

# Palpebral ptosis caused by Pachydermoperiostosis

## *Ptose palpebral causada por Paquidermoperiostose*

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### ABSTRACT

*Pachydermoperiostosis is a rare disorder characterized by the involvement of skin and bone, and in some cases it can have a mild adverse effect in the eyelid. Although the etiology is still unclear, idiopathic or hereditary cases, in an autosomal dominant inheritance, have been reported. This study is a case report of a patient with severe blepharoptosis due to pachydermoperiostosis, which describes the surgical procedure and the physiopathology of the condition.*

**Keywords:** *Blepharoptosis/physiopathology; Blepharoptosis/surgery; Osteoarthropathy, primary hypertrophic/etiology; Case reports*

### RESUMO

A paquidermoperiostose é uma síndrome caracterizada por acometimento cutâneo e ósseo, e em alguns casos ocorre comprometimento palpebral leve. É uma síndrome rara, idiopática ou hereditária, com provável herança autossômica dominante de penetrância variável. Descreve-se o caso de um paciente com ptose grave por paquidermoperiostose elucidando sua fisiopatologia e conduta cirúrgica aplicada.

**Descritores:** Blefaroptose/fisiopatologia; Blefaroptose/cirurgia; Osteoartropatia hipertrófica primária/etiologia; Relatos de casos

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## INTRODUCTION

**P**rimary hypertrophic osteoarthropathy (PHO) or pachydermoperiostosis is a rare syndrome characterised by digital clubbing; periostosis (abnormal bone formation in the periosteum and periarticular tissue oedema), especially in the distal shafts of long bones; and pachyderma (thickening of skin in the face and scalp).<sup>(1,2)</sup>

It is associated with pain, polyarthritis, coarse facial features, “cutis verticis gyrata” (skin wrinkling in the face and scalp), seborrhoea, mild ptosis, hyperhidrosis, facial acne, and “elephant foot”.<sup>(3,4)</sup>

The aim of this paper is to report a case of PHO with severe ptosis and describe its surgical treatment.

## CASE REPORT

LB, a 31 year-old male patient from Belo Horizonte/MG, was referred to the Oculoplastic Surgery Department of the Ophthalmic Centre of Minas Gerais complaining of difficulty opening the left eyelid and discharge in the left eye (LE).

He reported having been diagnosed with pachydermoperiostosis by a rheumatologist when he was 18 years old. The patient had a history gastric ulcer treated surgically and evidence of patent ductus arteriosus on echocardiography, managed by a cardiologist who indicated surgical treatment for the condition.

Radiologic studies showed new periosteal tissue formation in the extremities and long bones.

On examination, the patient had a scar on the upper right eyelid and glabella due to a surgical procedure performed at another hospital 8 months earlier. His corrected visual acuity was 20/25 in both eyes.

He had severe ptosis on the LE (Figure 1A). The distance from the eyelid margin to the corneal reflex was 2 mm in the right eye (RE) and 0 mm in the LE. His palpebral fissure measured 9 mm in the RE and 2 mm in the LE. Excursion of the levator palpebrae superioris was 17 mm in both eyes.



**Figure 1A:** severe unilateral ptosis .

An increase in the vertical length of the upper left tarsus and lower tarsi was also observed, with tarsal hypertrophy and increased tarsal thickness (Figure 1B) confirmed by slit lamp biomicroscopy, as well as papillary reaction in the left tarsal conjunctiva.

The remainder of the eye examination was normal.

In addition to these ocular changes, the patient had digital clubbing, significant thickening of the skin with deep scalp furrows



**Figure 1B:** hypertrophy and increased thickness of the tarsal

and coarse facial features, hypertrophy of the extremities and “elephant feet”. He complained of joint pain and occasional dyspnea.

The patient underwent surgical correction of his LE ptosis through pentagonal resection of the entire lateral portion of the upper left eyelid supplemented by partial resection of the tarsus, conjunctiva and Muller’s muscle (Fasanella-Servat procedure). Resection of a segment of the entire thickness of the lower eyelids (Bick procedure) was also performed, associated with a tarsal strip and fixation in the lateral orbital bone edge. Eyelid biopsy found sebaceous gland hyperplasia.

The patient was prescribed ketorolac and eye lubricants to treat the discharge and papillary reaction.

Postoperatively, the patient’s eyelid slit measured 6.5 mm, and he was satisfied with the cosmetic result of the procedure (Figure 2).



**Figure 2:** one month postoperatively.

## DISCUSSION

PHO was first described in 1868 in two brothers by Friedreich, who considered it as an case of acromegaly<sup>(2)</sup>. Tourine et al (1935) characterised the condition as a distinct syndrome and stressed its similarities with pulmonary osteoarthropathy<sup>(3)</sup>.

Disease onset has a bimodal distribution with a peak in the first year of life and another at 15 years, coinciding with the period of rapid growth during puberty; it then progresses gradually over the next 5-20 years and eventually stabilises<sup>(5)</sup>.

PHO is more common in African Americans, and men are significantly more affected than women (7:1).

Three clinical forms of PHO have been described: (1) the complete form, with pachyperiostosis and pachyderma of the extremities and scalp and digital clubbing; (2) the incomplete form, with bone changes but without cutis verticys gyrata (isolated pachyderma of the extremities or face not affecting the scalp); and (3) the fruste form, with one or more skin changes but minimal or no bone abnormalities.

PHO can also be classified as: primary PHO, also called the Touraine-Solente-Gole Syndrome; and secondary PHO, known as the Pierre-Marie-Bamberger syndrome<sup>(6)</sup>. The secondary form is associated with chronic lung disease, hereditary congenital cyanotic heart disease, hepatobiliary disease, and paraneoplastic syndromes, and its clinical presentation is milder. Its onset occurs later, without familial or hereditary occurrence, and its bone changes are more acute and painful. Radiographic findings present certain differences. In primary PHO the periosteal reaction is irregular and poorly defined, while in secondary PHO linear bone deposits are observed.

The patient presented here can be classified as having the complete form of PHO, as there is evidence in the literature of an association between primary PHO and patent ductus arteriosus. Martinez-Lavin et al.<sup>(7)</sup>, after analysing their patients, concluded that PHO can be associated with patent ductus arteriosus.

According to the literature, PHO has been associated with a variety of disorders, including: gastrointestinal disorders (including gastric carcinoma, Crohn's disease, peptic ulcer disease, chronic gastritis, and Minetrier disease), myelofibrosis, gynecomastia, compressive neuropathy, hypoplastic internal genitalia, psoriatic onychopathy, periodontal defects, and spondylolisthesis.

Ophthalmic changes include mild ptosis, corneal scarring, corneal dystrophy of Bowman's membrane, cataract, and presenile macular dystrophy.<sup>(8,9)</sup>

Ptosis occurs due to thickening and hypertrophy of the tarsal plates, which in turn is caused by meibomian gland hyperplasia, increased sweat glands, and a thickened dermis. These changes increase eyelid thickness and length and produce mechanical ptosis, which can be associated with detachment of the levator aponeurosis.<sup>(6,8)</sup>

Ophthalmic involvement in PHO is uncommon, and our patient had severe mechanical ptosis. A review of the Brazilian ophthalmic literature found only one case report on the condition in the state of Ceará, published in Brazilian Archives of Ophthalmology in May/June 2005. Research on pachydermoperiostosis is more commonly done by rheumatologists and radiologists.

Correction of ptosis should be done with pentagonal resection of the tarsus to produce horizontal shortening of the upper eyelid and, if necessary, reinsertion of the levator aponeurosis. In the case presented here we also performed tarsal-conjunctival mullerectomy to reduce the vertical size of the tarsus, associated with the Bick procedure to produce horizontal shortening. We also proposed to supplement the procedure with resection of the levator palpebrae aponeurosis, as suggested by Alves et al.<sup>(6)</sup>, but the patient was already satisfied with the result.

The pathophysiology of PHO is still unclear, but its genetic nature has been demonstrated.

One hypothesis would be related to an intrinsic primary defect of platelets, facilitating degranulation and release of growth factors<sup>(1,2)</sup>. Other proposed aetiologies include a genetic influence, abnormal fibroblast activity, or changes in peripheral blood flow<sup>(10)</sup>.

Differential diagnosis includes acromegaly, diseases associated with coarse facial changes (the "lion face" in Hansen's disease, myxedema, congenital syphilis), and diseases that produce symmetrical periostitis (lymphoma, diffuse leishmaniasis, rheumatoid arthritis, chronic osteomyelitis, Paget's disease)<sup>(2)</sup>.

Treatment is usually done with analgesics and NSAIDs or

corticosteroids to relieve the associated polyarthritis. In some cases vagotomy can be performed to improve joint pain. Bisphosphonate can be a solution when other therapies fail. Use of sympathetic blockers and sympathectomy have been described to improve hyperhidrosis. Plastic surgery of the face and scalp can be performed for cosmetic reasons. Redundant skin and deep wrinkles can be excised through rhytidectomy.

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