bone involvement is quite rare in thalassemia intermedia. There are no cases in the literature who presented with otalgia and was subsequently diagnosed with thalassemia intermedia. Our patient had suffered from left ear pain and hearing loss for 10 months. In this case report, a 44-year-old male patient is presented who was diagnosed with thalassemia intermedia based on the examinations performed and had temporal bone involvement on magnetic resonance imaging.

Keywords: Thalassemia intermedia, otalgia, temporal bone.

Introduction

Thalassemia is a group of hereditary hematological diseases in which there is an impairment of globin chain synthesis due to an unbalanced synthesis of alpha- and beta-globin chains during the production of hemoglobin molecule (1-3).Beta thalassemia is a heterogeneous hereditary hemoglobin disorder characterized by a decrease in beta-globin synthesis and divided into three groups as thalassemia major, minor and intermedia. Thalassemia intermedia is caused by a defective gene which causes a partial depression of the beta-globin protein production. In this disease, the lifespan of erythrocytes is decreased to a level that causes anemia but no regular blood transfusions are required (1-3).

The disease has a broad clinical spectrum ranging from one form that remains completely asymptomatic until adulthood to another form that causes growth and development retardation between the ages of 2-6 years. The degree of clinical severity in beta thalassemia is related to ineffective erythropoiesis, anemia and iron accumulation. Erythroid hypertrophy aggravates ineffective erythropoiesis in medullary and extramedullary regions, resulting in characteristic deformities in the head and face. Cortical thinning and pathological fractures of the long bones may be seen (2-4). Ear complaints are very rare in patients with beta thalassemia. In this article, we present a patient who presented with the complaint of otalgia and was subsequently diagnosed with beta thalassemia intermedia.

Case Report

A 44-year-old male patient admitted to our clinic with the complaints of pain and hearing loss in the left ear that had been present for 10 months. The otoscopic examination of the patient revealed an intact left tympanic

İletişim Bilgisi / Correspondence
Uzm. Dr. Gökhan Celbek, Yenimahalle Devlet Hastanesi, İç Hastalıkları Kliniği, Ankara
E-mail: drgokhancelbek@hotmail.com

Geliş tarihi / *Received*: 02.03.2014 **Kabul tarihi / Accepted:** 03.04.2014 **Çıkar Çatışması / Conflict of Interest:** Yok / None

membrane and a red-purple appearance was observed behind the tympanic membrane. No pulsation was observed (Figure 1).

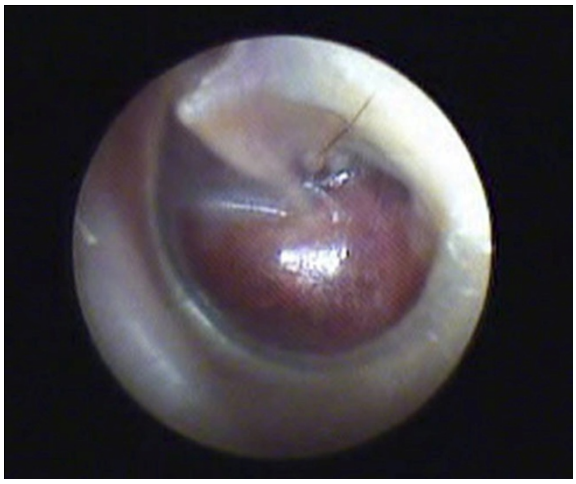

Figure 1. The otoscopic examination of the patient revealed an intact left tympanic membrane and a red-purple appearance was observed behind the tympanic membrane.

The patient had a history of surgery in the right ear 10 years ago and the graft membrane was intact. Pure tone audiogram revealed a mild hearing loss of 42/32 dB in the right ear and a severe hearing loss of 70/30 dB in the left ear (Figure 2).

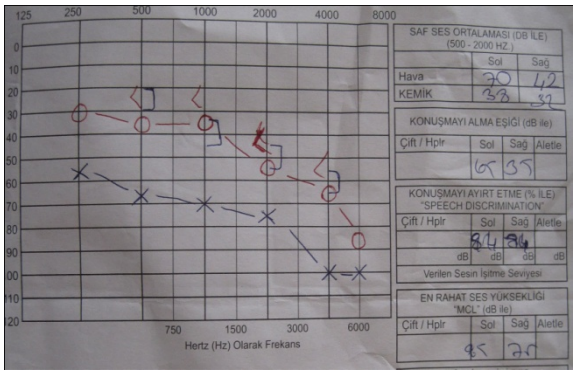

Figure 2. Pure tone audiogram

The computerized tomography of the temporal bone revealed a soft tissue filling the antrum and middle ear on the left side (Figure 3). The MR imaging of the temporal bone revealed a dense effusion in mastoid cells and middle ear on the left side and a hypointense active bone marrow signal on T1A and T2A sequences instead of normal medullary signals in the clivus, bilateral petrous apexes and other bony structures of the skull base (Figure 4).

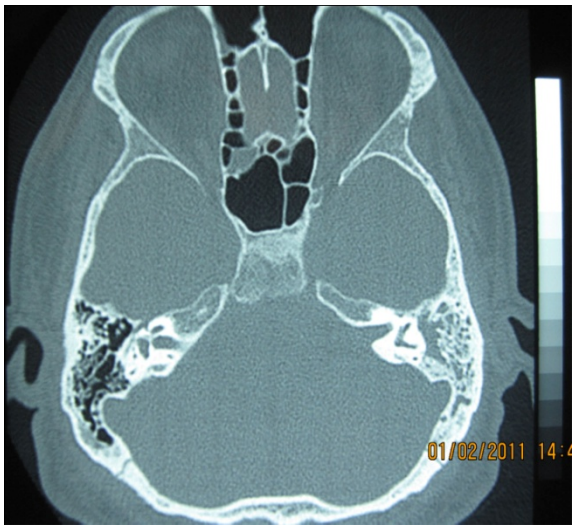

Figure 3. The computerized tomography of the temporal bone

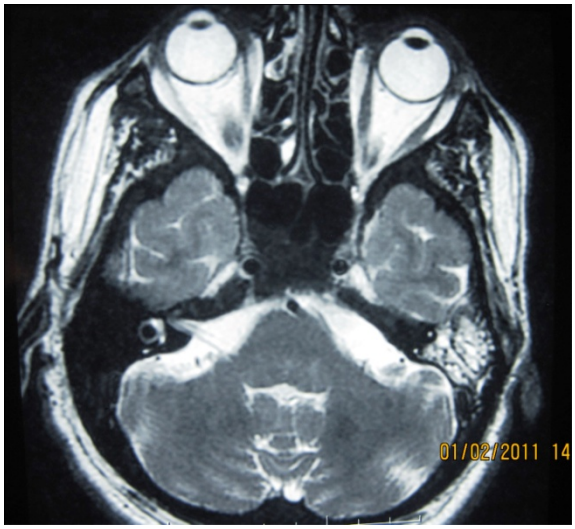

Figure 4. The MR imaging of the temporal bone

The patient was consulted to the hematology clinic with these findings. The complete blood count results were as follows: hemoglobin, 14.1 g/dl (13.6- 17.2); hemotocrit, 41.8% (39.5-50.3), MCV was 77.3 fl (80- 100), and MCH was 26.1 pg (27- 34), platelet count, 214 x 103 /µL (156-373); and erythrocyte count, 5.41 x 103 /µL (4.38- 5.77). The bone marrow aspiration smear revealed a marked increase in M:E ratio and megaloplastic / dysplastic changes in the erythroid series that were considered as primarily attributable to rapid proliferation. The hemoglobin electrophoresis of the patient showed that the hemoglobin F level was 83.4% (1-2%). Upper abdominal ultrasonography revealed hepatosplenomegaly. Thoracic and abdominal computerized tomography demonstrated a 35x36 mm soft tissue

bilaterally at the level of T9-11 vertebrae, which was considered as extramedullary hematopoiesis. The patient was diagnosed with beta thalassemia based on these findings and taken under treatment and follow-up by the hematology clinic. No ENT treatment was deemed necessary and the patient was taken under follow-up. The examination performed during 6-month follow-up revealed a decrease in ear pain and in the red-purple appearance behind the tympanic membrane.

Discussion

In patients with beta thalassemia intermedia, extramedullary hematopoiesis may be observed as erythropoietic tissue masses in the spleen, liver, lymph nodes, thorax and vertebrae due the increased bone marrow activity compensatory to chronic anemia and occurrence of normal blood cells outside the bone marrow. Lesions in close proximity to the chest and vertebrae may cause several neurological disorders, including spinal cord compression, paraplegia and cauda equina syndrome (1,5,6).

In their article published in 2007, Malik et al(5). reported that a 27-year-old male with thalassemia intermedia presented with paraplegia and spinal magnetic resonance imaging revealed a mass located in the paravertebral space between spinal levels T5 and T10 was associated with extramedullary hematopoiesis.

Similarly, in our case, extramedullary hematopoiesis was found as bilateral 35x36 mm soft tissue masses at the level of T9-11 vertebrae on thoracic and abdominal computerized tomography. These masses caused no neurological disorders.

In their case report published in 1992, Sheikha et al (7) reported the presence of temporal bone involvement in 3 siblings with thalassemia intermedia. One of these siblings, an 18-year-old male patient had been suffering from hearing loss in the left ear for 3 years. However, he had no ear pain.

Our patient had complaints of ear pain and hearing loss. Our literature review revealed no cases who presented with ear pain and was subsequently diagnosed with thalassemia intermedia.

Craniofacial abnormalies with maxillary hypertrophy may be seen in patients with thalassemia intermedia. Sheikha et al(7). noted that their patient had thalassemic facies with maxillary hypertrophy. Our patient did not have such a facial appearance.

Sheikha et al. (7) reported that the otoscopic examination of their case revealed intact pars tensa and granulation tissue superior to pars flaccida in the left tympanic membrane. The otoscopic examination of our patient revealed an intact left tympanic membrane and a red-purple appearance was observed behind the tympanic membrane. No pulsation was observed.

In case of red-purple discoloration in the tympanic membrane, differential diagnosis should include ear diseases such as acute otitis media, glomus tympanicum, serous otitis media, tympanic membrane hemangioma, vascular malformations of the middle ear, and temporal bone fractures as well as hematological diseases such as acute leukemia, lymphoma, and hemorrhagic diatheses as possible causes. For this purpose, complete blood count, hemostasis parameter tests and peripheral blood smear should be performed and magnetic resonance imaging should always be requested. Thereby, increase in morbidity and mortality is avoided by the prevention of an unnecessary surgical operation. In the case presented by Sheikha et al(7). mastoid exploration had been performed with the prediagnosis of chronic suppurative otitis media with cholesteatoma. Soft tissue was observed in the mastoid. A similar tissue was also observed in mesotympanum and hypotympanum around ossicular chain in the middle ear. Histopathologically, the tissue was considered as a hematopoietic tissue.

Physical examination of patients with beta thalassemia may reveal hepatosplenomegaly

(3). Physical and ultrasonography examination of our patient showed hepatosplenomegaly.

The complete blood count of patients with thalassemia intermedia generally reveal values between 7- 10 g/dl, 50- 80 fl and 16- 24 pg for Hb, MCV and MCH, respectively (1). In our patient Hb value was 14.1 g/dl, MCV was 77.3 fl, and MCH was 26.1 pg.

Temporal bone involvement in hematological diseases can be best demonstrated using magnetic resonance imaging. On the computerized tomography scan of the temporal bone, soft tissue may mimic cholesteatoma, causing unnecessary large surgical interventions. In the case presented by Sheikha et al(7). temporal bone computerized tomography showed an osteolytic cavity in the left mastoid and the patient underwent cholesteatoma surgery. In our case, the computerized tomography of the temporal bone revealed a soft tissue filling the antrum and middle ear on the left side. In our literature review, we could not find any studies where an MRI scan of a patient with beta thalassemia was presented. In our case, the MR imaging of the temporal bone revealed a dense effusion in mastoid cells and middle ear on the left side and signal changes of hypointense active bone marrow on T1A and T2A sequences instead of normal fatty medullary signal changes were detected in the clivus, bilateral petrous apexes and other bony structures of the base of the skull included in the area of examination. Surgical intervention was deemed unnecessary based on the MR imaging of the temporal bone.

In conclusion, beta thalassemia is an important hematological disease in all Mediterranean countries, including Turkey. Patients may very rarely admit to otolaryngology clinics with otological complaints due to temporal bone involvement. It should be kept in mind that a red-purple discoloration in the tympanic membrane may be a sign of a hematological disease and the patients should be evaluated in collaboration with the hematology clinic. In such cases, it should be kept in mind that computerized tomography of the temporal bone may cause misinterpretations and that magnetic resonance imaging should be used. Thereby, the delays in the diagnosis will be prevented as well as surgical interventions causing an unnecessary increase in mortality and morbidity will be avoided.

References

1. Galanello R, Origa R. Beta-thalassemia. Orphanet J Rare Dis. 2010; 21: 5-11.
2. Karakas Z. Beta Talasemi Intermedia. Turkiye Klinikleri J Pediatr Sci. 2007; 3: 34-41.
3. Aessopos A, Kati M, Meletis J Thalassemia intermedia today: should patients regularly receive transfusions? Transfusion. 2007; 47: 792-800.
4. Taher A, Isma'eel H, Cappellini MD. Thalassemia intermedia: revisited. Blood Cells Mol Dis. 2006; 37:12-20.
5. Malik M, Pillai LS, Gogia N, Puri T, Mahapatra M, Sharma DN, Kumar R. Paraplegia due to extramedullary hematopoiesis in thalassemia treated successfully with radiation therapy. Haematologica. 2007; 92: 28-30.
6. Castelli R, Graziadei G, Karimi M, Cappellini MD Intrathoracic masses due to extramedullary hematopoiesis. Am J Med Sci. 2004; 328: 299-303.
7. Sheikha A, Kameswaran M, Okafor BC, al-Saigh AA. Otological manifestations of thalassaemia intermedia: evidence of temporal bone involvement and report of a unique cholesteatoma-like lesion. J Laryngol Otol. 1992; 106: 316-321.