Mohammad Ali El-Darouti

Challenging Cases in Dermatology

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Foreword I

The stimulating book of Professor Mohammad Ali El-Darouti is coming just in time to fill up an important gap in medical education.

Most of the progresses in the management of frequent diseases have been the result of a scientific approach looking on populations of patients and using statistical methods in order to decrease the background noise of individual situations. This was the birth of the Evidence-Based Medicine (EBM). EBM is of major usefulness, and the new generation of dermatologists believes that it is a gold standard. Without EBM-based guidelines, they are lost.

However, in Dermatology, we are confronted every day with rare diseases for which EBM is not and will never be available. In addition, even in frequent diseases, EBM represents the medical science, but we are not dealing with populations but with individual situations, and we have to learn how to manage them.

Thus, all along the medical life, it is of key importance to practice not only EBM but also a "patient-based medicine", allowing to train oneself the techniques making possible to manage, at diagnostic and therapeutic levels, the unique individual situations of the real life.

Written by a clinician of outstanding experience, well-known for his pedagogic expertise, this book is a gem. It offers the experience of 35 years of professional life devoted to help patients in difficult situations. Each case is not only a very stimulating clinical enigma but also an educational approach pushing the reader to understand the thought process put on the patient, the diagnostic and the treatment.

This book, in addition to being a real reading pleasure, is a very important milestone for medical education of students and of experts. It pushes us again not to forget that we are dealing with individual patients, and that we have to educate our mind not only to apply guidelines but to take care of human beings.

Louis Dubertret

Foreword II

One of the great joys and challenges of clinical dermatology is to make an accurate diagnosis that allows optimal treatment of a patient's skin condition. As physicians, our diagnostic skills tend to improve with years of experience in the clinic, but our professional activities can also be sharpened and refined by learning from others. In that regard, there is no substitute for a great teacher who has wonderful cases to share, and therefore this book, prepared by Professor Mohammad El-Darouti, represents a truly magnificent compilation of fascinating dermatoses, both rare and common. Professor El-Darouti has a well-deserved international reputation as a superb clinical dermatologist, an astute dermatopathologist, a clever diagnostician, and a respected teacher, and all those elements come together in this unique book. The format challenges the reader to diagnose the cases, to think about the appropriate investigations, and to become better dermatologists. The combination of cases and material is highly original and the layout makes the reader want to keep reading page after page, just like a bestselling novel – except here we are dealing with real patients and their very real skin diseases. Reading this book is a joy; it is a superb example of continuing medical education that all dermatologists can fully appreciate and, most importantly, benefit a great deal from, professionally and personally. Be surprised, be amazed, but above all, be prepared to think differently in diagnosing the cases in this book. We can all be inspired and learn a lot.

London, UK

John McGrath, MD, FRCP, FMedSci

Preface

This book is not simply a collection of cases; it provides the distinctive feature of a strategic way of thinking and data analysis while discussing each case to reach a diagnosis. Every case is a unique story and the sequence of telling these stories is similar. However, it must be noted that no single template applies to all cases. Some cases present extremely rare diseases that are hardly reported once or twice in the literature. Others describe rare presentations of diseases that are not, themselves, uncommon. As such, this book offers extensive discussions of complex cases, surprising diagnoses, therapeutic challenges, and other exciting features.

A familiarity with such unusual cases will undoubtedly provide the dermatologist with increased knowledge, a wider perspective, and innovative techniques that can be used to solve diagnostic dilemmas. Readers will be able to make better selections from a list of puzzling probabilities.

I intentionally avoided writing the diagnosis as a title, as I wanted the readers to go through the journey of every case, starting with the clinical data, histological slides, and other available investigations. Data are collected and compiled in a few lines to make the bases of the final diagnosis. A brief discussion of each case follows and includes epidemiology, pathogenesis, clinical and histological features, differential diagnosis, definite diagnosis, prognosis, and management. The text allows for comparison of each case to similar cases, and notes distinguishing features of each condition under consideration. A brief summary of the management of each case is provided and a follow-up is described except where tracking was, unfortunately, lost for a few patients. Each case concludes with a message about the valuable lessons learned from the case. This book will take you down an extraordinary adventure with every case.

A wealth of diagnostic approaches and pearls of wisdom are provided and gleaned from over 35 years of clinical experience. Almost all patients are Egyptians from diverse geographical areas within Egypt and different socioeconomic classes.

This book not only adds new and rare diseases to a physician's general list of differential diagnoses, but also explains the best protocol for assimilating a patient's history, clinical findings, and lab results to reach key diagnoses and identify rarer diseases. Obviously, the longer the list of differential diagnoses in a dermatologist's repertoire, the less likely the physician will miss a serious or uncommon disease.

I always used to hear about "dreams coming true," and I was blessed to witness and enjoy many of my dreams coming true; however, never did I revel in those successes the way I have with this book. I truly hope you delight in this astounding and abundant journey.

Cairo, Egypt — Mohammad Ali El-Darouti

Contents

Part I

Genodermatoses

Hyperkeratotic Palms and Soles with Periorificial Keratosis

1

Case

Clinical Data

A 7-year-old boy (Figs. 1.1, 1.2, 1.3, 1.4, and 1.5) presented with excessively keratotic skin over the palms and soles, restricting his mobility and interfering with walking. He also had persistent flexion deformity of the fingers. There was a life-long history of thick scaly lesions affecting the scalp, forehead, peri-auricular, peri-oral, and peri-anal areas. He had a thin, short hair that was growing very slowly. His mental and physical growths were normal. Examination revealed no dental anomalies and no deafness.

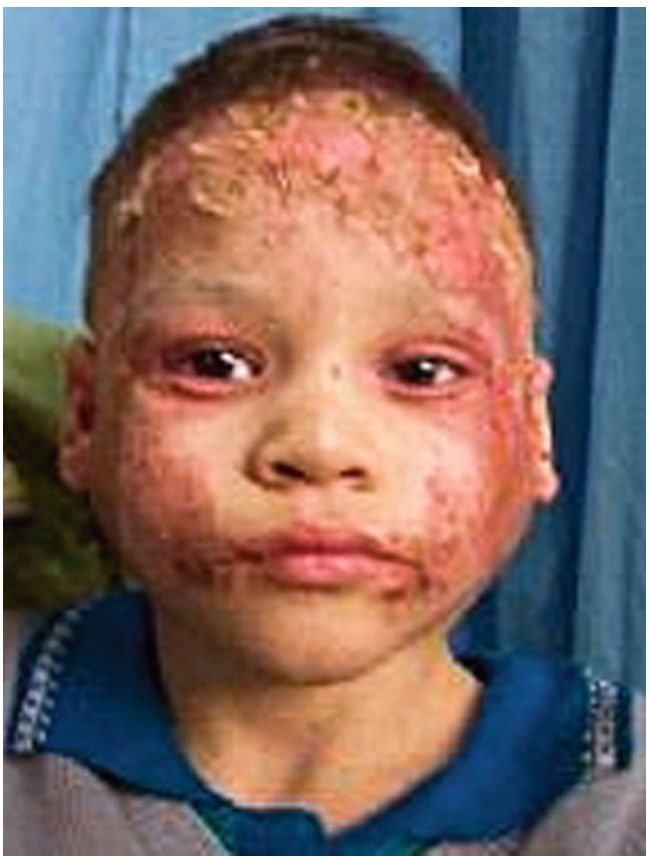

Fig. 1.1 Hyperkeratosis of the face involving perioral area, face and scalp

Differential Diagnosis

- **Vohwinkel syndrome:**
  - Diffuse palmoplantar keratoderma; honeycomb in pattern with linear hyperkeratosis on elbows and knees.
  - Pseudoainhum associated with star-fish-shaped hyperkeratotic plaques on dorsa of hands, feet, elbows and knees.
  - Deafness is a feature (unlike this case).
- **Tyrosinemia II (Richner-Hanhart syndrome):**
  - Painful callosities of the hands and feet, but no other skin lesions.
  - Severe mental retardation is a feature (unlike this case).
  - It may be associated with keratitis and corneal ulceration.
- **Pachyonychia congenita:**
  - Several types, all with distinctively thickened hyperkeratotic fingernails and toenails (all 20) (unlike this case).
  - Palmoplantar keratoderma is variable.
  - No ainhum, and no joint deformities.
- **Mal de Meleda:**
  - Malodorous transgradient diffuse palmoplantar keratoderma in glove and stocking distribution, with hyperhidrosis and nail changes (unlike this case).
  - Erythematous hyperkeratotic lesions on elbows, knees, groin, axillae and around the mouth (like this case).
  - Associated with pseudoainhum, brachydactyly, EEG abnormalities (unlike this case).
  - No joint deformity, and hair growth is normal (unlike this case).
- **Papillon Lefèvre syndrome:**
  - Diffuse palmoplantar keratosis. However, not disabling.

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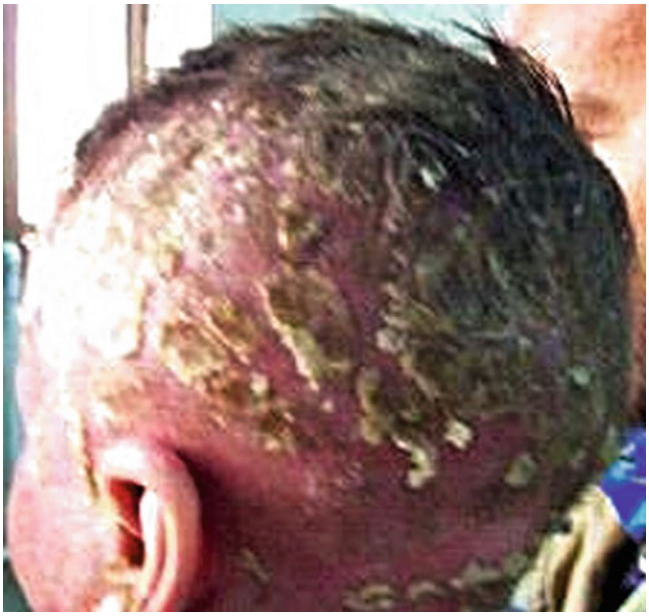


Fig. 1.2 Hyperkeratosis of the scalp with thin, sparse and light brown hair. Areas of alopecia are seen

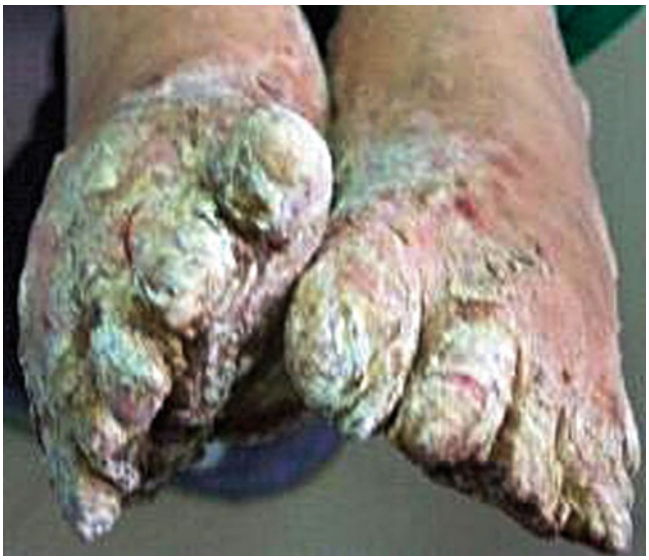


Fig. 1.3 Gross keratoderma of the soles

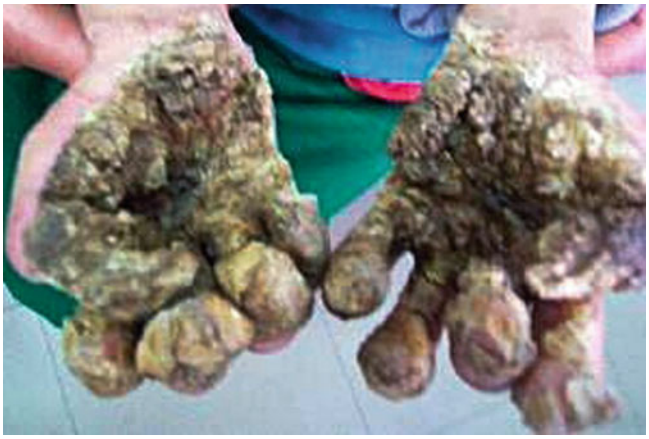


Fig. 1.4 Gross keratoderma of the palms

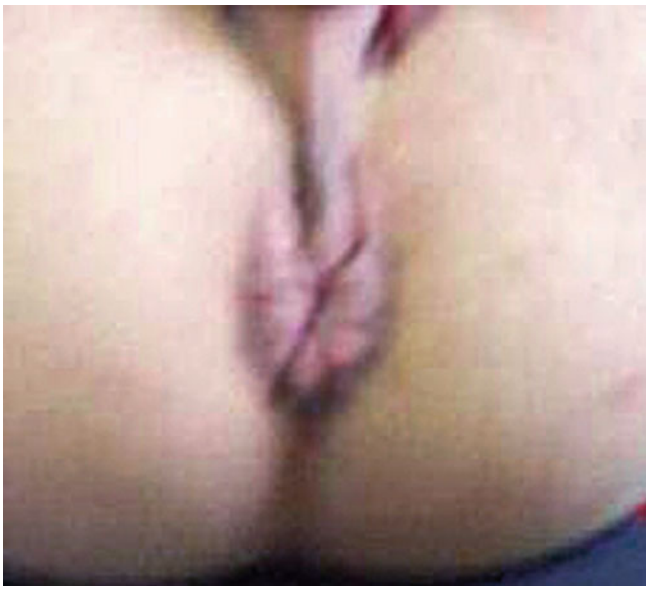
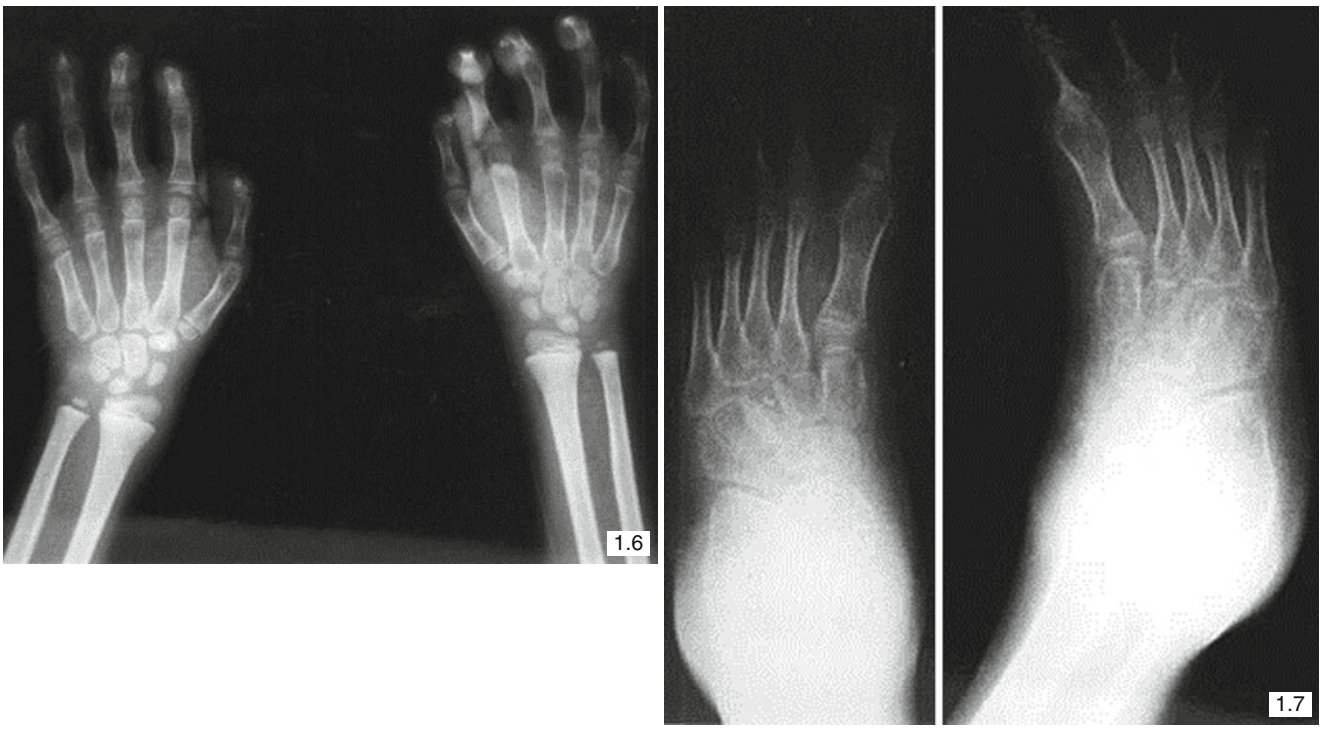


Fig. 1.5 Perianal hyperkeratotic plaques

  - Early, aggressive periodontitis (unlike this case).
  - Calcification of dura and choroid plexus.
  - Frequent pyogenic infections.
- **Olmsted syndrome:**
  - Diffuse mutilating palmoplantar keratosis with flexion deformity of the digits (like this case).
  - Progressive perioral, perianal, and perineal hyperkeratotic plaques (like this case).
  - Reported features of pseudoainhum, alopecia, nail and joint abnormalities.

Investigations

X-rays of the hands and feet showed joint deformity (Figs. 1.6 and 1.7).



Figs. 1.6 and 1.7 X-rays of the hands and feet showed joint deformity

Based on the Following Findings

- The distinctive combination of:
  1. Diffuse mutilating congenital palmoplantar keratoderma associated with Joint deformity.
  2. Perioral, perianal, and perineal keratotic lesions.
- The additional feature of hypotrichosis.
- The negative findings (absence of mental retardation, deafness, and periodontitis), which excluded other causes of palmoplantar keratoderma.

The Final Diagnosis was Olmsted Syndrome (Congenital Palmoplantar and Perioral Keratoderma).

About the Diagnosis

Definition

- Olmsted syndrome is a very rare genodermatosis, named after H.C. Olmsted, whose original description of the disorder in 1927 included a combination of bilateral, mutilating, palmoplantar keratoderma and periorificial hyperkeratotic plaques with flexion deformities of the digits, associated with occasional constriction or pseudoainhum leading to spontaneous amputation.
- Many features have been subsequently associated with this syndrome, and new features continue to be reported.

Epidemiology

- Extremely rare, with only 27 cases reported in the literature.
- Onset is in the first year of life.
- The majority of the published cases have been males (like this case); it has been rarely reported in females.

Pathogenesis and Etiology

- Pathogenesis is unknown, but there is cytochemical evidence of hyperproliferation of the epidermis.
- This syndrome seems to be of sporadic occurrence, although a familial case possibly due to autosomal dominant transmission and another case with X-linked dominant inheritance in two monozygotic male twins have been reported.
- Cambiaghi et al. reported transmission in two monozygotic male twins. They suggest that this condition is inherited as an X-linked dominant trait with reduced expression in female subjects.

- Kress et al. found a defect in the expression of mature epidermal keratins (types 1 and 10) and persistence of acidic keratins (types 5 and 14) in the involved epidermis.

Clinical Features

- **Main features:**
  - Severe progressive mutilating symmetric palmoplantar keratoderma.
  - Flexion deformities of the digits.
  - Digital constriction or spontaneous amputation.
  - Hyperkeratotic Periorificial, perianal, and perineal lesions.
- **Other reported features:**
  - Very slow hair growth.
  - Nail and tooth anomalies.
  - Corneal dystrophy.
  - Follicular papules on trunk and extremities.
  - Linear keratoses on the flexor forearms.
  - Growth retardation.
  - Psychomotor retardation.
  - Squamous cell carcinoma.

Histological Features

- Non-specific.

Investigations

- X-rays of the hands and feet demonstrate the joint deformity.

Differential Diagnosis

- Vohwinkel syndrome.
- Tyrosinemia II (Richner-Hanhart syndrome).
- Pachyonychia congenita.
- Mal de Meleda.
- Papillon Lefèvre syndrome.
- Clouston's hidrotic ectodermal dysplasia.

Definite Diagnosis

- The characteristic combination of:
  1. Diffuse mutilating congenital palmoplantar keratoderma associated with Joint deformity.
  2. Periorificial keratosis.
- Exclusion of other causes of congenital palmoplantar keratoderma (Negative findings of mental retardation, deafness, and periodontitis).

Prognosis

- The disease has a slow but progressive course.
- The keratoderma becomes extremely thick and may be disabling and interfering with normal walking.
- Fissuring around the toes that heals with a constricting band of tissue, leading to autoamputation of the digits, has been reported.
- The periorificial lesions may or may not improve with age.
- Patients with Olmsted syndrome may show a higher susceptibility to develop epidermal tumors such as squamous cell carcinoma and epithelioma cuniculatum.

Treatment

- There is no satisfactory treatment for this condition.
- Topical treatment:
  - Offers only symptomatic relief of pain and fissures by reducing the thickness of the keratotic palmoplantar skin lesions.
  - Topical tretinoin slightly improved the keratosis but proved irritant in one case.
  - Salicylic acid, urea, boric acid, corticosteroids, shale oil, emollients, antimicrobials, and wet dressings, have been tried with varying results.
- Systemic treatment:
  - Systemic retinoids and keratolytics showed unsatisfactory results.
  - Antihistamines, vitamins E, antimicrobials, corticosteroids have also been used anecdotally with no consistent benefits.
- For nonresponding patients, full-thickness excision of hyperkeratotic plaques followed by skin grafting is another therapeutic option to alleviate the pain. This treatment may improve flexion contracture of the fingers, but the risk of recurrence persists.

Management of This Case

Systemic retinoids and Urea 40 % cream under occlusion showed satisfactory results.

Message

- Urea 40 % cream under occlusion was found to be very effective in the treatment of the palmoplantar keratoderma due to Olmsted syndrome.
- To the best of the author's knowledge, this treatment has not been previously reported.

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Indurated, Dark, Hairy Plaques, with Arthritis and Deafness

2

Case 1

Clinical Data

A 48-year-old male presented with recurrent bouts of non-itchy wheals with fever, chills, and malaise, lasting almost 24 h. Onset started during infancy. Examination revealed multiple large plaques showing hyperpigmentation, hypertrichosis, and induration (Figs. 2.1, 2.2, and 2.3). He had history of recurrent cellulitis, joint pain, and progressive hearing loss.

Differential Diagnosis

- **Morphea:**
  - Shows indurated plaques, but usually without hypertrichosis.
  - Not associated with systemic symptoms of fever, chills and malaise.
  - No loss of hearing.
- **Kaposi Sarcoma:**
  - Shows brown erythematous hyperpigmented plaques, but without hypertrichosis.
  - No systemic symptoms and
  - No loss of hearing.
- **Winchester Syndrome:**
  - Skin changes of thickenings, hyperpigmentation, and hypertrichosis.
  - Shows dwarfism (unlike this case), joint contracture among other anomalies.
- **POEMS Syndrome:**
  - Skin changes of thickenings, hyperpigmentation, and hypertrichosis.
  - Other elements of the syndrome (Polyneuropathy, Organomegaly, Endocrinopathy, M-Paraproteinemia and Skin lesions) are usually seen.
- **Muckle Wells Syndrome:**
  - Bouts of urticaria, fever, and chill (like this case).
  - Progressive hearing loss (like this case).

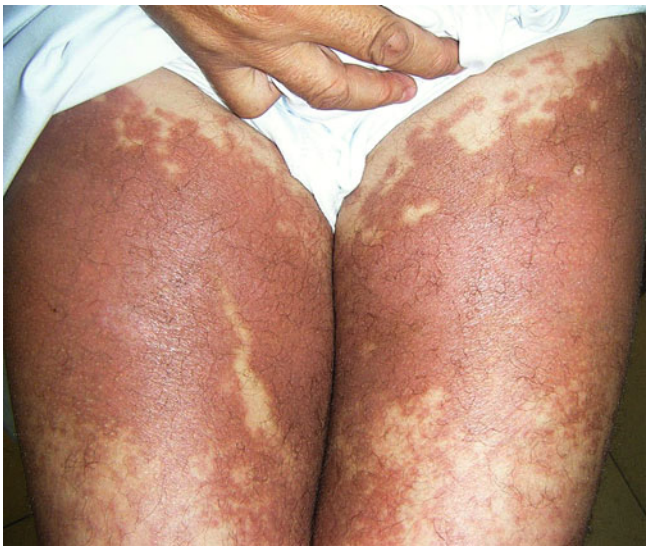

Fig. 2.1 Erythematous hyperpigmented plaques on both legs. The lesions show hypertrichosis

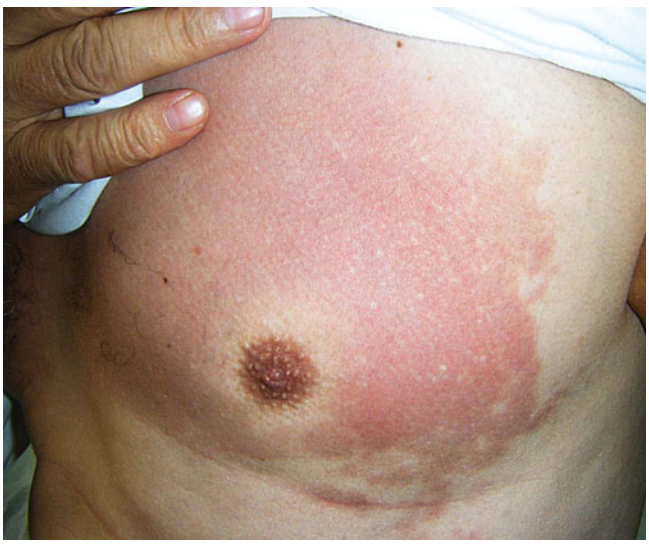

Fig. 2.2 Erythematous plaques over the left breast

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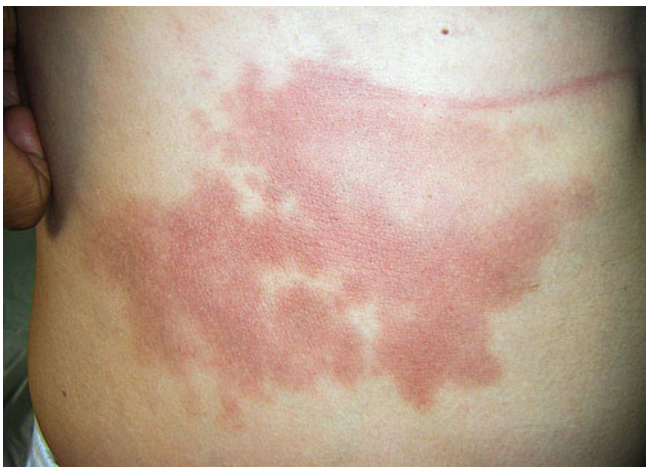

Fig. 2.3 Faint, erythematous urticarial eruptions on the trunk

Biopsy Findings (of Case 1)

Biopsy findings were of mild epidermal hyperplasia, remarkable dermal thickening replacing the subcutaneous fat, dilated blood vessels, perivascular and interstitial infiltrate of plasma cells, eosinophils and mast cells with large numbers of spindle-shaped cells (Figs. 2.4, 2.5, 2.6, and 2.7).

Investigations

- Lab investigations:
  - ESR: High.
  - CRP: High.
  - Serum amyloid protein (SAP): High.
  - Interleukin-6: High.
- Radiological investigations:
  - Abdominal Sonography: Hepatosplenomegaly.
- Other investigations:
  - Hearing test: Impaired hearing.

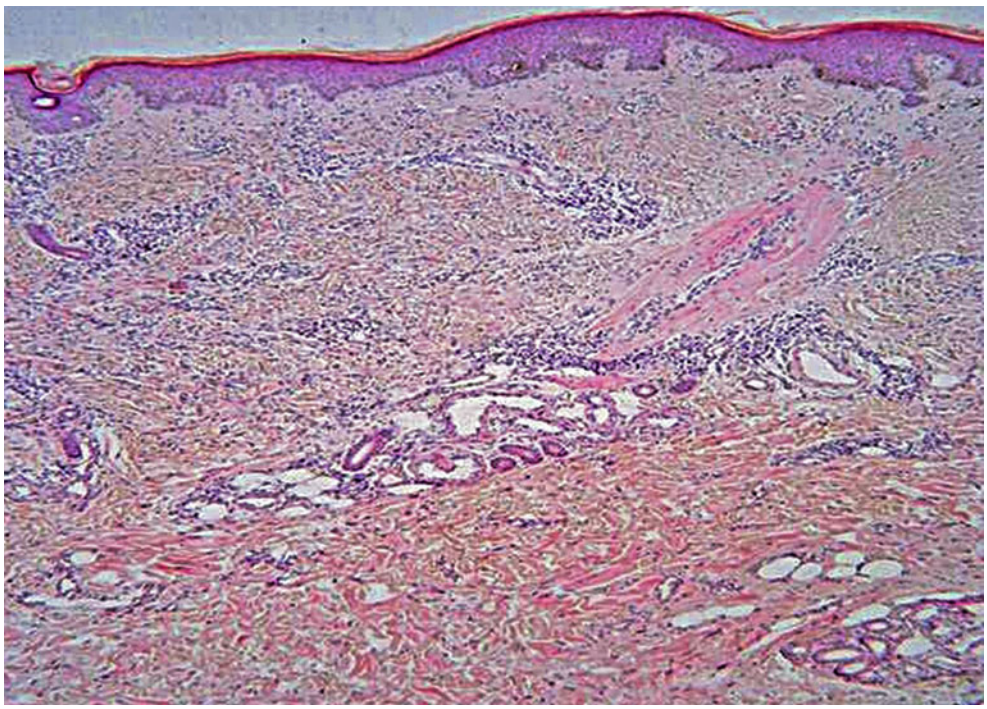

Fig. 2.4 Mild epidermal hyperplasia, remarkable dermal thickening replacing the subcutaneous fat

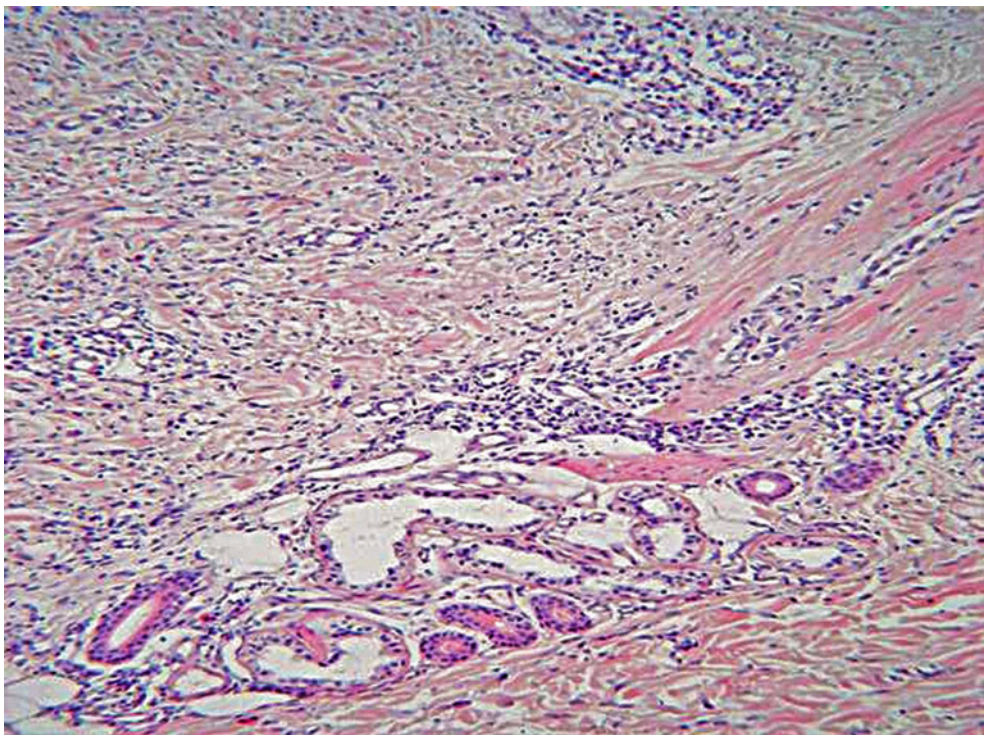

Fig. 2.5 Dilated blood vessels

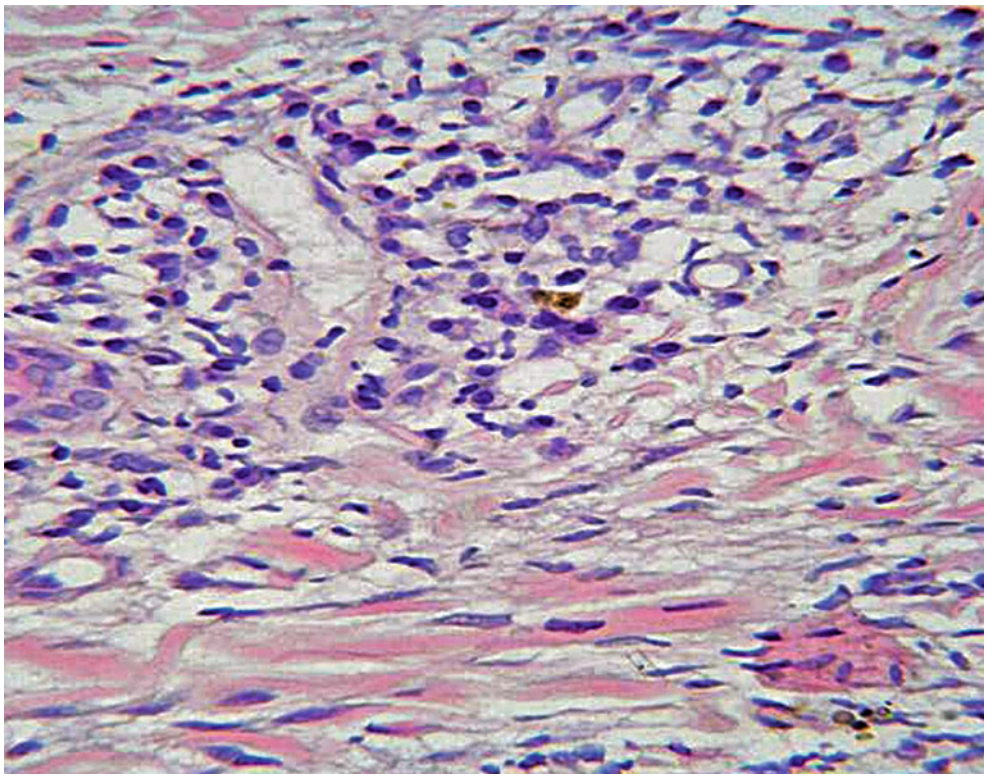

Fig. 2.6 Perivascular and interstitial infiltrate of plasma cells, eosinophils and mast cells. Large numbers of spindle-shaped cells are seen