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PACHYONYCHIA CONGENITA WITH TUBERCULOUS PLEURAL EFFUSION: A RARE CASE REPORT FROM A TERTIARY CARE CENTER

General Medicine

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ABSTRACT

Background Pachyonychia congenita is a rare inherited keratin disorder characterized by hypertrophic nail dystrophy and palmoplantar keratoderma. Due to its rarity and similarity to other nail disorders, it is frequently misdiagnosed. The coexistence of pachyonychia congenita with systemic infections such as tuberculosis is extremely uncommon.**Case Presentation** We report a case of a 44-year-old male who presented with fever, shortness of breath, and generalized weakness for 15–20 days. Dermatological examination revealed hypertrophic dystrophy of multiple fingernails and toenails along with palmoplantar keratoderma. Chest radiography demonstrated left-sided pleural effusion. Pleural fluid analysis showed exudative effusion with elevated adenosine deaminase (ADA) levels (159 U/L) and lymphocytic predominance, consistent with tuberculous pleural effusion. Cytological examination showed no evidence of malignancy.**Conclusion**This case highlights the importance of careful dermatological examination and comprehensive clinical evaluation in identifying rare disorders such as pachyonychia congenita. Early recognition helps avoid misdiagnosis and facilitates appropriate management, especially when associated with systemic conditions like tuberculosis.

KEYWORDS

CASE STUDY

A 44-year-old male presented to the Department of General Medicine with complaints of fever with chills, progressive shortness of breath, and generalized weakness for approximately 15–20 days. On admission, the patient was hemodynamically stable but showed mild respiratory distress. The vital parameters recorded were pulse rate of 106 beats per minute, respiratory rate of 17 breaths per minute, blood pressure of 118/81 mmHg, body temperature of 99.2°F, and oxygen saturation of 90% on room air.

On dermatological examination, the patient exhibited marked hypertrophic dystrophy of the fingernails and toenails, characterized by thickened nail plates, yellowish discoloration, and prominent subungual hyperkeratosis. These changes were evident in multiple nails of both hands and feet. The clinical appearance of the fingernails is shown in Figure 1, demonstrating significant nail plate thickening and dystrophy. Similarly, the toenails showed severe hypertrophic changes with excessive keratin accumulation, as illustrated in Figure 2.

Examination of the plantar surfaces of the feet revealed diffuse hyperkeratotic plaques with thickened skin and roughened surfaces around the toes, consistent with palmoplantar keratoderma. These findings are presented in Figure 3, which demonstrates the characteristic hyperkeratotic lesions of the plantar region.

Baseline hematological investigations revealed a hemoglobin level of 11.9 g/dL, total leukocyte count of $10.4 \times 10^3/\mu L$, and platelet count of $489 \times 10^3/\mu L$. The erythrocyte sedimentation rate (ESR) was markedly elevated at 74 mm/hr, indicating an underlying inflammatory process. Liver function tests demonstrated mildly elevated transaminases with AST (82.2 U/L) and ALT (61.4 U/L).

Radiological evaluation using chest X-ray (posteroanterior view) revealed a homogeneous opacity in the left mid and lower lung zones with blunting of the left costophrenic angle, suggestive of left-sided pleural effusion.Subsequent pleural fluid aspiration and analysis showed a protein concentration of 5.3 g/dL and glucose level of 43.3 mg/dL, indicating an exudative pleural effusion. The adenosine deaminase (ADA) level was markedly elevated at 159 U/L, strongly suggestive of tuberculous pleural effusion. Cytological examination revealed a total cell count of approximately 1500 cells/mm^3 with lymphocytic predominance (80%) and mesothelial cells accounting for 20%, with no malignant cells detected.

Based on the clinical findings, dermatological examination supported by photographic documentation (Figures 1–3), radiological evidence of pleural effusion, and pleural fluid analysis demonstrating lymphocytic exudative effusion with elevated ADA, the patient was diagnosed with pachyonychia congenita with concomitant tuberculous pleural effusion.

TABLE – 1
GENERAL DETAILS

Table 1. Demographic Characteristics and Clinical Presentation

Parameter	Findings
Age	44 years
Sex	Male
Duration of symptoms	15–20 days
Pulse rate	106 beats/min
Respiratory rate	17 breaths/min
Blood pressure	118/81 mmHg
Body temperature	99.2 °F
Oxygen saturation	90% on room air

Table 2. Dermatological Examination Findings

Clinical Feature	Observation
Fingernails	Hypertrophic nail dystrophy with thickened nail plates and yellow discoloration
Toenails	Severe nail thickening with excessive keratin accumulation
Subungual Hyperkeratosis	Present in multiple fingernails and toenails
Plantar lesions	Diffuse hyperkeratotic plaques with thickened skin
Palmoplantar Keratoderma	Present

Table 3. Hematological and Biochemical Investigations

Investigation	Result	Reference Range	Interpretation
Hemoglobin	11.9 g/dL	13–17 g/dL	Mild anemia
Total leukocyte count	$10.4 \times 10^3/\mu L$	$4–11 \times 10^3/\mu L$	Within normal range
Platelet count	$489 \times 10^3/\mu L$	$150–410 \times 10^3/\mu L$	Thrombocytosis
ESR	74 mm/hr	0–15 mm/hr	Markedly elevated
AST (SGOT)	82.2 U/L	<50 U/L	Elevated
ALT (SGPT)	61.4 U/L	<50 U/L	Elevated

Table 4. Radiological and Pleural Fluid Analysis Findings

Investigation	Parameter	Result	Interpretation
Chest X-ray (PA view)	Lung field	Homogeneous opacity in left mid and lower zone	Suggestive of pleural effusion
Chest X-ray	Costophrenic angle	Blunting of left CP angle	Left-sided pleural effusion
Pleural fluid analysis	Protein	5.3 g/dL	Exudative effusion
Pleural fluid analysis	Glucose	43.3 mg/dL	Reduced
Pleural fluid analysis	ADA	159 U/L	Strongly suggestive of tuberculous effusion
Cytology	Total cell count	1500 cells/mm³	Increased
	Lymphocytes	80%	Lymphocytic predominance
	Mesothelial cells	20%	Present

FIGURES

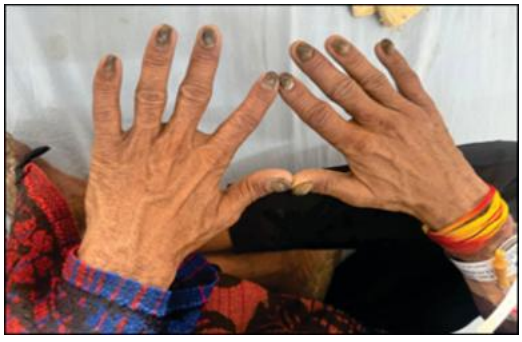

Figure 1: Hypertrophic dystrophy of fingernails showing marked nail thickening and subungual hyperkeratosis.

Figure 2: Severe dystrophic toenails with extensive keratin accumulation.

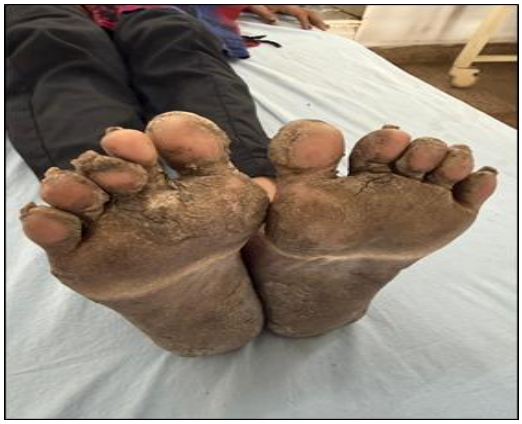

Figure 3: Plantar keratoderma with thickened hyperkeratotic plaques.

DISCUSSION

Pachyonychia congenita (PC) is a rare inherited disorder of keratinization characterized by hypertrophic nail dystrophy, palmoplantar keratoderma, and other ectodermal abnormalities[1] [2]. The condition results from mutations in keratin genes such as *KRT6A*, *KRT6B*, *KRT6C*, *KRT16*, and *KRT17*, which play a crucial role in maintaining the structural integrity of epithelial tissues[3] [4]. Disruption of these keratin proteins leads to abnormal keratinization and fragility of epithelial cells, resulting in the characteristic clinical manifestations of the disease.

The most prominent clinical feature of pachyonychia congenita is hypertrophic nail dystrophy, which typically affects both fingernails and toenails[1] [5]. The nails appear thickened, discolored, and distorted due to excessive subungual keratin deposition. In the present case, the patient demonstrated marked nail thickening with yellowish discoloration and irregular nail plates involving both hands and feet, as shown in Figures 1 and 2. These findings are consistent with the classical description of pachyonychia congenita reported in previous studies[2],[5].

In the present case, the patient also exhibited diffuse plantar hyperkeratosis with thickened keratotic plaques surrounding the toes, as illustrated in Figure 3. Similar findings have been described in several reports of pachyonychia congenita, where plantar keratoderma is often responsible for significant morbidity[2],[5].

Due to its rarity and overlapping clinical features, pachyonychia congenita is frequently misdiagnosed as more common conditions such as onychomycosis, psoriasis, or lichen planus[1] [3]. Patients are often treated empirically with antifungal medications before the correct diagnosis is established. In the present case, fungal infection was excluded by laboratory examination, and the characteristic clinical findings strongly supported the diagnosis of pachyonychia congenita.

Other hereditary palmoplantar keratoderma syndromes, such as Papillon–Lefèvre syndrome, may also present with similar hyperkeratotic lesions and therefore form an important differential diagnosis. Papillon–Lefèvre syndrome is an autosomal recessive disorder caused by mutations in the cathepsin C gene and is characterized by palmoplantar hyperkeratosis along with severe early-onset periodontitis leading to premature loss of teeth[11].

An additional significant finding in this patient was the presence of left-sided pleural effusion detected on chest radiography. Pleural fluid analysis demonstrated an exudative effusion with high protein levels, lymphocytic predominance, and markedly elevated adenosine deaminase (ADA) levels, which are typical features of tuberculous pleural effusion[6]. Tuberculosis remains a common infectious disease in developing countries, and pleural effusion is a well-recognized extrapulmonary manifestation of the disease[6].

Although pachyonychia congenita primarily affects the skin and nails, the coexistence of systemic conditions such as tuberculosis can complicate the clinical presentation. The presence of respiratory symptoms, including fever and breathlessness, in this patient prompted further investigation, which ultimately confirmed the diagnosis of tuberculous pleural effusion.

Recognition of the characteristic dermatological features of pachyonychia congenita, supported by photographic documentation, is essential for accurate diagnosis[1],[2],[5]. At the same time, clinicians must remain vigilant for associated systemic conditions, particularly in patients presenting with additional symptoms such as fever or respiratory distress.

Early diagnosis of pachyonychia congenita allows appropriate symptomatic management and prevents unnecessary treatments[1] [5]. Reporting rare cases such as this contributes to the existing literature and enhances awareness among clinicians regarding the varied clinical presentations of this uncommon disorder.

CONCLUSIONS

Pachyonychia congenita is a rare inherited keratinization disorder characterized by hypertrophic nail dystrophy and palmoplantar keratoderma. Because of its clinical resemblance to more common

conditions such as onychomycosis and psoriasis, the disease is frequently misdiagnosed. Careful clinical examination and exclusion of fungal infections are essential for establishing the correct diagnosis.

The present case demonstrates the classical dermatological features of pachyonychia congenita with associated tuberculous pleural effusion, highlighting the importance of comprehensive clinical evaluation when patients present with systemic symptoms. Early recognition of characteristic findings can prevent unnecessary treatments and allow appropriate management and patient counseling. Reporting such rare cases contributes to improved awareness and understanding of this uncommon disorder among clinicians.

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