Background Pachydermoperiostosis (PDP) is an uncommon genetic syndrome characterized by distinctive digital clubbing, periostosis and pachydermia. PDP is a chronic condition that, while not life-threatening, decreases the patient's quality of life. Among other clinical manifestations, the thickened skin of the upper third of the face gives a leonine appearance to the face. There is no accepted therapy for alleviating the aesthetic defects associated with pachydermia.

Observation The aim of this study was to evaluate the role and long-term effect of botulinum toxin type A (BTX-A) in improving facial manifestations in patients with PDP. Three patients with PDP were treated with BTX-A. The main outcome measures were physician rating of wrinkle severity in relaxation, at baseline and after treatment. Secondary measures were patient global assessment of improvement. Case 1 and Case 3 were successfully treated with BTX-A; Case 2 reported an exacerbation of the eyelid ptosis possibly related to treatment. Case 1 underwent 4 sets of injections over a 48 week period which has never been described.

Key message We suggest temporal improvement of cosmetic appearance of patients with PDP could be achieved by BTX-A injection. Repeated treatments remained effective. The pathogenesis of PDP is thickening and folding of skin, we speculate that the reason that BTX-A contributes to the aesthetic improvement could be the inhibitory effects that BTX-A exerts on muscles. As the injected muscles relax, the surrounding tissue relaxes as well. In addition, injection of botulinum toxin type A into the dermal–subdermal layer has been anecdotally reported to improve skin texture and turgor. Although the pachydermia improved after treatment, the mechanism is still unclear. It remains to be confirmed the optimum technique for injection of BTX-A, and the optimum doses and treatment repetition regimen in PDP patients.