

Pachydermoperiostosis - a case report

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Abstract Pachydermoperiostosis is a rare autosomal dominant condition but autosomal recessive families probably can also occur. At least two gene mutations have been implicated, namely HPGD and SLCO2A1. This condition usually presents at puberty with progressive enlargement of the joints due to pachydermia, periostosis, and clubbing. Disease progresses for 5–20 years before stabilizing. We describe a case of 22 year old male who presented with thickened skin on the face and scalp (resembling cutis verticis gyrata), palmoplantar hyperhidrosis and clubbing.

Key words

Pachydermoperiostosis, cutis verticis gyrata, hyperhidrosis, clubbing.

Introduction

Pachydermoperiostosis is a rare autosomal dominant condition but autosomal recessive inheritance families probably can also occur. At least two gene mutations have been implicated, namely HPGD and SLCO2A1.¹ This condition usually presents at puberty with progressive enlargement of the joints due to pachydermia, periostosis, and clubbing. Other features include thickened skin on the face and scalp (resembling cutis verticis gyrata), palmoplantar hyperhidrosis, and acro-osteolysis. Disease progresses for 5–20 years before stabilizing.²

There are two subtypes namely Primary and Secondary. Primary cases are rare inherited disorder and present soon after puberty. Secondary Pachydermoperiostosis occurs predominantly in men aged 30-70 years. Bone changes are the most obvious features and skin changes are often relatively mild.³

Case report

A 22 year old unmarried male, presented with asymptomatic furrows on scalp and forehead for four years. The patient was in usual state of health four years back, when he started developing furrows on the scalp and forehead, which gradually got deeper. There was no history of trauma or any pre-existing dermatoses at these sites. He did not have history of seizures, headache, dyspnea, cough, chest pain or any other chronic illness including neuropsychiatric disorder. However, he gave a history of swelling of fingers and increased sweating but could not recall the exact duration. None of his family members suffered from such condition.

On clinical examination, his vitals were normal, systemic examination showed no abnormalities.

On cutaneous examination, there were deep furrows on the forehead as well as on the scalp (**Figure 1**). There was digital clubbing of finger and toe nails (**Figure 2**). There were no other skeletal abnormalities. Generalized hyperhidrosis was noted. There was no hypertrophy of tongue.

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Figure 1 Deep furrows of the forehead and scalp



Figure 2 Clubbing of fingers and toes

On investigation, Complete blood counts and thyroid function tests were within normal limits. X-ray chest and both hands, as well as ultrasonographic studies of abdomen and pelvis showed no abnormalities. Skin biopsy was not performed. Patient was counselled regarding the condition.

Due to the facial involvement, botulinum toxin was injected but no significant improvement was noted.

Discussion

The primary hypertrophic osteoarthropathy (HOA) (pachydermoperiostosis) is a rare hereditary disease characterized by skin manifestations (pachydermia), digital clubbing and proliferation of periosteum (periostitis) with sub-periosteal new bone formation. The

secondary form is associated with severe diseases like bronchiectasis, cystic fibrosis, congenital heart diseases, biliary cirrhosis, inflammatory bowel disease etc., and sometimes occurs as a part of paraneoplastic syndrome.⁴

It is a hereditary disease and many case reports have been published where there was family history suggestive of pachydermoperiostosis.^{5,6} However, our patient did not have a family history.

As this disease usually occurs in young adults, there could be a huge psychological impact on the quality of life.⁷

The treatment is usually symptomatic and is especially required if the patient has arthritis. Zhang H et al. has reported successful treatment of two cases of pachydermoperiostosis with

etoricoxib, aescin, and arthroscopic synovectomy.⁸

Cosmetic procedures such as facelift and botulinum toxin has been shown to improve facial appearance.¹

Our case is one of the few cases that decided on botulinum toxin treatment despite the high cost factor. This shows that the psychological impact is huge in this condition.

Conclusion

Although pachydermoperiostosis is a genetic disease and the clinical feature cause disfigurement of the body, treatment option like botulinum and athopaedic surgical techniques along with some days help alleviate the patient for the psychological trauma. A thorough history, examination and examination is mandatory to confirm the diagnosis

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