CASE REPORT

A RARE CASE OF PACHYONYCHIA CONGENITA TYPE-1 FROM PAKISTAN

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Pachyonychia congenita (PC) is a rare, genodermatosis inherited in an autosomal dominant pattern. Less than 500 cases have been reported in the literature worldwide. The disease commonly affects the nails with the typical findings of subungual hyperkeratosis and discoloured nails, skin thickening of the palms and soles causing focal palmoplantar hyperkeratosis and keratoderma. In some patients, the oral mucosa may also be affected by leukokeratosis, natal teeth and hoarseness of voice. There may be the presence of keratosis pilaris, epidermal cysts and steatocystoma multiplex. We present a classic case of pachyonychia congenita type 1 that fulfils the diagnosis with the typical clinical findings of subungual hyperkeratosis, palmoplantar keratoderma and oral leukokeratosis. Our case is one of the first few cases reported from a developing country like Pakistan in the literature.

Keywords: Pachyonychia congenita; Genodermatosis; Subungual hyperkeratosis

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INTRODUCTION

Pachyonychia congenita (PC) is a rare, autosomal dominant genodermatosis, first described by Muller in 1904.[1] Since the discovery of the disease only 450 cases have been reported in literature. This disease mainly affects the nails and palmoplantar skin but other areas like the oral mucosa, larynx, tongue, teeth and hair may also be affected.[2] The current classification is based on the genetic mutations of one of the five keratins: KRT6a, KRT6b, KRT6c, KRT16, and KRT17.[3] We present a case of PC in a young male who was treated with acitretin.

CASE REPORT

A 23-years-old male, resident of Karachi, Pakistan, presented to the outpatient clinic of dermatology department with the complaint of social embarrassment due to thickened nails of both the hands and feet along with thickening of the skin of palms and soles for the past 12 years. According to the patient, he was in his usual state of health 12 years back when his nails gradually started to thicken with yellowish discoloration. All twenty nails were affected simultaneously. This thickening and discolouration was not associated with any pain or pruritus. The patient denied any history of trauma or infection of the affected nails. The patient initially ignored the symptoms but now wanted to seek medical attention due to his social anxiety and embarrassment. His family history was non significant and no any other family member affected with the same disease. He was a product of non-consanguineous marriage. On examination of the hands and feet, there was markedly gross subungual hyperkeratosis (wedge-shaped nails) and yellowish discoloration of all twenty nails with mild focal palmoplanter keratoderma over the pressure points (figure 1, 2 and 3). On further inquiry, the lesions were not painful but, there was slight discomfort with the palmoplantar keratoderma. The examination of the oral cavity revealed leukokeratosis. There was gross brown staining of the teeth due to betel nut addiction along with poor dental hygiene (figure 4). The examination of the scalp and hair revealed normal hair with no abnormal texture and normal trichoscopic findings (figure 5). The body was examined for cutaneous cysts, keratosis pilaris and steatocystoma multiplex which was normal. He complained of hyperhidrosis which was apparent on examination of his palms and soles. His voice however, was normal with no hoarseness or any other change overtime. There was no retention of natal teeth.

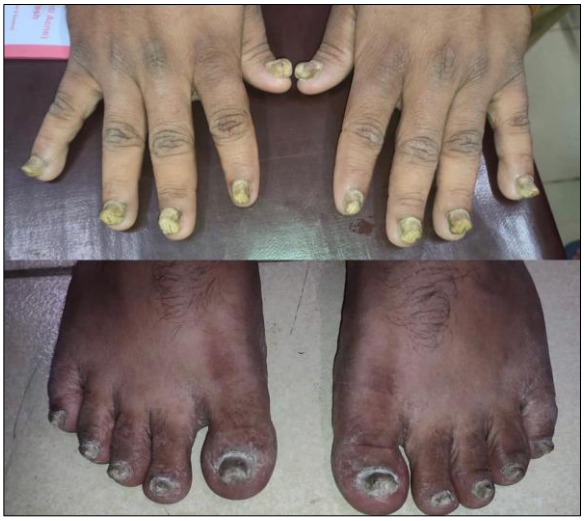

Figure-1: Grossly thickened and discolored nails of the hands and feet

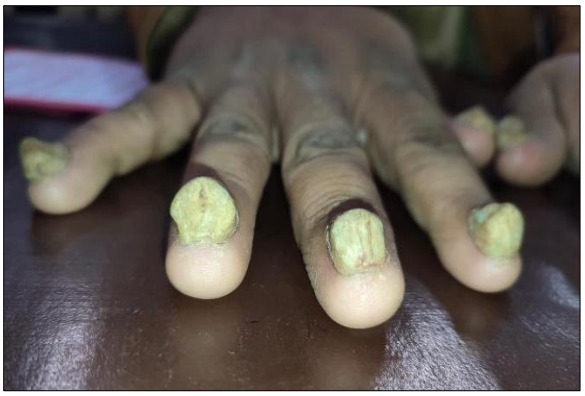

Figure-2: Severe subungual hyperkeratosis causing significant elevation of the nail from the nail bed and hoof like nails

Figure-3: Focal palmar hyperkeratosis

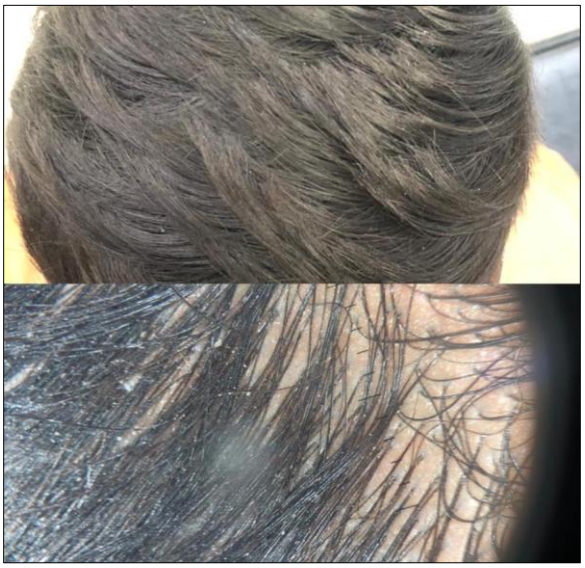

Figure-4: Normal hair texture and trichoscopic findings

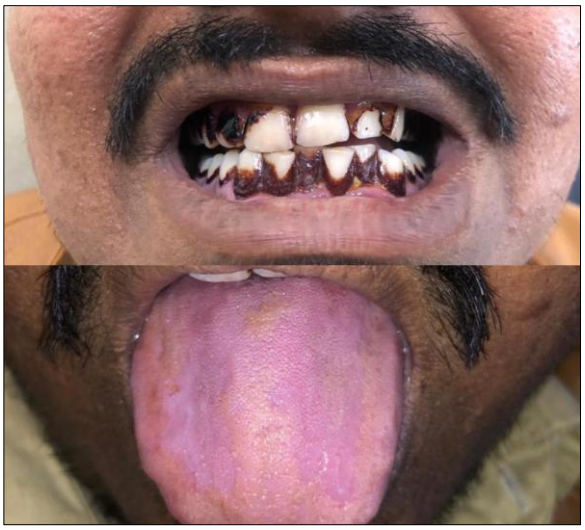

Figure-5: Dental staining due to betel and mild leukokeratosis

We made a clinical diagnosis of Jadassohn-Lewandowsky type pachyonychia congenita (PC type 1; mutations in K6a or K16). Due to the poor financial status of the patient and unavailability of genotyping facilities in Pakistan the plan to perform genotyping testing was deferred. This disorder is clinically identified and our patient is a classic case with principle features of pachyonychia congenita. He was advised regular cutting, filing and grinding of the nails after soaking along with application of topical keratolytics and emollients for the keratoderma. He was counselled about the disease, its features and the availability of very few treatment options. Considering the option of oral acitretin, our patient was worked up with liver function tests, ultrasound abdomen and lipid profile. He had a deranged lipid profile due to which the commencement of oral acitretin was deferred. We plan to follow our patient routinely along with regular monitoring of his lipid profile.

DISCUSSION

PC is characterized by onychodystrophy, subungual hyperkeratosis of the distal nails, palmoplantar hyperkeratosis and keratoderma. PC may also show features of oral leukokeratosis, natal teeth, hoarseness and laryngeal involvement, coarse and twisted hair, follicular keratosis and cutaneous cysts.[2] The prevalence of PC is 0.9 cases per million with an overall affected population between 5000 to 10,000 patients worldwide.[4] Around 30% of the cases can arise sporadically without a family history as seen in our patient.

According to the keratin mutations, PC is divided into two subtypes; PC-1 ((Jadassohn–Lewandowski type) characterized by oral leukokeratosis and mutations in KRT6a or KRT16;

and PC-2 (Jackson–Lawler type) due to mutations in KRT6B or KRT17, featuring cutaneous cysts, eruptive vellus hair and natal teeth.[1] PC-1 is the more common variant with mainly nail dystrophy and thickening present in 90-98% of cases of PC-1.[1] Another common association is the palmoplantar keratoderma which is the most debilitating symptom affecting quality of life in most patients.[4] In most patients the pachyonychia presents by the first year of life. According to the study by Samuelov L, it is seen that 20 nails are involved in 50% of the cases which was the case in our patient.[4] Painful palmoplantar keratoderma usually develops between 1-9 years of age. As it is the most debilitating symptom, most treatment plans are targeted to manage it.[5] Topical options, such as keratolytics, emollients, retinoids and steroids have been used but have limited results.[5] Oral retinoids are considered the primary treatment option for PC. They reduce hyperkeratosis and hence pain associated with it. In a study by Gruber *et al*., it was found that there was a 50% and 14% improvement in hyperkeratosis and pachyonychia respectively.[5]

Response to treatment varies from patient to patient and the treatment needs to be modified according to the patient's clinical phenotype. Being one of the rare disorders, it is important to highlight the clinical cases reported in the literature. Our case appears to be one of the first few cases reported from Pakistan. This case highlights the importance of correlating clinical features and devising a treatment plan best suited for the patients to improve their quality of life.

AUTHORS CONTRIBUTION

NS: Wrote most of the paper. SK: Drafted the paper. HT: Provided revisions to the scientific content of the manuscript. RM: Provided revisions to the scientific content of the manuscript. PM: Provided revisions to the grammatical/ stylistic errors of the manuscript

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