Case Report

Bagcilar Medical Bulletin 2018;3(1):14-16
DOI: 10.5350/BMB20180227073455

Co-occurrence of Chiari Type 1 Malformation, Syringomyelia, Anterior Thoracic Meningocele And Neurofibromatosis Type 1: A Very Rare Case Report

Abdurrahim Tas[2], Azmi Tufan[1], Ozgur Yusuf Aktas[1], Burak Eren[1], Ilker Gulec[1], Cihan Isler[3], Mustafa Safi Vatansever[1], Feyza Karagoz Guzey[1]

ABSTRACT

Background: Neurofibromatosis type 1 (NF-1) is an autosomal dominant hereditary disease associated with neurological and skeletal dysplasias. It is known that anterior thoracic meningocele, which is a rare pathology, may be associated with NF-1. Chiari type 1 malformation (CM-1) is a developmental disorder possibly caused by mesodermal deficiency frequently leading to syringomyelia. CM-1 may also be associated with NF-1. However, no case in which all of these four pathologies occur together has been reported in the literature.

Case Report: We present a 45-year-old female with known NF-1 without any neurological signs or symptoms. In her thoracic imaging performed for multinodular goiter investigation, an anterior thoracic meningocele was seen. Her spinal and brain magnetic resonance imaging (MRI) investigations revealed presence of CM-1 and syringomyelia as well. Although the patient was not neurologically compromised, surgical treatment for CM-1 and syringomyelia was offered because of the width of the syringomyelia. However, the patient did not accept the operation.

Conclusion: Although NF-1 may be associated with CM-1/syringomyelia or anterior thoracic meningocele, there was no case with all of these pathologies together reported in literature. This case reminded us that NF-1 cases, even in the absence of any neurological complaints, must be evaluated for a possible presence of spinal and brain pathologies.

Keywords: Anterior meningocele, Chiari malformation, neurofibromatosis type 1, syringomyelia

[1]Health Sciences University, Bagcilar Training and Research Hospital, Department of Neurosurgery, Istanbul, Turkey
[2]Health Sciences University, Gazi Yasargil Training and Research Hospital, Department of Neurosurgery, Diyarbakir, Turkey
[3]Istanbul University, Cerrahpasa Medical Faculty, Department of Neurosurgery, Istanbul, Turkey

Corresponding author:
Abdurrahim Tas,
Health Sciences University, Gazi Yasargil Training and Research Hospital, Department of Neurosurgery, Diyarbakir, Turkey
E-mail address: drabdurrahimtas@gmail.com

Date of submission: February 27, 2018
Date of acceptance: March 06, 2018

Citation: Tas A, Tufan A, Aktas OY, Eren B, Gulec I, Isler C, et al. Co-occurrence of chiari type 1 malformation, syringomyelia, anterior thoracic meningocele and neurofibromatosis type 1: a very rare case report. Bagcilar Medical Bulletin 2018;3(1):14-16.
https://doi.org/10.5350/BMB20180227073455

ÖZ

Chiari tip 1 malformasyonu, siringomiyeli, anterior torakal meningosel ve nörofibromatozis tip 1 birlikteliği: Çok nadir bir olgu sunumu

Amaç: Otozomal dominant kalıtsal bir hastalık olan nörofibromatozis tip 1 (NF-1) nörolojik bulgular ve iskelet displazileriyle gider. Nadir bir patoloji olan anterior torakal meningoselle NF-1 birlikteliği iyi bilinen bir durumdur. Chiari tip 1 malformasyonu (NF-1) olasılıkla embriyonal dönemde ortaya çıkan mezodermal yetersizlik nedeniyle gelişen bir hastalıktır ve sıklıkla siringomiyeliye neden olur. CM-1 de NF-1 ile birlikte bulunabilir. Ancak literatürde bu dört patolojinin birlikte görüldüğü bir olgu bildirilmemiştir.

Olgu sunumu: Bilinen NF-1 öyküsü olan ve bir yakınması olmayan 45 yaşında kadın olgu sunuldu. Multinodüler guatr incelemesi için yapılan torakal görüntülemede anterior torakal meningosel saptandı. Beyin ve tüm spinal manyetik rezonans görüntülemeleri (MRG) CM-1 ve siringomiyeli varlığını ortaya koydu. Hastada bir nörolojik bulgu olmamasına rağmen siringomiyelinin geniş olması nedeniyle CM-1 ve siringomiyeliye yönelik cerrahi girişim önerildi. Ancak hasta kabul etmedi.

Sonuçlar: Nörofibromatozis tip 1, CM-1/siringomiyeli kompleksi veya anterior torakal meningoselle birlikte görülebilirse de, literatürde bu dört patolojinin birlikte görüldüğü bir olgu bildirilmemiştir. Bu olgu bize herhangi bir nörolojik bulgusu olmasa da NF-1 olgularının beyin ve omurga patolojileri açısından incelenmesi gerektiğini hatırlatmıştır.

Anahtar kelimeler: Anterior meningosel, Chiari malformasyonu, nörofibromatozis tip 1, siringomiyeli

Introduction

Anterior thoracic meningocele is a quite rare pathology, and most of the cases had a comorbidity of NF-1, which is an autosomal dominant hereditary disease associated with neurological and skeletal dysplasias (1). It is thought that CM-1 is caused by mesodermal deficiency during the early embryonic period, and it is frequently seen together with syringomyelia. Also, co-occurrence of CM-1 and NF-1 is a well-known entity (2). However, there has been no case with all of these four entities in literature.

We report a case with CM-1, syringomyelia, NF-1, and anterior thoracic meningocele.

Case Report

A 45-year-old female with a known NF-1 and NF-1 family history was referred to our outpatient clinic because of the presence of cystic lesions related to her thoracic spinal canal seen on her thoracic imaging performed for multinodular goiter. There were no neurological complaints or findings in her examination except scoliosis at the cervicothoracic junction (Figure 1). A brain and whole-spine MRI was performed. A multiloculated anterior thoracic meningocele and erosion of the vertebral bodies were seen on her thoracic spinal MRI (Figure 2 c-e). In addition, there was a ca. 6 mm tonsillar herniation from the foramen magnum and a wide cervicothoracic syringomyelia (Figure 2a and b). There was no other pathology on the brain MRI.

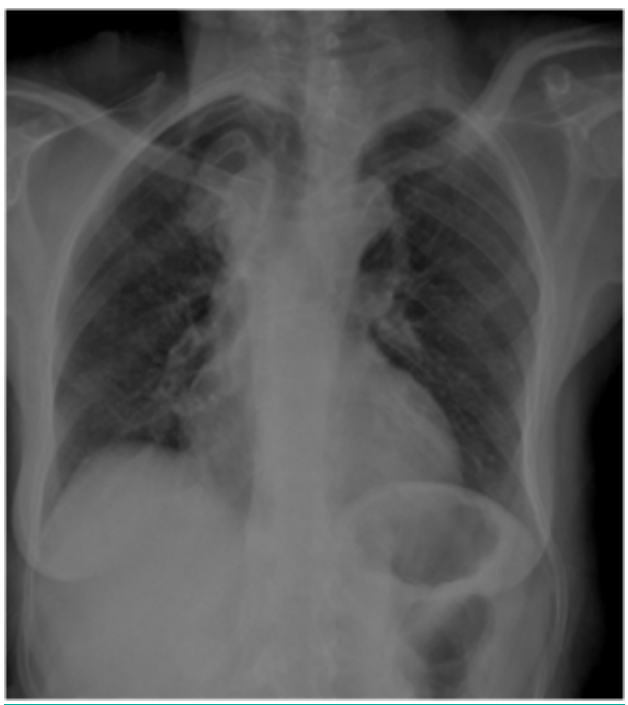

Figure 1: Thorax radiograph demonstrating cervicothoracic scoliosis.

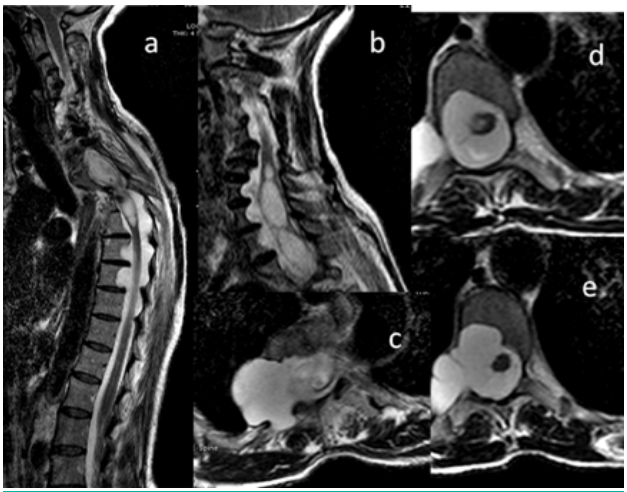

Figure 2: Sagittal cervical and thoracic spinal MRI sections showing wide syringomyelia (a and b), tonsillar herniation (b), and axial thoracic MRI sections from T5, T7 and T8 levels, respectively, showing anterior thoracic meningocele herniating from the right neural foramina and erosion of the vertebral bodies. (c, d, and f).

Although the patient was not in any way neurologically compromised, surgical treatment for CM-1 and syringomyelia was offered because of the width of the syringomyelia. However, the patient did not accept the operation. She was followed for 15 months. There were no findings at her neurological examination on her last outpatient visit. However, we subsequently lost contact with the patient.

Discussion

Chiari type 1 malformation is a well-investigated developmental disease that frequently causes syringomyelia. Mesodermal dysplasia develops during the early embryonic period in both NF-1 and CM-1. Therefore, it is thought that co-occurrence of these two entities is a logical condition (2). In a study, it is reported that 5.4% of children with CM-1 also had NF-1, and 8.6% of children with NF-1 had CM-1 (3).

Anterior thoracic meningocele is a rare pathology. While usually not presenting with any complaints, it may cause chest pain, radicular pain, headache, coughing, and dyspnea (3). It was seen radiologically as a well-shaped mass in the posterior mediastinum. Its relationship with the spinal

subarachnoid space may be shown with MRI or myelography (4,5). It has been reported that about 70% of the cases are associated with NF-1 (1). According to the most plausible theory, there were dysplastic sites on the spinal dura mater and a widening of the neural foramina in the NF-1 cases. Meninges may herniate from these weak sites by the force of the pulsatile pressure of the cerebrospinal fluid (CSF), and growing anterior meningoceles may develop (3,6,7). Anterior meningoceles frequently develop in the thoracic region because of weaker paravertebral muscles and lower intrathoracic pressure (8).

Although NF-1 is frequently associated with both CM-1 and anterior thoracic meningocele, the co-presence of these 3 pathologies in a patient is very rare. There was only one such case in the literature: Vanhauwaert and Deruytter (9) reported a case with a huge anterior thoracic meningocele and NF-1 that had been treated for CM-1 with foramen magnum decompression before. However, there was no syringomyelia in that case, and the sole case with NF-1, anterior thoracic meningocele, CM-1, and syringomyelia in the literature is the one presented in this article.

It is recommended to follow asymptomatic cases of anterior thoracic meningocele. Surgery is required in cases with symptomatic or growing cysts. In these patients, posterior, posterolateral, or anterolateral operations may be performed according to the size of the cyst (10). Vanhauwaert and Deruytter (9) reported good clinical results with a cystoperitoneal shunt in their case with a cyst so large that it caused a decrease in functional pulmonary capacity.

In our case, surgery was offered to the patient because of her wide syringomyelia. However, she did not accept the operation, and her follow-up was uneventful during 15 months.

Conclusions

Neurofibromatosis type 1 is related to both CM-1 and the syringomyelia complex and anterior thoracic meningocele because of mesodermal deficiency. However, it is extremely rare to see these four pathologies occurring together. This case is the first one in literature.

Contribution Categories	Name of Author
Follow up of the case	A.T., F.K.G., B.E., A.T., C.I.
Literature review	A.T., F.K.G., B.E., I.G., M.S.V., O.Y.A.
Manuscript writing	A.T., F.K.G., B.E., I.G., M.S.V., O.Y.A., A.T., C.I.
Manuscript review and revision	A.T., F.K.G., B.E., I.G., M.S.V., O.Y.A., A.T., C.I.

Conflict of Interest: Authors declared no conflict of interest.

Financial Disclosure: Authors declared no financial support.

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