International Journal of Science and Research (IJSR)
ISSN: 2319-7064
SJIF (2022): 7.942

Jadassohn - Lewandowsky Syndrome: A Case Report of a Rare Disease

Dr Sharavi Budanur, Dr Rashmi Sriram

1. Introduction

Pachyonychia congenita is a very rare genetic disorder affecting skin and nails. It was first documented by muller in 1904. It has an autosomal dominant mode of inheritance and is classified into four types of which, the type 1/Jadassohn–lewandowsky type and the type 2/ Jackson lawler types are of importance. The clinical features reported in all types include toe nail dystrophy, plantar keratoderma and plantar pain.

Here we report a case of Pachyonychia Congenita type 1 who visited outpatient department in our hospital

2. Case Report

An 18 year old male patient presented with multiple elevated skin lesions over his chest and abdomen with focal thickening over bilateral palms and soles present since childhood. The lesions were insidious in onset, not associated itching. He complained of mild to moderate intensity pain on palms and soles. He was born out of a second degree consanguineous marriage with unremarkable birth and family history.

On examination – There were multiple well defined hyperpigmented, hyperkeratotic papules, few coalescing to form plaques over chest and abdomen. There was diffuse hyperkeratosis over palms and soles

Mucosal examination revealed asymptomatic oral leukokeratosis on dorsum on tongue

All the nails showed subungual hyperkeratosis with brownish black discoloration

Routine laboratory investigations including complete hemogram, liver function test and renal function test were within normal limits. Punch biopsy was performed from a hyperkeratotic papule on abdomen and from the diffuse hyperkeratosis over palm. Histopathology of the papule revealed epidermis with focal papillomatosis, acanthosis and lamellated hyperkeratosis, increase in basal melanin pigmentation was also noted. The sample from palm showed features of orthokeratotic hyperkeratosis with focal parakeratosis. Epidermis also showed spongiosis and mild acanthosis suggestive of epidermolytic hyperkeratosis. The upper dermis showed fibrocollagenous tissue with mononuclear infiltrate. With the above clinical history, cutaneous examination revealing hyperkeratotic papules and plaques over chest and abdomen, supported by histopathological reports, a diagnosis of pachyonychia congenita was made. Patient was started on tablet Acitretin 10mg daily night for 3 weeks with moisturizers for topical application

However Genetic and molecular biological studies for confirming the diagnosis could not be carried out due to lack of infrastructure facilities.

3. Discussion

Pachyonychia congenita is a rare genodermatosis with autosomal dominant mode of inheritance. An estimated 5, 000 – 10, 000 cases have been reported worldwide. It as associated with mutations of keratin genes KRT6A, KRT6B, KRT16, KRT17[2]. Keratins are structural proteins which are very essential for maintaining integrity of the cell. Mutations in the genes encoding these keratins lead to increased cell fragility

Feinstein et al classified this into four clinical types[3]

***Type I* pachyonychia congenita aka Jadassohn - Lewandowsky syndrome –** It is the commonest type seen in around 50% cases. Clinical features of this include painful palmoplantar hyperkeratosis, follicular hyperkeratosis, cysts and oral leukokeratosis. The nails have distal prominent thickening with hypercurvature due the prominent nail bed keratosis[3],

***Type II* pachyonychia congenita**, aka **Jackson - Lawler syndrome** – clinical features in this type include natal teeth (15 - 50%), cutaneous cystic lesions, disorders of scalp and eyebrow hair, corneal dystrophy, steatocystoma multiplex[4],

***Type III* pachonychia congenita aka Schafer–Branuer syndrome** – the patients present with angular cheilitis, corneal dyskeratosis, and cataracts.

***Type IV* pachyonychia congenita –** In this type, along with features of type 3, patients also present with laryngeal hoarseness, mental retardation and hoarseness of voice[5]

Jadassohn Lewandowsky syndrome results from mutations in genes encoding epidermal keratinocyte keratins, specifically the 1A and 1B helical encasing regions of keratins. K6a, K6b, K16, and K17 are the most frequent sites of mutations. A characteristic finding of this disease is subungual hyperkeratosis leading to the elevation and increased transverse curvature of the nail plate. This elevation is most pronounced distally resulting in an omega or pincer nail deformity The affected nail plates are discoloured, thick, and friable, and may even fail to reach the distal fingertip[6]. The diagnosis of this disease is mainly on clinical findings or through genetic analysis identifying variations in one of the five keratin genes. offsprings of affected individuals have 50% chance of inheriting the disorder. Prenatal testing for a pregnancy at increased risk can be done[7].

There is no specific management for pachyonychia congenita and management chiefly revolves around management of hyperkeratosis. Foot care involves paring down hyperkeratotic areas with razors. Humectants like propylene glycol and α - hydroxy acids like salicylic acid may help in removal of the outer keratin layers. Periodic soaks in dilute bleach can be used to reduce microbial colonization. Systemic retinoids like Isotretinoin and Etretinate can be used to treat and reduce hyperkeratosis. In our case too, patient was treated with tablet acitretin 10 mg daily night for 3 weeks. Patient showed excellent response to treatment with acitretin. A few studies have also reported using botulinum toxin A for successful management of pain and hyperkeratosis of palms and soles in patients with pachyonychia congenita[8]

Though pachyonychia congenita is transmitted through autosomal dominant mode of inheritance, a few sporadic cases have been mentioned in literature. A case report by Kishan Kumar YH et al highlights a sporadic case of pachyonychia congenita in a 20 year old female born out of a consanguineous marriage with no similar lesions in family[9]. In our case too the patient was born out of a second degree consanguineous marriage and had no similar lesions in family.

4. Conclusion

The above case of pachyonychia congenita type 1 / Jadassohn Lewandowsky syndrome has been reported for its rarity and excellent response to treatment with acitretin. Genetic testing as a tool for diagnosing pachyonychia is often expensive causing financial burden to patients. The facilities and infrastructure for genetic testing are also very limited. We emphasize here the importance of recognising the clinical features of this rare disease and using histopathology as a diagnostic tool for treatment, thus saving time and reducing the cost of treatment for these patients.

References

[1] McLean WH, Hansen CD, Eliason MJ, Smith FJ. The phenotypic and molecular genetic features of pachyonychia congenital. *J invest Dermatol* 2011; 131: 1015–17.
[2] Smith FJD, Corden LD, Rugg EL, et al. Missense mutations in keratin 17 cause either pachyonychia congenita type 2 or a phenotype resembling steatocystoma multiplex. J Invest Dermatol.1997; 108: 220
[3] Feinstein A, Friedman J, Schewach - Millet MS. Pachyonychia congenita. J Am Acad Dermatol.1988; 19: 705.
[4] Rathore PK, Khullar V, Das A. Pachyonychia Congenita Type 1: Case Report and Review of the Literature. Indian J Dermatol.2016 Mar - Apr; 61 (2): 196 - 9.
[5] Su WPD, Chun SI, Hammond DE, Gordon H. Pachyonychia congenita: a clinical study of 12 cases and review of the literature. Pediatr Dermatol.1990; 7: 33.
[6] Agrawal, Sanjay, et al. "Pachyonychia congenita type - 1 (Jadassohn - Lewandowsky syndrome). " *Indian Journal of Paediatric Dermatology*, vol.15, no.3, Sept. - Dec.2014, p.137.
[7] Smith, Frances JD, et al. "Pachyonychia Congenita. " *GeneReviews®*, edited by Margaret P Adam et. al., University of Washington, Seattle, 27 January 2006.
[8] Milstone LM, Fleckman P, Leachman SA, Leigh IM, Paller AS, van Steensel MA, Swartling C. Treatment of pachyonychia congenita. J Investig Dermatol Symp Proc.2005 Oct; 10 (1): 18 - 20
[9] Kishan Kumar YH, Keerthi S. Pachyonychia congenita affecting nails only: A sporadic case or novel mutation of an uncommon genodermatoses. Int J Health Allied Sci 2017; 6: 191 - 3

Figures

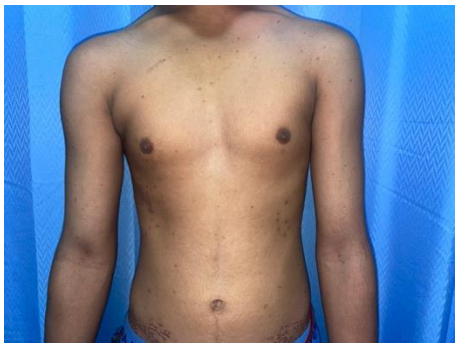

Figure 1: Well defined hyperpigmented, hyperkeratotic papules over chest and abdomen

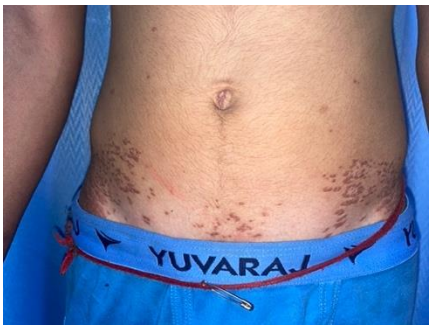

Figure 2: Well defined hyperpigmented, hyperkeratotic papules coalescing to form plaques over hips

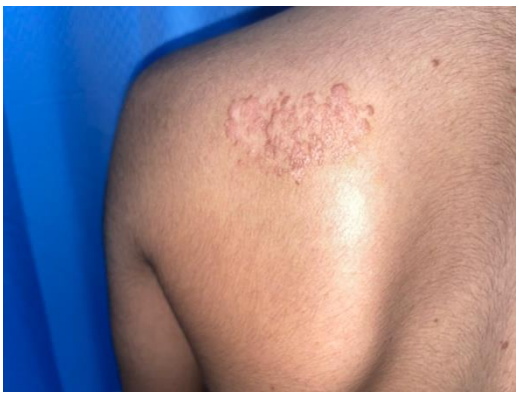

Figure 3: Well defined hyperpigmented, hyperkeratotic papules coalescing to form plaques over back

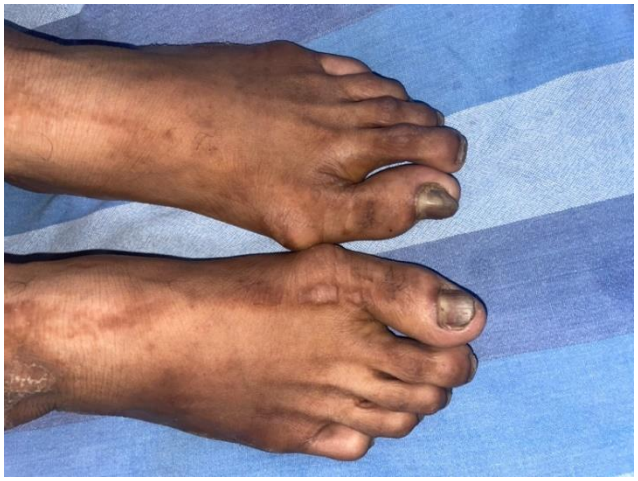

Figure 4: Subungual hyperkeratosis seen in nail beds of both feet

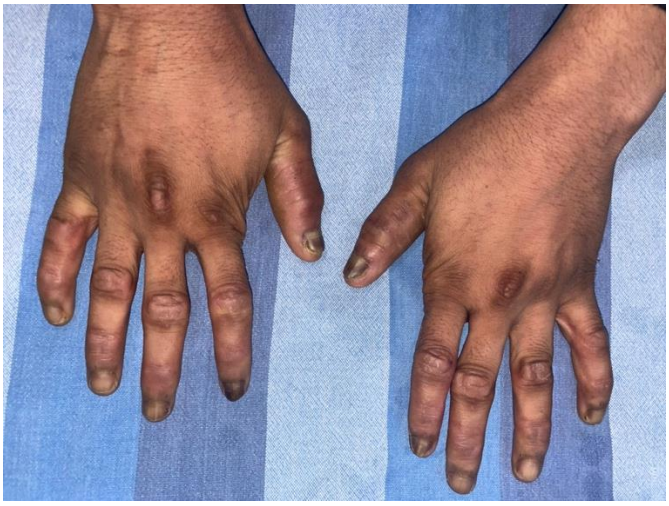

Figure 5: Subungual hyperkeratosis seen in nail beds of both hands

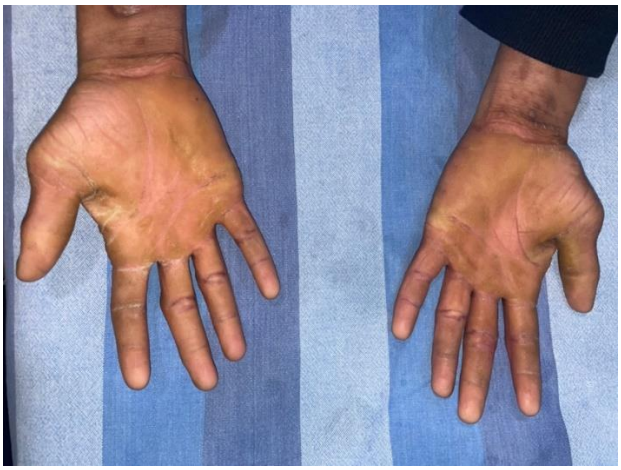

Figure 6: Diffuse hyperkeratosis over bilateral palms

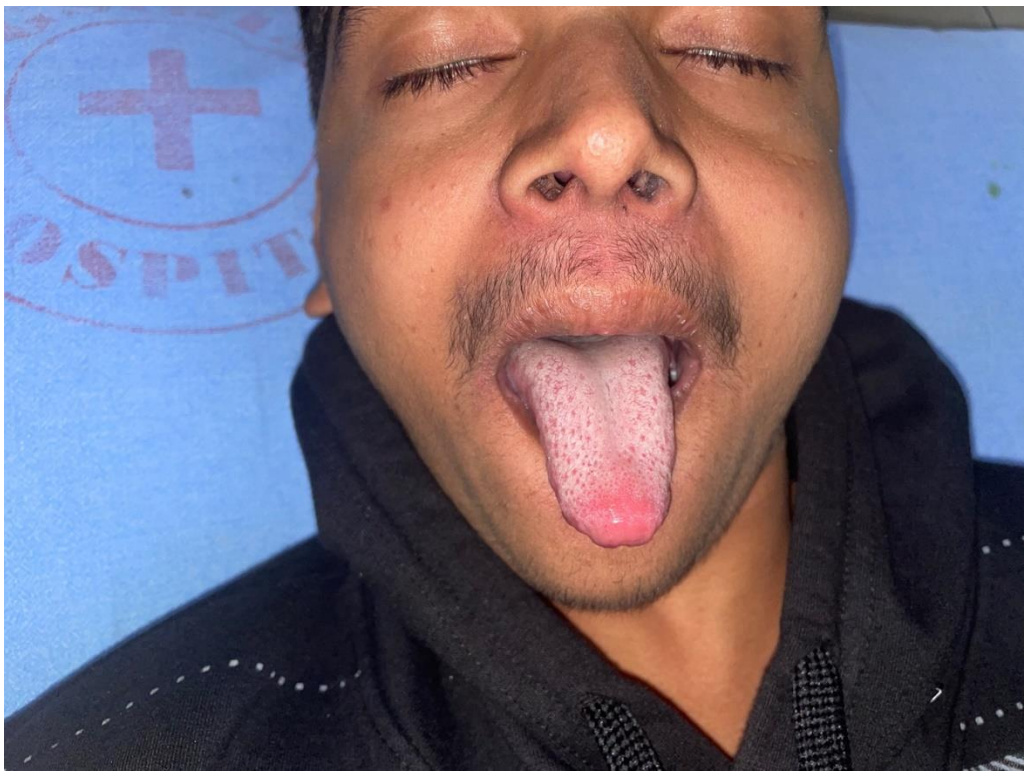

Figure 6: ORAL LEUKOKERATOSIS ON TONGUE

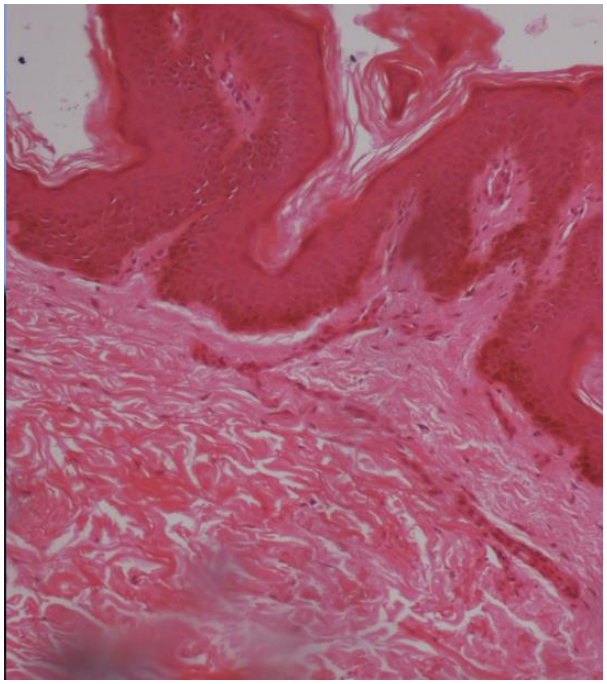

Figure 7: Epidermolytic Hyperkeratosis

Figure 8: Palmoplantar keratoderma