DOI: 10.4274/tjo.55707

Case Report / Olgu Sunumu

Pachydermoperiostosis as a Rare Cause of Blepharoptosis

Nadir Bir Blefaroptozis Nedeni Pakidermoperiostozis

Özlem Yalçın Tök*, Levent Tök*, M. Necati Demir**, Sabite Kaçar***, Elif Nisa Ünlü****, Firdevs Örnek*****
*Süleyman Demirel University Faculty of Medicine, Department of Ophthalmology, Isparta, Turkey
**Yıldırım Beyazıt University Faculty of Medicine, Department of Ophthalmology, Ankara, Turkey
***Türkiye Yüksek İhtisas Education and Research Hospital, Department of Gastroenterology, Ankara, Turkey
****Süleyman Demirel University Faculty of Medicine, Department of Radiology, Isparta, Turkey
*****Ankara Training and Research Hospital, Ministry of Health, Department of Ophthalmology, Ankara, Turkey

Summary

A 37-year-old male patient diagnosed with pachydermoperiostosis at another center came to our clinic to rectify his blepharoptosis. The physical examination of the patient revealed skeleton and skin symptoms typical for pachydermoperiostosis. There was thickening and extending horizontal length of the eyelids, an S-shaped deformity on the edges of the eyelids, and symmetric bilateral mechanical blepharoptosis. In order to treat the blepharoptosis, excision of the thickened skin and the orbicular muscle as well as levator aponeurosis surgery was performed. The esthetic result was satisfactory. Pachydermoperiostosis is a rare cause of blepharoptosis. Meibomius gland hyperplasia, increase of collagen substance in the dermis, mucin accumulation are reasons of thickening the eyelid and finally this causes the development of mecanical blepharoptosis. Even though many two or three phased surgical procedures can be applied to these cases, satisfactory results can be achieved with levator surgery and excision of the thickened skin and the orbicular muscle as well. (Turk J Ophthalmol 2014; 44: 416-8)

Key Words: Pachydermoperiostosis, primary hypertrophic osteoarthropathy, ptosis

Özet

Başka bir merkezde pakidermoperiostozis tanısı konmuş 37 yaşındaki erkek hasta, bilateral blefaroptozisinin düzeltilmesi amacıyla kliniğimize başvurdu. Hastanın fizik muayenesinde pakidermoperiostozisin tipik bulguları olan iskelet ve cilt bulguları mevcuttu. Göz kapaklarında ise kalınlaşma, horizontal boyunda uzama, kapak kenarlarında S şeklinde deformite ve simetrik bilateral mekanik blefaroptozis vardı. Hastaya blefaroptozisi düzeltmek amacıyla kalınlaşmış cilt ve orbiküler kas eksizyonu ve levator aponevroz cerrahisi uygulandı. Estetik sonuç tatmin edici idi. Pakidermoperiostozis nadir görülen bir blefaroptozis nedenidir. Kapaklarda meibomian bez hiperplazisi, dermiste kollajen içeriğinin artması ve müsin birikimi nedeniyle kalınlaşma ve sonuçta mekanik bir blefaroptozis gelişir. Bu olgularda iki ya da üç basamaklı pek çok cerrahi işlem uygulanabileceği gibi levator cerrahisi ve kalınlaşmış cilt ve orbiküler kas eksizyonu ile de yüz güldürücü sonuca ulaşılabilir. (Turk J Ophthalmol 2014; 44: 416-8)

Anahtar Kelimeler: Pakidermoperiostozis, primer hipertrofik osteoartropati, ptozis

Introduction

Pachydermoperiostosis is a rare disease involving the skin and the skeletal system. Though the reason is unknown, it is suggested to be inherited in an autosomal dominant manner.[1] It is nine times more common in men than in women.[2] Findings present at puberty, and progression continues until adulthood. The disease is characterized by hypertrophic skin changes, clubbing of fingers, and periostosis. Dermatological findings are relatively severe and involve face, skull, skin, palms, and soles. Soft tissue hyperplasia leads to thickening and folding of the skin. Sebaceous gland hyperplasia causes enlargement of the sweat glands, leading to hyperhydrosis. The periostosis of bones and periosteal new bone formation stimulate the cortical thickening of the small bones of the hands and feet as well as

Address for Correspondence/Yazışma Adresi: Özlem Yalçın Tök MD, Süleyman Demirel University Faculty of Medicine, Department of Ophthalmology, Isparta, Turkey
Phone: +90 246 211 92 86 E-mail: esattok@yahoo.com **Received/Geliş Tarihi:** 08.09.2013 **Accepted/Kabul Tarihi:** 30.01.2014

of the distal ends of extremity long bones.[3] Thickening of the wrists and ankles is observed, and clubbing occurs in the distal phalanges. In the present study, we discuss the findings and treatment of a patient with pachydermoperiostosis, which is a rare cause of blepharoptosis.

Case Report

A 37-year-old male patient was referred to the oculoplastic clinic with a complaint of dropping of both eyelids. The general appearance of the patient revealed remarkable details. He had facial enlargement with deep folds in the skin of the forehead (Figure 1). His skin was thickened with coarse furrows. There were no signs of prognatism or macroglossia. Enlargement of the hands and feet and clubbing of the fingers were noted (Figure 2). Patient's plain radiographs of the extremities demonstrated symmetrical new bone formation involving the distal portions of the radius, tibia, fibula and the femur bilaterally. Contour irregularities and exophytic bone bulges were monitored at proximal epiphyseal-metaphyseal levels. Metacarpal thickening and contour irregularities were seen in the hands (Figure 3). The soft tissues of the hands were also increased.

We were informed that his complaints started at puberty. The patient was diagnosed with pachydermoperiostosis at age 19. We also learned that his brother also had a milder form of the disease. According to ocular examination, the best-corrected visual acuity was 20/20 bilaterally. Anterior segment examination revealed no pathology other than bilateral papillary reaction of the tarsal conjunctiva.

Eye movements, pupillary responses, and color vision were normal. Eyelids were considerably thickened, and there was an S-shaped curve in the upper eyelid margin. With the eyelids closed, the upper eyelids tended to override the lower eyelids, particularly in the lateral areas. The tarsal plate appeared quite lax and was everted easily. Vertical height of the tarsal plate was 14 mm bilaterally. Horizontal length of the eyelid was 42 mm bilaterally. Levator function was measured to be 14 mm bilaterally. Palpebral aperture was 3 mm bilaterally. Margin reflex distance was -1 mm bilaterally, and severe symmetrical blepharoptosis was present. A mechanical blepharoptosis was considered to have developed due to the thickened skin and enlarged tarsal plate.

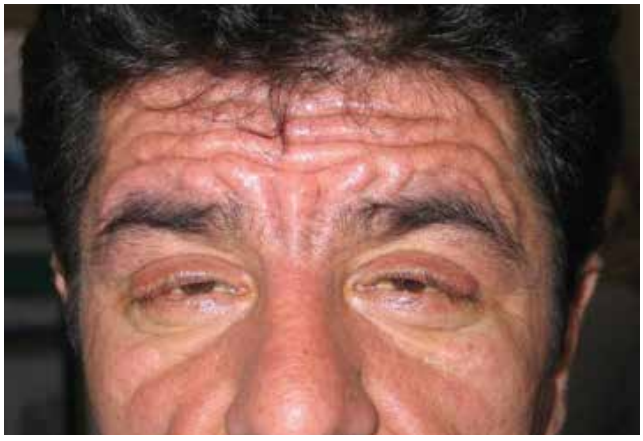

Figure 1. View of bilateral symmetrical ptosis and of skin folds

The patient underwent excision of the redundant skin and the orbicularis muscle as well as levator aponeurosis surgery. Intraoperatively, it was observed that the vessels lying between the Müller's muscle and levator muscle were greatly enlarged. Severe hemorrhage occurred. There were no postoperative problems. At final examination 28 months later, the result was observed to be satisfactory (Figure 4).

Figure 2. Clubbing of the digits and enlargement of the hand in pachydermoperiostosis compared with normal adult hand

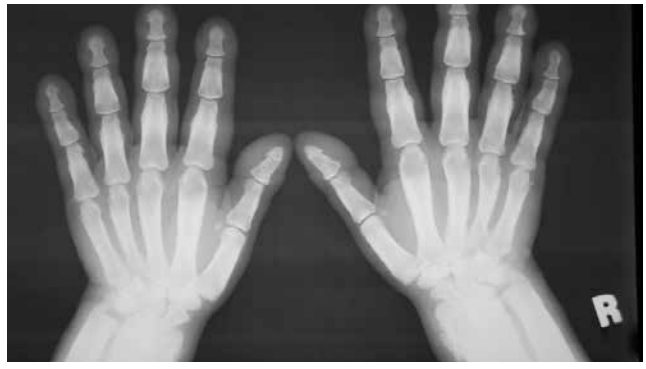

Figure 3. Patient's hand radiographs demonstrating contour irregularities and exophytic bone bulges at proximal epiphyseal-metaphyseal levels, thickening and contour irregularities at the metacarpus

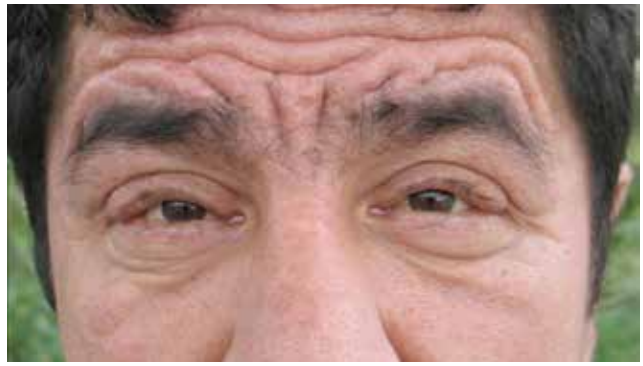

Figure 4. Postoperative view of the patient after 28th month of surgery

Discussion

The most common ocular associations of pachydermoperiostosis are blepharoptosis and tarsitis. Cutaneous changes also influence the eyelids, leading to thickening of the lids. The findings on light microscopy of the eyelids in pachydermoperiostosis are thickening and sclerosis of the connective tissue between the orbicularis oculi muscle and the tarsal plate as well as a thickened tarsal plate due to meibomian gland hyperplasia and epidermalization of the conjunctival epithelium.[2] Sebaceous hyperplasia and dermal mucin deposits are the most prominent histopathological findings responsible for the thickening of the eyelids and blepharoptosis.[1]

Pachydermoperiostosis might be confused with many diseases, including acromegaly. However, normal serum growth hormone levels and typical bony changes can assist in making a differential diagnosis. The disease differs from hypertrophic pulmonary osteoarthropathy due to absence of an underlying bronchial lesion. In the presence of symmetrical involvement of small and large joints, it might also be confused with rheumatoid arthritis. Because of the facial appearance, the differential diagnosis from leonine facies of leprosy, congenital syphilis, and myxoedema should be made.[1,4] Patients with pachydermoperiostosis should be presented to a wide range of medical specialists, including those in endocrinology, dermatology, rheumatology, and orthopaedics for multidisciplinary evaluation. Treatment in cases of pachydermoperiostosis targets the alleviation of symptoms. Non-steroidal anti-inflammatory drugs, corticosteroids, colchicine, and tamoxifen may alleviate polyarthritis. Retinoic acid can be used for dermal changes.[5] In addition, botulinum toxin type A can be injected to improve the appearance of wrinkles.[6] Surgical treatment is limited to the correction of cosmetic indications or existing deformities.

Blepharoptosis is often not severe in pachydermoperiostosis. In the literature, several methods have been described to correct the blepharoptosis. Arinci and colleagues reported excision of the excess skin and orbicularis muscle in a fusiform shape, levator aponeurosis plication, and wedge resection of the lateral third of the upper lid.[4] Kirkpatrick et al.[1] described tarsal excision via a conjunctival approach and wedge excision to shorten the horizontal length of the lid.

In pachydermoperiostosis, the increased skin thickness and the enlarged and heavy tarsal plate exceed the limit of the levator muscle, leading to the development of blepharoptosis. In the present study, without shortening the vertical and horizontal length of the lid, excision of the orbicularis muscle and skin was performed, and the weight of the lid was reduced. The levator complex was strengthened by levator surgery. At the end of a 28-month follow-up period, a satisfactory result was obtained. Due to the progressive nature of the pachydermoperiostosis, if blepharoptosis develops in the future, a horizontal lid shortening may be required in this patient.

References

1. Kirkpatrick JN, McKee PH, Spalton DJ. Ptosis caused by pachydermoperiostosis. Br J Ophthalmol. 1991;75:442-6.
2. Neufeld KR, Price K, Woodward J. Massive eyelid thickening in pachydermoperiostosis with myelofibrosis. Ophthal Plast Reconstr Surg. 2009;25:316-8.
3. Thapppa DM, Sethuraman G, Kumar GR, Elangovan S. Primary pachydermoperiostosis: a case report. J Dermatol. 2000;27:106-9.
4. Arinci A, Tümerdem B, Karan MA, Erten N, BüyükbabanI N. Ptosis caused by pachydermoperiostosis. Ann Plast Surg. 2002;49:322-5.
5. Hussain W, Coulson IH. Hyperhidrosis in association with keratoderma and strikingly large hands and feet: a rare case of pachydermoperiostosis. Clin Exp Dermatol. 2010;35:16-7.
6. Ghosn S, Uthman I, Dahdah M, Kibbi AG, Rubeiz N. Treatment of pachydermoperiostosis pachydermia with botulinum toxin type A. J Am Acad Dermatol. 2010;63:1036-41.