

# A Rare Case of Pachydermoperisotosis (PDP) and Its Ocular Manifestations

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## ABSTRACT

A case of 56 years old Pakistani male visited OPD with complaint of thickening of both eyelids, ptosis and left lower lid ectropion caused by rare condition named pachydermoperiostosis (PDP). PDP is a rare autosomal dominant condition but autosomal recessive families probably can also occur. It is manifested by clubbing of digits, hyperhidrosis of palm and feet, peri-ostosis, acro-osteolysis and pachydermia. Ocular features include blepharoptosis, floppy eyelids, eyelid and palpebral conjunctival hypertrophy, mechanical ectropion, meibomian gland dysfunction, tear film abnormalities, punctate epithelial erosions and ocular surface disease. Surgical management was given by full-thickness wedge resection leading to horizontal tightening and this was done along with shortening of levator and its advancement. Histopathology demonstrated chronic non-specific inflammation and foreign body giant cell reaction.

**Key Words:** Eyelids, Blepharoptosis, Pachydermoperiostosis, Clubbing.

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## INTRODUCTION

Pachydermoperiostosis (PDP) is another name of primary hypertrophic osteoarthropathy. In 1868, Friedreich reported the first case of PDP.<sup>1</sup> Precise prevalence and incidence of the disease are still unknown. In males, this disease is 9 times more common and more severe than females.<sup>1</sup> PDP is a very rare hereditary disorder. It is autosomal dominant condition but autosomal recessive inheritance is also reported.<sup>2</sup> There are two subtypes; primary and secondary. Primary cases are rare inherited disorder and present soon after puberty. It is manifested by clubbing of digits, hyperhidrosis of palm and feet, peri-ostosis, acro-osteolysis and pachydermia.<sup>3</sup> Most prominent feature of PDP is facial involvement which includes thickening and furrowing of the face and scalp skin (resembling cutis verticisgyrate) and

sebaceous glands over activity. Ocular features include lengthening and thickening of the eyelids and chronic inflammation of tarsus secondary to palpebral tissues hypertrophy, which may results in mechanical ectropion. We report a case of Pachydermoperiostosis associated with both eyes, blepharoptosis and left lower lid ectropion.

## Case Report

We report this case considering the Declaration of Helsinki. A 56-year old male presented with complaint of heaviness in eyelids and problem in keeping both eyes open. He was offspring of consanguineous parents. In his family, his nephew had similar complaint of drooping of eyelids. On presentation, he had coarse facial features, oily facial skin and skin of scalp and forehead were wrinkled and thickened. Patient's both eyelids were massively thickened. He had bilateral blepharoptosis with marginal reflex distance (MRD) of 2mm and poor (0 mm) levator function. Patient was using predominantly the frontalis muscles to open his eyes. His left lower lid had mechanical ectropion (Figure: 1). Patient's hands and feet were larger than the normal and he had digital

clubbing (Figure: 2). He also had excessive sweating of the palms and soles. His vision was 20/120 in both



**Figure 1:** Showing thickened and wrinkled forehead and scalp skin, thickened eyelids with bilateral blepharoptosis, left lower lid was having mechanical ectropion.



**Figure 2:** Showing enlarged hands with distal clubbing.

eyes due to nuclear cataract. Extra-ocular movements and rest of the intraocular examination were normal. Patient's X-rays showed peri-osteosis, acro-osteolysis and soft tissue swelling of limbs joint (Figure: 3). Diagnosis of Pachydermoperiostosis associated with bilateral ptosis and left eye ectropion was made. In the surgical management of left eye, excision of excess skin was done after an incision at upper lid crease. The full-thickness wedge resection was done along with Levator aponeurosis shortening (15 mm) and advancement (2 mm) and then lateral tarsal strip was performed. Tissue from left upper and lower eyelid was submitted for histopathology evaluation. The result demonstrated chronic non-specific inflammation and foreign body giant cell reaction. After 6 months his both eyes were operated for cataract with gap of 1 month. Patient was referred to endocrinologist and dermatologist for further management.



**Figure 3:** Showing X-rays feet indicating Peri-osteosis, acro-osteolysis and soft tissue swelling.

## DISCUSSION

Hypertrophic osteoarthropathy is of two types. Primary hypertrophic osteoarthropathy, which is another name of Pachydermoperiostosis (PDP), accounting for 5% of all hypertrophic osteoarthropathy cases. Secondary hypertrophic osteoarthropathy is related to underlying cardiopulmonary disorders and malignancies and occurs predominantly in men of 30-70 years of age.<sup>4,5</sup> Clinically Pachydermoperiostosis are of three types: the complete includes peri-osteosis and pachyderma, incomplete includes only periosteosis but no pachydermia and for me fruste has pachyderma

but with mild or no periosteal involvement.<sup>6</sup> Our case belongs to complete type of PDP. The exact cause of PDP is yet not known.<sup>7</sup> In familial PDP cases, homozygous and compound heterozygous germ line mutations in the HPGD gene is detected, which encodes 15-hydroxyprostaglandin dehydrogenase (15-PGDH). Prostaglandin degradation mainly depend upon 15-PGDH and when it gets defective or absent, it results in excessive levels of prostaglandins, particularly PGE2 which contribute to the pathogenesis of PDP.<sup>4</sup> The prostaglandin transporter gene (SLCO2A1) mutations have been documented with pachydermoperiostosis. Mutations in SLCO2A1 deactivate PGE2 transport and leads to deregulation of PGE2.

Ocular manifestation of this disease is of particular importance which includes blepharoptosis, floppy eyelid syndrome, eyelid and palpebral conjunctiva hypertrophy, mechanical ectropion, meibomian gland dysfunction, tear film abnormalities, punctate epithelial erosions, and ocular surface disease.<sup>8</sup> In its surgical management, (as we did in our case), excision of the upper eyelid skin is done to correct both the vertical and horizontal dimensions of the eyelids.<sup>9</sup> Three step method of eyelid shortening, tarsectomy, and blepharoplasty was described in cases of delayed healing. However, upper and lower eyelid blepharoplasties and full-thickness wedge resections can be performed on both sides simultaneously.<sup>10</sup> We combined shortening and thinning of eyelid with shortening and advancement of levator muscle which effectively corrected ptosis, lid laxity and ectropion. Histopathology confirmed the diagnosis of Pachydermoperiostosis.

## CONCLUSION

Pachydermoperiostosis is a rare disease. It can present with ocular manifestations along with other systemic features. As in this case, patient presented with ptosis and mechanical ectropion but on further systemic evaluation, it was found that he had clubbing, periosteosis and pachyderma. Although rare, but such cases should be kept in mind while evaluating and managing the patients with ptosis, ectropion and eyelid thickening.

**Conflict of Interest:** Authors declared no conflict of interest.

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## Authors' Designation and Contribution

Fariha Sher Wali; Assistant Professor: *Concepts, Design, Literature search, Data acquisition, Statistical analysis, Manuscript preparation, Manuscript review.*

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