Atlas of Pediatric Dermatoscopy

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Editors

Springer

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Preface

We are pleased to introduce this *Atlas of Pediatric Dermatoscopy* as the first book entirely devoted to the use of dermatoscopy in pediatric skin conditions.

Dermatoscopy in the past decade has revolutionized the approach to pigmented lesions by improving their diagnostic accuracy. In recent years, its use has been extended to a variety of inflammatory and infectious cutaneous diseases as well as skin appendage disorders (the terms *inflammoscopy*, *entodermoscopy*, *trichoscopy*, and *onychoscopy* have been introduced to define the use of dermatoscopy in inflammatory, infectious, hair, and nail disorders, respectively). In these cases, dermatoscopy, if performed by trained physicians, may enhance the clinical diagnosis and reduce the need of semi-invasive or invasive procedures such as skin scrapings and/or biopsy.

Being noninvasive, dermatoscopy is particularly suitable for its use in pediatric patients, in which standard invasive diagnostic procedures may be difficult to carry out.

The atlas, full of clinical and dermatoscopic images, covers the most frequent pediatric skin and appendage disorders in which dermatoscopy may be useful. For each condition, a brief epidemiological and clinical overview, along with a detailed dermatoscopic description and its histopathological correlation, is presented.

We wish that this book will encourage dermatologists, pediatricians, and GPs to consider the use of dermatoscopy in their clinical practice. For those who are already familiar with this technique, we hope that this atlas will expand their knowledge in the field of pediatric dermatology.

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Part I

Introduction

Instrumentation

1

Giuseppe Micali and Francesco Lacarrubba

The continuing evolution of imaging systems over the past years has improved the diagnostic accuracy of neoplastic and other skin disorders. Dermatoscopy (or dermoscopy) is a simple technique that allows a noninvasive, magnified, in vivo visualization of microstructures of the skin (epidermis, dermo-epidermal junction, and papillary dermis) not visible to the naked eye.

The most used device to perform a dermatoscopy examination is the handheld dermatoscope that, similarly to an otoscope, is user-friendly and relatively inexpensive (about 1000 Euros) (Fig. 1.1) [1]. Dermatoscopy requirements include a high-quality lens for ×10 magnification and a transilluminating lighting system, typically using an incident light source. For an optimal observation, it is necessary to apply a liquid (oil, alcohol, water) between the magnifying lens and the skin ("epiluminescence" technique) decreasing light reflection, refraction, and diffraction and making the epidermis translucent [2]; however, in some cases (as for some hair shaft disorders), the use of "dry dermatoscopy" is recommended. Modern devices use cross-polarized light that absorbs all the scattered waves avoiding the use of a liquid medium. In addition, these instruments offer the capability of viewing the skin without direct skin contact (noncontact dermatoscopy). Image storage may be obtained by connection to a digital camera using specially designed adaptors (Fig. 1.2) [3].

Alternatively, more expensive (about 15,000–25,000 Euros), computer-assisted digital systems are represented by the videodermatoscopes that may be equipped with lenses that ensure higher magnifications, up to ×1000 (Fig. 1.3). Images are obtained with a high-resolution color video camera using both non-polarized and polarized light and are visualized on a monitor. They can be transferred into a database allowing for accurate saving and storage and monitoring for geometric, chromatic, or structural evaluations. Videodermatoscopes can also be equipped with systems able to arrange automatic computerized analysis of pigmented lesions [4]. Several variables, such as circularity, maximum diameter, symmetry, and internal clusters of color, can be investigated to provide diagnostic suggestions. However, the role of these systems is limited, and further advances are necessary to improve the correct identification of equivocal lesion [5].

Recently, low-cost videomicroscopes (about 30–100 Euros) are available on the web for nonmedical use in entomology, botany, and microelectronics. These low-resolution, USB digital devices are able to provide magnification ranging from ×10 to ×1000 and to storage images in a personal computer. They have been demonstrated to be useful for the diagnosis of scabies, being able to identify its typical signs similarly to a standard videodermatoscope [6], whereas they seem to have a limited role in other conditions such as hair and scalp abnormalities [7]. Their use is certainly not recommended in the evaluation of pigmented lesion because of the low color quality and/or resolution provided.

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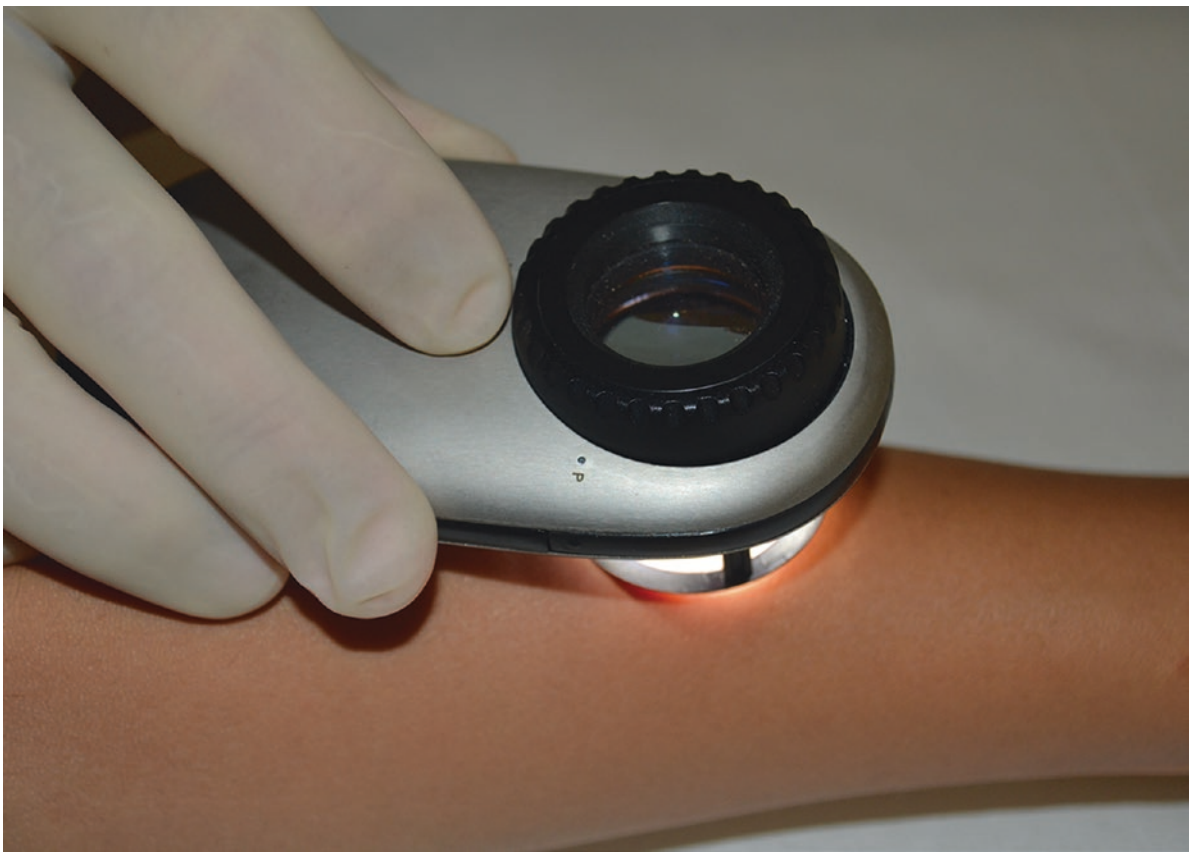

Fig. 1.1 Handheld dermatoscope

Fig. 1.2 Handheld dermatoscope connected to a digital camera

Fig. 1.3 Videodermatoscope

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Part II

Melanocytic Lesions

Spitz/Reed Nevus

2

Graziella Babino, Elvira Moscarella, Elisabetta Fulgione, Elena De Col, Vincenzo Piccolo, and Giuseppe Argenziano

2.1 Definition

Spitz nevus is a benign melanocytic neoplasm characterized by a proliferation of large spindle and/or epithelioid cells [1]. Reed nevus, described by Reed et al. [2] as "pigmented spindle cell nevus," is nowadays considered by many pathologists as indistinguishable from the pigmented variant of Spitz nevus. Here we refer to the two melanocytic lesions under the unique term of "Spitz/Reed nevus."

2.2 Etiology

Etiology of Spitz/Reed nevi has not been fully elucidated. However, there is evidence that cell proliferation is primarily mediated by mutations affecting genes in the RAS-RAF-MEK-ERK-MAP kinase pathway, namely, HRAS-activating mutations [3].

2.3 Epidemiology

Spitz/Reed nevus typically occurs in childhood and adolescence, although it may be present at birth or develop after 20 years of age. Caucasians seem to be more frequently affected. The most common location is the head/neck region, followed by lower extremities, trunk, and upper extremities [4, 5].

2.4 Clinical Features

Classical Spitz nevus is a solitary, asymptomatic, amelanotic, or hypopigmented papule or, less frequently, a nodule, a plaque, or a polypoid formation. The pigmented variant clinically presents as a solitary, dark brown to black papule or plaque [1, 2]. In addition, a number of clinical variants including halo, combined, dysplastic, and multiple disseminated and agminated Spitz nevi have been described [4].

2.5 Differential Diagnosis

Clinical differential diagnosis of Spitz/Reed nevi includes either benign or malignant lesions, such as Clark nevus, dermatofibroma, angioma, pyogenic granuloma, primary adnexal tumor, solitary mastocytoma, and pseudolymphoma. Spitzoid melanoma, a morphologic type of melanoma with Spitzoid features, is the most important differential diagnosis.

Moreover, a series of Spitzoid lesions, presenting varying features of clinical and histopatologic atypia and unknown malignant potential, has been described. These have been referred to with a variety of terms such as "Spitz nevus with atypia," "atypical Spitz nevus," "atypical Spitz tumors" (AST), and "melanocytic tumors of uncertain malignant potential" (MELTUMP) [6, 7].

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2.6 Diagnosis

The diagnosis is based on clinical-dermatoscopic correlation and histopathology.

2.7 Dermatoscopy

Six main dermatoscopic patterns can be ascribed to Spitz/Reed nevus [1, 2, 4]. The "vascular pattern," usually seen in hypo-/nonpigmented lesions, is characterized by the presence of dotted and monomorphic vessels, regularly distributed throughout the lesion and sometimes grouped and surrounded by regularly intersecting white lines, the so-called reticular depigmentation (Fig. 2.1). The "starburst pattern" shows peripheral globules fused with the central body of the lesion, giving regular "on focus" radial projections (so-called streaks) (Fig. 2.2). In the "globular pattern," brown to black, large globules are distributed throughout the lesion and at the periphery in multiple lines (Fig. 2.3). This pattern differs from the peripheral globular pattern seen in growing Clark nevi, in which globules are distributed in a single line at the periphery of the lesion. The "homogenous pattern" shows a diffuse dark brown to black-bluish color, which lacks evidence of clear-cut streaks at the periphery (Fig. 2.4). The "reticular pattern" is characterized by the presence of a regular brown to black network, sometimes showing a heavy pigmentation at the center of the lesion that can be removed by tape stripping ("superficial black network") (Fig. 2.5). Sometimes, atypical or multicomponent or "melanoma-like" pattern can be detected with blue-whitish veil (as a result of a deep dermal pigmentation with an overlying epidermal hyperplasia), irregular globules, and irregular streaks (Fig. 2.6).

2.8 Histopathological Correlation

Spitz/Reed nevi are plaque-shaped, well circumscribed, and typically composed of sharply demarcated melanocytic nests within a hyperplastic epidermis. Spindle or epithelioid melanocytes are highly cohesive and adjacent to keratinocytes. Melanin pigment is present mainly within spindle cells, dermal melanophages, and single intraepidermal dendritic melanocytes [8]; pigmented parakeratosis can be present, thus accounting for the presence of a superficial black network [9]. Overall, differently from melanoma, ulceration, dermal sheets of cells, and significant mitotic activity are lacking.

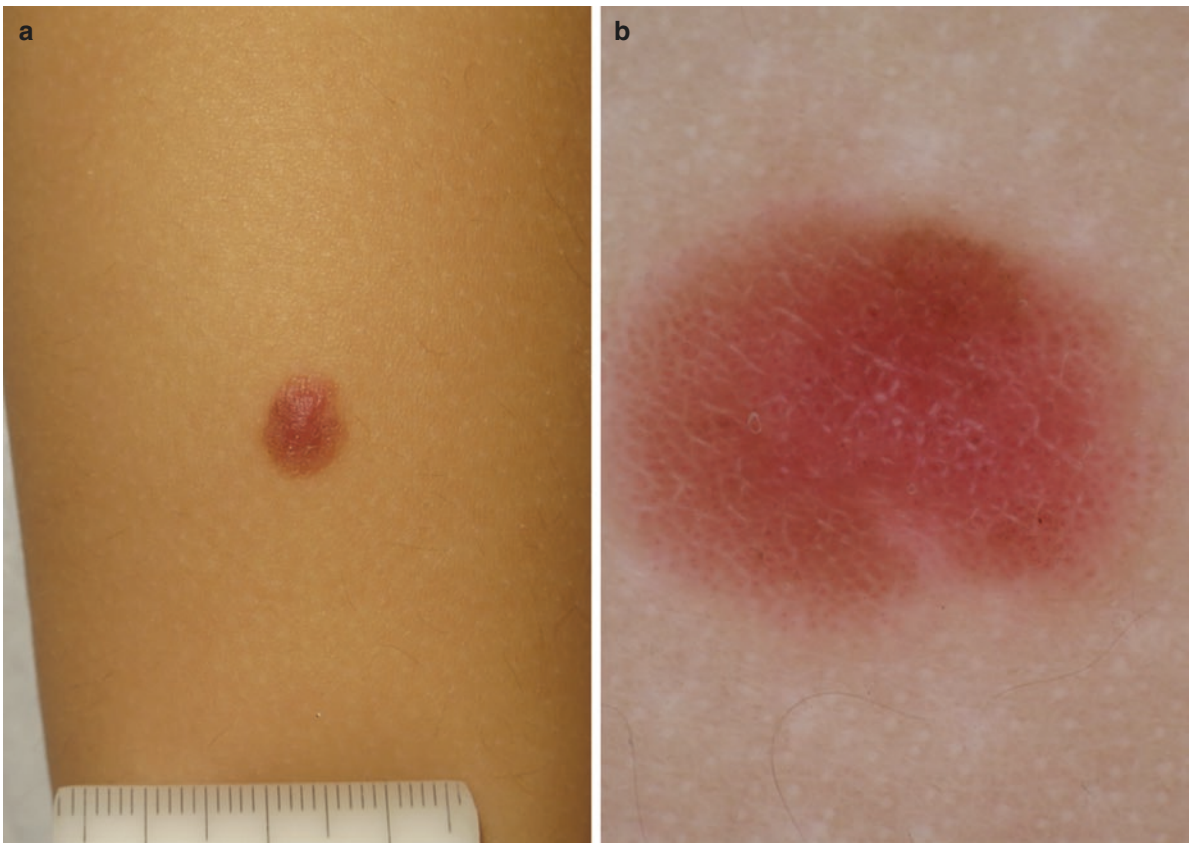

Fig. 2.1 Spitz/Reed nevus—(**a**) Hypopigmented lesion of the leg in a 4-year-old girl. (**b**) Dermatoscopy (×10): dotted vessels (*vascular pattern*)

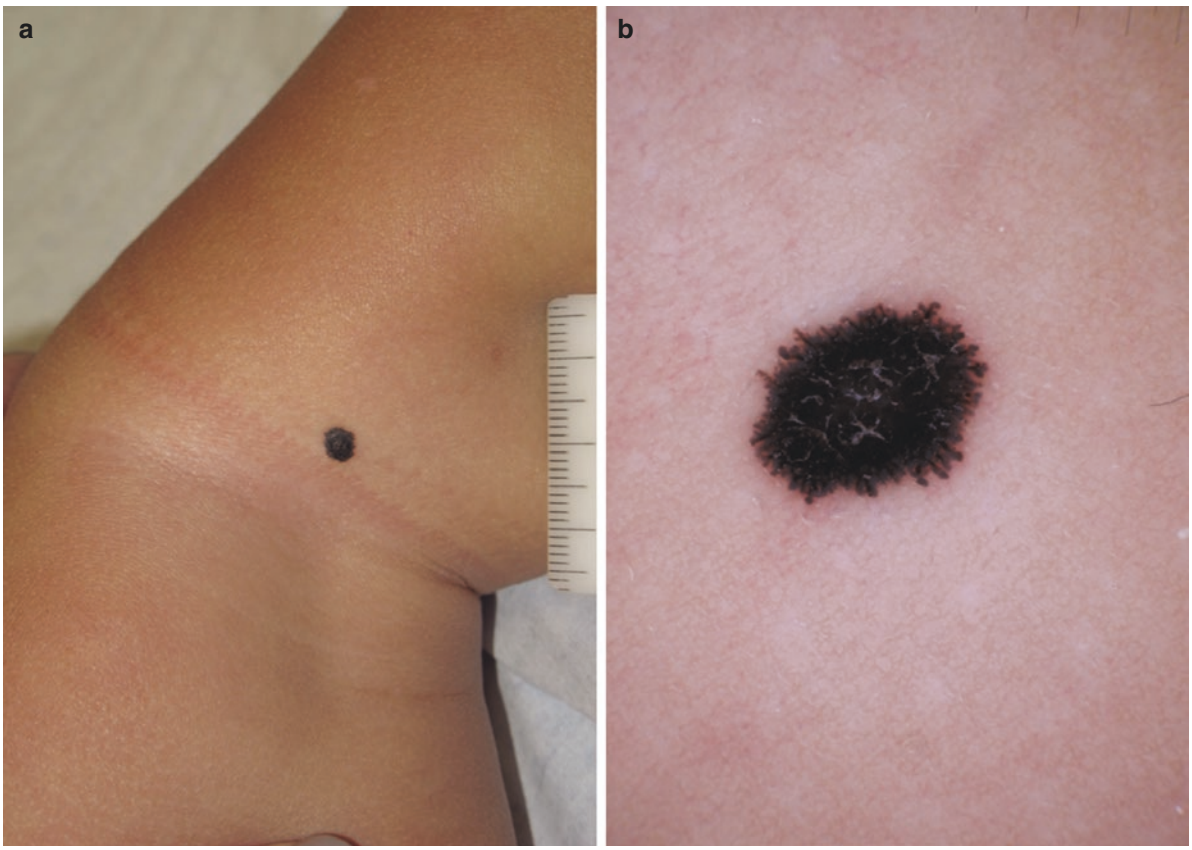

Fig. 2.2 Spitz/Reed nevus—(**a**) Heavily pigmented lesion of the leg in a 5-year-old boy. (**b**) Dermatoscopy (×10): peripheral globules (*starburst pattern*)

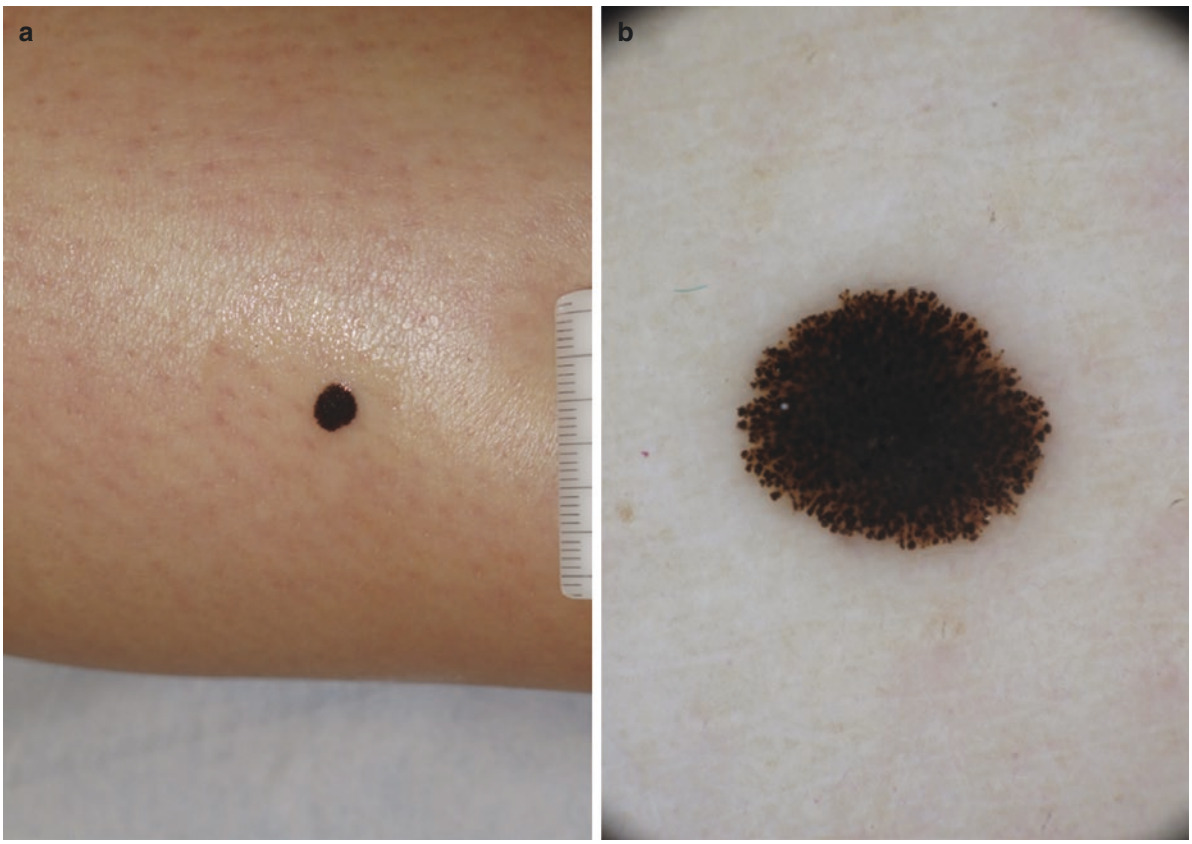

Fig. 2.3 Spitz/Reed nevus—(**a**) Dark brown macule of the leg in a 13-year-old girl. (**b**) Dermatoscopy (×10): diffuse brown background with brown to black, large globules distributed throughout the lesion and at the periphery (*globular pattern*)

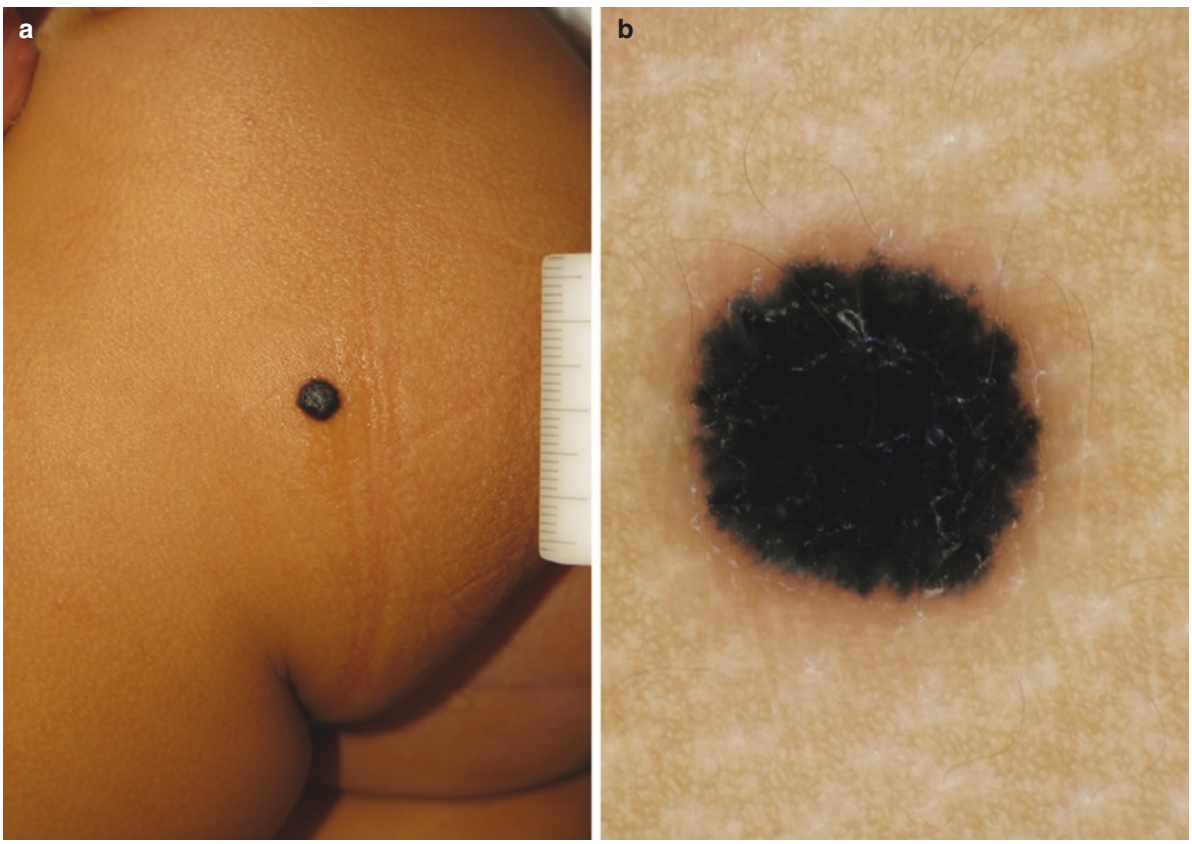

Fig. 2.4 Spitz/Reed nevus—(**a**) Heavily pigmented plaque of the buttock in a 1-year-old boy. (**b**) Dermatoscopy (×10): diffuse dark brown to black-bluish color (*homogenous pattern*)

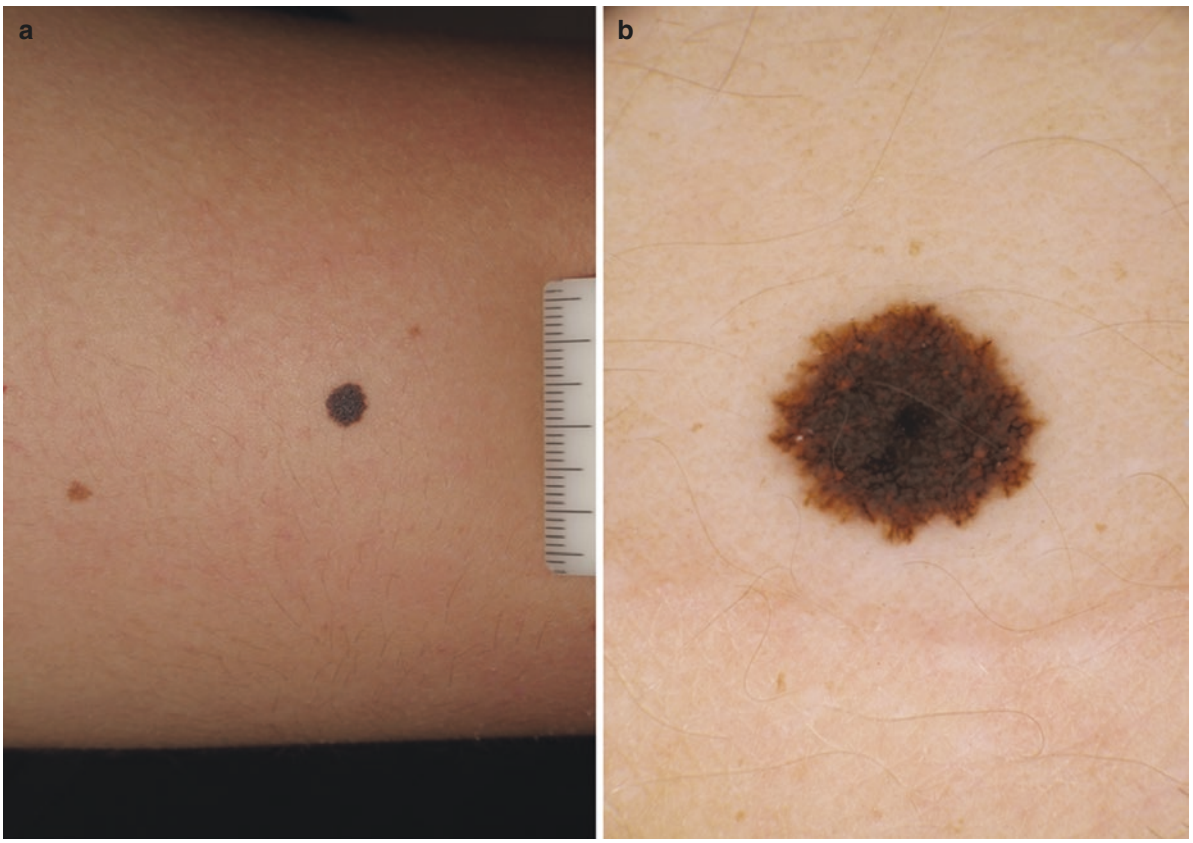

Fig. 2.5 Spitz/Reed nevus—(**a**) Pigmented lesion of the upper arm in a 12-year-old boy. (**b**) Dermatoscopy (×10): regular black network (*reticular pattern*)

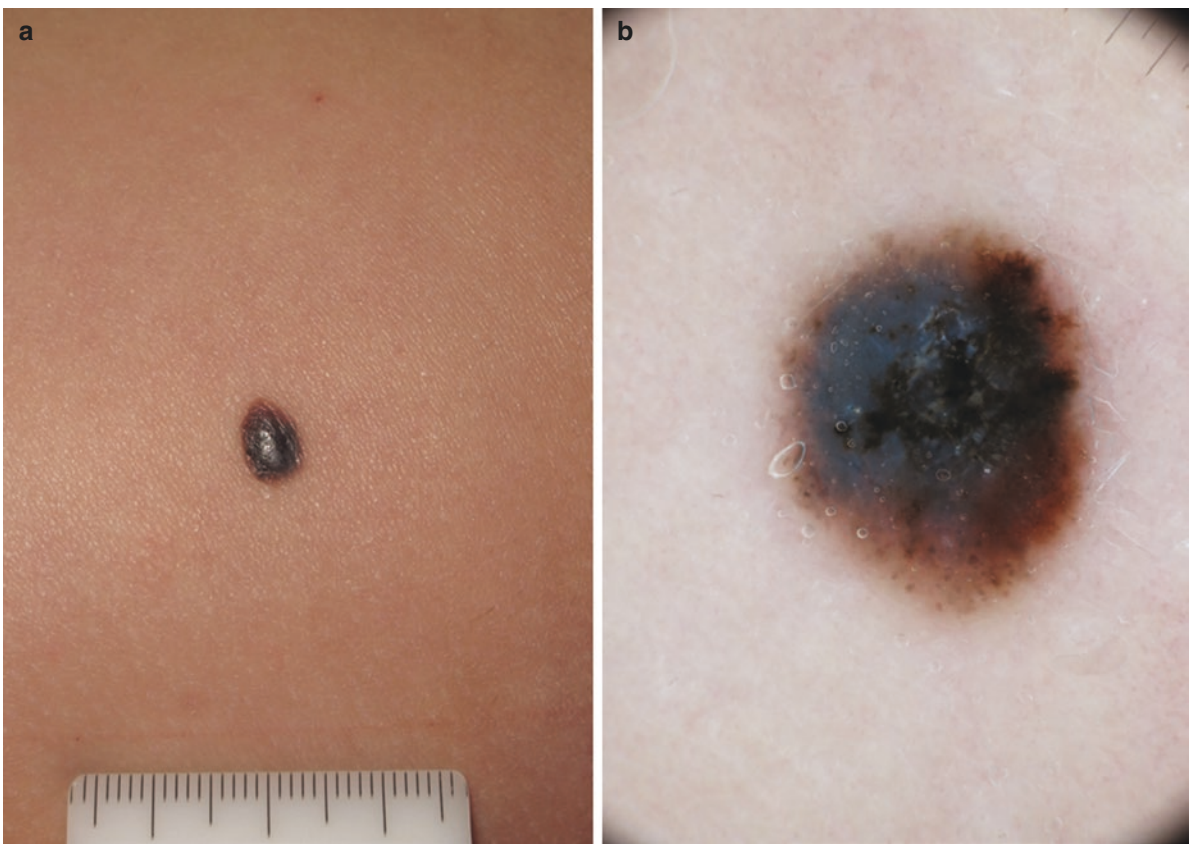

Fig. 2.6 Spitz/Reed nevus—(**a**) Pigmented nodular lesion of the upper arm in a 11-year-old girl. (**b**) Dermatoscopy (×10): multicomponent or "melanoma-like" pattern (*atypical pattern*)

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Congenital Nevi

3

Elena De Col, Elvira Moscarella, Graziella Babino, Elisabetta Fulgione, Vincenzo Piccolo, and Giuseppe Argenziano

3.1 Definition

Congenital melanocytic nevi (CMN) are defined as nevi present at birth or appeared within the first year of life and persistent throughout life.

They are classically subdivided into three groups based on their size: small (<1.5 cm), medium (1.5–20 cm), and large or giant (>20 cm); this group can be further classified as G1 (21–30 cm), G2 (31–40 cm), and G3 (>40 cm) [1]. Recently, a categorization scheme for standardized description of CMN has been proposed, including projected adult size, anatomic localization, morphologic features, and number of associated satellite nevi [2].

3.2 Epidemiology

The prevalence of CMN ranges between 0.2 and 6% in the worldwide literature [3]; the incidence of giant CMN is estimated in <1:20.000 newborns [4].

3.3 Etiology

The etiology is largely unknown; however recent studies highlighted that CMN are the result of a mutation in utero, after the embryo has already begun to develop. Early mutations can hit a multipotent progenitor cell, leading to multiple CMN and, sometimes, to involvement of other organ systems [5].

3.4 Clinical Features

CMN are usually larger than common nevi, with color and shape variegation and tendency to become palpable and covered by terminal hair. The giant variant involves a large area of the skin surface and may be associated with "satellite nevi" that are smaller CMN present at birth or appearing later [6].

The large and giant CMN, especially those with satellitosis, may be associated with systemic involvement, such as neurocutaneous melanocytosis [2, 5, 6]. This is a melanocytic proliferation involving the central nervous system that can negatively affect the prognosis as a result of increased intracranial pressure. Where this complication occurs, death is reported in more than half of patients within 3 years from the onset of symptoms [5, 7].

The current evidence-based literature data suggest a risk of CMN evolution toward malignant melanoma ranging between 0.05 and 10.7% [5, 8]. This risk is correlated with the size of CMN and the patient age, since melanomas tend to develop earlier and more frequently in giant CMN. In particular, the risk of melanoma development in small and medium CMN is 1%, compared to up to about 10% in large and giant CMN [8, 9]. Moreover, in children younger than 10 years, the estimated incidence is about 0.7 per million and increases up to 13.2 per million in those aged between 15 and 19 years [10].

Several benign proliferations have been described within CMN. Among these, the so-called proliferative nodules can be

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difficult to differentiate from melanoma from a clinical and histopathological point of view. They mostly arise in large or multiple nevi [5]. They can appear at any time in childhood, when they generally grow over a period of weeks and then stabilize [5].

3.5 Differential Diagnosis

The clinical and dermatoscopic features make the diagnosis of CMN rather univocal, especially in medium and large variants. Sometimes these must be distinguished from Becker nevus (acquired homogeneous brown lesion with numerous terminal hairs and irregular edges often localized on a hemithorax), cafè-au-lait macules (brown and homogeneous flat lesion, congenital or acquired without terminal hair), epidermal nevus (congenital or acquired nasty plaque devoid of terminal hair), and nevus spilus (considered a variant of CMN characterized by a variable number of brown macules or papules superimposed on a patch of brown pigmentation) [11]. Moreover, the possible different colors and shapes of CMN may induce to consider the differential diagnosis with malignant melanoma.

3.6 Diagnosis

The clinical diagnosis is generally not difficult because of the presence of a pigmented lesion with possible different size and color from birth or shortly thereafter.

3.7 Dermatoscopy

The typical pattern of CMN is globular and defined as "cobblestone pattern," characterized by the presence of large and angulated globules resembling cobblestones [12]. Globular pattern is most frequently found in the CMN of the head, neck, and trunk, while in the extremities it is sometimes possible to detect a reticulated or mixed pattern (reticular-globular) [13]. Additional dermatoscopic criteria include milia-like cysts, perifollicular hypopigmentation, hypertrichosis, comma vessels, and, occasionally, blue-white structures that make difficult the differential diagnosis with malignant melanoma (Figs. 3.1, 3.2, and 3.3) [12].

3.8 Histopathological Correlation

Histopathologically, CMN may show a junctional, compound, or intradermal pattern, depending on the age at which they are removed. In particular, they are often junctional at birth and early infancy [14], with a prominent melanocytic hyperplasia located in the epidermis and adnexal epithelium [15], whereas CMN removed after the neonatal period usually show nevus cells in the lower two-thirds of the dermis extending between collagen and around nerves, vessels, and adnexa [16]. At routine hematoxylin and eosin-stained sections, a full-thickness dermal involvement is typical of the larger congenital nevi [17], while a patchy distribution of nevus cells in the dermis may be observed in the smaller ones [18]. In any case, the involvement of eccrine glands and septa is considered the most specific feature of a true CMN [19].

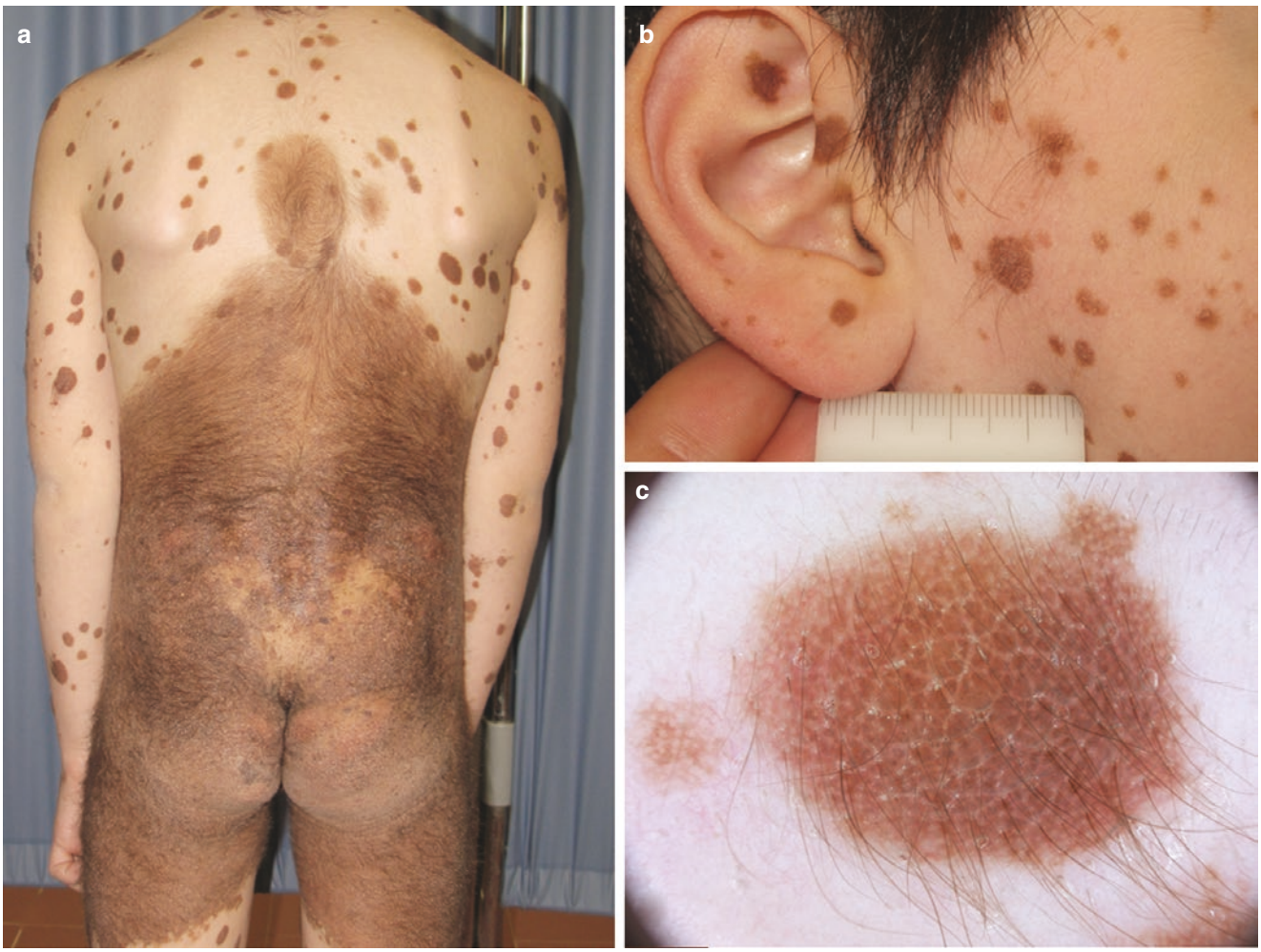

Fig. 3.1 (**a**) Giant congenital nevus in a 5-year-old boy. The lesion was present since birth, involving the trunk and several body areas with multiple satellites; neurologic development of the child was normal. (**b**) Close-up of the face showing multiple small- to medium-sized congenital nevi. (**c**) Dermatoscopy (×10): *cobblestone pattern*

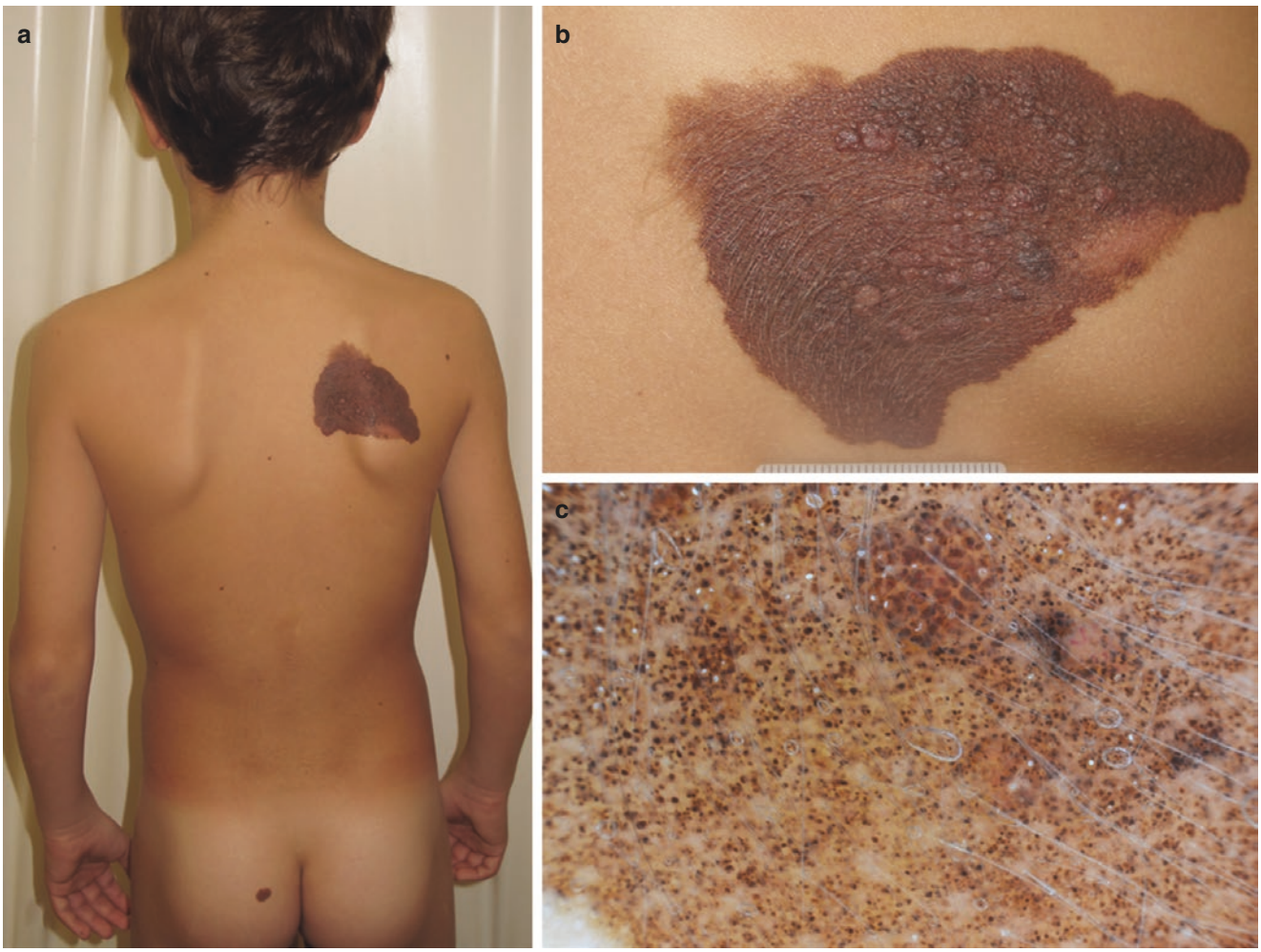

Fig. 3.2 (**a**) Medium size congenital nevus in a 6-year-old boy. The lesion was present since birth on the right scapular area. (**b**) Close-up image highlighting the presence of multiple small nodules, lighter than the underling nevus, soft on palpation, categorized as benign proliferations. (**c**) Dermatoscopy (×10): *globular pattern* all over the lesion

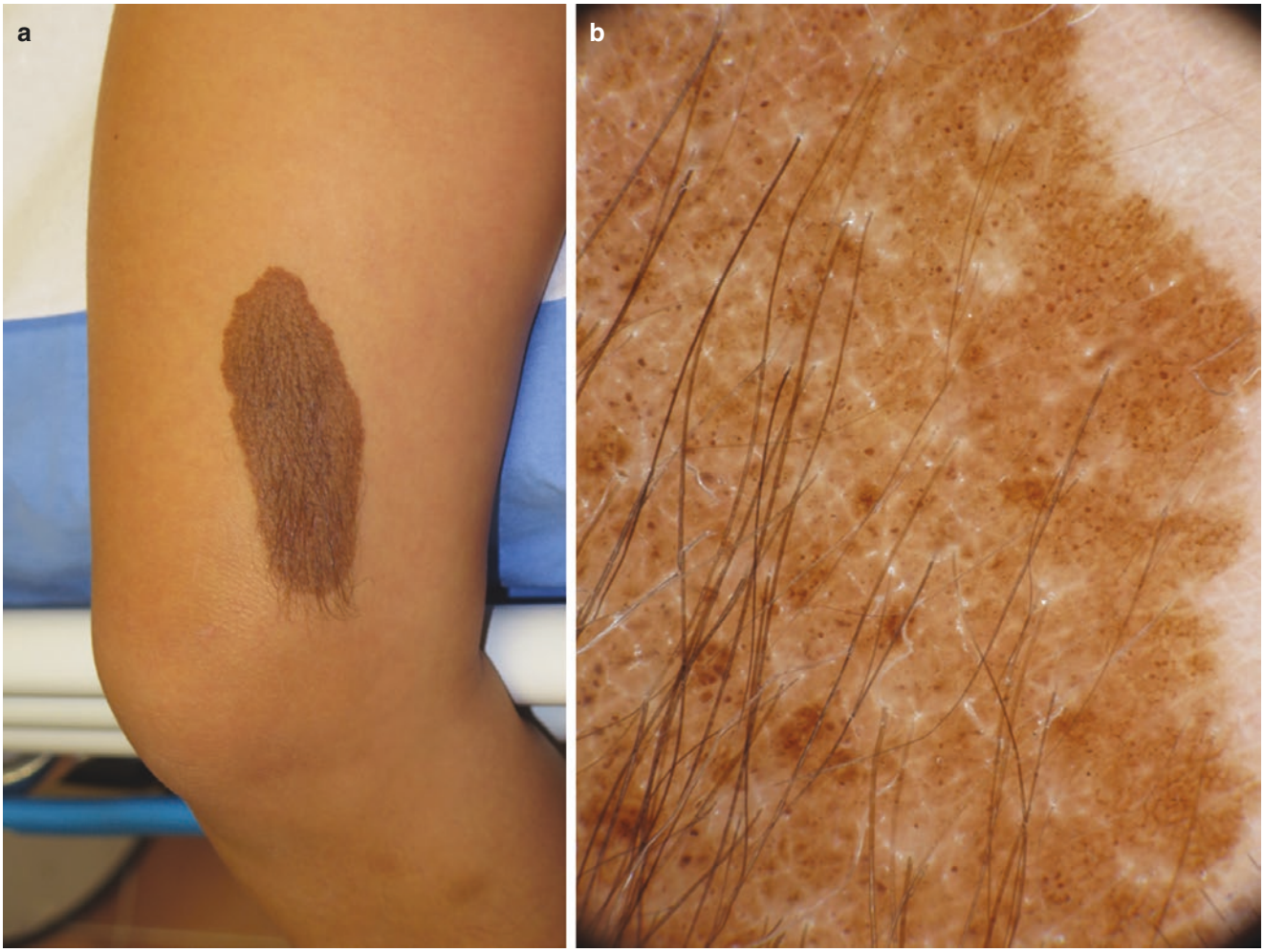

Fig. 3.3 (**a**) Medium-sized congenital nevus located on the leg in a 10-year-old boy. (**b**) Dermatoscopy (×10): mixed *globular-homogeneous* and *reticular pattern*

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Childhood Melanoma

4

Elisabetta Fulgione, Elvira Moscarella, Graziella Babino, Elena De Col, Vincenzo Piccolo, and Giuseppe Argenziano

4.1 Definition

Pediatric melanoma is defined as melanoma diagnosed between birth and 18 years of age.

4.2 Epidemiology

Pediatric melanoma is extremely rare, accounting for approximately 2% of childhood malignancies [1–3]. In the United States, 1–4% of all melanomas arise in patients younger than 20 years; however, only 0.3% occurs before puberty [3].

4.3 Etiology

Causes and risk factors for pediatric melanoma are less well understood but may be similar to that of adult melanoma. Children with fair skin, light hair, and freckles are at higher risk [4]. According to a recent review, approximately 50% of childhood melanoma occurs in association with a preexisting nevus, about 20% of them in association with acquired melanocytic nevi or small- to intermediate-sized congenital melanocytic nevi (CMN), and the remaining in large to giant CMN [5]. Other studies report on a much lower frequency of nevus-associated melanomas [6, 7]. Kanzler et al. [8] reported that the risk of malignant transformation of small and intermediate CMN virtually does not exist before puberty, and Ferrari et al. [1] recently reported only 9 cases of nevus-associated melanoma in their series of 33 childhood melanomas. Similarly, Bonifazi et al. [9] reported on approximately one third of nevus-associated melanomas (mostly in giant CMN) in their review of 289 cases of childhood melanoma.

4.4 Clinical Features

When arising "de novo," there is evidence that childhood melanoma often lacks the typical clinical features of pigmented melanoma and that a considerable proportion of melanoma in children may be hypomelanotic to amelanotic and raised, mimicking clinically pyogenic granuloma, non-pigmented Spitz nevus, and atypical Spitz tumors [10]. In these cases, the clinical EFG (E, elevation; F, firm on palpation; G, growing progressively for more than a month) rule might be helpful in summarizing the clinical symptoms of such melanomas.

4.5 Differential Diagnosis

Many melanocytic neoplasms in childhood pose diagnostic challenges to pathologists and sometimes cannot be unequivocally classified as benign nevi or melanoma, such as atypical Spitz tumors (AST).

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4.6 Diagnosis

Histopathology is mandatory for the diagnosis of childhood melanoma. Expert consensus is necessary in doubtful cases.

4.7 Dermatoscopy

In pigmented melanoma, a multicomponent pattern is most frequently retrieved (Fig. 4.1). In amelanotic melanoma, a polymorphous vascular pattern composed by a combination of dotted vessels and linear-irregular vessels, often associated with remnants of pigmentation, is usually detected. Less frequent features, but highly specific for nodular amelanotic melanoma, are milky red globules characterized by a reddish-whitish color, irregular size, and blurred borders. In amelanotic and hypomelanotic lesions, the differential diagnosis with nonpigmented Spitz nevus may be challenging. Both amelanotic melanoma and Spitz nevus may reveal dotted vessels as the only dermatoscopic feature in addition to the clinical history of a rapid growth (Fig. 4.2). Moreover, differential diagnosis also includes benign non-melanocytic lesions, such as pyogenic granuloma (PG). PG typically reveals a reddish homogeneous area surrounded by a white collarette, eventually intermingled by whitish lines [11]. However, excision is always warranted in doubtful cases.

4.8 Histopathological Correlation

The multicomponent pattern seen at dermatoscopy, which is highly suspicious of malignancy for the concomitant presence of multiple features within a lesion, has been shown to truly correlate with morphological alterations of the skin histopathologically detectable. Therefore, the atypical pigment network is related to the atypical melanocytic hyperplasia along the dermo-epidermal junction associated with prolonged epidermal crests; irregular globules are suggestive of atypical melanocytic nests in the lower epidermis or in the papillary dermis; black or gray dots are induced by intensely pigmented macrophages in the dermis; gray-blue areas correlate with the presence of lymphocytes and/or melanophages at the level of the dermis. Moreover, the polymorphous vascular pattern is associated with intense vascularization, with vessels of different forms and sizes.

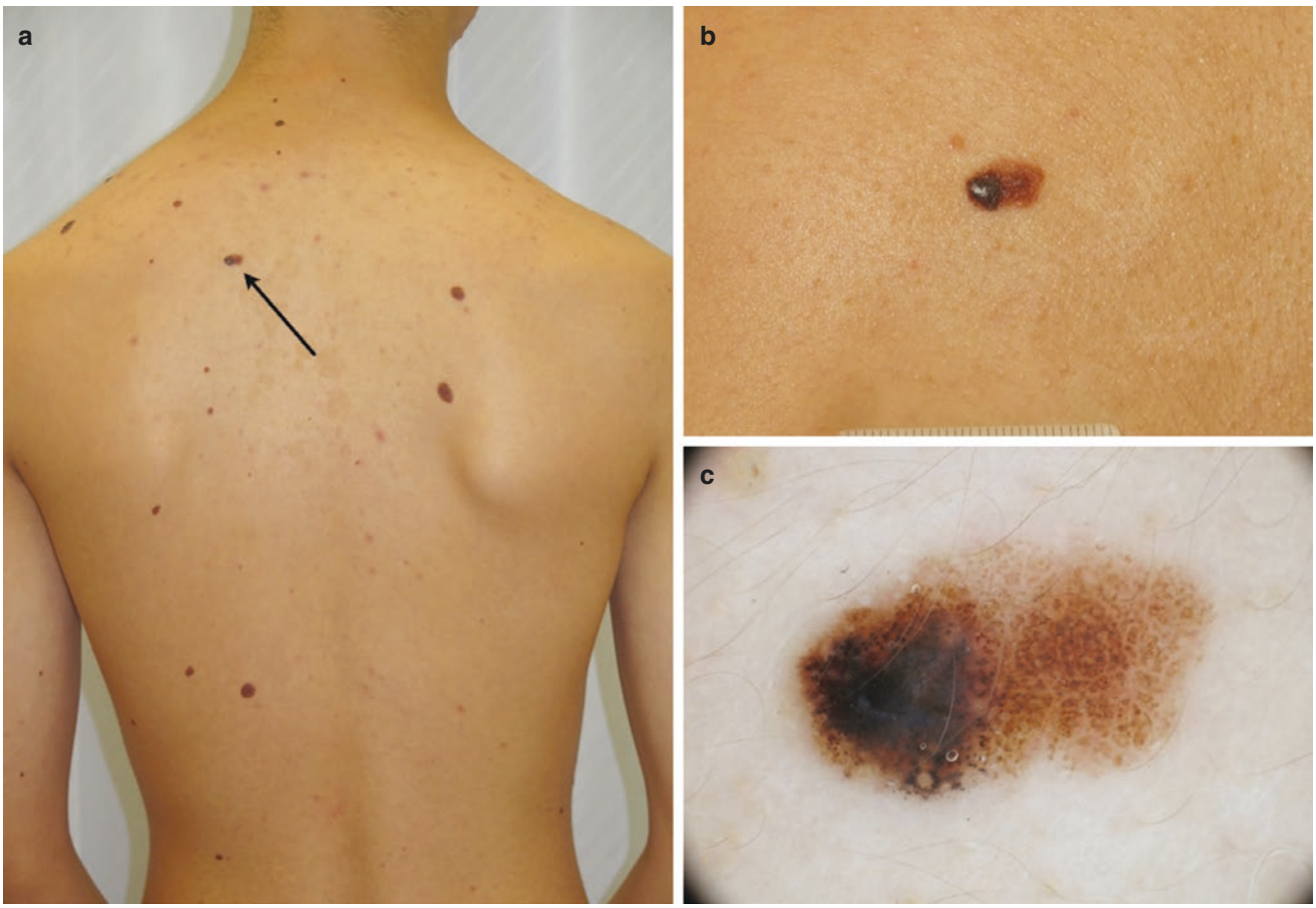

Fig. 4.1 Nevus-associated invasive melanoma (0.9 mm Breslow thickness) in a 16-year-old boy. (**a**) Close-up of the back showing multiple melanocytic nevi: melanoma is marked by a black arrow. (**b**) Close-up of melanoma, appearing as an asymmetric lesion with an eccentric, bluish, palpable area. (**c**) Dermatoscopy (×10): regular *cobblestone pattern* with an asymmetric black blotch and blue-white veil corresponding to the clinically palpable area

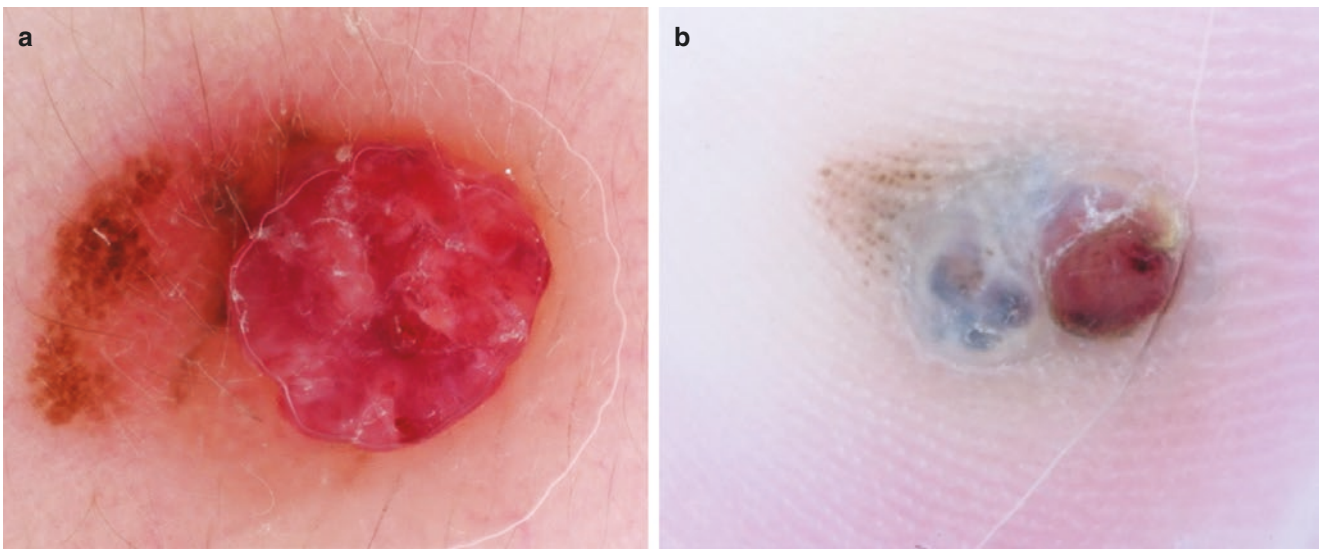

Fig. 4.2 (**a**) Dermatoscopy (×10) of an atypical Spitz tumor of the cheek in a 9-year-old girl: the lesion is asymmetric, with a flat area characterized by a brown, reticular pattern and a nodular, amelanotic component revealing milky red areas. (**b**) Dermatoscopy (×10) of an invasive, spitzoid melanoma (2.0 mm Breslow thickness) of the right foot in a 4-year-old girl: the lesion is asymmetric, with brown *globular pattern* and eccentric nodular component showing blue and red structureless areas

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Part III

Skin Infections/Infestations

Molluscum Contagiosum

5

Francesco Lacarrubba, Anna Elisa Verzì, Franco Dinotta, and Giuseppe Micali

5.1 Definition

Molluscum contagiosum is a highly contagious cutaneous viral infection commonly seen in children.

5.2 Epidemiology

Molluscum contagiosum is found worldwide, with an annual incidence reported between 2 and 20% [1, 2]. It predominantly affects children, as well as sexually active adults, and immunocompromised individuals, with no significant difference in sex distribution [3]. Transmission of the virus occurs directly by skin-to-skin contact, and lesions can spread in other body areas by autoinoculation. More than 40% of patients can identify a close friend or relative with the infection [4]. Vertical transmission via an infected genital tract has been rarely reported as congenital disease [5].

5.3 Etiology

Molluscum contagiosum virus (MCV) is a member of the *Molluscipoxvirus* genus in the family *Poxviridae*. MCV has four major viral types based upon DNA analysis: MCV-1, MCV-1a, MCV-2, and MCV-3. MCV-1 is commonly the cause of molluscum contagiosum in children, while MCV-2 tends to be more sexually transmitted and infects older patients [2]. The pathogenesis of the lesions requires complex interactions between virus and its host, and several anti-chemotactic and anti-apoptotic proteins have been postulated to have a role [6].

5.4 Clinical Features

The incubation period of the virus is from 2 to 8 weeks with a range extending to 6 months [7]. Typically, lesions appear as small, pink to skin-colored, dome-shaped papules, with central umbilication (Figs. 5.1a, 5.2a, 5.3a, and 5.4a). They usually have an average size of 3–5 mm in diameter, although giant lesions may sometimes be observed [8, 9]. Lesions may be solitary, but they are often multiple and grouped together. Any cutaneous area may be involved. Common sites are the face, neck, axillae, antecubital and popliteal fossae, chest, thighs, and buttocks. After trauma, or spontaneously, inflammatory changes result in suppuration and crusting. In immunosuppressed individuals, infection is typically more severe and widely disseminated.

5.5 Differential Diagnosis

Molluscum contagiosum can mimic keratoacanthoma, verruca vulgaris, condyloma acuminatum, syringoma, hidrocystoma, and comedo.

5.6 Diagnosis

The diagnosis of molluscum contagiosum is based on observation of the characteristic umbilicated papules. However, in some patients it may be a challenge. Tzanck preparation, performed on a scraping from a representative lesion, followed by microscopy examination shows a typical pattern of numerous discrete ovoid intracytoplasmic inclusion bodies,

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called molluscum bodies or Henderson-Patterson bodies [1]. Reflectance confocal microscopy has been suggested as a new technique for the diagnosis in selected cases [10].

5.7 Dermatoscopy

Dermatoscopy shows a typical pattern consisting of multiple, yellowish-white, lobulated, amorphous central structures surrounded by a crown of linear, fine, and sometimes blurred vessels, some of them branching. These vessels usually do not cross the center of the lobules (Figs. 5.1b, 5.2b, 5.3b, and 5.4b) [11, 12]. Dermatoscopic features seem to vary depending on the size of the lesions with large lesions showing the typical polylobular structure and small ones showing four-leaved clover-like structures [13].

5.8 Histopathological Correlation

The central polylobular structure observed by dermatoscopy correlates to the well-defined, lobulated, endophytic hyperplasia, with fibrous septa between the lobules seen at histopathology (Fig. 5.5) [13, 14]. Moreover, the vascular pattern corresponds histopathologically to the dilated vessels in the dermis.

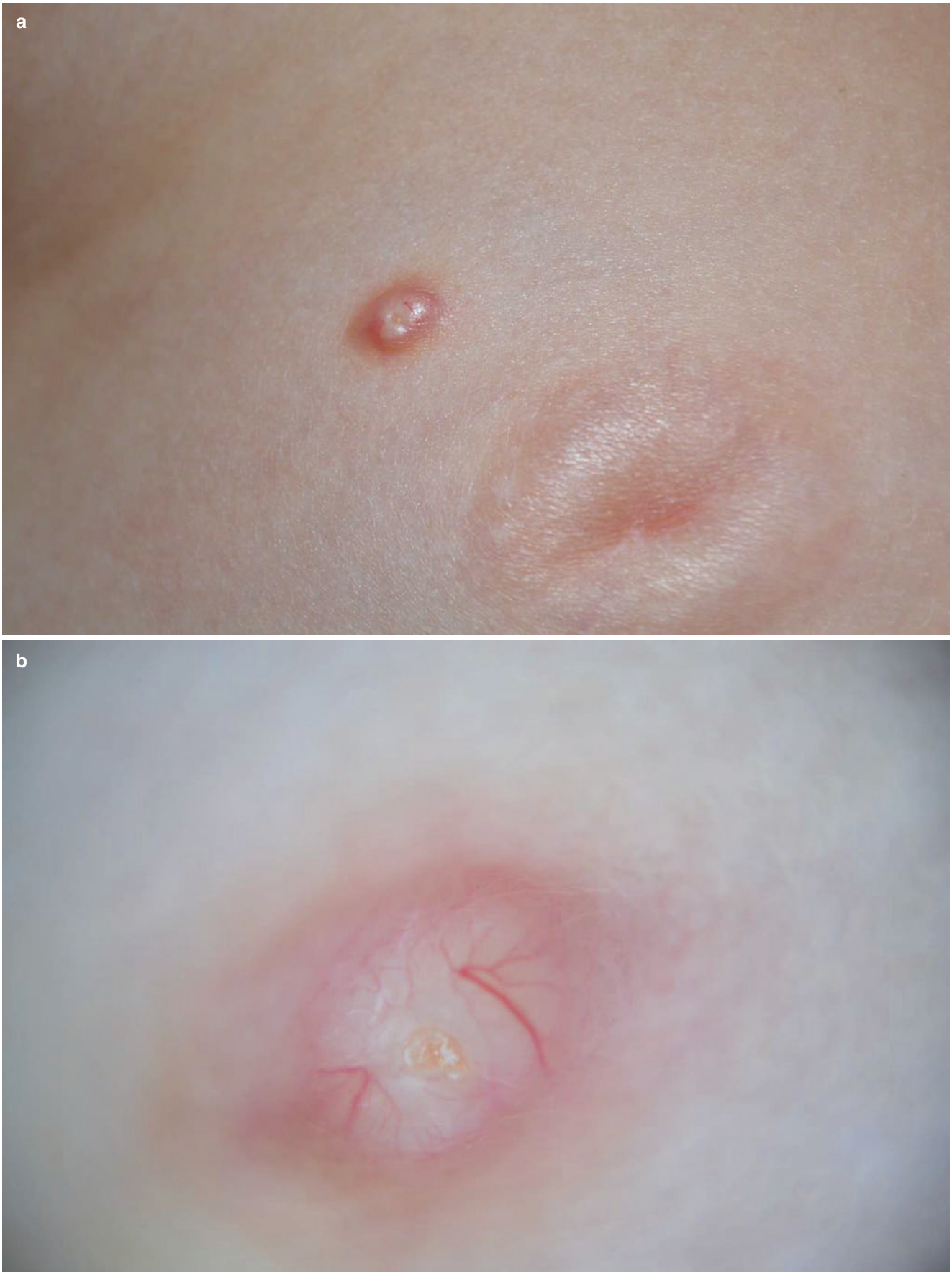

Fig. 5.1 (**a**) Molluscum contagiosum. Pink-whitish papule with central umbilication on the chest of an 8-year-old boy. (**b**) Dermatoscopy (×10): yellowish-white central structureless areas with linear and branching vessels at the periphery

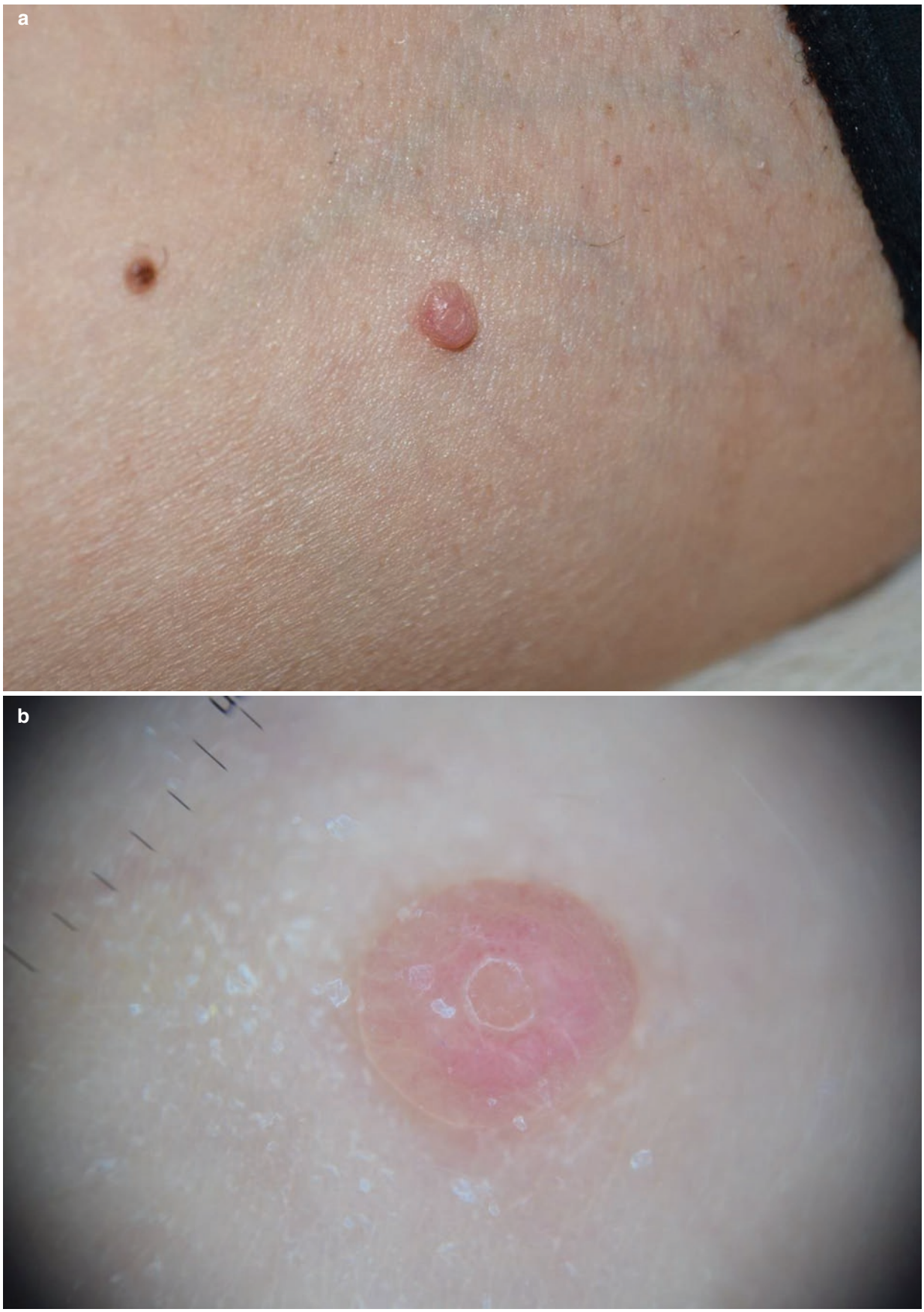

Fig. 5.2 (**a**) Molluscum contagiosum. Pink, dome-shaped papule with central umbilication on the thigh of a 14-year-old girl. (**b**) Dermatoscopy (×10): amorphous central structures surrounded by fine and blurred vessels

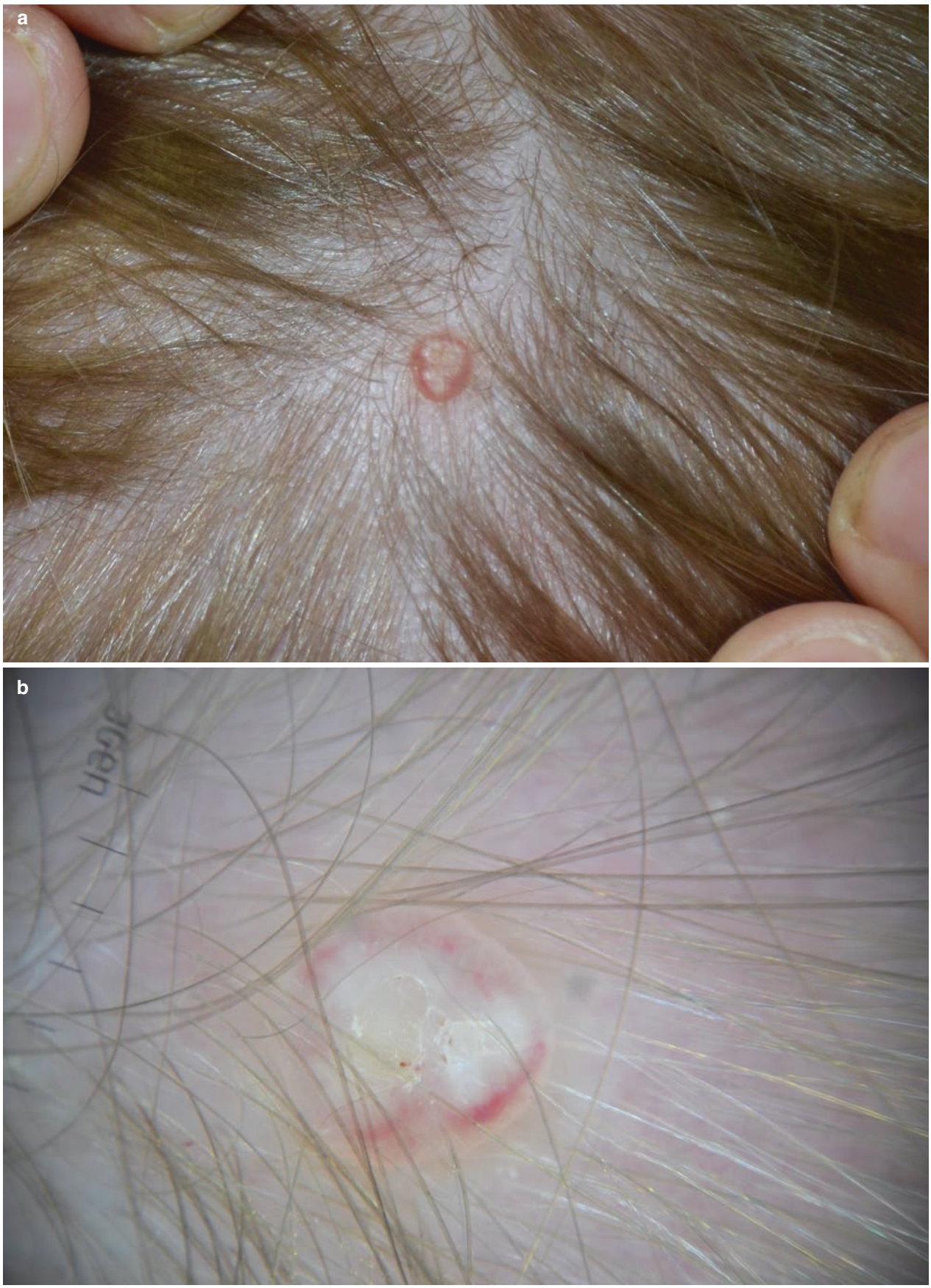

Fig. 5.3 (**a**) Molluscum contagiosum. Pink-whitish papule on the scalp of a 5-year-old boy. (**b**) Dermatoscopy (×10): yellowish-white, lobulated, amorphous central structures (four-leaved clover-like structures) surrounded by a crown of linear and blurred vessels

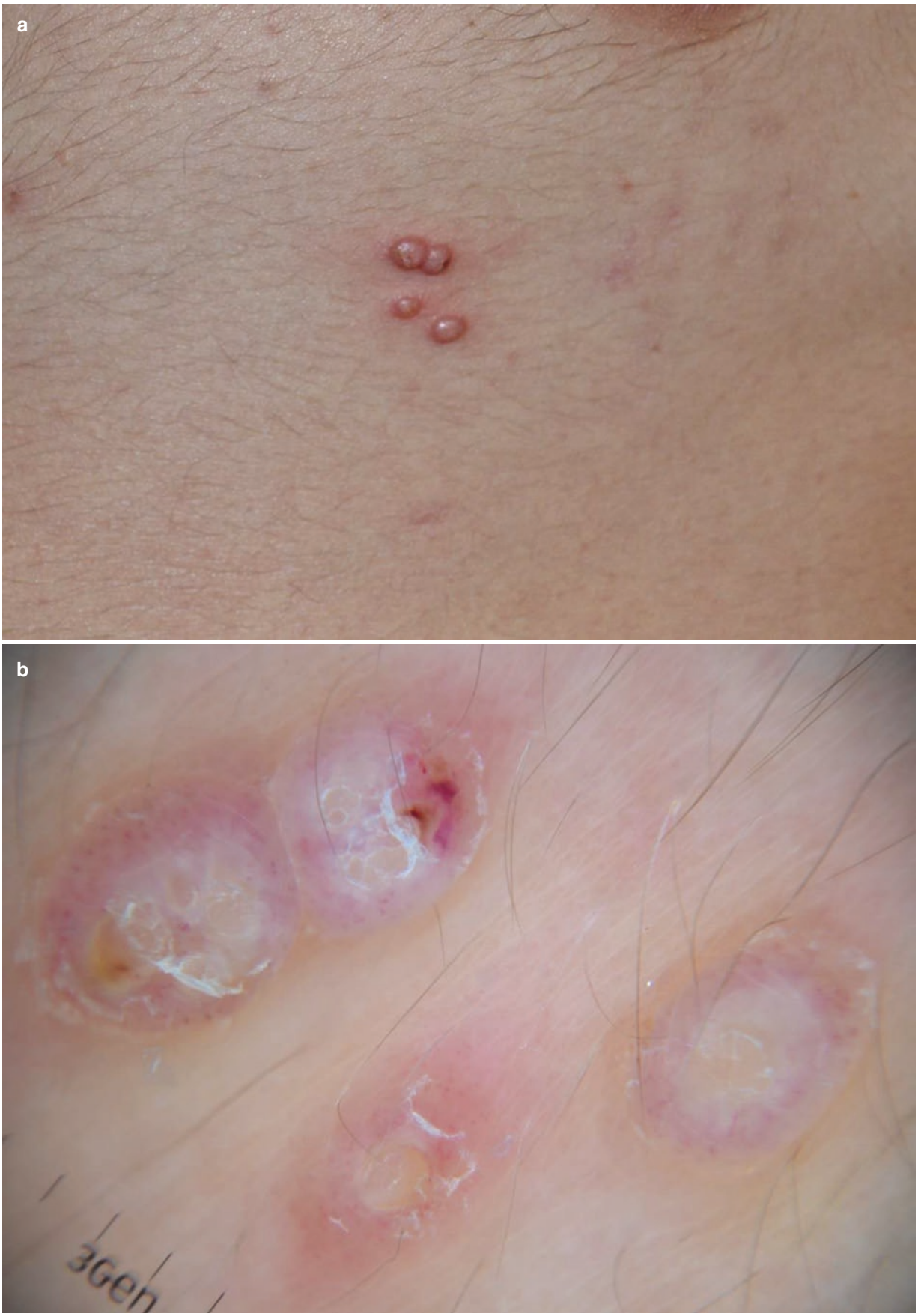

Fig. 5.4 (**a**) Molluscum contagiosum. Multiple pink-whitish papules on the trunk of a 14-year-old boy. (**b**) Dermatoscopy (×10): all lesions show yellowish central structureless areas with linear and branching vessels at the periphery

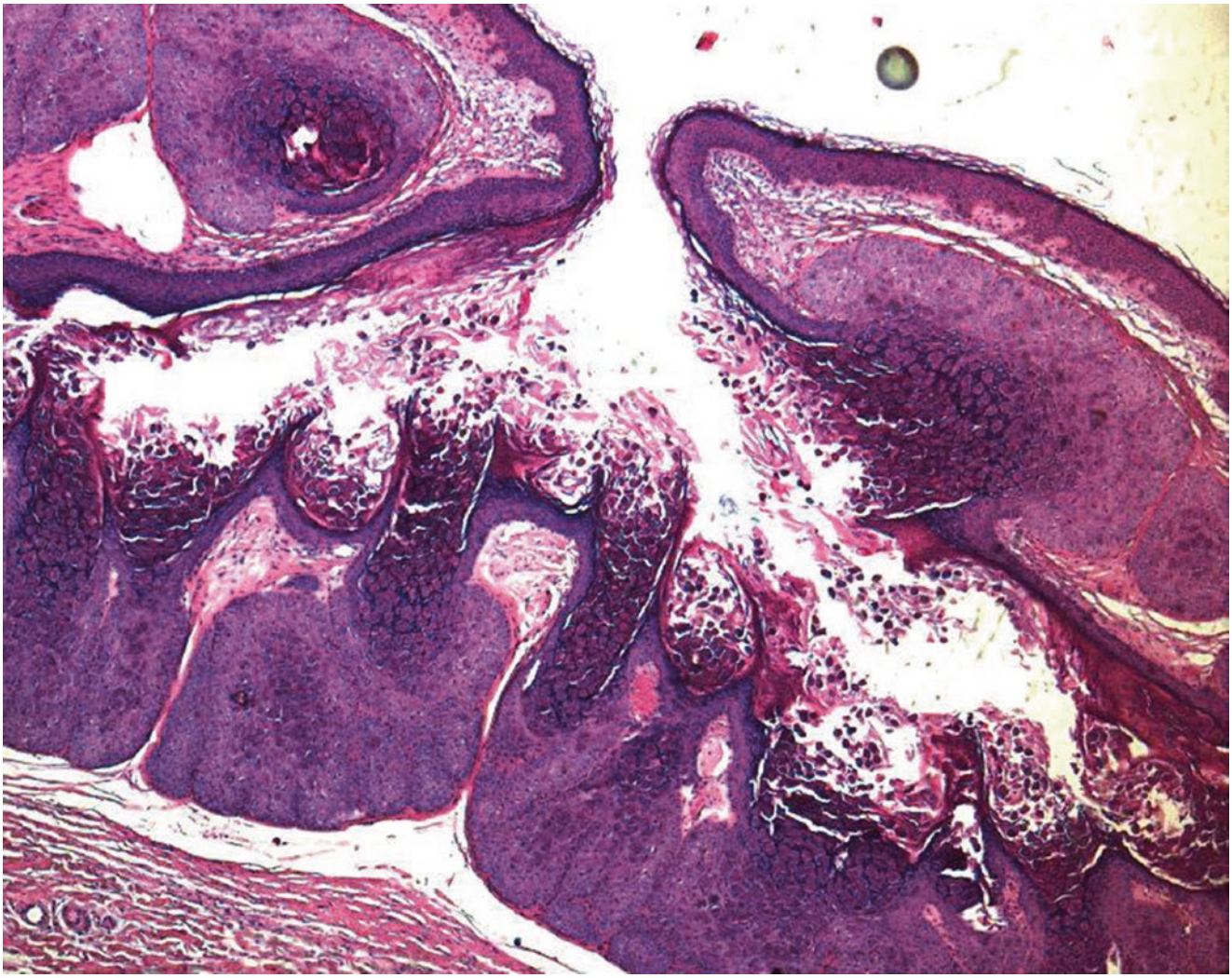

Fig. 5.5 Molluscum contagiosum. Histopathology: well-defined lobulated, endophytic hyperplasia, with fibrous septa between the lobules [H&E staining; magnification: ×100]

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Cutaneous and Anogenital Warts

6

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6.1 Definition

Warts are common benign epithelial keratinocytes proliferations caused by human papillomavirus (HPV) infection that may involve the skin and the mucous membranes.

6.2 Epidemiology

6.2.1 Cutaneous Warts

They are very common in children although limited epidemiologic data exist [1]. The prevalence of cutaneous warts is estimated to range from 5% to 21.7%, reaching up to 44% in some primary school classes [2–5]. Lesions often resolve without any treatment. In a retrospective study on 214 patients, warts resolved in 65% of children by 2 years and in 80% within 4 years, regardless of treatment [6].

6.2.2 Anogenital Warts

They are less commonly seen in children than in adult population. In a study on 211 preschool children, anogenital warts were observed in about 2% of children [7].

6.3 Etiology

6.3.1 Cutaneous Warts

Lesions are caused most frequently by the cutaneous HPV types 1, 2, 3, 4, 7, 10, 27, and 57 [8]. Infection is predominantly by direct skin-to-skin contact but may be indirect through contaminated surfaces and objects.

6.3.2 Anogenital Warts

HPV-6 and 11 are typically associated, but HPV-16, 18, 31, 33, and 35 are found occasionally. Similarly to cutaneous warts, transmission occurs via skin-to-skin contact; therefore in children, the possibility of sexual abuse should always be excluded. Vertical transmission has also been suggested including antenatal, perinatal, or postnatal route [9].

6.4 Clinical Features

6.4.1 Cutaneous Warts

Cutaneous lesions are classified according to morphology and anatomic localization. Common warts (verrucae vulgaris) are exophytic papules or plaques of variable size with hyperkeratotic surface (Fig. 6.1a). They are usually located in sites prone to trauma, i.e., on the hand and fingers, but they may occur anywhere. In periorificial areas, they appear pedunculated and filiform (Fig. 6.2a). Palmar and plantar warts are endophytic/exophytic papules often painful at pressure (Fig. 6.3a). They may coalesce into large plaques named as mosaic warts. Flat warts (verrucae planae) are often multiple, pinkish, brownish, or skin-colored slightly elevated papules sometimes in a linear array that commonly involve the face, hands, and arms (Fig. 6.4a).

6.4.2 Anogenital Warts

Lesions in the anogenital region are also known as condylomata acuminata. They appear as exophytic, cauliflower-like, pedunculated, or broad-based papules or plaques with brownish or skin-colored surface (Figs. 6.5a and 6.6a).

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6.5 Differential Diagnosis

6.5.1 Cutaneous Warts

In children, molluscum contagiosum, localized hyperkeratosis (callus), comedones, nevi, acrochordons, acrokeratosis verruciformis, and lichen nitidus may resemble cutaneous warts.

6.5.2 Anogenital Warts

It mainly includes molluscum contagiosum, pearly penile papules or vestibular papillomatosis, Fordyce's spots, angiokeratoma of Fordyce, lymphangiomas, lichen nitidus and epidermoid cyst.

6.6 Diagnosis

The diagnosis of cutaneous and anogenital warts is usually based on typical clinical appearance.

6.7 Dermatoscopy

6.7.1 Cutaneous Warts

The main dermatoscopy pattern suggestive of common warts is the presence of irregular whitish structures (mosaic pattern), typically associated with irregularly distributed, hemorrhagic reddish to black dots (Fig. 6.1b) [10–12]. In exophytic lesions, multiple papillomatous projections (finger-like) containing elongated and dilated vessels that are surrounded by whitish halos may be observed (Fig. 6.2b) [10–12]. Linear, hairpin, and coiled vessels can be found [13]. Lesions may also reveal a daisy flower pattern consisting of radial petallike keratotic projections with a central mass of keratin [13]. Palmoplantar warts are characterized by the presence of verrucous, yellowish structureless areas with multiple irregularly distributed red to brown to black dots or linear streaks due to hemorrhages (Fig. 6.3b). These dots are helpful criteria to distinguish plantar warts from calluses, which lack blood spots and show a translucent central core or a homogeneous opacity [14]. Skin lines are typically interrupted in palmoplantar warts [13]. Flat warts reveal regularly distributed, tiny red dots on a light brown to yellow background (Fig. 6.4b) [12]. These features are helpful to distinguish them from acne comedones, which typically reveal a central white to yellow pore corresponding to the hair follicle opening, and from verruca plana-like keratosis which shows a brain-like appearance [15].

6.7.2 Anogenital Warts

Dermatoscopy patterns of anogenital warts vary according to the clinical presentation. Papular lesions show a whitish network circumscribing areas centered by dilated glomerular or dotted vessels (mosaic pattern) (Fig. 6.5b), whereas cauliflower-like warts reveal multiple, irregular whitish projections arising from a common base and comprising elongated and dilated vessels that are usually more pronounced at the periphery (Fig. 6.6b) [16–18]. For the evaluation of genital lesions, videodermatoscopy is much more preferable to dermatoscopy with a handheld device, both to achieve higher magnification and resolution and to obviate physician's close contact with the skin surface [18, 19].

6.8 Histopathological Correlation

6.8.1 Cutaneous Warts

Histopathologically, the variable amount of acanthosis, papillomatosis, and hyperkeratosis correlates to the various dermatoscopic patterns seen in cutaneous warts (Fig. 6.7). The presence of elongated and tortuous vessels as well as hemorrhages/extravasation of erythrocytes corresponds to the dotted, linear, hairpin, and coiled vessels and reddish/black dots seen at dermatoscopy, respectively.

6.8.2 Anogenital Warts

The whitish reticular network and the glomerular vessels dermatoscopically characterizing papular lesions are related to hyperkeratosis and acanthosis and tortuous and dilated capillaries in the papillary dermis, respectively (Fig. 6.8). As regards cauliflower-like lesions, the whitish projections correspond to marked papillomatosis, hyperkeratosis, and acanthosis [18, 19].

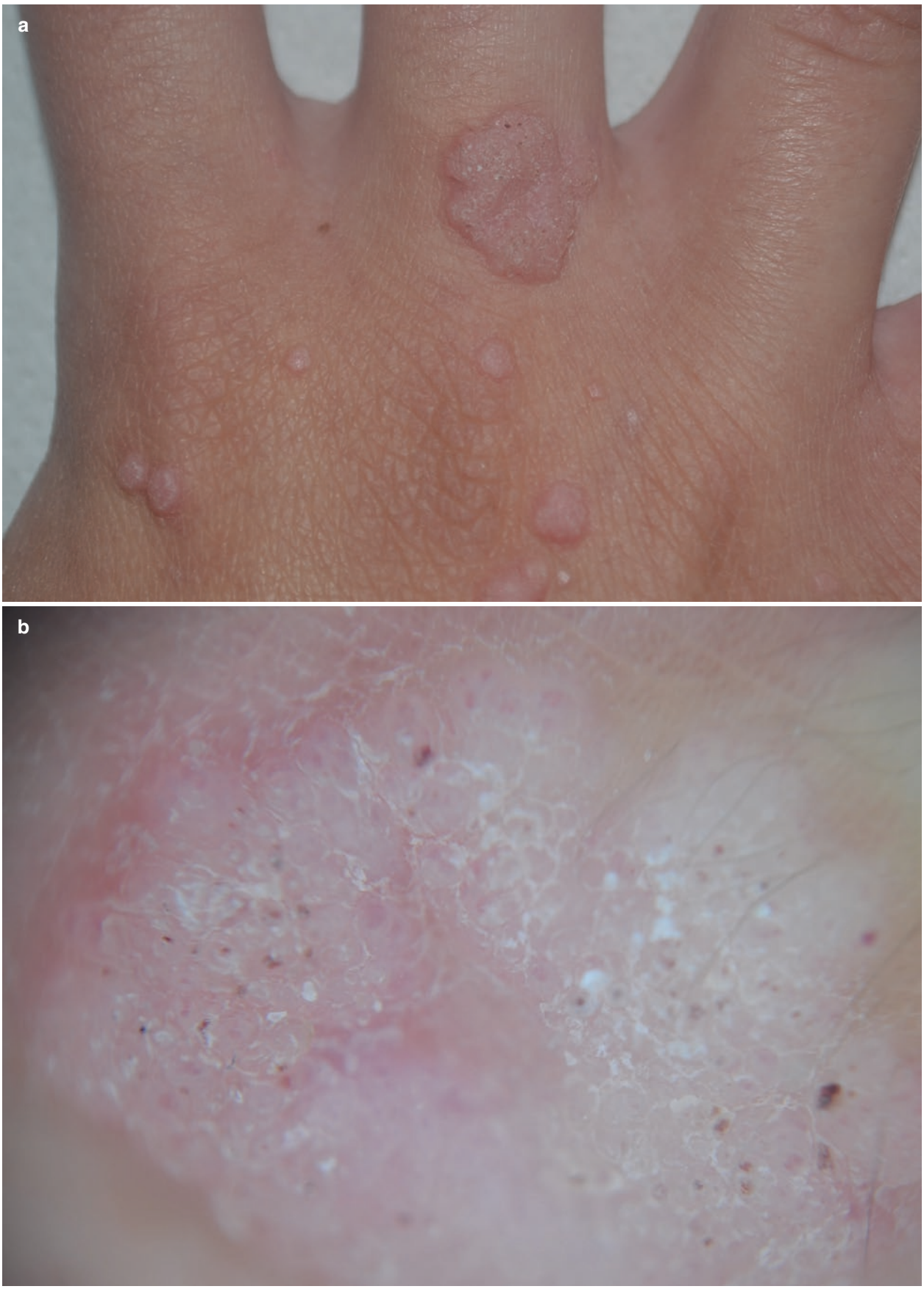

Fig. 6.1 (**a**) Cutaneous warts. Multiple hyperkeratotic papules and plaques on the right hand in a 9-year-old boy. (**b**) Dermatoscopy (×10): rounded structures resembling a puzzle (*mosaic pattern*), associated with irregularly distributed, hemorrhagic reddish dots

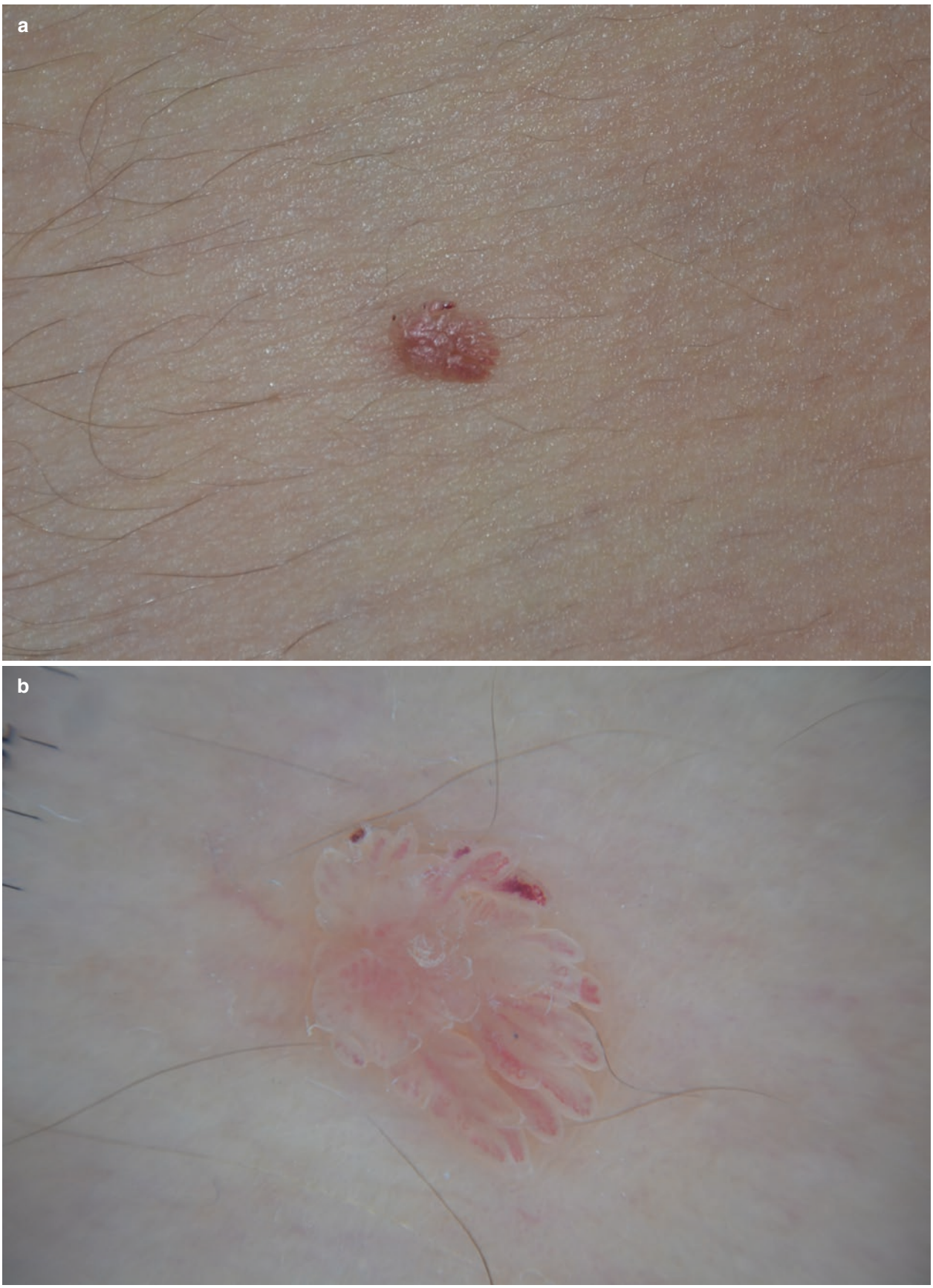

Fig. 6.2 (**a**) Cutaneous wart. Slightly hyperkeratotic papule with filiform sprouts. (**b**) Dermatoscopy (×10): multiple papillomatous projections (*finger-like*) containing central red hairpin and coiled vessels that are surrounded by whitish halos

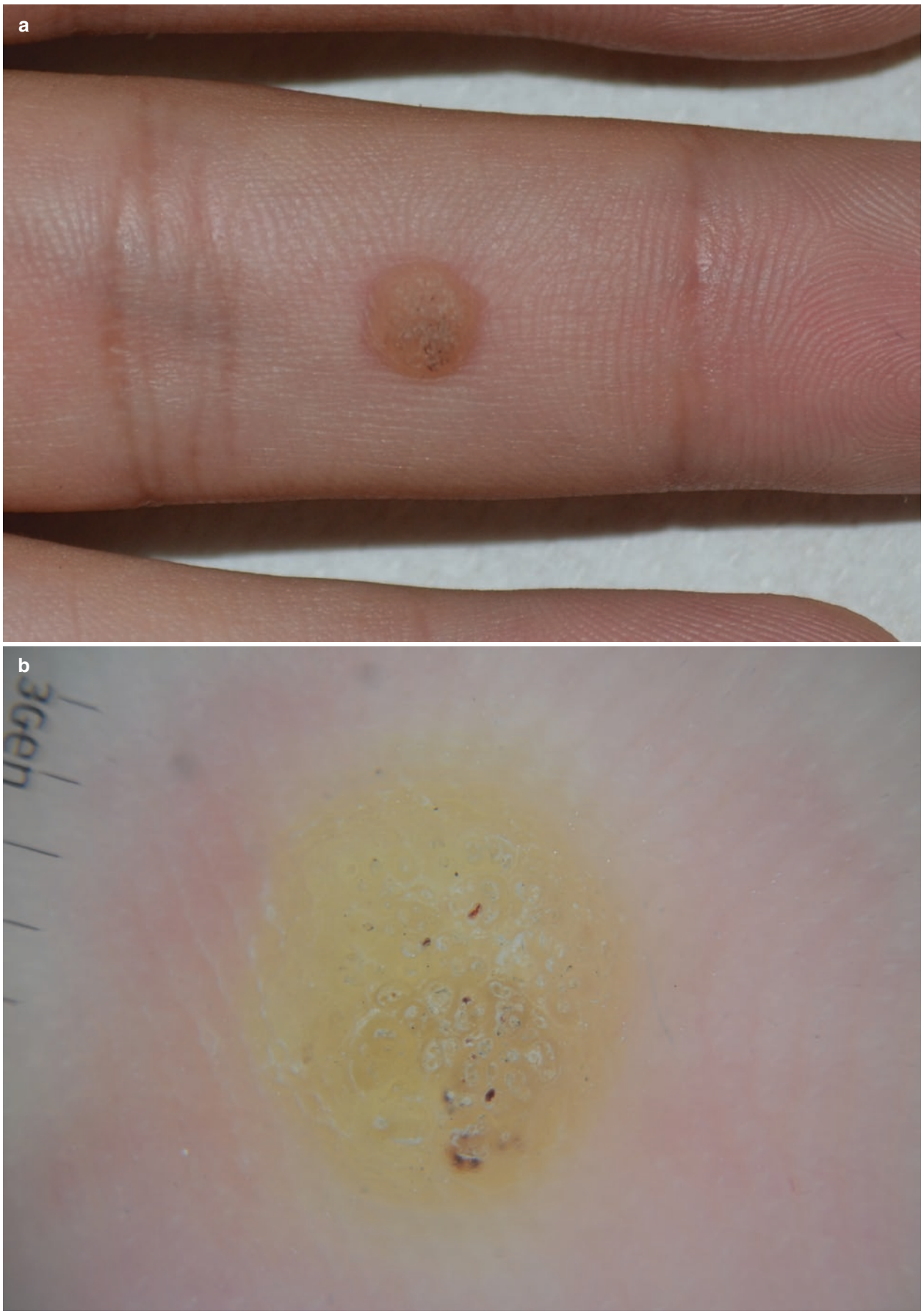

Fig. 6.3 (**a**) Palmar wart. Hyperkeratotic papule on the flexor surface of the finger. (**b**) Dermatoscopy (×10): verrucous, yellowish structureless area with irregularly distributed brownish dots and interrupted skin lines

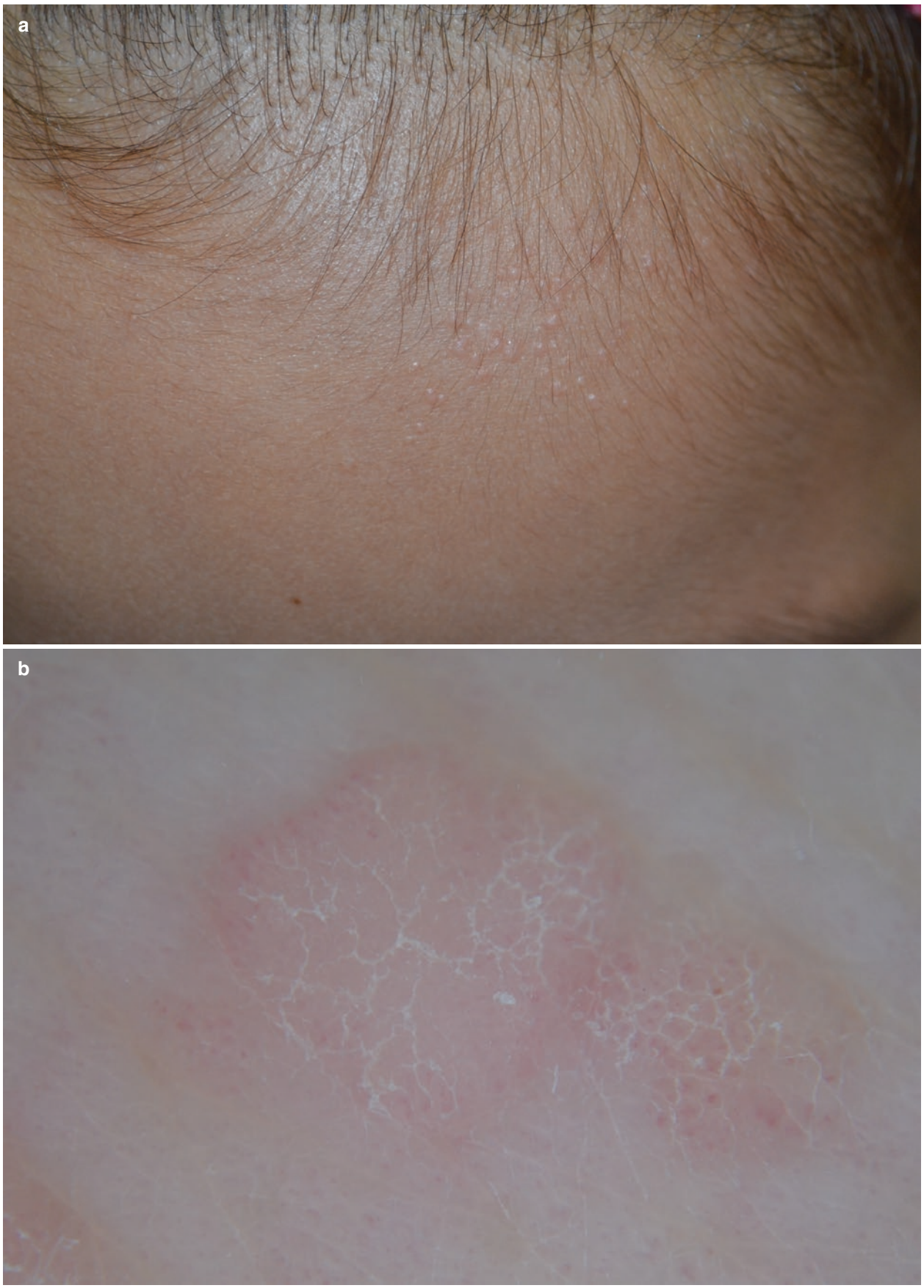

Fig. 6.4 (**a**) Flat warts. Multiple, skin-colored, slightly elevated papules on the forehead. (**b**) Dermatoscopy (×10): multiple red dots on a light yellowish background

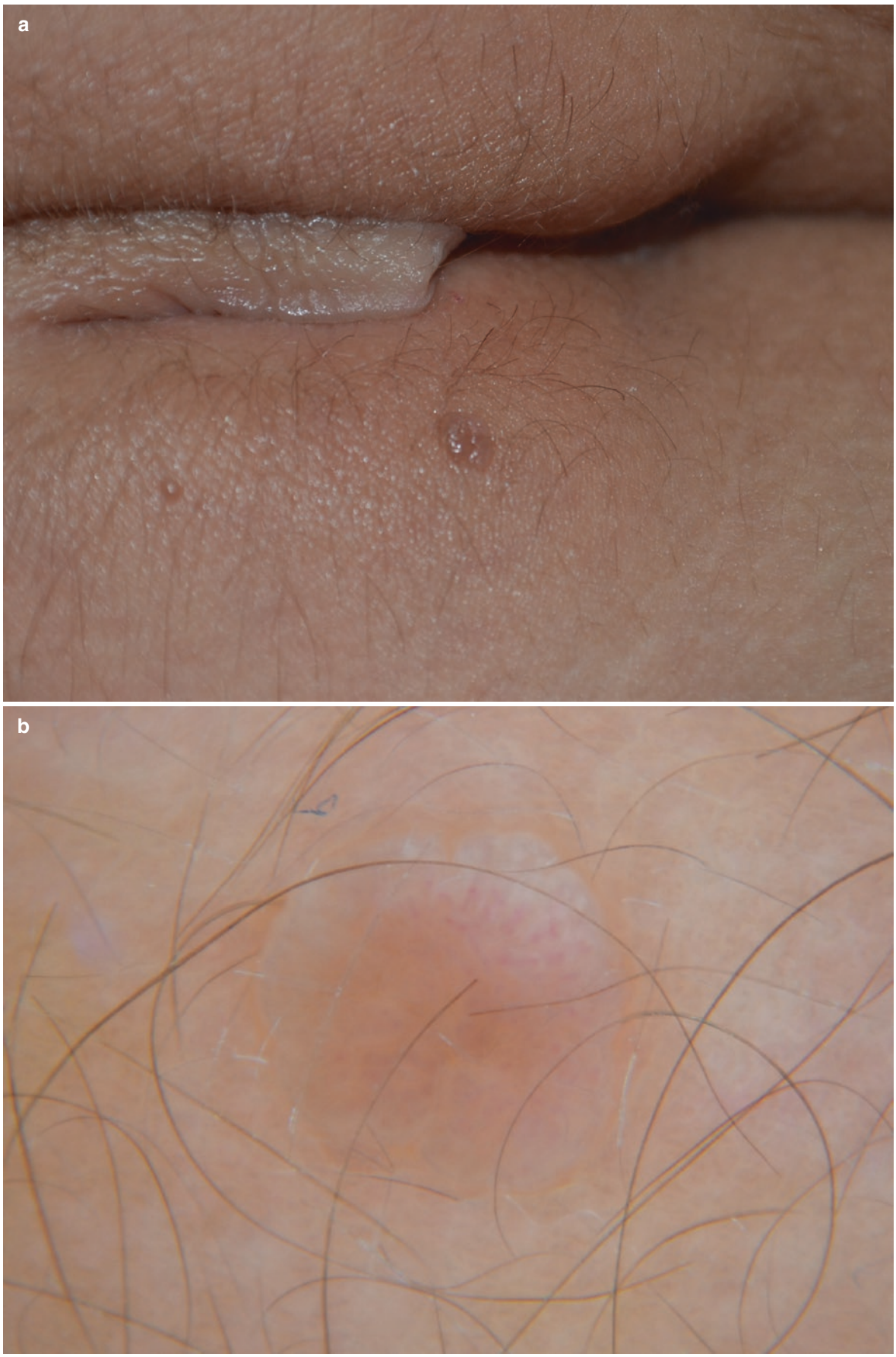

Fig. 6.5 (**a**) Genital warts. Broad-based skin-colored papules located on the right labium majus. (**b**) Dermatoscopy (x10): whitish network and dilated dotted vessels at the periphery of the lesion

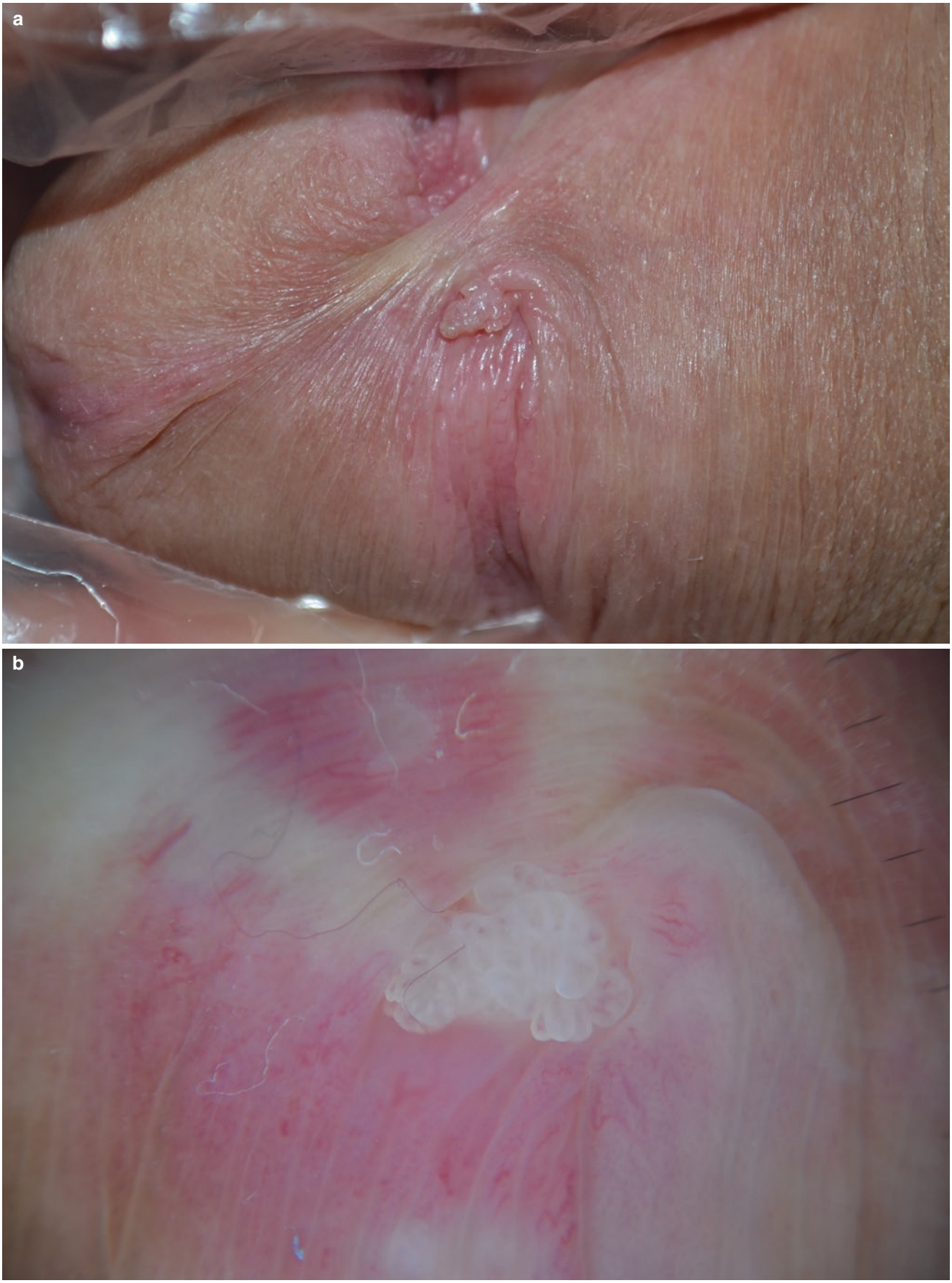

Fig. 6.6 (**a**) Genital warts. Exophytic and cauliflower-like skin-colored papule located on the frenulum. (**b**) Dermatoscopy (×10): multiple, irregular whitish projections filled with elongated and dilated vessels

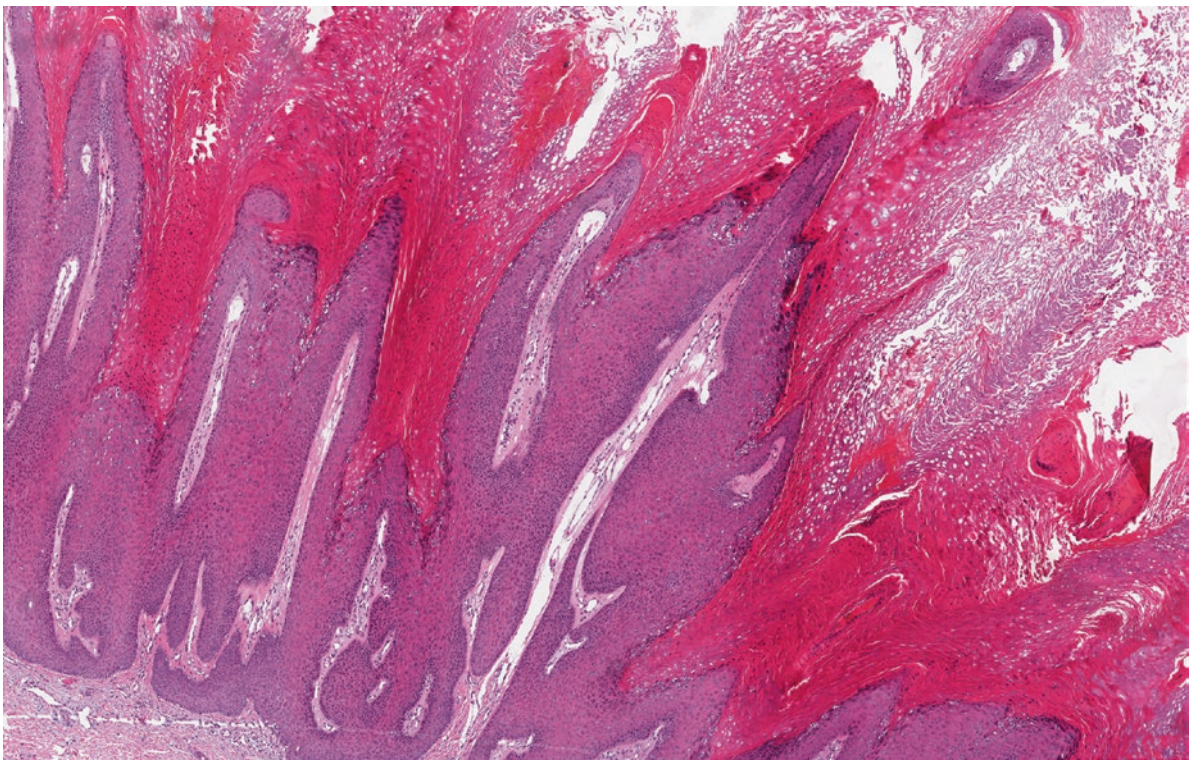

Fig. 6.7 Cutaneous wart. Histopathology: hyperkeratosis, acanthosis, and papillomatosis with elongated vessels [H&E staining; magnification: ×100]

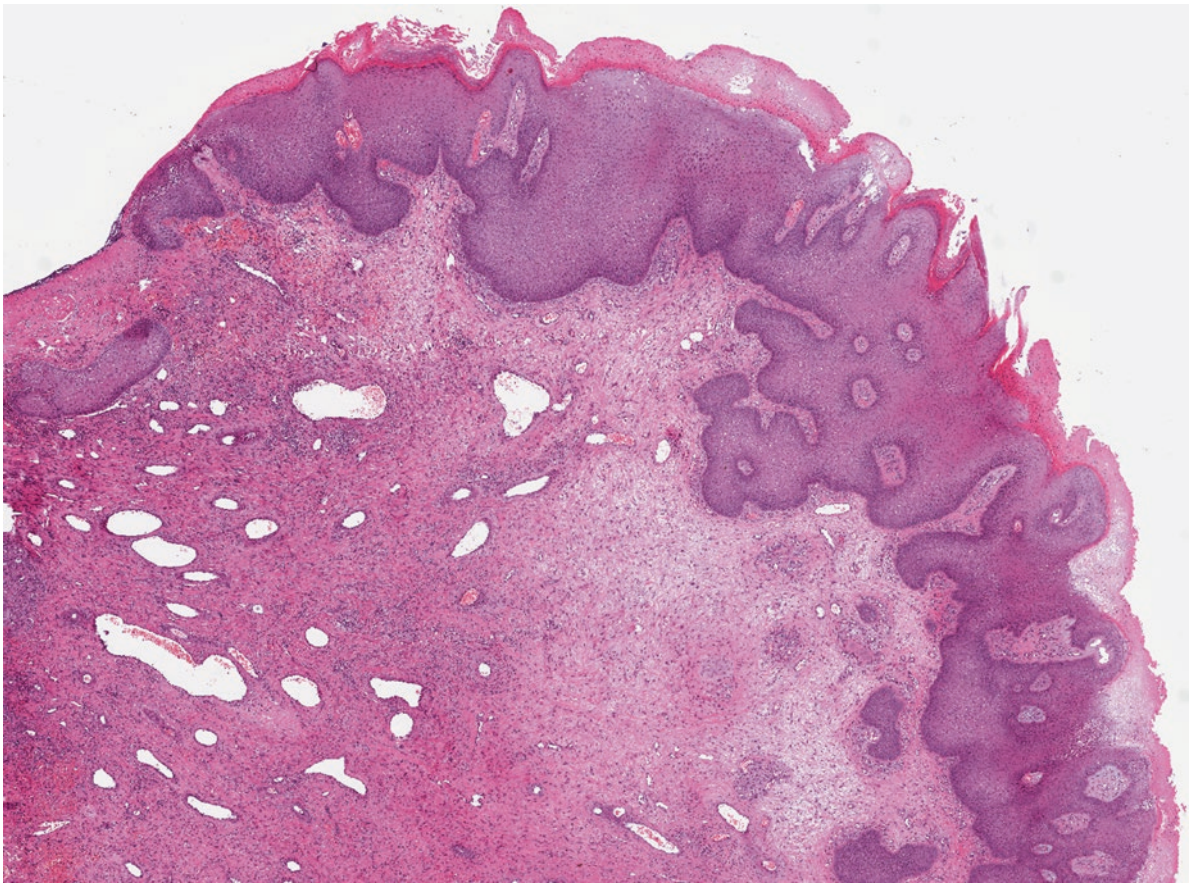

Fig. 6.8 Genital wart. Histopathology: hyperkeratosis, acanthosis, and tortuous and dilated capillaries in the papillary dermis [H&E staining; magnification: ×50]

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Tinea Capitis

7

Francesco Lacarrubba, Simona Boscaglia, and Giuseppe Micali

7.1 Definition

Tinea capitis (TC), also known as scalp ringworm, is a superficial infection of the scalp caused by dermatophytes that have predilection for hair shafts and follicles. It is a typical fungal infection that involves principally children with variation about epidemiology, etiology, and distribution according to different geographic areas [1].

7.2 Epidemiology

The incidence of TC is variable from 1% to 11% according to geographic region and timing of data collection in different studies [2]. It also depends on different factors such as migration, living conditions, tourism, and cultural habits [3]. Children aged 3–7 years are more frequently affected without gender difference. The higher prevalence in children is due to the absence of scalp sebum secretion and Malassezia colonization that represent two protective factors that reduce the dermatophyte ability to cause the infection. Other risk factors that are generally present in children include promiscuity in crowed places (school), direct contact with pets, and poor hygiene [4, 5].

7.3 Etiology

The prevalence of the causative agent is variable depending on the geographic areas. In the Mediterranean countries, *Microsporum canis*, a zoophilic dermatophyte, is the most frequent agent causing external colonization of the hair ("ectothrix" invasion). Recently, especially in urban areas, an increase of anthropophilic dermatophytes, such as *Trichophyton tonsurans* and *soudanense*, has been observed, probably due to immigration fluxes [6]. In this case, the hair shaft is filled with hyphae and spores ("endothrix" invasion).

7.4 Clinical Features

Clinically, TC may present with different manifestations depending on the type of pathogen involved and host's defenses. It is necessary to distinguish between noninflammatory and inflammatory forms [1, 7].

The noninflammatory forms (Figs. 7.1a, 7.2a, and 7.3a) include the *ectothrix type* and the *endothrix type*. The first is characterized by the presence of patchy, circular areas of alopecia, where broken-off hairs with gray color, due to arthrospores coating and fine scaling, may be observed. The second type may appear either as multiple alopecic areas with fine scaling and follicles filled by broken-off hairs or black dots or as diffuse scaling resembling dandruff without alopecic areas.

The inflammatory forms mainly include *Kerion celsi*, a severe form of TC which presents with multiple confluent painful inflamed plaques associated with pustules and crusts on alopecic areas, and *favus*, due to *Trichophyton schoenleinii* and endemic in Africa. The latter shows a typical lesion known as scutula, a cup-shaped and yellow crust composed by hyphae and keratin located around follicular openings [1].

7.5 Differential Diagnosis

Any condition that may cause alopecia or scalp scaling should be distinguished by TC. In particular, in children common disorders are alopecia areata, trichotillomania, psoriasis, and seborrheic dermatitis [7].

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7.6 Diagnosis

The diagnosis of TC is classically obtained through the microscopic direct examination of skin scraping for hyphae and/or through mycological cultures that allow the identification of the specific agent [8]. At Wood's lamp examination, a green florescence may be observed in infections due to *Microsporum* spp., which are not present in TC due to other species (e.g. *Trichophyton* spp.) [7].

7.7 Dermatoscopy

Dermatoscopy may be very helpful for a rapid diagnosis of TC, although it does not allow the identification of the responsible agent. *Comma hairs* represent a typical dermatoscopic feature (Fig. 7.1b); the invasion by fungus determines an impairment of the hair shaft that consequently is bended and broken, showing a C-shaped or a corkscrew form (in Afro-Americans) (Fig. 7.3b) [8–13]. Another typical feature is represented by *Morse code hairs* (or *interrupted hairs*), characterized by hair hair shaft presenting alternating whitish and brownish bands (Fig. 7.2b). At high magnification (x100), the light areas can be appreciated as empty spaces where the shaft is less resistant and more prone to bend and break (Figs. 7.4 and 7.5) [9]. Finally, *zig-zag hairs* have been described. In the inflammatory forms of TC, there are less specific features as erythema, scaling, pustules, and crusts [8, 9].

7.8 Histopathological Correlation

Biopsy is not a common procedure in the diagnosis of TC. Histopathology shows the signs of cutaneous reaction to the fungal invasion as edema, vasodilatation, inflammatory infiltrate, and epidermal disarrangement in which hyphae may be evidenced.

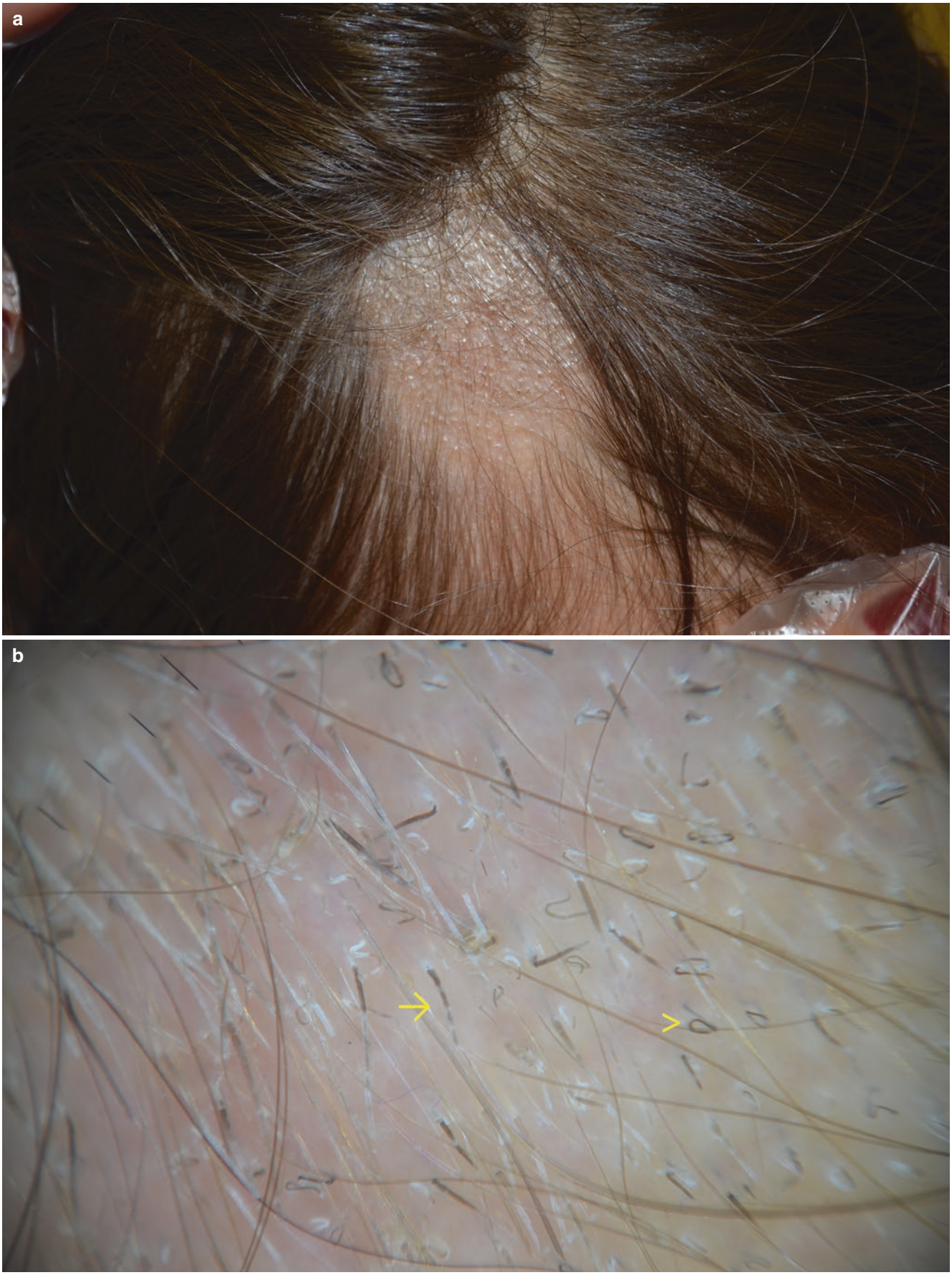

Fig. 7.1 (**a**) Tinea capitis. Ovular alopecic area of the temporal region of the scalp in a 7-year-old female. (**b**) Dermatoscopy (×10): *comma hairs* (arrowhead) and *Morse code hairs* (arrow)

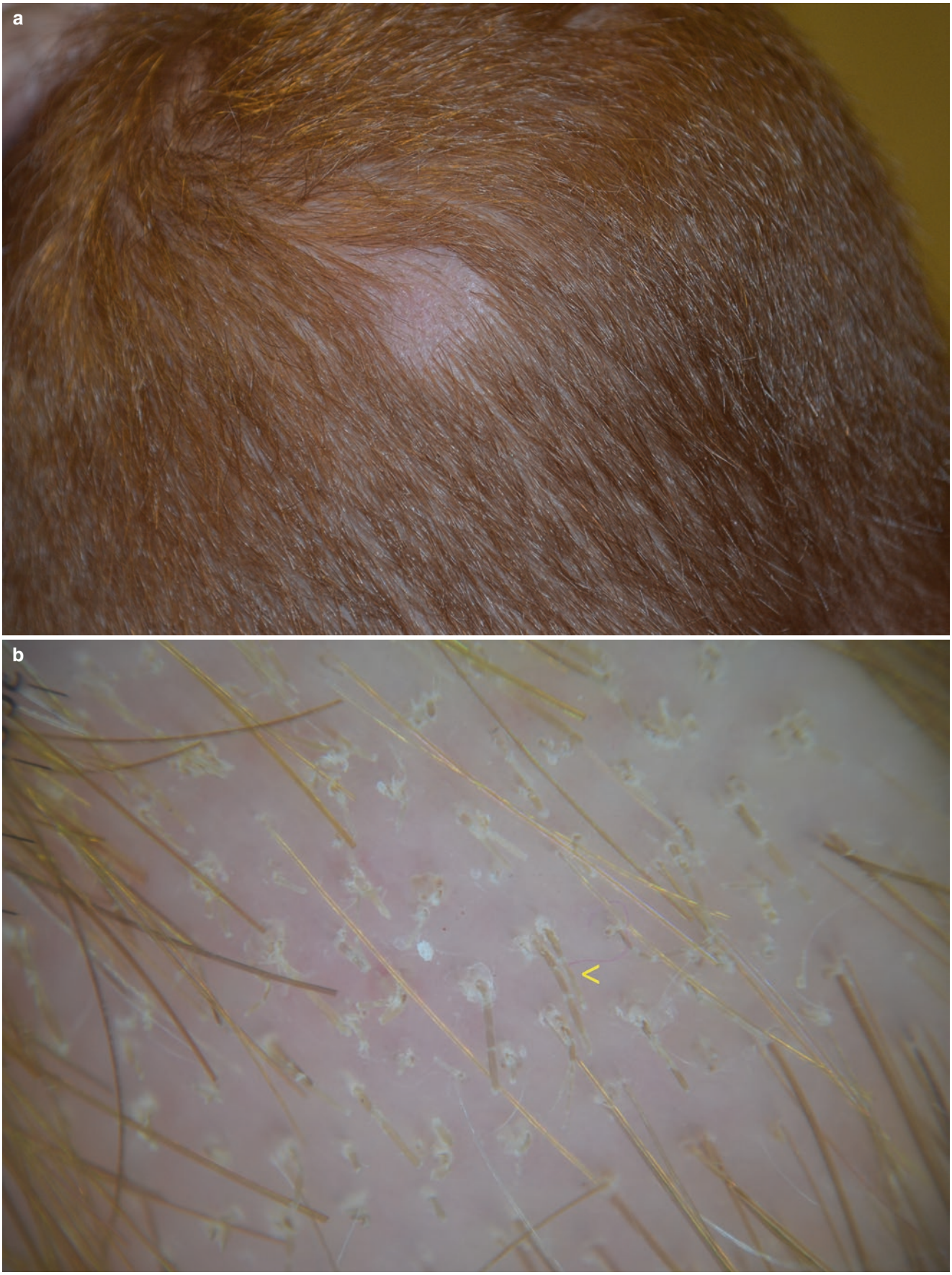

Fig. 7.2 (**a**) Tinea capitis. Small roundish alopecic area of the vertex in a 6-year-old boy. (**b**) Dermatoscopy (×10): diffuse scaling and *Morse code hairs* (arrowhead)

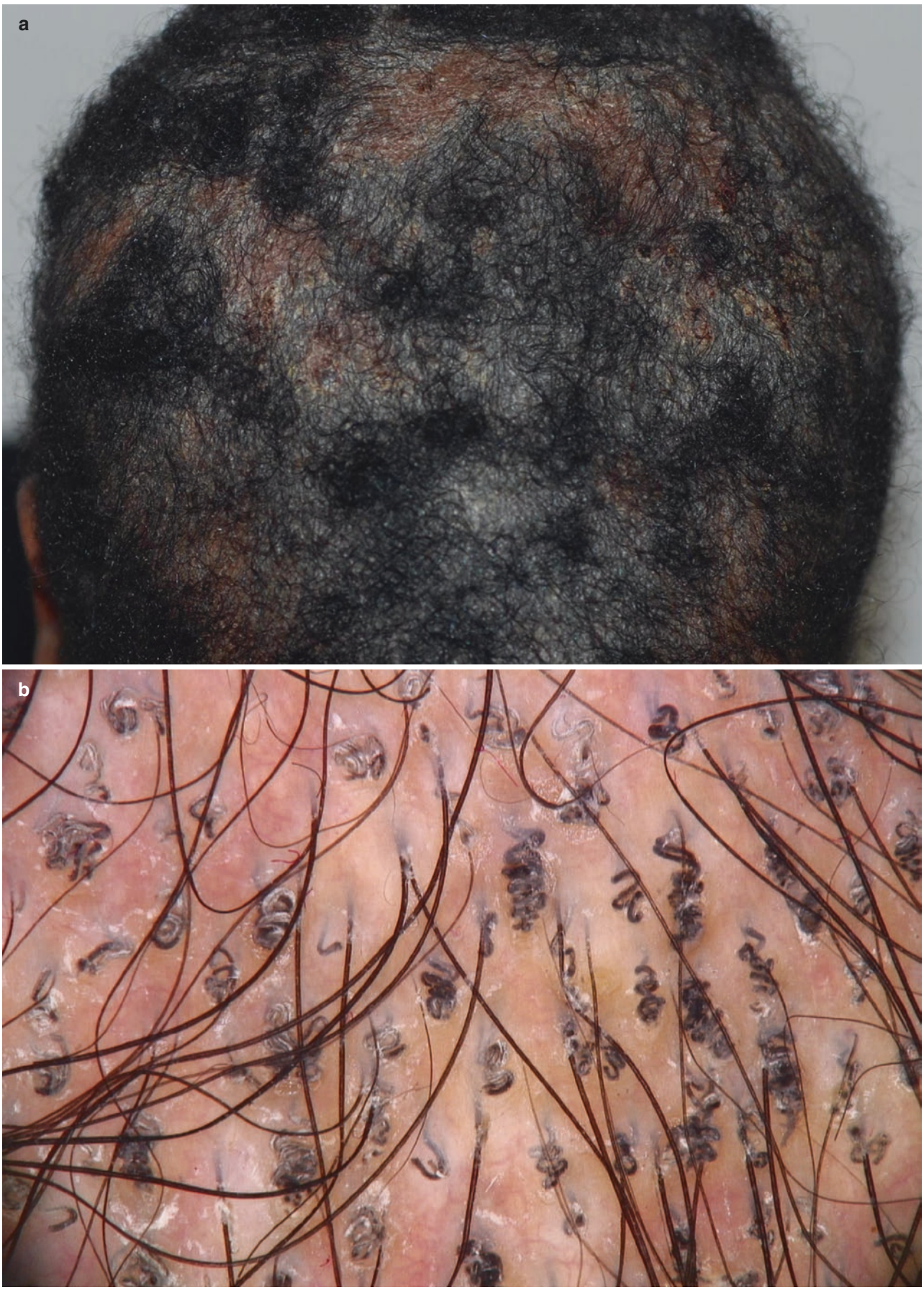

Fig. 7.3 (**a**) Tinea capitis. Multiple alopecic patches with scaling in an African-American boy. (**b**) Dermatoscopy (×10): multiple *corkscrew hairs* (Courtesy of Bianca Maria Piraccini, MD)

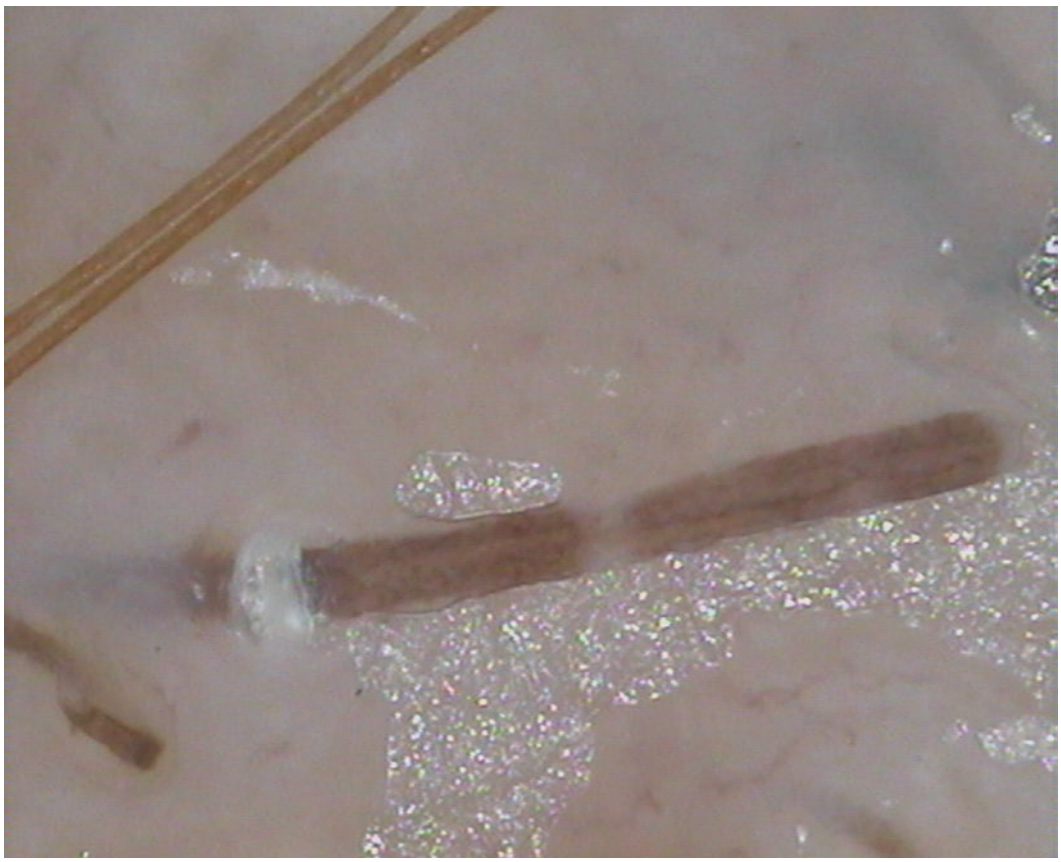

Fig. 7.4 Tinea capitis. High-magnification dermatoscopy (×150) of a *Morse code hair*

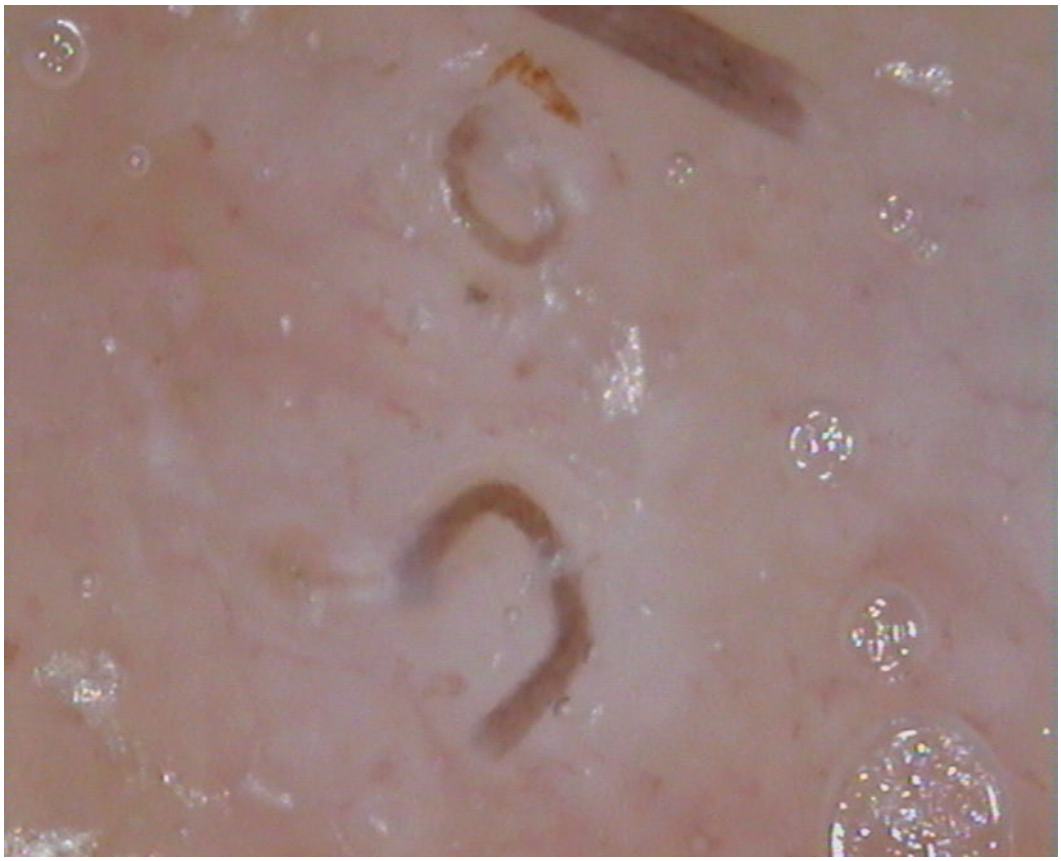

Fig. 7.5 Tinea capitis. High-magnification dermatoscopy (×150) of a *comma hair*

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Scabies

8

Giuseppe Micali, Giorgia Giuffrida, Enrica Quattrocchi, and Francesco Lacarrubba

8.1 Definition

Scabies is a common highly contagious ectoparasitosis caused by the mite *Sarcoptes scabiei hominis*.

8.2 Epidemiology

Scabies is diffused worldwide with a prevalence that is estimated to be up to 300 million [1]. People in resource-poor communities and vulnerable institutions, such as hospitals, nursing homes, schools, and prisons, are especially susceptible to scabies as well as to the secondary complications of infestation [2]. The most common source of transmission is prolonged skin-to-skin contact with an infected individual. It takes about 15–20 min of close contact for successful direct transmission, and intrafamilial transmission is frequently reported [3].

8.3 Etiology

Sarcoptes scabiei hominis is an arthropod belonging to the class of Arachnida, an obligate parasite that completes its entire life cycle on humans. The evolutionary cycle of the mite takes about 15 days. The fertilized female digs a serpentine burrow in the epidermis, advancing about 2 mm per day. Spawning takes place at night, from the fourth day after fertilization, at a rate of about 1–5 eggs per day up to a total of 20–25 eggs. Following hatching, after 3–4 days of incubation, the larvae migrate to the skin surface and burrow into the intact stratum corneum to make short tunnels.

An infested host contains about 10–15 adult female mites on his body. However, in crusted or Norwegian scabies, common in immunosuppressed individuals, the mites can reach over one million.

8.4 Clinical Features

The typical sign of scabies is represented by the burrow, a serpiginous line ranging from 1 to 10 mm in length and ending in a tiny papule, the site of the adult mite. Other nonspecific findings are represented by papules, nodules, pustules, and scratching marks (Figs. 8.1, 8.2, 8.3, 8.4a, and 8.6a). Lesions are more frequently detected on the web space of the hand, the flexural aspects of the wrist, the axillae, the umbilicus, the belt line, the nipples, the buttocks, and the penile shaft. In children, especially infants and babies, the head, the palms, and the soles, generally spared in adults, are typically involved [4, 5]. Secondary bacterial infection commonly occurs [6].

8.5 Differential Diagnosis

It includes atopic dermatitis, neurodermatitis, papular urticaria, folliculitis, dermatitis herpetiformis, prurigo nodularis, and bites of mosquitoes, fleas, or other mites [7, 8].

8.6 Diagnosis

The standard technique for the diagnosis of scabies consists of identification of the mite, eggs, or feces by microscopic examination of scales obtained by skin scraping. This technique may be too discomforting and may cause fear, especially in younger patients. As the results generally depend

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on the scraped areas, repeated tests are sometimes necessary for a conclusive diagnosis. Moreover, scraping may be complicated with accidental infections from blood-transmissible agents such as HIV or HCV.

8.7 Dermatoscopy

Dermatoscopy at ×10 shows a typical sign of the "jet with contrail," where the jet-shaped triangular structure corresponds to the pigmented anterior part of the mite (mouthparts and two anterior pairs of legs) and the contrail-shaped segment corresponds to the burrow (Figs. 8.4b, 8.5a, 8.6b) [9–11]. Higher magnifications (>×100) allow the detection of details such as mites in migration, eggs, and feces, usually not appreciable at lower magnifications (Figs. 8.5b, 8.6c, 8.7, and 8.8) [5, 10, 11].

8.8 Histopathological Correlation

Histopathological examination of a burrow may reveal the presence of mites, larvae, eggs, and/or excrements in the superficial layers of the epidermis. Moreover, a dermal infiltrate composed of lymphocytes, histiocytes, mast cells, and eosinophils may be associated.

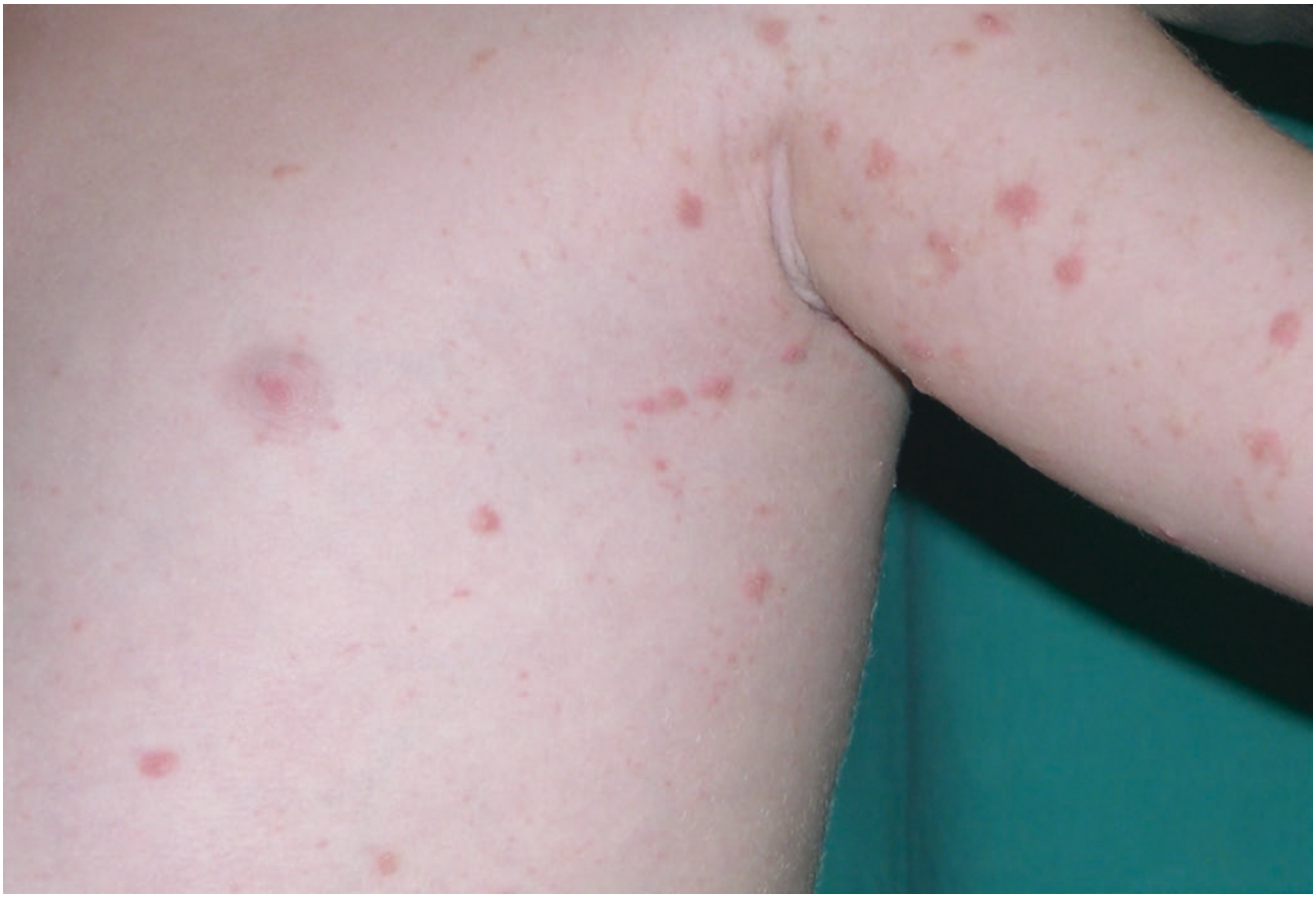

Fig. 8.1 Scabies. Multiple erythematous papules of the trunk and the upper extremity in a 5-year-old boy

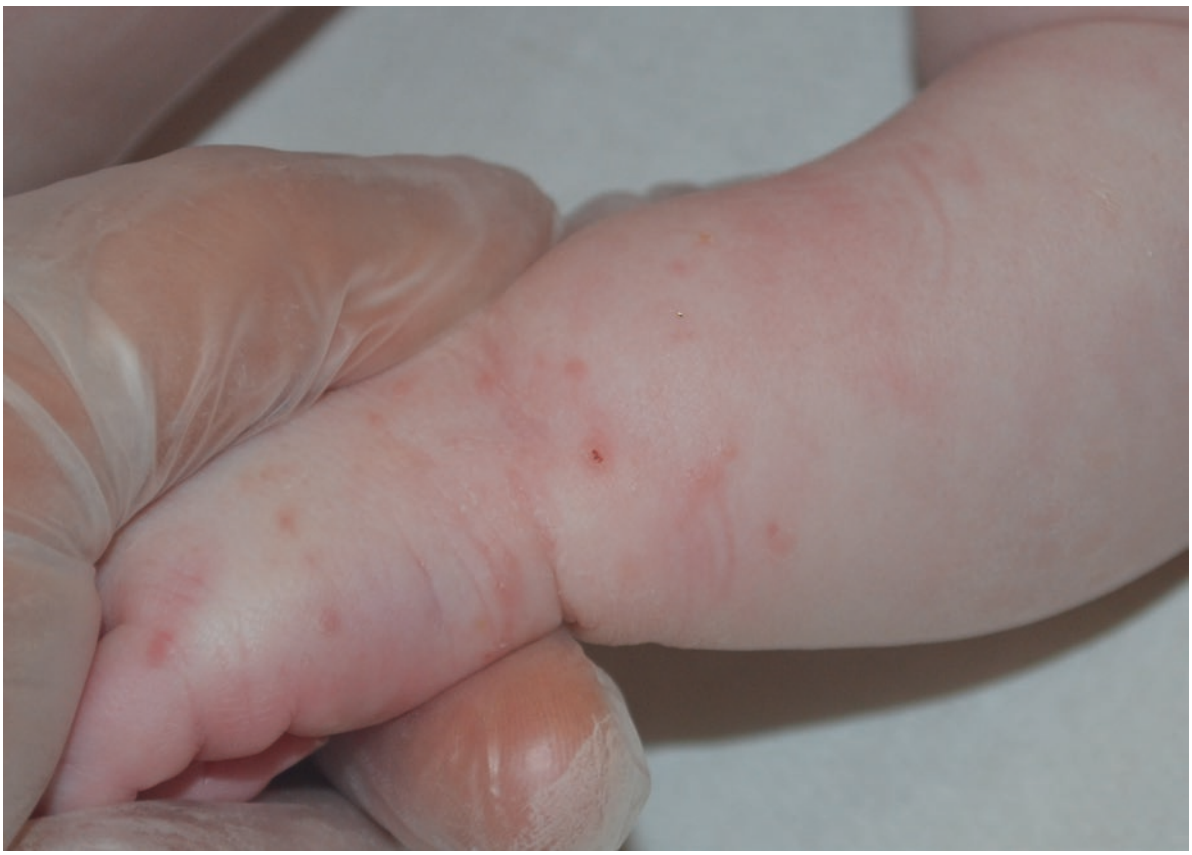

Fig. 8.2 Scabies. Multiple erythematous papules and scratching marks of the wrist in a 1-year-old boy

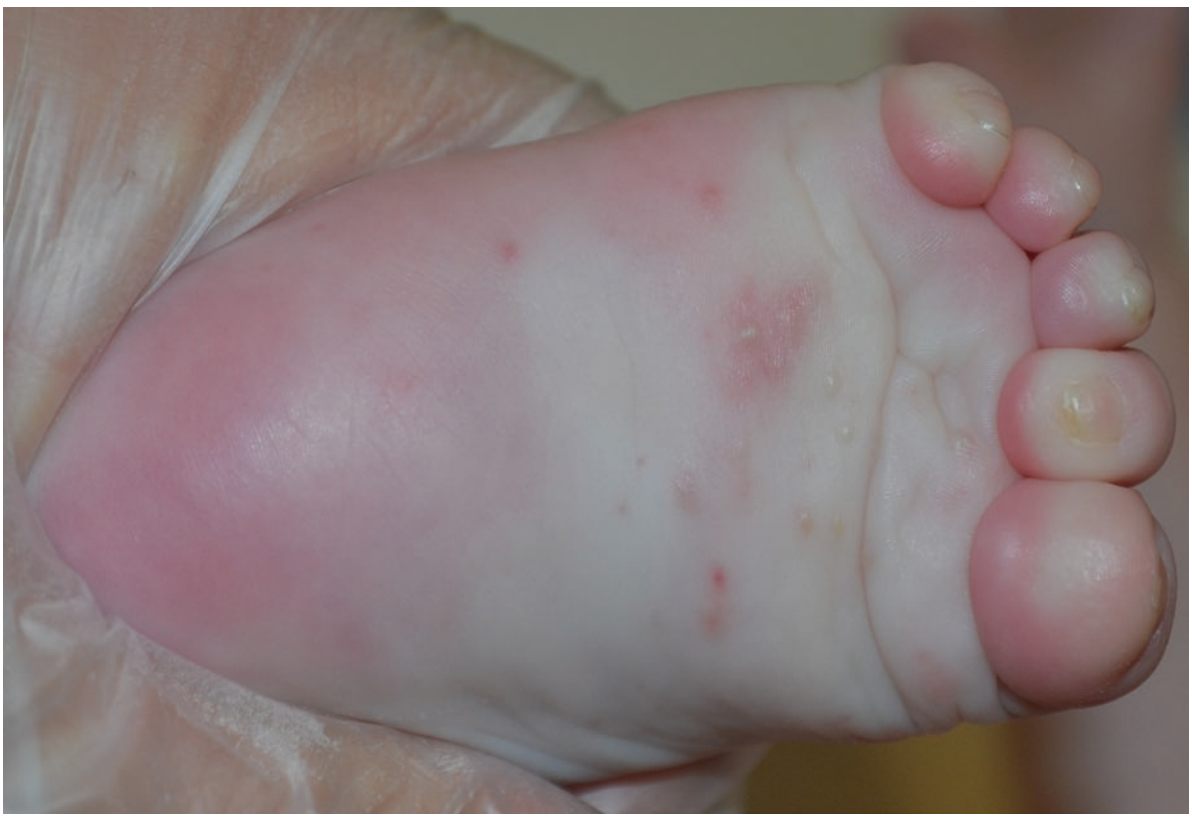

Fig. 8.3 Scabies. Multiple papulopustular lesions of the right sole in a 10-month-old girl

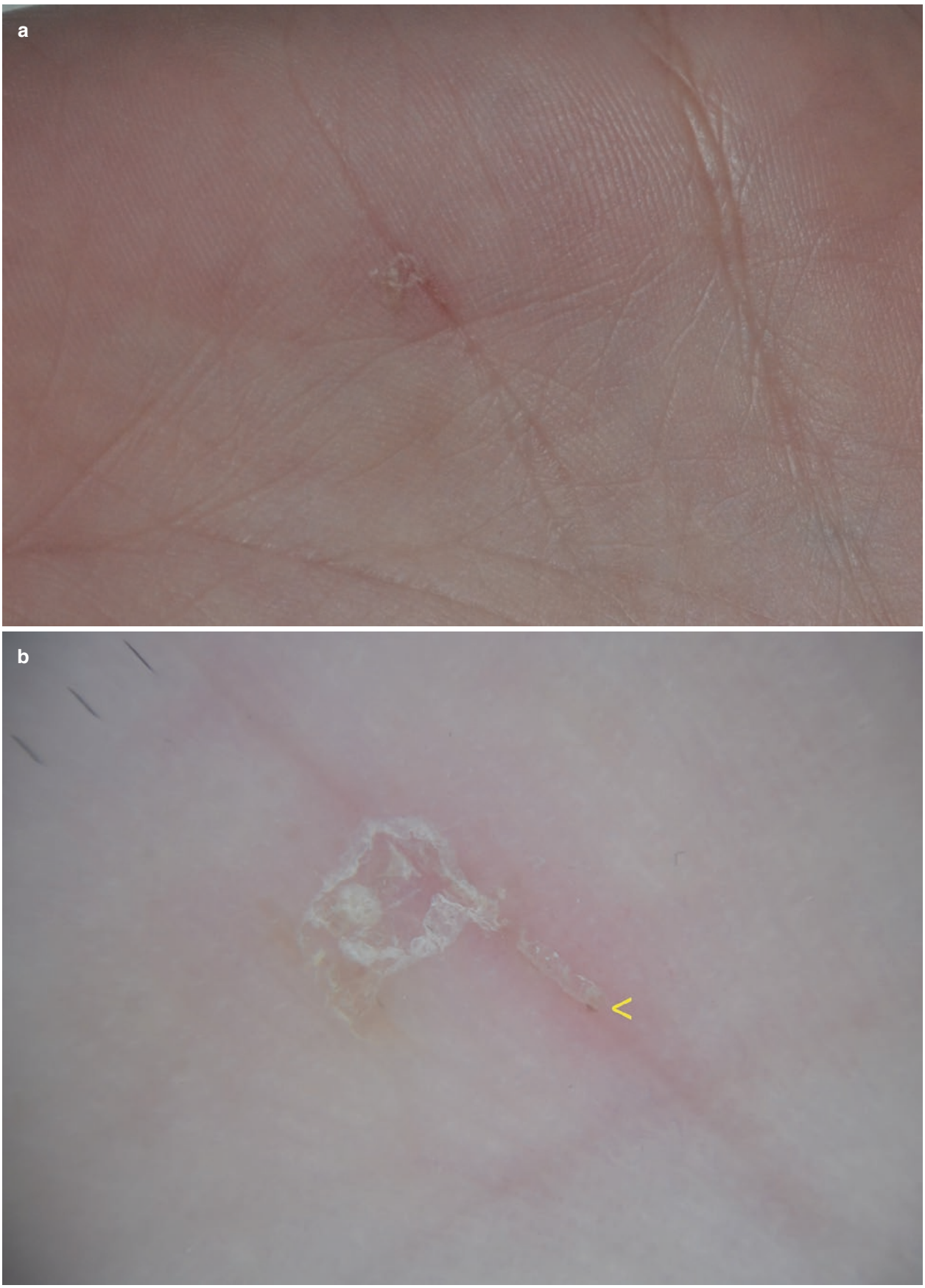

Fig. 8.4 (**a**) Scabies. Solitary scaling lesion of the palm in a 16-year-old boy with diffuse itching. (**b**) Dermatoscopy (×10): well-defined burrow with a slight pigmented extremity corresponding to the anterior part of *Sarcoptes scabiei* (arrowhead)

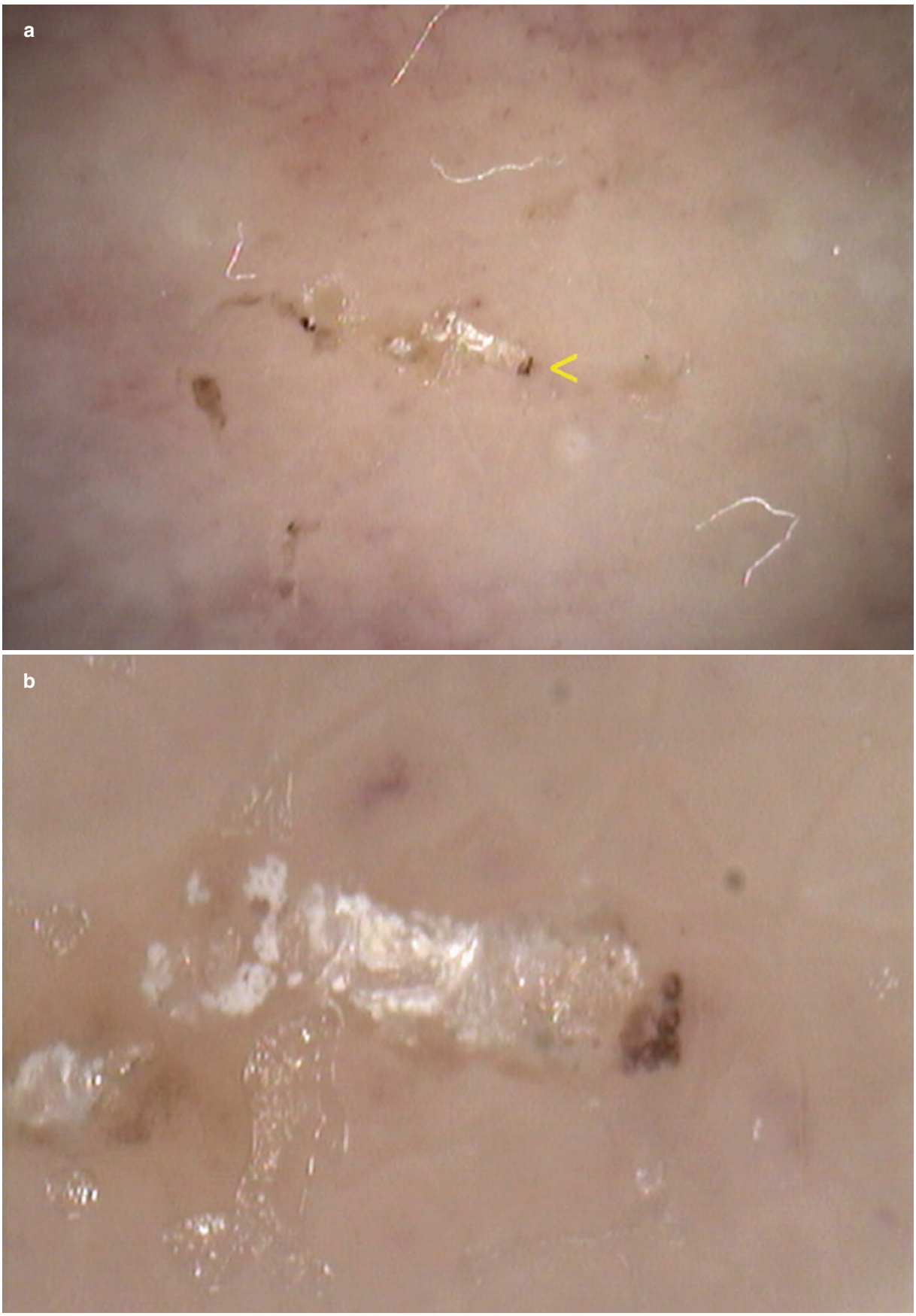

Fig. 8.5 (**a**) Scabies. Dermatoscopy (×10): brownish jet-shaped triangular structure (arrowhead) located at the end of a contrail-shaped segment, respectively, corresponding to the anterior part of the mite and the burrow (*jet with contrail*). (**b**) High magnification dermatoscopy (×200): the roundish body of the mite with its brownish area, corresponding to mouthparts and two anterior pairs of legs, is evident

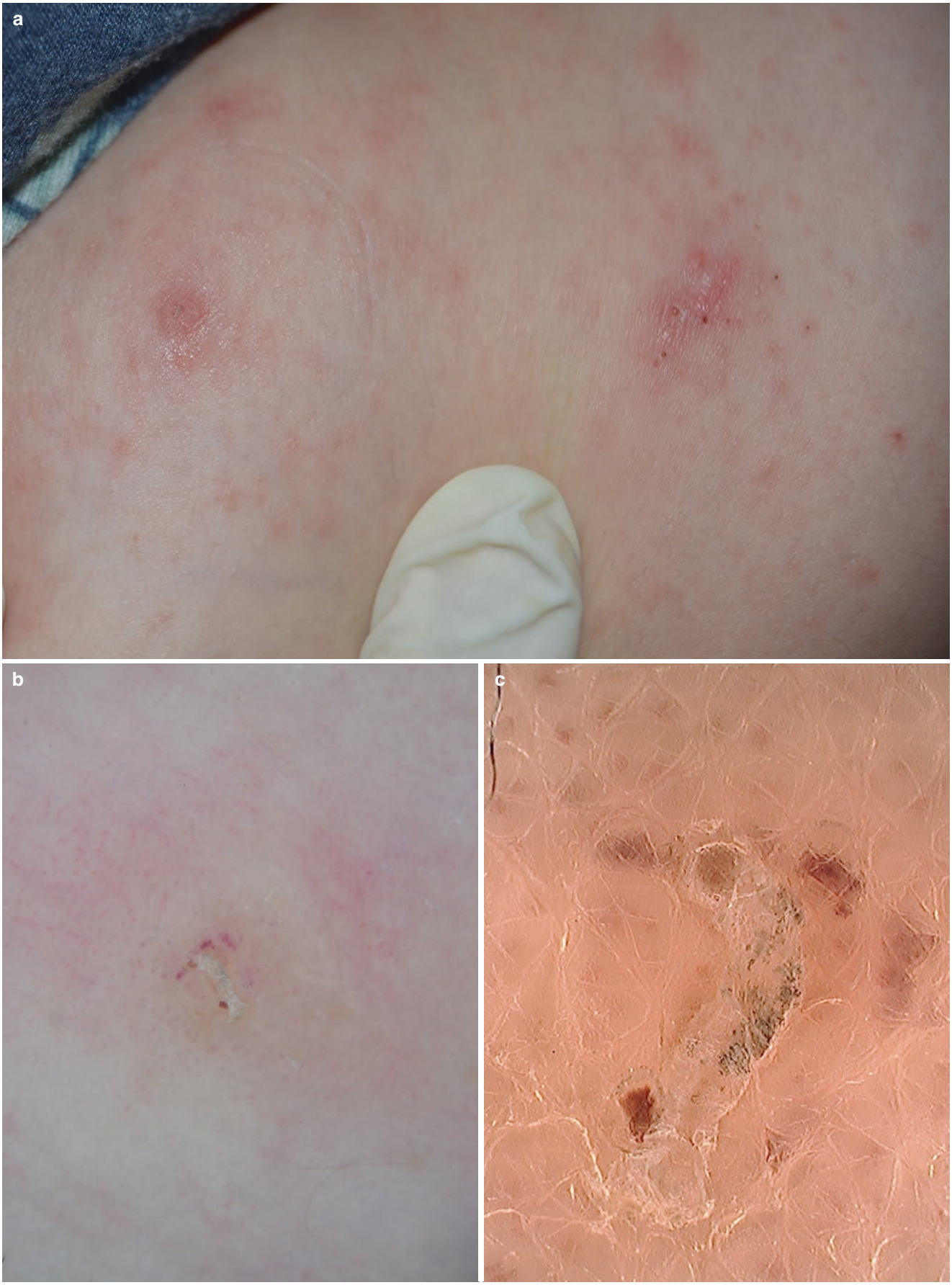

Fig. 8.6 (**a**) Scabies. Erythematous and crusted papules of the thigh in a 6-year-old girl. (**b**) Dermatoscopy (×10): contrail-shaped segment corresponding to the burrow. (**c**) At higher magnification (×200), it is evident that the burrow is empty

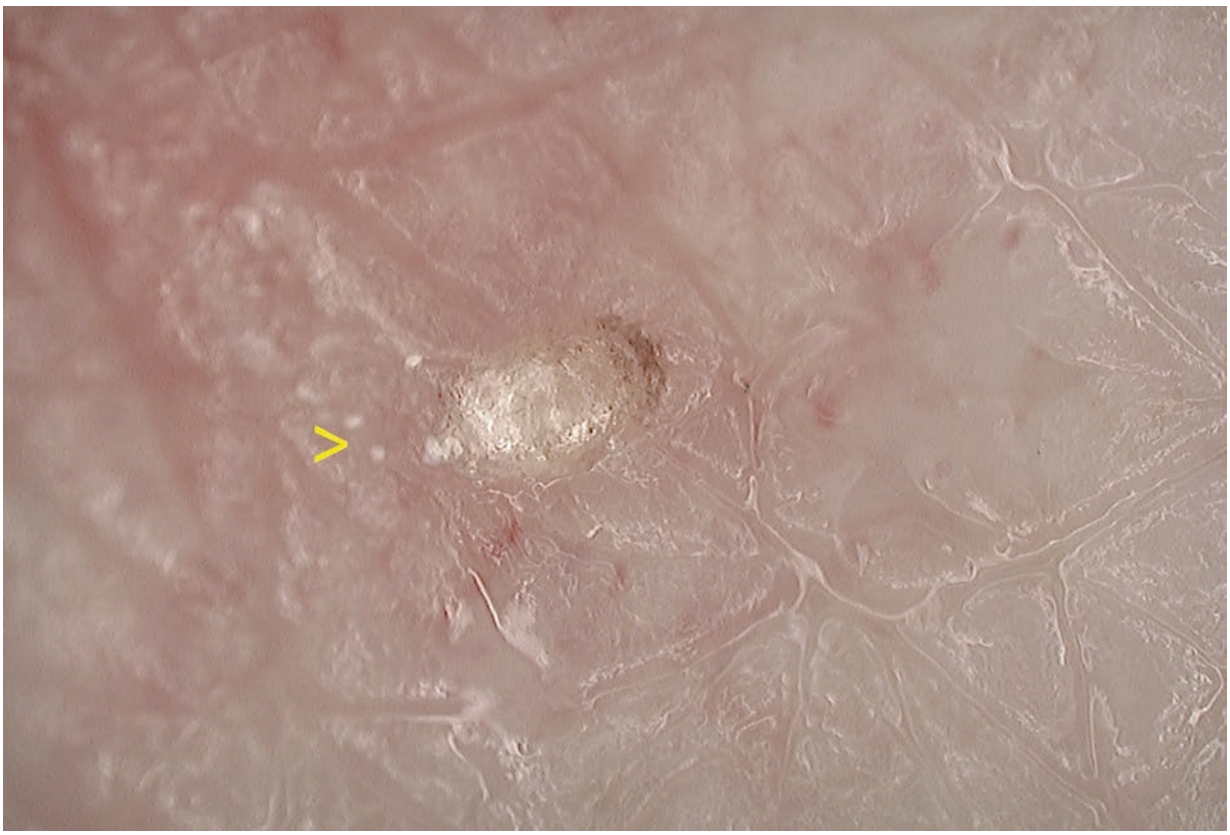

Fig. 8.7 Scabies. High magnification dermatoscopy (×400): roundish body of the mite with its triangular, brownish anterior part and small, white, roundish feces (arrowhead) within the burrow

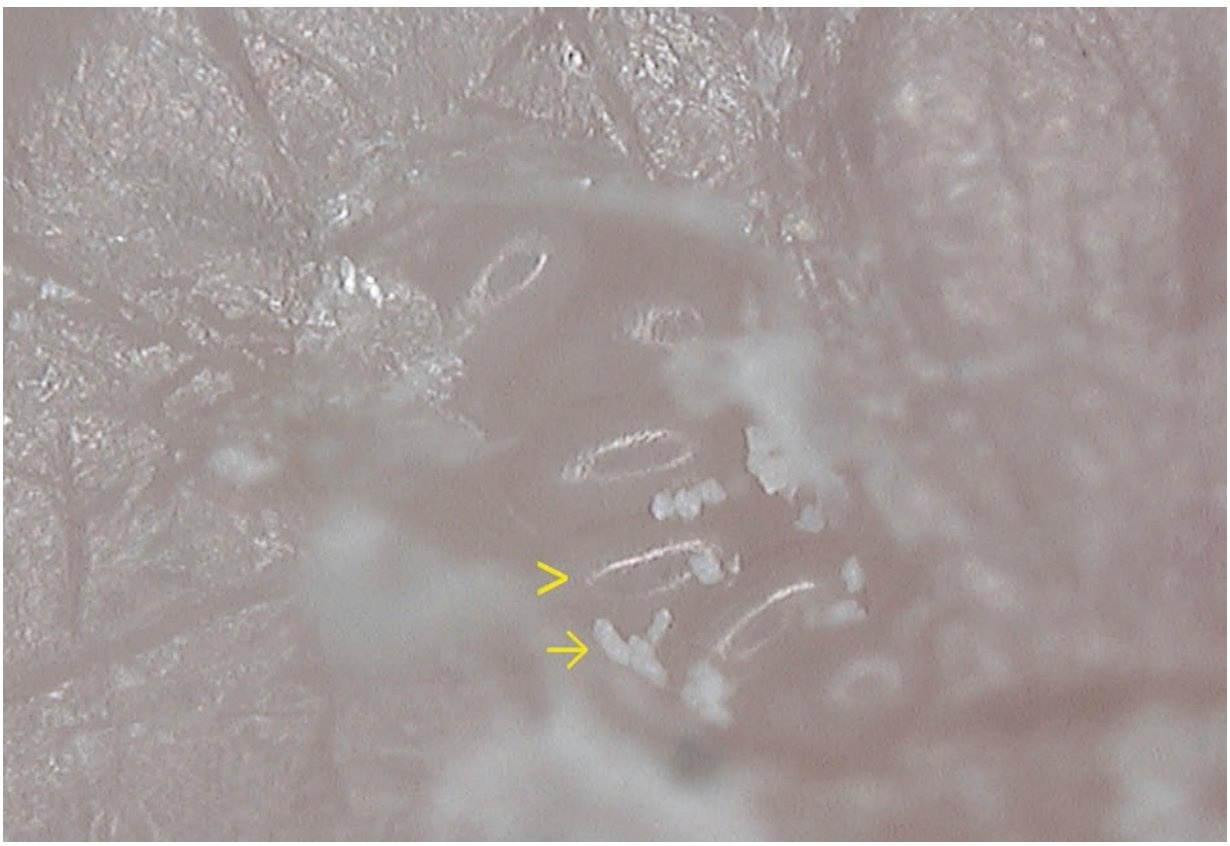

Fig. 8.8 Scabies. High magnification dermatoscopy (×500): translucent ovular eggs (arrowhead) and white roundish feces (arrow) within the burrow

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Pediculosis

9

Francesco Lacarrubba, Simona Boscaglia, Franco Dinotta, and Giuseppe Micali

9.1 Definition

Pediculosis is an infestation of lice, blood-feeding ectoparasitic insects that can occur in different species of warm-blooded animal. Head lice and pubic lice represent the most frequent forms of pediculosis affecting humans [1].

9.2 Epidemiology

The prevalence of head lice infestation is variable, ranging from 1% to 20% in different countries [2]. They have a predilection for children aged 3–12 years, with a preference for female gender, as a result of close contact, hairbrush sharing, and long hairs [3]. Low socio-sanitary conditions, promiscuity, and overcrowding are additional risk factors. For these reasons epidemics are typically observed in close communities, in particular nurseries, and in schools. Lower incidence in Afro-Americans might be explained by curly hair shafts hampering parasite grasping [1]. Pubic lice infestation is rare in children after puberty; transmission occurs by intimate contact, so it is considered a sexually transmitted disease and often gives rise to suspicion of child abuse. Less frequently the contagion is possible by clothes and towels [2] or with hand contact from the genital area [3].

9.3 Etiology

Pediculosis is caused by host-specific ectoparasites belonging to phylum Arthropoda. In particular, head lice infestation is caused by *Pediculus humanus* var. *capitis* [2], which settles on the hair and feeds sucking host blood. Transmission is often direct, less frequently through carriers such as inanimate objects [1, 2]. Pubic lice infestation, due to *Phthirus pubis*, in adults involves the low abdomen hairs generally as a result of intercourse with an infected partner. In children, due to lack of terminal hairs in other body regions, *Phthirus pubis* usually affects the margin of the scalp and the eyelashes (phthiriasis palpebrarum) [4].

9.4 Clinical Features

Pruritus, due to lice bites and triggered by saliva's hypersensitivity, is the main symptom of pediculosis, although some patients may be asymptomatic. The presence of excoriations due to scratching and superinfections is common (Figs. 9.1 and 9.2) [2, 5]. In head lice, pruritus is mainly localized in occipital and retroauricular regions and may be associated with lymphadenopathy. In phthiriasis palpebrarum, blepharitis is a common complaint.

9.5 Differential Diagnosis

In head lice, it is necessary to differentiate nits from the so-called pseudonits, conditions that mimic pediculosis, such as hair casts, debris of hair spray or gel, and scales of seborrheic dermatitis or other scaling conditions. In contrast to nits, they are freely movable along the hair shaft [1]. Phthiriasis palpebrarum should be differentiated from some inflammatory conditions causing scaling and blepharitis, including atopic and contact dermatitis and seborrheic dermatitis [6].

9.6 Diagnosis

The diagnosis of both head lice and pubic lice is generally clinical. In head lice, with the aid of a magnifying glass and a comb, it is possible to detect the lice and/or their nits adherent to hair shafts close to the follicular openings, especially in ret-

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roauricular and occipital areas (Fig. 9.3) [1, 3]. *Phthirus pubis* is easily identifiable with a magnifying glass. In phthiriasis palpebrarum, it is sometimes difficult to clinically detect the parasites because of their deep burrowing in the lid margin [4, 5].

9.7 Dermatoscopy

The identification of the adult parasite is difficult in head lice (Fig. 9.4), while pubic lice are generally easily detected (Fig. 9.5). Dermatoscopy unequivocally shows the presence of the nits fixed to the hair shaft (head lice and pubic lice) or the eyelashes (phthiriasis palpebrarum) [1, 7]. With dermatoscopy, it is possible to differentiate between vital and empty nits, providing useful information with therapeutic implications: vital, full nits appear as ovoid, brown structures with a convex extremity, whereas empty nits appear as ovoid, translucent structures with flat free endings (Figs. 9.6, 9.7, and 9.8) [1, 8]. Moreover, dermatoscopy allows to differentiate nits from pseudonits (Figs. 9.9 and 9.10) [1, 9]. In phthiriasis palpebrarum, the lice may be observed between the eyelashes (Fig. 9.11) [10].

9.8 Histopathological Correlation

No biopsy is necessary to diagnose pediculosis.

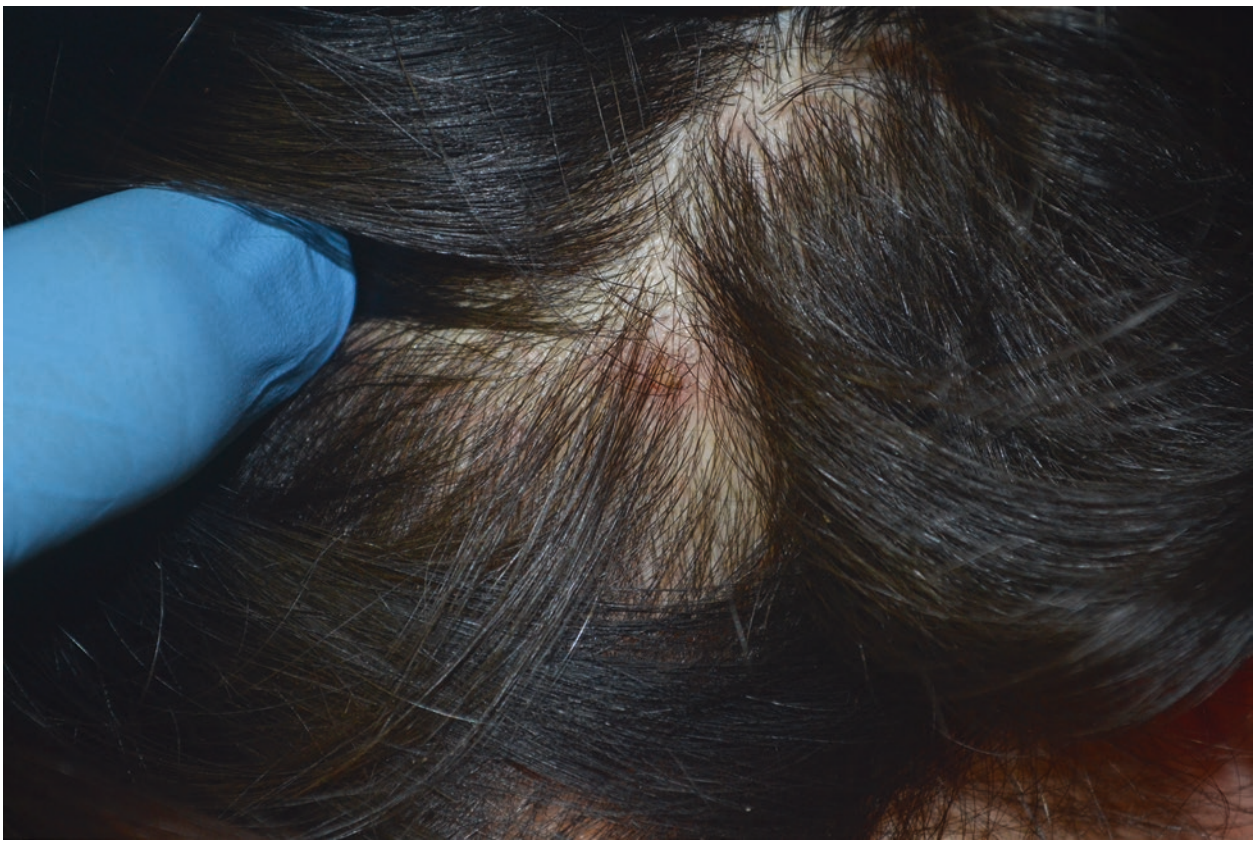

Fig. 9.1 Head lice. Scalp excoriations due to scratching in a 10-year-old girl

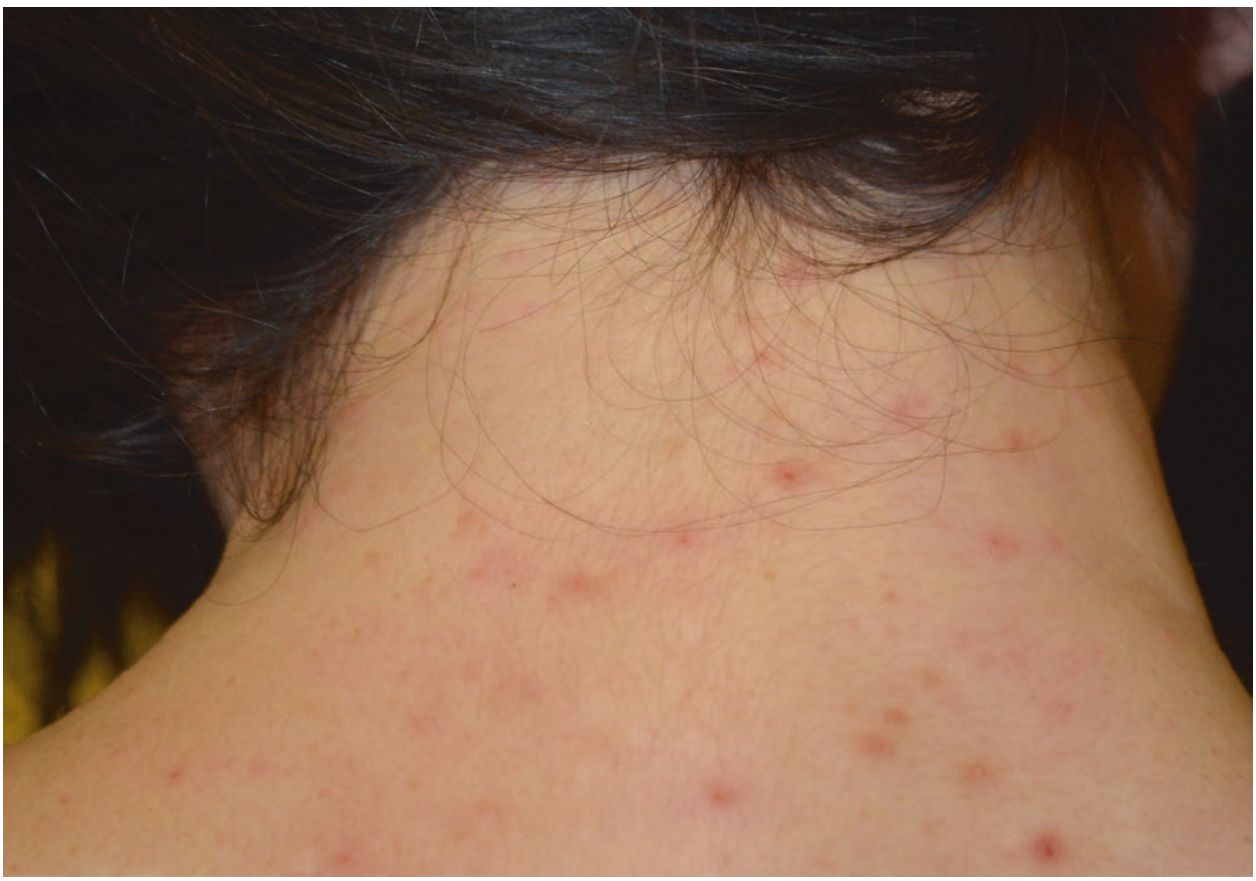

Fig. 9.2 Head lice. Pruritic eruption of the neck in a 12-year-old girl

Fig. 9.3 Head lice. Nits (arrowheads) adherent to hair shaft of the occipital area in a 10-year-old girl

Fig. 9.4 Head lice. Dermatoscopy (×20) of *Pediculus capitis* on the scalp

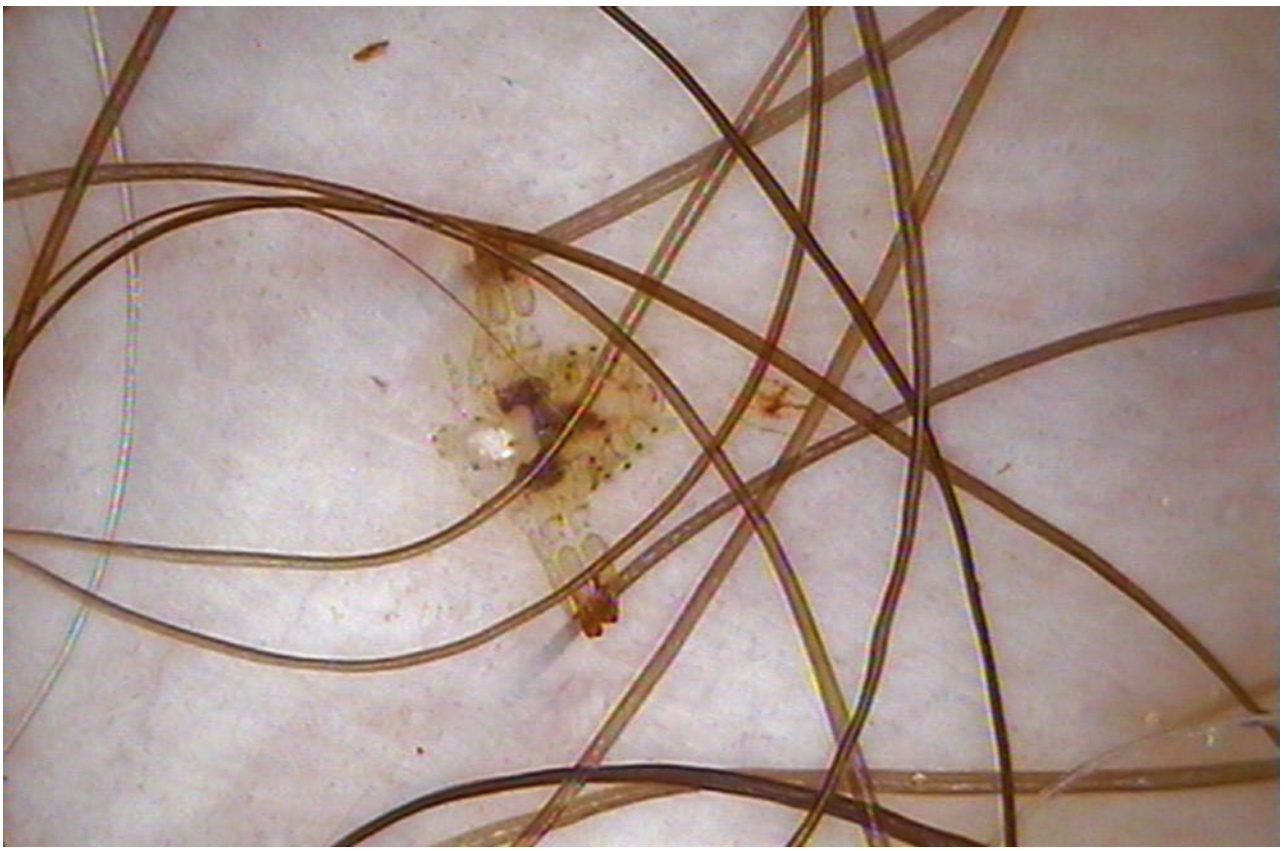

Fig. 9.5 Pubic lice. Dermatoscopy (×20) of *Phthirus pubis* on the pubic area of a 15-year-old boy

Fig. 9.6 Head lice. Dermatoscopy (×50) of vital nit fixed to the hair shaft appearing as an ovoid, brown structure with a convex extremity

Fig. 9.7 Head lice. Dermatoscopy (×50) of empty nit fixed to the hair shaft appearing as an ovoid, translucent structure with flat free ending

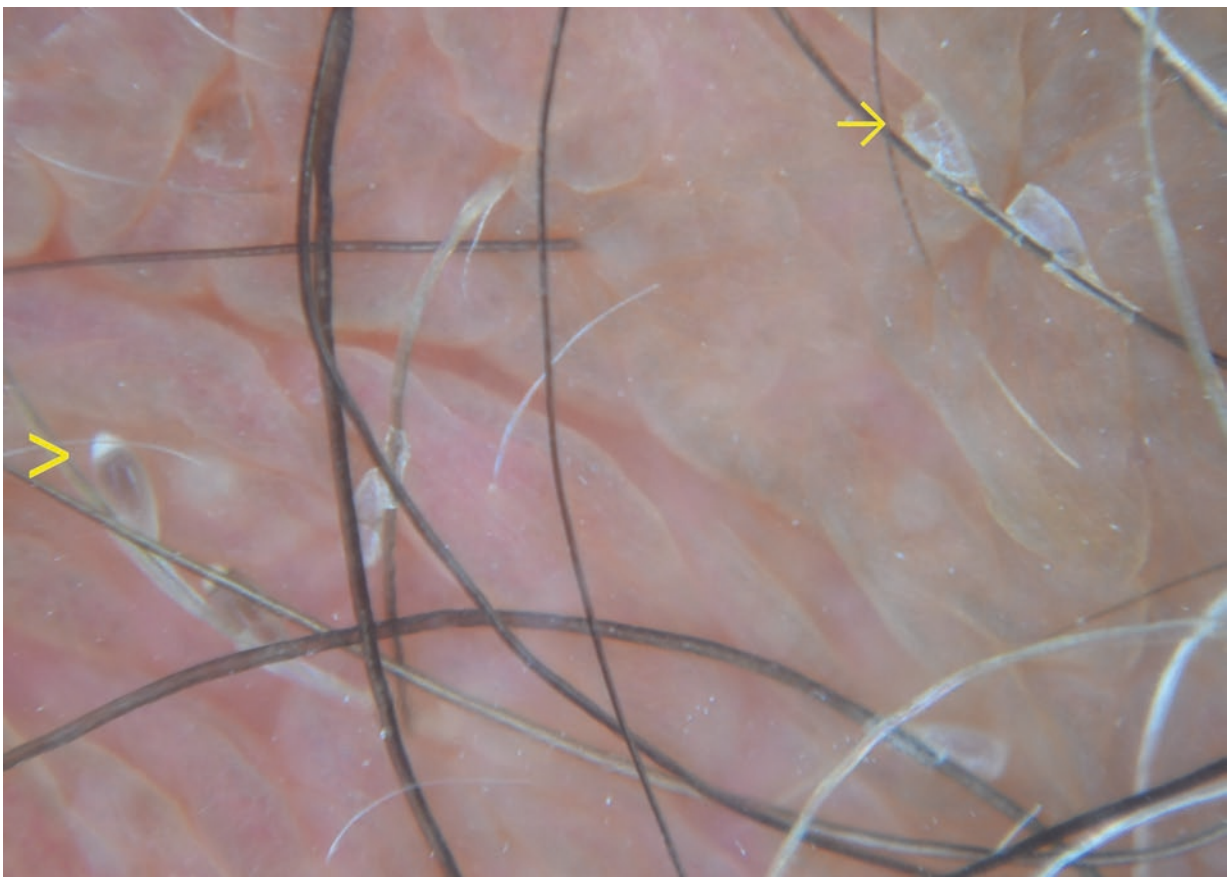

Fig. 9.8 Pubic lice. Dermatoscopy (×20) of full (arrowhead) and empty (arrow) nits on the scrotum of a 16-year-old boy

Fig. 9.9 Pseudonit. Dermatoscopy (×50) of a scale in seborrheic dermatitis: amorphous, whitish structure on the hair shaft

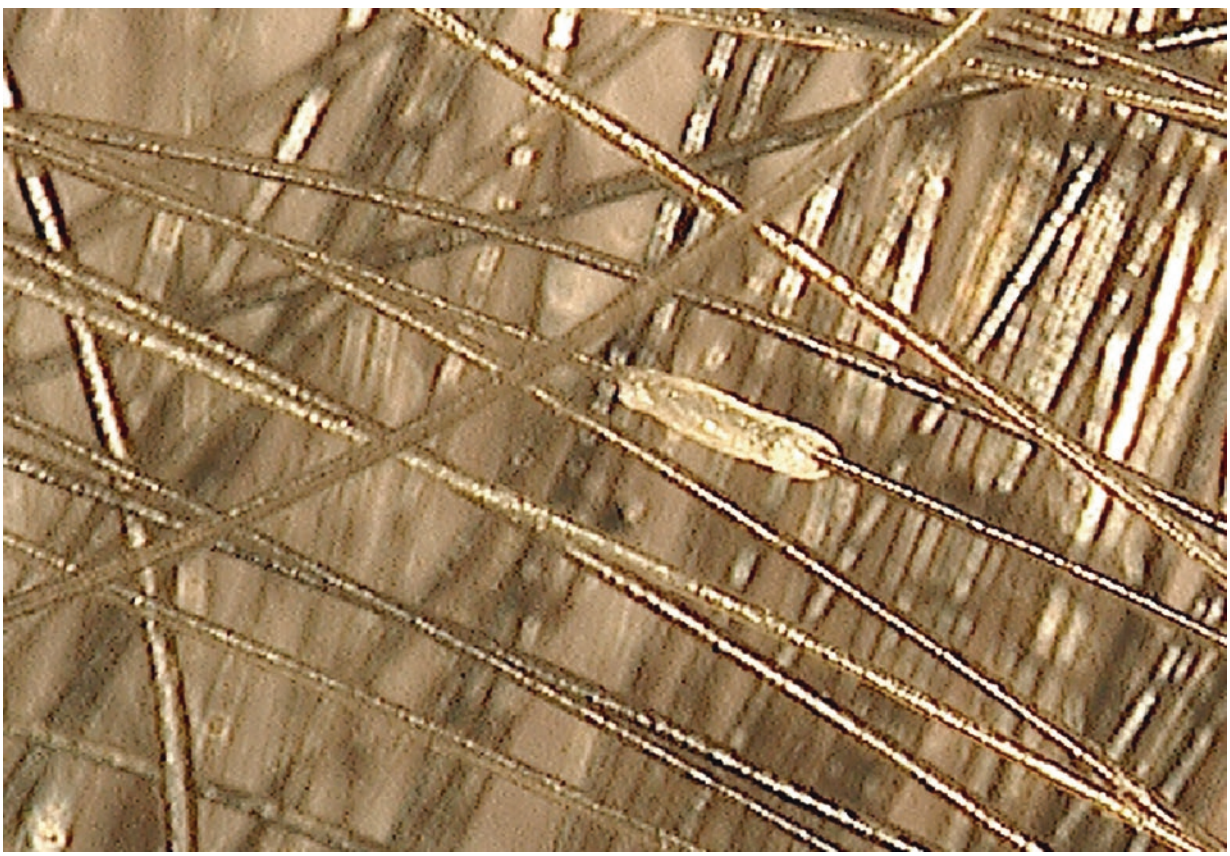

Fig. 9.10 Pseudonit. Dermatoscopy (×50) of hair cast: whitish, tubular structure around the hair shaft

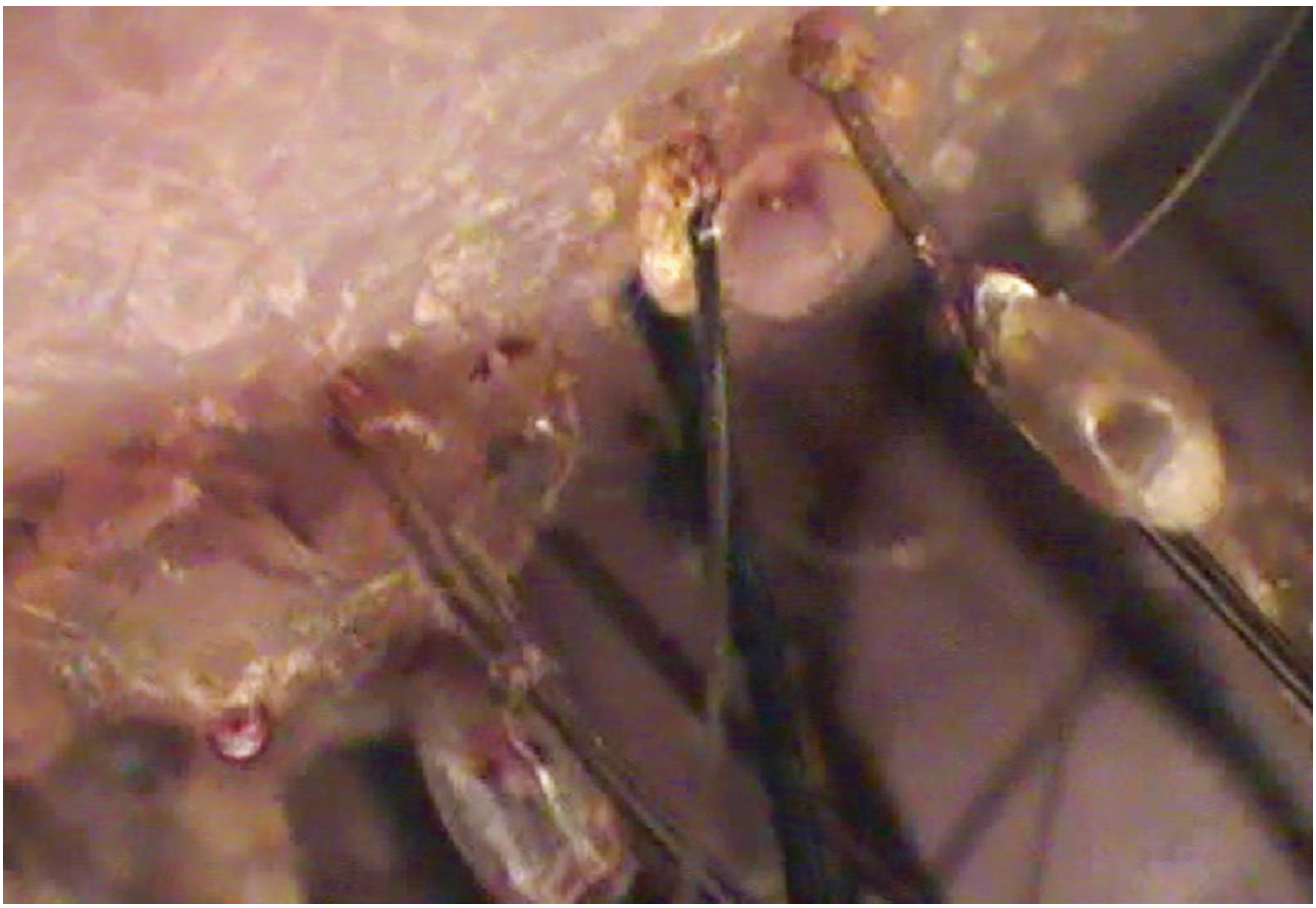

Fig. 9.11 Phthiriasis palpebrarum. Dermatoscopy (×80) of *Phthirus pubis* and full and empty nits on the eyelashes of a 6-year-old boy

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Cutaneous Leishmaniasis

10

Iria Neri

10.1 Definition

Leishmaniasis is a disease caused by protozoan parasites of *Leishmania* spp. and can be classified in three different forms: cutaneous (CL), mucocutaneous, and visceral leishmaniasis.

10.2 Epidemiology

Almost 1.5 million new cases of CL are registered each year and the global prevalence reaches 12 millions worldwide.

10.3 Etiology

In CL, the protozoa are transmitted by phlebotomine sandflies bites, commonly at nighttime. Infected dogs represent parasite reservoirs and contribute to human transmission of CL. Leishmania is an obligate intracellular parasite of macrophages, where it replicates and then spreads to the host. The incubation period is usually 1–12 weeks and the infection may run a subclinical course.

10.4 Clinical Features

CL usually involves the exposed areas and presents with an asymptomatic erythema at the site of phlebotomine bite that soon turns into an infiltrated erythematous papule. It then enlarges and becomes an indolent nodule or plaque characterized by a central ulcer/erosion and crust, with underlying keratin plugs (Figs. 10.1a, 10.2a and 10.3a). Lesions heal in a period of weeks to years leaving an atrophic scar. Satellite lesions can develop from the first lesion.

10.5 Differential Diagnosis

Differential diagnosis mainly includes a variety of skin lesions such as cutaneous tuberculosis, granulomatous foreign body reaction, cutaneous sarcoidosis, cutaneous lymphoma, pyoderma gangrenosum, keratoacanthoma, and squamous cell carcinoma.

10.6 Diagnosis

It requires the identification of typical Leishman-Donovan bodies in Giemsa-stained skin smears or, in challenging cases, its detection by microbiological cultures or PCR. In some cases, skin biopsy may be needed.

10.7 Dermatoscopy

Dermatoscopy can be useful for a noninvasive diagnosis of CL [1–3]. The most common finding is a diffuse erythema associated with yellowish-orange areas (Figs. 10.1b, c, 10.2b and 10.3b), a pattern that may be observed in other granulomatous lesions (foreign body reaction, lupus

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vulgaris, and sarcoidosis) [4]. In initial CL, yellow tear-like structures and polymorphous vessels (hairpin-like, arborizing, corkscrew, comma-shaped, linear irregular, dotted vessels, glomerular-like) arranged in a radial pattern are generally detected. Advanced lesions display hyperkeratosis and white peripheral projections resembling a starburst-like pattern around a central ulcer/erosion or crust [1]. Finally, dermatoscopy may also be useful for treatment monitoring of CL. In case of treatment response, dermatoscopy shows the presence of orange-brown structureless areas and tree-like vessels (Fig. 10.2c and 10.3d).

10.8 Histopathological Correlation

The orange-brown areas correspond to inflammatory mixed infiltrates of lymphocytes, plasma cells and histiocytes, small epithelioid granulomas, and multinucleated giant cells (Fig. 10.4) [1].

The yellowish-orange tear-like structures present in the early lesions correlate to keratin-filled follicular plugs caused by obstruction due to the growing lesion. In old lesions, the white starburst-like pattern corresponds to parakeratotic hyperkeratosis due to the central ulcer/erosion healing process [1].

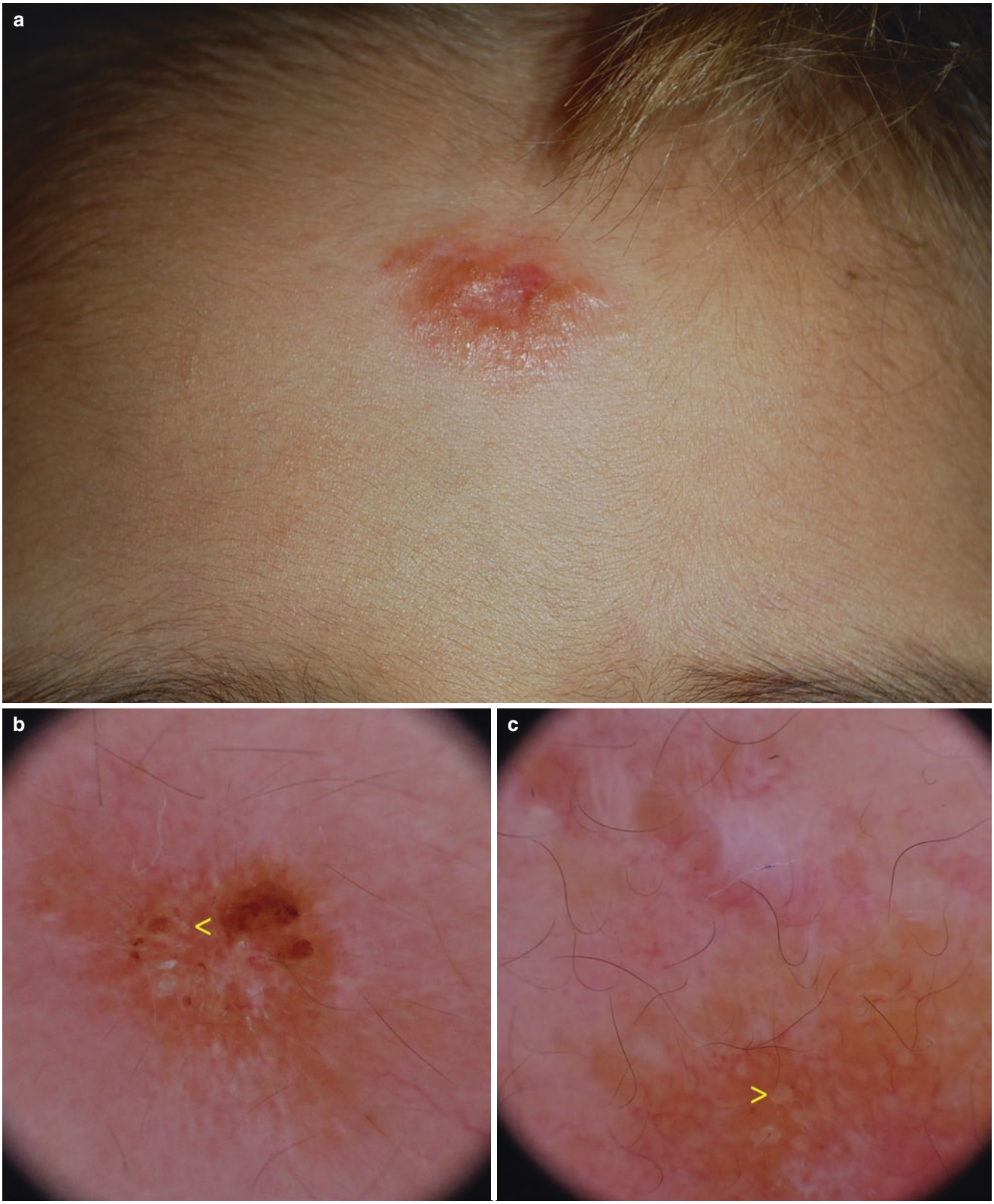

Fig. 10.1 (**a**) Cutaneous leishmaniasis. Erythematous nodule of the forehead. (**b**) Dermatoscopy (×10): generalized erythema associated with yellowish-orange areas and *tear-like* structures (arrowhead). (**c**) Dermatoscopy (×10) of another area of the lesion: yellow *tear-like* structures (arrowhead) displayed on an orange-brown background

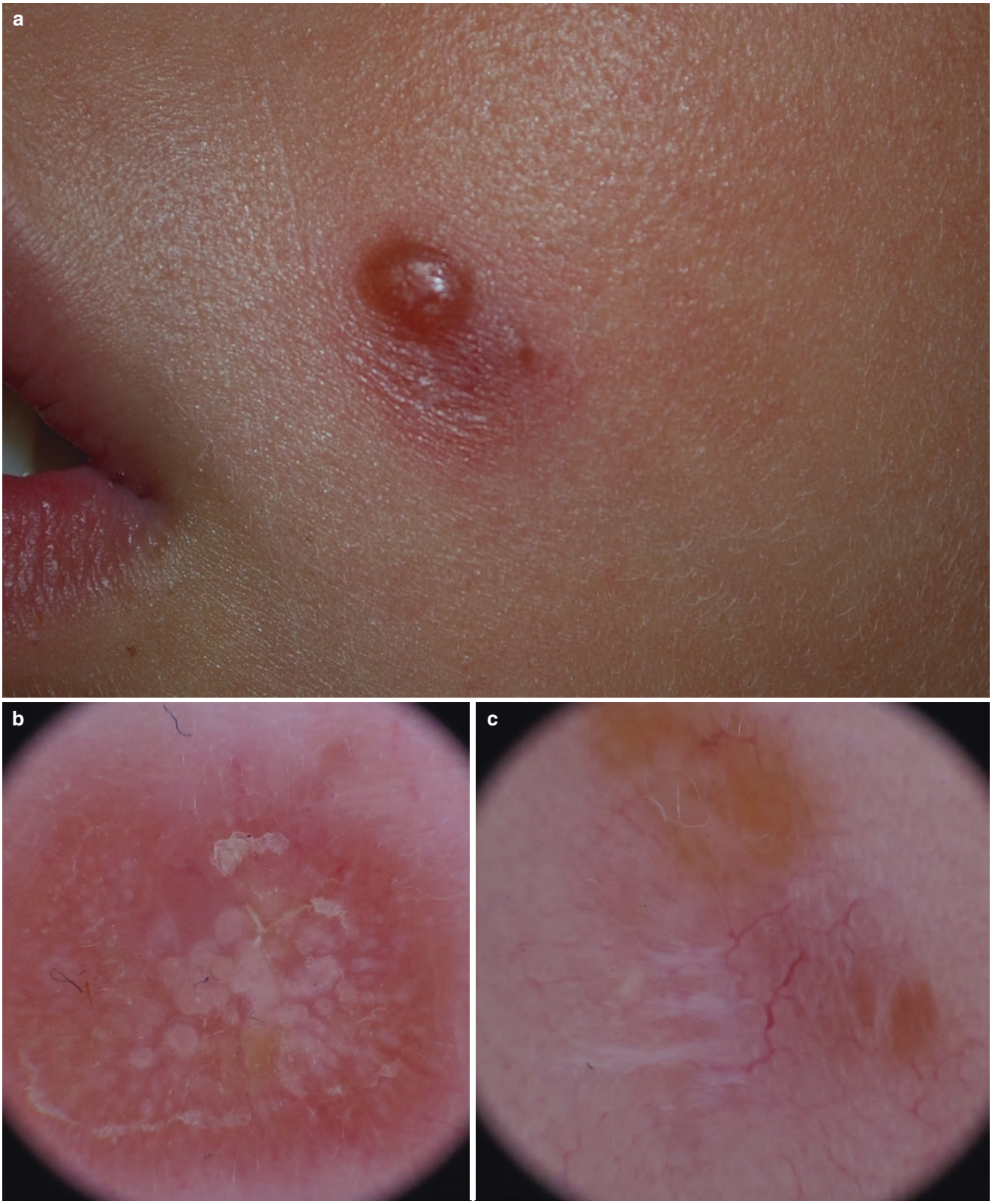

Fig. 10.2 (**a**) Cutaneous leishmaniasis. Erythematous nodule of the cheek. (**b**) Dermatoscopy (×10): *tear-like* structures on an orange-brown background. (**c**) Dermatoscopy (×10) of the lesion after treatment with paromomycin 15% ointment: brown structureless areas and treelike vessels

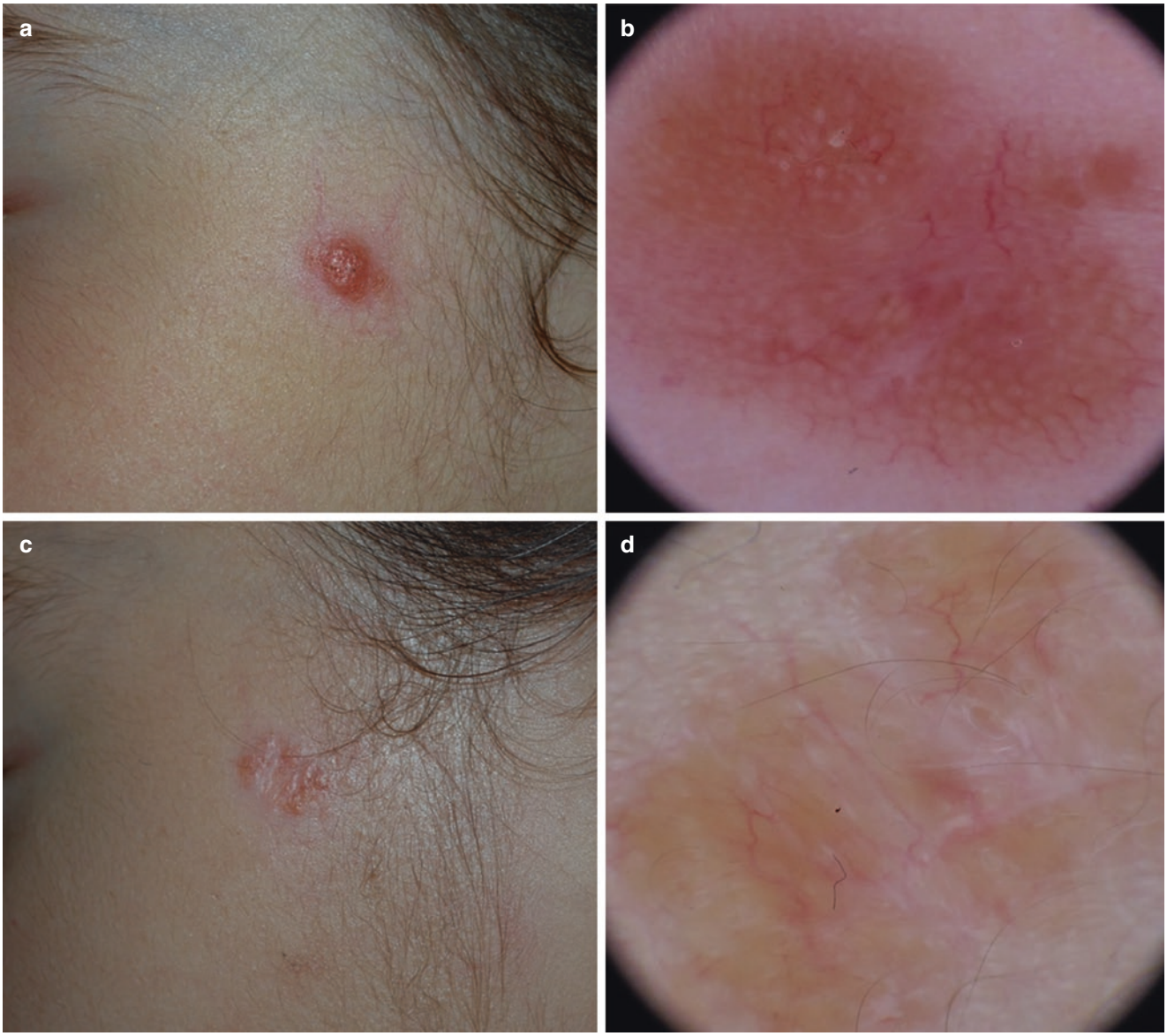

Fig. 10.3 (**a**) Cutaneous leishmaniasis in a 5-year old girl: red nodule of the left cheek. (**b**) Dermatoscopy (×10): yellow *tear-like* structures and treelike vessels are detectable within a red-orange background. (**c**) Clinical regression almost obtained after 1 month of treatment with paromomycin 15% ointment. (**d**) Dermatoscopy (×10) after treatment with paromomycin: structureless areas with an orange-brownish hue with treelike vessels are present in the absence of tear-like structures

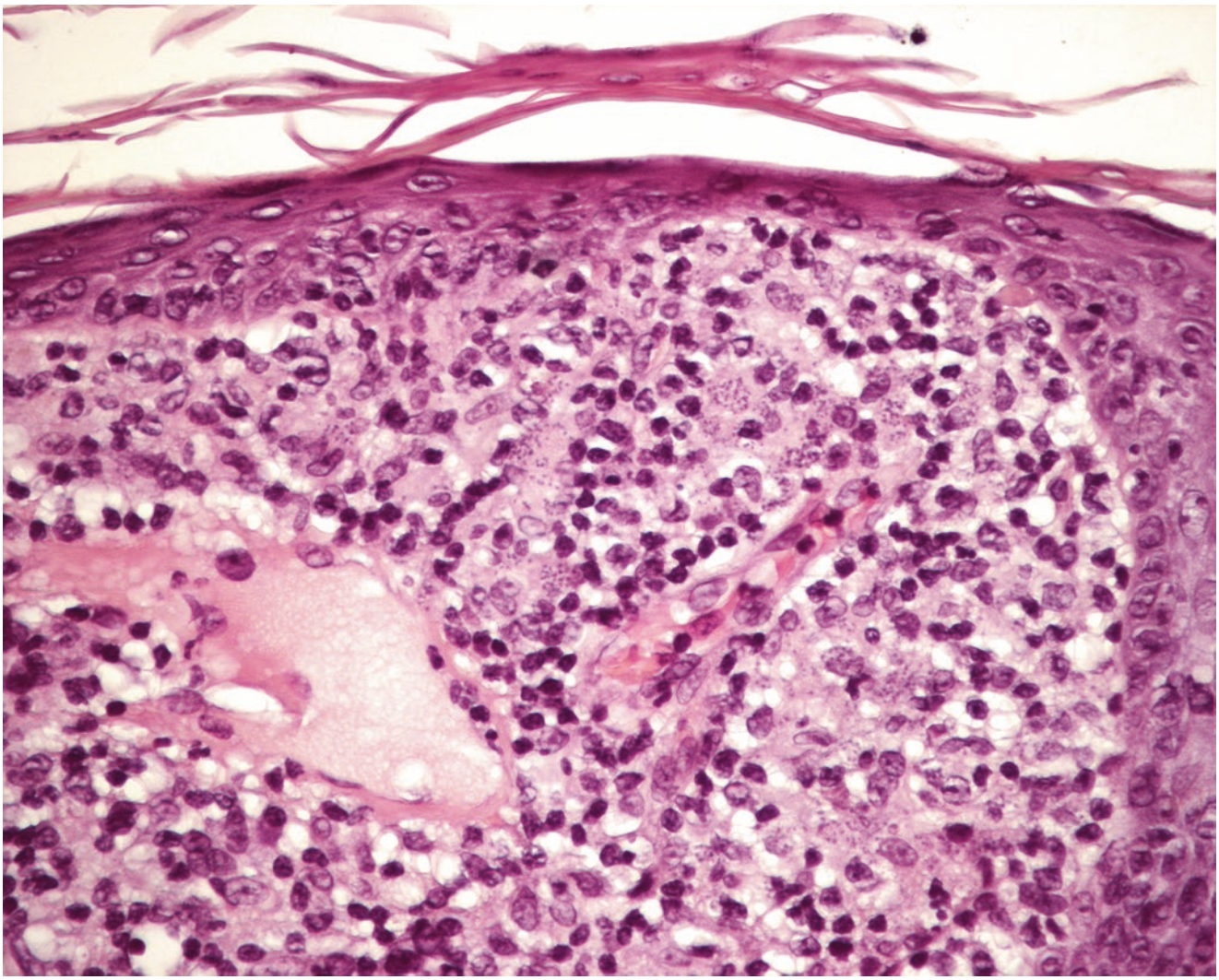

Fig. 10.4 Cutaneous leishmaniasis. Histopathology showing dense granulomatous infiltrate in the superficial dermis consisting of epithelioid histiocytes, lymphocytes, and plasma cells. Amastigotes of Leishmania are detectable within macrophages cytoplasm [H&E staining; magnification: x200]

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Part IV

Inflammatory Skin Disorders

Psoriasis

11

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Maria Letizia Musumeci, and Francesco Lacarrubba

11.1 Definition

Psoriasis is a common chronic, inflammatory, erythematous-desquamative skin disorder with a relapsing-remitting course that can involve any part of the body.

11.2 Epidemiology

In the worldwide population, the estimated prevalence is 2–4% [1]. In childhood, the prevalence is approximately between 0.7 and 2%, with a linear progressive increase with age [2]. The mean age at the diagnosis in children is 11 years, with a slight female predominance [3].

11.3 Etiology

Psoriasis has a complex multifactorial etiopathogenesis, not yet completely clear, in which both genetic and extrinsic factors contribute to activate an immunological reaction. A major role is played by T cells, stimulated by external triggers such as infections, traumas, or stress, that activate and maintain an inflammatory reaction through the production and release of many cytokines (TNF-alfa, IL-17, IL-23) [4]. In childhood, possible trigger factors are represented by viral and bacterial infections or hormonal abnormalities (obesity) [5].

11.4 Clinical Features

In childhood, the most frequent variety is represented by plaque psoriasis in which the classic lesions appear as well-demarcated erythematous plaques topped by white scales with a preferential localization on the extensor aspects of limbs, the trunk and the scalp (Figs. 11.1a and 11.2a). Generally, these lesions are thinner and smaller compared to adults. A clinical variant frequently observed in children is inverse psoriasis, localized in flexural areas with a symmetrical distribution, in which scaling is absent (Fig. 11.3a). In babies, psoriasis involving the inguinal folds can simulate a diaper dermatitis. Another clinical variant more frequent in the pediatric age is guttate psoriasis, characterized by small, roundish diffuse lesions, preferentially located on the trunk and often related to upper respiratory tract infections, typically streptococcal (Fig. 11.4a) [6].

11.5 Differential Diagnosis

Differential diagnosis in children mainly includes atopic dermatitis, psoriasiform drug eruption, pityriasis rubra pilaris, pityriasis rosea, fungal infections, and seborrheic dermatitis [7].

11.6 Diagnosis

The diagnosis of psoriasis is mainly based on clinical features. In doubtful cases, histopathological examination may be required. Dermatoscopy and reflectance confocal microscopy, may provide useful clues helpful for a noninvasive diagnosis [8].

11.7 Dermatoscopy

Dermatoscopy in psoriasis may be useful for both diagnosis and treatment monitoring, especially in children, in which any rapid and noninvasive tool is particularly well-accepted [8]. Dermatoscopy shows a typical vascular pattern characterized

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by the presence, at ×10, of "dotted" or "pinpoint" capillaries (Figs. 11.1b, 11.2b, and 11.3b) corresponding to dilated, elongated, and convoluted capillaries with a typical glomerular or "bushy" aspect at higher magnifications (>×100) (Figs. 11.1c, 11.2c, 11.3c, and 11.4b). Using digital systems, it is possible to estimate the diameter of each "bush" (50–110 μm), whose size is significantly increased compared to that of normal skin (15–25 μm). This is particularly useful for evaluating treatment response [9]. At the edge of the plaques, elongated capillary loops with "hairpin" aspect and arranged in a parallel fashion to the cutaneous surface may be detected. A limiting factor is the presence of scaling which may significantly interfere with the recognition of the vascular structures.

11.8 Histopathological Correlation

Histopathologically, dotted and bushy capillaries observed, respectively, at low and high magnification dermatoscopy, correspond to a typical aspect observed in all stages of psoriasis, i.e., dilated capillaries within elongated dermal papillae (Fig. 11.5) [6].

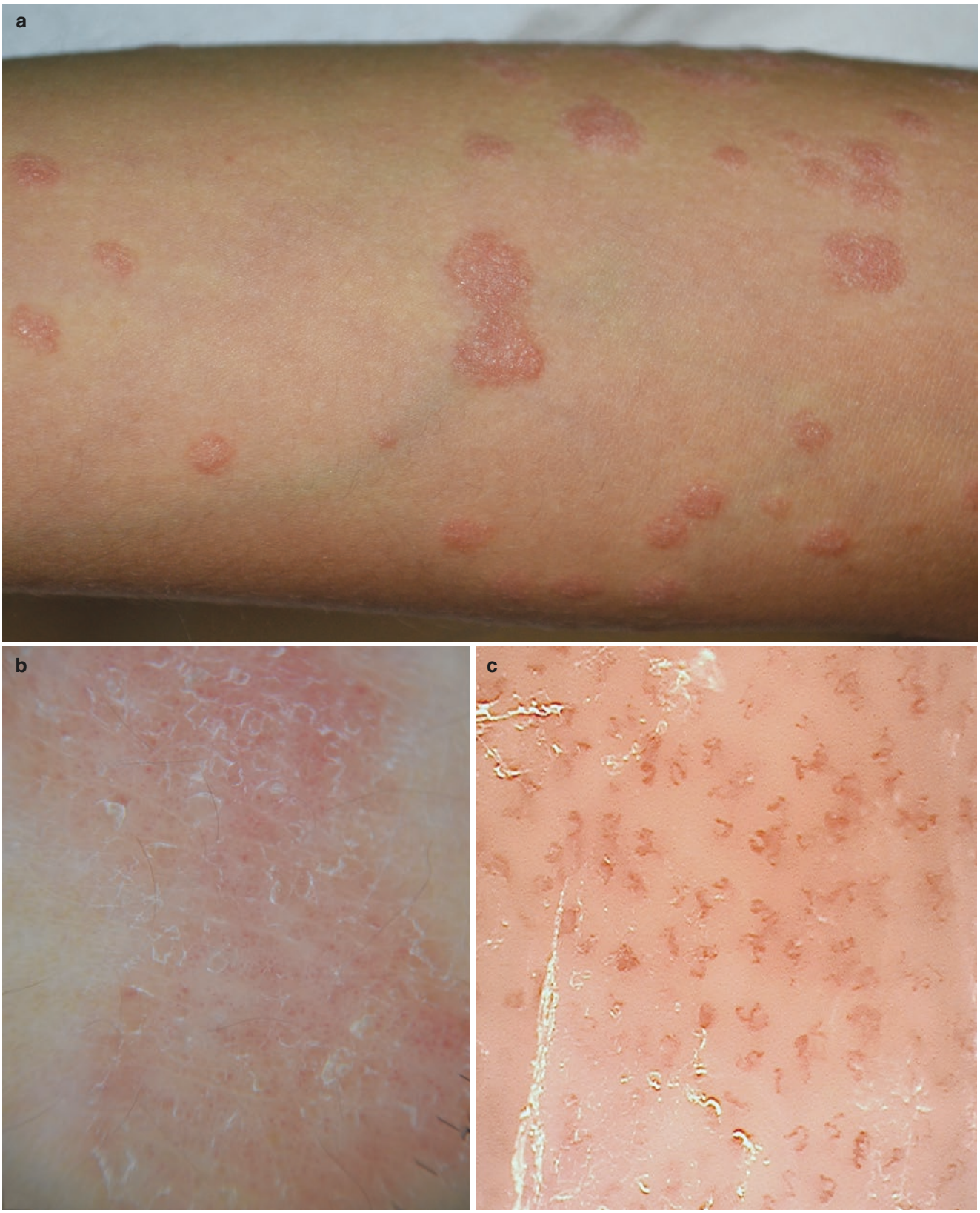

Fig. 11.1 (**a**) Psoriasis. Multiple, well-demarcated erythematous-desquamative plaques of the leg in an 11-year-old boy. (**b**) Dermatoscopy (×10): dotted capillaries associated to whitish scales. (**c**) High-magnification dermatoscopy (×100): dilated, elongated, and convoluted vessels (*glomerular* or *bushy* aspect)

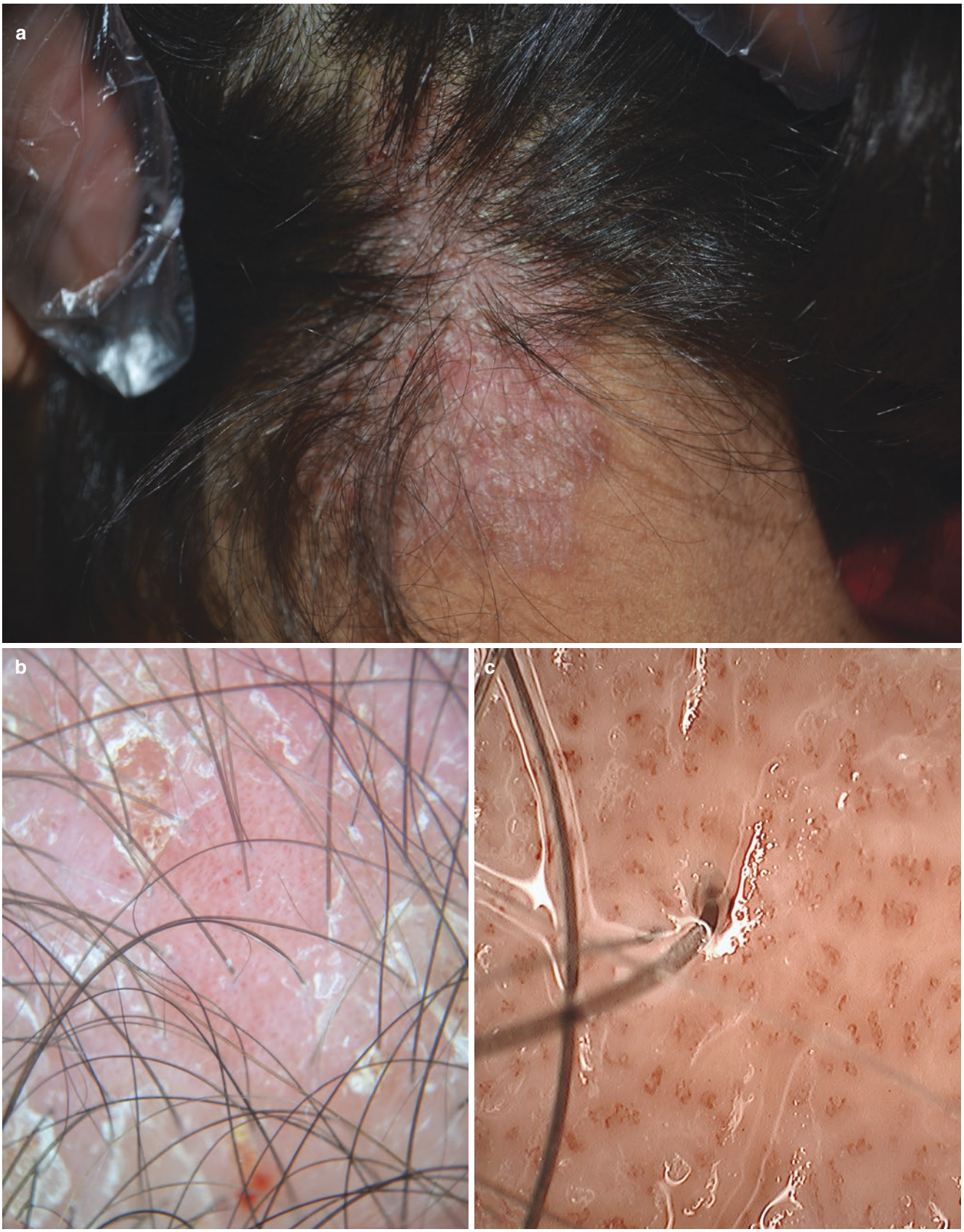

Fig. 11.2 (**a**) Scalp psoriasis. Large erythematous-desquamative plaque involving the occipital area and the neck in a 12-year-old girl. (**b**) Dermatoscopy (×10): dotted capillaries, scales, and crusting due to scratching. (**c**) High-magnification dermatoscopy (×100) performed in a non-scaling area: dilated, elongated, and convoluted vessels (*glomerular* or *bushy* aspect)

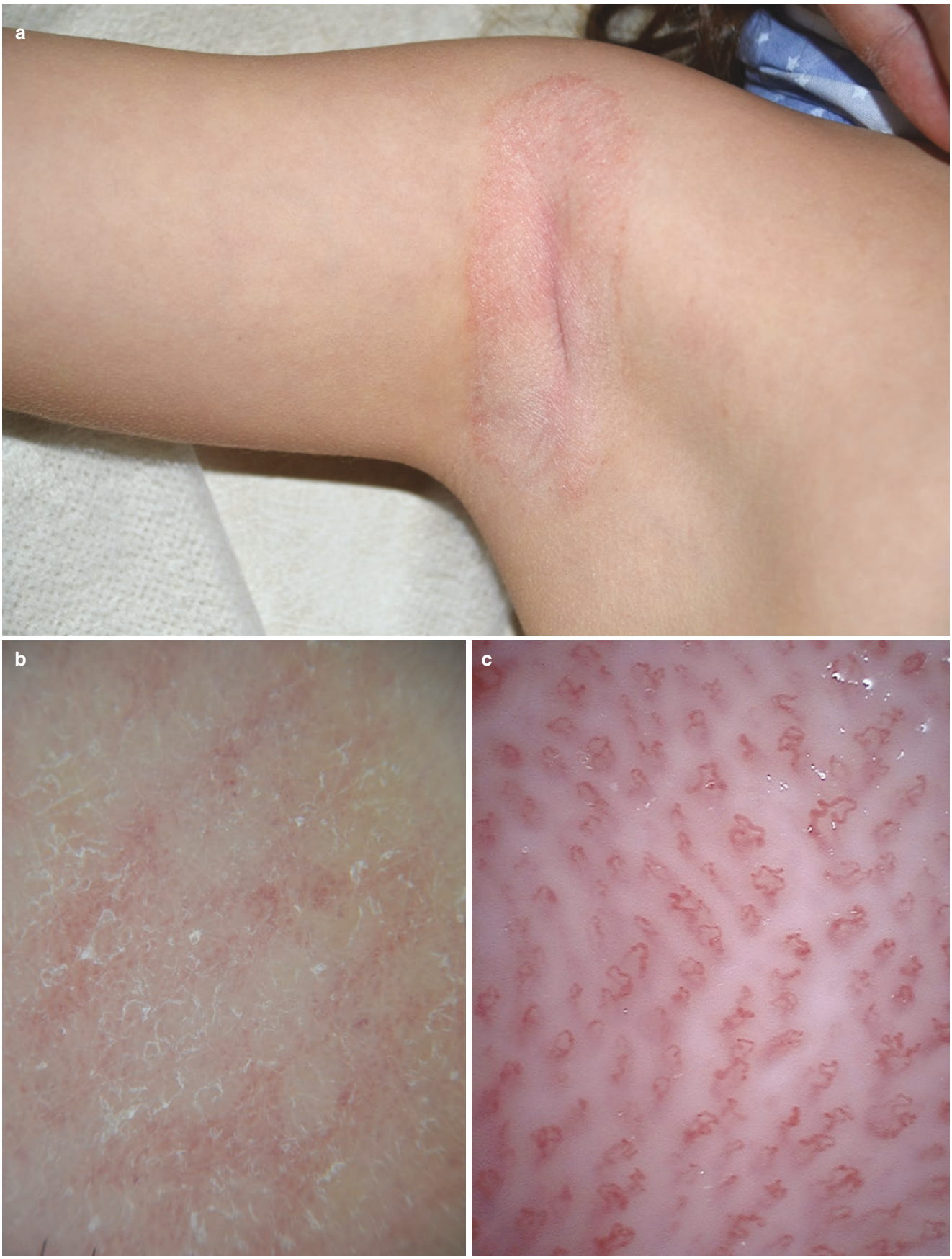

Fig. 11.3 (**a**) Inverse psoriasis. Large erythematous plaque involving the axillary area in a 7-year-old girl. (**b**) Dermatoscopy (×10): dotted capillaries and sparse scaling. (**c**) High-magnification dermatoscopy (×150): dilated, elongated, and convoluted vessels (*glomerular* or *bushy* aspect)

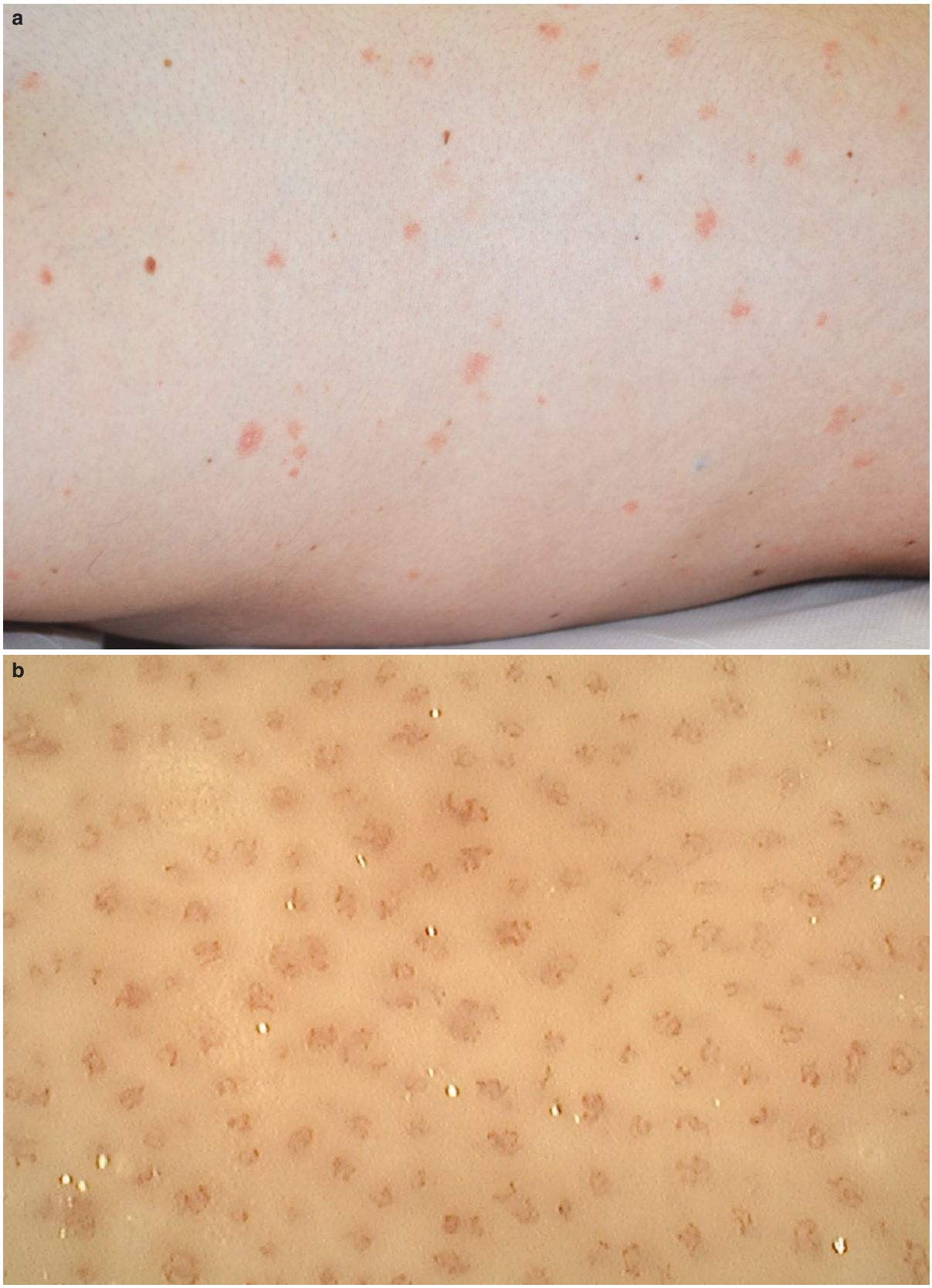

Fig. 11.4 (**a**) Guttate psoriasis. Small, roundish erythematous lesions on the trunk in a 15-year-old boy. (**b**) High-magnification dermatoscopy (×150): dilated, elongated, and convoluted vessels (*glomerular* or *bushy* aspect)

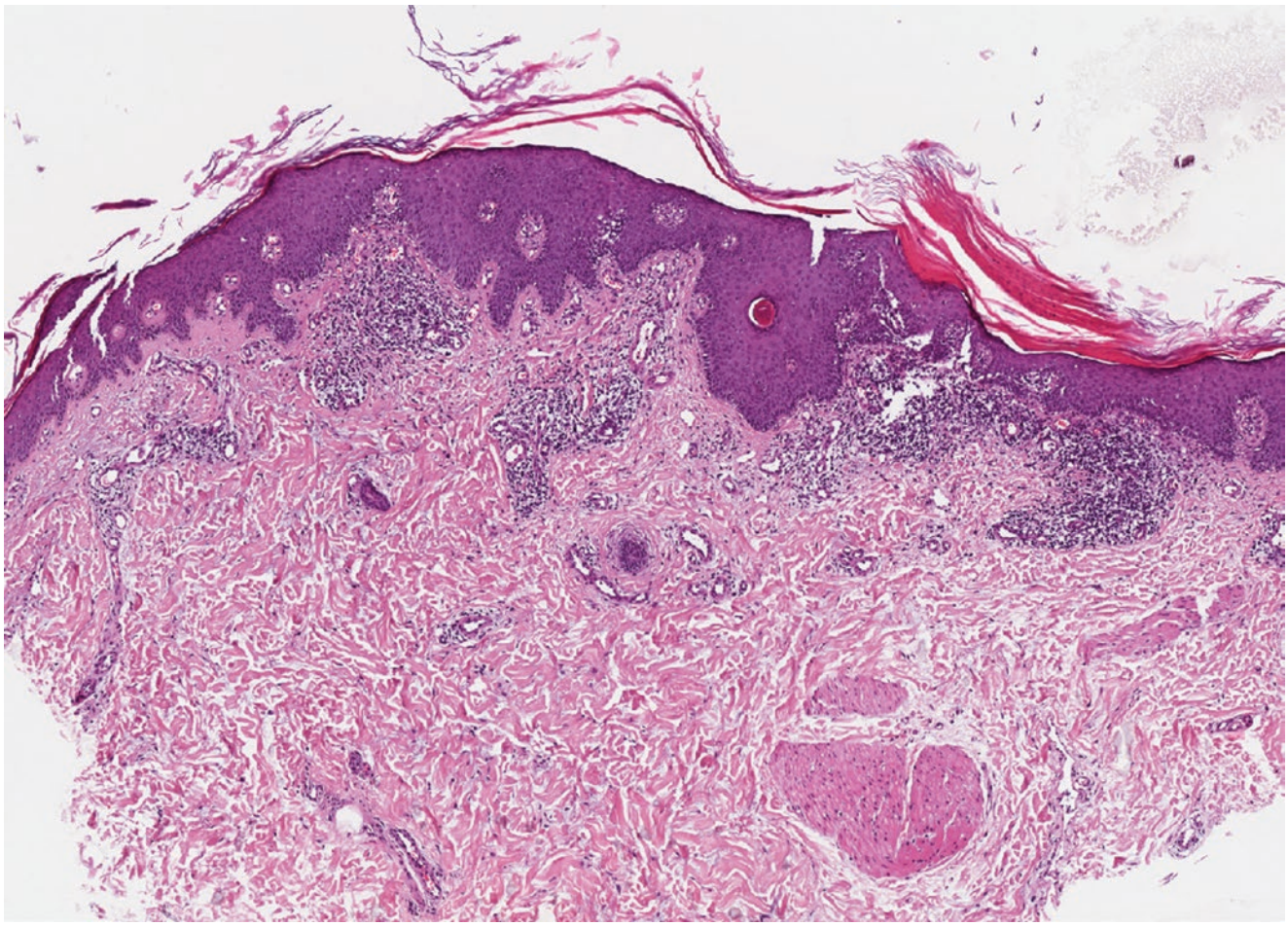

Fig. 11.5 Psoriasis. Histopathology showing the presence of epidermal hyperplasia, parakeratosis, acanthosis, and papillomatosis, with dilated vessels in the papillary dermis [H&E staining; magnification: ×100]

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Lichen Planus

12

Enzo Errichetti and Giuseppe Stinco

12.1 Definition

Lichen planus (LP) is an idiopathic inflammatory disorder affecting the skin, hair, nails, and mucosal membranes that often have a chronic course with relapses and periods of remission [1].

12.2 Epidemiology

Although LP more commonly affects middle-aged adults (about 1% of all new dermatology patients), it may occur at any age, including childhood [1, 2]. Its exact incidence in the pediatric population is not known and ranges between 0.1 and 10% of all LP cases [1]. Neither racial nor sex predilection has been noted [1]. Familial or inherited LP may occur in up to 10% of cases and usually manifests at an earlier age [1].

12.3 Etiology

LP is thought to be a T-cell-mediated autoimmune condition possibly targeting basal keratinocytes which express altered unknown self-antigens [1, 3]. Both CD4+ and CD8+ T cells are found in early LP lesions, while progression of disease usually leads to preferential accumulation of CD8+ cells, which are considered to play a key role in epidermal basal layer damage [1, 3]. Several triggering factors have been reported to initiate such an inflammatory response, mainly including viruses, drugs, and contact allergens [1]. A role of genetic factors is also suggested by the possible occurrence of familial cases [1].

12.4 Clinical Features

The typical cutaneous lesion of LP consists of a flat-topped, purple, polygonal, and pruritic papules (the four Ps), sometimes showing a shallow central depression; LP lesions are often grouped and tend to coalesce (Fig. 12.1a). Whitish dots or reticulated networks referred to as Wickham striae may be visible over the surface of well-developed papules, especially if examined by magnifying lens after applying an oil drop. LP usually heals with hyperpigmentation, which is more prominent among patients with dark skin (Fig. 12.2); conversely, post-inflammatory hypopigmentation develops only rarely. LP lesions mainly tend to involve the flexural areas of the wrists, arms, and legs in a bilateral symmetrical fashion. Numerous clinical variants have also been described, including [4] inverse LP, annular LP, hypertrophic LP, LP pigmentosus (Fig. 12.3), atrophic LP, ulcerative LP, bullous LP, LP pemphigoides, erythrodermic LP, linear LP, actinic LP, and follicular LP.

12.5 Differential Diagnosis

The main differential diagnoses of childhood LP include [4] guttate psoriasis, pityriasis rosea, lichen nitidus, pityriasis lichenoides, scabies, lichenoid sarcoidosis, lichen scrofulosorum, guttate morphea, lichen sclerosus, granuloma annulare, Majocchi's pigmented purpuric dermatosis, annular lichenoid dermatitis of youth, linear psoriasis, lichen striatus, lichen simplex chronicus, psoriasis vulgaris, and verrucous sarcoidosis.

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12.6 Diagnosis

The diagnosis of LP is usually not difficult when dealing with cases presenting with classical polygonal papules at sites of predilection and associated with characteristic mucous membrane lesions [1, 4]. However, considering the significant morphological variability of LP, such typical clinical findings may not always be evident, thereby requiring histology to make a definitive diagnosis [1, 4]. Dermatoscopy has been recently shown to be helpful to identify LP diagnostic clues not visible to the naked eye, thereby improving its noninvasive diagnosis, even in clinically atypical cases with unusual presentation [5–16].

12.7 Dermatoscopy

The main dermatoscopic feature of LP is represented by Wickham striae, which may appear as pearly-whitish (more commonly), yellowish, or blue-white (especially in dark-skinned patients) structures possibly showing various morphologies, i.e., reticular (the most common), linear, "radial streaming," annular, round, "leaf venation" (delicate secondary striae branching from the centered whitish venation, linked together at either end, mimicking the crystal structure of snow), and "starry sky" (clustered, follicular white dots) aspects (Figs. 12.1b, c and 12.4) [5–14]. Additional findings visible in active LP lesions include (1) violet, reddish, pink, brown, or yellow background; (2) vessels with dotted, globular, and/or linear appearance, mainly detectable at the periphery of the lesion and less commonly showing a perifollicular or diffuse pattern (Fig. 12.1b); (3) white/yellow dots; and (4) some pigmented structures (dots, globules, and/or reticular or cloud-like areas) (Fig. 12.1c) [5–14]. Even though all the aforementioned findings may sometimes coexist in a single lesion, dermatoscopy features usually vary according to disease stage [5–15]. Indeed, while early papules usually show a subtle WS over a reddish background, mature lesions are mainly characterized by both well-represented WS and peripheral vessels (Fig. 12.1b), which, however, tend to fade over time, concomitantly to the gradual appearance of pigmented structures (initially around WS contour and then diffusely—Fig. 12.1c) [5–15]. In long-standing lesions, the pigmented findings are often the only visible dermatoscopic clue [5–15].

Some morphological variants of LP may present peculiar dermatoscopic features: annular LP lesions usually show WS, capillaries or pigmented structures (according to their duration) on their active border; hypertrophic LP displays comedo-like structures filled with yellow plugs or round corneal structures ("corn pearls") and, less commonly, other abovementioned features [16]; LP pigmentosus typically features pigmented findings (Fig. 12.5).

Apart from diagnostic purposes, dermatoscopy may also be used to assess the likelihood of post-inflammatory pigmentation persistence (with homogeneous, structureless and light brown areas devoid of granularity being correlated with a shorter duration and granular pigmentation being associated with a longer course) and to monitor the evolution of LP lesions after therapy [15].

12.8 Histopathological Correlation

Histologically, active classical LP lesions show a band-like lymphocytic infiltrate in close proximity to the dermoepidermal junction and several epidermal changes, i.e., degeneration of the basal layer, irregular acanthosis, wedge-shaped hypergranulosis, and compact orthokeratosis, with the last two findings thought to be correlated with the typical WS seen on dermatoscopy (Fig. 12.6) [17]. Dilatation of papillary and subpapillary vessels is also present in elder lesions, and respectively corresponds to dotted and linear vessels visible on dermatoscopic examination [17]. In regressing lesions and LP pigmentosus, dermal pigment incontinence and melanophages are the predominant histological features, which dermatoscopically correlate with homogeneous hyperpigmentation and granular gray-bluish/brownish structures, respectively [17]. Comedo-like structures and round corneal structures ("corn pearls"), commonly seen on dermatoscopic examination of hypertrophic LP, are due to dilated hypergranulotic infundibula with orthokeratosis [17].

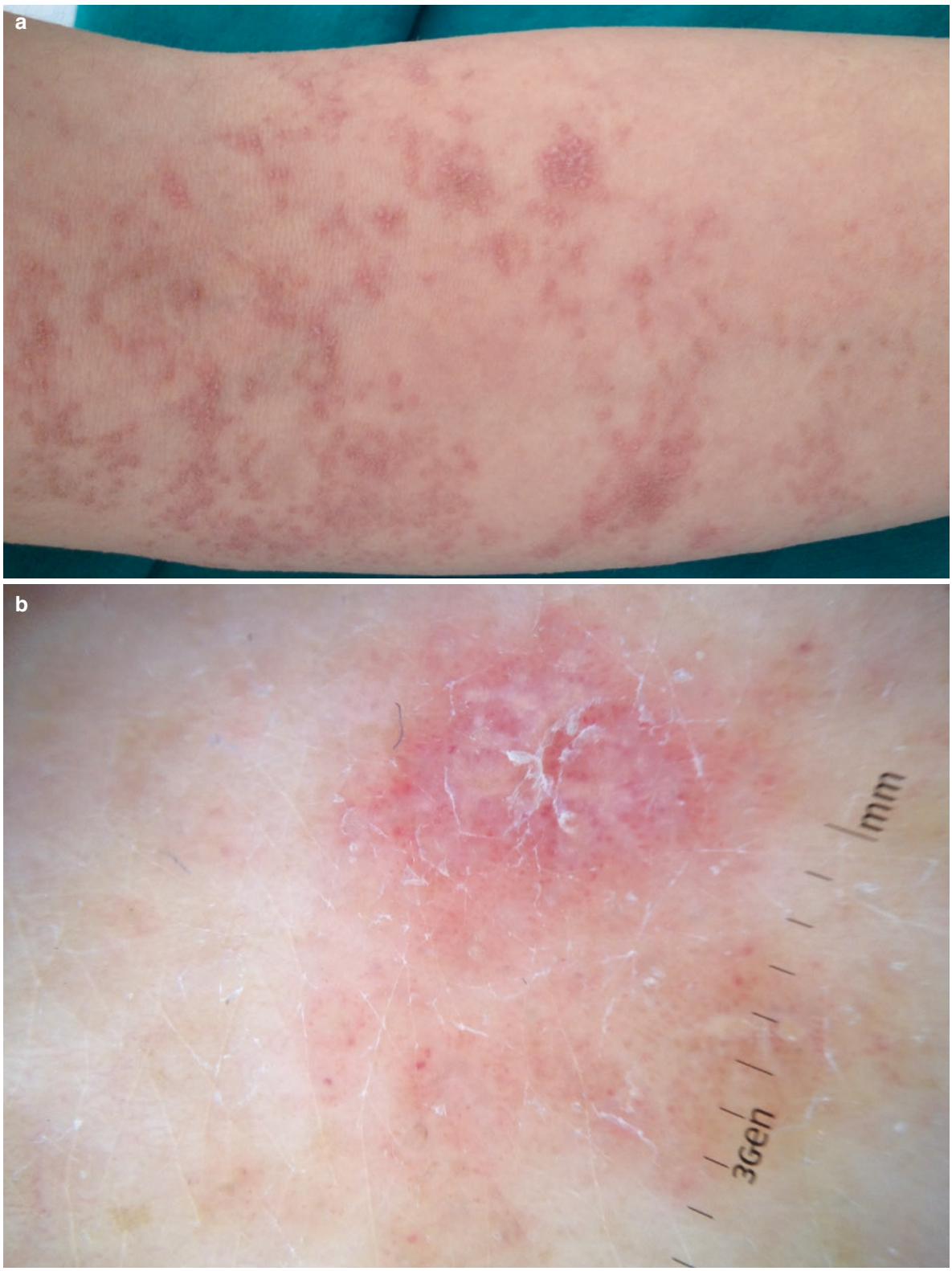

Fig. 12.1 (**a**) Classic lichen planus. Several flat-topped, purple, and polygonal papules having the tendency to coalesce on the arms of a 14-year-old girl. (**b**) Dermatoscopy (×10) of a mature lesion: typical Wickham striae (in this case displaying a yellowish-whitish shade and a reticular morphology) surrounded by dotted vessels. (**c**) Dermatoscopy (×10) of a regressing lesion: Wickham striae surrounded by brownish dots; vascular structures are less evident compared to mature lesions

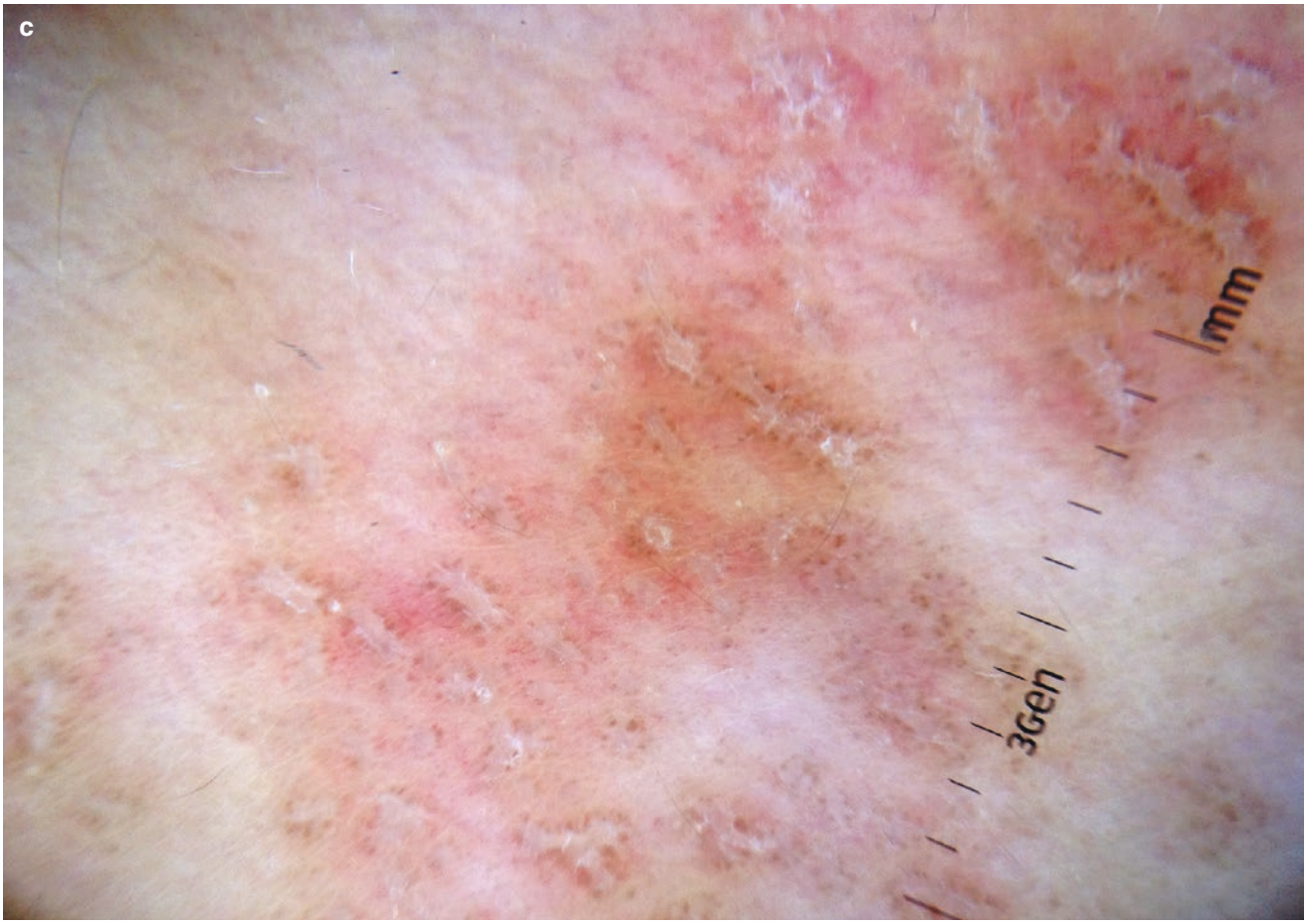

Fig. 12.1 (continued)

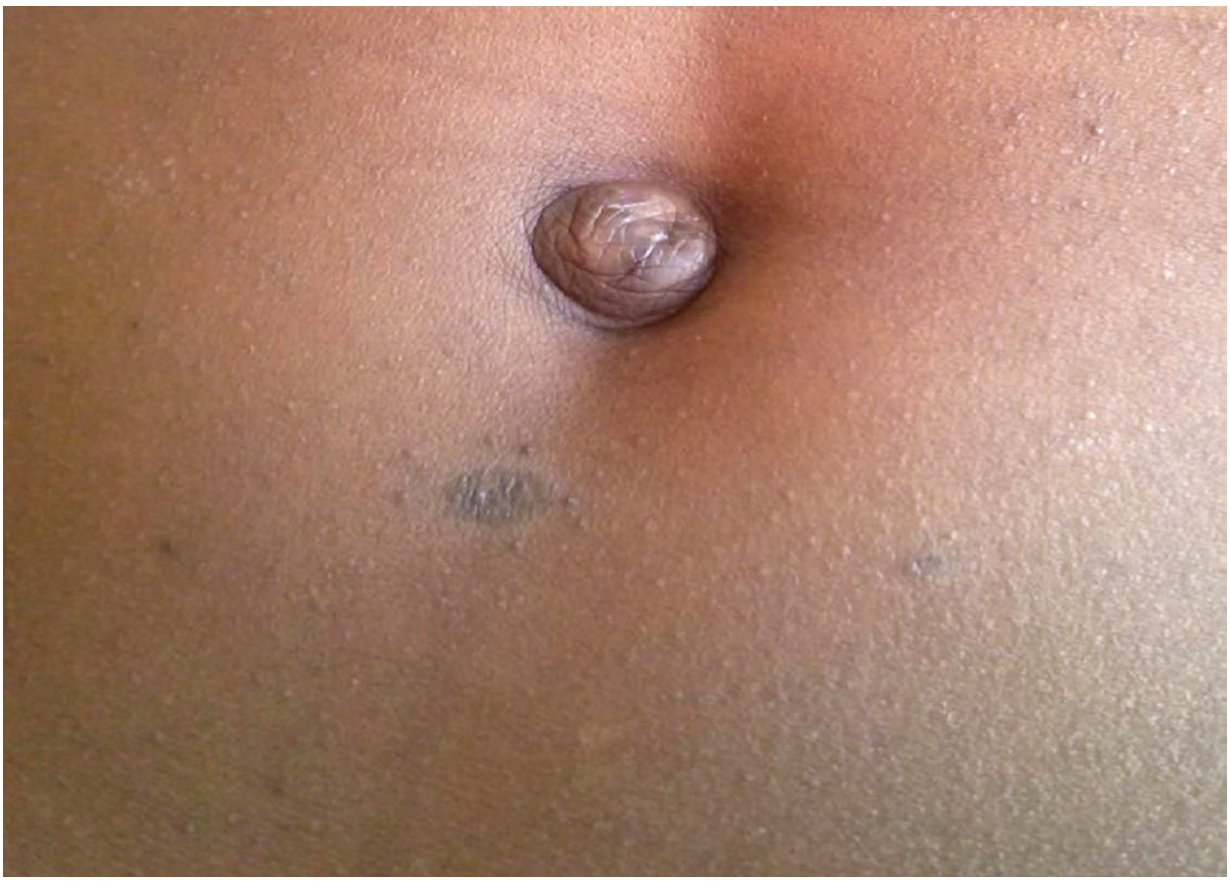

Fig. 12.2 Classic lichen planus. In dark-skinned patients, both active and healing lesions tend to show a darker hue

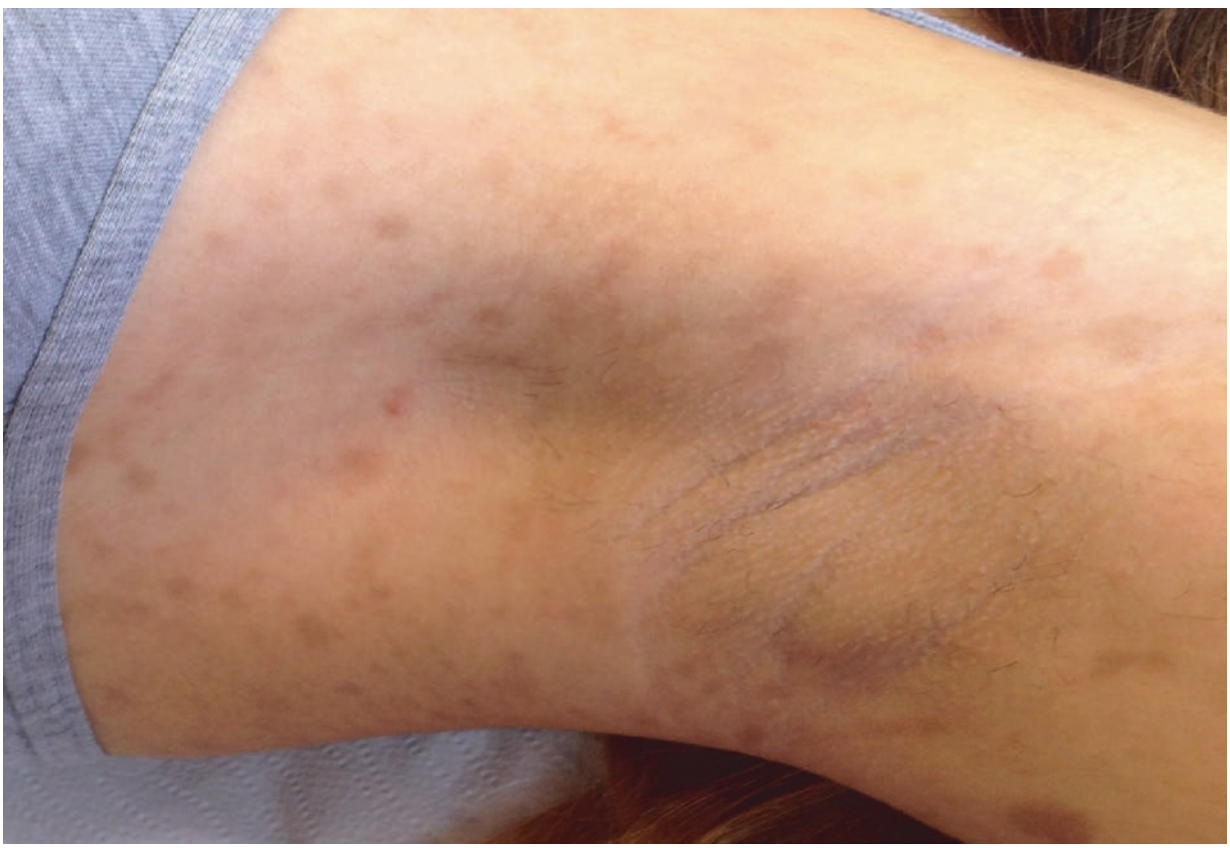

Fig. 12.3 Lichen pigmentosus. Typical brownish well-defined macules in a 12-year-old girl

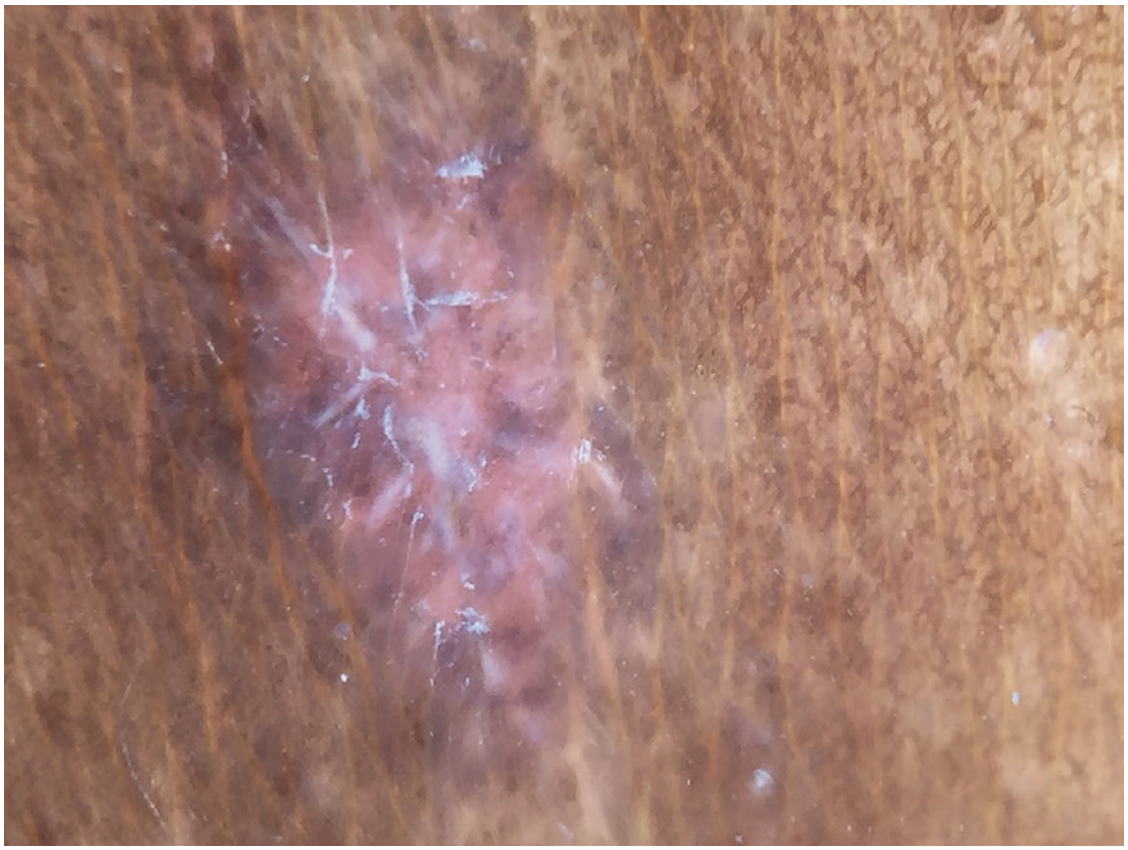

Fig. 12.4 Lichen planus in dark-skinned patient. Dermatoscopy (×10): Wickham striae showing a whitish-bluish hue

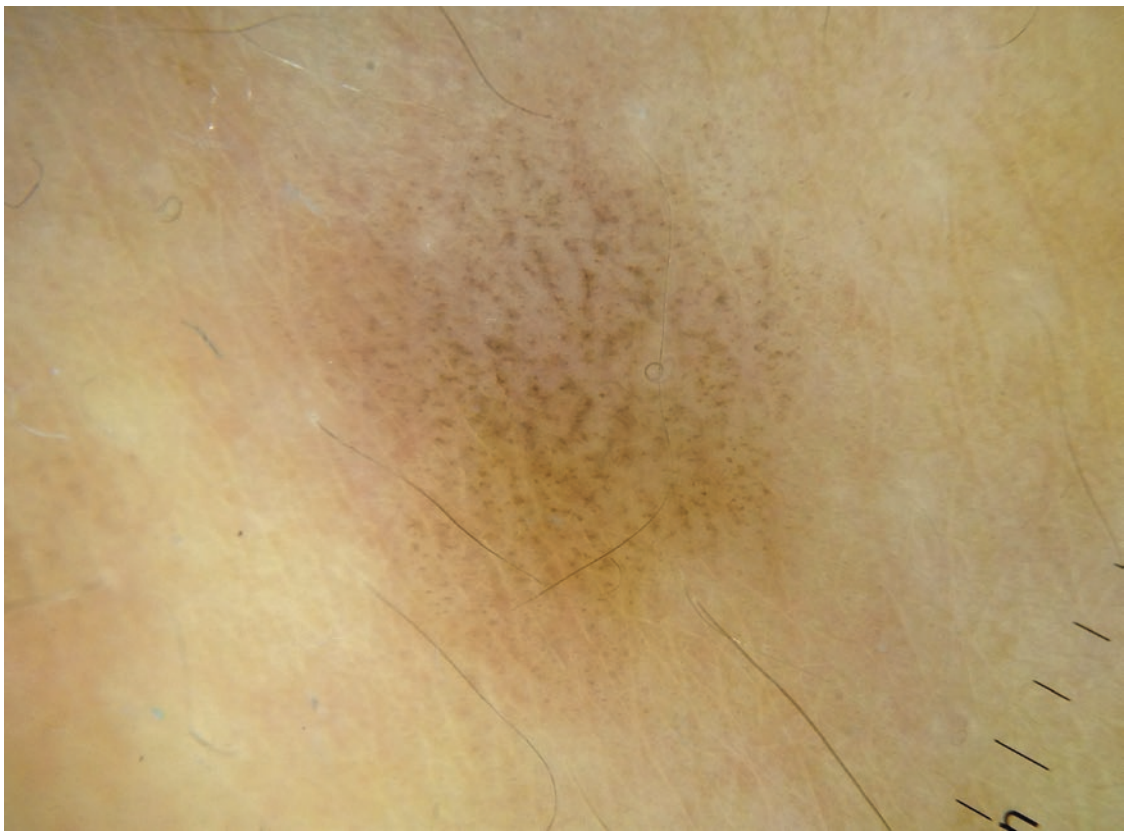

Fig. 12.5 Lichen planus pigmentosus. Dermatoscopy (×10): brownish-gray dots over a subtle brownish background. Pigmented structures are often the only dermatoscopic clue also in long-standing lesions of classic lichen planus

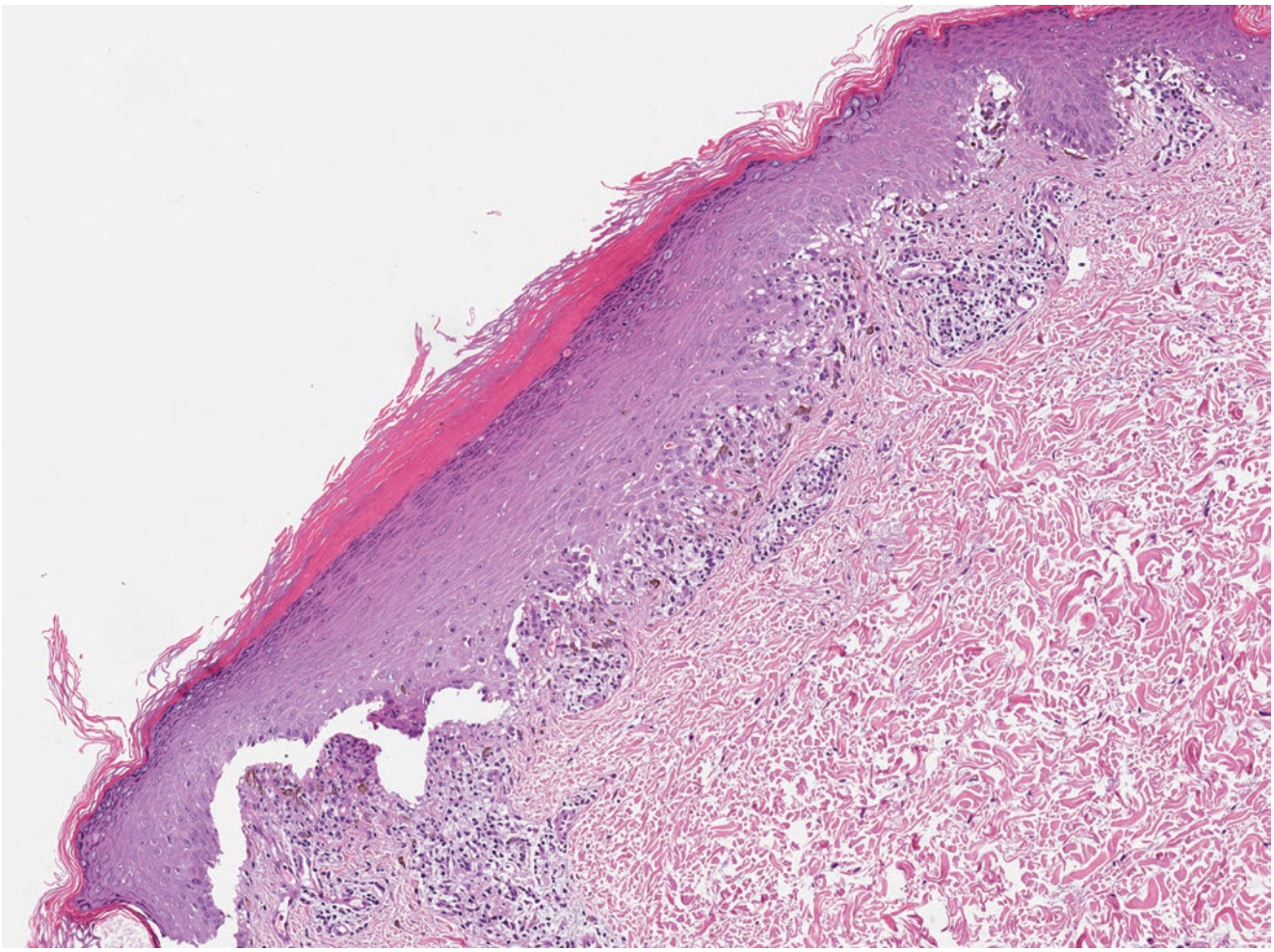

Fig. 12.6 Classic lichen planus. Histopathology: band-like lymphocytic infiltrate close to the dermoepidermal junction and epidermal changes (degeneration of the basal layer, irregular acanthosis, wedge-shaped hypergranulosis, and compact orthokeratosis) [H&E staining; magnification ×100] (Courtesy of Giuseppe Micali, MD)

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Lichen Nitidus

13

Enzo Errichetti and Giuseppe Stinco

13.1 Definition

Lichen nitidus (LN) is an idiopathic, benign, chronic, inflammatory dermatosis first described by Pinkus in 1901 which is typically characterized by spontaneous resolution in a few years [1].

13.2 Epidemiology

LN is a quite uncommon condition, with an estimated incidence of about 0.3 cases per 100,000 people [1]. It mainly affects children and young adults, even though cases in any age group have been reported [1, 2]. Neither racial nor gender predilection exists, albeit generalized variants appear to occur predominantly in females [1, 2].

13.3 Etiology

Although the exact etiology of LN is not known, the occurrence of familial cases has led some authors to speculate the existence of a genetically determined susceptibility to develop LN following environmental triggering factors [3]. According to some studies, LN could be induced by allergens causing epidermal and dermal antigen-presenting cells to activate a cell-mediated response, leading to lymphocyte accumulation and development of typical inflammatory papules [1].

13.4 Clinical Features

LN is typically characterized by tiny (1–2 mm in diameter), flesh-colored or hypopigmented (especially in dark-skinned individuals), asymptomatic, shiny papules (Fig. 13.1a), which mainly affect the abdomen, chest, extremities, and genitalia (especially in men), although atypical localizations such as mucous membranes, nails, palms, and soles have also been reported [1–3]. A linear arrangement of lesions may occur as a result of the Köbner phenomenon (skin lesions appearing on lines of trauma) [1–3]. Many unusual clinicopathological variants have been described, i.e., actinic, generalized, linear, keratodermic, perforating, purpuric/hemorrhagic, and vesicular [1–3].

13.5 Differential Diagnosis

The main differential diagnoses of LN in pediatric population include [4] micropapular sarcoidosis, papular granuloma annulare, lichen scrofulosorum, lichen planus, lichen

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spinulosus, lichen sclerosus, follicular eczema (Fig. 13.2), frictional lichenoid dermatitis (Fig. 13.3), keratosis pilaris, warts, and molluscum contagiosum.

13.6 Diagnosis

The diagnosis of LN is usually clinical, based on its distinctive features [1, 5]. However, when lesions are restricted to genitalia/unusual sites or in case of atypical presentations, histopathology may be required for a definitive diagnosis [1, 5]. Recently, dermatoscopy has been showed to be a useful supportive tool for the nonivasive recognition of LN [6, 7].

13.7 Dermatoscopy

Dermatoscopic examination of LN generally shows smooth, roundish, homogeneous, whitish clouds (one for each papule) usually presenting sharply demarcated margins and often lacking physiological skin markings (Fig. 13.1b) [6, 7]; the same pattern has been reported on the penis, even though lesions in such an area usually retain normal skin furrows [6]. On the other hand, when located on palmoplantar areas, LN lesions may reveal parallel linear scales as well as oval or elongated, well-defined depressions (pits) surrounded by ring-shaped, silvery white scales [7]. The possible detection of such dermatoscopic pattern on palms and soles might depend on the thickness of stratum corneum, persistent mechanical stress, and more hyperkeratotic disease presentation in these areas [7].

13.8 Histopathological Correlation

LN is characterized by a dense lymphohistiocytic inflammatory cell infiltrate (which may be granulomatous) lying in close proximity to the thinned epidermis and enveloped by bordering elongated rete ridges ("claw clutching a ball" appearance) (Fig. 13.4), which dermatoscopically corresponds to the whitish homogeneous cloud seen in each LS lesion [1]. Additional histological findings include basal cell hydropic degeneration and parakeratosis without hypergranulosis (Fig. 13.4), with the latter feature likely being responsible for the detection of scaling on dermatoscopic examination of palmoplantar LN [1, 7].

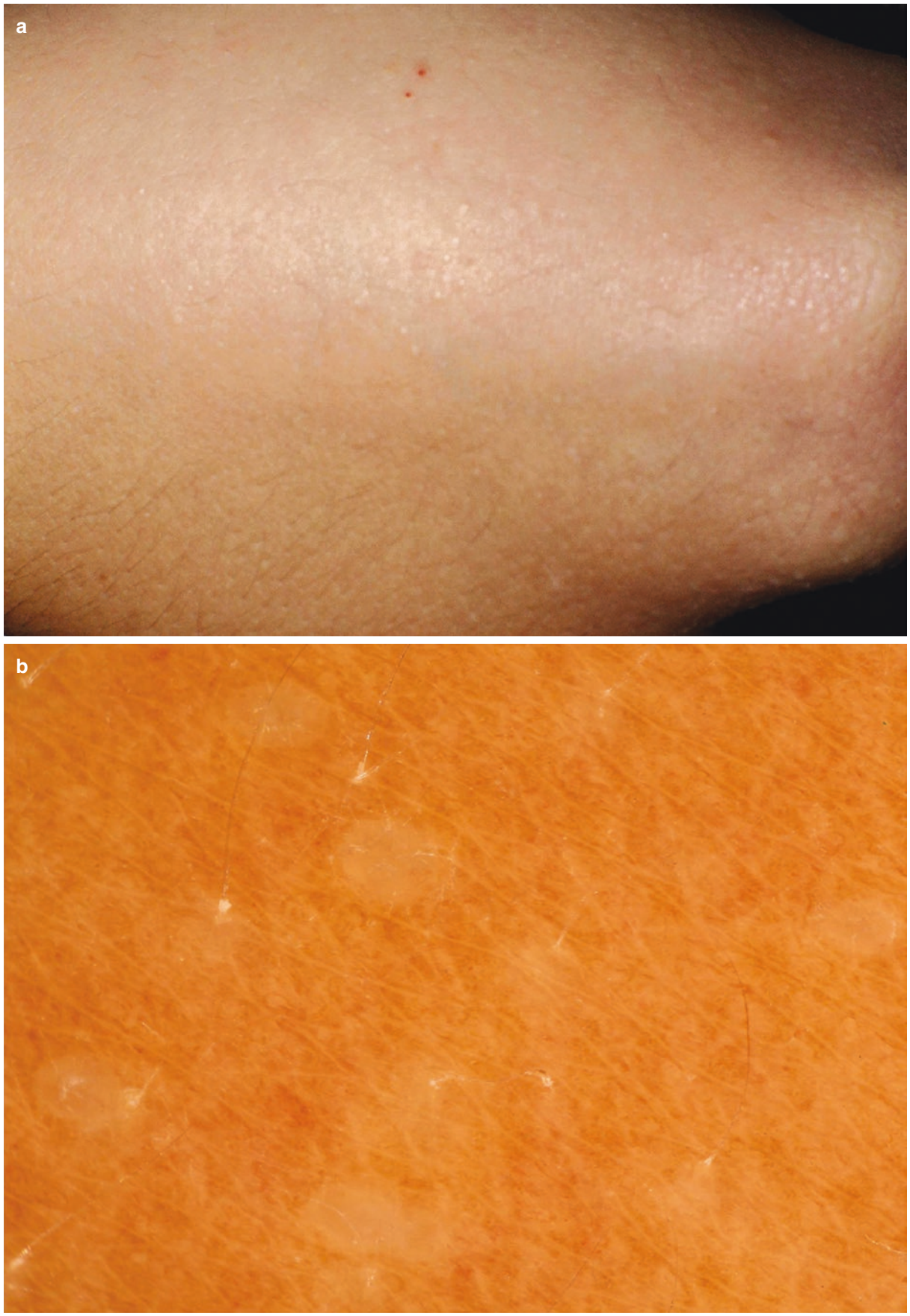

Fig. 13.1 (**a**) Lichen nitidus. Minute, discrete, flesh-colored papules localized on the right forearm/elbow in a 14-year-old girl. (**b**) Dermatoscopy (×10): smooth, roundish, homogeneous, whitish clouds (one for each papule) presenting sharply demarcated margins and lacking physiological skin markings

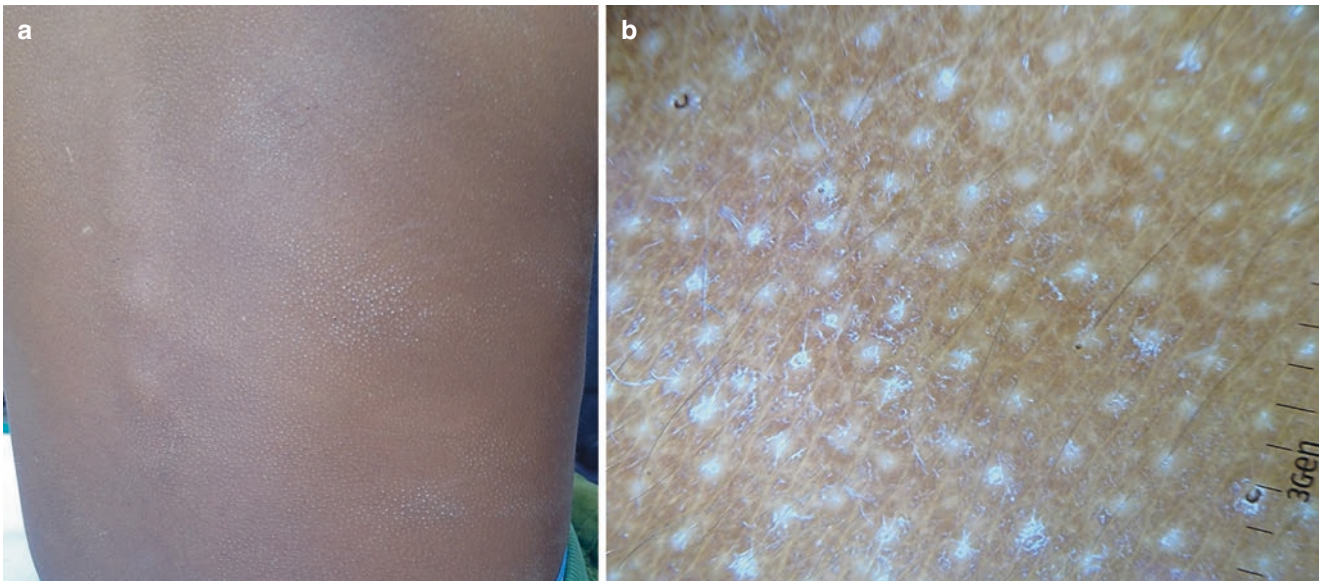

Fig. 13.2 (**a**) Follicular eczema. Several tiny hypopigmented papules on the back of a boy. (**b**) Dermoscopy (×10): roundish, keratotic, whitish areas with blurry margins and equidistant from one another (due to the follicular nature of the condition); some lesions are also centered by a hair

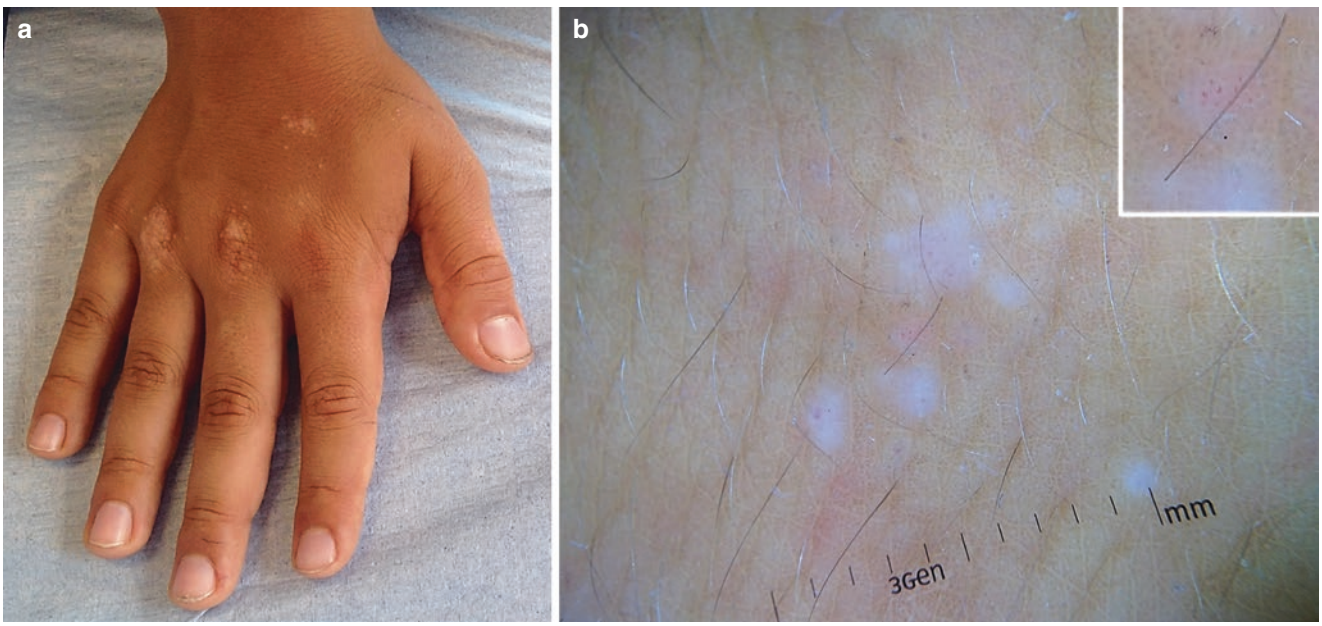

Fig. 13.3 Frictional lichenoid dermatitis. (**a**) Many hypopigmented flat papules on the dorsal aspect of the right hand of a boy. (**b**) Dermoscopy (×10): discrete, blurry, whitish areas with retention of the normal skin furrows (unlike lichen nitidus, in which they are often absent) and regularly arranged dotted vessels (box)

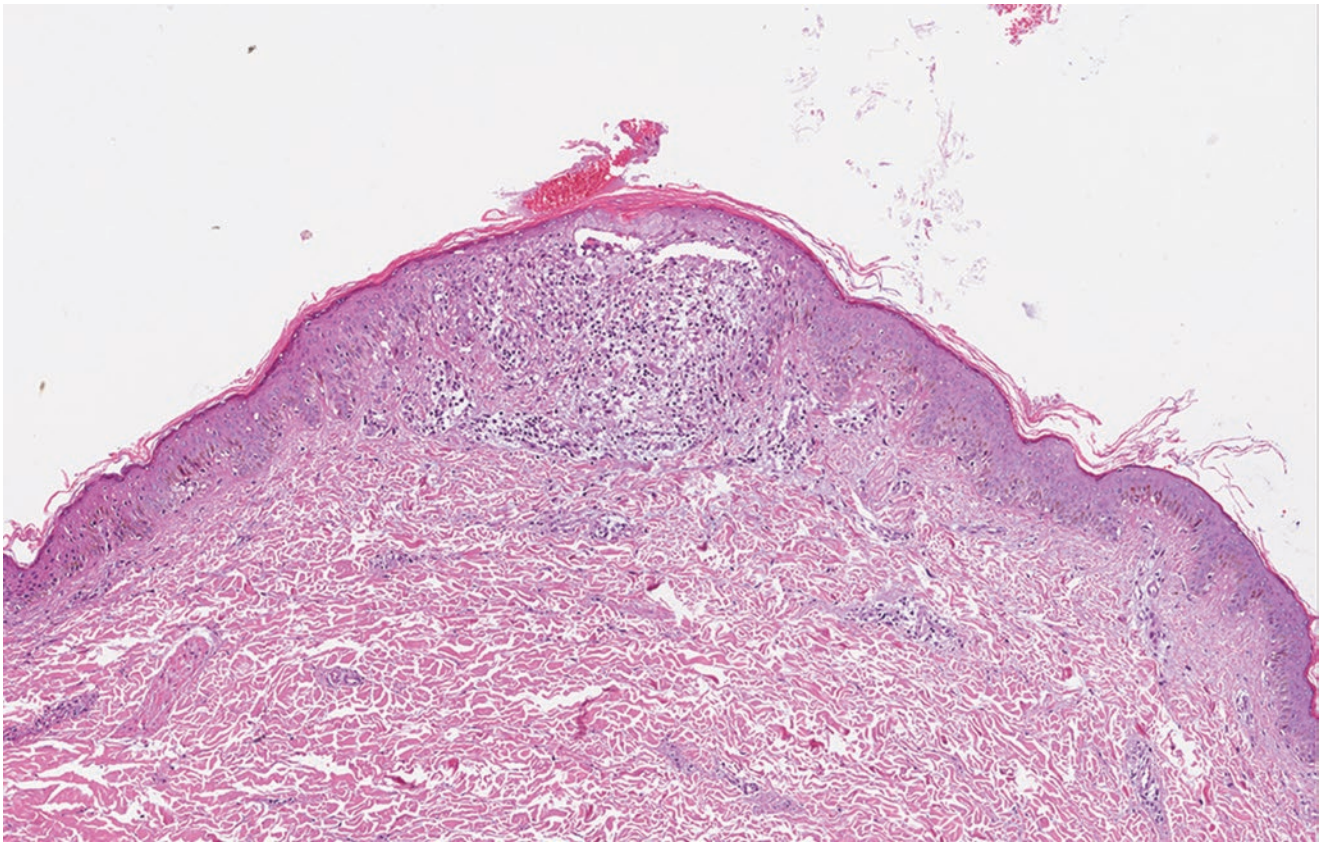

Fig. 13.4 Lichen nitidus. Histopathology: dense lymphohistiocytic inflammatory cell infiltrate lying in close proximity to the thinned epidermis [H&E staining, magnification ×100] (Courtesy of Giuseppe Micali, MD)

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Lichen Sclerosus

14

Iria Neri

14.1 Definition

Lichen sclerosus (LS) is a chronic inflammatory dermatosis causing atrophy and scarring that may involve both genital and extragenital areas.

14.2 Epidemiology

Females are mainly affected (85–90%) and the association with autoimmune disorders is found in 15% of patients. LS has a bimodal presentation: before puberty and in postmenopausal women. Extragenital manifestations occur in almost 11% of cases [1].

14.3 Etiology

The etiology is unclear, but an autoimmune mechanism has been hypothesized in association with HLA class I antigens and HLA-A29/B44. The role of human papillomavirus, *Borrelia burgdorferi*, hormonal changes, and trauma (Koebner phenomenon) has been postulated.

14.4 Clinical Features

LS initially presents with small, pink to ivory white polygonal papules that coalesce into porcelain-white plaques, determining atrophy and scarring as a result of increased skin fragility along with linear fissurations (Figs. 14.1a, 14.2a, 14.3a, and 14.4a). Purpura and ecchymosis may also occur [1]. Itching and pain are frequent complaints. In males, the glans and urinary meatus may be affected, causing phimosis and stenosis. In females, genital LS commonly involves the perianal and perineal region. Extragenital lesions may be observed in both sexes and are usually asymptomatic. The isomorphic response (Koebner phenomenon) may be detected.

14.5 Differential Diagnosis

The main differential diagnoses of LS are represented by vitiligo, post-inflammatory depigmentation, vulvar or penile intraepithelial neoplasia, and morphea in the case of extracutaneous manifestation. An overlap between LS and morphea (plaque-type and generalized) has been reported in the literature; therefore patients with morphea should be carefully screened for concomitant LS, including the inspection of the anogenital area [2].

14.6 Diagnosis

Diagnosis can be usually performed clinically with the aid of a dermatoscopic evaluation. In doubtful cases, a biopsy for histopathological examination is suggested.

14.7 Dermatoscopy

The main dermatoscopy features of genital LS are the presence of whitish patches associated to linear branching or dotted vessels (Fig. 14.1b) [3–5]. Minor findings include erosions, hemorrhages, gray-blue dots with a characteristic arrangement, scales, and comedo-like openings.

Dermatoscopy is a helpful tool also for the diagnosis of extragenital LS, revealing a homogeneous whitish area surrounded by an erythematous or hyperchromic halo (Figs. 14.2b,

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14.3b, and 14.4b). Multiple roundish to oval yellow circles (comedo-like openings) can be noticed on the whitish background, especially in early lesions (Figs. 14.2b and 14.3b) [3–9]. The differential diagnosis with morphea becomes much easier when performing dermatoscopy, which shows in this case accentuated whitish beams and linear branching vessels, with the lack of comedo-like openings [8, 9].

14.8 Histopathological Correlation

On histological examination, the homogeneous whitish areas with associated vascular changes seen on dermatoscopy correspond to epidermal atrophy, fibrosis, and vascular ectasia. The presence of gray-blue dots is related to dermal melanophages, while comedo-like openings are the result of ortohyperkeratosis and follicular plugging (Fig. 14.5) [3–7].

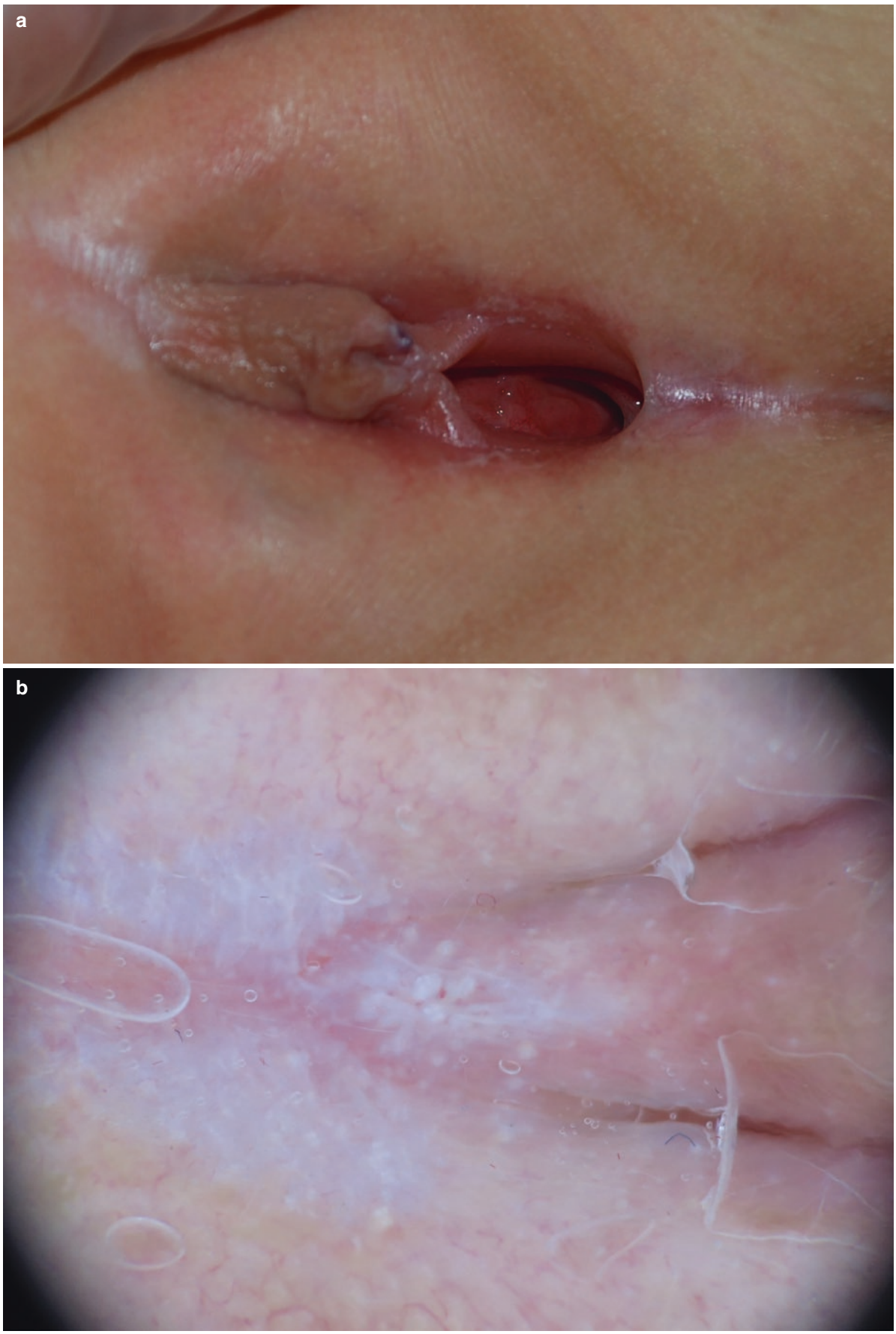

Fig. 14.1 (**a**) Vulvar lichen sclerosus in a 5-year-old girl: atrophic, porcelain, white plaques of the anterior labial commissure and prepuce of clitoris, with the concomitant involvement of the external surface of labia minus and perineum. (**b**) Dermatoscopy (x10): non-coalescent, whitish, oval areas with a central fine capillary network over a structureless background

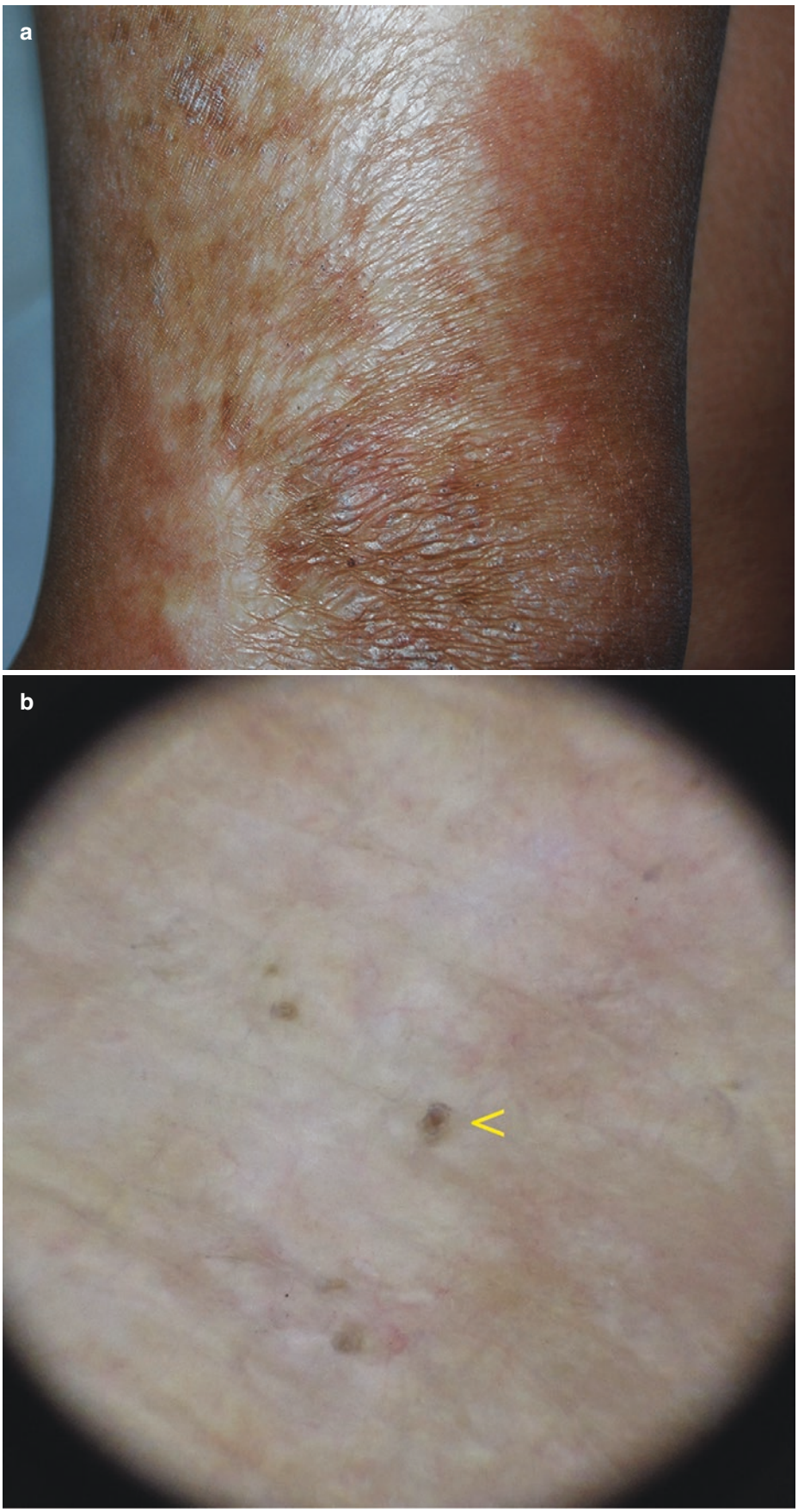

Fig. 14.2 (**a**) Extragenital lichen sclerosus on the legs in an 8-year-old dark-skinned girl. (**b**) Dermatoscopy (×10): patchy structureless whitish areas with comedo-like openings (arrowhead)

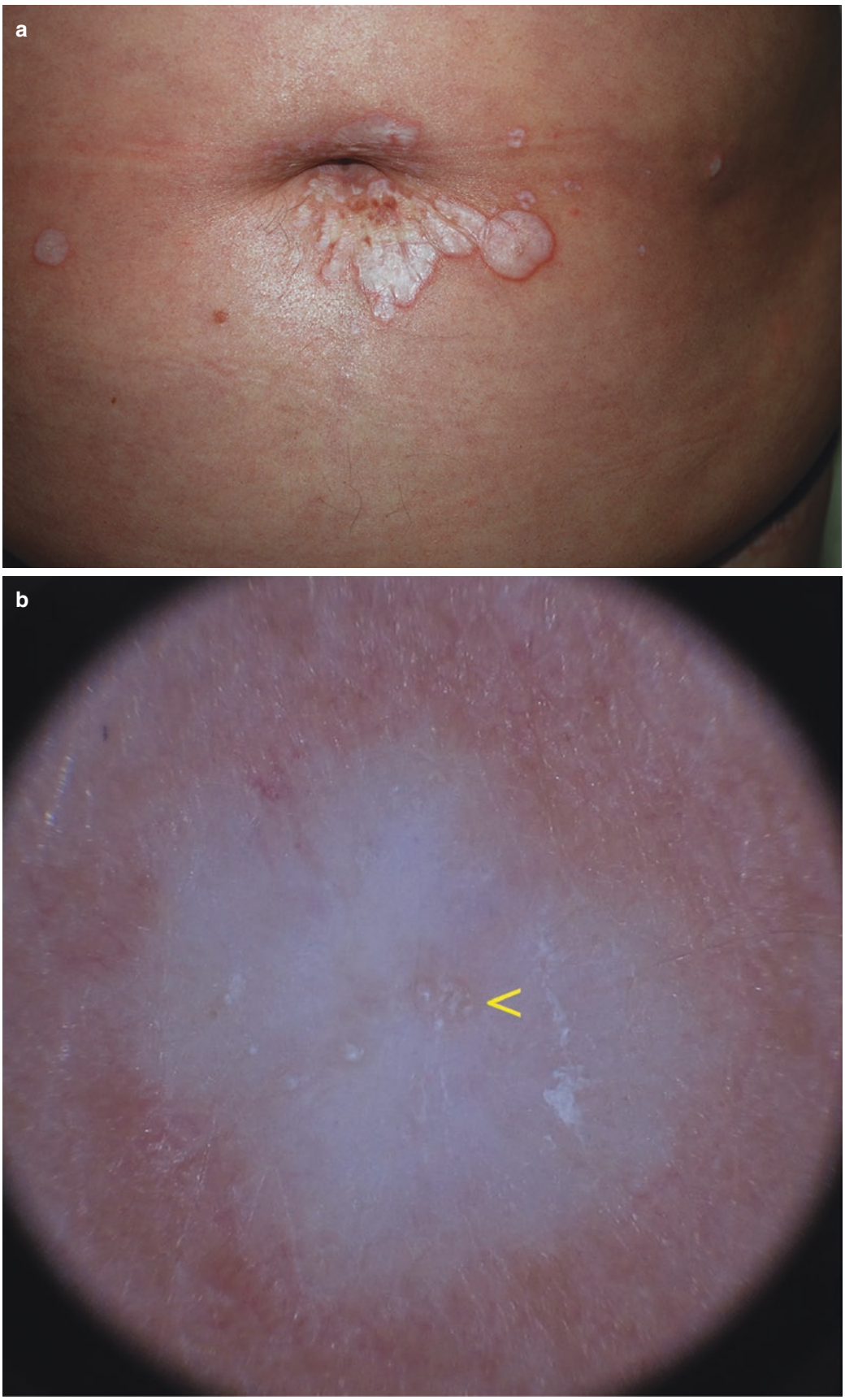

Fig. 14.3 (**a**) Extragenital lichen sclerosus on the abdomen in 17-year-old girl. (**b**) Dermatoscopy (×10): structureless whitish areas and white-yellow circles (comedo-like openings, arrowhead); the lesion is surrounded by an erythematous halo

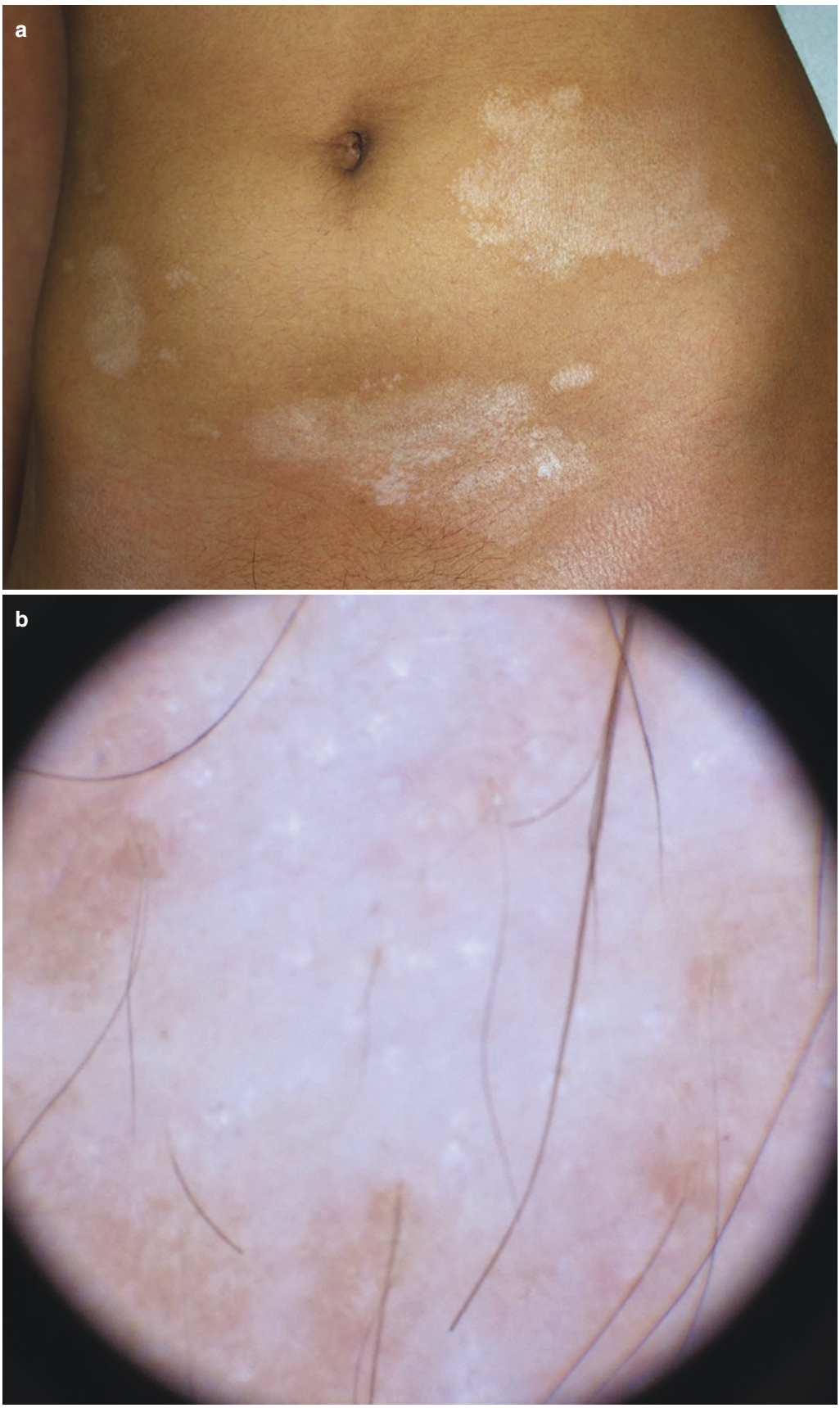

Fig. 14.4 (**a**) Extragenital lichen sclerosus on the abdomen in 15-year-old boy. (**b**) Dermatoscopy (×10): patchy white to yellow structureless areas with multiple whitish globules

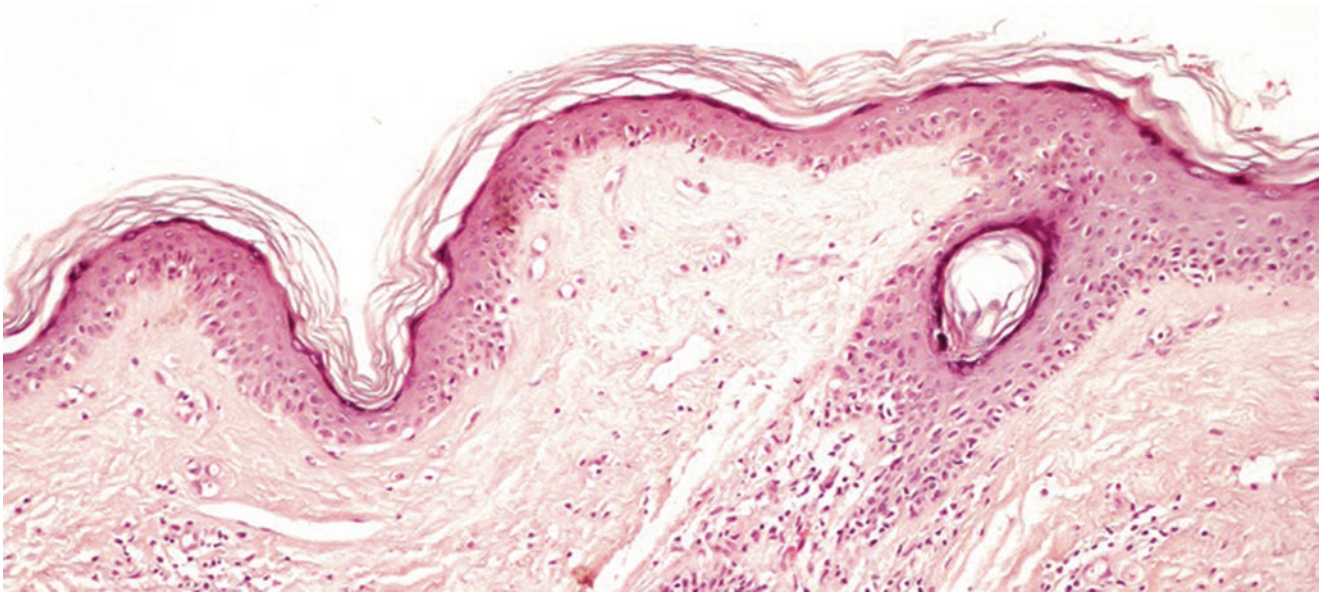

Fig. 14.5 Extragenital lichen sclerosus. Histopathology: laminated and basket woven orthokeratosis of the epidermis, slight perivascular lymphocytic inflammatory infiltrate and homogenization of the collagen in the superficial and mid-dermal layers [H&E staining; magnification: ×100]

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Lichen Aureus and Majocchi's Disease

15

Enzo Errichetti and Giuseppe Stinco

15.1 Definition

Lichen aureus (LA) and Majocchi's disease (MD) are two uncommon subtypes of pigmented purpuric dermatosis (capillaritis) which predominantly affect children/adolescents or young adults [1].

15.2 Epidemiology

LA is estimated to represent 0.05% of dermatological diseases in pediatric patients, while there are no precise data on the prevalence of MD in children [2]. Whereas LD mainly occurs in males, MD shows a female predilection [2]. All races are equally affected [2, 3].

15.3 Etiology

The etiopathogenesis of both LA and MD is not known [1–5]. Capillary fragility has been speculated to play a key role, yet precise physiopathological mechanisms are not clear [1–5]. Asymptomatic infections, trauma, and drugs have been reported as possible triggers [1–5]. Moreover, a role of vascular pressure increase has been supposed [1–5]. A few familial cases of MD have been described, thus supporting the role of a possible genetic background, at least in a minority of patients [5, 6].

15.4 Clinical Features

LA is typically characterized by asymptomatic to mildly pruritic, small, golden, copper- or rust-colored, lichenoid papules often coalescing into sharply demarcated plaques which range in size from 1 to 20 cm (Fig. 15.1a) [1–4]; lesions may be solitary or multiple but are usually unilateral [1]. A zosteriform pattern has been rarely described [1–4].

Clinically, MD presents with a variable number of asymptomatic or slightly pruritic, annular, linear, serpiginous or arciform, 1–3 cm in diameter, erythematous patches showing tiny telangiectases along with petechiae inside the border, and central clearing sometimes associated with atrophy (Fig. 15.2a) [1–3, 5–7]. Lesions occur symmetrically on the lower extremities, but may extend to the trunk and upper extremities [1–3, 5–7].

The course of both LA and MD is chronic and is characterized by recurrences and remissions over a period of months to years [1–7].

15.5 Differential Diagnosis

In children, purpuric contact dermatitis, necrobiosis lipoidica, traumatic bruises, fixed drug eruption, and purpuric mycosis fungoides should be ruled out for LA, whereas for MD differential diagnosis includes granuloma annulare, angioma serpiginosum, tinea corporis, erythema annulare centrifugum, and multiple erythema chronicum migrans [1–7].

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15.6 Diagnosis

The diagnosis of both LA and MD is usually made on clinical ground by recognizing their peculiar features [1–7]. A skin biopsy is helpful in doubtful cases [1–7]. The use of dermatoscopy has been recently showed to be useful to support the noninvasive diagnosis of both conditions [8–12].

15.7 Dermatoscopy

LA and MD display similar dermatoscopic findings, viz., irregular or roundish, nonblanchable, purpuric, reddish dots, globules, and/or patches over a red-brownish or red-coppery background [8–12], which, in the authors' experience, is usually diffuse/more prominent in LA and more focal/subtle in MD (Figs. 15.1b and 15.2b). Additionally, some telangiectatic vessels may also be seen in MD, while LA may show gray spots, globules, and a network of brownish to gray interconnected lines as well [8–12].

15.8 Histopathological Correlation

The histological hallmarks of both LA and MD consist of extravasation of red blood cells and presence of various amounts of hemosiderin deposits in macrophages (Fig. 15.3) [2, 4–7], which respectively correlate with purpuric spots and red-brownish/coppery background seen on dermatoscopy [8–12]. Other histological features include a lymphohistiocytic perivascular dermal infiltrate in MD and a dense band-like lymphohistiocytic dermal infiltrate along with hyperpigmentation of the basal cell layer and dermal incontinentia pigmenti in LA [2, 4–7], with the last two findings being dermatoscopically correlated with brownish-gray reticular lines and gray dots [8–12]. Of note, both LA and MD may show an increased number of dilated dermal vessels, which, however, are more pronounced in the latter condition, thus explaining the presence of telangiectases on dermatoscopic examination [2, 4–7].

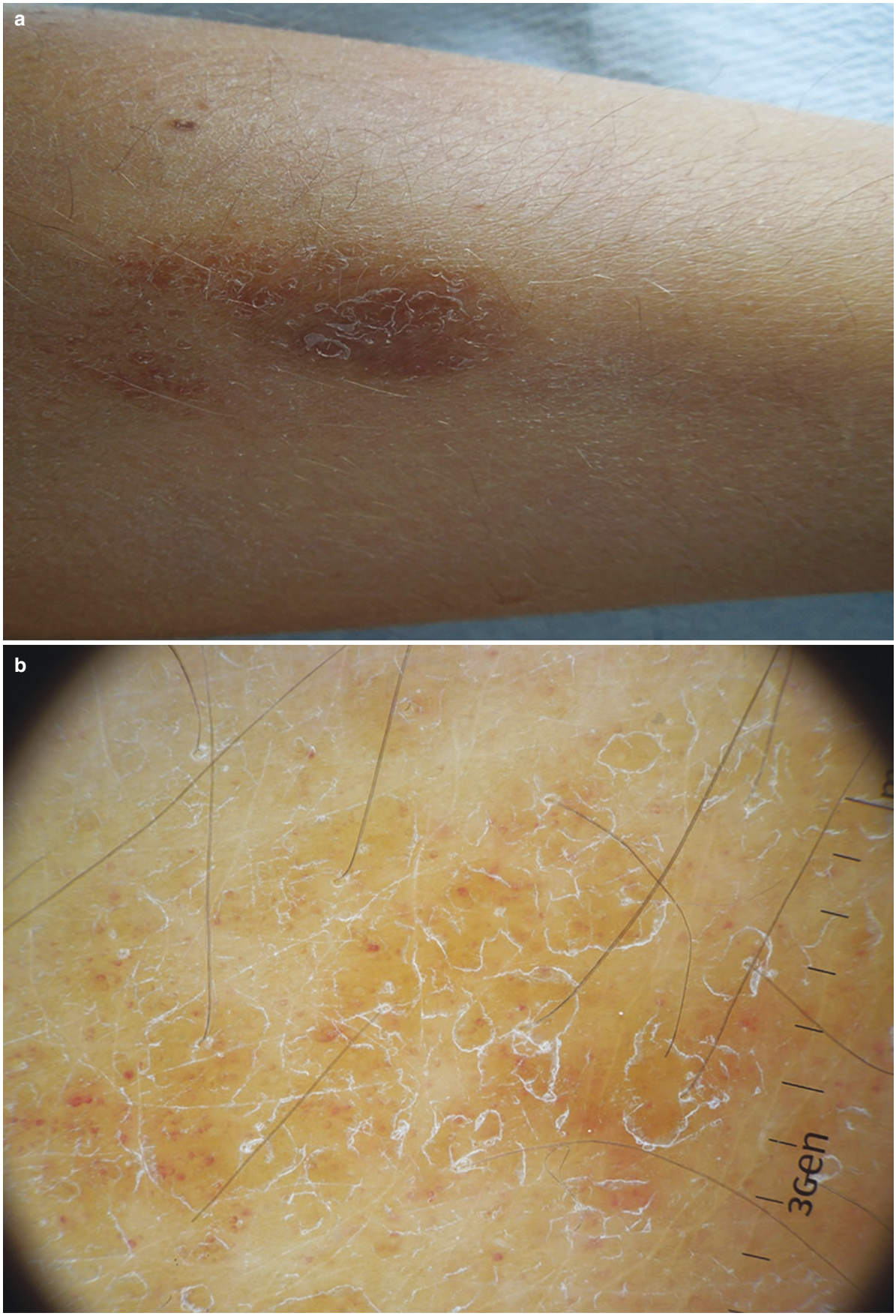

Fig. 15.1 (**a**) Lichen aureus. Solitary, rust-colored, sharply demarcated plaque with a few satellite papules on the left leg of a 12-year-old girl. (**b**) Dermatoscopy (×10): roundish, nonblanchable, purpuric, reddish dots/globules along with whitish scaling over a diffuse coppery background

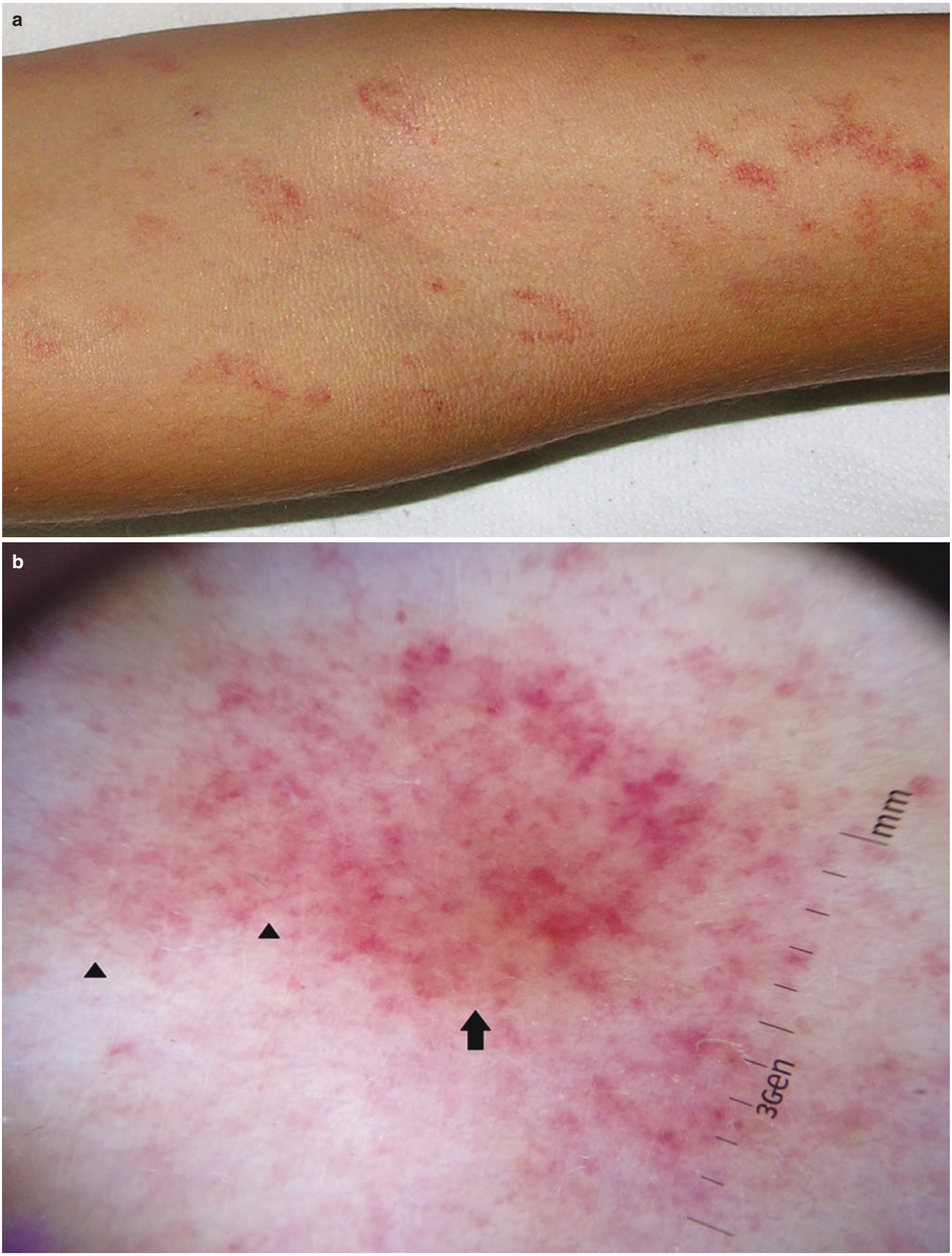

Fig. 15.2 (**a**) Majocchi's disease. Several annular, linear and arciform, erythematous patches on the right arm of an 11-year-old boy. (**b**) Dermatoscopy (×10): several roundish, nonblanchable, purpuric, reddish dots/globules and a few telangiectatic vessels (arrowheads); a focal coppery area is also evident (arrow)

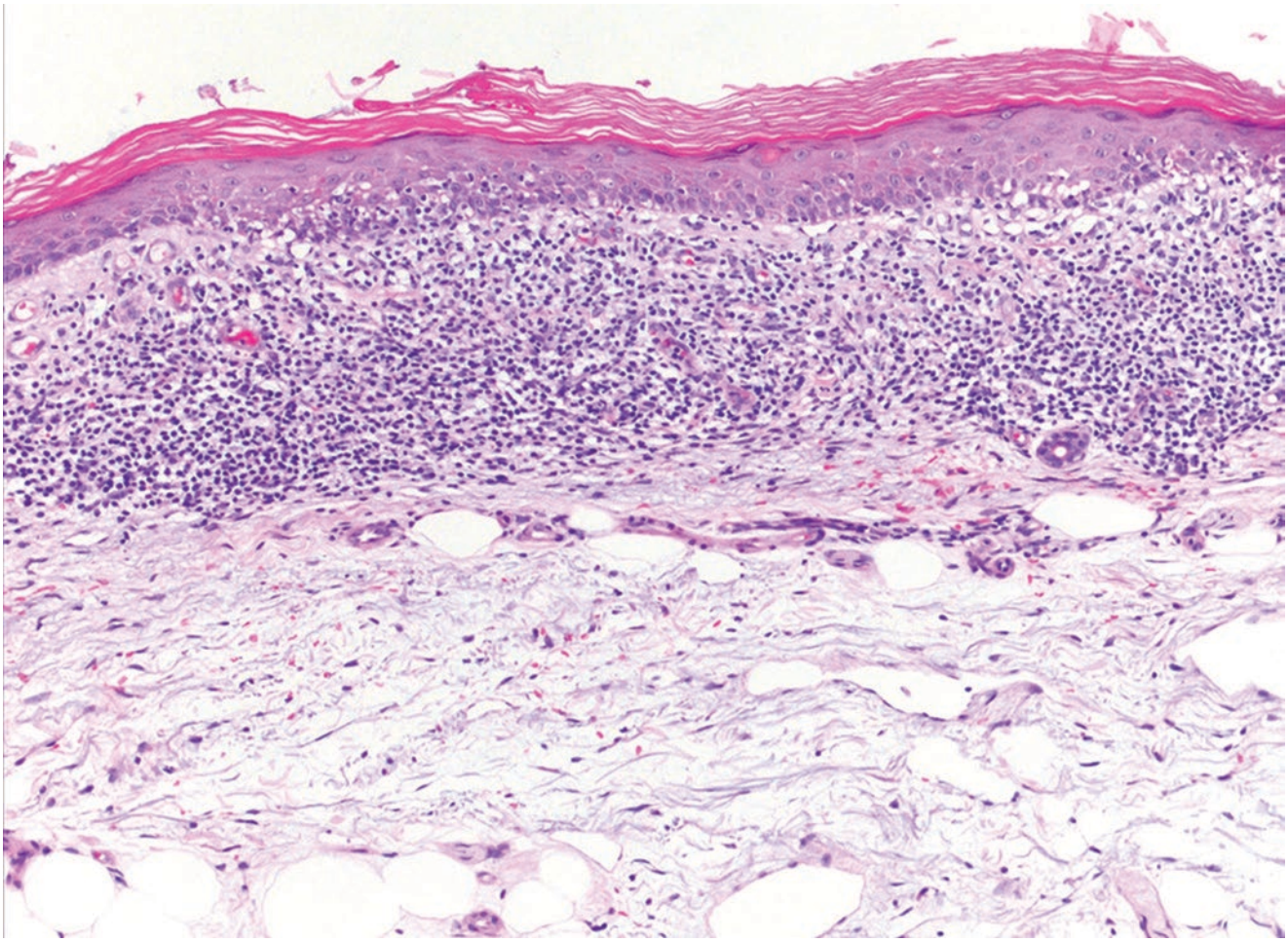

Fig. 15.3 Lichen aureus. Histopathology: typical extravasated erythrocytes (responsible for the presence of reddish purpuric dots/globules) along with a band-like lymphohistiocytic dermal infiltrate [H&E staining; magnification ×100; courtesy of Carla Di Loreto, MD, Institute of Anatomic Pathology, University of Udine, Italy]

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Morphea

16

Enzo Errichetti and Giuseppe Stinco

16.1 Definition

Morphea, also known as localized scleroderma, is a self-limited or chronically relapsing connective tissue disorder characterized by excessive collagen deposition leading to thickening of the dermis and/or subcutaneous tissues [1]. Differently from systemic sclerosis, it typically does not show sclerodactyly, Raynaud phenomenon, nailfold capillary changes, telangiectasias, and progressive internal organ involvement [1].

16.2 Epidemiology

Morphea is a rare condition, with an estimated incidence around 0.4–2.7 per 100,000 people [1, 2]. Up to half of all cases occur in pediatric patients, with linear and plaque/superficial circumscribed subtypes representing by far the most common clinical forms in this group [1, 2]. Similarly to other connective tissue diseases, there is a female predominance, which is, however, less marked in children, especially in subjects under 10 years of age (female to male ratios of 1.5:1) [1]. Although morphea affects all races, it appears to be more common in whites [1, 2].

16.3 Etiology

The etiopathogenesis of morphea is still unclear, but an autoimmune mechanism has been supposed based on the frequent presence of autoantibodies and on the personal and/or familial history of autoimmune disease in affected individuals [1]. Endothelial cell injury (due to antiendothelial cell antibody-dependent cytotoxicity or autologous complement activation) is currently thought to be the inciting event in the pathogenesis of the disease, with subsequent increase in levels of adhesion molecules, recruitment of inflammatory cells (mainly lymphocytes), and overproduction of fibrogenic cytokines (i.e., IL-4, IL-6, and TGF-beta) [1, 3]. Trauma, radiation, medications, and infections have all been proposed to act as triggering events in the development of morphea in susceptible individuals [1].

16.4 Clinical Features

Individual lesions of morphea typically begin with an erythematous, edematous, inflammatory phase, which is followed by the development of central sclerosis presenting as thickening and ivory white discoloration; an erythematous-to-violaceous border (lilac ring) encircling the sclerotic area is typically visible in active lesions (Fig. 16.1a) [1, 4, 5]. Over months or years, sclerotic lesions soften and become atrophic with hypo- or hyperpigmentation (Fig. 16.2a); loss of hair follicles and sweat glands is usually present [1, 4, 5]. Morphea includes several morphological subtypes [1, 4, 5]: circumscribed plaque-type (single or multiple round to oval lesions >1 cm in diameter involving up to two anatomical regions); guttate variant (multiple roundish lesions having a diameter less than <1 cm usually affecting the trunk); keloidal or nodular subtype (nodules resembling keloids in the presence of typical plaque-type morphea); bullous variant (tense subepidermal bullae developing overlying plaque-type, linear, or deep morphea lesions); linear subtype (blaschkoid linear induration of the skin/subcutaneous tissues), generalized plaque-type (multiple roundish lesions >1 cm in diameter involving at least three anatomical sites); deep subtype (primarily involving the subcutaneous fat and underlying structures); and pansclerotic variant (circumferential and often deep involvement of the majority of body surface areas with sparing of fingers, toes, and nipples).

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16.5 Differential Diagnosis

The main differential diagnoses of morphea in pediatric population vary according to disease stage [6]: (1) early inflammatory lesions (erythema chronicum migrans, annular lichenoid dermatitis of youth, fixed drug eruption, and granuloma annulare); (2) sclerotic lesions (lichen sclerosus et atrophicus, radiation fibrosis, stiff skin syndrome, progeria, and morpheaform dermatofibrosarcoma protuberans); and (3) atrophic lesions (parapsoriasis/mycosis fungoides, atrophoderma of Pasini and Pierini, linear atrophoderma of Moulin, and atrophic lichen planus).

16.6 Diagnosis

Diagnosis of morphea in pediatric population is based on the detection of typical clinical features, though histological assessment is needed in case of atypical/subtle presentation or early/late lesions in order to distinguish such a condition from its main differential diagnoses [1, 4, 5].

16.7 Dermatoscopy

The most peculiar dermatoscopic feature of sclerotic/active morphea lesions consists of whitish fibrotic beams, which are frequently crossed by linear branching vessels (Fig. 16.1b) [7–10]. Of note, such findings are also visible in clinically non-sclerotic patches, thereby facilitating the recognition of this dermatosis even in early phases [7–10]. Pigment network-like structures are also often evident in morphea (Fig. 16.1b), while "comedo-like openings" and whitish patches are seen in a minority of cases only [8–10]. According to a relatively recent study comparing dermatoscopic features of lichen sclerosus and morphea, the presence of "comedo-like openings" and whitish patches would be more indicative of a diagnosis of lichen sclerosus, while the detection of fibrotic beams would be more characteristic of morphea [8]. Interestingly, some authors have emphasized the usefulness of dermatoscopy in therapeutic monitoring because it would allow an accurate assessment of fibrosis reduction (whitish beams) and regression of neovessels (branching vessels) typical of morphea (Fig. 16.2b) [7].

16.8 Histopathological Correlation

The histological features of morphea vary according to disease stage [11]. The active inflammatory phase (Fig. 16.3) is typically characterized by a perivascular/interstitial inflammatory infiltrate of lymphocytes/plasma cells in reticular dermis and subcutaneous tissues and dermal vessel dilatation/neovascularization, which are responsible for the presence of linear branching vessels on dermatoscopy, and thickening of preexisting collagen bundles/deposition of newly formed collagen, dermatoscopically corresponding to whitish fibrotic beams [11]. Sometimes, hyperpigmentation of the basal cell layer may be present as well, thus resulting in pigment network-like structures on dermatoscopic assessment [11]. In the sclerotic stage, the inflammatory infiltrate typically decreases, but collagen bundles in the reticular dermis/subcutis become thick, closely packed, and hyalinized, with consequent increase in the number of fibrotic beams on dermatoscopy [11].

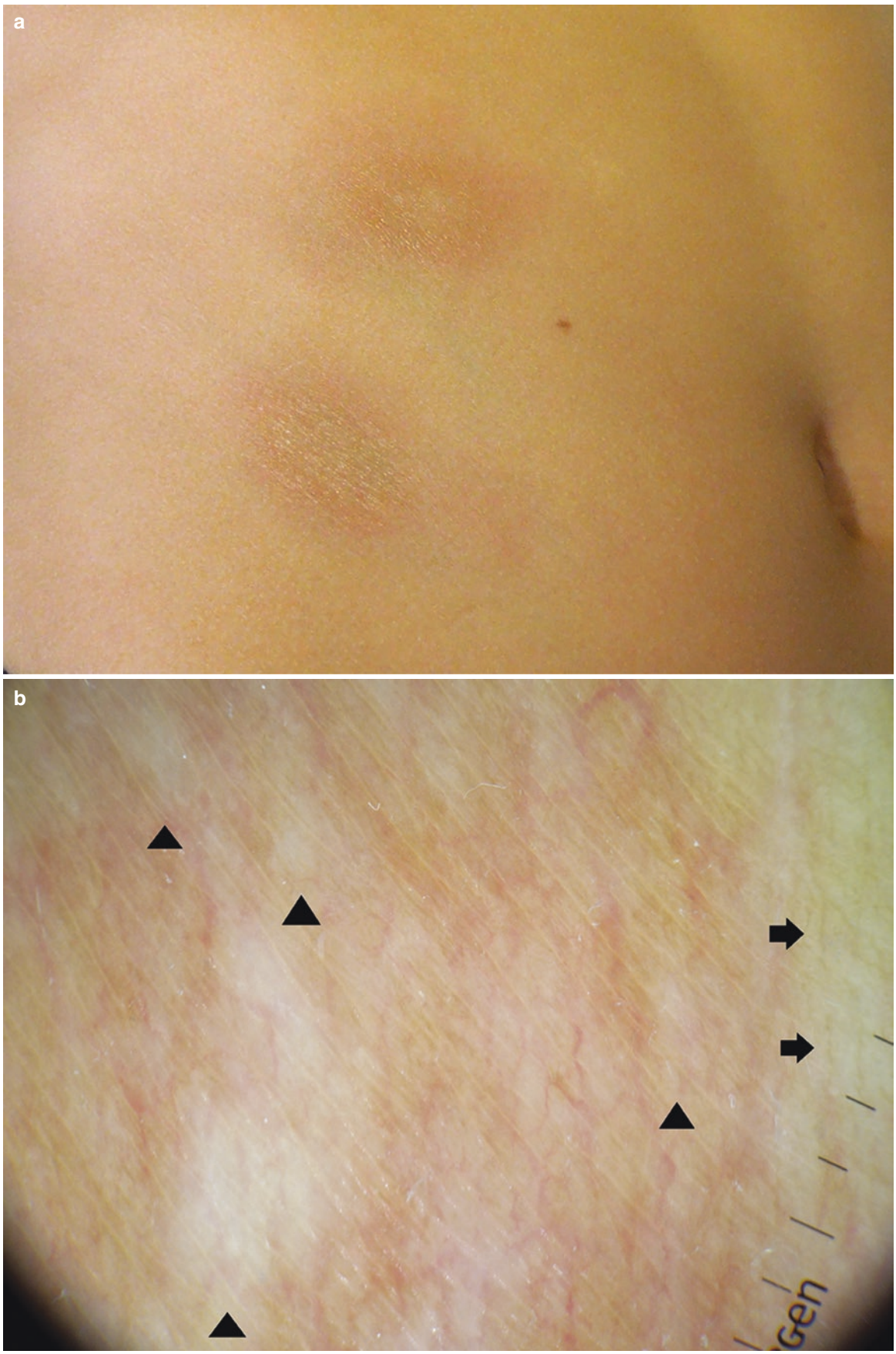

Fig. 16.1 (**a**) Morphea. Two sclerotic plaques with an ivory white-colored center and a slightly erythematous border on the abdomen of a 7-year-old boy. (**b**) Dermatoscopy (×10) of the active border of a plaque: whitish beams (arrowheads) and linear branching vessels; a few brownish lines (arrows) are also present

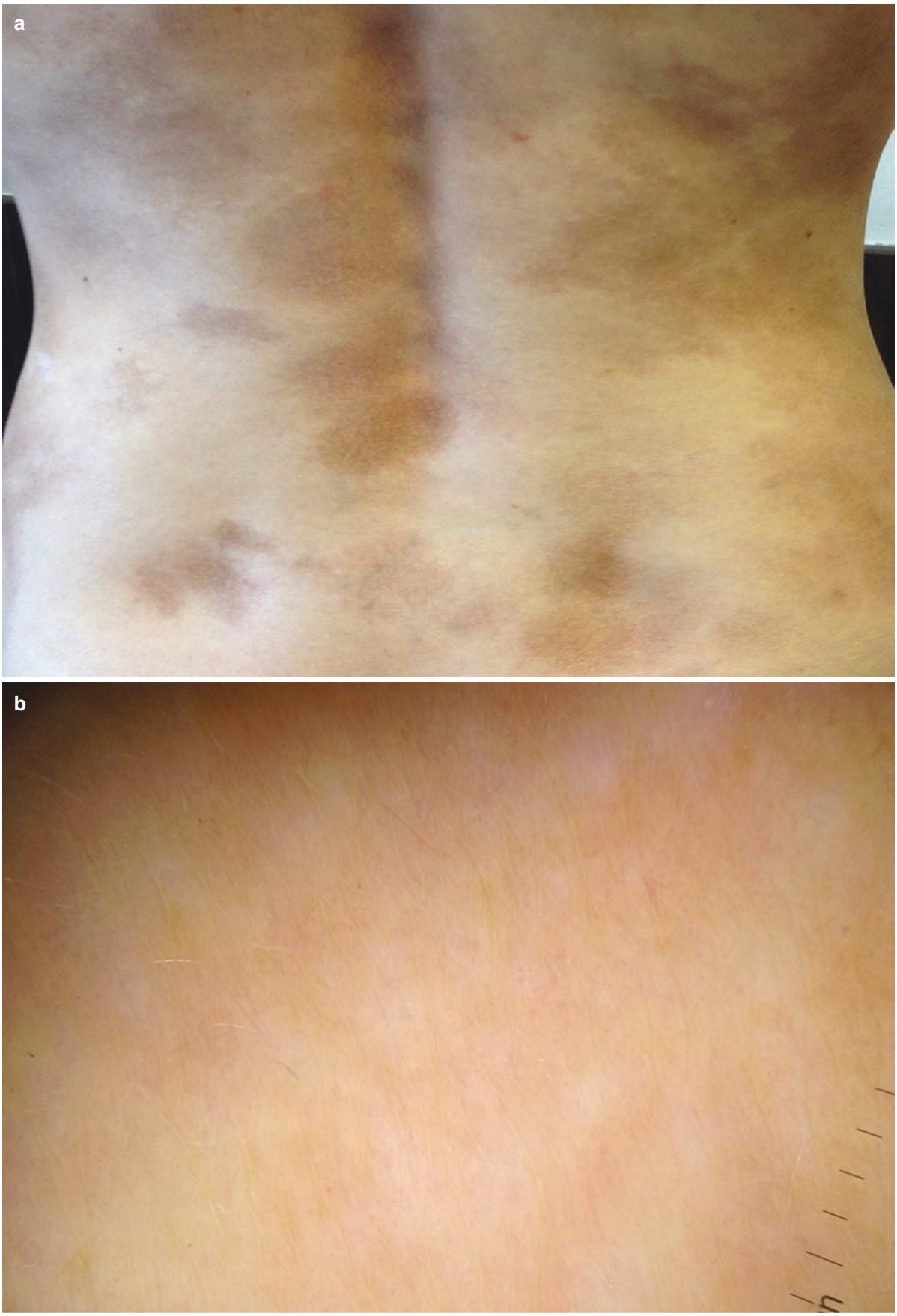

Fig. 16.2 (**a**) Morphea. Several long-standing hyperpigmented patches on the back of a 14-year-old girl. (**b**) Dermatoscopy (×10) of an inactive patch: less intense whitish beams and no significant vessels

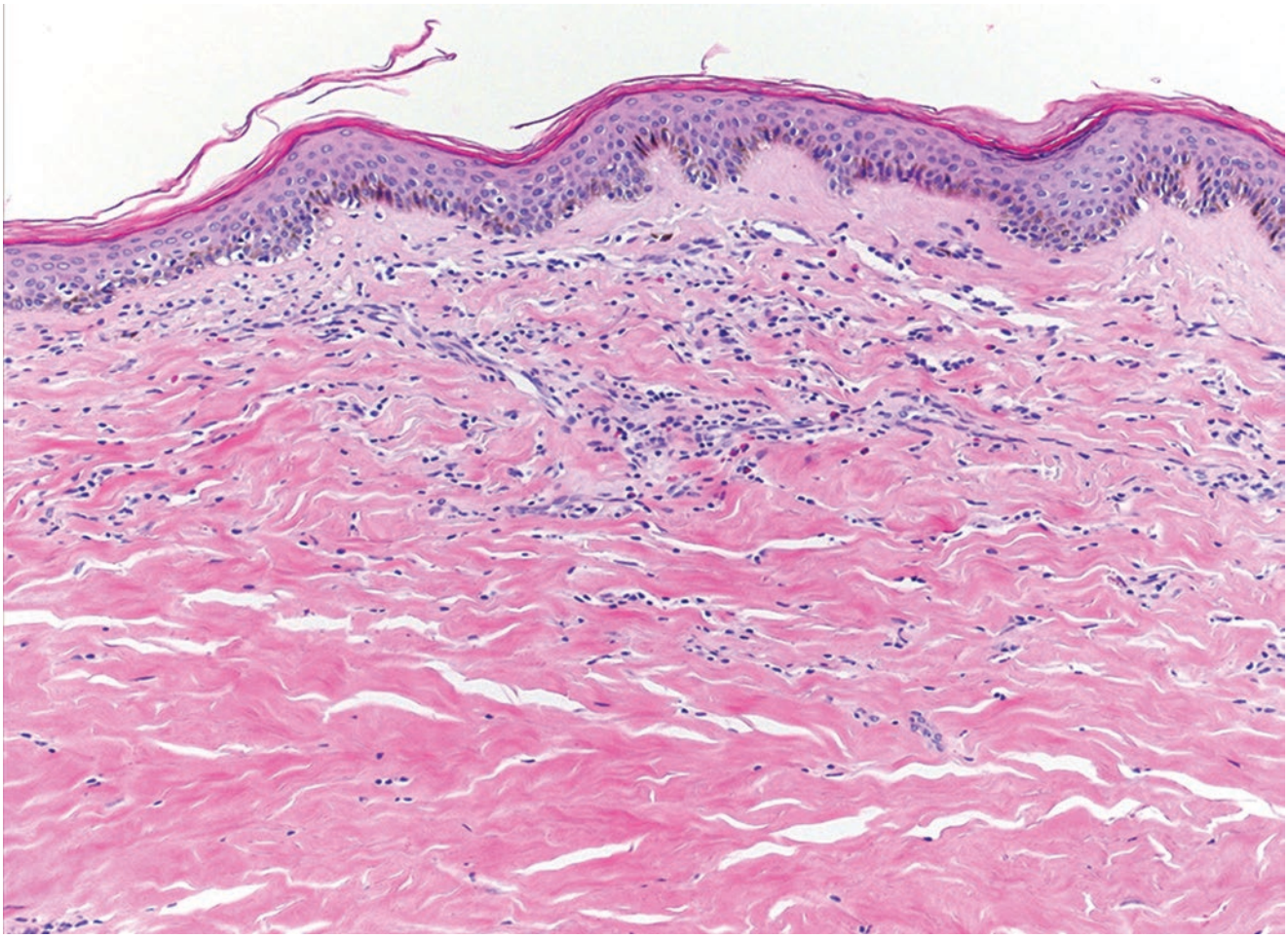

Fig. 16.3 Morphea. Histopathology of an active lesion. Perivascular/interstitial inflammatory infiltrate of lymphocytes/plasma cells in reticular dermis, dermal vessels dilatation, hyperpigmentation of the basal cell layer, and thickening of collagen bundles [H&E staining; magnification ×100; courtesy of Carla Di Loreto, MD, Institute of Anatomic Pathology, University of Udine, Italy]

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Pityriasis Rosea

17

Enzo Errichetti and Giuseppe Stinco

17.1 Definition

Pityriasis rosea (PR), which literally means "fine pink scale," is an acute, self-limiting exanthematous disease usually presenting a duration of 6–8 weeks, which was first described by Gibert in 1860 [1].

17.2 Epidemiology

PR is a very common condition, with an estimated annual incidence of around 160 cases per 100,000 [1]. In temperate climates, there may be a seasonal variation, with most cases occurring in spring and winter [1]. Even though PR may be seen in any age group, it is more common in subjects aged between 10 and 35 years [1]. It is slightly more frequent in females, with a female-to-male ratio of 1.5–2:1 [2]. No racial predominance has been reported [1, 2].

17.3 Etiology

PR is thought to be a reactive viral exanthem based on the following evidences: seasonal occurrence, clinical course, description of epidemic occurrence, possible presence of prodromal symptoms, and low recurrence rate [3]. In particular, recent evidence suggests that it could be associated with reactivation of herpes viruses 6 and 7 [3]. Although other infectious agents have been speculated to play a trigger role in its development, recent analyses demonstrated that no definitive association exists [3, 4].

17.4 Clinical Features

Classic PR usually presents with a single truncal skin lesion, known as "herald patch," which is followed by the onset of numerous smaller lesions [1]. The primary lesion consists of a well demarcated, 2–4 cm in diameter, oval or round, salmon-colored, erythematous, or hyperpigmented (especially in individuals with darker skin) patch presenting a fine peripheral scaling collarette (Fig. 17.1a) [1]. After a period ranging from a few days to a couple of months, secondary lesions develop along the lines of cleavage in a "Christmas tree" pattern mainly on the trunk and proximal extremities (Figs. 17.1a and 17.2a) [1]. From a morphological point of view, they may resemble the herald patch in miniature or present as small, red, usually nonscaly papules [1]. In approximately 20% of cases, PR presents with atypical features: absence of "herald patch" or typical scaling, inverse (peripheral) localization, localized forms, and morphological variants (urticarial, pustular, purpuric, follicular, and erythema multiforme-like) [1].

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17.5 Differential Diagnosis

PR in pediatric population should mainly be differentiated from lichen planus, guttate psoriasis, pityriasis lichenoides, tinea corporis, drug eruption, disseminated contact dermatitis, nummular eczema, and scabies [1, 5].

17.6 Diagnosis

The diagnosis of PR is usually clinical, based on its distinctive features [1]. However, recognizing atypical instances may be quite challenging, thereby requiring histological assessment to differentiate PR from similar conditions [1, 5]. Of note, dermatoscopy has been reported to be useful to improve the clinical diagnosis of PR [6–10].

17.7 Dermatoscopy

Both the herald patch and the secondary lesions of PR typically show a characteristic peripheral whitish scaling localized just inside the border ("collarette" sign) as well as dotted vessels, which, differently from psoriasis, are distributed in an irregular or focal pattern; diffuse or localized yellowish-orange structureless areas may be visible as well (Fig. 17.1b) [6–10]. In the authors' experience, the dermatoscopic "collarette" sign may be appreciated even when it is clinically hardly detectable (Figs. 17.1c and 17.2b), and such a feature is often the only clue in dark skin patients, as vascular findings and yellowish-orange structureless areas are usually not visible in these skin complexions (Fig. 17.2b).

17.8 Histopathological Correlation

Histopathologically (Fig. 17.3), peripheral scaling area of PR lesions is characterized by (1) epidermal changes, including slight acanthosis, focal spongiosis, and focal parakeratosis in mounds [11], with the last finding correlating with the dermatoscopic "collarette" sign, and (2) dermal changes, such as superficial perivascular lymphohistiocytic infiltrate and red cell extravasation, with consequent hemosiderin deposition [11], which is responsible for the presence of yellowish-orange areas seen on dermatoscopic examination. The possible presence of dilated dermal blood vessels within inconstantly elongated dermal papillae justifies the detection of focal/irregular dotted vessels on dermatoscopy [11].

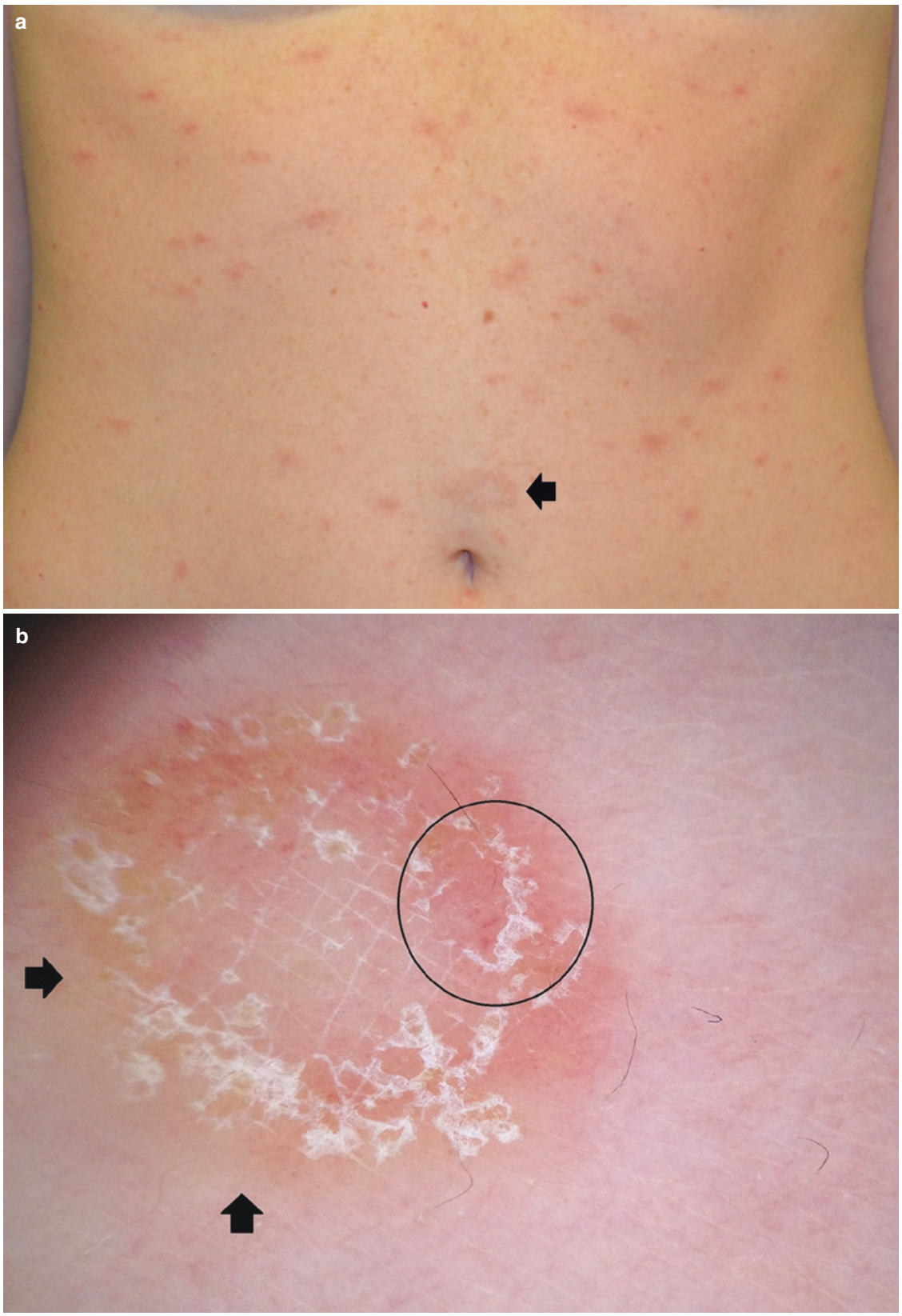

Fig. 17.1 (**a**) Pityriasis rosea. Several roundish, scaling, erythematous lesions on the trunk of a 15-year-old girl; the lesion pointed out by the arrow was the first one to appear ("herald patch"). (**b**) Dermatoscopy (×10) of a lesion: peripheral whitish scaling localized in the border ("collarette" sign) and dotted vessels that are distributed in an irregular or focal pattern (circle). Yellowish-orange structureless areas are also visible (arrows). (**c**) Dermatoscopy (x10) of a lesion with no clinically evident peripheral scaling: the "collarette" sign is detected

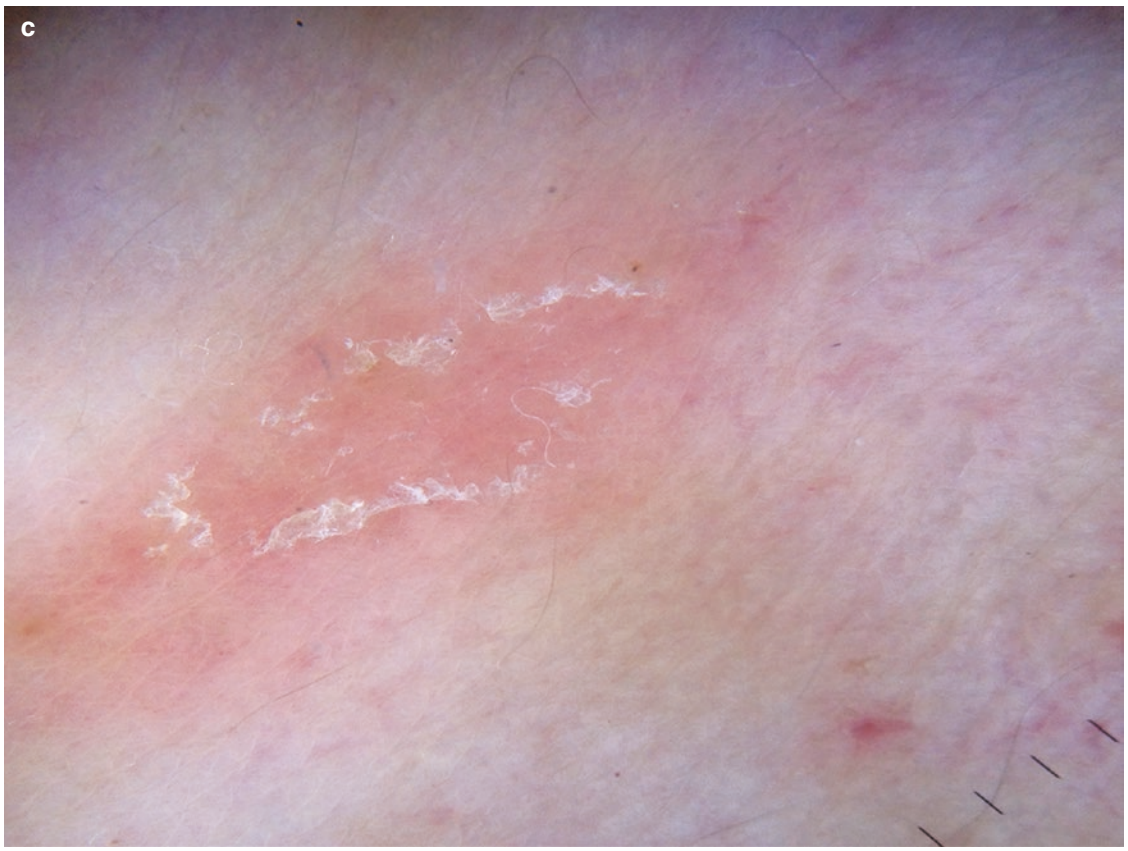

Fig. 17.1 (continued)

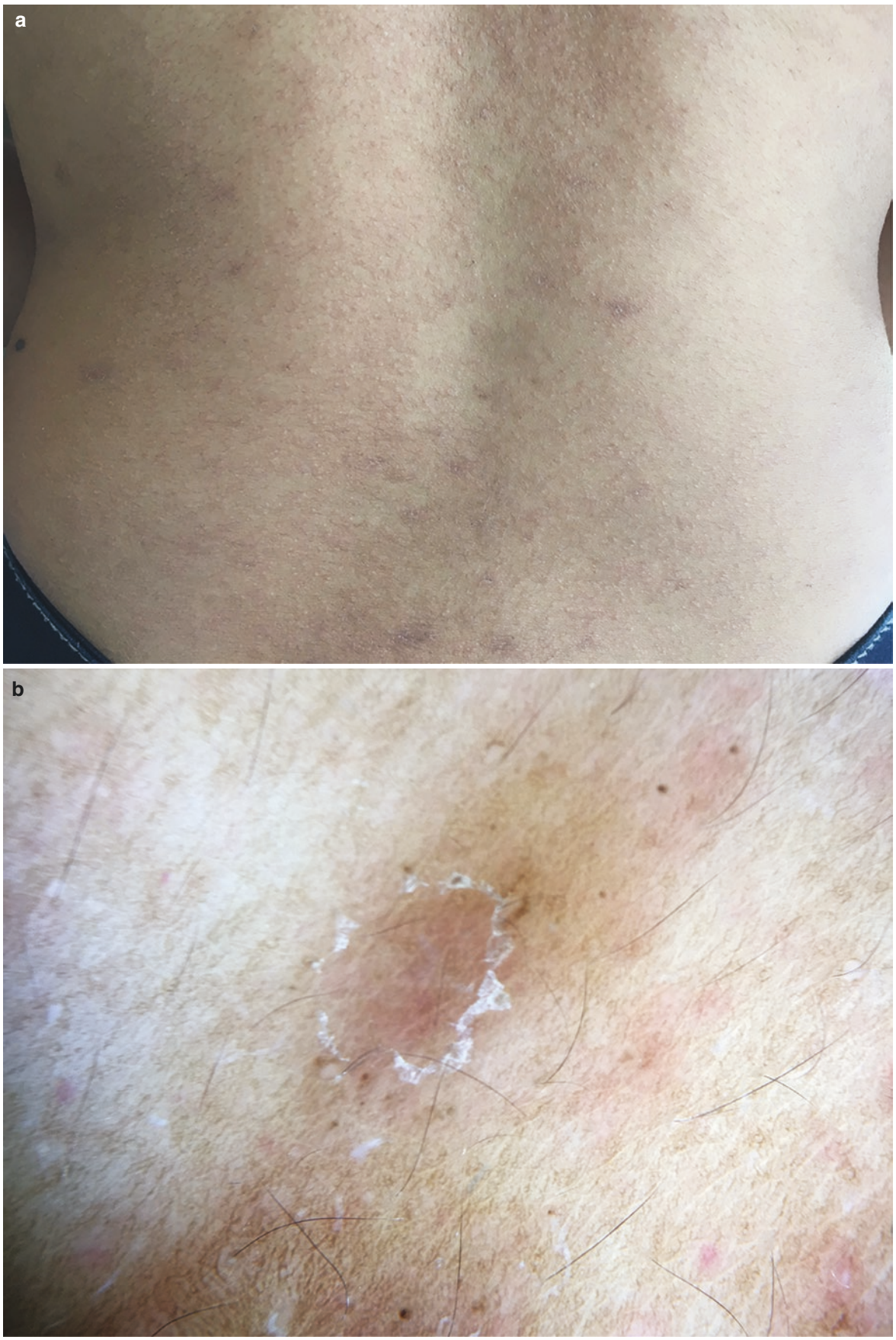

Fig. 17.2 (**a**) Pityriasis rosea. Several scaling brownish lesions on the back in a dark-skinned 14-year-old patient. Many of them do not show the peripheral scaling collarette. (**b**) Dermatoscopy (×10): the "collarette" sign is often the only dermatoscopic clue in dark-skinned patient

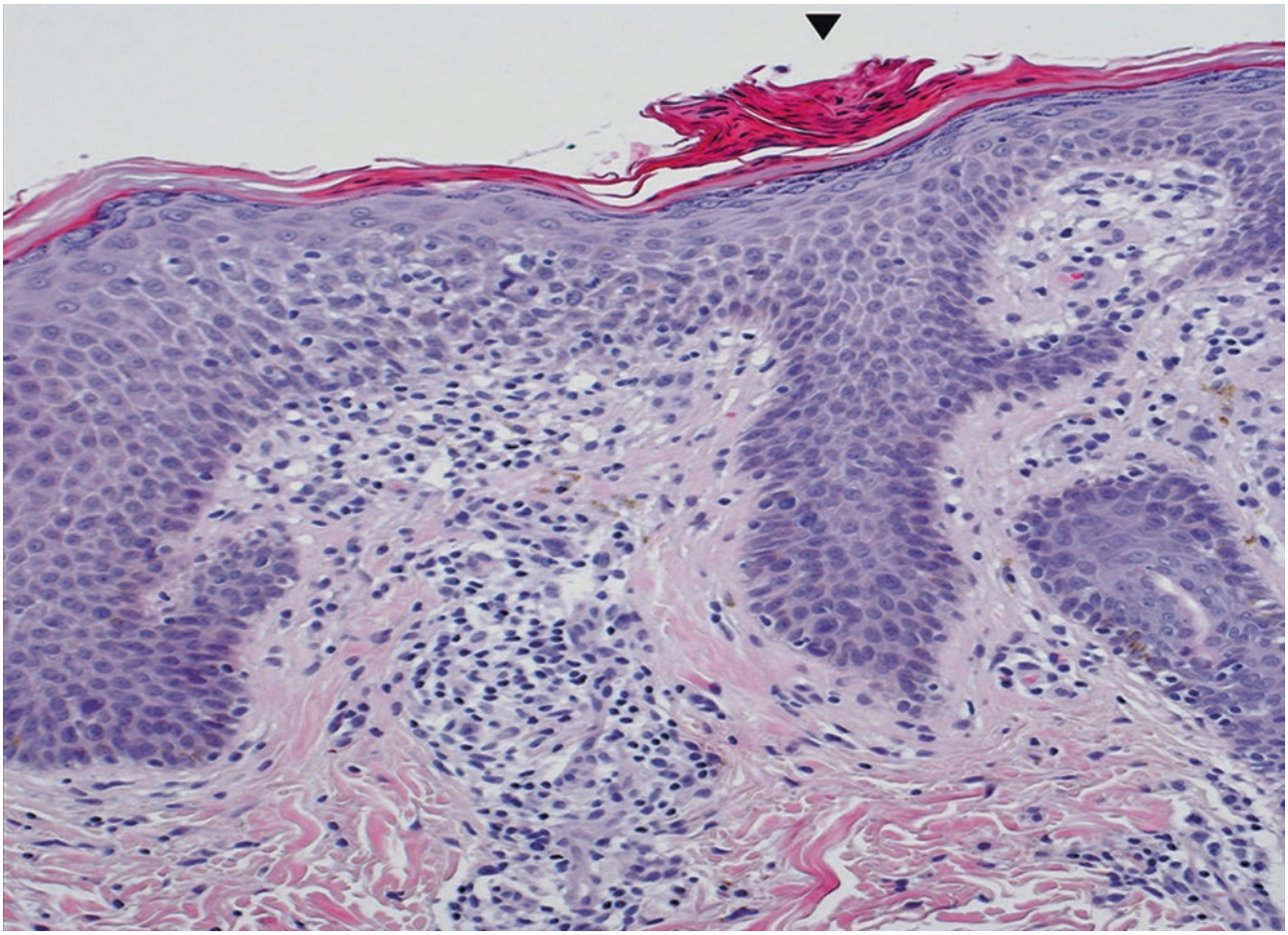

Fig. 17.3 Pityriasis rosea. Histopathology: epidermal changes, i.e., slight acanthosis, focal spongiosis, and focal parakeratosis in mounds (arrowhead), and dermal changes, i.e., superficial perivascular lymphohistiocytic infiltrate and red cell extravasation [H&E staining; magnification ×100]

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Pityriasis Lichenoides

18

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18.1 Definition

Pityriasis lichenoides (PL) is a term used to define a disease spectrum encompassing two main variants, pityriasis lichenoides et varioliformis acuta (PLEVA) and pityriasis lichenoides chronica (PLC), and a third much rarer and aggressive form known as febrile ulceronecrotic Mucha-Habermann disease [1], which is not addressed in this chapter. Despite their names, PLEVA and PLC mainly differ in their clinical and histological features rather than disease duration, as both of them last for an average of 18–20 months [2]. Of note, intermediate or overlapping forms exist [1].

18.2 Epidemiology

Even though PL may affect all ages, it appears more often during the first three decades of life [1]. Epidemiologic data collected from the review of various pediatric cases/series showed that it peaks at 5 and 10 years of age [1]. While no significant gender differences have been found in adult patients, there is a male predominance in the pediatric population; all races are thought to be affected equally [1]. Recent data suggested that PLEVA is the most common pattern in childhood (57% compared with 37% PLC and 6% mixed pattern) [2].

18.3 Etiology

Three main etiopathogenetic theories for both PLC and PLEVA have been suggested: (1) inflammatory reactions induced by infectious agents, (2) inflammatory responses secondary to a T-cell dyscrasia, and (3) immune complex-mediated hypersensitivity vasculitides. Several triggering factors have been reported, mainly including medications, such as chemotherapeutic agents, oral contraceptives, astemizole or herbs, and infections (especially viral) [1].

18.4 Clinical Features

Although both PLEVA and PLC are clinically characterized by a polymorphic appearance, with lesions at various stages of evolution, they exhibit a different lesion morphology [1, 2].

In detail, early PLEVA lesions are asymptomatic, 2–3 mm in diameter, erythematous macules promptly turning into edematous pinkish papules, which may centrally become vesiculopustular, hemorrhagic, necrotic, ulcerated, and/or crusted before spontaneously regressing in a few weeks (Fig. 18.1a); residual dyschromias (mainly hypopigmentation) and varioliform scars are often seen [1]. Trunk, thighs, and upper arms are the most common affected sites [1, 2]. Constitutional symptoms such as fever, headache, malaise, and arthralgia may sometimes precede or accompany cutaneous manifestations [1].

PLC classically presents with asymptomatic, firm, 3–10 mm in diameter, erythematous papules developing a reddish-brown hue and a centrally adherent mica-like scale that may be typically detached “en bloc” by gentle scraping (Fig. 18.2a) [1]; postinflammatory hyper- or hypopigmented macules are often observed [1]. PLC more commonly affects trunk and proximal part of extremities [1, 2].

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18.5 Differential Diagnosis

The main differential diagnoses of PLEVA include chickenpox, arthropod bites, leukocytoclastic vasculitis, and, particularly, lymphomatoid papulosis in its early and late stages, while PLC should mainly be distinguished from guttate psoriasis, pityriasis rosea, lichenoid sarcoidosis, and lichen planus [1, 2].

18.6 Diagnosis

The diagnosis of both PLEVA and PLC is usually suspected on the basis of clinical history/appearance and confirmed by skin biopsy [1, 2]. Dermatoscopy has been demonstrated to facilitate the noninvasive recognition of both such conditions [3, 4].

18.7 Dermatoscopy

Dermatoscopic examination of PLEVA may show a central whitish area or a crust having an amorphous brownish appearance, both surrounded by a well-defined ring of pinpoint and/or linear vessels with a targetoid aspect (Fig. 18.1b). At higher magnification, such vascular structures appear dilated and convoluted, with some of them showing a glomerular pattern or linear arrangement; moreover, nonblanchable reddish globules may also be visible [3].

Turning to PLC, it has been found that orange-yellowish structureless areas and nondotted vessels (including milky-red areas/globules, linear irregular and/or branching vessels) are the most frequent dermatoscopic characteristics of active lesions, as they may be observed in nearly 90% of cases (Fig. 18.2b) [4]. Additional dermatoscopic findings are represented by whitish scaling, focally distributed dotted vessels, and hypopigmented areas. This last feature is probably more typical of long-standing lesions, which often display focal postinflammatory hypopigmentation [4–6].

18.8 Histopathological Correlation

Histologically, PLEVA lesions are typically characterized by dermal lymphohistiocytic infiltrate, extravasated erythrocytes, dermal edema, and several epidermal changes, including disappearance of the dermal-epidermal junction, thinning of the granular layer, confluent parakeratosis, ballooning of keratinocytes, intraepidermal vesiculation, and variable degree of necrosis of keratinocytes [1]. It is likely that these last two histological findings are correlated with the presence of the central whitish area or amorphous brownish crust visible on dermatoscopic examination [3]. On the other hand, the vascular pattern seen by dermatoscopy corresponds to dilation and engorgement of blood vessels and microhemorrhages in the papillary dermis [3].

With regard to PLC, histology usually shows perivascular chronic inflammatory cell infiltrate in the superficial dermis, focal disappearance of the dermal-epidermal interface, slight acanthosis, extravasated erythrocytes (and consequent hemosiderin degradation products deposition), and hyperkeratosis with focal parakeratosis (Fig. 18.3) [1]. These last two features are respectively correlated with orange-yellowish structureless areas and whitish scaling seen on dermatoscopy [4]. Nondotted vascular structures and focal dotted vessels detectable by dermatoscopic examination are likely to be the result of dilatation of superficial dermal vessels and dilated blood vessels within focally elongated dermal papillae, respectively [4].

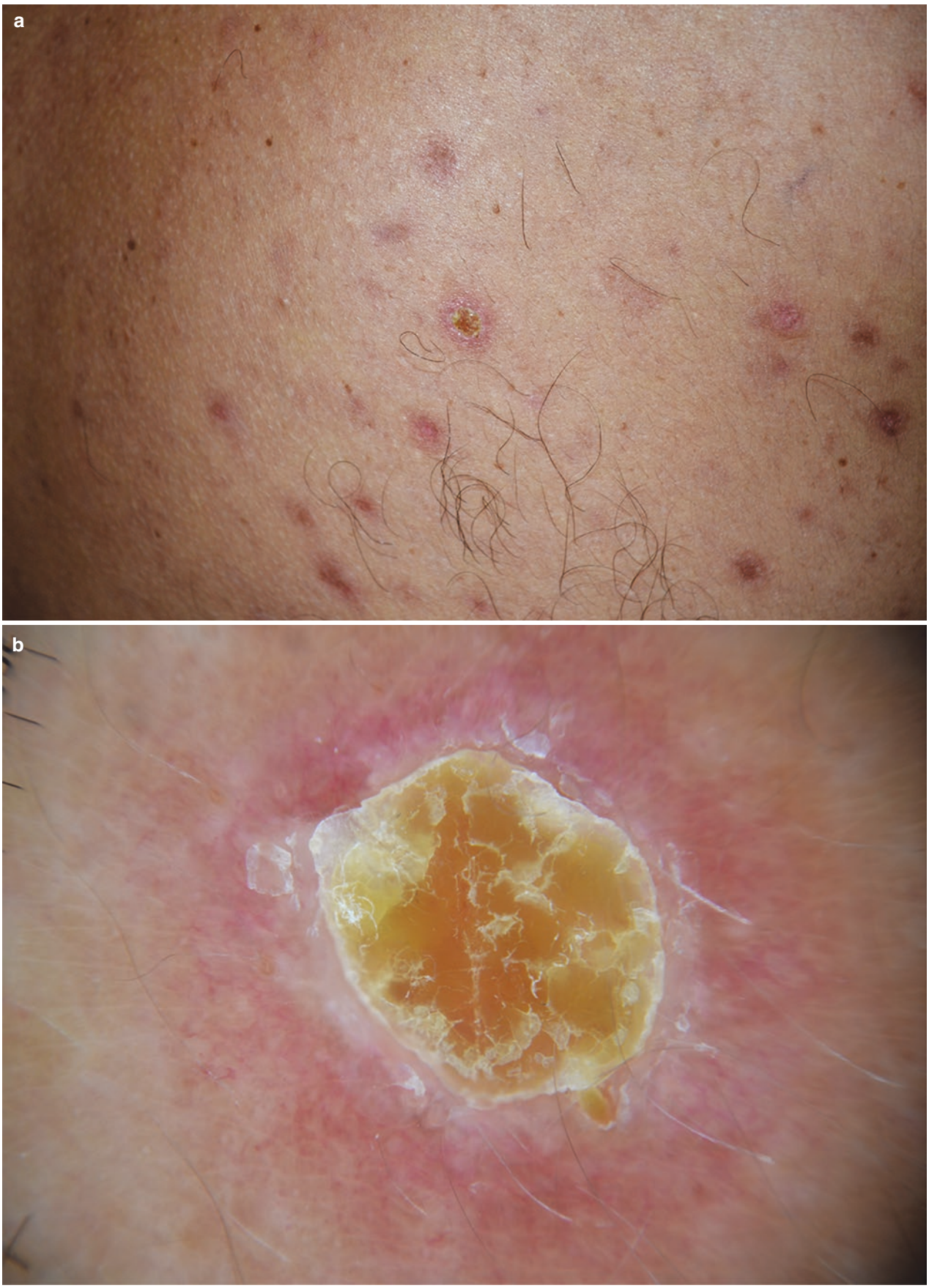

Fig. 18.1 (**a**) Pityriasis lichenoides acuta. Multiple erythematous maculopapular lesions, some of them covered by crusts, located on the trunk of a 16-year-old boy. (**b**) Dermatoscopy (×10): central amorphous yellowish area surrounded by linear vessels with a targetoid aspect (Courtesy of Giuseppe Micali, MD)

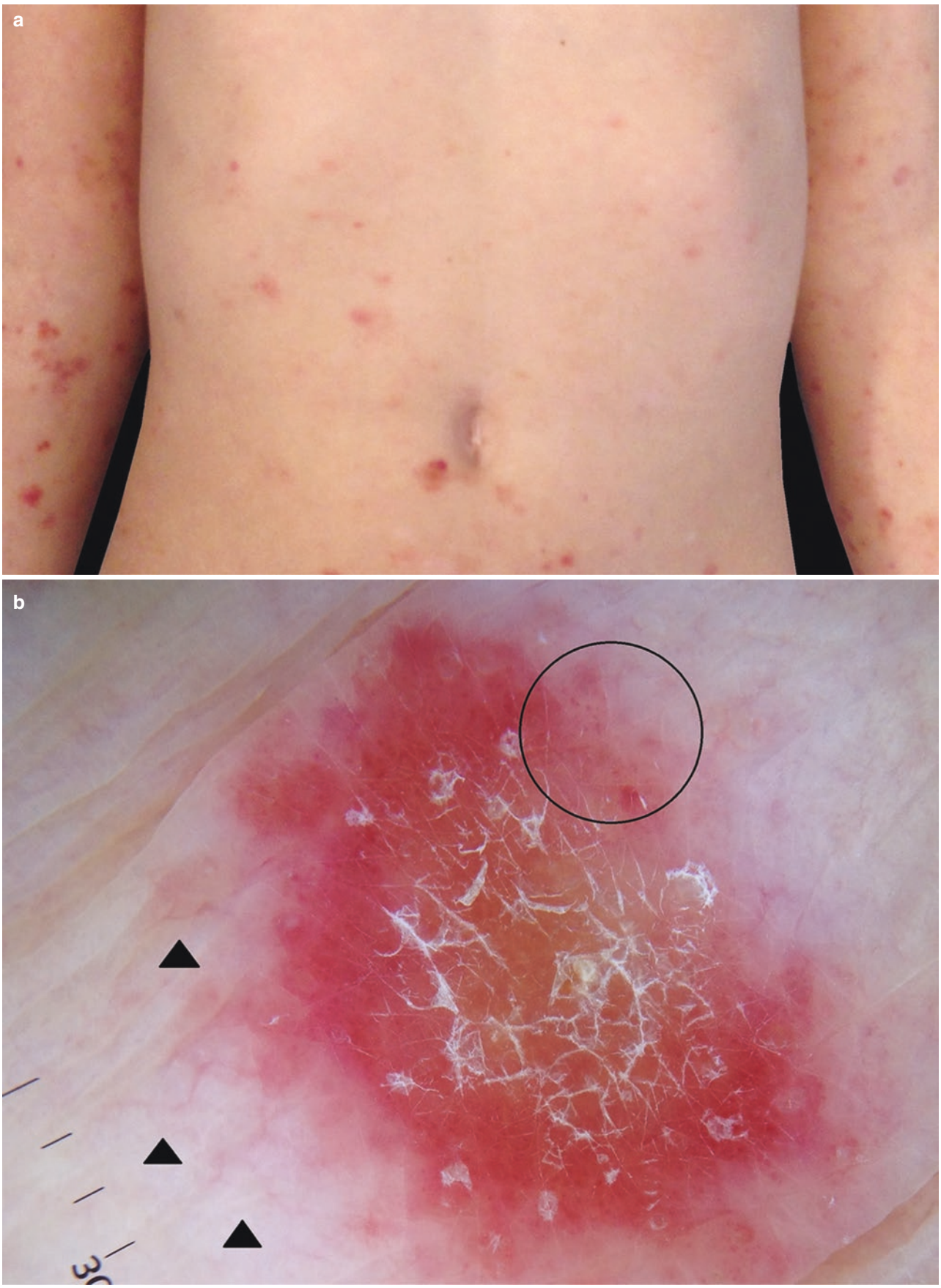

Fig. 18.2 (**a**) Pityriasis lichenoides chronica. Diffuse erythematous brownish papules in an 8-year-old boy. (**b**) Dermatoscopy (×10): central orange-yellowish structureless area, whitish scaling, focally distributed dotted (black circle), and linear irregular/branching vessels (arrowheads) at the periphery

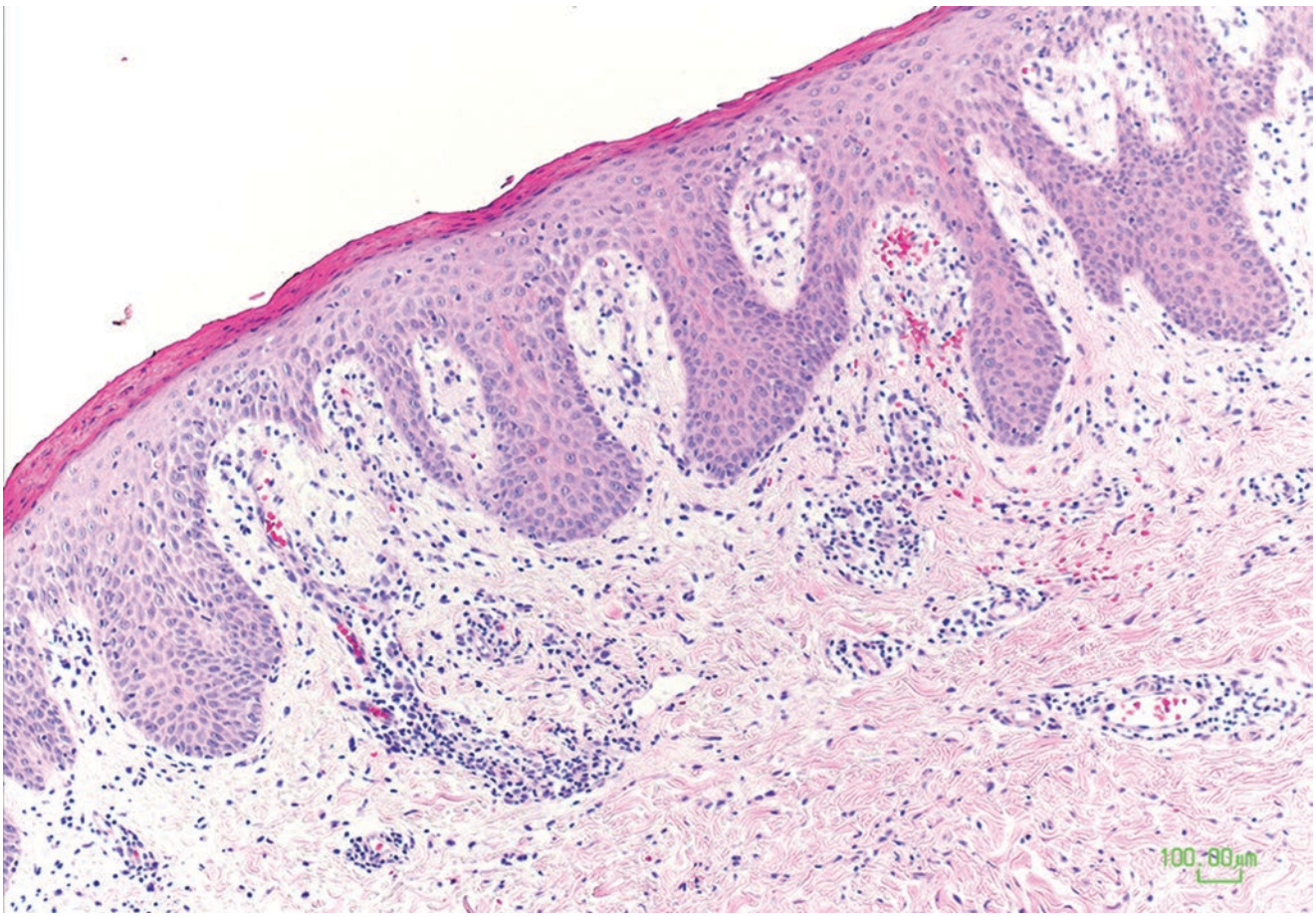

Fig. 18.3 Pityriasis lichenoides chronica. Histopathology: perivascular chronic inflammatory cell infiltrate in the superficial dermis, focal disappearance of the dermal-epidermal interface, slight acanthosis, extravasated erythrocytes, and hyperkeratosis with focal parakeratosis [H&E staining; magnification ×100; courtesy of Carla Di Loreto, MD, Institute of Anatomic Pathology, University of Udine, Italy]

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Darier's Disease

19

Enzo Errichetti and Giuseppe Stinco

19.1 Definition

Darier's disease (DD), also known as keratosis follicularis, is an uncommon autosomal dominantly inherited acantholytic genodermatosis first reported by Darier and White in 1889 [1].

19.2 Epidemiology

The prevalence of DD has been reported to range from 1/30,000 to 1/100,000 [1]. The majority of patients present a positive family history, though spontaneous mutations are not uncommonly seen [1]. Disease expressivity is typically variable, but penetrance is quite high (greater than 95%) [1]. DD usually develops between the ages of 6 and 20 years, albeit several adult-onset cases have been described; neither racial nor gender predilection has been reported [1].

19.3 Etiology

DD is due to mutations in the gene ATP2A2, which encodes the sarcoplasmic/endoplasmic reticulum Ca2+-ATP isoform 2 protein (SERCA2), a pump that maintains a low cytoplasmic Ca2+ level by transporting calcium ions from the cytosol to the lumen of the endoplasmic reticulum [1–3]. Alterations in calcium regulation are thought to affect the synthesis, folding, and/or trafficking of desmosomal proteins, including desmoplakin and cytokeratins 10 and 14, and to impair cell cycle checkpoints [1–3]. High temperatures, high humidity, excessive sweating, pregnancy and delivery, surgery, exposure to UV rays, medications, bacterial/fungal skin infections, and mechanical irritation are known to be exacerbating factors for DD [1].

19.4 Clinical Features

DD presents with discrete, greasy, hyperkeratotic, skin-colored, reddish-brown, or yellowish-brown papules which may sometimes coalesce into crusted plaques; seborrheic areas and skin creases are the main involved sites (Figs. 19.1a and 19.2a) [1]. Symptoms include itch and pain. Nail abnormalities, acral lesions, and mucous membrane changes are commonly detected and may be the presenting signs of the disease [1]. Clinical variants include erosive, vesiculobullous, hyperkeratotic, comedonal, freckled "Groveroid," hypopigmented, and segmental forms [1]. DD may be accompanied by extracutaneous manifestations including psychiatric conditions such as mental retardation, epilepsy, or bipolar disease [1].

19.5 Differential Diagnosis

The main differential diagnoses of DD in pediatric patients include [4] acne, infectious folliculitis, seborrheic dermatitis, follicular eczema, pityriasis lichenoides chronica, and pityriasis rubra pilaris.

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19.6 Diagnosis

Usually DD is diagnosed based on its clinical appearance and positive family history, but it is not uncommonly mistaken for other dermatoses, thus requiring a skin biopsy or genetic confirmation to reach a definitive diagnosis [1, 4]. Recent data suggests that dermatoscopy may be of aid in the noninvasive recognition of DD [5–9].

19.7 Dermatoscopy

In the first dermatoscopic description of DD, "giant pseudocomedones," consisting in dilated oval openings with raised or flat borders and central brown or yellowish hyperkeratotic plugs, were found to be the most frequent feature, with or without a variable vascular pattern, including red dots, red lines, or erythema [5]. However, recent data suggests that DD may display a more complex pattern characterized by polygonal, starlike, or roundish-oval-shaped yellowish/brownish structures, surrounded by a more or less thin whitish halo, on a pinkish homogeneous structureless area, with or without the presence of whitish scales and dotted and/or linear vessels (Figs. 19.1b, c, and 19.2b) [6–8]. Such a pattern has been described in type 1 segmental DD as well [9]. Of note, the starlike structures may also be visible in other dermatological disorders, including Dowling-Degos disease, acantholytic dyskeratotic acanthoma, and, especially, Grover's disease; however, these conditions rarely occur in pediatric population [6, 7].

19.8 Histopathological Correlation

Histologically, DD is characterized by well-defined hyperparakeratotic areas, acanthosis, dyskeratosis with corp ronds and grains, focal acantholysis with suprabasal cleft formation, and superficial dermal chronic inflammation (Fig. 19.3) [8, 9]. In terms of dermatoscopic-pathological correlation, the central yellowish-brownish area and its whitish halo would correspond to the compact hyperkeratosis and acanthosis, respectively, while the pinkish background and vessels may be due to dermal inflammation [8, 9].

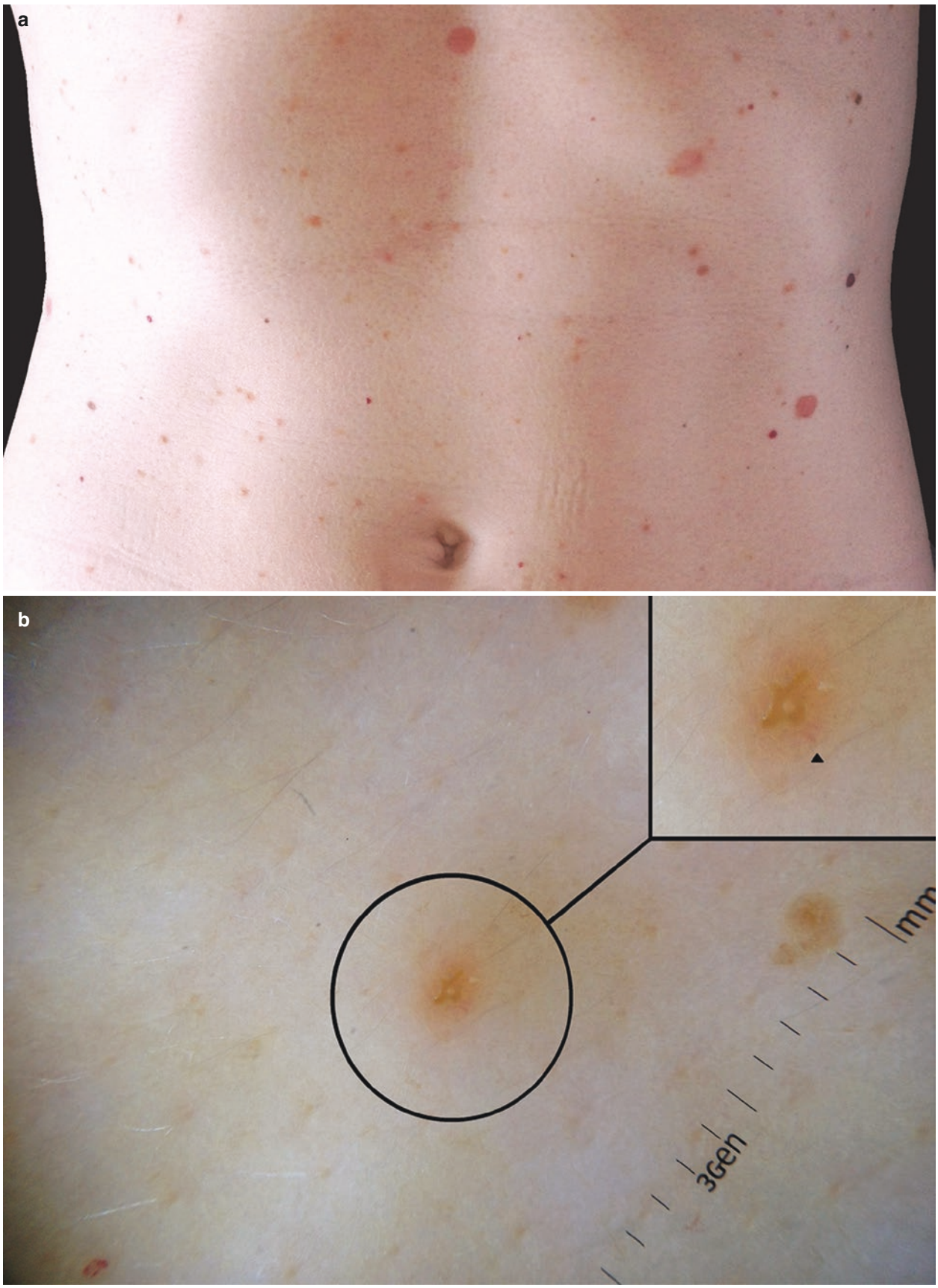

Fig. 19.1 (**a**) Darier's disease. Multiple discrete, hyperkeratotic, yellowish-brown papules on the abdomen of a 16-year-old girl. (**b**) Dermatoscopy (×10) of a lesion: centrally located starlike yellowish/brownish area, surrounded by a thin whitish halo and overlying a pinkish background; few linear vessels are also visible at the periphery (arrowhead in the box). (**c**) Dermatoscopy (×10) of another lesion: central roundish yellowish/brownish area and peripheral whitish halo (box)

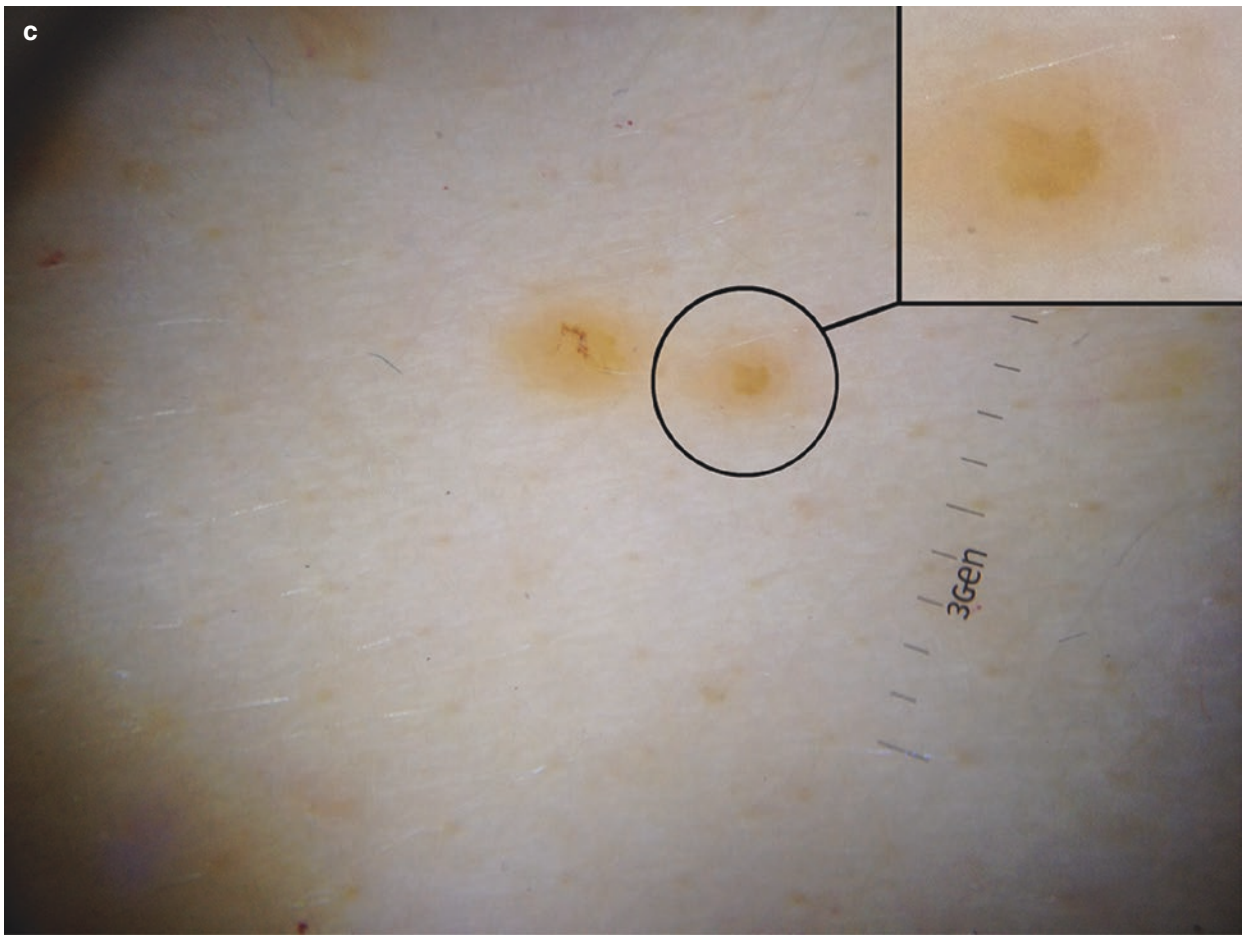

Fig. 19.1 (continued)

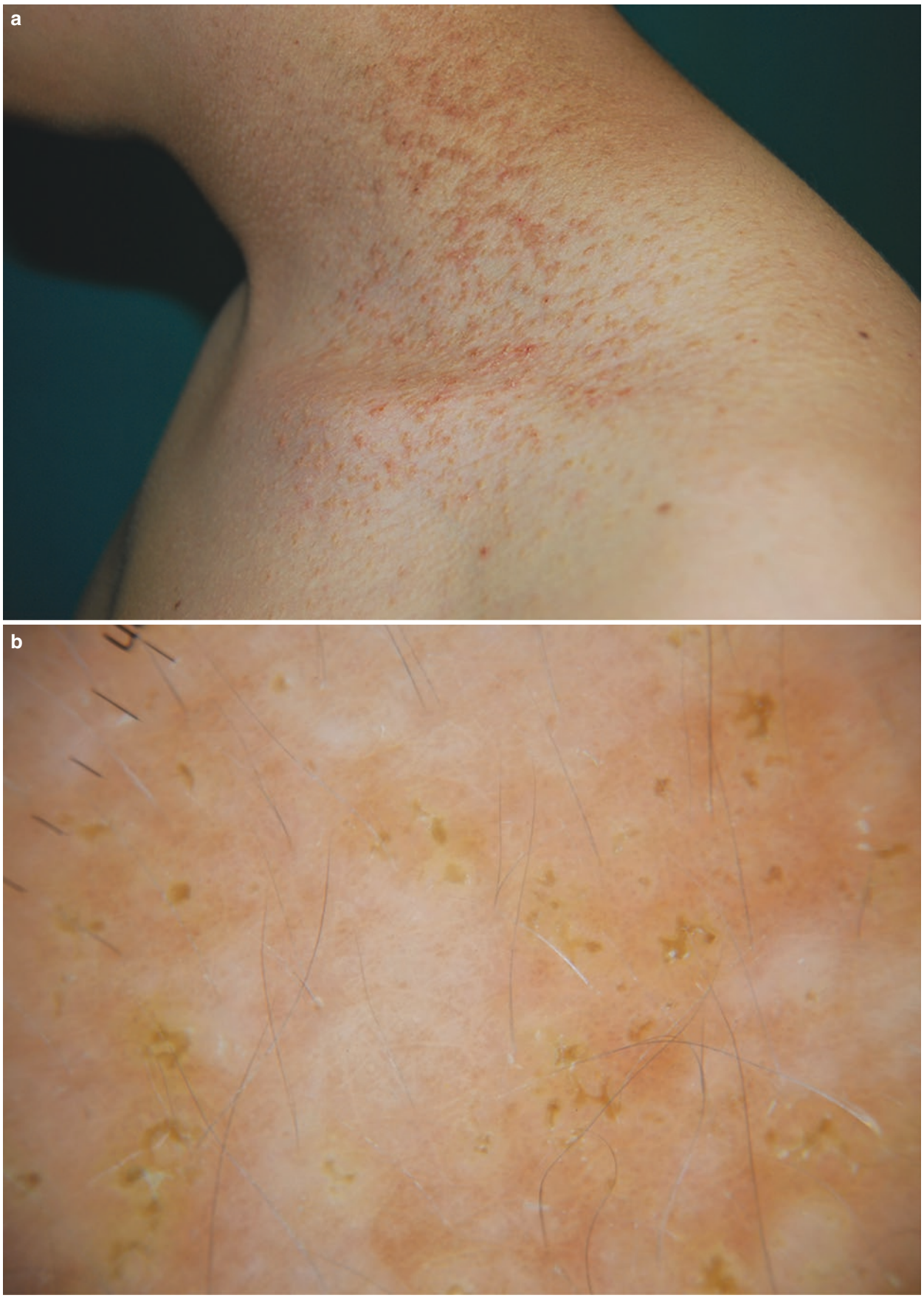

Fig. 19.2 (**a**) Darier's disease. Multiple hyperkeratotic, yellowish-brown papules of the neck in a 15-year-old boy. (**b**) Dermatoscopy (×10): several polygonal, starlike, or roundish-oval-shaped yellowish/brownish areas surrounded by a thin whitish halo (Courtesy of Giuseppe Micali, MD)

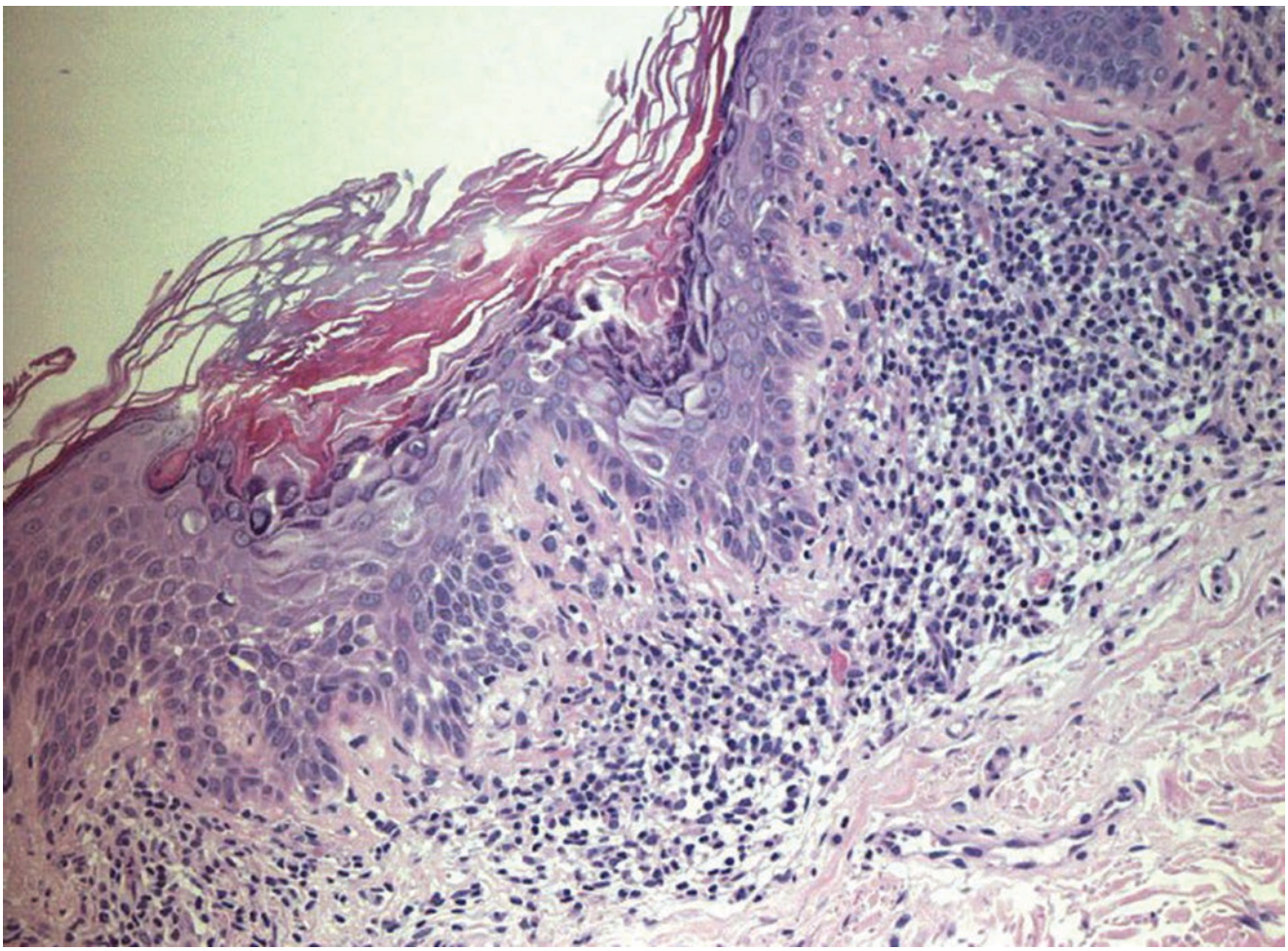

Fig. 19.3 Darier's disease. Histopathology: hyperparakeratotic plugging, acanthosis, focal dyskeratosis, acantholysis, and superficial dermal inflammation [H&E staining; magnification ×100]

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Mastocytosis

20

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20.1 Definition

The term mastocytosis encompasses a spectrum of uncommon disorders characterized by mast cell proliferation and accumulation in one or more organs, with the skin being the most frequently involved one [1–4]. According to World Health Organization (WHO) criteria, there are three cutaneous variants: maculopapular cutaneous mastocytosis [i.e., urticaria pigmentosa (UP) and telangiectasia macularis eruptiva perstans (TMEP)], diffuse cutaneous mastocytosis, and mastocytoma [1–4]. Nodular and plaque-type mastocytoses are no longer recognized as separate entities, but they are now considered as clinical subtypes of either maculopapular cutaneous mastocytosis or mastocytoma (based on the number of the lesions – see clinical features section) [1–4].

20.2 Epidemiology

Even though the real incidence of cutaneous mastocytosis is unknown, such a condition is thought to represent about 0.1–0.8% of all new dermatological consultations [1–4]. Apart from TMEP, which mainly affects young adults, the majority of cases occurs during infancy or early childhood and usually resolves by puberty; there is no preference for gender or race [1].

20.3 Etiology

There is growing evidence that mast cell accumulation in tissues is the direct consequence of activating mutations (especially D816V mutation) in the proto-oncogene KIT, which encodes the receptor for mast/stem cell growth factor (c-KIT) [1]. However, such mutations are not always detected in the pediatric population, so etiology and pathogenesis of mastocytosis in these cases remains to be determined [1–4]. Interestingly, aberrant mast cells present in skin lesions release, among other mediators, a significant amount of mast/stem cell growth factor (c-KIT ligand), which in turn stimulates both mast cell and melanocyte proliferation and melanogenesis, thereby explaining the common detection of skin hyperpigmentation in cutaneous mastocytosis lesions [1].

20.4 Clinical Features

UP presents with red-brownish maculae, papules, nodules, and/or plaques with various sizes [1–3]. Differently from adults, which display small (3–5 mm in diameter), macular/slightly palpable papular lesions mainly affecting the trunk and thighs, involvement in children is usually more extensive, with lesions being more hyperpigmented than erythematous, larger (between 5 and 15 mm in size), and more raised (Fig. 20.1a) [1, 3]. Especially in children (because of the higher number of lesional mast cells compared to adults), lesions may become erythematous and/or urticated after scratching or rubbing (Darier's sign) [1]. Such a sign, however, is not pathognomonic for cutaneous mastocytosis as it may be seen in other conditions, such as leukemia cutis, juvenile xanthogranuloma, and Langerhans cell histiocytosis [5].

TMEP is clinically characterized by telangiectasic, brownish-erythematous macules with irregular borders and a diameter ranging from 2 to 6 mm; the chest and limbs are the most commonly involved sites [1, 6]. Darier's sign is usually absent [1, 6].

Diffuse cutaneous mastocytosis presents with a diffuse leathery (pachydermatous) thickening of the skin that may be erythematous or yellow-brown in color ("peau d'orange" or "peau de chagrine" appearance); nodules may arise at sites of dense mast cell infiltrates [1, 7]. Moreover, blisters

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commonly develop spontaneously or after minor trauma or scratching (Darier's sign) [1, 7].

Mastocytoma typically presents as one or few (up to five) tan-brown nodular lesions mainly involving the distal extremities (Figs. 20.2a, 20.3a and 20.4a) [1, 3]. Darier's sign is typically positive (Fig. 20.2b). Occasionally, attacks of flushing can be induced by rubbing a solitary mastocytoma [1, 3].

20.5 Differential Diagnosis

The differential diagnosis of maculopapular cutaneous mastocytosis (i.e., UP and TMEP) includes multiple melanocytic nevi, Langerhans cell histiocytosis, juvenile xanthogranulomas, nodular scabies, café au lait spots, multiple cutaneous lentiginosis, and postinflammatory hyperpigmentation [1, 8]. Diffuse cutaneous mastocytosis should be differentiated from generalized bullous diseases, diffuse smooth muscle hamartoma (Michelin tire baby syndrome), bullous congenital ichthyosiform erythroderma, incontinentia pigmenti, and acrodermatitis enteropathica [1, 7, 8]. Finally, mastocytoma may resemble juvenile xanthogranuloma, Spitz nevus, pseudolymphoma/lymphocytoma cutis, congenital melanocytic nevus, and café au lait spots (resolving lesions) [1, 8].

20.6 Diagnosis

Diagnosis of the various forms of cutaneous mastocytosis in pediatric populations usually relies on the typical clinical features, measurement of serum tryptase concentrations, and/or the presence of Darier's sign (especially for pediatric UP, diffuse cutaneous mastocytosis, and mastocytoma) [1–4]. However, because of normal serum tryptase levels, lacking of Darier's sign and/or atypical clinical presentation, the distinction from other similar dermatoses may sometimes be challenging, thus requiring histological assessment to reach a definitive diagnosis [1–4]. Recently, dermatoscopy has been showed to be a useful tool in assisting the noninvasive diagnosis of both maculopapular cutaneous mastocytosis and mastocytoma [9–13]; on the other hand, there is no report on dermatoscopy of diffuse cutaneous mastocytosis.

20.7 Dermatoscopy

The dermatoscopic pattern of cutaneous mastocytosis varies according to the disease subtype [9–13]. For maculopapular variants, the most frequent dermatoscopic finding of UP is represented by a homogeneous light-brown pigmentation (especially in recent lesions) and/or pigment network (especially in older mature lesions) (Figs. 20.1b, c). TMEP is typically characterized by reticular vessels on an erythematous/brownish base ("reticular vascular" pattern), sometimes associated with a brownish network [9–13]. Dermatoscopy, however, does not provide a reliable differentiation of these two conditions as, although, rarely, UP may show a reticular vascular pattern [10]. Interestingly, in a relatively recent study, the presence of such a pattern, along with serum tryptase levels and plaque-type lesions, would represent the best combination to predict the need for maintained antimediator therapy [9]. Less common vascular findings visible in both UP and TMEP include erythematous halo, sparse dotted vessels, and thin tortuous linear vessels [9–14]. Finally, UP nodules/plaques may sometimes display yellow-orange blots [9].

As regards mastocytoma, a study conducted on 11 cases has demonstrated the presence of diffuse or multifocal yellow-orange areas (yellow-orange blots) (Fig. 20.3b) as a constant feature being found in 100% of cases, with no other dermatoscopic findings observed [9]. However, such results should be interpreted cautiously as the authors considered only patients with solitary lesions, while subjects with more than one lesion were included in the group of nodular/plaque mastocytosis [9]. In our experience, we found in solitary mastocytoma additional less common features, i.e., diffuse light-brown discoloration and brownish network-like appearance especially in regressing phases (Fig. 20.4b).

20.8 Histopathological Correlation

Both UP and TMEP are characterized by dermal mast cell infiltrates, which are more dense and diffusely distributed in the papillary dermis in the former and more subtle and mainly restricted to superficial perivascular areas in the latter [14]. Additionally, basal cell hyperpigmentation of the overlying epidermis is a common feature in maculopapular cutaneous mastocytosis, especially UP [14]. Of note, such a hyperpigmentation may be subtle and involve diffusely the basal layer, thus giving rise to the homogeneous brownish discoloration seen on dermatoscopy, or may be more marked on the rete ridges, thereby explaining the dermatoscopic detection of pigment network pattern in some lesions [9, 14]. Interestingly, basal cell hyperpigmentation is more prominent in older lesions in the maculopapular variants and less so in mastocytomas [14]. This might explain the more frequent presence of pigmented reticular reinforcement in older UP lesions and detection of brownish areas in regressing mastocytomas [14]. Regarding the latter condition, the main histological feature includes the presence of tumorlike dense mast cell deposits filling the entire dermis and often extending into the subcutis, which are likely to correspond to the yellow-orange areas visible on dermatoscopy (Fig. 20.5) [9, 14].

Finally, vessels seen on dermatoscopic examination in cutaneous mastocytosis correspond histologically to dilatation and vascular proliferation induced by mast cell mediators [9, 14].

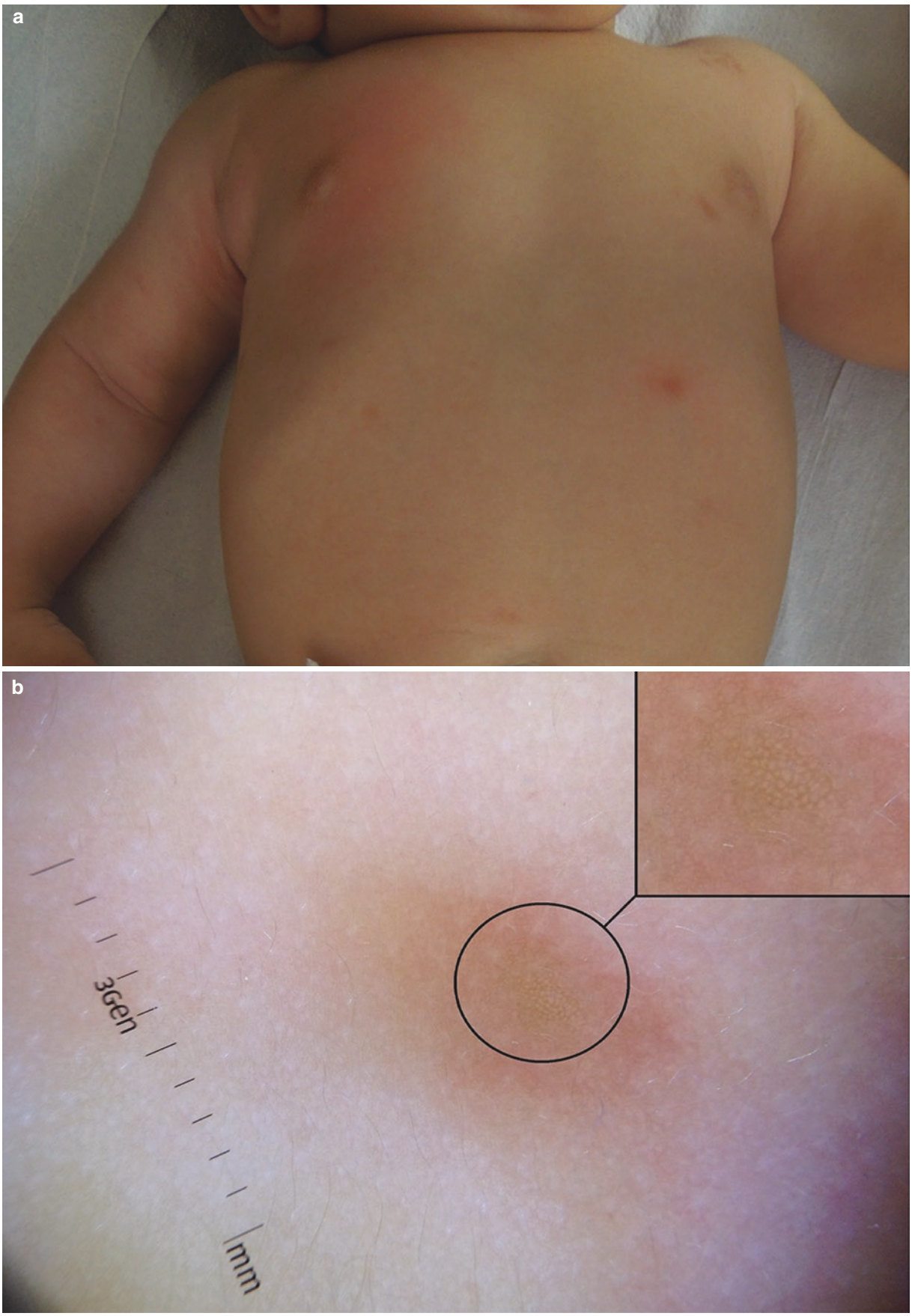
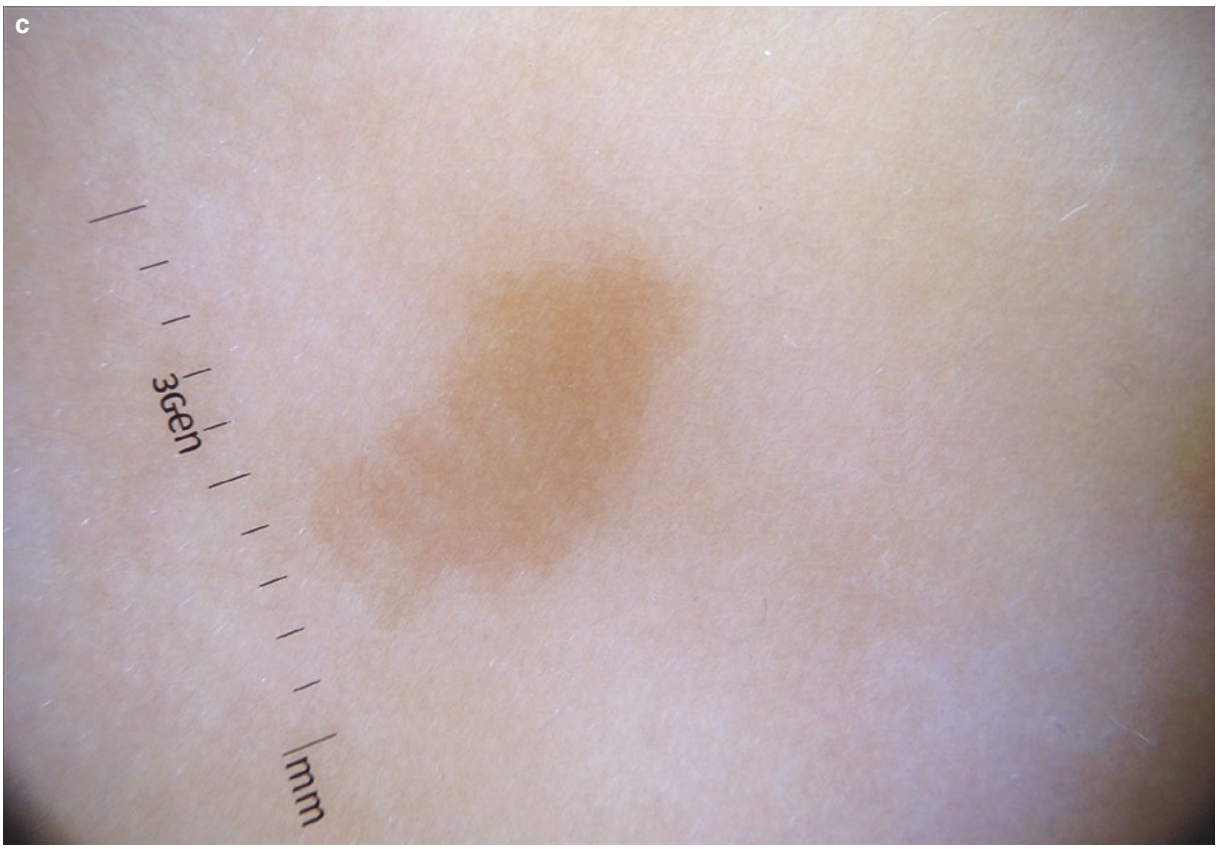


Fig. 20.1 (**a**) Urticaria pigmentosa. Diffuse brownish papules in a child. (**b**) Dermatoscopy (×10) of a mature papule: diffuse brownish discoloration along with a pigmented reticular reinforcement (box). (**c**) Dermatoscopy (×10) of a recent lesion: diffuse brownish background



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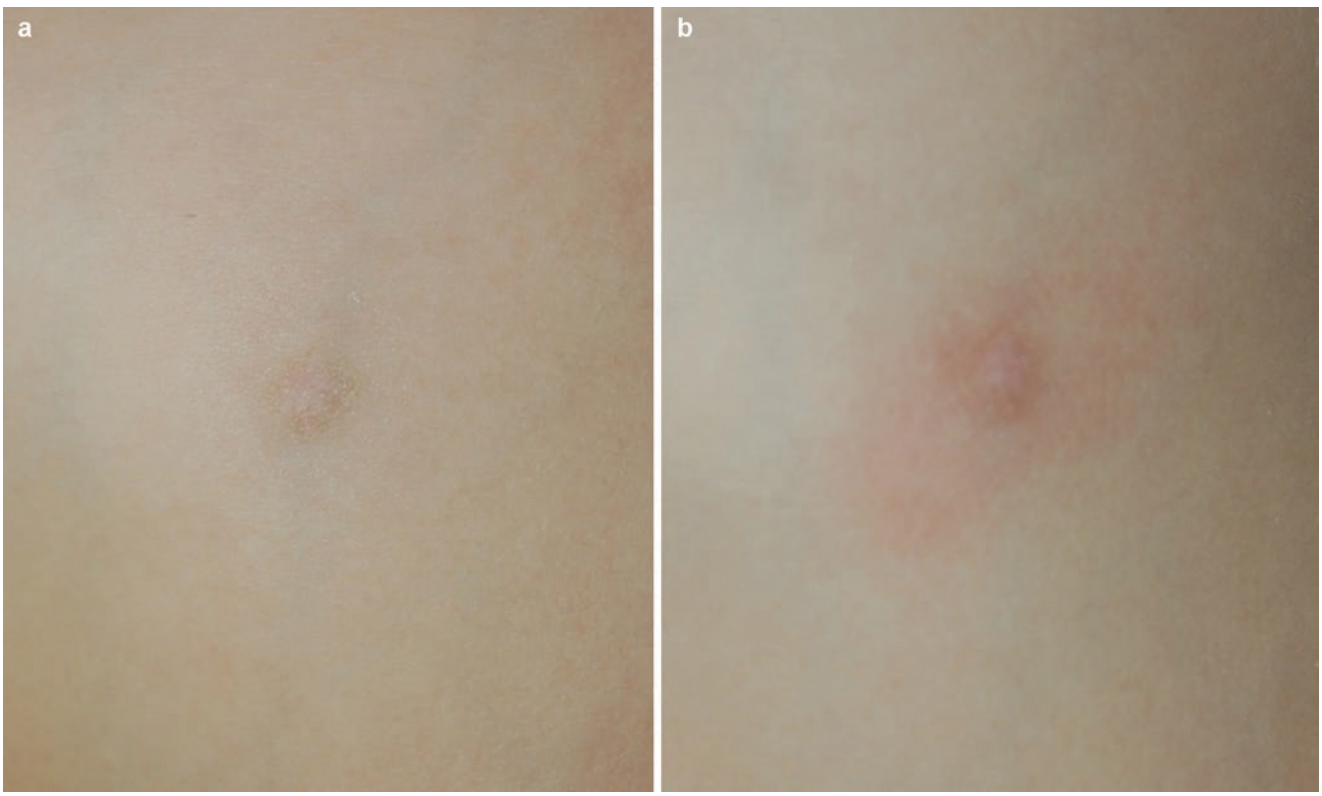

Fig. 20.2 Mastocytoma. (**a**) Slightly palpable papule on the abdomen. (**b**) The lesion becomes erythematous and urticated after scratching [Darier's sign] (Courtesy of Giuseppe Micali, MD)

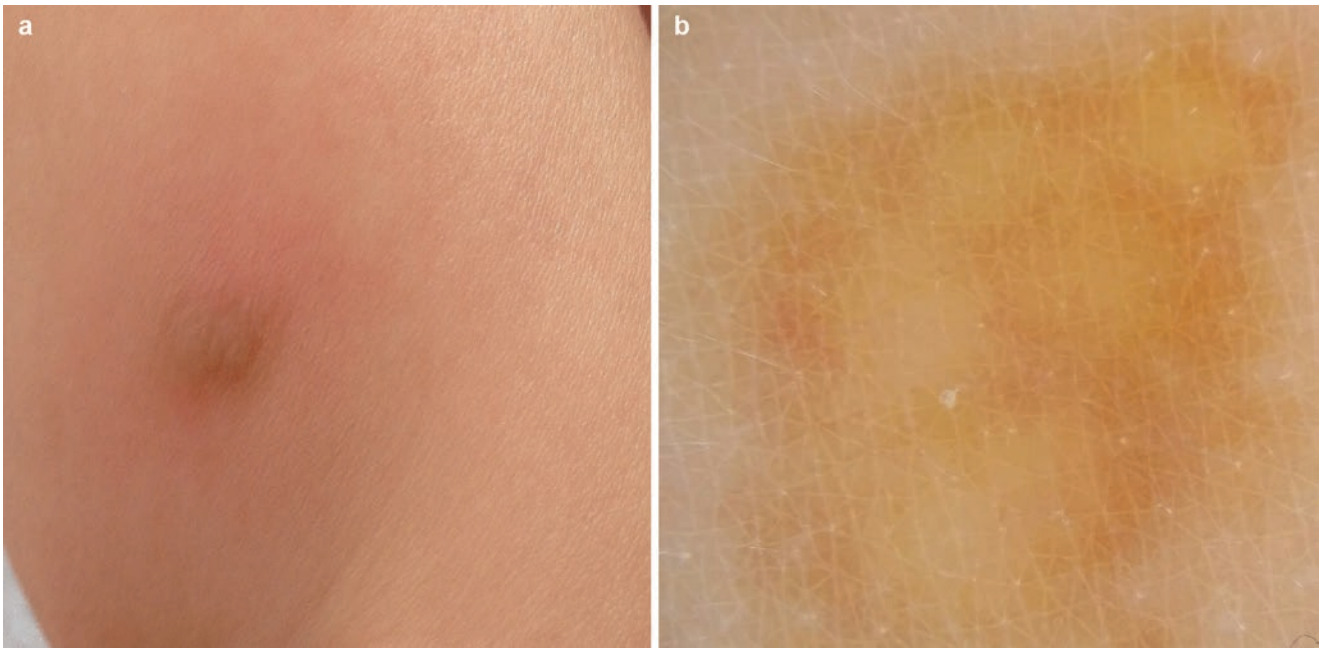

Fig. 20.3 (**a**) Mature mastocytoma presenting as a brownish nodular lesion on the left lower limb of a 6-month-old infant. (**b**) Dermatoscopy (×10): multifocal yellow-orange areas (yellow-orange blots) and peripheral brownish background

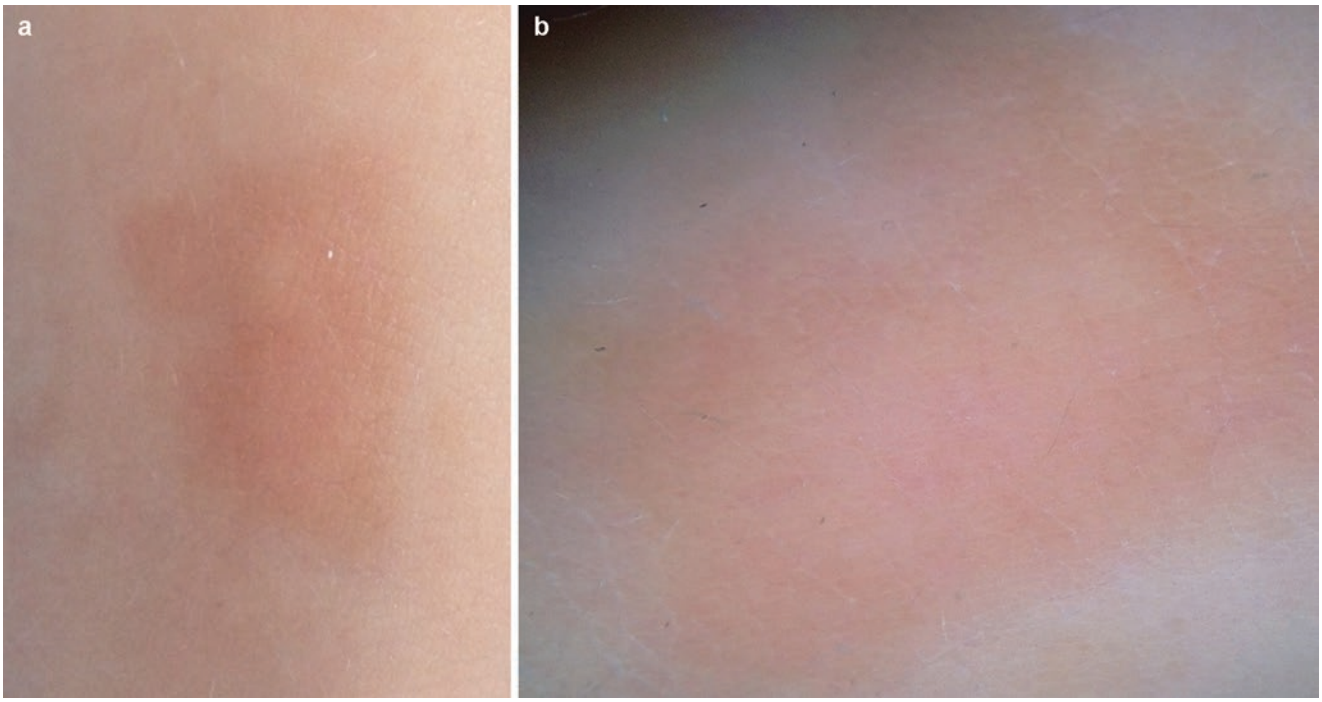

Fig. 20.4 (**a**) Healing mastocytoma on the right leg of a 5-year-old girl. (**b**) Dermatoscopy (×10): light-brown discoloration with a slight erythema in the center of the lesion

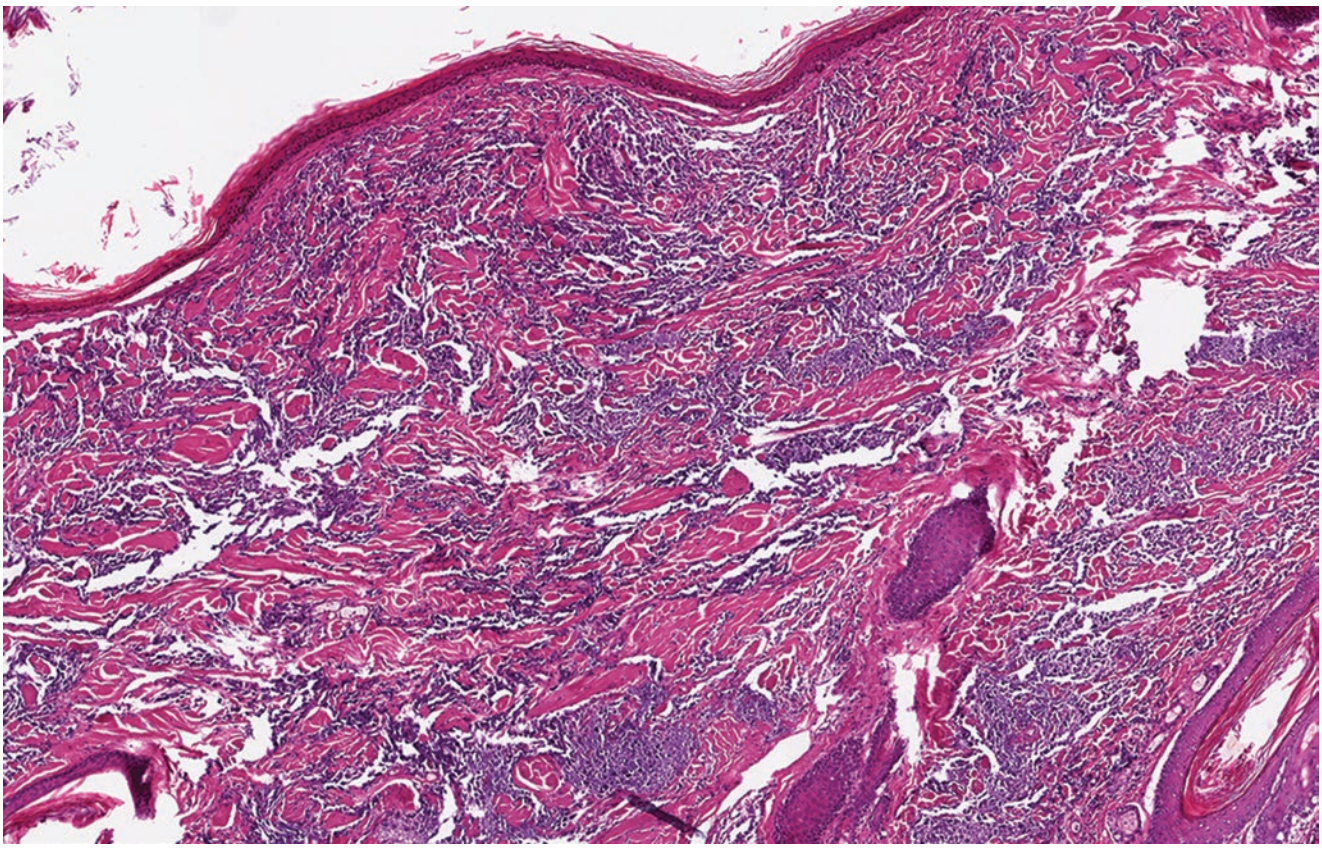

Fig. 20.5 Mastocytoma. Histopathology: dermal mast cell infiltrate diffusely distributed in the papillary dermis and basal cell hyperpigmentation (Courtesy of Giuseppe Micali, MD) [H&E staining; magnification ×100]

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Part V

Appendage Disorders

Alopecia Areata

21

Francesco Lacarrubba, Anna Elisa Verzì,
Federica Dall'Oglio, and Giuseppe Micali

21.1 Definition

Alopecia areata is a non-scarring hair loss disorder characterized by an unpredictable course, with possible spontaneous regrowth and sudden relapse at any given time.

21.2 Epidemiology

Overall, it is considered a common type of hair loss being responsible for 0.7–3.8% of dermatology referrals [1]. Its incidence is estimated approximately 2% in the general population [2]. In the pediatric population, alopecia areata occurs in 26.2% of patient complaining of hair loss [3]. Males and females appear to be equally affected [4, 5]. A study of 191 patients showed regrowth in roughly 40–70% of patients in those classified as limited-patch stage [6]. Unfortunately, compared with adults, early onset of alopecia areata in childhood may result in a greater degree of disease and further progression [6]. In approximately 16% of patients with alopecia areata, a personal history of concomitant autoimmune disorders, such as thyroid dysfunction, diabetes mellitus, vitiligo, and psoriasis, is revealed [4, 7].

21.3 Etiology

The exact etiology of alopecia areata is still unknown. The current evidence supports an immune-mediated origin involving T-cells, and strong genetic contribution, as confirmed by familiar cases [8, 9]. Environmental influences (e.g., viral infections, trauma, stress) may be responsible for triggering the disease [10].

21.4 Clinical Features

The typical clinical manifestation is a circular, well-circumscribed, patchy area of hair loss without any signs of inflammation and scarring (Figs. 21.1a, 21.2a, 21.3a, and 21.4a). Small "exclamation-mark" hairs at the periphery can usually be noted. The skin within the bald patch appears normal or slightly reddened. The scalp is the most involved site, but the disease may occur in any hair-bearing regions (eyebrow, eyelash, facial, extremity, axillary, and pubic areas) [11]. The exclusive involvement of the lower occipital scalp in a band-like pattern is known as ophiasis pattern alopecia areata. In some cases, patches may become confluent leading to total scalp hair loss (alopecia totalis) (Fig. 21.5a). Alopecia universalis occurs when there is a complete scalp and body hair loss. Regrowth is often unpigmented, but the hairs gradually resume their normal caliber and color. Nail abnormalities may also occur.

21.5 Differential Diagnosis

Multiple hair loss disorders can mimic alopecia areata. In children, trichotillomania and tinea capitis represent the main disorders that should be ruled out.

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21.6 Diagnosis

The diagnosis of alopecia areata is usually at clinical observation [9]. In active expanding alopecia patches, the hair-pull test may be positive at periphery.

21.7 Dermatoscopy

In alopecia areata, dermatoscopy may be very useful both for diagnostic purpose and for therapeutic monitoring [12–16].

The typical dermatoscopic features include the presence of dystrophic hair shafts, yellow dots, and hypopigmented vellus hairs (Figs. 21.1b, 21.2b, 21.3b, 21.4b, and 21.5b) [13–16]. Dystrophic hair shafts may appear as broken hairs, exclamation-mark hairs, and black dots, are indicative of anagen alterations, and correlate with strong disease activity. In particular, exclamation-mark hairs (1–5 mm long, appearing with thin proximal ends and thicker distal ends) represent the most specific signs of acute alopecia areata [14]. Black dots may be also observed in children in trichotillomania and tinea capitis. Yellow dots are commonly seen both in acute and chronic forms of alopecia areata and are usually regularly distributed within the alopecic patch. They may be observed in children in congenital hypotrichoses. The recognition of vellus hairs (sometimes appearing as coiled hairs) (Fig. 21.4b) represents an early sign of regrowing (disease remission) [13].

21.8 Histopathological Correlation

Hair follicles exhibit both abnormal hair cycling and inflammation. T-cells attack anagen hair follicles, which are subsequently maintained in a dystrophic state. This results in hair production of inappropriate integrity and size, corresponding to broken hairs and exclamation-mark hairs seen in dermatoscopy, as well as incompletely differentiated hair shaft remnants lodged in the follicular infundibulum, corresponding to black dots. Moreover, follicular infundibula appear dilated and filled with sebum and degraded keratinous material, dermatoscopically corresponding to yellow dots [15].

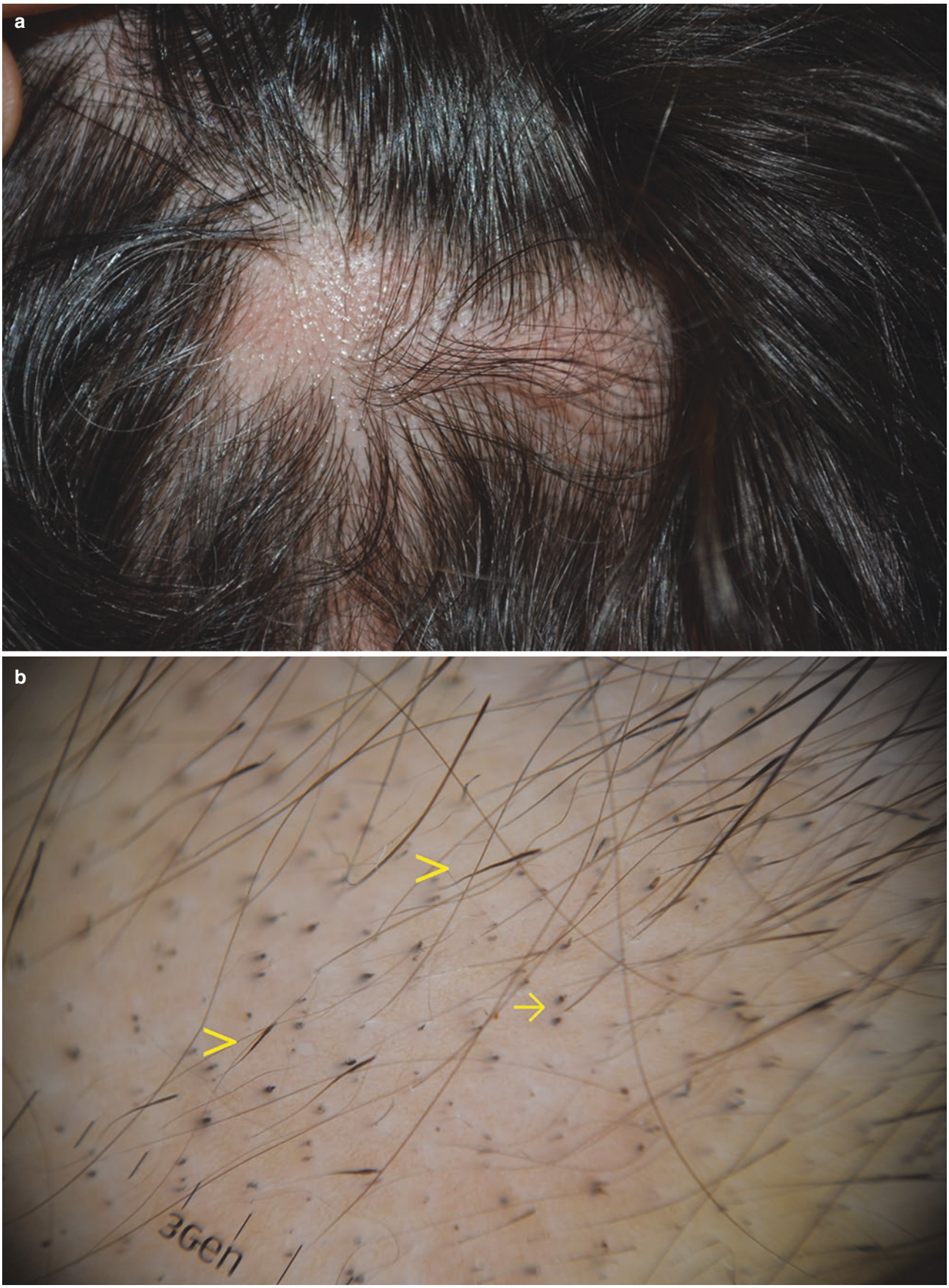

Fig. 21.1 (**a**) Alopecia areata. Patchy areas of hair loss in a 14-year-old girl. (**b**) Dermatoscopy (×10): several *black dots* (arrow) and *exclamation-mark hairs* (arrowheads)

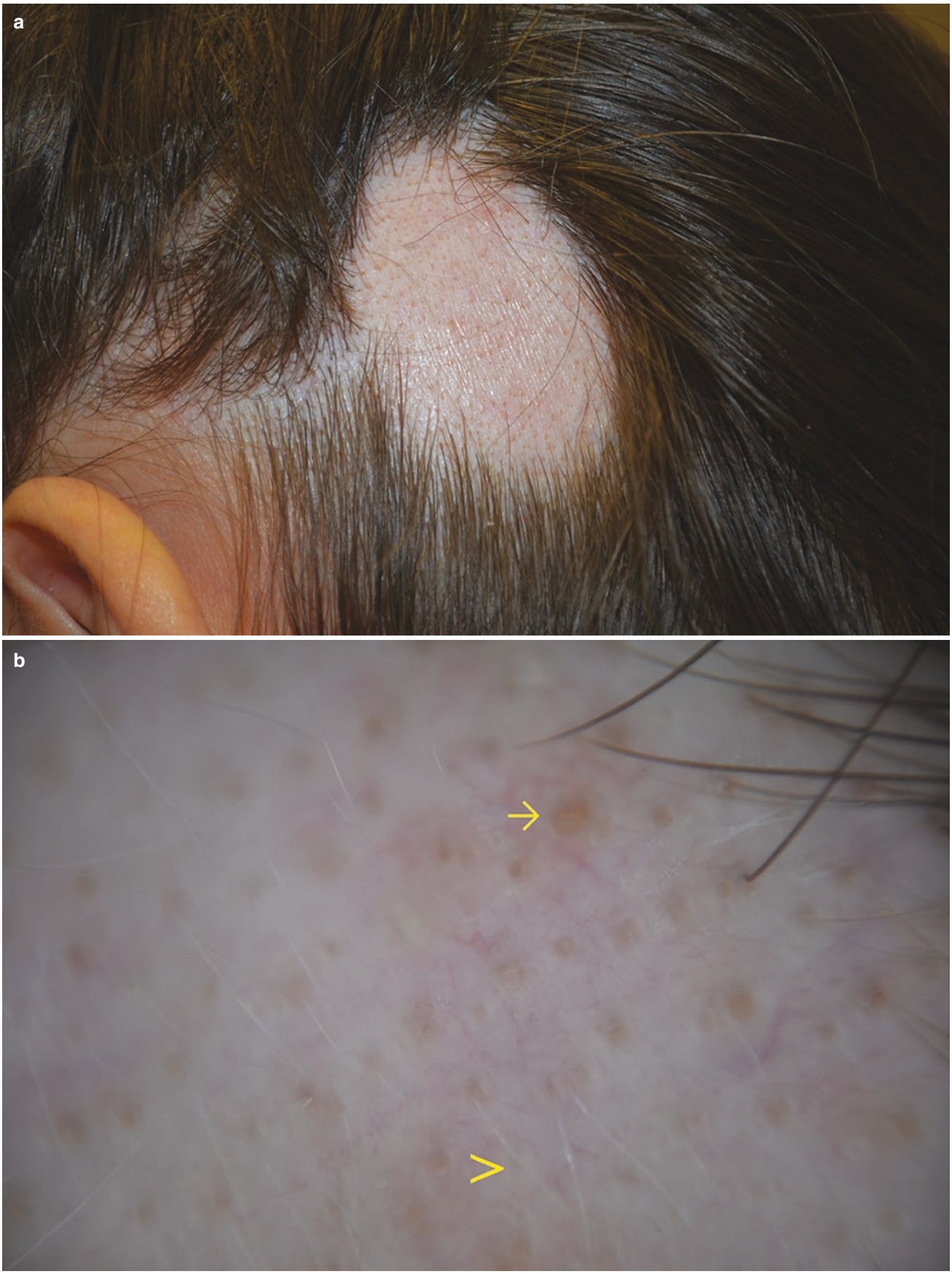

Fig. 21.2 (**a**) Alopecia areata. Circular, well-defined, patchy area of hair loss in a 12-year-old girl. (**b**) Dermatoscopy (×10): multiple *yellow dots* (arrow) and hypopigmented *vellus hairs* (arrowhead)

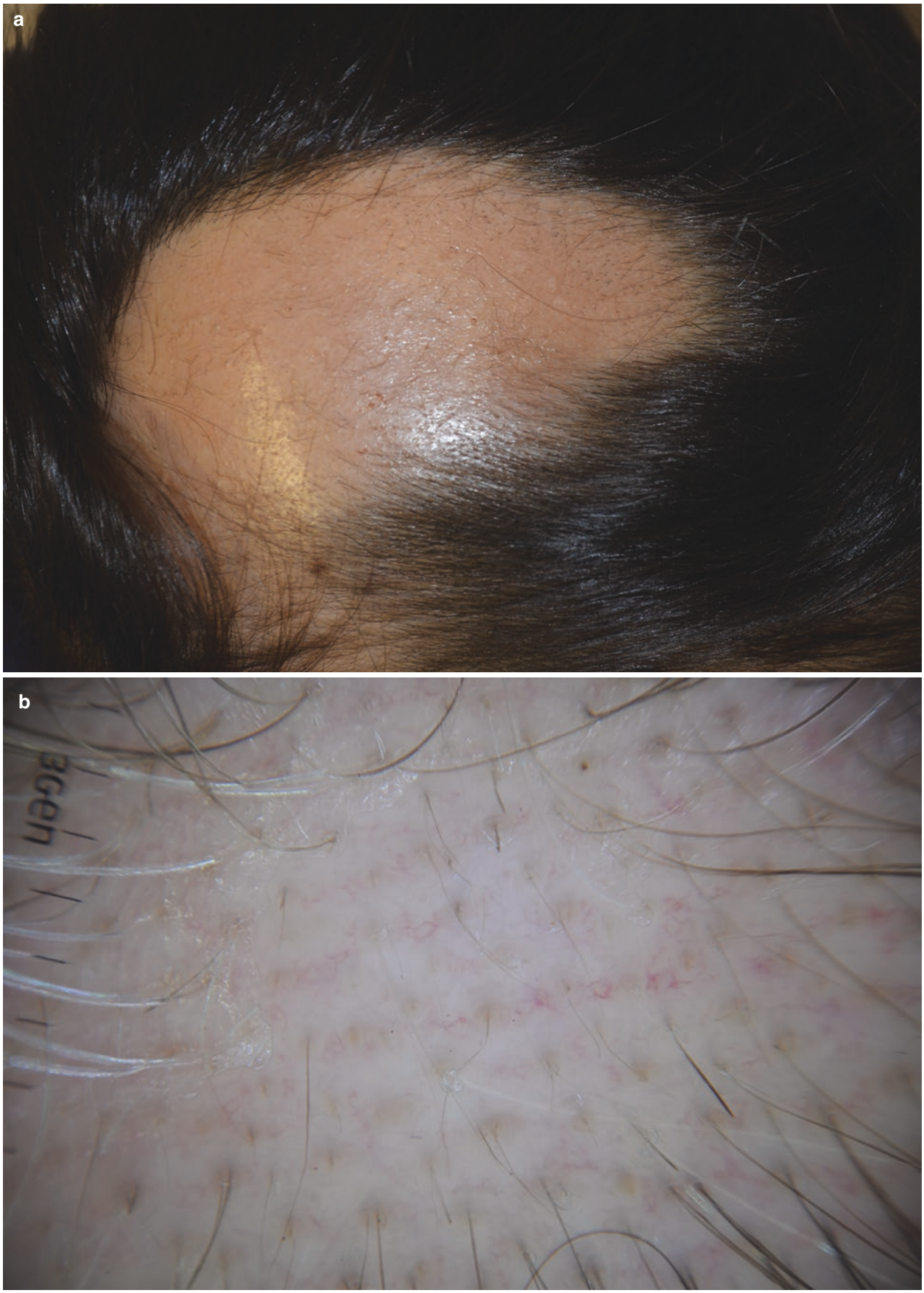

Fig. 21.3 (**a**) Alopecia areata. Large, patchy area of hair loss in an 8-year-old girl. (**b**) Dermatoscopy (×10): *yellow dots*, *exclamation-mark hairs*, and regrowing *vellus hairs*

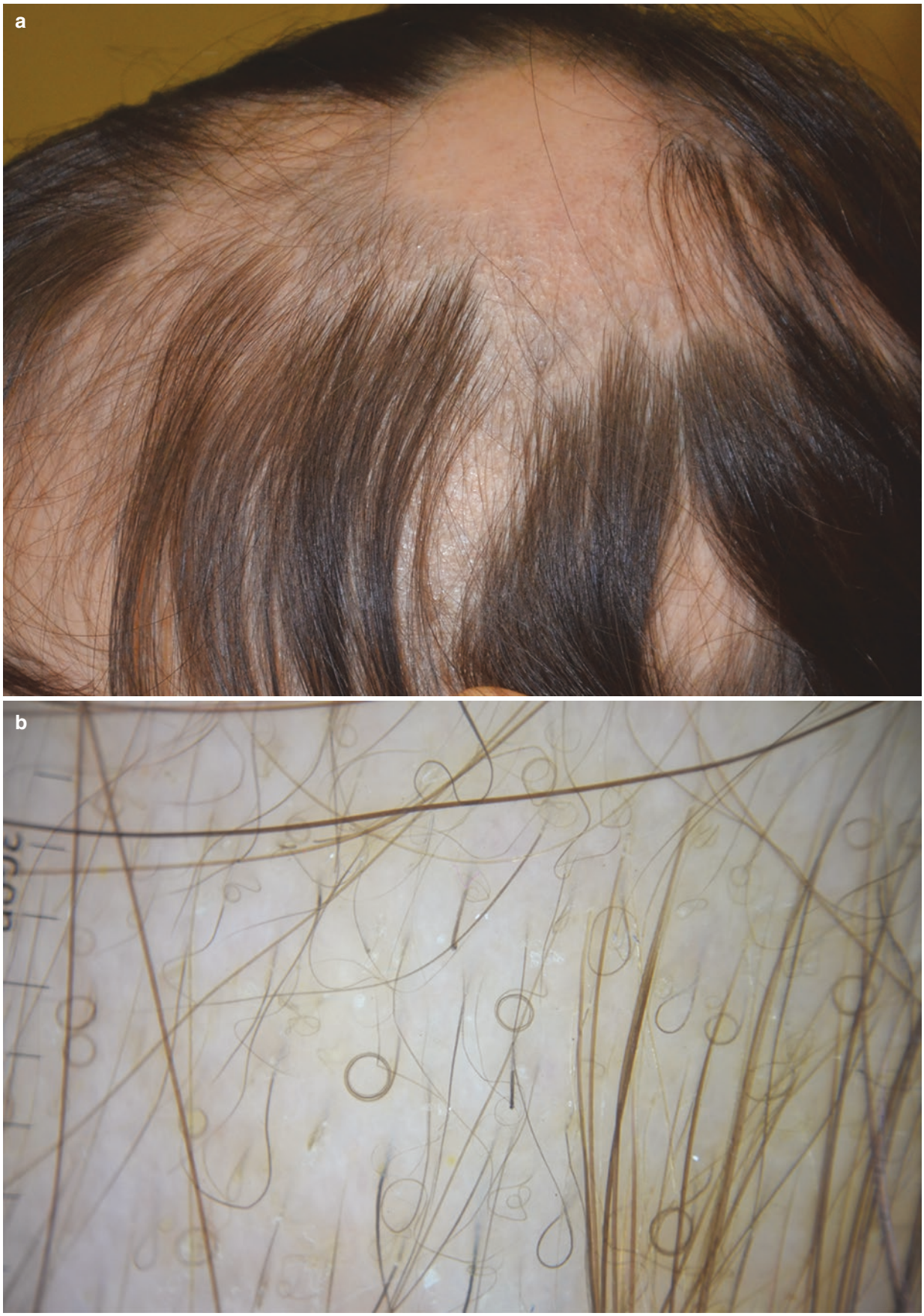

Fig. 21.4 (**a**) Alopecia areata. Multiple patchy areas of hair loss in a 9-year-old girl. (**b**) Dermatoscopy (×10): *exclamation-mark hairs* as well as regrowing *vellus hairs* appearing as *coiled hairs*

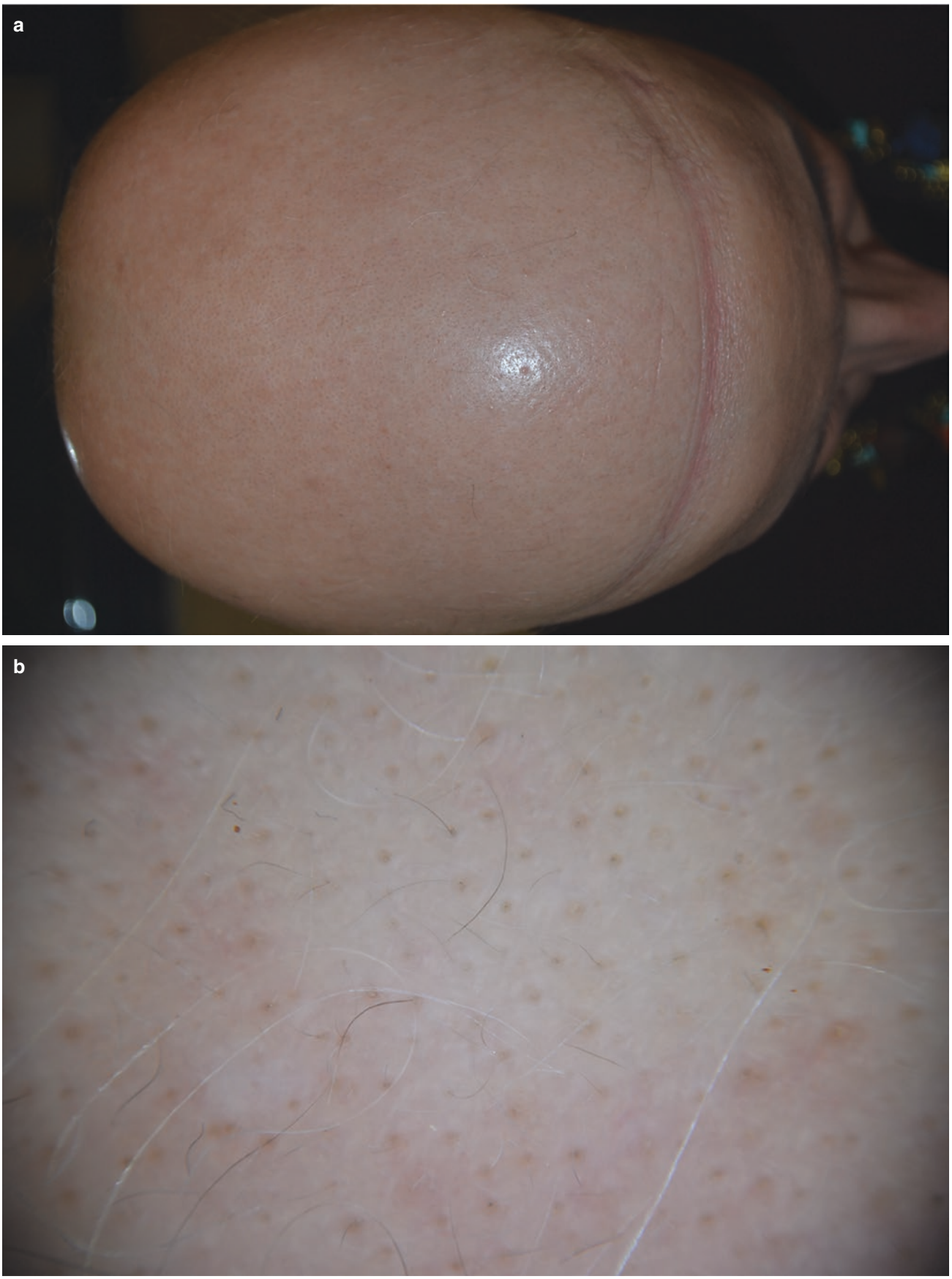

Fig. 21.5 (**a**) Alopecia areata. Total scalp hair loss (alopecia totalis) in a 16-year-old boy. (**b**) Dermatoscopy (×10): *yellow dots* and *vellus hairs*, some of them hypopigmented

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Trichotillomania

22

Giuseppe Micali, Anna Elisa Verzì, Federica Dall'Oglio, and Francesco Lacarrubba

22.1 Definition

Trichotillomania is defined as hair loss due to patient's repetitive self-pulling of hair. Although it is thought to be expression of an obsessive-compulsive disorder, recent findings also suggest that trichotillomania may be similar to tic disorders [1].

22.2 Epidemiology

The true prevalence of trichotillomania is unknown being grossly underestimated and underrecognized. The lifetime prevalence is estimated to be between 0.6 and 4.0% [2]. In a prospective study of 210 children complaining of hair loss and scalp disorders, 7% of patients presented trichotillomania [3]. The disorder frequently occurs in early childhood as well as in pre- or early adolescence.

22.3 Etiology

The etiology of trichotillomania is complex. It may be classically interpreted as a sign of an underlying psychosocial problem and several triggers were associated with its onset. In a study conducted on 33 patients, triggering factors were identified in about 50% of cases including physical appearance, family-related issues, school-related issues, and concurrent illness [4]. Several neurobiologic factors have also been implicated [5]. Many patients pull their hair unconsciously while they are occupied with other activities, e.g., watching television.

Moreover, cases of parents who used to pull the child's hair as a release for an own hair-pulling urge, known as trichotillomania by proxy, have been reported [6, 7].

22.4 Clinical Features

Physical examination of affected areas shows single or multiple, ill-defined patches of incomplete alopecia displaying short and broken hairs of varying lengths (Figs. 22.1a and 22.2a). Perifollicular erythema, hemorrhage, or excoriations may also be present. Recurrent picking can lead to superficial bacterial infections, masking the picture [8].

The most common affected area is the scalp, although eyelashes, eyebrows, and pubic hair may also be involved [8].

Sometimes, patients ingest the pulled hairs (trichophagia) leading to a rare complication named trichobezoar. This habit is present in approximately 5–30% of adult patients, but it seems less frequent in children [9].

22.5 Differential Diagnosis

In the pediatric population, alopecia areata is the main differential diagnosis for trichotillomania. Sometimes it could be a challenge. In addition, the coexistence of trichotillomania and alopecia areata in the same patient has been reported [10, 11]. A prompt and correct diagnosis of the association is quite difficult but crucial.

Other causes of hair loss with similar clinical aspect include tinea capitis, telogen effluvium, and traction alopecia [5].

22.6 Diagnosis

The American Psychiatric Association's *Diagnostic and Statistical Manual of Mental Disorders*, Fifth Edition (DSM-5) places trichotillomania in the category of obsessive-compulsive and related disorders [12]. The specific criteria

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are (1) recurrent pulling out of one's hair, resulting in hair loss; (2) repeated attempts to decrease or stop the hair-pulling behavior; (3) clinical significant distress or impairment in social, occupational, or other important areas of functioning from hair pulling; (4) no relation of hair pulling or hair loss with another medical condition (e.g., a dermatologic condition); and (5) no relation with another mental disorder (e.g., attempts to improve a perceived defect or flaw in appearance, such as may be observed in body dysmorphic disorder).

22.7 Dermatoscopy

At dermatoscopy, the recognition of multiple hairs broken at different lengths and fraying of the extremities (trichoptilosis) are the most consistent features (Figs. 22.1b, 22.2b, c, d, and 22.3) [13–20]. Moreover, damaged hair shafts have been variably described based on their appearance: "flame hairs" appear as semitransparent, wavy, and cone-shaped hair residues resembling flames; sometimes, the remaining distal part of hair shaft may contract and coil ("coiled hairs"); if a diagonal fracture occurs, the distal ends of the hair shafts appear "empty" inside ("tulip hairs"); the "V-sign" can be found when two hairs emerging from the same follicular unit are pulled simultaneously. Regrowing hairs may coexist with features of ongoing hair pulling. Moreover, yellow dots, black dots, and micro-exclamation-mark hairs may also be detected, although nonspecific of trichotillomania.

The interfollicular area may reveal the presence of hair residue particles, called hair powders. Often, red dots corresponding to follicular microhemorrhages may suggest the history of forced pull off [11, 21].

22.8 Histopathological Correlation

At histologic examination, catagen and telogen hairs without evidence of inflammation are typically increased in number. Pigment casts and empty anagen follicles are often seen. Torn-away follicles may be seen with remnants of hair bulbs. The perifollicular hemorrhage near the hair bulb or in the interfollicular space may be diagnostic.

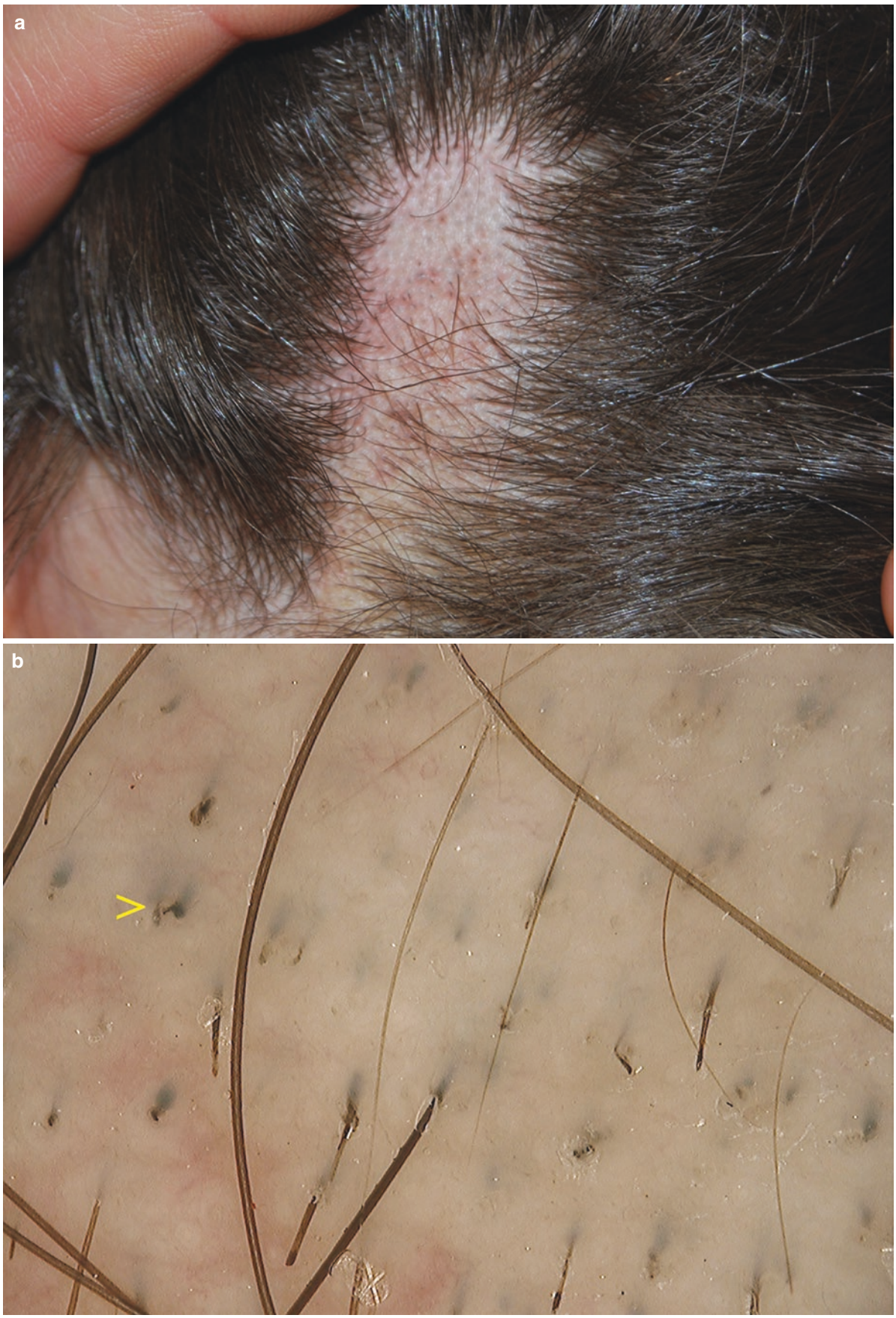

Fig. 22.1 (**a**) Trichotillomania. Ill-defined patch of incomplete alopecia of the scalp in a 10-year-old girl. (**b**) Dermatoscopy (×10): multiple *broken hairs*, *black dots*, and *flame hairs* (arrowhead)

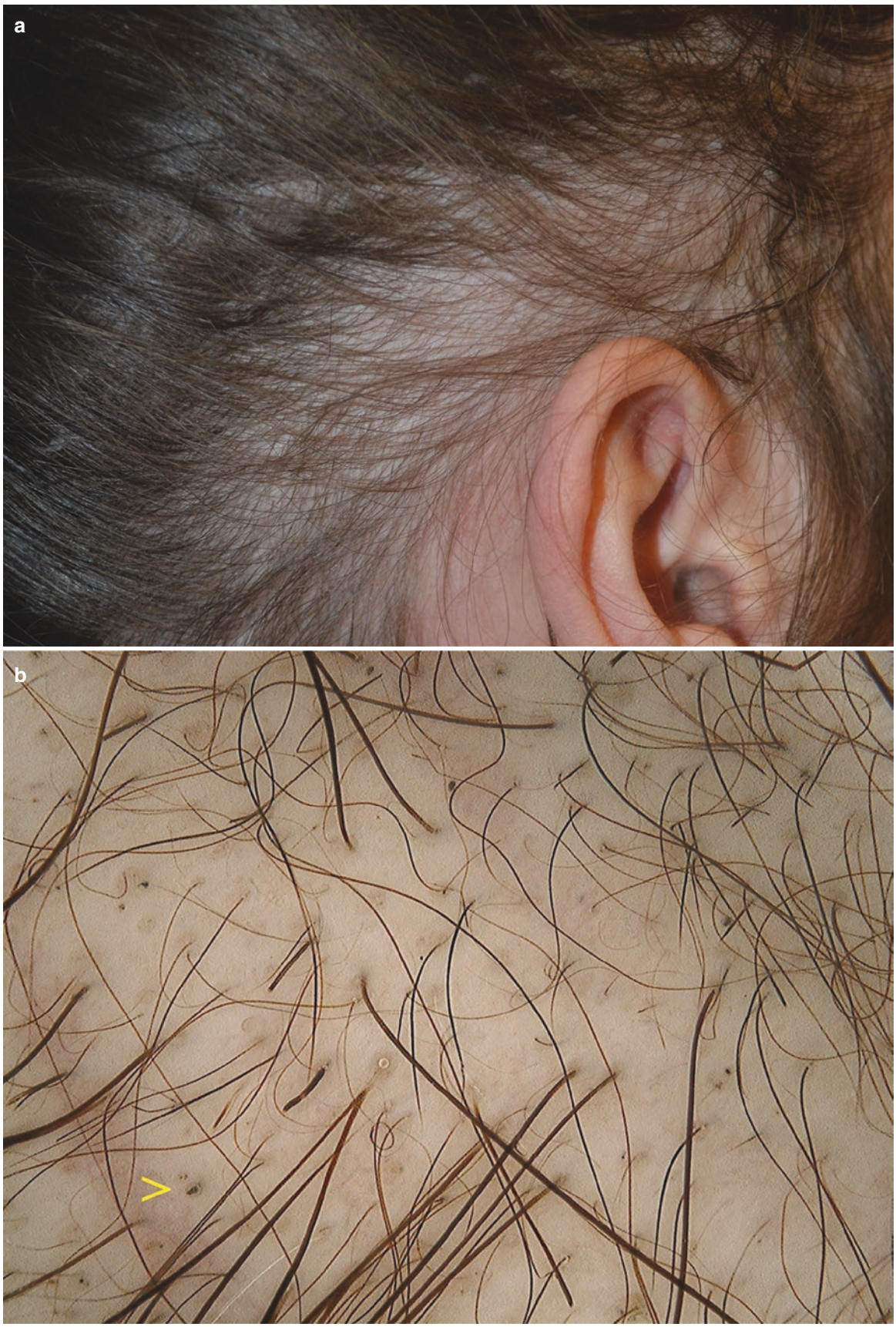

Fig. 22.2 (**a**) Trichotillomania. Ill-defined area of incomplete alopecia of the scalp in a 12-year-old girl. (**b**) Dermatoscopy (×10): multiple hairs broken at different lengths, few *black dots* (arrowhead), and several fine, regrowing hairs. (**c**) High magnification dermatoscopy (×70): *broken hairs* and regrowing *coiled hairs* (arrowhead). (**d**) High magnification dermatoscopy (×100) of another area: fraying of the hair extremity (trichoptilosis)

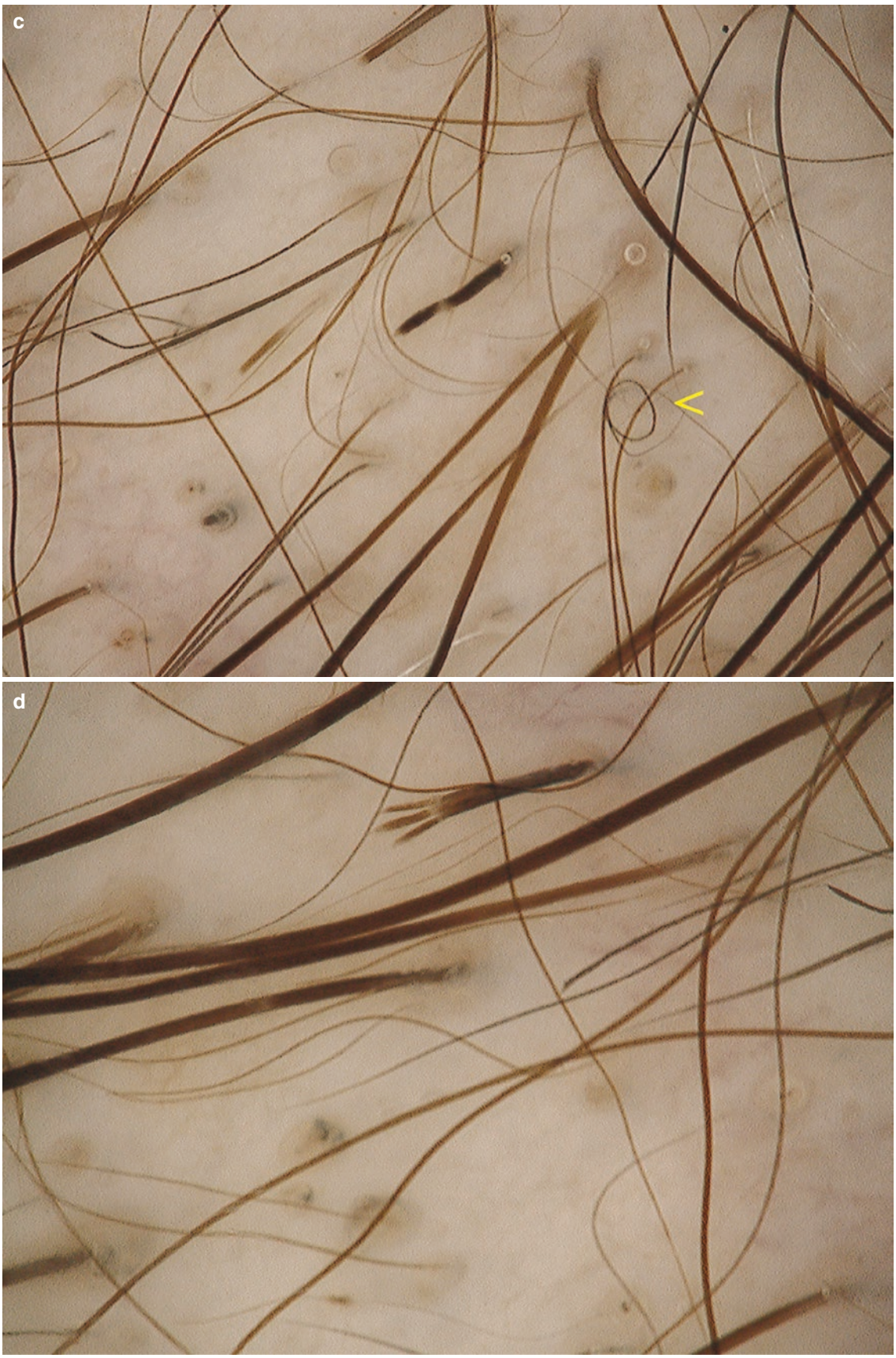

Fig. 22.2 (continued)

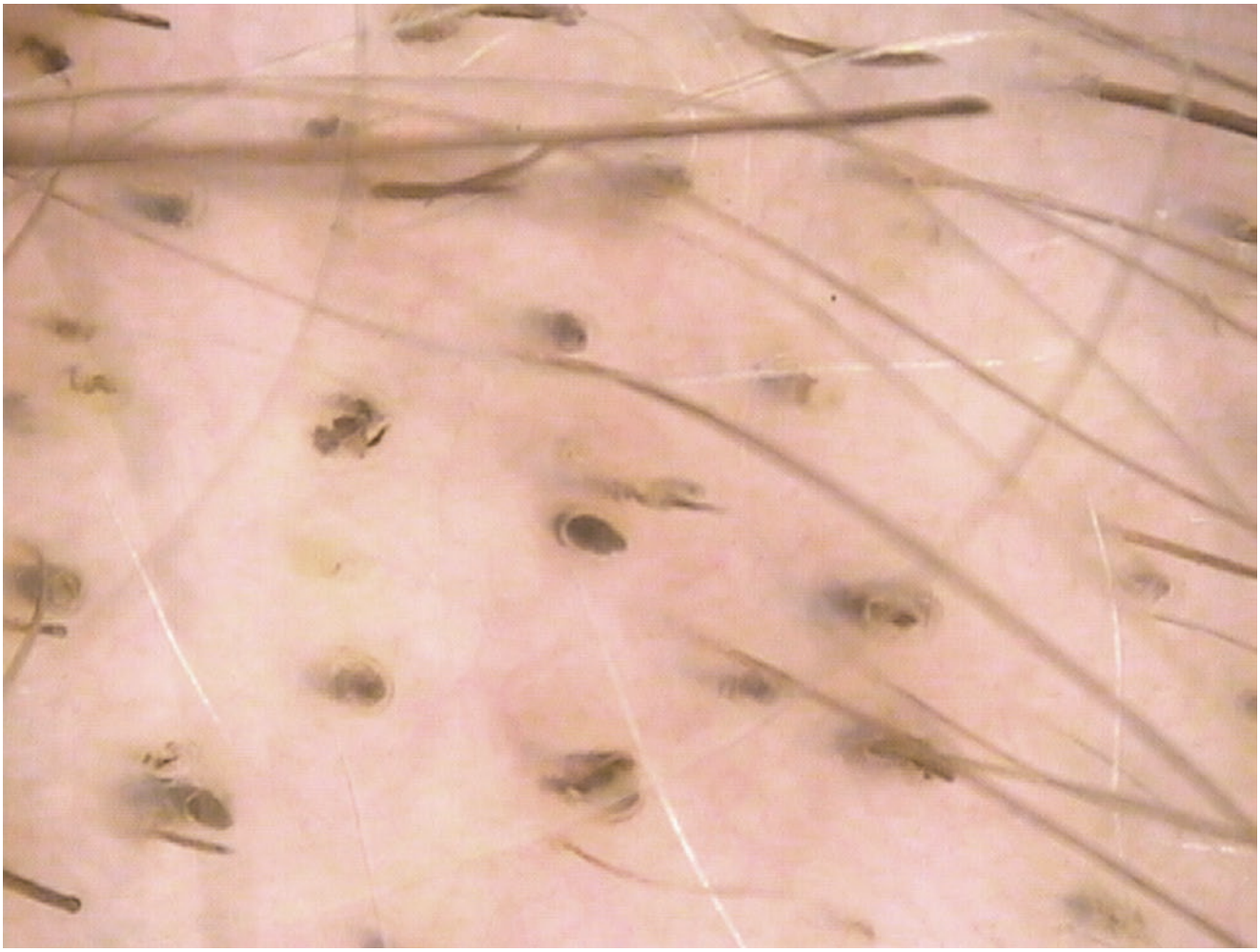

Fig. 22.3 Trichotillomania. High magnification dermatoscopy (×100): typical semitransparent, wavy, and cone-shaped hair residues (*flame hairs*)

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Congenital Triangular Alopecia

23

Francesco Lacarrubba, Anna Elisa Verzì, Maria Rita Nasca, and Giuseppe Micali

23.1 Definition

Congenital triangular alopecia, also known as temporal triangular alopecia and Brauer nevus, was first described in 1905 by Raymond Sabouraud [1]. It is a benign, nonprogressive, circumscribed, and non-scarring form of alopecia usually confined to the frontotemporal scalp.

23.2 Epidemiology

The exact incidence of congenital triangular alopecia among general population is not known [2]. It is rarely reported in the literature, probably due to the benign and asymptomatic behavior of the condition as well as to undiagnosis or misdiagnosis [3–6].

23.3 Etiology

The etiology of congenital triangular alopecia is currently unclear. It has been postulated that it may reflect mosaicism and may be a paradominant trait [7, 8]. Although considered congenital, it is usually noted in children between 2 and 4 years of age, but may present at birth or develop in adulthood [9]. It usually appears sporadically, although a small number of familial cases have been reported [10].

23.4 Clinical Features

Congenital triangular alopecia usually presents as nonprogressive, triangular-, oval-, or lancet-shaped patches of alopecia involving unilaterally or, less frequently, bilaterally temporal region of the scalp (Figs. 23.1a and 23.2a) [6]. Rare is the occurrence over the parietal area and vertex [11]. The skin of the affected areas is normal in appearance with no atrophy, scarring, or inflammation.

23.5 Differential Diagnosis

Congenital triangular alopecia may be misdiagnosed as androgenetic alopecia, alopecia areata, traction alopecia, trichotillomania, tinea capitis, sebaceous nevus, and aplasia cutis congenita.

23.6 Diagnosis

History, clinical features, and typical location may help distinguish congenital triangular alopecia from other circumscribed alopecias.

23.7 Dermatoscopy

Examination using a polarized light dermatoscope allows the visualization of a carpet of short vellus hairs as well as normal follicular openings (Figs. 23.1b, c, and 23.2b) [12–16]. The lesion is surrounded by normal terminal hairs. In addition, absence of features diagnostic for other types of localized alopecia (abnormal hair shafts, altered follicular orifice, perifollicular changes) can be demonstrated [14].

23.8 Histopathological Correlation

Histopathology confirms the presence in the alopecic area of normal numbers of hair follicles and miniaturized telogen hairs in the dermis with no evidence of scarring or perifollicular inflammation [3].

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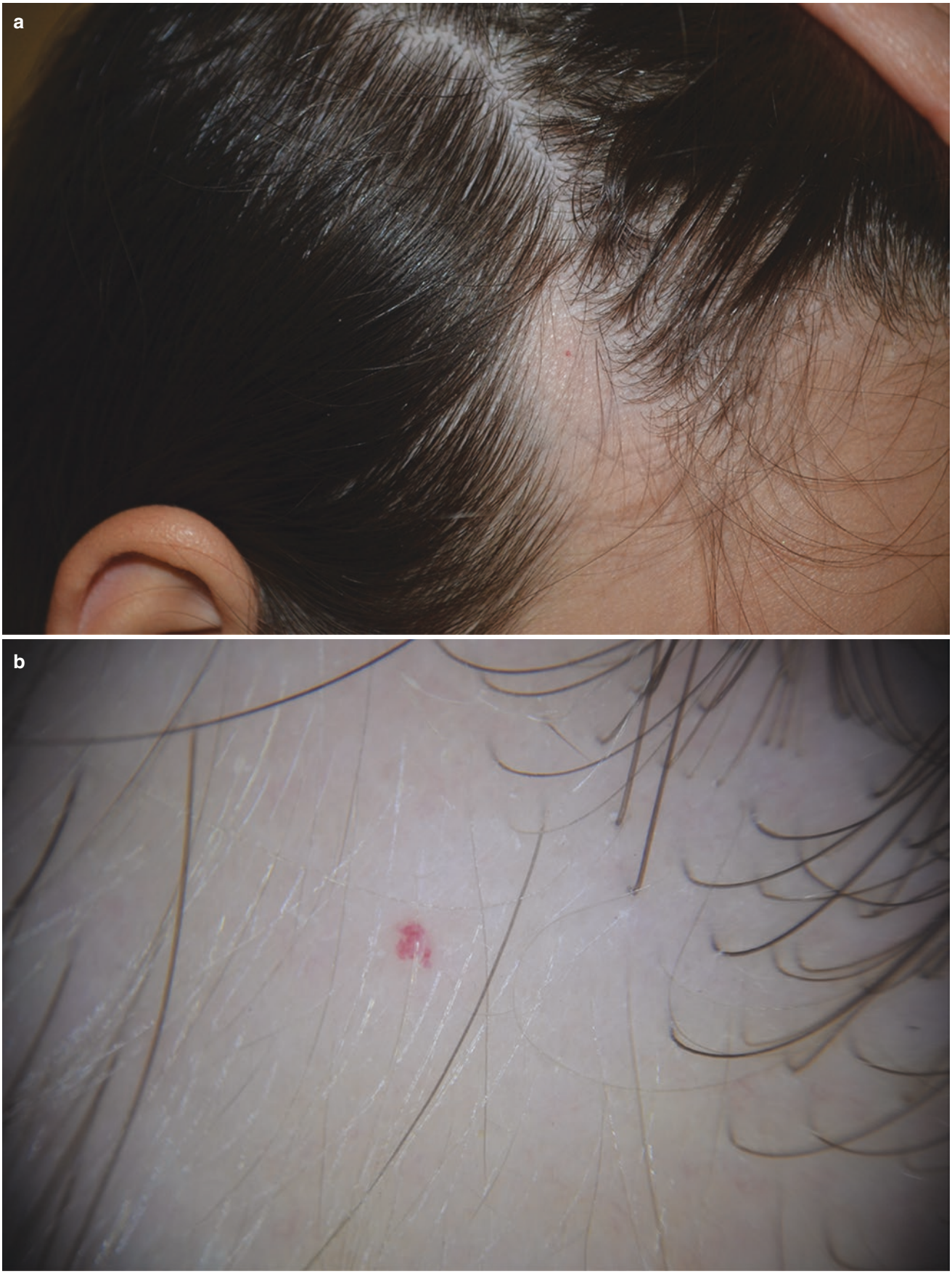

Fig. 23.1 (**a**) Congenital triangular alopecia. Triangular-shaped patch of alopecia involving the temporal region of the scalp in an 8-year-old girl. (**b**) Dermatoscopy (×10): carpet of short vellus hairs and normal follicular openings. (**c**) Dermatoscopy (×10) of the periphery of the alopecic patch: short vellus hairs surrounded by normal terminal hairs

Fig. 23.1 (continued)

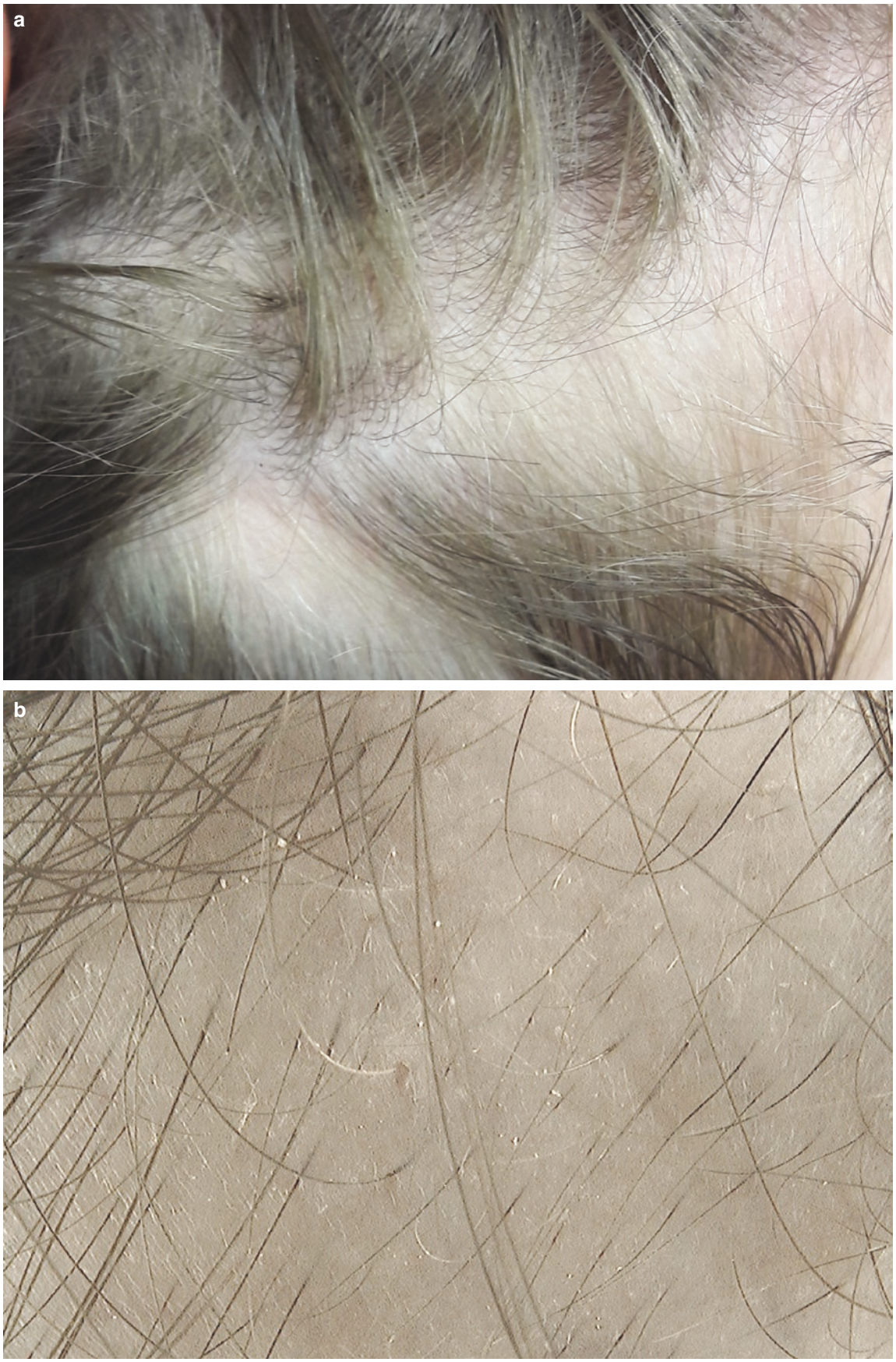

Fig. 23.2 (**a**) Congenital triangular alopecia. Ill-defined patch of alopecia involving the temporal area in a 13-year-old girl. (**b**) Dermatoscopy (×10): short vellus hairs and normal follicular openings

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Hair Shaft Disorders

24

Fernanda Torres and Antonella Tosti

24.1 Introduction

Hair shaft disorders can be congenital or acquired, which can be caused by trauma. Patients presenting a congenital hair defect usually present it at birth, or it usually becomes evident in the first months of life [1]. Patients can present with localized or diffuse alopecia, and some hair shaft defects can also simulate hair loss and should be considered in patients with abnormal hair growth.

This chapter will describe the relevant dermatoscopic findings in the most common pediatric hair shaft defects.

24.2 Hair Shaft Disorders with Increased Fragility

Patchy or diffuse alopecia can be observed in this group of patients.

24.2.1 Monilethrix

Monilethrix is an inherited autosomal dominant condition with variable expression due to mutation in the keratin hair basic 6. Dermatoscopy of the hair reveals uniform small elliptical nodes of normal thickness separated by regular multiple constrictions (Figs. 24.1 and 24.2) [2, 3].

The same dermatoscopic findings can be observed in other areas such as the eyebrows or body hairs (Figs. 24.3 and 24.4).

24.2.2 Pseudomonilethrix

Under the microscope, monilethrix can be confused with pseudomonilethrix, which is caused by overlapping hairs pressed under the cover slide prepared for microscopic examination, and thus dermatoscopy prevents misdiagnosing these two conditions [4].

24.2.3 Trichorrhexis Invaginata (Netherton Syndrome)

Netherton syndrome is a rare autosomal recessive disease caused by a mutation in the SPINK5 gene that is characterized by trichorrhexis invaginata, erythroderma, ichthyosis, atopy, and failure to thrive [5, 6]. Trichorrhexis invaginata is a constant feature in this syndrome: the hair shaft shows multiple knots along its length. The knots consist of a proximal

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cup-shaped portion and a distal ball-shaped portion resembling the ball and cup joint of bamboo, which is easily detected by dermatoscopy (Fig. 24.5) [5]. In that way dermatoscopy allows an early diagnosis of this syndrome, decreasing the need for the expensive SPINK5 testing.

24.2.4 Pili Torti

Pili torti are flattened and present twisting of the hair shaft through 180° in groups of 3–10 at irregular intervals (Fig. 24.6) [7]. A small number of these hairs can be frequently found in scarring alopecias and in association with other hair shaft abnormalities. Pili torti are also a feature of several rare genetic syndromes such as Menkes syndrome. Dermatoscopy shows irregular twisting and flattening of the hair shaft.

24.2.5 Trichothiodystrophy

These patients present short, brittle hair with low sulfur content. It encompasses a heterogeneous group of autosomal recessive disorders. The affected hairs show alternating light and dark bands along the hair shaft (tiger-tail pattern) under the polarized microscope. Dermatoscopy is useful to select the hair that should be examined under the microscope; these present a nonhomogeneous structure resembling grains of sand and a wavy contour [8].

24.2.6 Trichorrhexis Nodosa

This condition is the most common defect of the hair shaft. It can be congenital or acquired, and although it is not specific for a disease, it can represent an important clue to a possible metabolic disorder. Dermatoscopy shows broken hair shafts with a brushed tip (Fig. 24.7) [8].

24.2.7 Ectodermal Dysplasia

Rakowska et al. performed hair dermatoscopy in 16 patients with ectodermal dysplasia and observed predominance of pilosebaceous units with one hair (69%), abnormalities of hair shaft pigmentation (gray hair with single dark hairs, 56%), pili torti, trichothiodystrophy, trichorrhexis nodosa, and, rarely, cicatricial alopecia [9].

24.3 Hair Shaft Disorders Without Increased Fragility

Changes in hair texture and luster can be observed, but usually not hair loss.

24.3.1 Wooly Hair Syndrome

Dermatoscopy reveals hair shafts resembling a “crawling snake,” with short wave cycles and also some broken hairs [8].

24.3.2 Pili Trianguli et Canaliculi (Uncombable Hair)

This diagnosis is usually based on scanning electron microscopy, but dermatoscopy is a valid and simple alternative. The hair shaft has a triangular or reniform shape and commonly presents with a longitudinal groove or flattening [10].

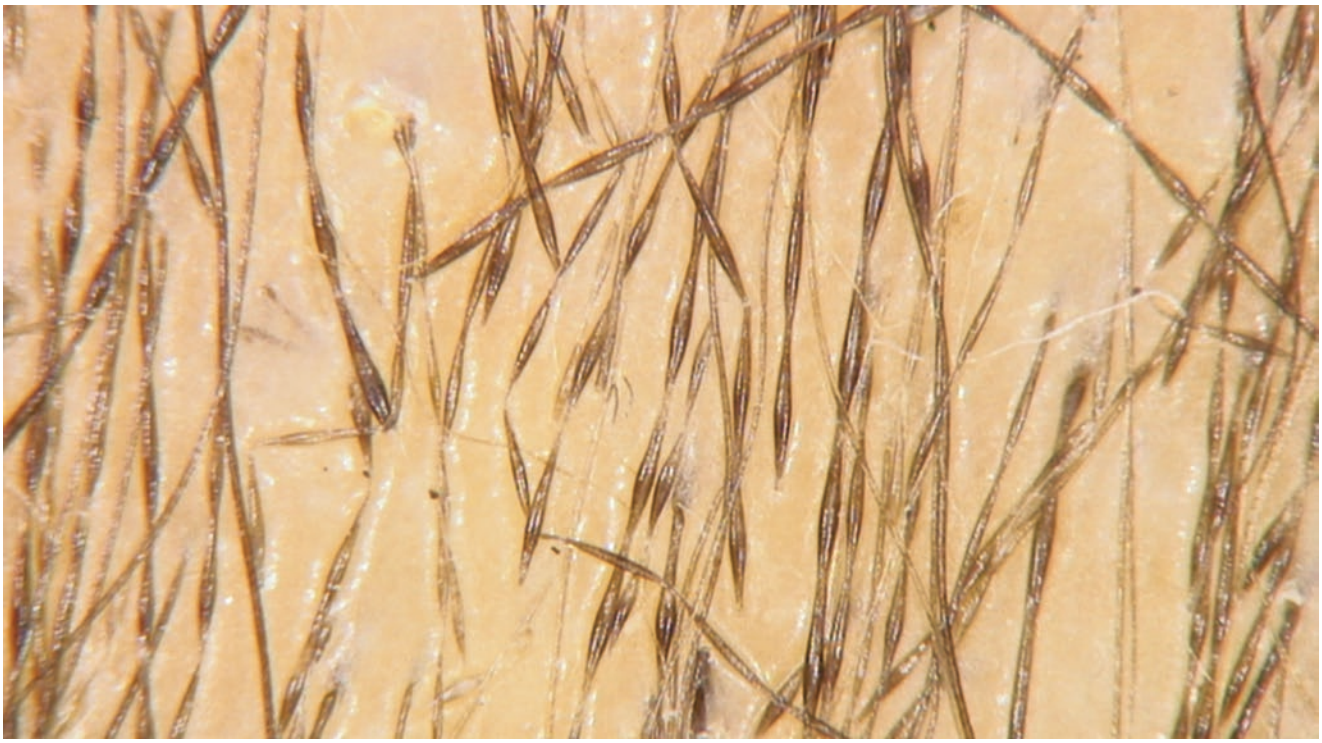

Fig. 24.1 Monilethrix. Dermatoscopy (×20): uniform small elliptical nodes of normal thickness separated by regular constrictions

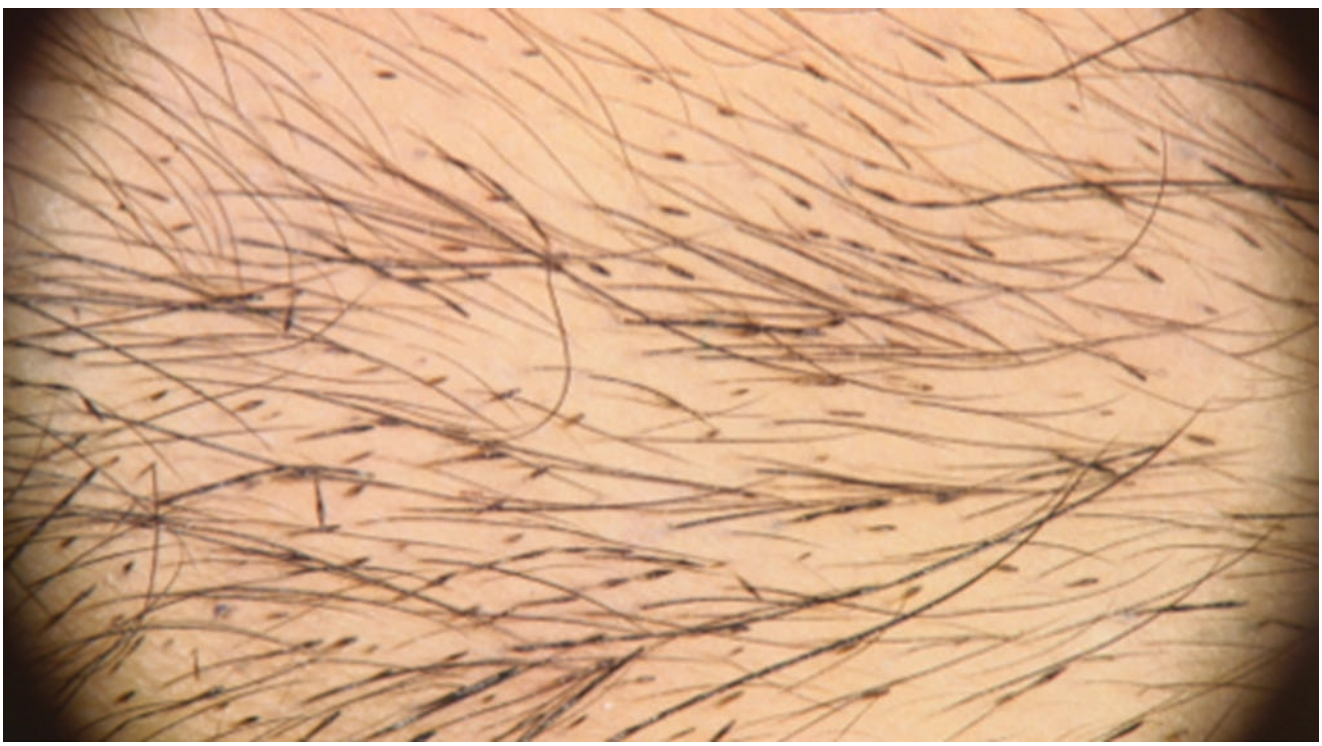

Fig. 24.2 Monilethrix. Dermatoscopy (×20) of the eyebrows

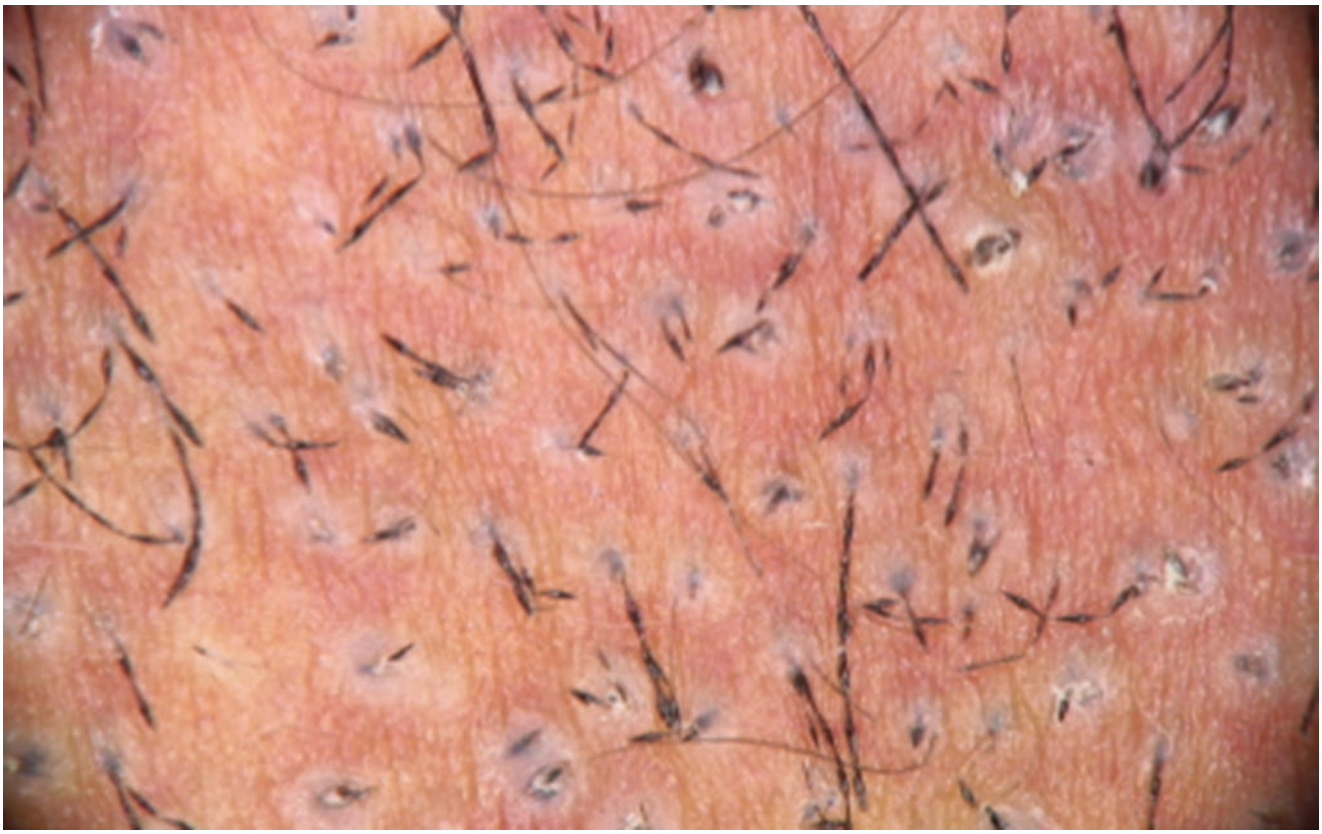

Fig. 24.3 Monilethrix. Dermatoscopy (×20) of the nuchae

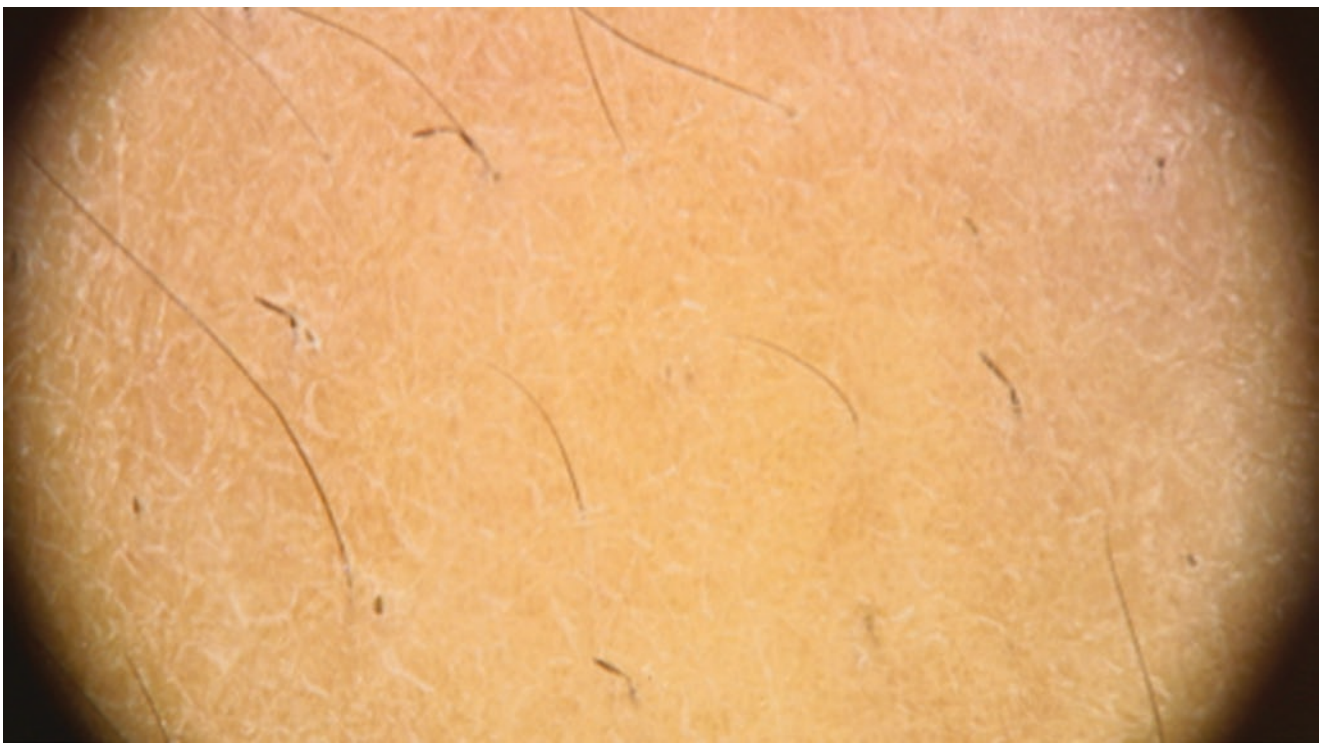

Fig. 24.4 Monilethrix. Dermatoscopy (×20) of the leg

Fig. 24.5 Bamboo hair in trichorrhexis invaginata. Dermatoscopy (×20): knots with a proximal cup-shaped portion and a distal ball-shaped portion

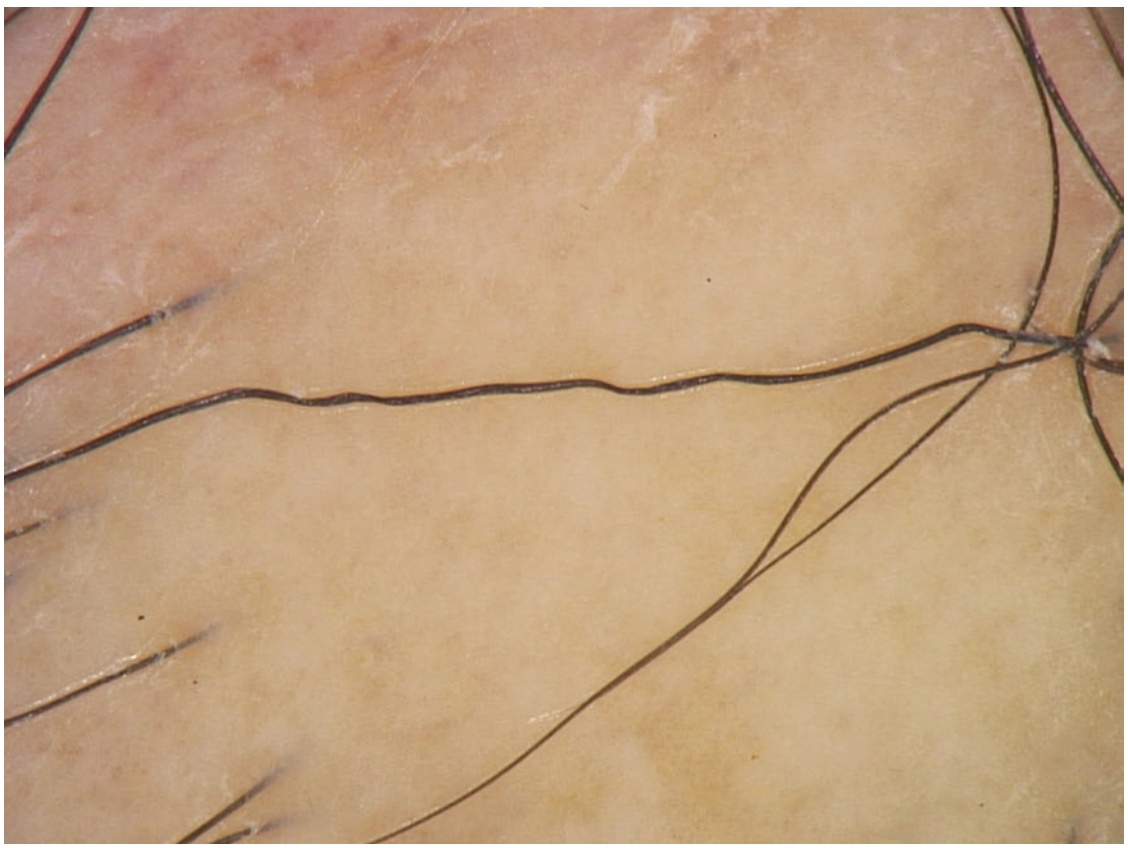

Fig. 24.6 Pili torti. Dermatoscopy (×20): flattened and twisted hair shaft

Fig. 24.7 Trichorrhexis nodosa. Dermatoscopy (×70): broken hairs with a brushed tip

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Nail Disorders

25

Bianca Maria Piraccini and Michela Starace

25.1 Technique

Nail dermatoscopy, or onychoscopy, can be performed "dry" or with "immersion" technique (in order to render the stratum corneum translucent) depending on the suspected disease: in case of nail plate alterations, it is better to use the dry method, while in case of color alterations, the use of gel is recommended because it increases visibility.

The unique anatomy of the nail apparatus makes onychoscopy technically difficult to be performed and not easy to be interpreted. The main technical problem is the convexity and hardness of the nail plate, which makes difficult to obtain complete contact of the lens to the surface. The usual technical problem to see all the nail at once is avoidable in children, in which the small size of the nail permits obtaining all the part of the nail unit in same picture.

25.2 Dermatoscopy of Normal Nails in Children

Onychoscopy should not be limited to the nail plate, which is only a part of the nail apparatus, but should include the proximal and lateral folds, the hyponychium, and the nail bed, the latter visible through the plate. The nails of pediatric population are thin, soft, and completely formed and their size is related to age. The normal nail is rectangular in shape: the fingernail has a longitudinal major axis and transversal minor axis, while the toenail in children has a trapezoid shape with always a longitudinal major axis and is wider at the distal free edge.

The normal nail plate is well displayed at ×10 magnification. It appears pale pink in color because of the presence of the vessels of the underlying nail bed, and its surface is smooth and shiny. It is translucent and adherent to the nail bed and with a free edge of regular thickness.

The nail bed is visible below the transparent plate and appears pale pink in color. It represents the epithelium and connective tissue on which the nail plate rests.

The normal proximal nail fold at ×10 magnification appears pale pink in color, and its epithelium has a smooth surface. The cuticle is easily visible as a transparent transverse band that seals the nail plate from the outside environment at the proximal nail fold.

The hyponychium is the distal part of the nail unit, at the junction of the free edge of the nail plate and the nail bed. It can be observed by putting the lens under the nail plate free margin: the epithelium shows the digital creases and, at ×40 magnification, the capillary of the dermis appears as red dots, due to their arrangement perpendicular to the skin.

25.3 Alterations of the Nail Shape

The alterations of the shape of the nails include abnormalities in size, proportion, or curvature.

25.3.1 Abnormal Size

Anonychia and micronychia represent the total or partial absence of the nail. They can be congenital or acquired. In nail-patella syndrome, which is due to a mutation of the gene *LMX1B* localized on chromosome 9q34.1 [1, 2] and is inherited in an autosomal dominant pattern, nail hypoplasia is associated with bone and kidney abnormalities. Nail changes may be limited to the fingernails, usually the thumbs, with hypoplasia or absence of the nail plate. Dermatoscopy

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reveals the characteristic finding of the lunula with a triangular shape with the horizontal basis of the triangle at the level of the proximal margin and the point of the triangle directed toward the distal margin. The color of the triangular lunula is white, while its dimension can be different in different digits, being more pronounced in the thumbs that usually are the more affected, and less evident going toward the fifth digit (Fig. 25.1).

25.3.2 Abnormal Proportion

In children, alterations of nail proportion can be congenital, such as in racquet nails where the nail plate is wider than long and the nail appears abnormally broad, or acquired as in onychophagia, which affects children and adolescents who start to bite the nails possibly because of stress, anxiety, or boredom. In onychophagia the nail plate becomes shorter and irregular in shape with the transverse axis longer than longitudinal axis. Dermatoscopic features of nail biting include (a) short nail plate, with irregular distal margin and exposure of the nail bed epithelium to mechanical and chemical damage of the teeth and saliva; (b) dilated proximal nail fold capillaries due to chronic trauma; (c) scaling and crusts, wounds, and diffuse inflammation of the periungual folds, associated with hemorrhages; and (d) skin maceration on the lateral folds. Moreover, in severe cases it is possible to observe the hyponychium and nail bed skin. One or more bands of melanonychia can be associated to melanocytic activation by trauma of nail matrix (Fig. 25.2).

25.3.3 Abnormal Curvature

The normal nail plate is curved transversally and, to a lesser extent, longitudinally. In cases of abnormal curvature of a single digit, it is important to exclude a nail bed tumor, such as osteochondroma. It usually appears between the ages of 10 and 25 years, sometimes preceded by a history of trauma and appears as a firm and tender nodule under the nail plate clinically similar to subungual exostosis. Onychoscopy of osteochondroma shows the subungual mass under the nail plate with a superficial view of the nail bed vessels that are easily visible with a reticular aspect (Fig. 25.3).

25.4 Alterations of the Nail Surface

Different signs on the surface of the nail plate, reflecting a damage of nail matrix, can be detected.

25.4.1 Pits

They are small depression of the nail plate surface. Dermatoscopy is very helpful to distinguish diseases appearing with pitting, such as psoriasis and alopecia areata, especially in cases where pitting is the only sign. The pits of psoriasis are large, deep, and irregular in shape, size, and distribution (Fig. 25.4), while the pits of alopecia areata are regular in shape, size, and distribution (Fig. 25.5).

25.4.2 Beau's Lines and Onychomadesis

Beau's lines are transverse nail plate surface depressions resulting from a mild trauma to the proximal nail matrix with transient reduced nail growth. When the trauma is more intensive and the damage involves the whole nail matrix with a total arrest of the nail plate production, there is onychomadesis, which appears as a transverse detachment of the nail from the proximal nail fold. In children, Beau's lines and onychomadesis can be present in one digit as a result of bacterial or viral infection or in case of traumas such as finger sucking or a habit tic. In finger sucking dermatoscopy shows onycholysis of the distal nail plate and maceration of the periungual skin due to saliva (Fig. 25.6). If the trauma from biting is at the level of the proximal nail fold, Beau's lines can appear until onychomadesis. In habit tic nail deformity, the clinical aspect is characterized by longitudinal furrows of the nail plate with multiple transverse lines. Onychoscopy permits enhanced visualization of the typical findings of the transverse lines that are parallel from each other (Fig. 25.7). The cuticle is absent or uplifted with hyperkeratosis. The periungual tissue may show scales and crusts. Macrolunula can be associated, resulting from the periodic trauma, as well as a band of longitudinal melanonychia due to melanocytic activation.

In cases where several nails are interested by Beau's lines and/or onychomadesis, a systemic cause should be suspected, such as coxsackievirus-induced hand-foot-mouth disease.

25.4.3 Trachyonychia

It is a benign inflammatory nail condition of the proximal nail matrix that can be due to many diseases including alopecia areata, lichen planus, eczema, and psoriasis. TND can affect one or all nails ("20-nail dystrophy"). The affected nails show diffuse roughness with longitudinal and regular fissuring and are usually opaque. Nail thinning with koilonychia may be present. At dry onychoscopy, the nail plate shows fine longitudinal striations covered by thin scales and mild thinning (Fig. 25.8).

25.4.4 Longitudinal Furrow

It is a single longitudinal depression that runs through the nail plate from the proximal to the distal part with a variable depth and width on the basis of the disease. Longitudinal furrowing results from a compression of the nail matrix usually by a benign tumor that is visible under the proximal nail fold. In children, the most typical tumor that appears with longitudinal furrowing is fibrokeratoma, a common benign tumor of the proximal nail fold. The clinical aspect fibrokeratoma is reflected by dermatoscopy that shows a periungual filiform mass, pink in color, sometimes with a hyperkeratotic tip. When the tumor is localized under the nail plate, onycholysis or erythronychia is produced (Fig. 25.9). When multiple fibrokeratomas are present in fingernails and/or toenails, named Koenen tumors, they are one of the major criteria of tuberous sclerosis.

25.5 Alterations of the Nail Color

Alterations of the nail color can result from the deposition of a pigment on the nail plate or from the production of the pigment from the nail matrix. In the latter case, the color is within the nail plate.

25.5.1 Leukonychia

Children usually are affected by true leukonychia that is due to trauma to the distal matrix. In true leukonychia, the milky white discoloration is within the nail plate and results from the presence of foci of parakeratotic cells within the nail plate. The presence of nuclei impairs nail plate transparency and reflects light, resulting in the white color. Depending on the shape, leukonychia can be punctate or transverse. Punctate leukonychia is typical of several fingernails: onychoscopy shows a normally smooth nail plate surface and spots of white discoloration inside the nail plate (Fig. 25.10). Transverse leukonychia is quite rare in children and typically restricted to the first toenails. This variety of true leukonychia is due to trauma from the shoes to a thick nail plate, which transmits the trauma to the distal nail matrix, resulting in periodic defective keratinization with the production of one or more transverse white bands that move distally with nail growth [3]. The shape of the white spots inside the nail plate is in this case transverse.

25.5.2 Splinter Hemorrhages

They are due to rupture of the blood capillaries of the nail bed and appear as longitudinal red to brown striae in the distal nail. Their shape reflects the shape of the capillaries of the nail bed, which run along the creases. Dermatoscopy allows an enhanced visualization of the splinter hemorrhages, which appear deep red in color and a few millimeters in length. Splinter hemorrhages may result from mechanical trauma or may be associated with other diseases, such as psoriasis (Fig. 25.11) or onychomycosis.

25.5.3 Subungual Hematoma

One of the best uses of onychoscopy is to distinguish blood from melanin [4]. Subungual blood extrusion due to trauma is very common in the toenails. Although the round shape of the hematoma is usually easy enough to distinguish it from a band of melanic nail pigmentation with the naked eye, the patient usually becomes aware of the presence of a brown-black nail pigmentation of one toenail that last for a long time. Dermatoscopy shows the round shape of hematoma, associated with a homogeneous color in the red-brown pigmentation, a globular pattern, a peripheral fading, and multiple blood globules or splinter hemorrhages around the hematoma. The color of hematoma depends on the time from the occurrence of the trauma. A recent hematoma is deep under the plate and red purple to black in color, with irregular margins but generally round at the proximal edge and with a streaked and filamentous distal end [5]. A new term is coined as *pseudopods* to refer to the distal end of a nail hemorrhage (Fig. 25.12) [6]. Older lesions are more superficial on the ventral nail plate and roundish, red brown in color, often surrounded by small globules of paler color or dots of coagulated blood with fading around the center of the lesion.

25.5.4 Melanonychia

The term melanonychia refers to black-brown-gray pigmentation of the nail due to the presence of melanin within the nail plate. Usually it appears as a longitudinal band which starts from the proximal margin and extends to the distal margin of the nail following the growth of the nail. Onychoscopy can be useful for the evaluation and the management of melanonychia in a three-step process: (1) establish if the pigment is melanin or not; (2) determine if the pigmentation of melanin is due to an activation or proliferation; and (3) determine if the proliferation is benign or malignant [7].

Dermatoscopy is useful to differentiate melanotic from non-melanotic pigmentation, particularly the quite common nail brown-black discolorations due to subungual hematoma or fungal infection [8]. Generally, melanic pigmentation is brown black and within the nail plate, and the aspect is a longitudinal band, whereas exogenous pigmentation

includes different types of color of the substance which adhere to the nail plate and not always have a longitudinal appearance. Onychoscopy of melanotic nail pigmentation is more difficult to interpret, as there are still not uniform criteria that allow us to differentiate melanonychia due to benign melanocytic proliferation, i.e., nail matrix lentigo or nevus, from melanonychia due to malignant melanocytic proliferation [7–9].

In case of melanonychia of multiple nails, a melanocytic activation, generally due to racial, systemic, or traumatic (nail biting) factors, should be suspected [9]. Dermatoscopy shows a gray background of the band with thin grayish regular and parallel lines (Fig. 25.13).

Nail matrix nevi are typically seen in childhood and it may be congenital or acquired. Clinical and dermatoscopic parameters used in adults are not valid with children. The size and the degree of pigmentation of the band vary considerably; dark bands are associated with pseudo-Hutchinson's sign, because the dark nail plate pigmentation is visible through the transparent nail fold [9–11]. Criterion suggesting biopsy in children is a rapid evolution of the band in growth and color. Dermatoscopic patterns that suggest a nevus are the presence of a brown background with longitudinal brown to black regular and parallel lines with regular spacing and thickness and, more important in children, black dots due to pigment accumulation in the nail plate (Figs. 25.14 and 25.15).

Nail melanoma in children is rare and its frequency depends on the race and exceptional in Caucasians. Twelve cases are described in literature, frequently in pigmented races [12–15]. Dermatoscopic patterns that suggest a melanoma in children are brown background; lines from brown to black; irregular degree of color pigmentation, spacing, or varying thickness; ending abruptly of the distal nail plate; and a parallelism disruption of the bands (Fig. 25.16). However, these features can also be seen in longitudinal melanonychia in children, and their specificity in young age is very low.

25.5.5 Erythronychia

It is a red band originating from the proximal to the distal nail plate, due to the presence of a subungual mass at the level of the lunula. The corresponding nail plate is normal or sometimes has longitudinal fissures. When single, the band indicates the presence of a subungual tumor, such as onychopapilloma or glomus tumor. These tumors are rare in young children and affect mostly adolescent ages. Onychopapilloma occurs on the thumb, sometimes other digits, but rarely involves the toes. It appears as a longitudinal pink-red band often associated or composed entirely of splinter hemorrhages. With clipping the onycholysis of the distal nail plate, the free margin of the band is occupied by hyperkeratosis [15]. Onychoscopy reveals a well-defined longitudinal red band with splinter hemorrhages, starting from the lunula and reaching to the distal margin where it causes a fissure and a filiform hyperkeratotic mass (Fig. 25.17) [16].

When erythronychia affects multiple nails, they are usually due to an inflammatory disease such as nail lichen planus or Darier's disease. Darier's disease is an autosomal dominant genodermatosis characterized by a persistent eruption of hyperkeratotic greasy papules mainly over the seborrheic sites of the body, usually associated with nail abnormalities and sometimes with mucous membrane lesions. Onychoscopy shows alternating parallel longitudinal red streaks with thinning of the corresponding nail plate and white bands. The bed vessels are more visible and associated with splinter hemorrhages. Distal splitting of the nail plate may be more or less marked, especially along the red bands.

25.6 Abnormal Detachment

The abnormal detachment of the nail plate from the nail bed is known as onycholysis. Air passes underneath the nail plate and the color changes from pink to white. Other pigments can be deposited under the nail plate, coloring this space from yellow, such as in onychomycosis, or green due to pyocyanin, or red due to hemosiderin. Onycholysis is not specific, as it can be observed in traumatic, inflammatory, infective, or neoplastic disorders.

25.6.1 Traumatic Onycholysis

It can involve the fingernails, but is very common in the toenails. The detachment of the nail plate due to traumas frequently appears bilateral and symmetrical. At onychoscopy the line of detachment of the plate from the bed is linear, regular, and smooth and surrounded by a normally pale pink bed, without hyperkeratosis (Fig. 25.18) [17, 18]. The subungual space is usually whitish to yellow, and small black drops corresponding to splinter hemorrhages can be present.

25.6.2 Nail Psoriasis

Typical clinical signs of nail psoriasis in children are onycholysis, pitting, and subungual hyperkeratosis. Dermatoscopy allows the detection of subclinical signs that can be very helpful for a definitive diagnosis of nail psoriasis in doubtful cases. In onycholysis due to psoriasis, onychoscopy shows the presence of a bright orange-yellow border surrounding the distal edge of the detachment (Fig. 25.19), a

slightly dented margin, and several splinter hemorrhages. Moreover, the salmon patches appear irregular in shape and size, with a color from red to orange [19, 20].

25.6.3 Nail Lichen Planus

It is uncommon in children, and usually milder than in adults, with persistent nail damage rare [21–23]. Onychoscopy enhances visualization of the longitudinal fissures of the nail plate due to lichen planus. Dermatoscopic observation of the proximal nail plate allows the evaluation the disease's course in a short time, since it shows the newly formed nail plate. Dorsal pterygium, rare in children, is the result of an irreversible damage to the nail matrix, with absent nail plate and adhesion of the dorsal skin to the nail bed, and formation of a v-shaped extension of the proximal nail fold (Fig. 25.20). Permanent anonychia is also rare.

25.6.4 Nail Lichen Striatus

It is rare and almost exclusively seen in children. Clinically, one nail is involved and shows lichenoid abnormalities with longitudinal ridging restricted to its medial or lateral portion that continues with skin lesions characterized by papules or verrucous scales along the Blaschko lines. Dry dermatoscopy permits to observe as longitudinal fissuring and distal splitting (Fig. 25.21).

25.6.5 Onychomycosis

It is rare in children [24]. Toenails are more affected than fingernails. Predisposing factors include dermatophyte infection in other members of family, nail abnormalities, traumatic factors, and immunodepression. Although mycological examination is needed to confirm the diagnosis [24], onychoscopy can be utilized to differentiate onychomycosis from other diseases causing onycholysis, such as traumatic onycholysis or nail psoriasis, Moreover, dermatoscopy may be used as a guide to identify the best spot to obtain adequate samples for mycological examination [25].

In children dermatoscopy may be particularly useful in distal subungual onychomycosis (DSO) and white superficial onychomycosis (WSO) [26]. In DSO the fungi reach the nail unit through the hyponychium and invade the space under the nail plate progressing proximally. The dermatoscopic patterns of DSO are (Fig. 25.22) jagged edge of the proximal margin of the onycholytic area, with sharp structures (spikes) directed toward the proximal fold; white-yellow longitudinal striae in the onycholytic nail plate; an overall appearance of the affected nail plate in parallel bands of different colors, resembling aurora borealis and in fact named *Aurora borealis pattern* [18, 24]; and a "ruin appearance" of the subungual hyperkeratosis due to the accumulation of dermal debris of fungal invasion, better visible with frontal dermatoscopy [27]. In WSO the fungi colonize the dorsal surface of the nail plate with the production of small white spots on one or several nails [28]; dermatoscopy shows a nail plate with several small white opaque and friable patches (Fig. 25.23). For a better result of the image, performing dry dermatoscopy is recommended because the use of a gel for interface induces a partial disappearance of the white discoloration that includes the scales irregularly spread on the nail surface. In punctate leukonychia, in which dermatoscopy shows single or multiple opaque white regular spots within the nail plate, no change is visible after the application of the gel.

25.7 Nail Tumors

Tumors in children are rare. The most frequent is represented by the subungual exostosis, a benign tumor of the bone of the distal phalanx occurring beneath the nails. Clinically it appears as a hard nodular lesion with a keratotic surface that elevates the nail plate, frequently in the great toenail. Usually, it affects children under 18 years of age who do sport with jumps because traumas increase the frequency. Dermatoscopy of the nail plate shows a white onycholysis of the distal part detached by the mass of the tumor. Frontal dermatoscopy is very helpful to better see the typical aspect of exostosis. It appears as a light red-colored nodule, pink or flesh-colored in color, which projects beyond the inner free edge of the nail (Figs. 25.24a, b). During the performing of the dermatoscopy, it is possible to notice the harness of the lesion on touching the lesion and the patient feels pain. The overlying nail can become brittle and may be lifted and the surface of the lesion may be hyperkeratotic. For this reason, it is better to use dermatoscopy with gel to observe nail plate onycholysis and dry dermatoscopy in the frontal view to better evidence the alteration of the surface of the lesion.

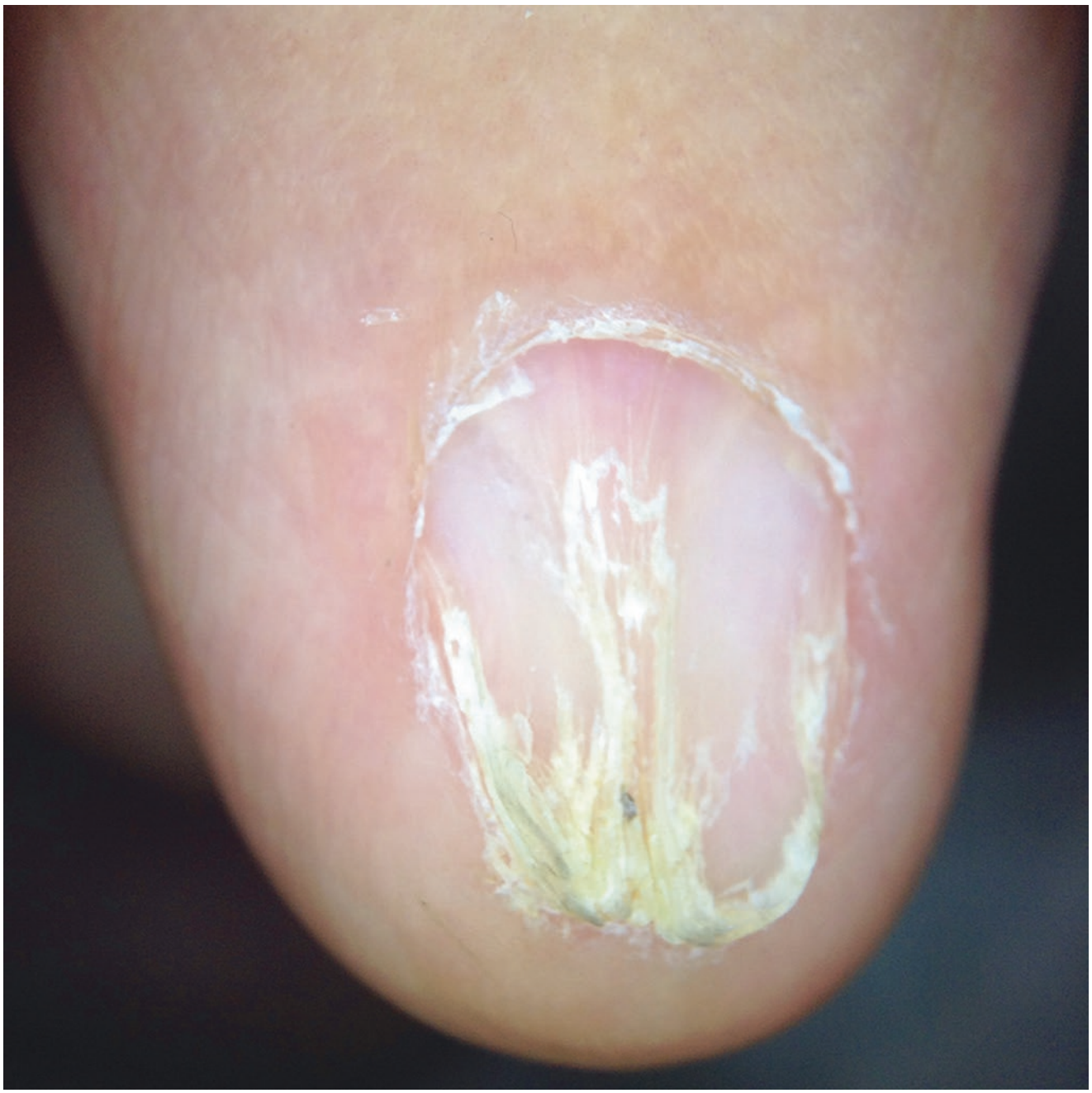

Fig. 25.1 Triangular lunula in a patient affected by nail-patella syndrome. Dermatoscopy (×10): horizontal basis of the triangle at the level of the proximal margin and point of the triangle directed toward the distal margin

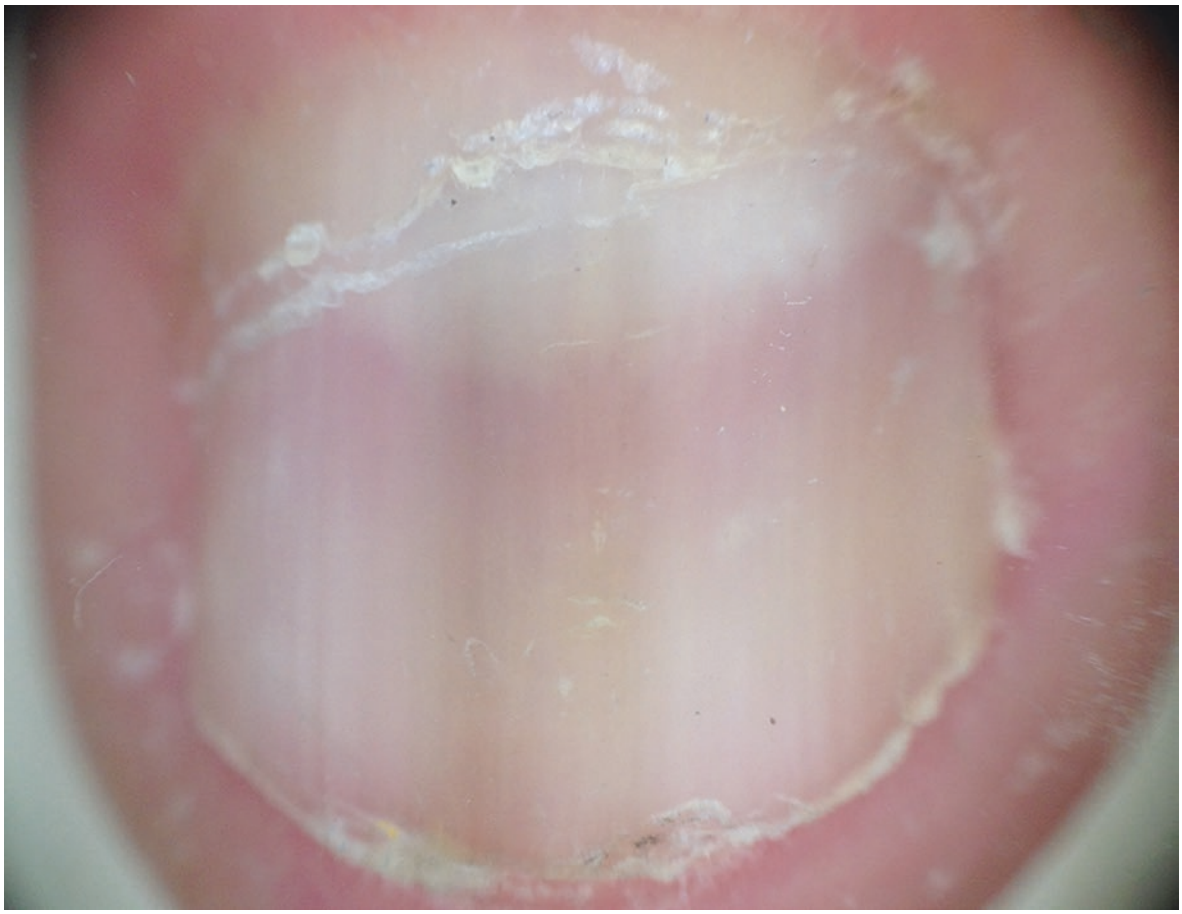

Fig. 25.2 Nail biting. Dermatoscopy (×10): band of melanonychia

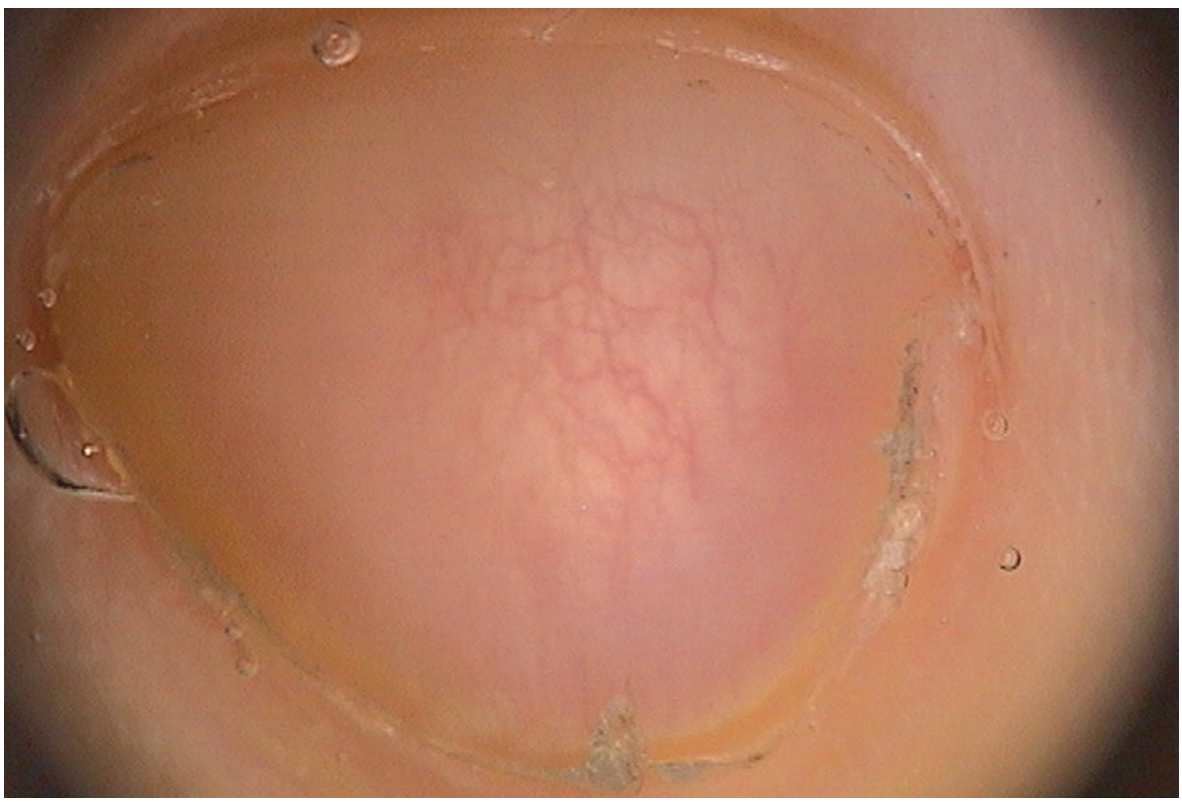

Fig. 25.3 Pseudoclubbing due to osteochondroma. Dermatoscopy (×20): longitudinally curved nail plate

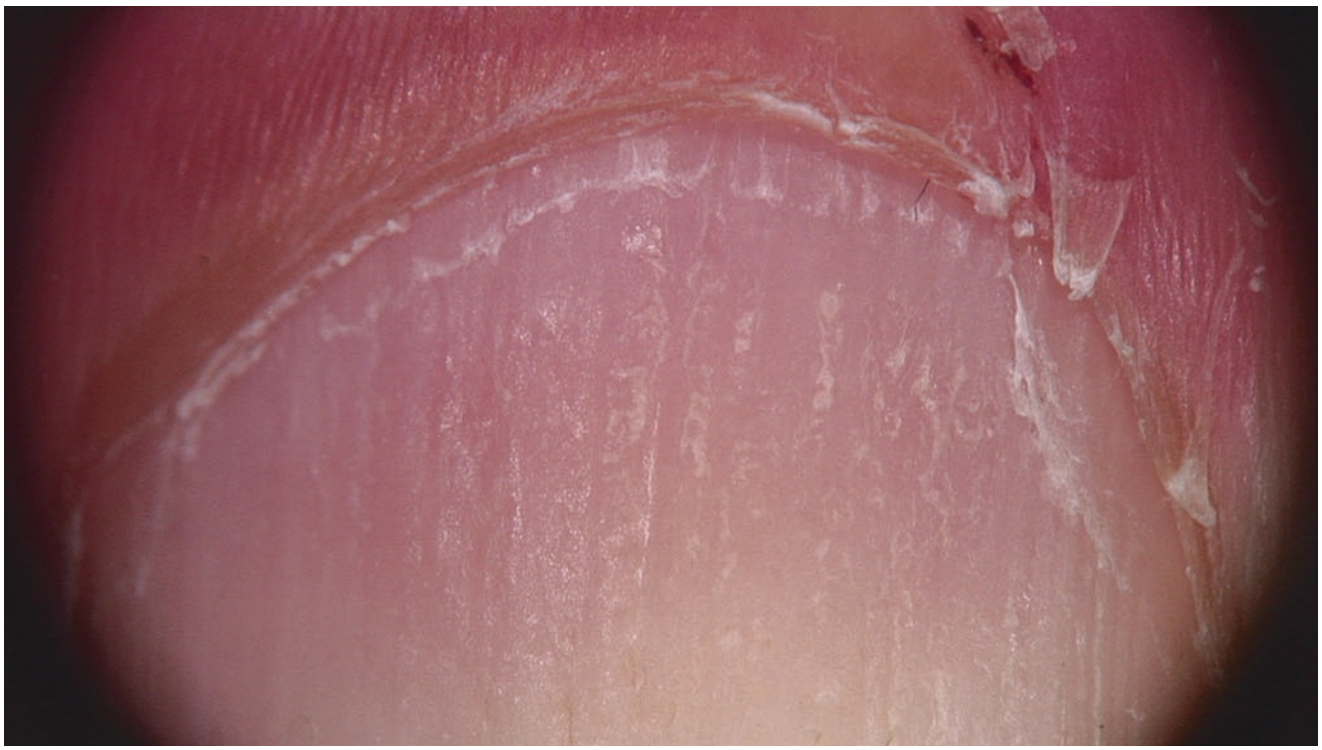

Fig. 25.4 Pitting in nail psoriasis: dermatoscopy (×20)

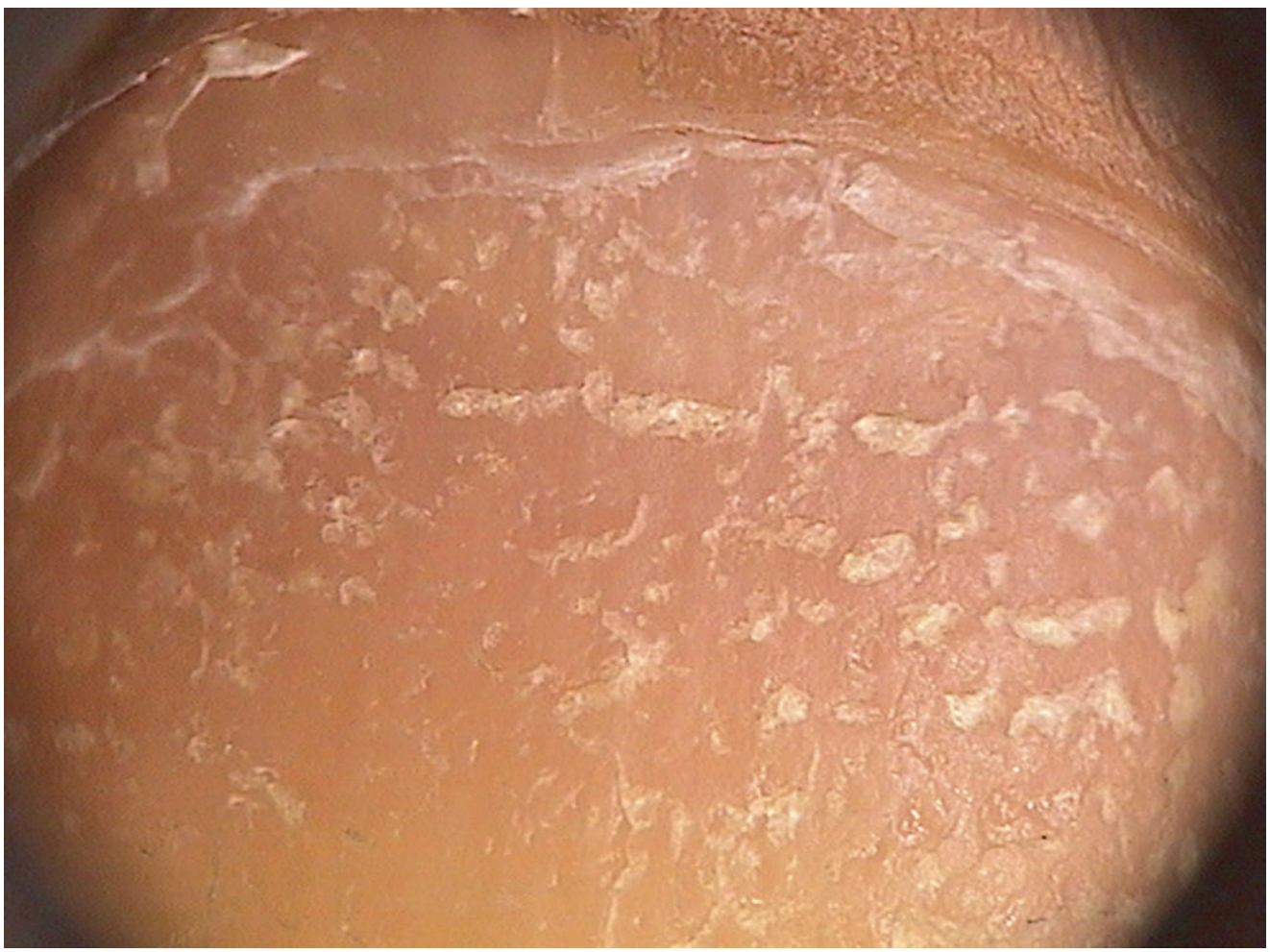

Fig. 25.5 Pitting in patient affected by alopecia areata: dry dermatoscopy (×10)

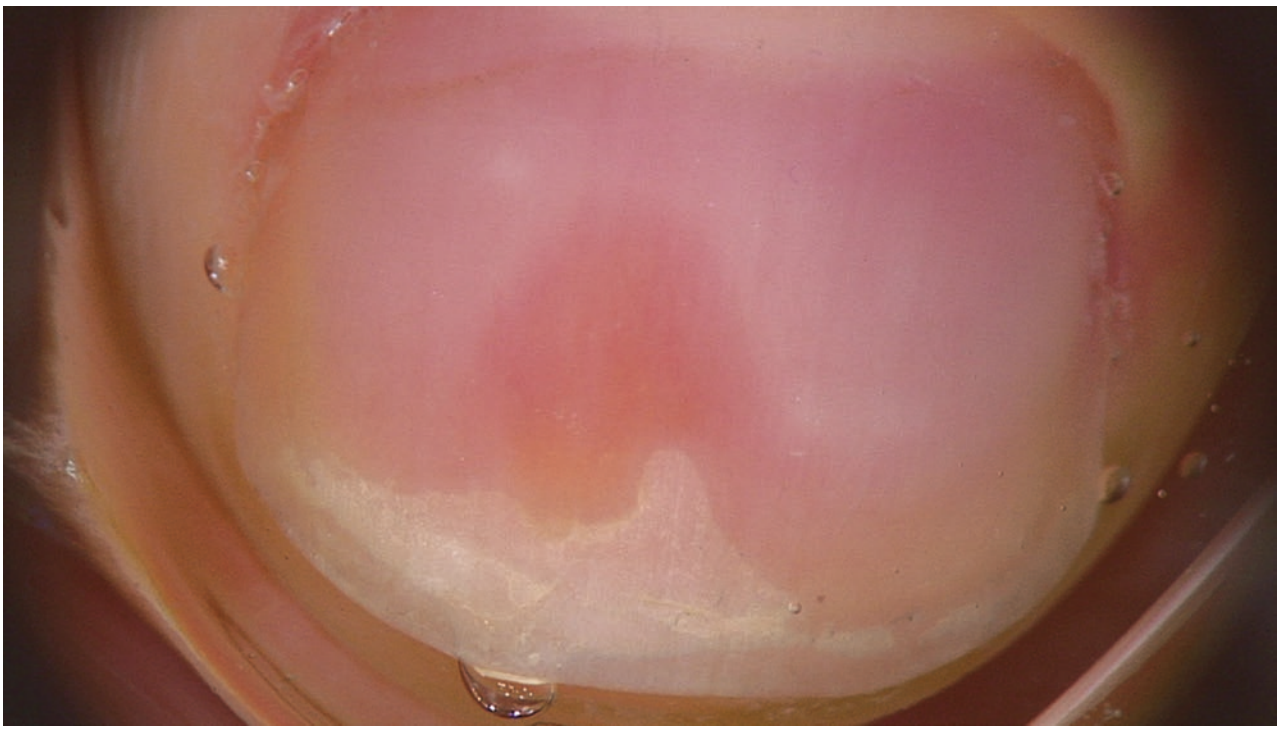

Fig. 25.6 Finger sucking. Dermatoscopy (×20): onycholysis of the distal nail plate

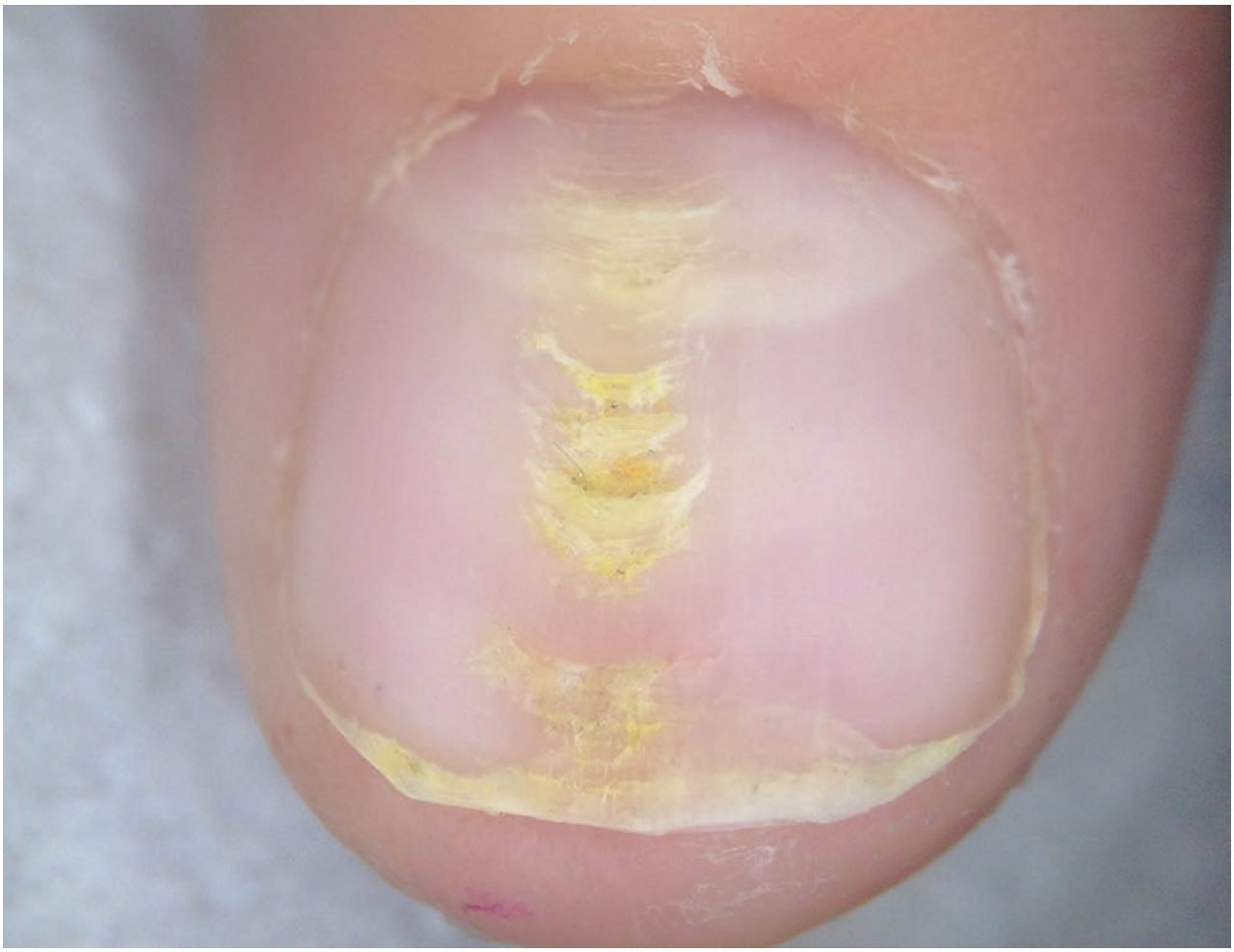

Fig. 25.7 Habit tic nail deformity. Dermatoscopy (×10): multiple transversal ridging

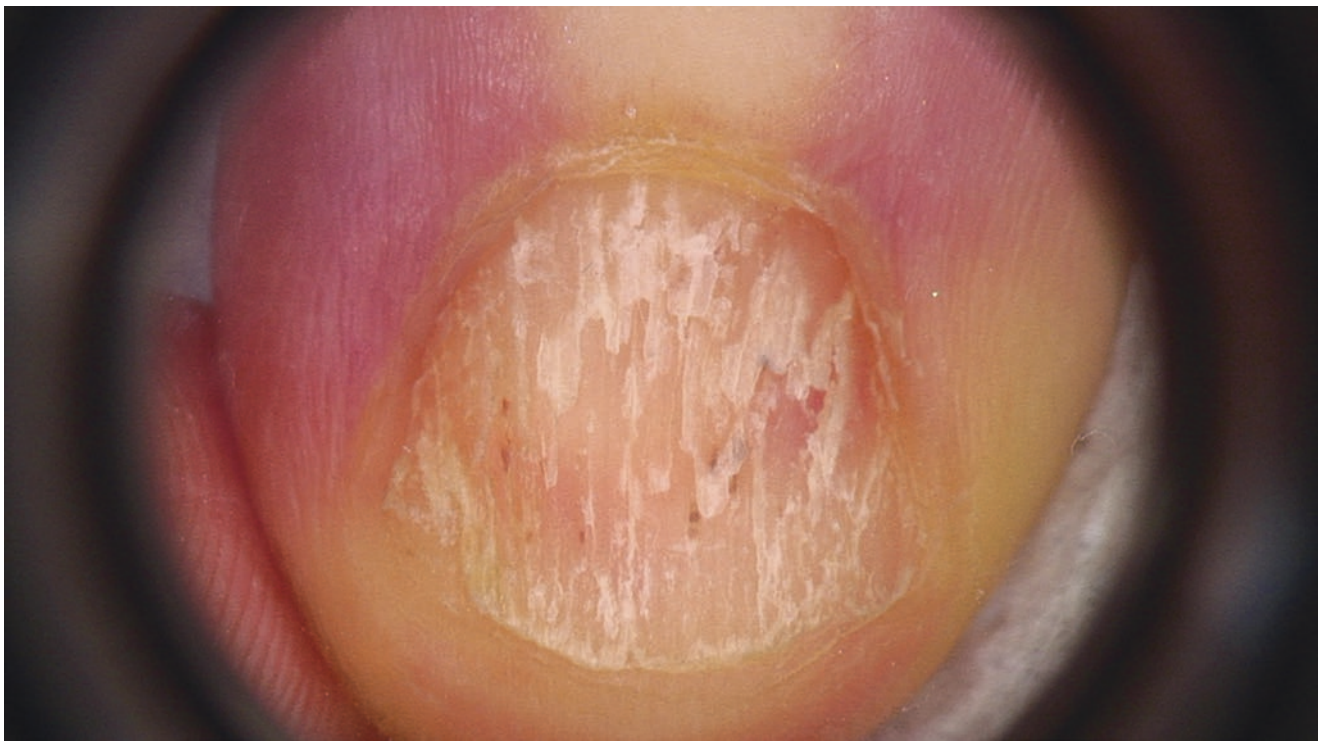

Fig. 25.8 Severe trachyonychia due to alopecia areata: dry dermatoscopy (×10)

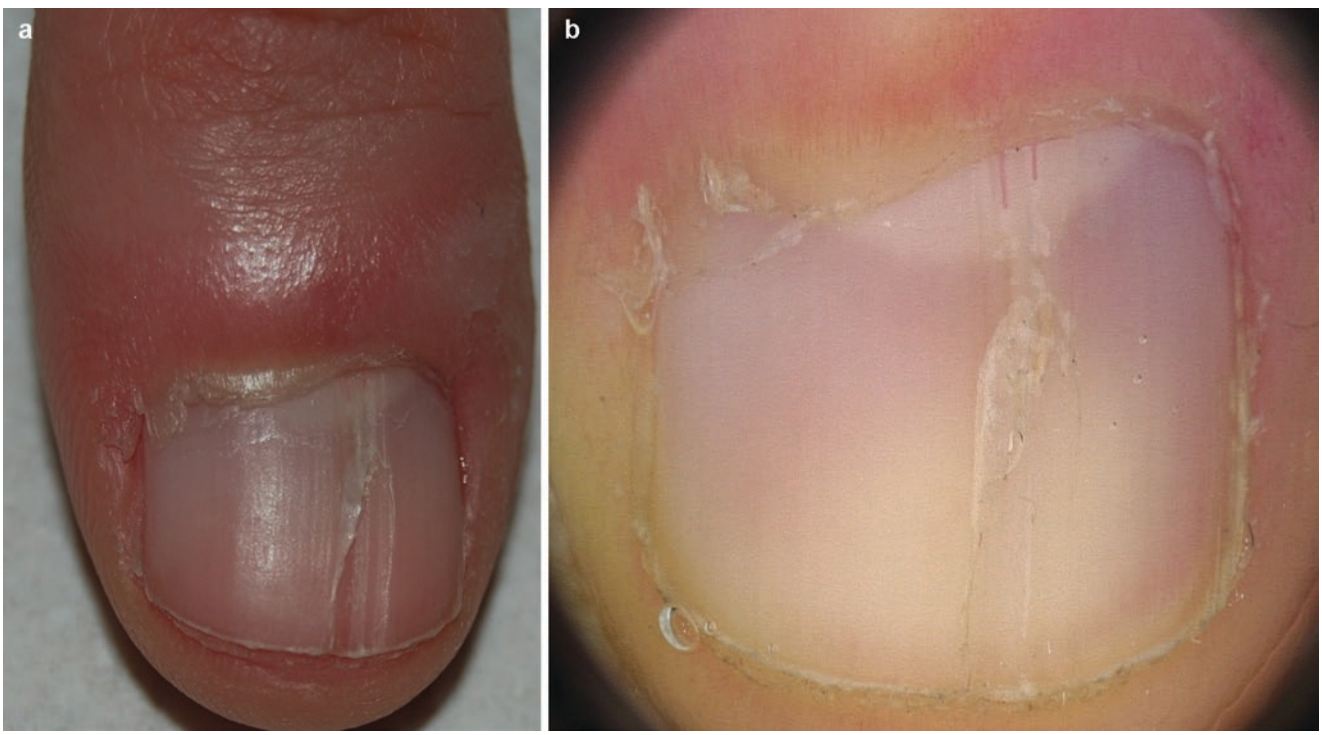

Fig. 25.9 (**a**) Fibrokeratoma under the nail plate with onycholysis. (**b**) Dermatoscopy (×10)

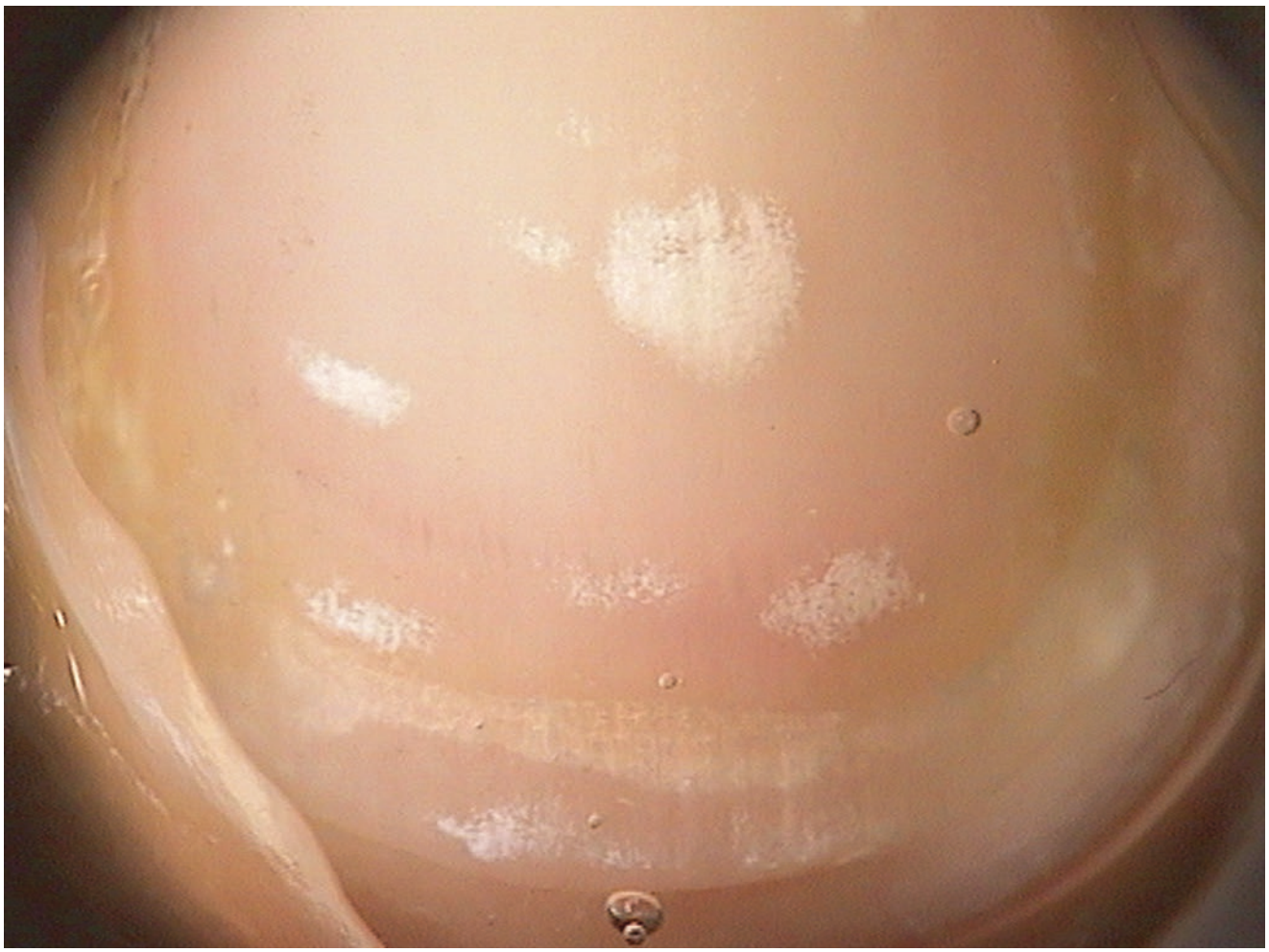

Fig. 25.10 Punctate leukonychia in fingernails: dermatoscopy with gel (×10)

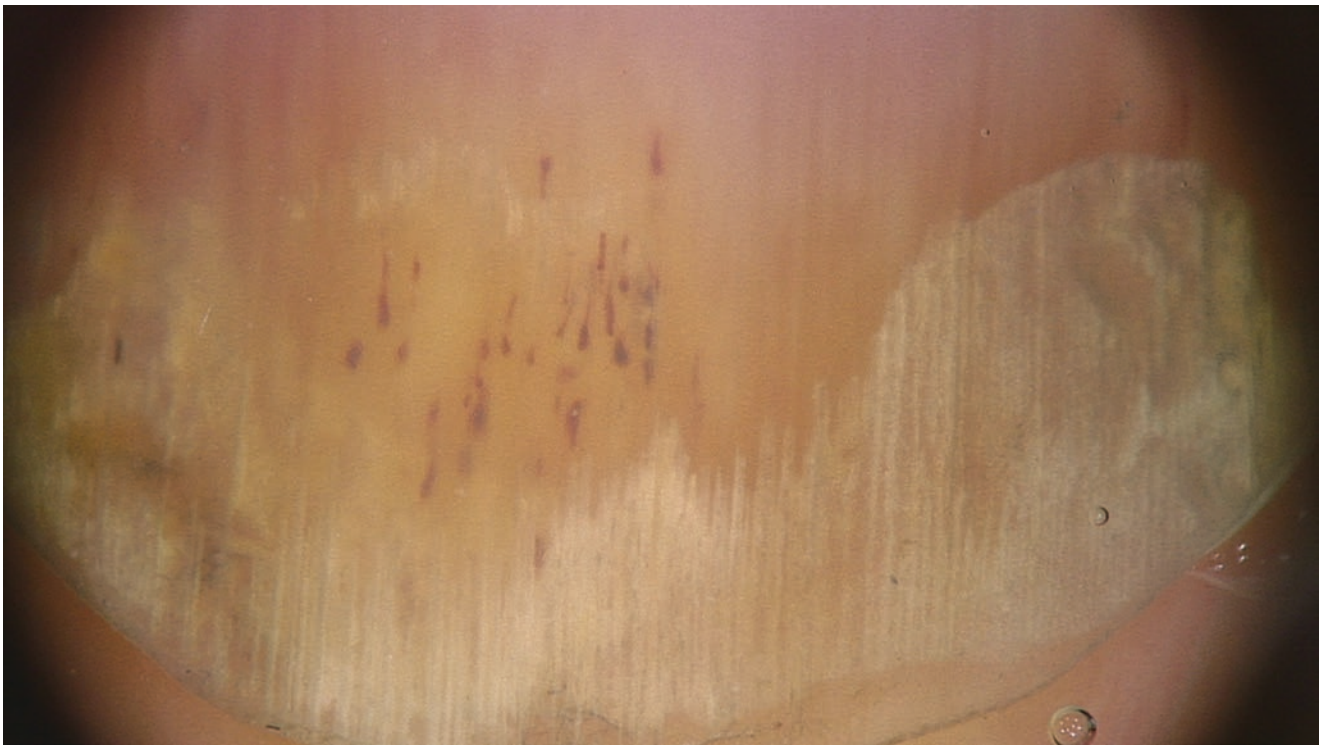

Fig. 25.11 Nail psoriasis. Dermatoscopy with gel (×20): multiple splinter hemorrhages

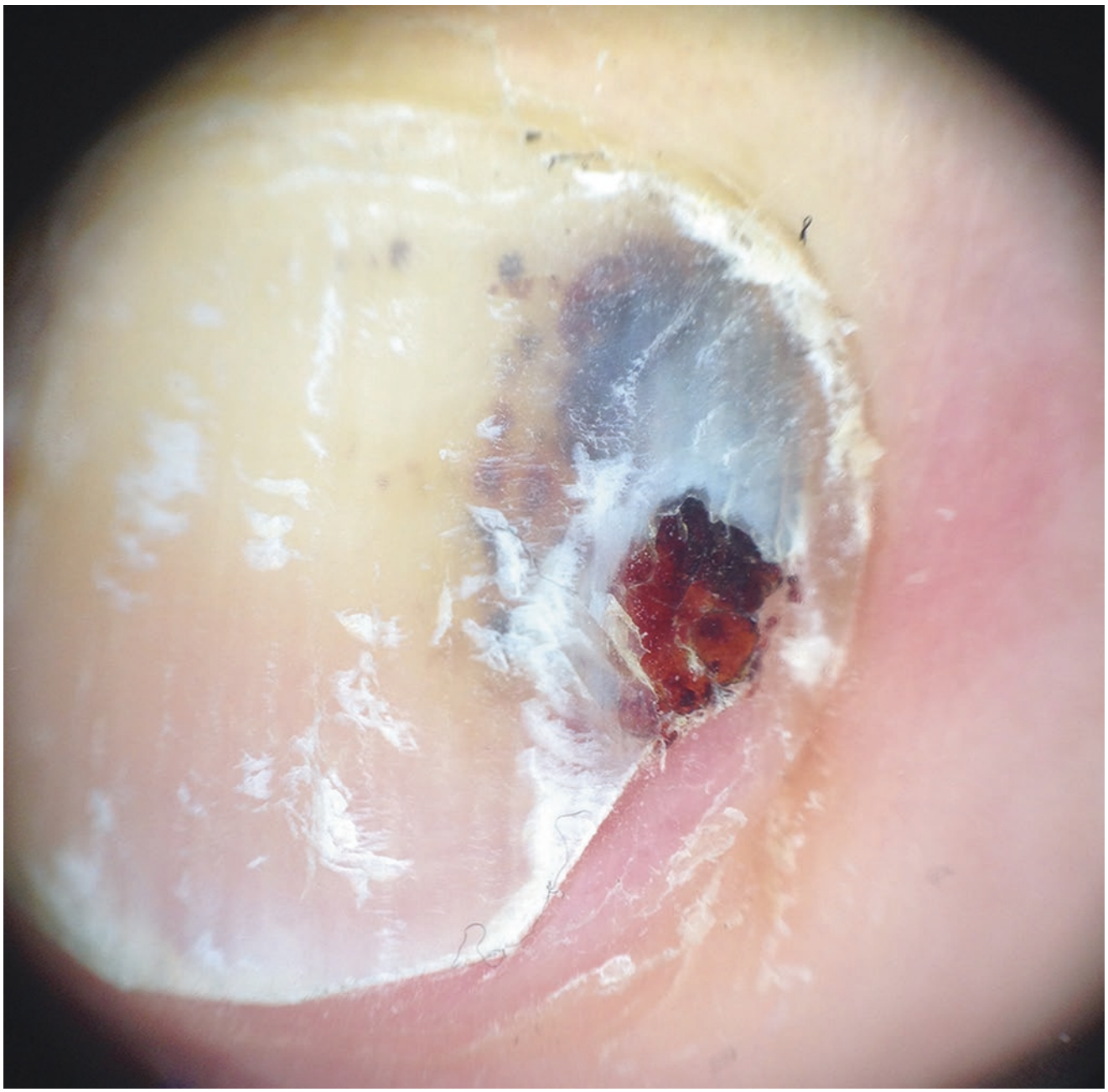

Fig. 25.12 Subungual hematoma. Dermatoscopy (×10): globular pattern and peripheral fading

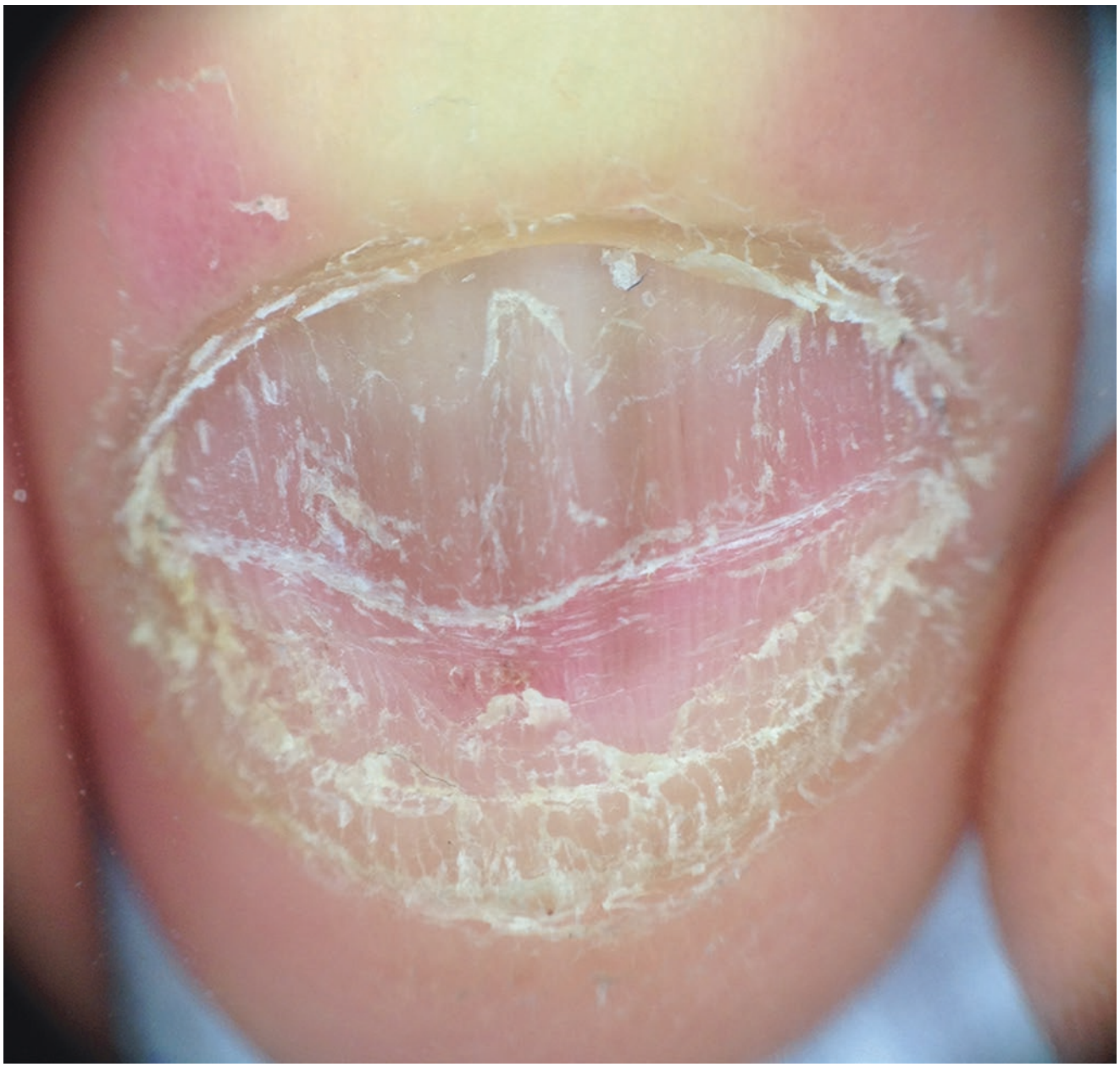

Fig. 25.13 Onychotillomania in a girl. Dermatoscopy (×10): longitudinal band due to melanocytic activation

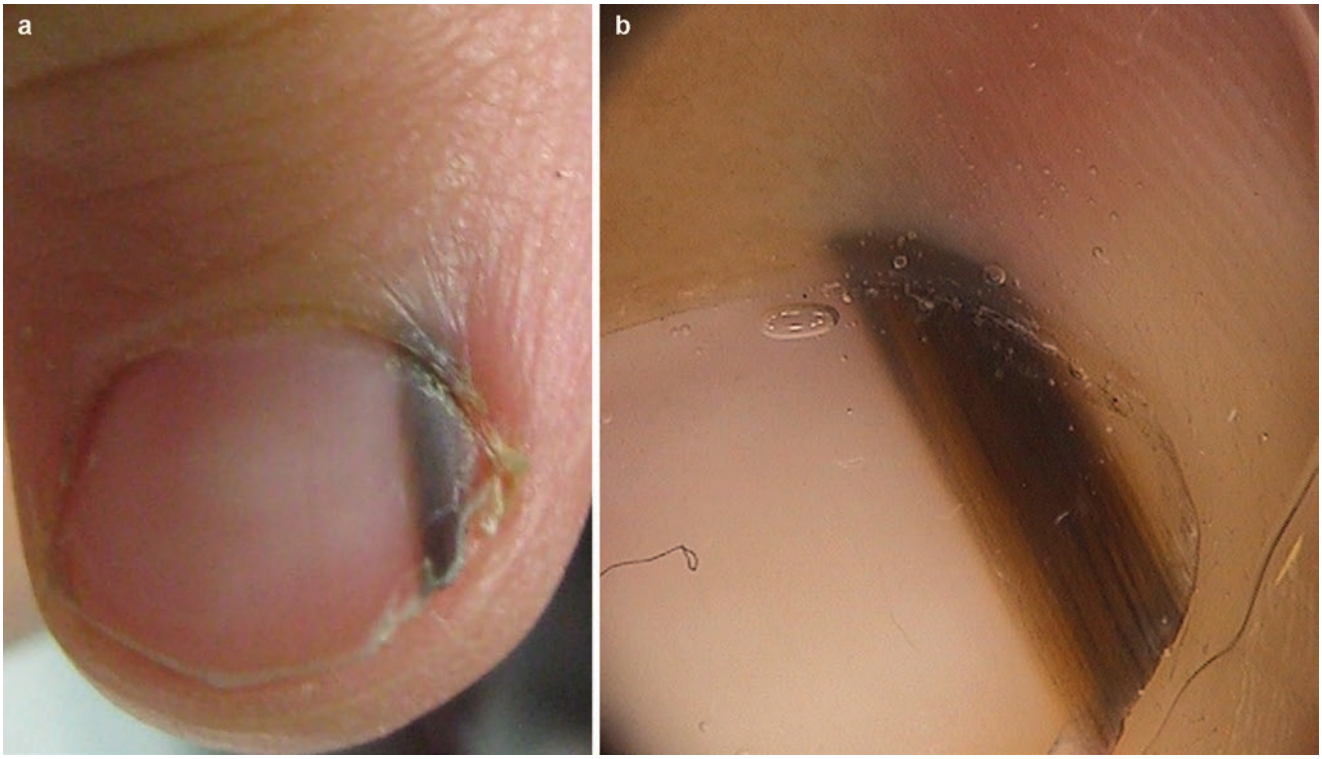

Fig. 25.14 Nail matrix nevus. (**a**) Longitudinal band of nail matrix nevus in a 2-year-old boy. (**b**) Dermatoscopy (×20)

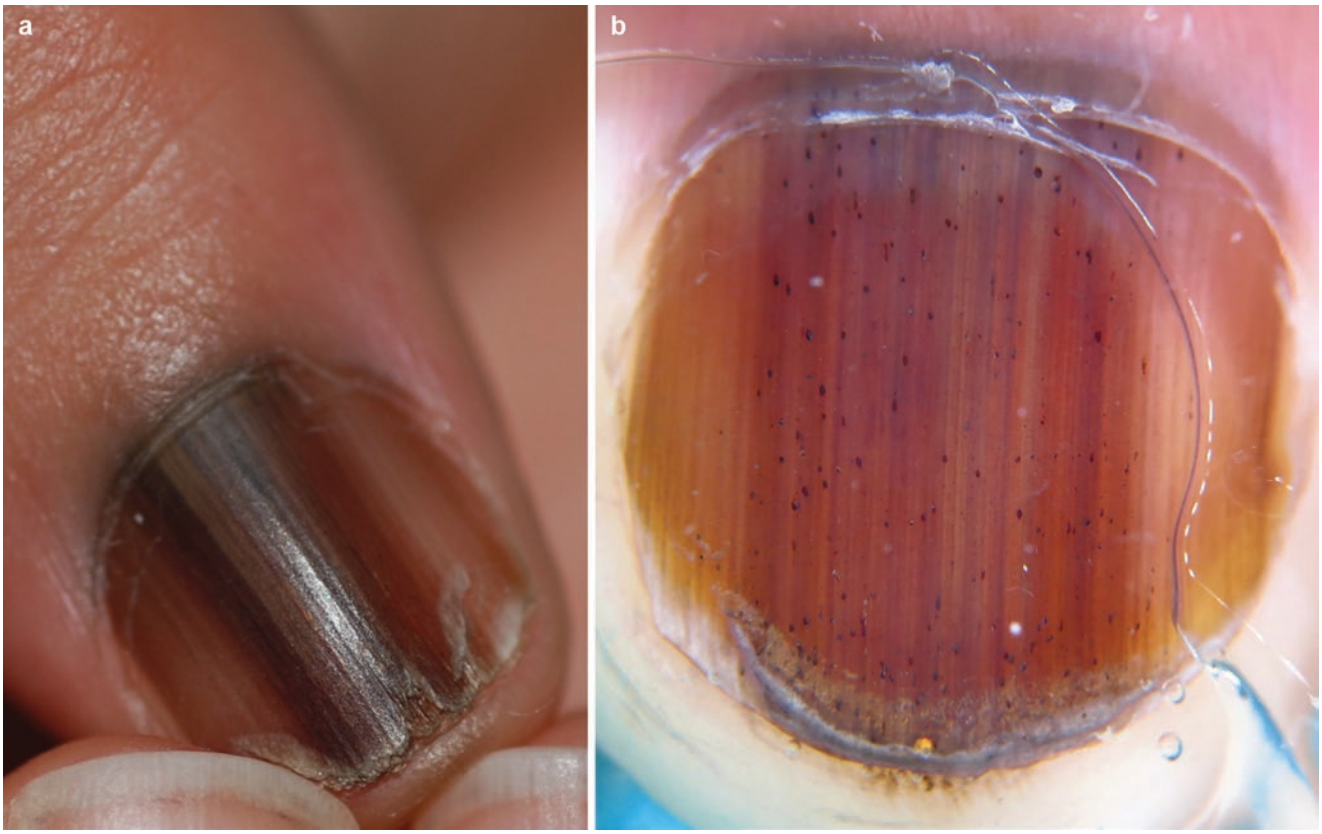

Fig. 25.15 (**a**) Nail matrix nevus of the whole nail. (**b**) Dermatoscopy (×20)

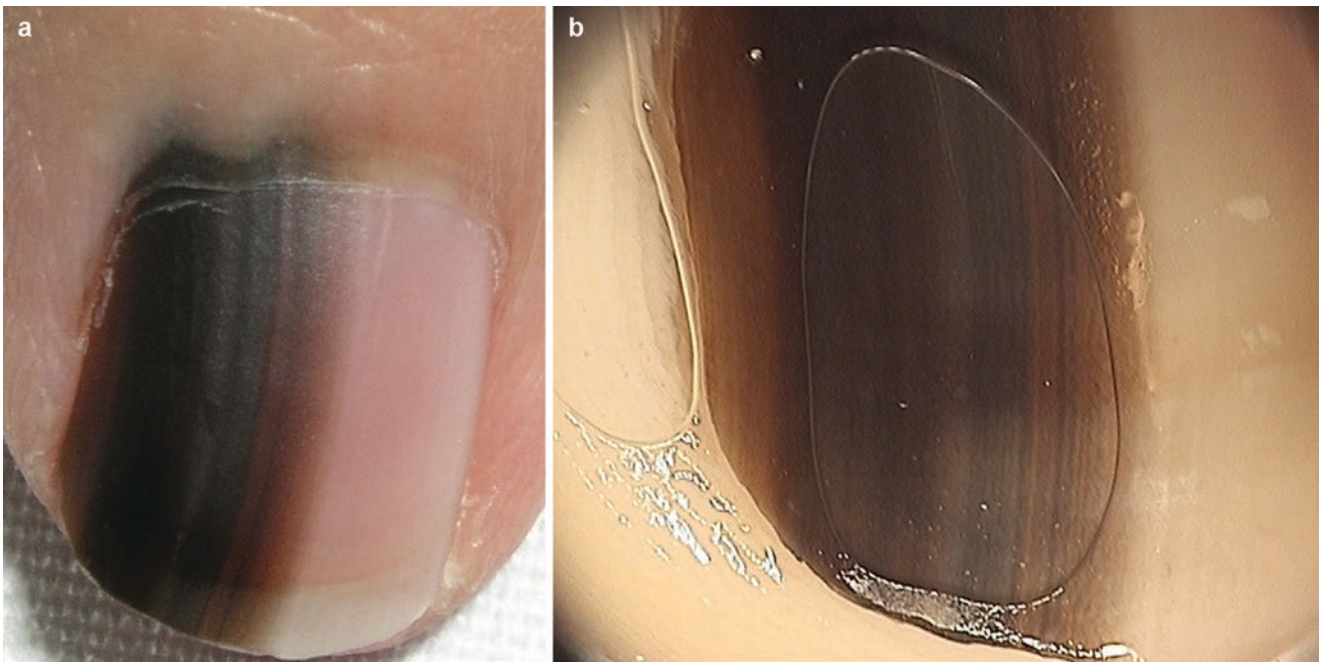

Fig. 25.16 (**a**) Nail melanoma in an 18-year-old girl. (**b**) Dermatoscopy (×20)

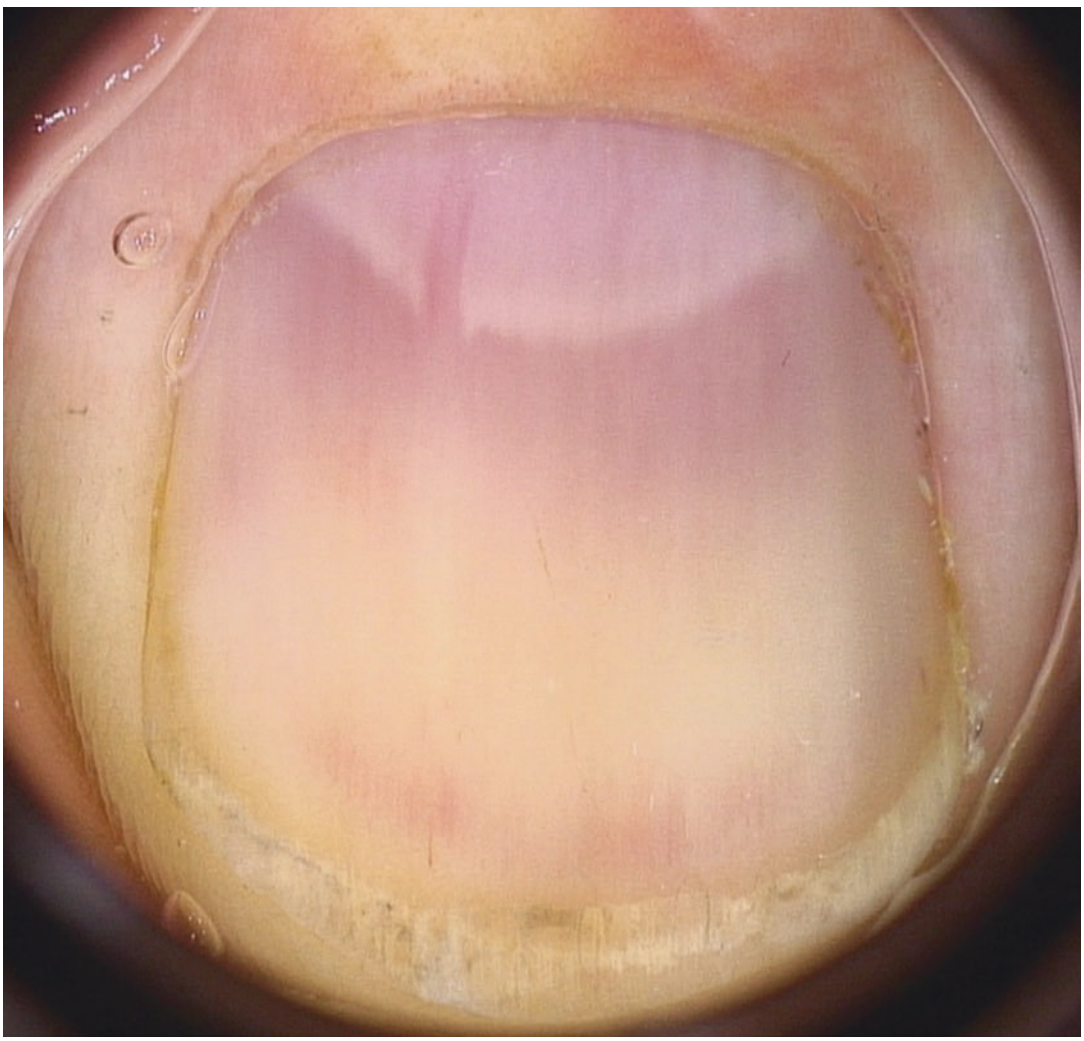

Fig. 25.17 Onychopapilloma. Dermatoscopy (×10) showing erythronychia

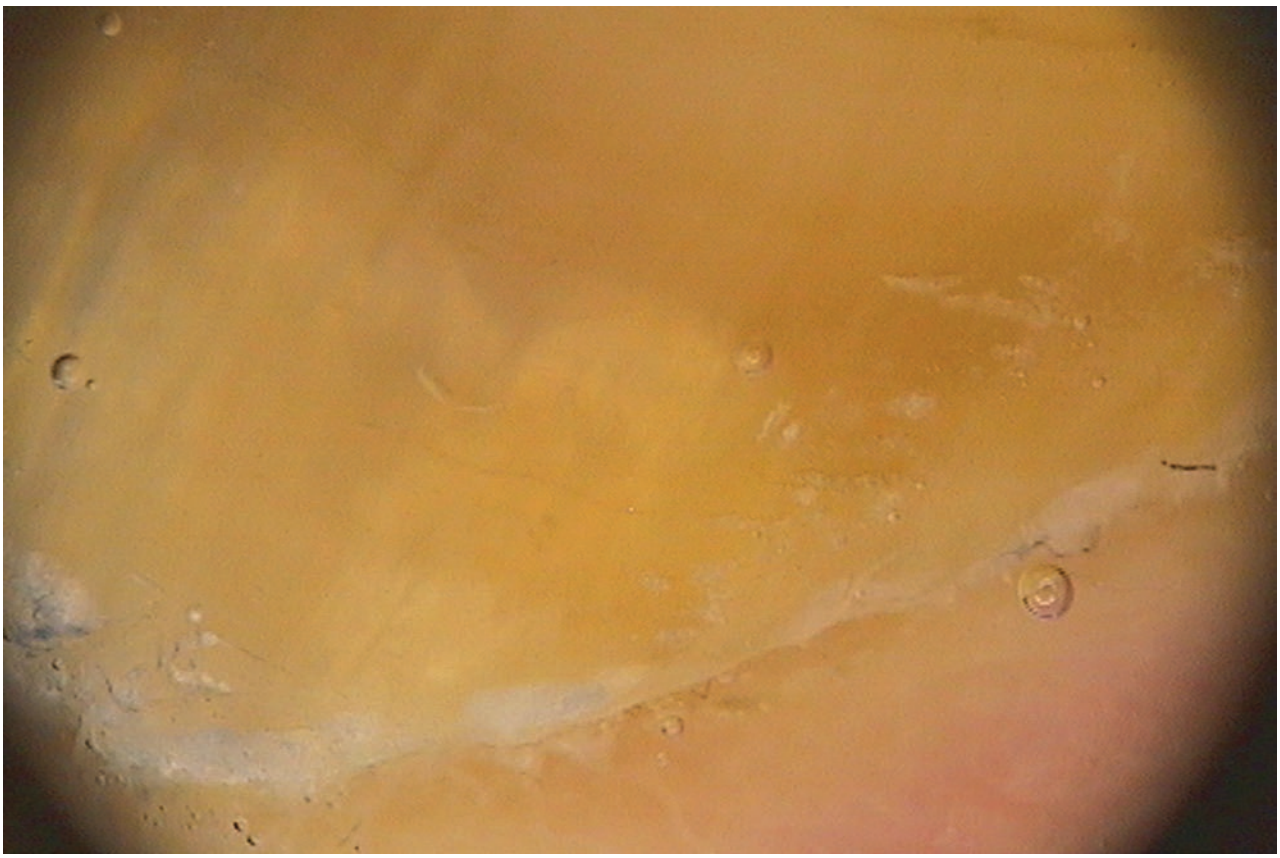

Fig. 25.18 Traumatic onycholysis. Dermatoscopy (×20) of the linear proximal margin of the onycholytic area

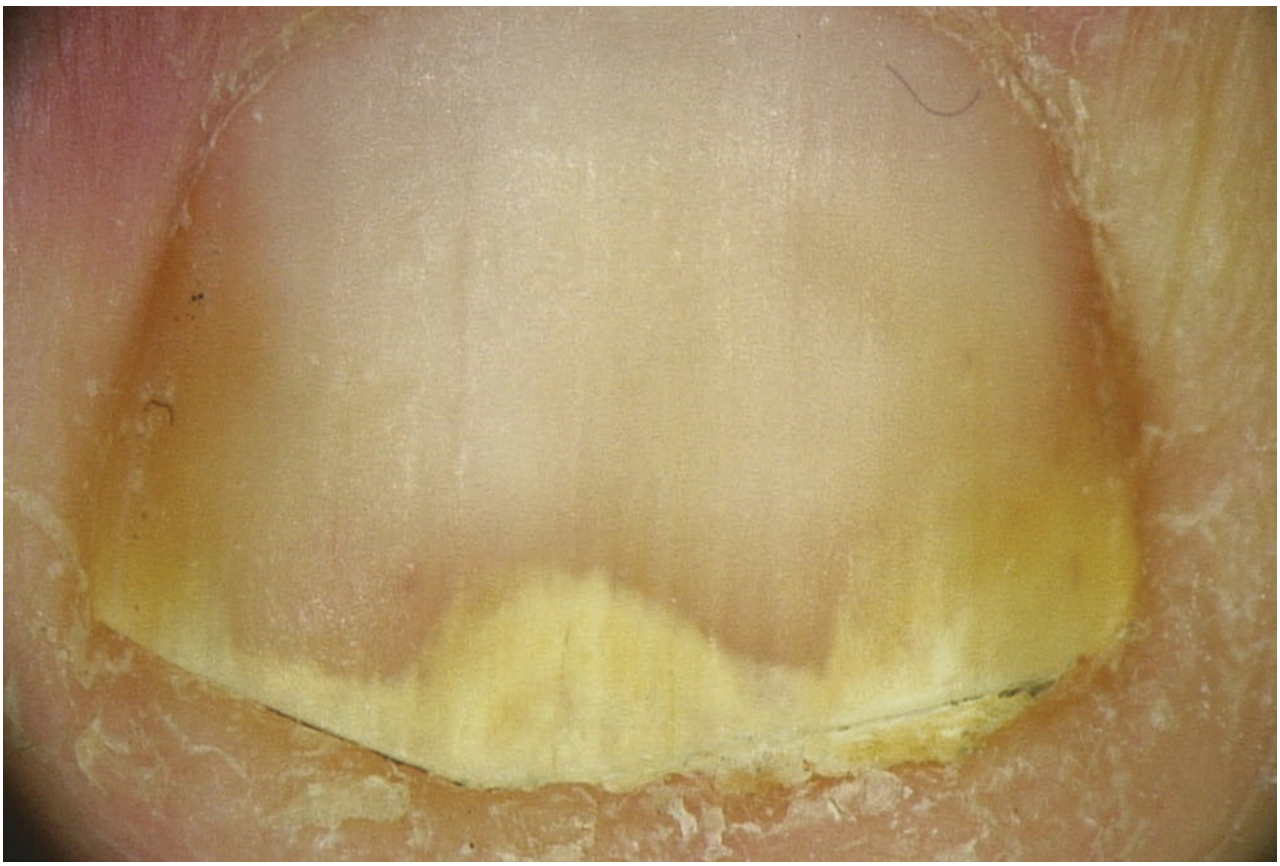

Fig. 25.19 Nail psoriasis. Dermatoscopy (×10) with a bright-orange-yellow border surrounding the distal edge of the detachment

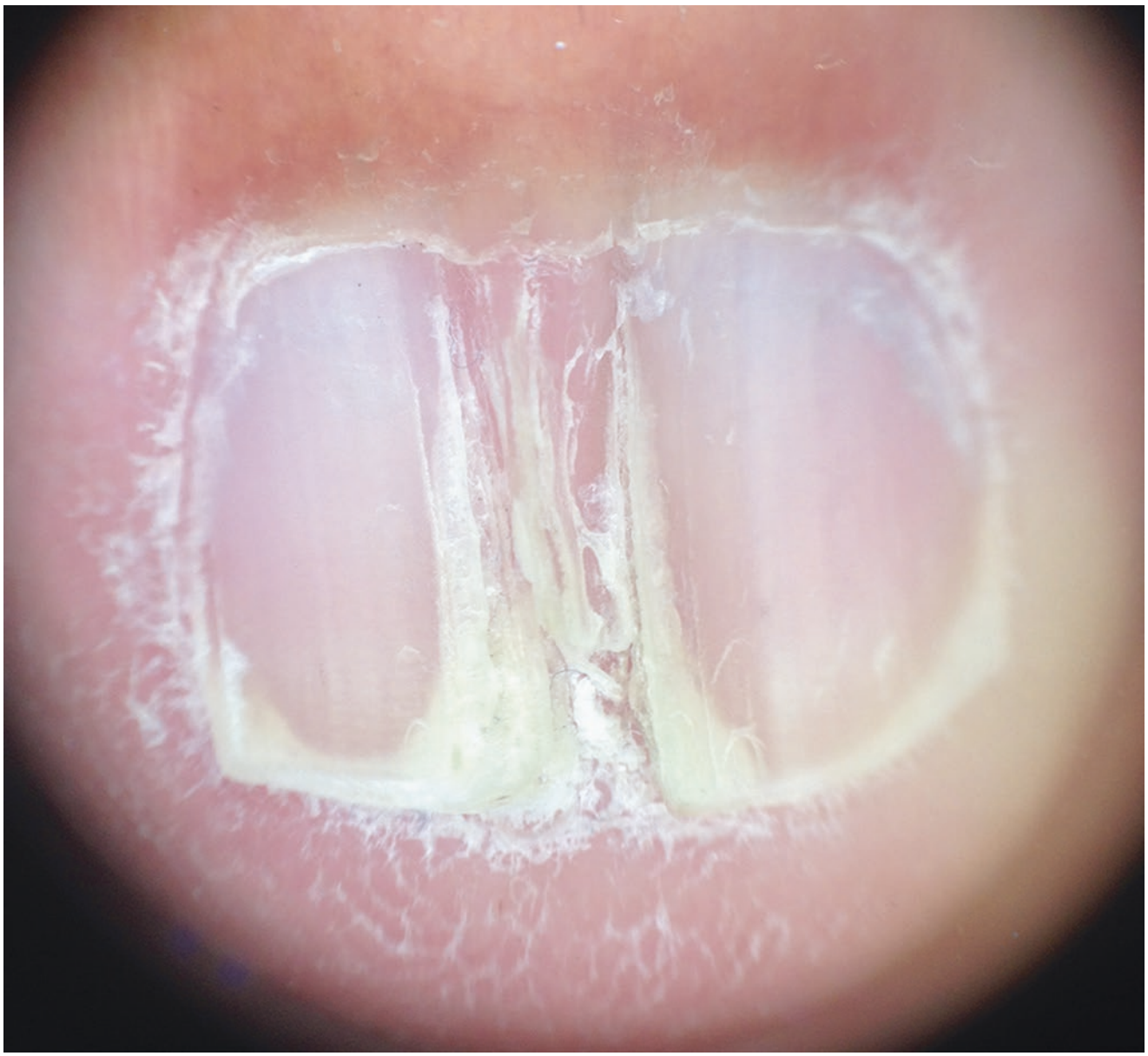

Fig. 25.20 Toenail lichen planus. Dry dermatoscopy (×10): dorsal pterygium

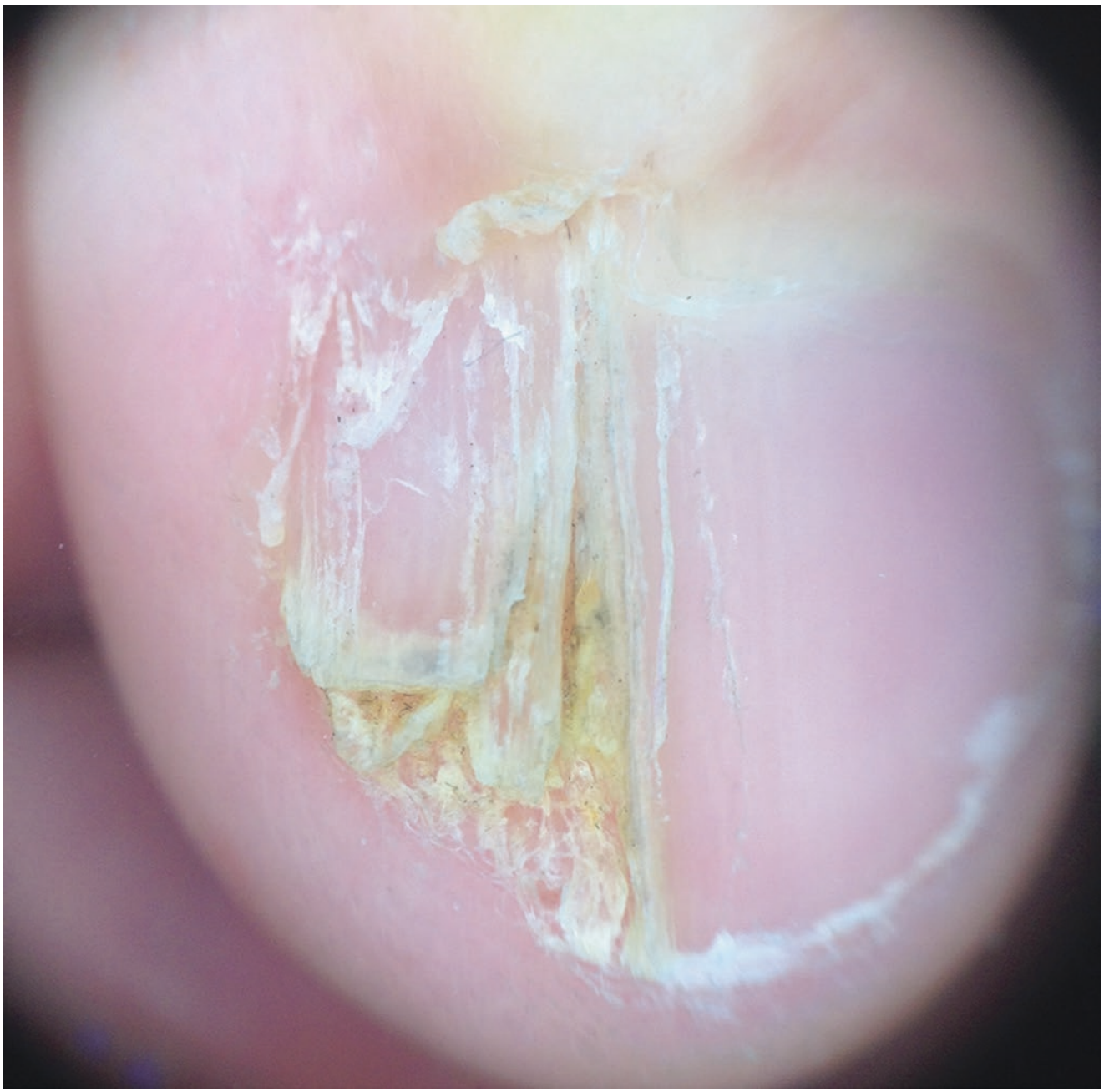

Fig. 25.21 Nail lichen striatus of the first right thumb. Dermatoscopy (×10) of the nail plate

Fig. 25.22 Distal subungual onychomycosis. Dermatoscopy (×20): fringed proximal margin of the onycholytic area and spikes

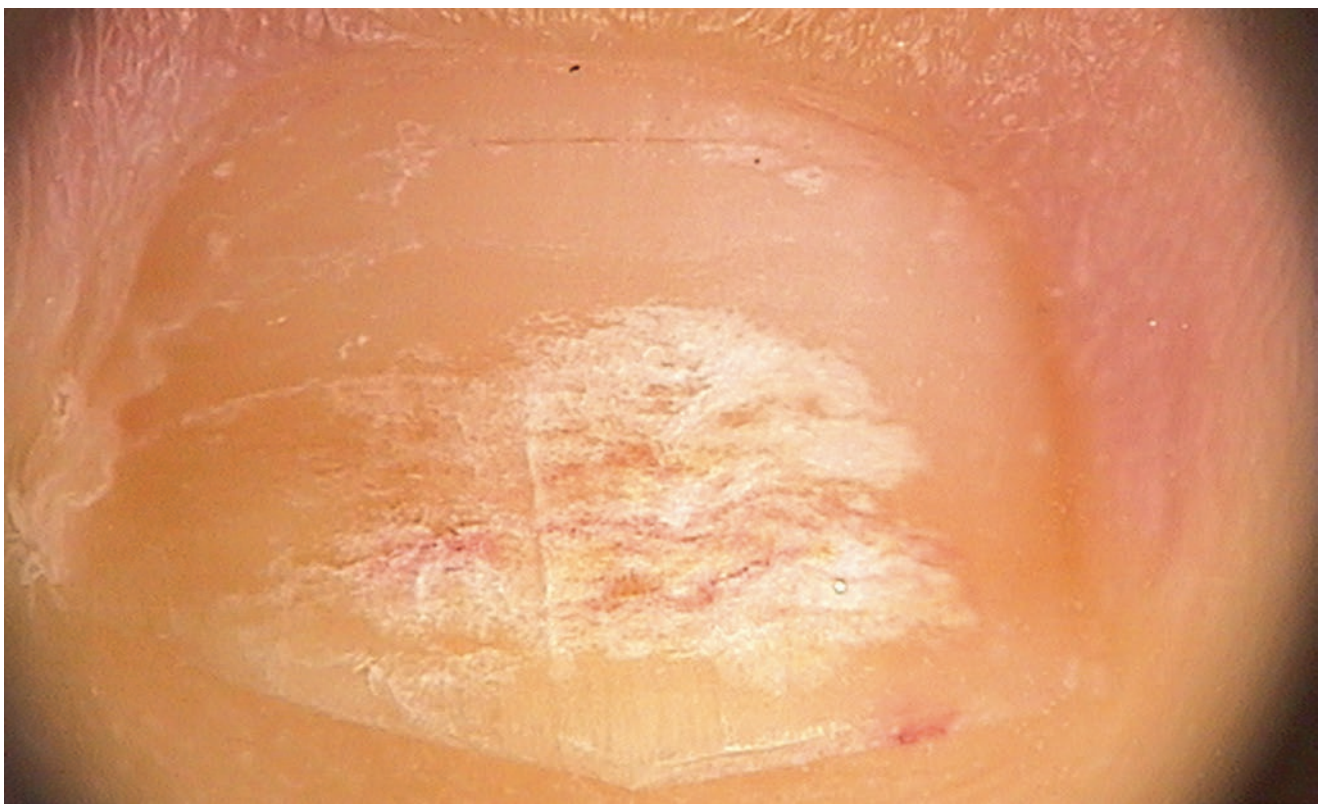

Fig. 25.23 White superficial onychomycosis. Dermatoscopy (×20): small, white, opaque, and friable patch on the nail plate

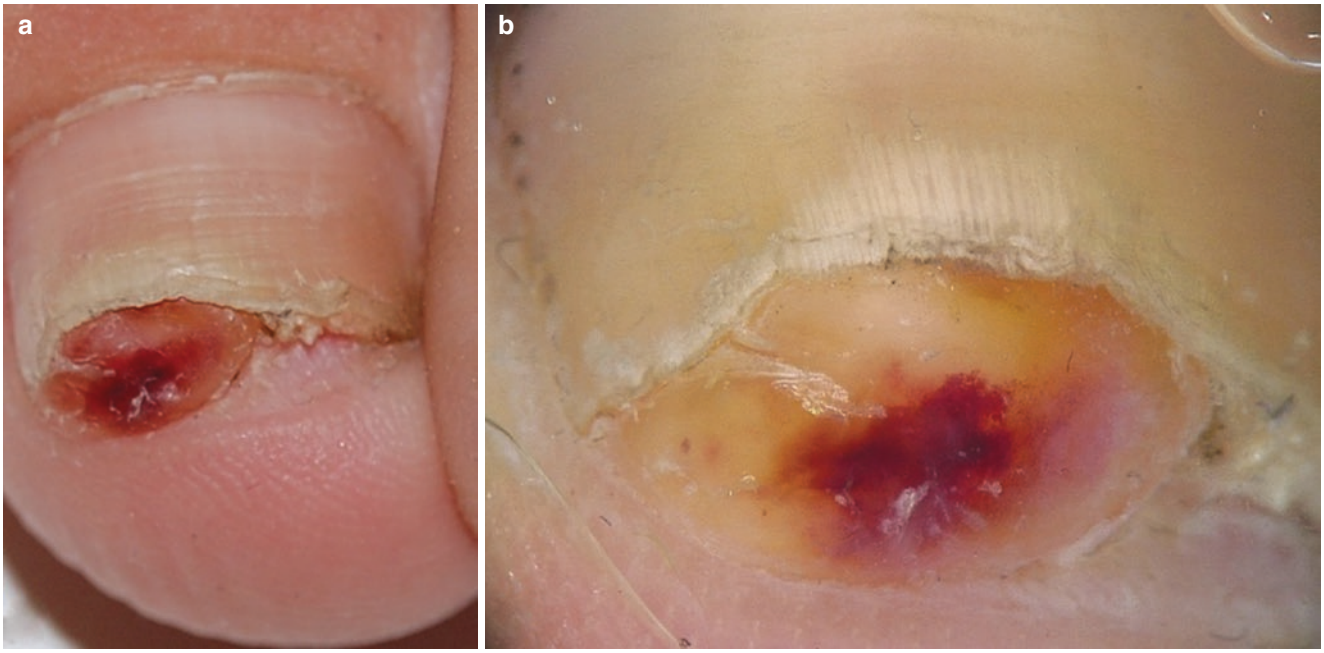

Fig. 25.24 (**a**) Subungual exostosis of the left toenail. (**b**) Frontal dermatoscopy (×40)

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Part VI

Miscellaneous Disorders

Pseudoxanthoma Elasticum

26

Giuseppe Micali, Simona Boscaglia, Maria Rita Nasca, and Francesco Lacarrubba

26.1 Definition

Pseudoxanthoma elasticum (PXE) is a rare genetic syndrome characterized by progressive mineralization and fragmentation of elastic fibers, called elastorrhexia, affecting different tissues (skin, retina, blood vessels) [1].

26.2 Epidemiology

The exact incidence of PXE is unknown. It is estimated to be from 1/25,000 to 1/100,000 [2]. This wide range is correlated with the difficult diagnosis of PXE, with many cases going unrecognized especially in the past. During the last decades, the incidence has considerably raised due to improved diagnostic techniques allowing an earlier identification of PXE. The first symptoms occur in childhood, with a peak around 10–15 years [3]. It has a female sex predilection (M/F = 1:2) and predominance in some ethnic groups [1].

26.3 Etiology

PXE is caused by mutation of the ABCC6 gene codifying an ATP transmembrane transporter protein, principally expressed in the liver and kidney. The pathogenesis is unclear. A "metabolic" hypothesis suggests that the deficiency of this protein in the liver causes an accumulation of circulating substrates, whereas a "cellular" hypothesis proposes a progressive oxidative stress due to the same deficiency [4].

26.4 Clinical Features

Cutaneous lesions of PXE often represent the first manifestations of the disease and in childhood may be the unique signs before appearance of ocular (visual impairment) and cardiovascular (myocardial infarction, angina pectoris) symptoms [3]. They present as soft, small, yellowish asymptomatic papules confluent in plaques with cobblestone aspect, also known as "Moroccan leather" or "chicken skin" (Figs. 26.1a, 26.2a, and 26.3a) [2]. Preferential localizations are the neck, axillae, antecubital and popliteal fossae, and inguinal and periumbilical areas. Mucosae may be involved with similar lesions, especially the lips and the anogenital mucosa [5].

26.5 Differential Diagnosis

Differential diagnosis of PXE includes cutis laxa, elastosis perforans serpiginosa, perforating periumbilical PXE, and fibroelastolytic papulosis. Some other dermatologic manifestations of hematologic conditions such as beta-thalassemia, sickle cell anemia, and coagulation factor deficiency should also be considered [4].

26.6 Diagnosis

The diagnosis of PXE is based on the criteria codified in 1994 and revised in 2000 by Plomp et al. [2] that are classified as major and minor. Major criteria include clinical or

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histological skin alterations, ocular findings detected by fundoscopy (peau d'orange or angioid streaks), and pathogenic mutation of both alleles of the ABCC6 gene or a first-degree relative with a definite diagnosis of PXE. Minor criteria include other ocular findings (one angioid streak shorter than one disk diameter; "comet signs" in the retina; "wing signs" in the retina) and pathogenic mutation of one allele of the ABCC6 gene. For a definitive diagnosis, the presence of at least two major criteria, not from the same category (skin, eye, or genetic), is necessary [2].

26.7 Dermatoscopy

Dermatoscopy of PXE shows the presence of multiple irregular yellowish areas alternating with prominent linear vessels (Figs. 26.1b, 26.2b, and 26.3b) [6]. In some fields, the yellowish areas coalesce to form parallel strands. Dermatoscopy may be useful for a noninvasive prompt diagnosis of PXE so to appropriately address diagnostic work-up [6, 7].

26.8 Histopathological Correlation

The specific histological pattern of PXE is represented by elastorrhexia, characterized by fragmentation and progressive mineralization of elastic fibers of the mid/low dermis (Fig. 26.4) [4]. Von Kossa stain confirms the presence of calcium deposits (Fig. 26.5). The yellowish hue observed at dermatoscopy may be related both to the elastolysis of elastic fibers and/or to the presence of calcium deposits [6]. The prominent superficial linear vessels may be due to a vascular rearrangement driven by the underlying dermal elastolysis [6].

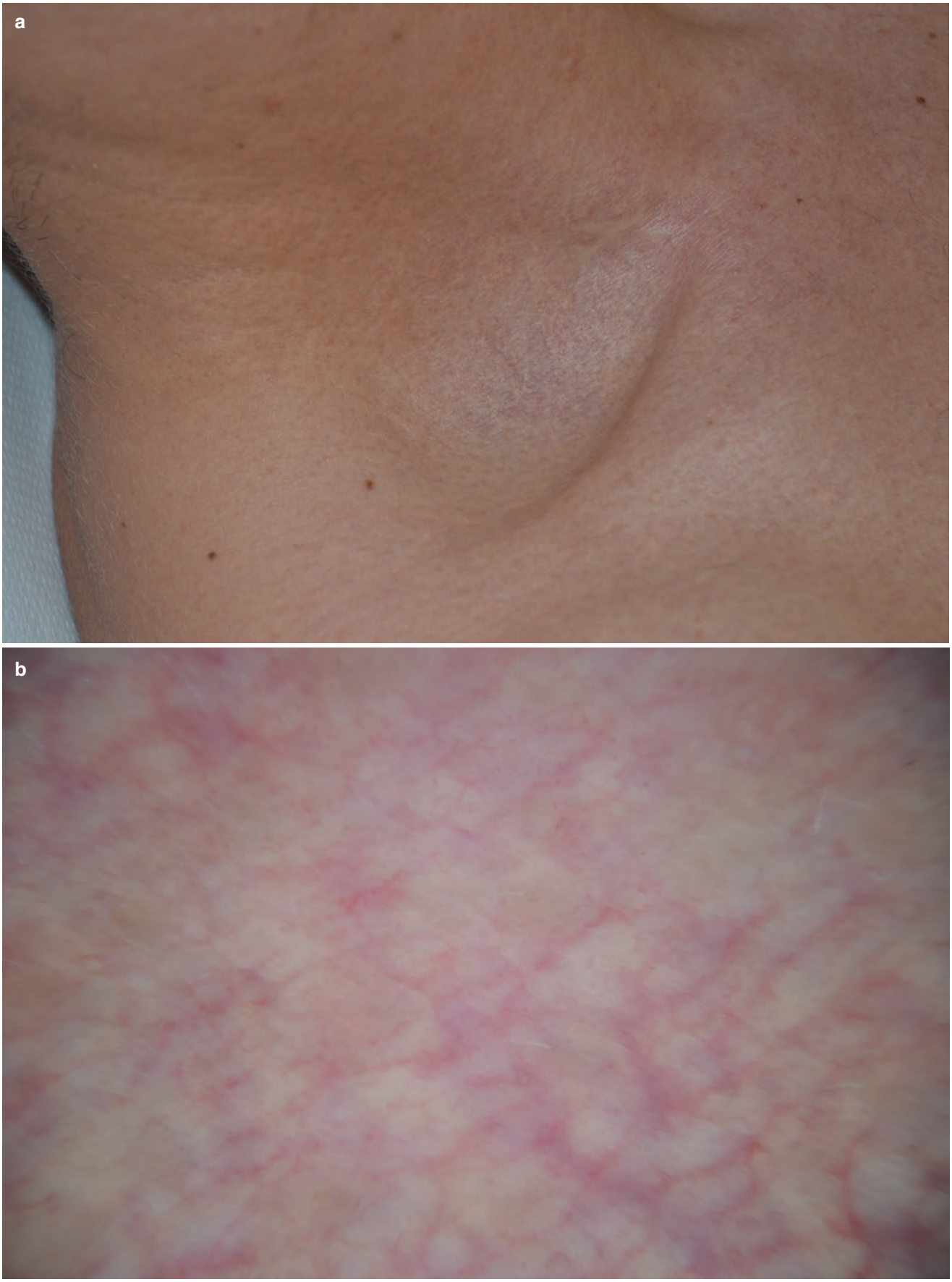

Fig. 26.1 (**a**) Pseudoxanthoma elasticum. Small, skin-colored/yellowish papules of the lateral neck in a 17-year-old boy. (**b**) Dermatoscopy (×10): irregular yellowish areas alternating with prominent linear vessels

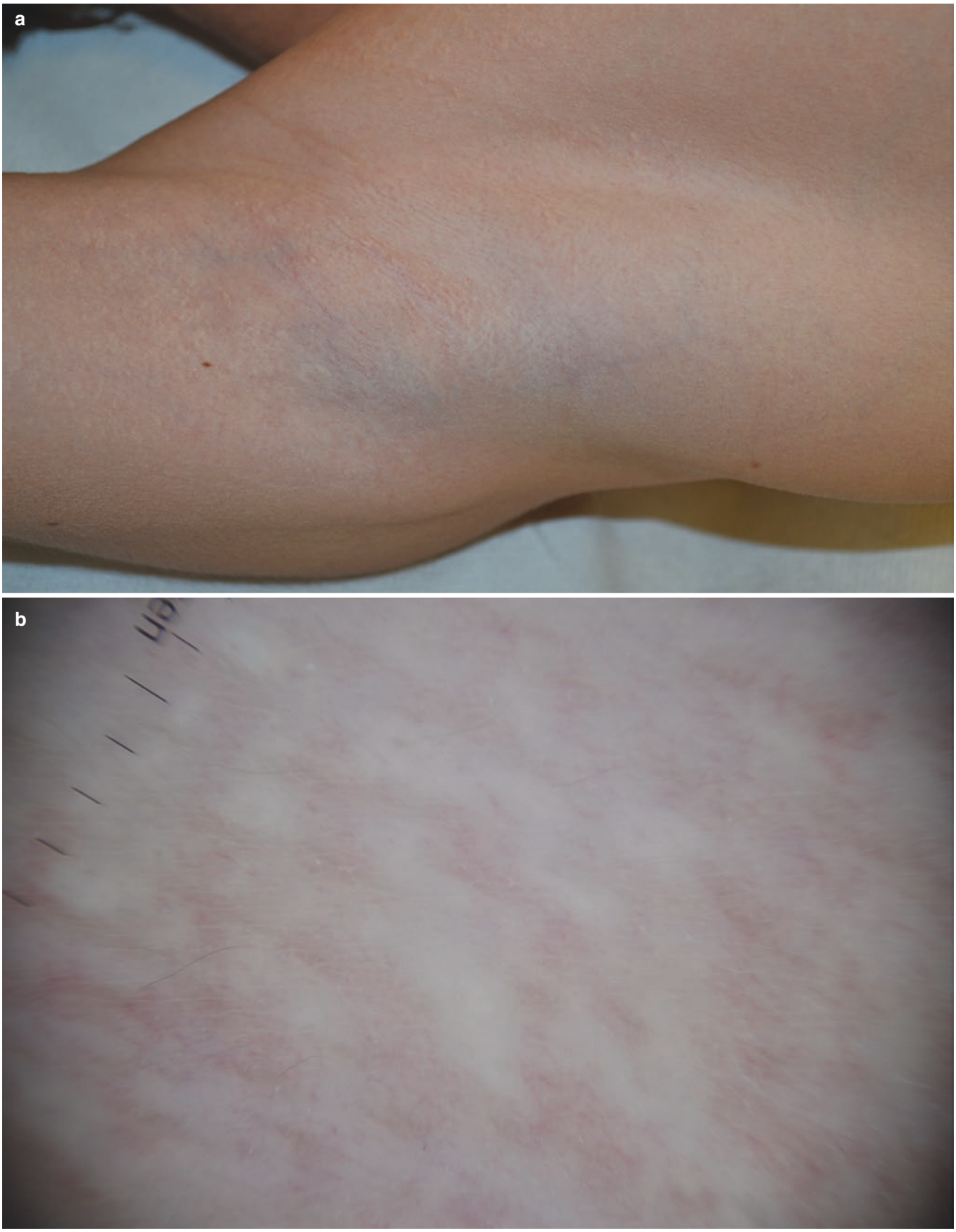

Fig. 26.2 (**a**) Pseudoxanthoma elasticum. Small, yellowish papules of the right axilla of a 13-year-old boy. (**b**) Dermatoscopy (×10): multiple irregular yellowish areas coalescing to form parallel strands alternating with linear vessels

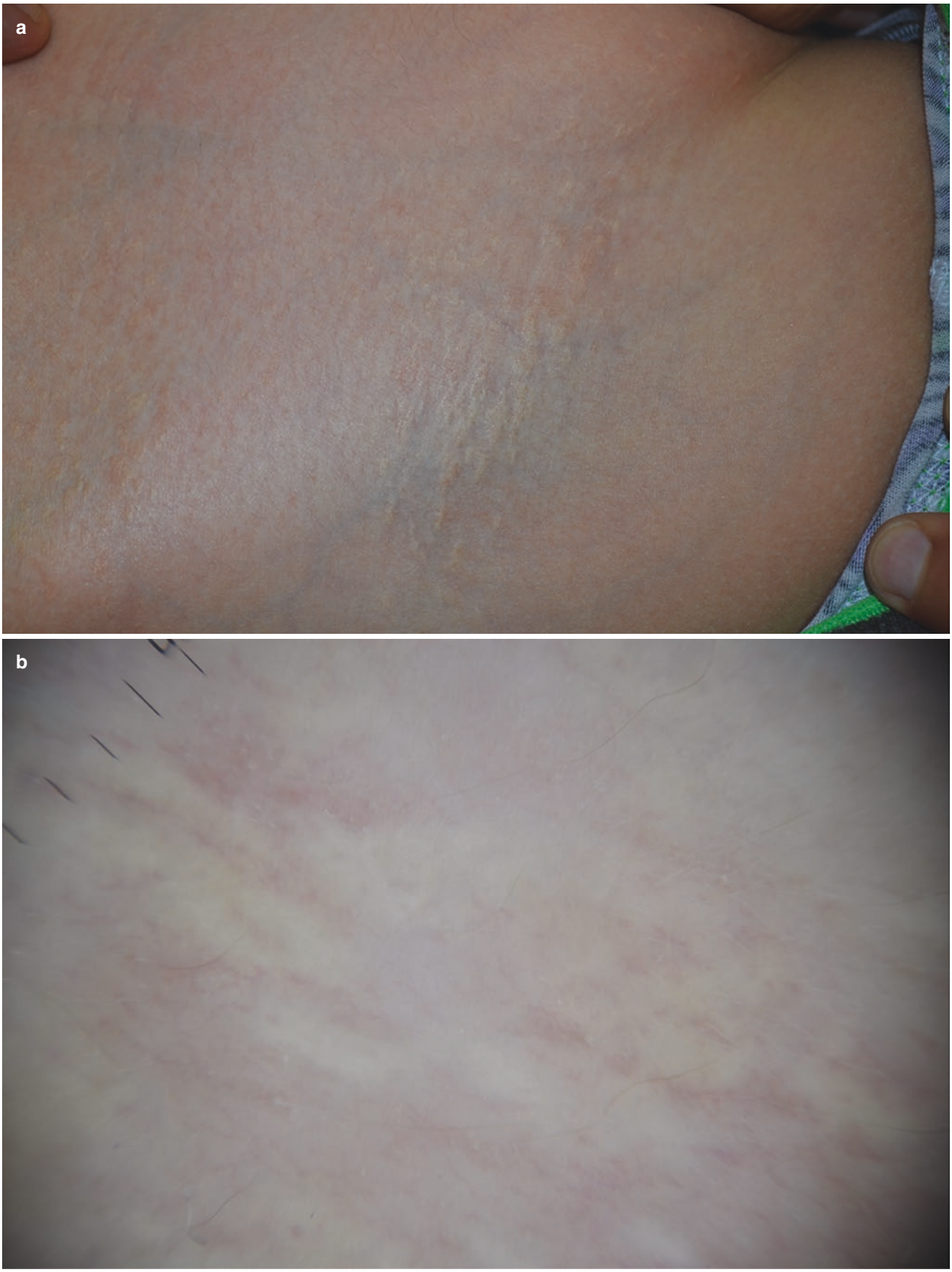

Fig. 26.3 (**a**) Pseudoxanthoma elasticum. Small, yellowish papules of the inguinal fold in the same patient of Fig. 26.2. (**b**) Dermatoscopy (×10): irregular yellowish areas alternating with linear vessels

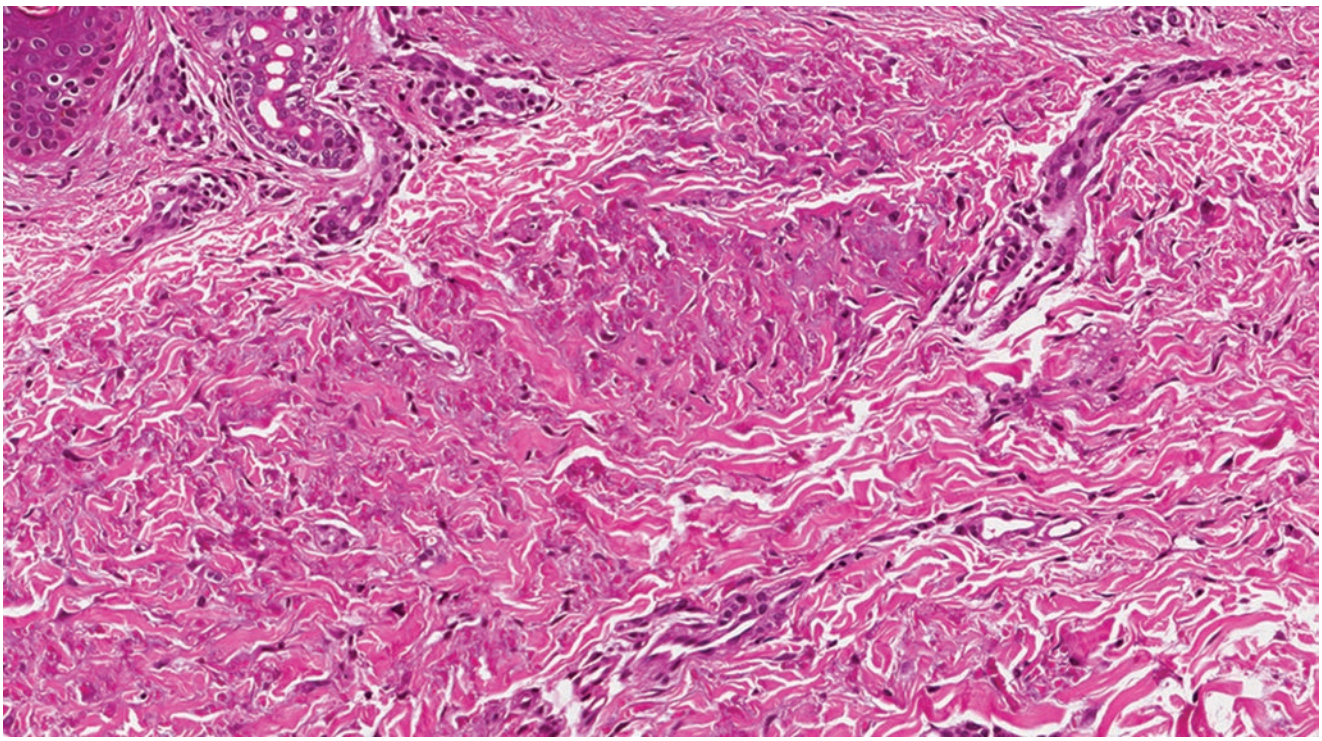

Fig. 26.4 Pseudoxanthoma elasticum. Histopathology: fragmentation and progressive mineralization of elastic fibers of the mid/low dermis (elastorrhexia) [H&E staining; magnification ×200]

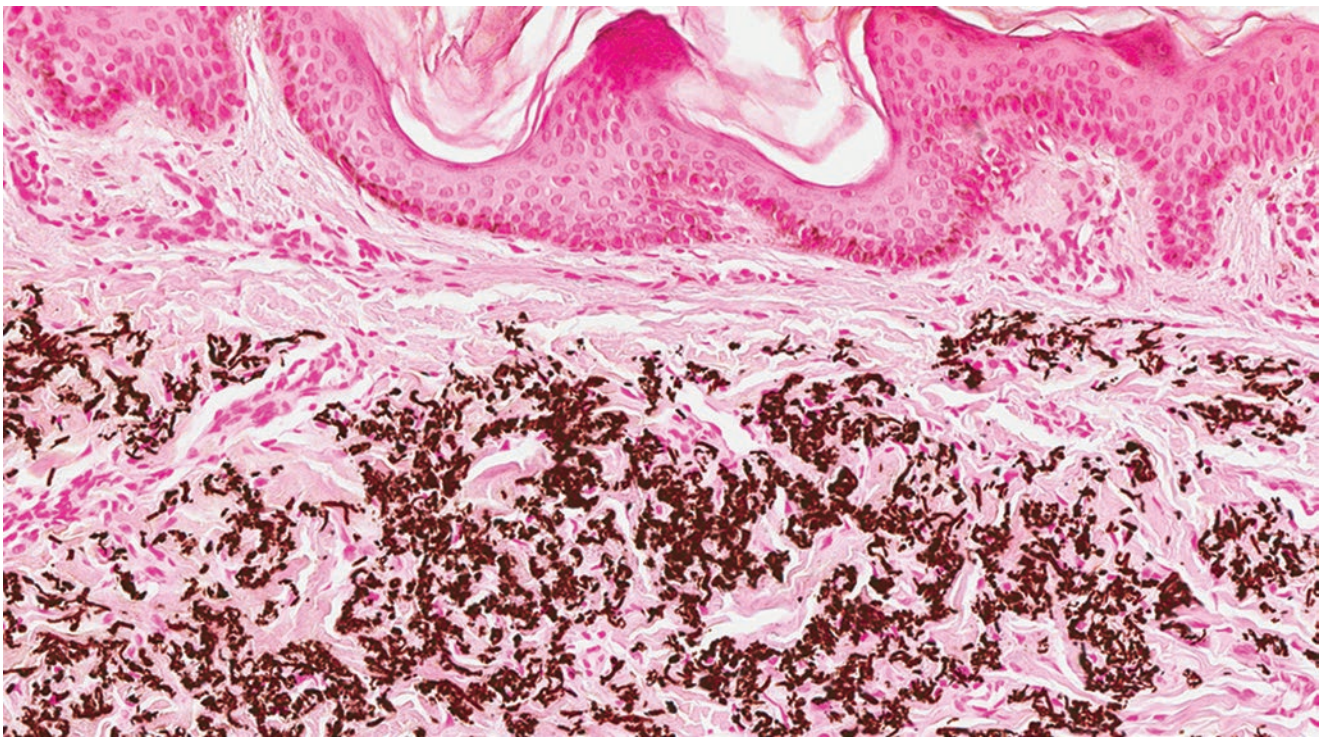

Fig. 26.5 Pseudoxanthoma elasticum. Histopathology: presence of calcium deposits in the dermis [von Kossa staining; magnification, ×150]

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Juvenile Xanthogranuloma

27

Giuseppe Micali, Anna Elisa Verzì, Aurora Tedeschi, and Francesco Lacarrubba

27.1 Definition

Juvenile xanthogranuloma (JXG) is one of the most common benign non-Langerhans cell histiocytoses in the pediatric population.

27.2 Epidemiology

JXG is considered an uncommon disease. In a tumor registry spanning 35 years, JXG accounted for 129 of 24,600 pediatric tumors (0.5%) [1]. The incidence may be underestimated since many cases are diagnosed on clinical examination without histologic confirmation. Lesions occur predominantly in infancy or early childhood, and indeed they appear during the first year of life in 40–70% of cases. Adults may rarely be affected [2].

27.3 Etiology

The exact etiopathogenesis is still unknown. An uncontrolled and disordered macrophage activation may be evoked in response to nonspecific tissue stimuli (infections or physical injuries) [3]. Unlike other xanthomatous lesions, JXG is not associated with dyslipidemias.

27.4 Clinical Features

JXG is characterized clinically by yellow, orange, red, or brown papulonodular lesions which may be single or multiple and vary in size (Figs. 27.1a, 27.2a, 27.3a, and 27.4a). The most common involved sites are the head and neck regions, but the trunk and extremities may also be affected [4]. Lesions may regress completely or may leave a residual atrophic or hyperpigmented scar. Extracutaneous sites, such as the lung, bone, testis, gastrointestinal tract, heart, eye, and oral cavity, may be involved.

27.5 Differential Diagnosis

The clinical differential diagnosis mainly includes Spitz nevi, mastocytomas, Langerhans cell histiocytosis, dermatofibromas, molluscum contagiosum, hemangioma, and neurofibroma.

27.6 Diagnosis

Diagnosis is usually based on clinical examination. Histopathology is infrequently performed because the clinical picture of cutaneous lesions is usually typical and spontaneous regression occurs.

27.7 Dermatoscopy

The typical dermatoscopic finding of JXG is a homogeneous orange-yellowish hue, which has been described as "setting-sun" appearance (Figs. 27.1b, 27.2b, 27.3b and 27.4b) [5–9]. Linear and branched vessels may also be detected. Of note, excessive pressure of the dermatoscope on the skin may limit blood flow to the lesion and hamper observation of the vascular component (Fig. 27.4c). Additional features include

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"clouds" of paler yellow globules, subtle pigment network, and whitish streak.

Dermatoscopic features have been related to the stage of the lesion [7–10]. In early and developed stages, the "setting-sun" appearance is almost always found. In late regressive stage, surrounding erythema decreases, and, as the vacuolated cells transform to more xanthomatized cells, paler yellow globules become more evident.

27.8 Histopathological Correlation

The "setting-sun" appearance seen at dermatoscopy corresponds to the presence of xanthomatous histiocytes (Fig. 27.5), whereas the "clouds" of paler yellow globules correlate with the collection of lipid-laden histiocytes in superficial dermis. In addition, the subtle pigment network and whitish streaks indicate foci of fibrosis.

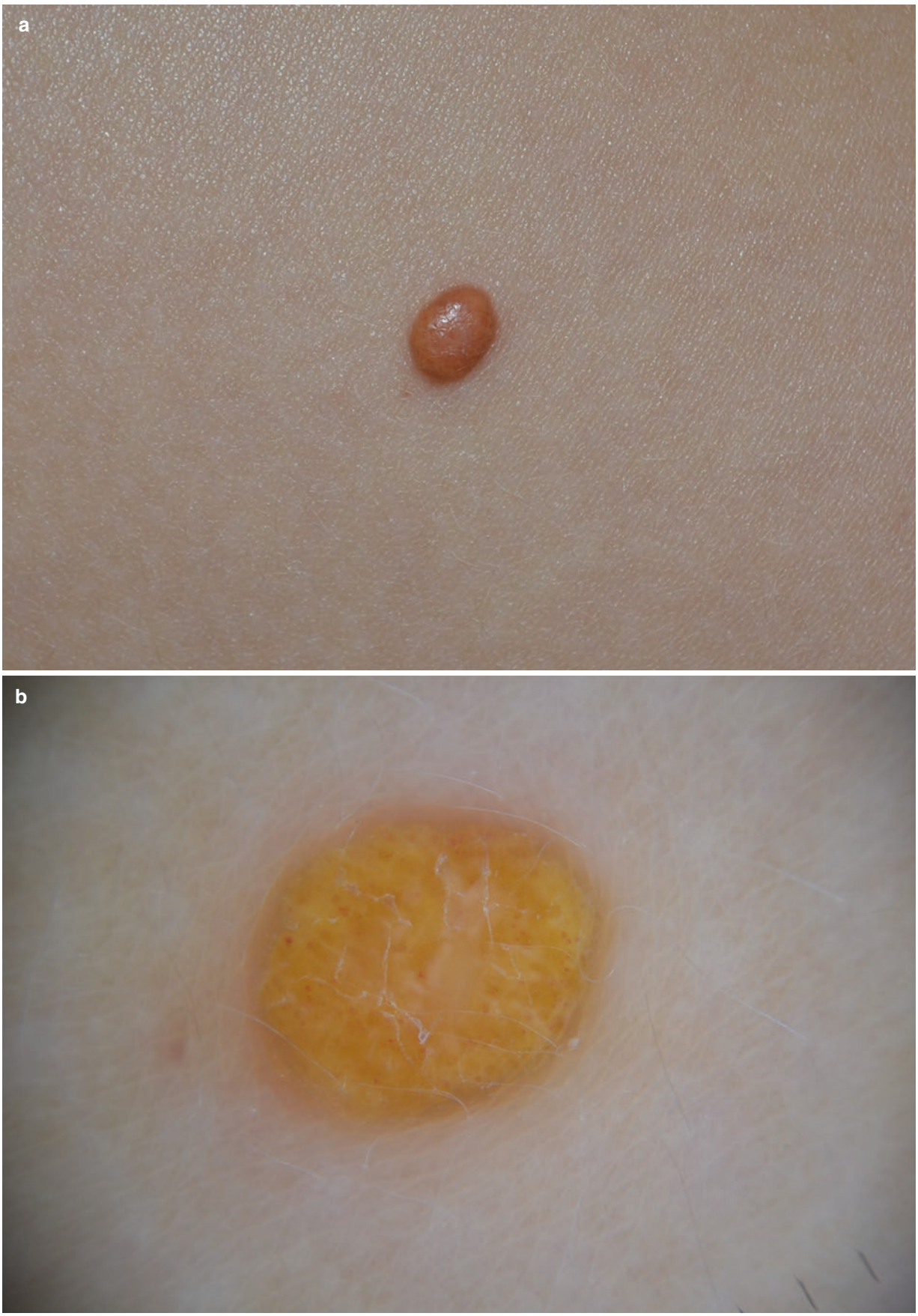

Fig. 27.1 (**a**) Juvenile xanthogranuloma. Orange-yellowish papule with a smooth surface located on the trunk of a 3-year-old boy. (**b**) Dermatoscopy (×10): orange-yellowish background *(setting-sun* appearance) with a few whitish streaks in the center

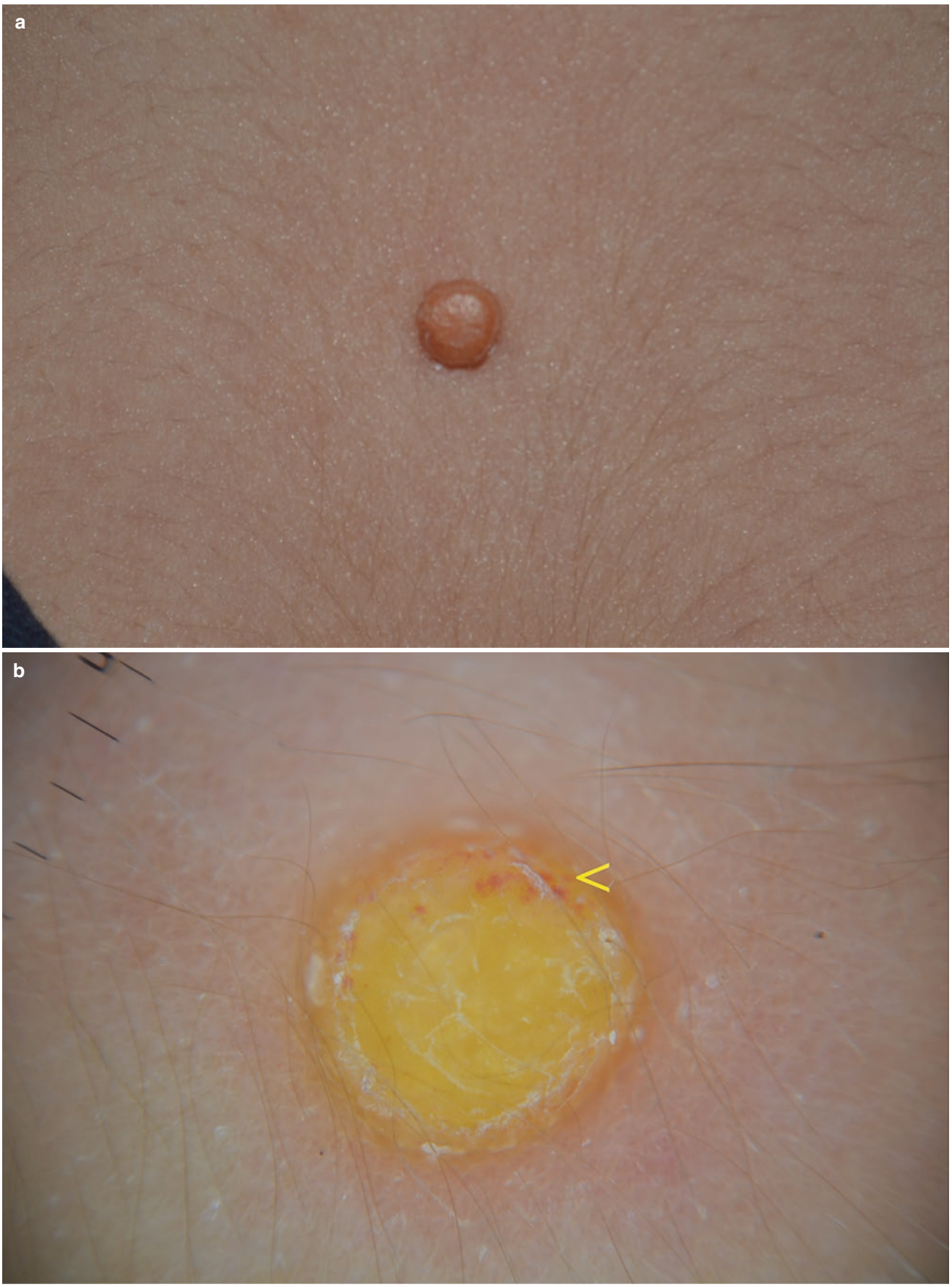

Fig. 27.2 (**a**) Juvenile xanthogranuloma. Orange-yellowish papule located on the abdomen in an 11-year-old boy. (**b**) Dermatoscopy (×10): yellowish background associated to fine desquamation and few dotted vessels at the periphery (arrowhead)

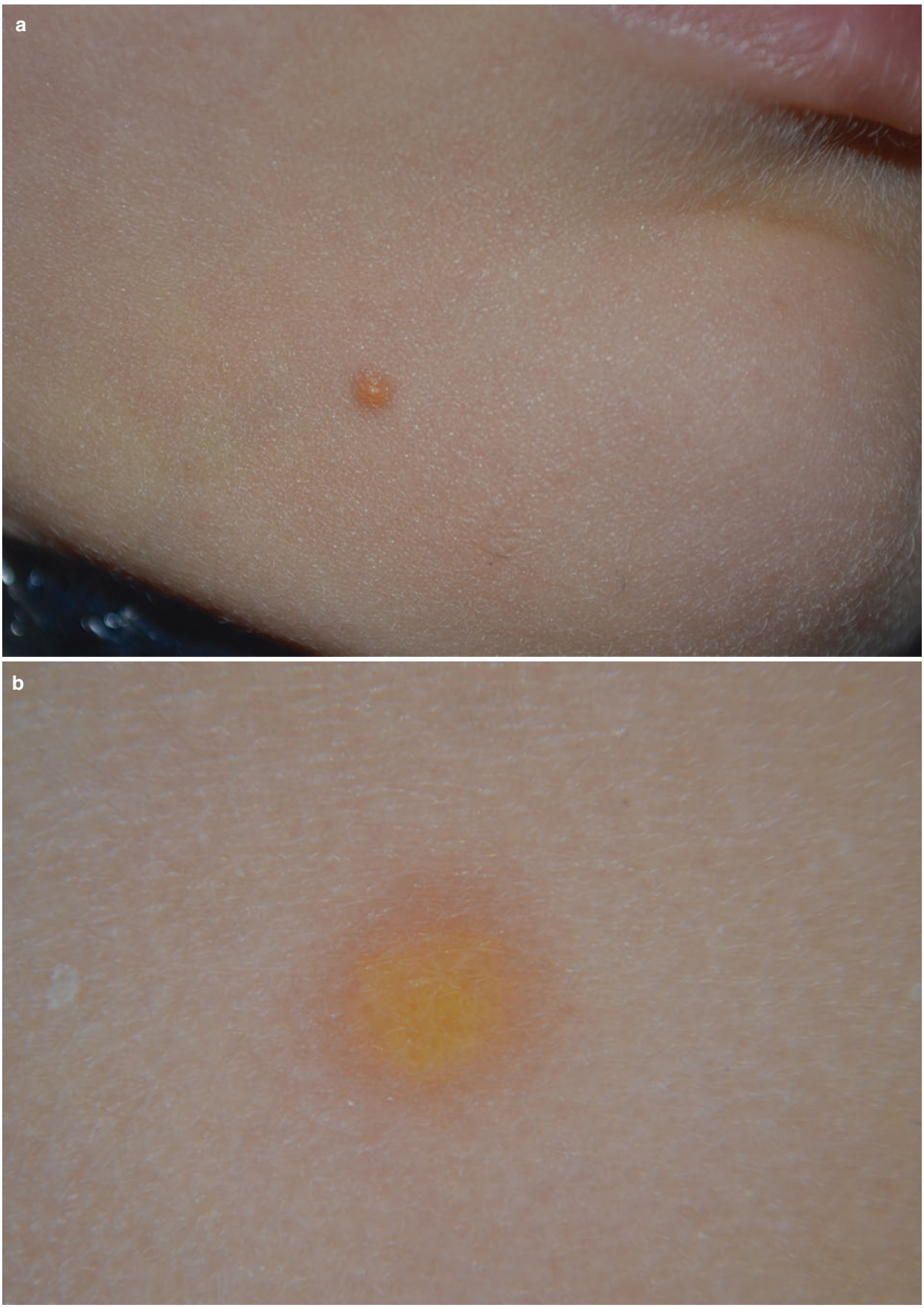

Fig. 27.3 (**a**) Juvenile xanthogranuloma. Small yellowish papule of recent onset located on the chin in an 8-year-old boy. (**b**) Dermatoscopy (×10): yellowish hue surrounded by a faint erythema

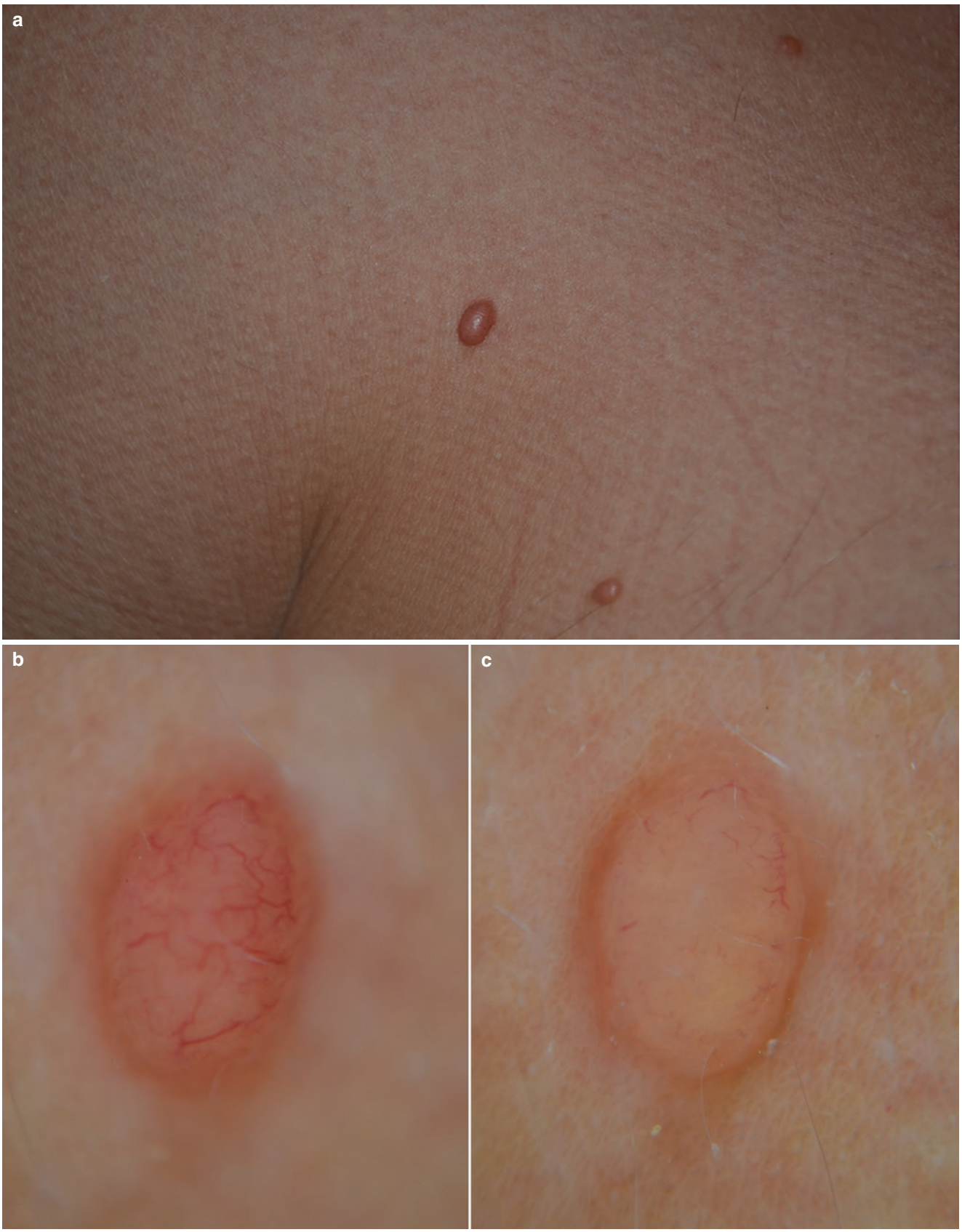

Fig. 27.4 (**a**) Juvenile xanthogranuloma. Two reddish papules of the shoulder in a 6-year-old boy. (**b**–**c**) Dermatoscopy (×10) of the biggest lesion: orange background associated to several telangiectatic vessels. A slight pressure of the dermatoscope on the lesion reduces the vascular component and enhances the visualization of the yellowish background

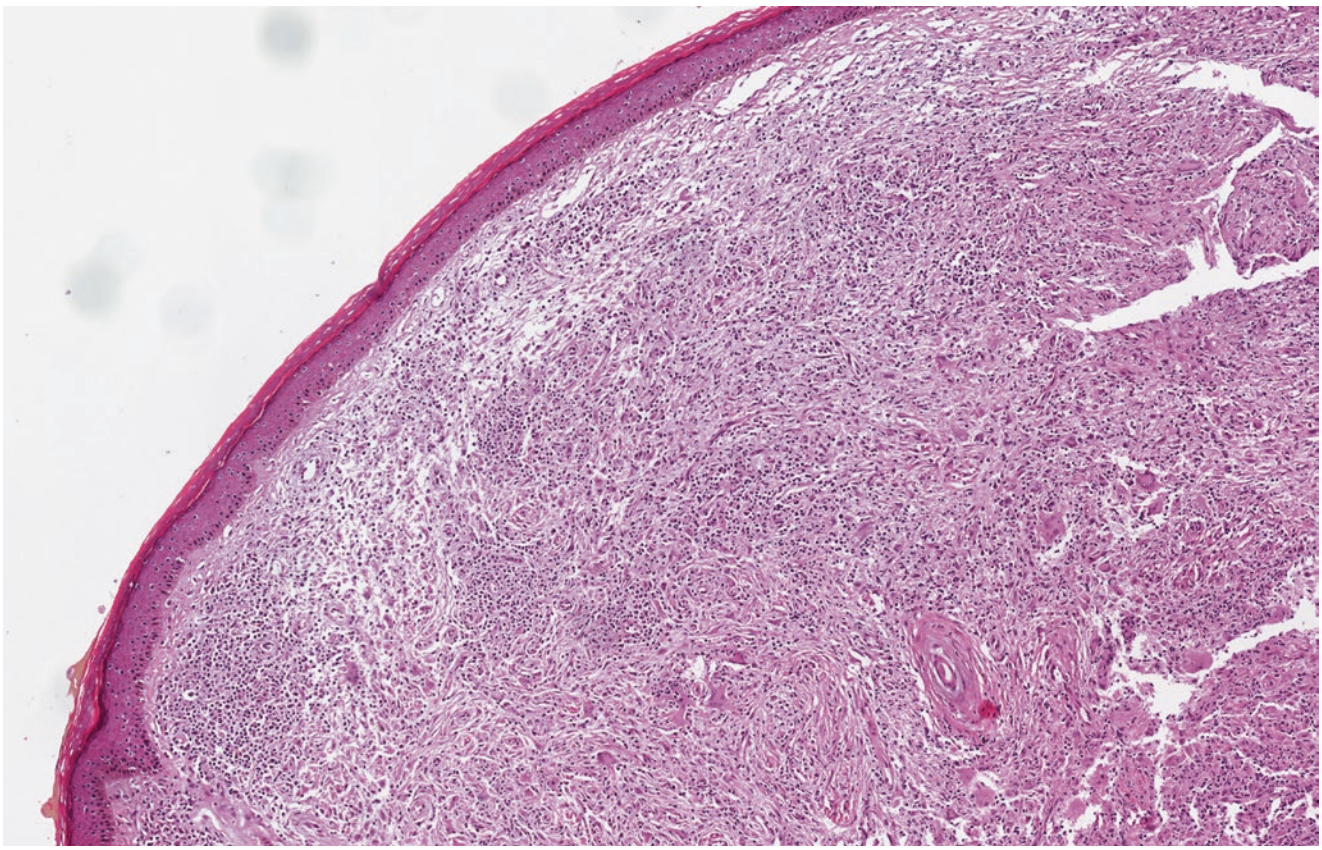

Fig. 27.5 Juvenile xanthogranuloma. Histopathology: presence of xanthomatous histiocytes in the dermis [H&E staining; magnification, ×100]

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Verrucous Epidermal Nevus

28

Enzo Errichetti and Giuseppe Stinco

28.1 Definition

Epidermal nevus is a general term encompassing a variety of hamartomatous ectodermal lesions characterized by epidermal changes either alone (keratinocytic nevi or "true" epidermal nevi) or in combination with overgrowth of skin appendages (organoid nevi) [1, 2]. Verrucous epidermal nevus (VEN) represents the most frequent variant of "true" epidermal nevi [2].

28.2 Epidemiology

Epidemiological data suggest that VEN occurs in about 1 in 1000 live births, with the majority of lesions appearing within the first year of life or by age 14 years, albeit cases with adult onset have been reported [2]. There is an equal male–female prevalence, and most cases are sporadic [2].

28.3 Etiology

VEN is thought to be the result of somatic mosaicism due to postzygotic mutations during embryogenesis, which have not yet been completely identified [1]. Recent reports pointed to a relation between such a condition and mutations in PIK3CA, KRAS, and FGFR3 genes [3, 4].

28.4 Clinical Features

VEN presents as localized or diffuse, closely set, skin-colored, brown, or gray-brown verrucous papules, often coalescing to form well-demarcated papillomatous plaques (Figs. 28.1a and 28.2a), which commonly have the appearance of being "stuck on" to the skin surface and display a linear arrangement according to either Blaschko's lines or tension lines (Fig. 28.3a) [1, 2]; they tend not to cross the midline [1, 2]. Initially, such lesions may be subtle and increase in extent/thickness as the child grows [1, 2]. Even though VEN is usually an isolated benign lesion, in some cases, particularly in extensive lesions (known as systemized epidermal nevi), it may be associated with abnormalities affecting other organs (epidermal nevus syndrome) [1, 2].

28.5 Differential Diagnosis

The main differential diagnoses of VEN include sebaceous nevus, congenital melanocytic nevi, acquired dermal melanocytic nevi, nevoid acanthosis nigricans, and verrucous stage of incontinentia pigmenti [2].

28.6 Diagnosis

The diagnosis of VEN is typically based on its distinctive distribution/morphological features, though differentiation from similar conditions, especially from other epidermal nevi, may sometimes be difficult on clinical ground only [1, 2]. Dermatoscopy has recently been shown to be a useful technique in the recognition of VEN [5].

28.7 Dermatoscopy

Dermatoscopic examination of VEN frequently shows brown circles, which consist of oval or round structures characterized by a hyperchromic brown edge surrounding a

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hypochromic area (Figs. 28.1b and 28.2b). In most cases, they occur adjacent to one another, are of variable dimensions, and may be localized either at the periphery or in the middle of the lesion [5]. In our experience, such findings are more commonly seen in less thick areas of the lesion (Fig. 28.1b), whereas they are observed more rarely in thick plaques (Fig. 28.2b). Other possible findings include features frequently visible in seborrheic keratosis and/or dermal melanocytic nevi, such as fissures, keratin-filled crypts, comedo-like openings, milia-like cysts, cerebriform appearance, whitish scaling, brown globules, brown-grayish dots (more common in traumatized VEN), papillomatous surface, and cobblestone appearance (Figs. 28.1b, 28.2b, and 28.3b) [5, 6].

28.8 Histopathological Correlation

Typically, the histological examination of VEN reveals hyperkeratosis, papillomatosis, acanthosis, hyperpigmentation of the basal cell layer, and elongation of the rete ridges (Fig. 28.4) [7]. The dermatoscopic feature of large brown circles is likely to correspond histologically to a peculiar disposition of pigmented keratinocytes surrounding the dermal papillae [5], while the association of diffuse basal cell hyperpigmentation with more or less marked acanthosis/papillomatosis may be related to the dermatoscopic detection of brownish globules and cobblestone-like appearance [7]. On the other hand, fissures, crypts, comedo-like openings, and cerebriform appearance observed on dermatoscopy are due to papillomatosis and epidermal invaginations filled or not with keratin (Fig. 28.4) [8]. Finally, milia-like dermatoscopic structures are the result of intraepidermal keratin-filled cysts [8].

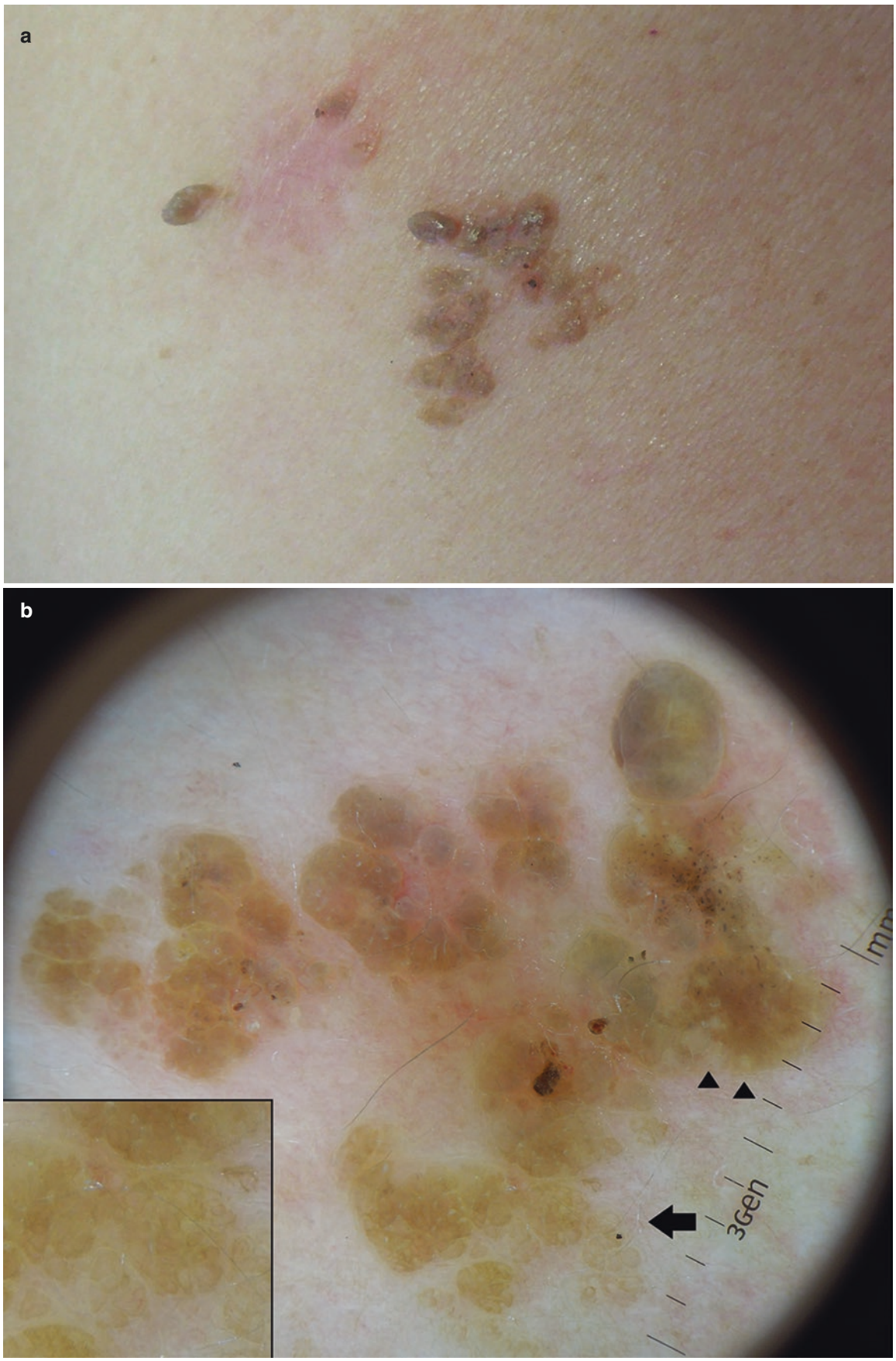

Fig. 28.1 (**a**) Verrucous epidermal nevus. Brownish papules coalescing into papillomatous plaques on the back in a 14-year-old girl. (**b**) Dermatoscopy (×10): brownish globules, brown-grayish dots, milia-like cysts (arrowheads), and brown circles (arrow and box)

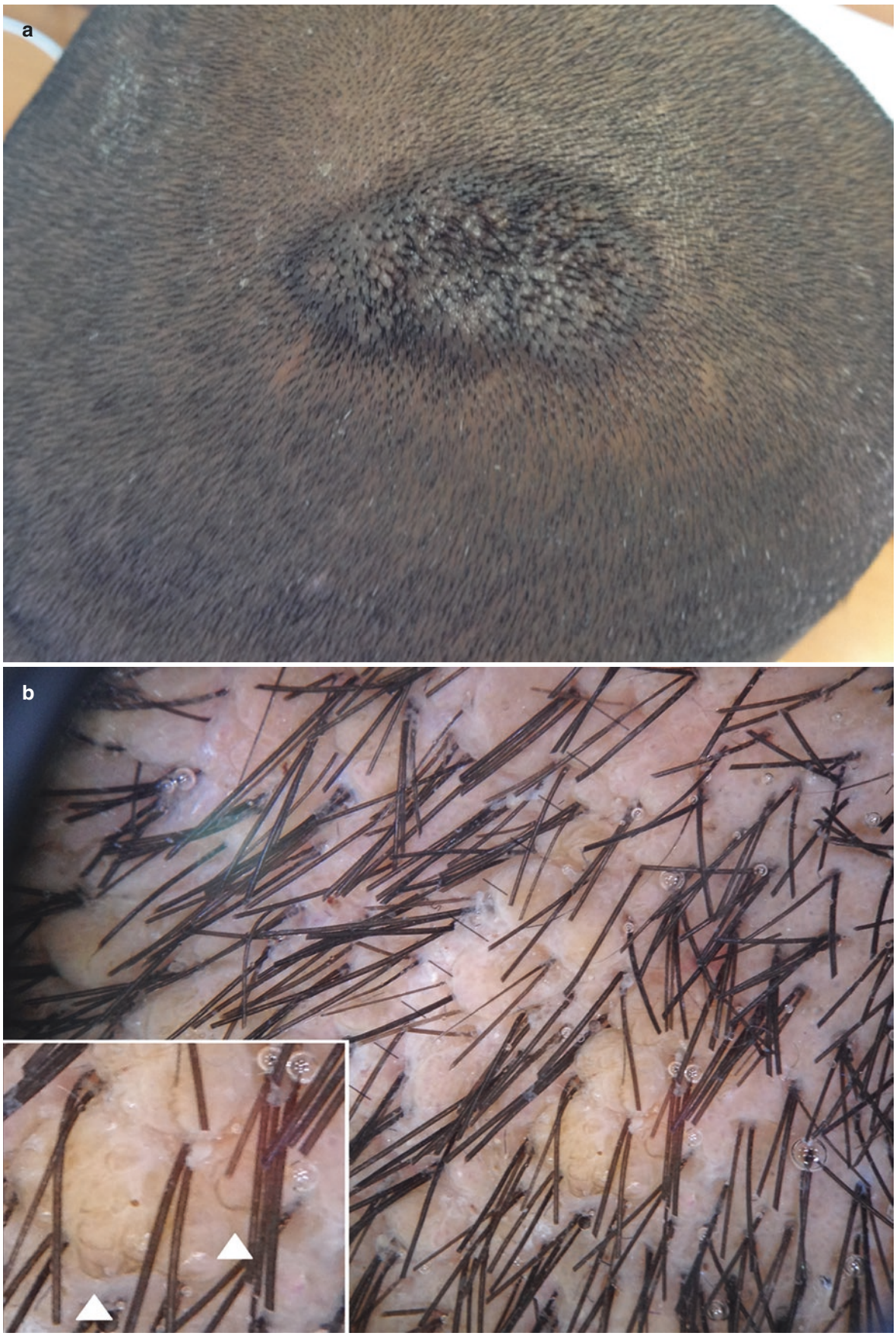

Fig. 28.2 (**a**) Verrucous epidermal nevus. Brownish plaque on the scalp in a 13-year-old dark-skinned boy. (**b**) Dermatoscopy (×10): brownish papillomatous surface with a few brown circles (arrowheads in the box)

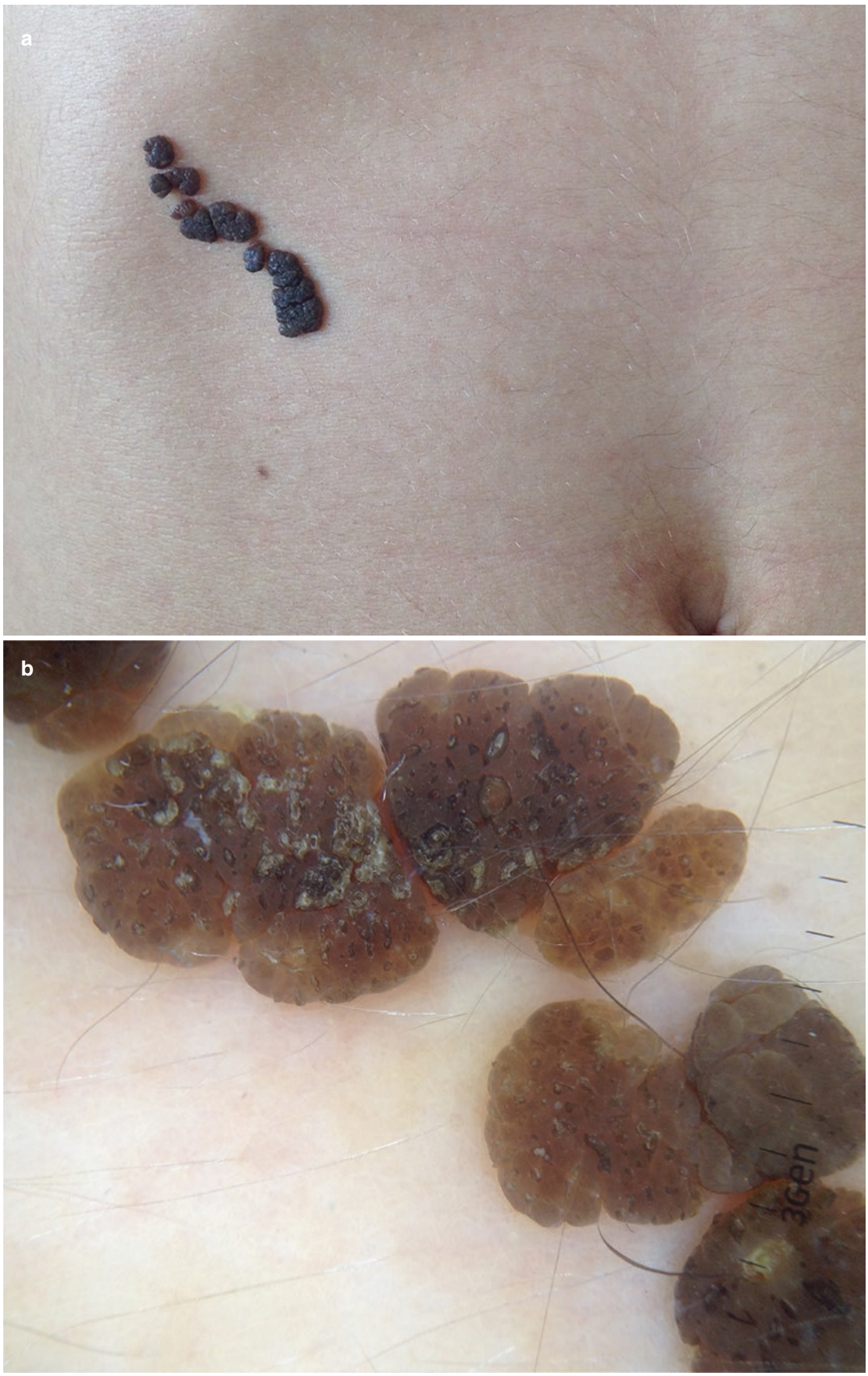

Fig. 28.3 (**a**) Verrucous epidermal nevus. Several brown papules of the trunk coalescing into a linear papillomatous plaque with a *stuck-on* appearance in a 10-year-old boy. (**b**) Dermatoscopy (×20): papillomatous surface, brownish globules, keratin-filled crypts, comedo-like openings, and whitish scaling

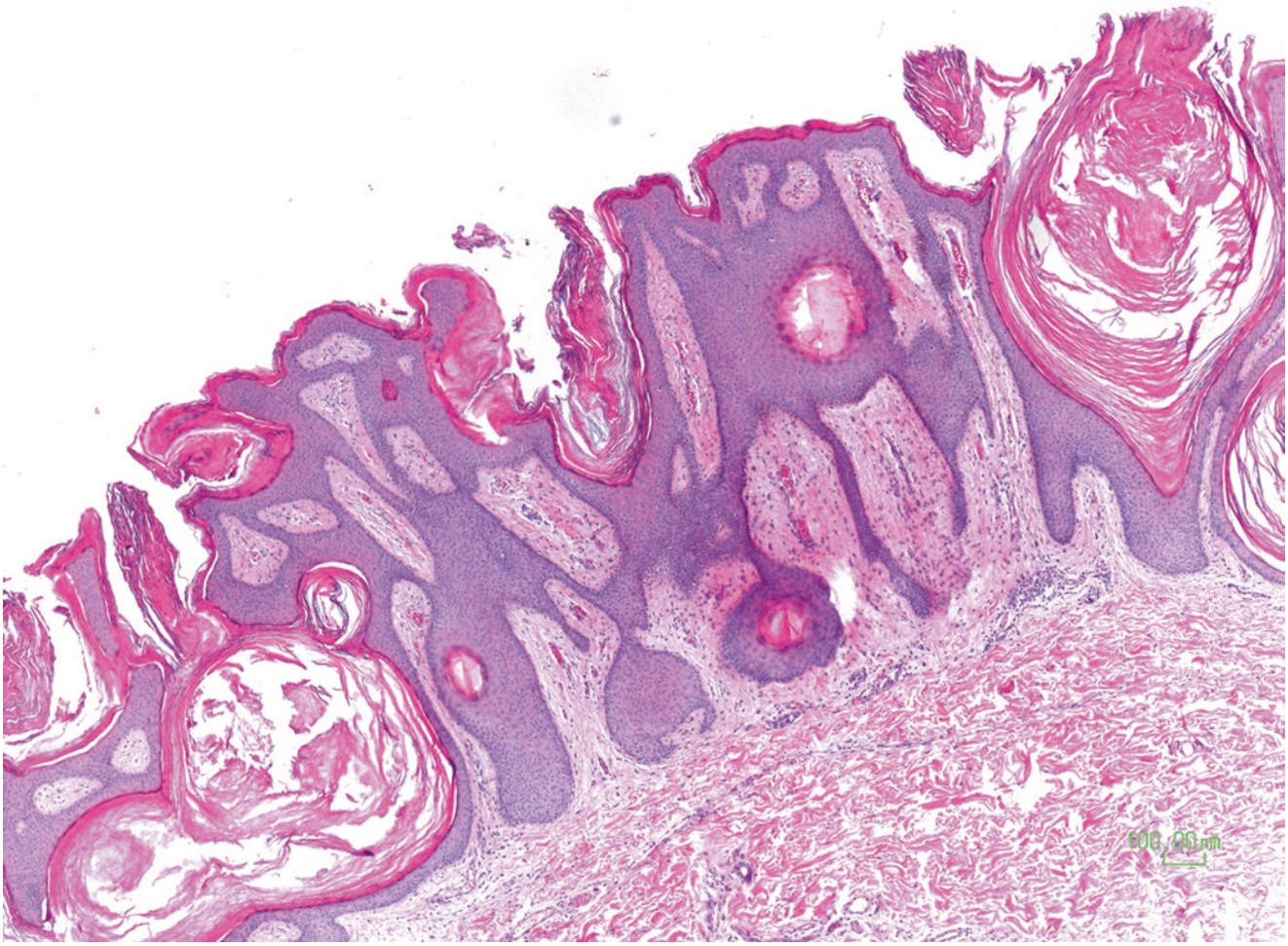

Fig. 28.4 Verrucous epidermal nevus. Histopathology: hyperkeratosis, papillomatosis, acanthosis, hyperpigmentation of the basal cell layer, and elongation of the rete ridges [H&E staining, magnification ×100; courtesy of Carla Di Loreto, MD, Institute of Anatomic Pathology, University of Udine, Italy]

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Sebaceous Nevus

29

Iria Neri

29.1 Definition

Sebaceous nevus (SN) is a congenital hamartoma occurring on the scalp or face, that combines a variety of epidermal, follicular, sebaceous, and apocrine gland abnormalities.

29.2 Epidemiology

SN usually presents at birth but may become evident later in life. SN can be found in about 0.3% of all neonates [1, 2] and has a prevalence of 0.8% among all dermatological patients.

29.3 Etiology

The etiology of SN is unknown. Recent studies show that postzygotic mutations in HRAS and KRAS genes may be related to both isolated SN and SN syndrome [3].

29.4 Clinical Features

SN typically evolves in successive clinical stages. At birth, the SN of the scalp presents as a congenital hairless area with a smooth, flesh-colored surface with a pink or orange cast (Figs. 29.1a and 29.2a). At puberty, it develops a velvety and verrucous surface as a consequence of hormonal stimulation (Figs. 29.3a and 29.4a). In 10–20% of SN, both benign and malignant superimposed neoplasm can occur, including syringocystadenoma papilliferum, apocrine cystadenoma, trichoblastoma, and basal cell carcinoma. Other malignant tumors, such as squamous cell carcinoma or adnexal carcinoma, are uncommon [4, 5].

The linear and extensive form of SN may be associated with ocular, bone, and neurological involvement in epidermal nevus syndrome or Schimmelpenning syndrome (OMIM 163200) defined by the association of SN with cerebral, ocular, or skeletal defects (1a). Other syndromes, such as didymosis aplasticosebacea, and SCALP (sebaceous nevus, central nervous system malformations, aplasia cutis congenita, limbal dermoid, and pigmented nevus) syndrome are very rare [6].

29.5 Differential Diagnosis

The differential diagnosis should be made with epidermal nevus and aplasia cutis congenita. Moreover, balloon cell nevus, a rare variety of benign melanocytic nevus, may simulate a SN for the presence of a yellowish hue [7].

29.6 Diagnosis

In newborns and infants, the diagnosis of SN is usually clinical. The dermatoscopic examination becomes important once the SN has fully developed, in order to identify possibly arising tumors within the original lesion. When a secondary neoplasm is suspected, a biopsy for histopathological examination is mandatory. Rarely, a verrucous proliferation within a SN can resemble a filiform wart on histology; in such cases, detection of HPV-DNA by PCR is diagnostic.

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29.7 Dermatoscopy

Dermatoscopy of a congenital area of alopecia on the scalp may be useful in differentiating early SN from aplasia cutis congenita in newborns, showing bright-yellow dots not associated with the hair follicles (Figs. 29.1b and 29.2b). Examination of a velvety plaque shows round to oval yellowish/orange structures of different size, sometimes gathered in clusters and/or associated with a whitish network (Fig. 29.3b). In the late stage, dermatoscopy of SN shows fissures and ridges arranged in a cerebriform pattern and/or verrucous proliferations (Fig. 29.4b). It may reveal a useful diagnostic tool for a more accurate preoperative diagnosis of neoplasms arising in SN [8–10]. The presence of round to oval yellowish structures may be also observed in balloon cell nevus [7]; however, in this case the concomitant observation of a brownish reticular or globular pattern suggests the correct diagnosis (Fig. 29.5).

29.8 Histopathological Correlation

No histopathological correlation is reported so far. The bright-yellow dots observed by dermatoscopy can be related to milia-like structures rather than sebaceous gland hyperplasia, because of the immaturity of the glands in infants. White dots and comedo-like openings, corresponding to sebaceous gland hyperplasia, appear later in life, in concomitance with the hormonal stimulation (Fig. 29.6) [9].

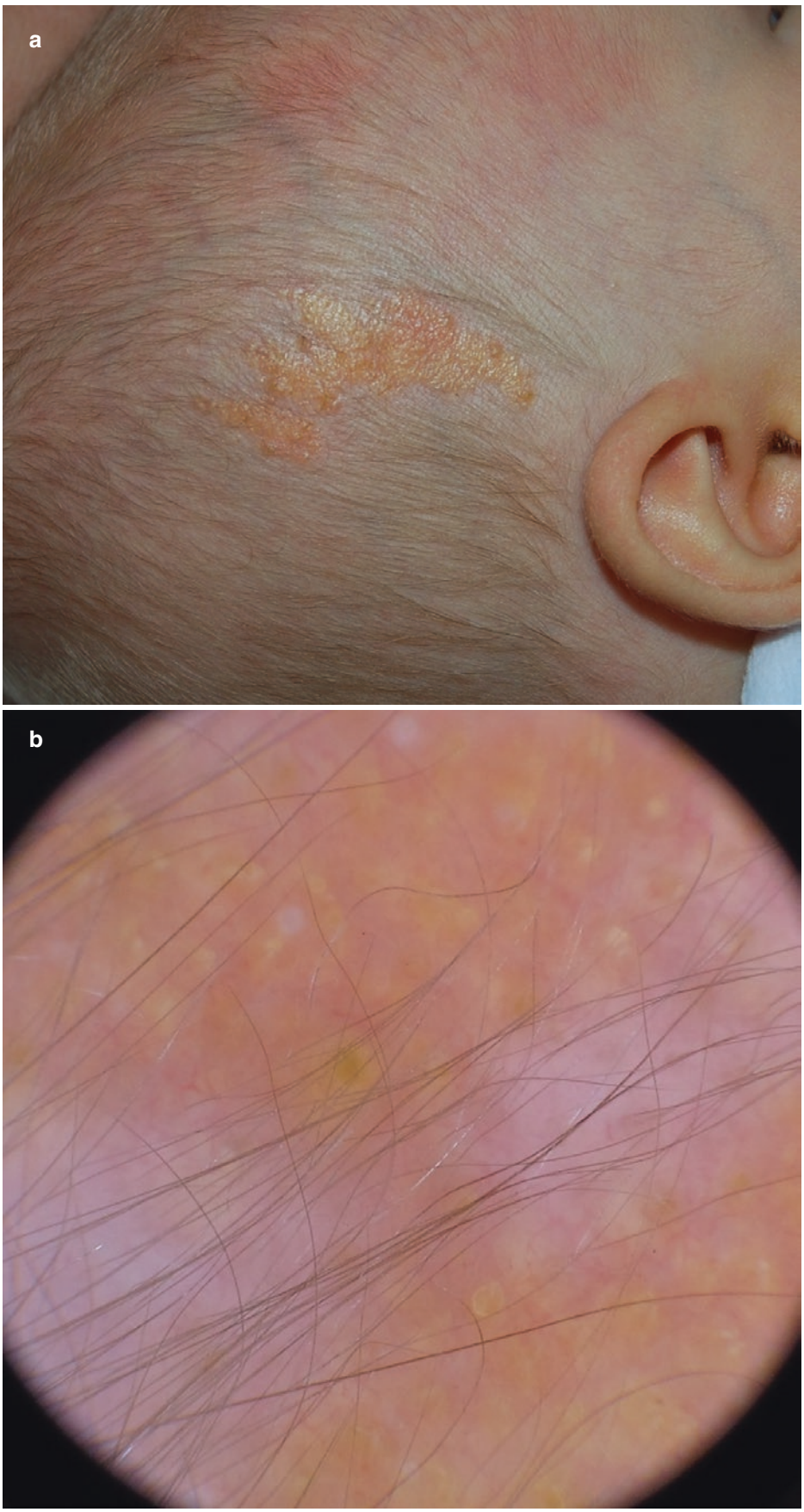

Fig. 29.1 (**a**) Sebaceous nevus. Congenital hairless patch of the scalp with a smooth, pink to orange surface. (**b**) Dermatoscopy (×10): multiple yellow and few whitish round structures on an orange background

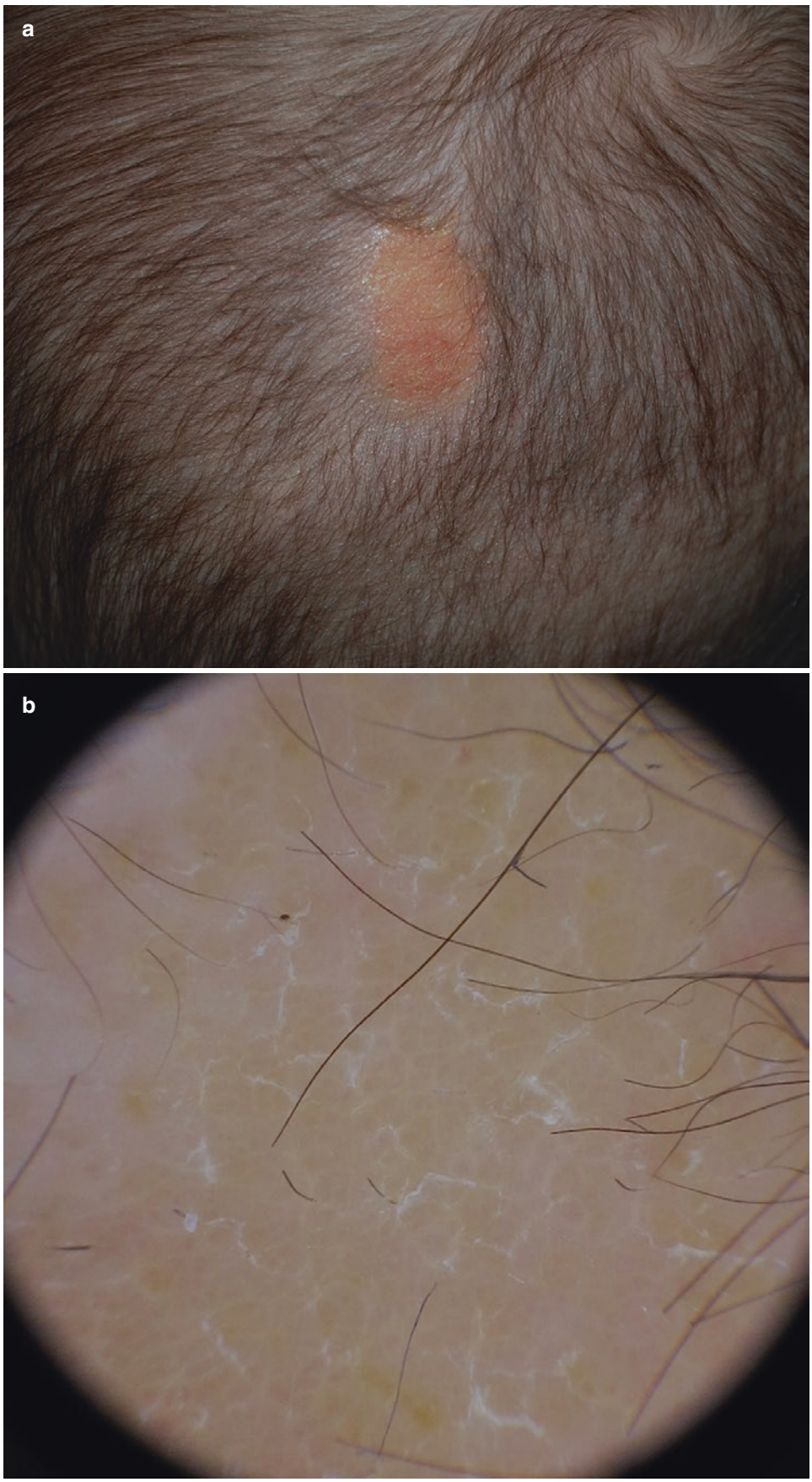

Fig. 29.2 (**a**) Sebaceous nevus. A 2-month-old boy with a congenital pink-colored, non-specific smooth area of alopecia, located close to the vertex. (**b**) Dermatoscopy (×10): structureless brownish orange polygonal areas associated with a whitish network

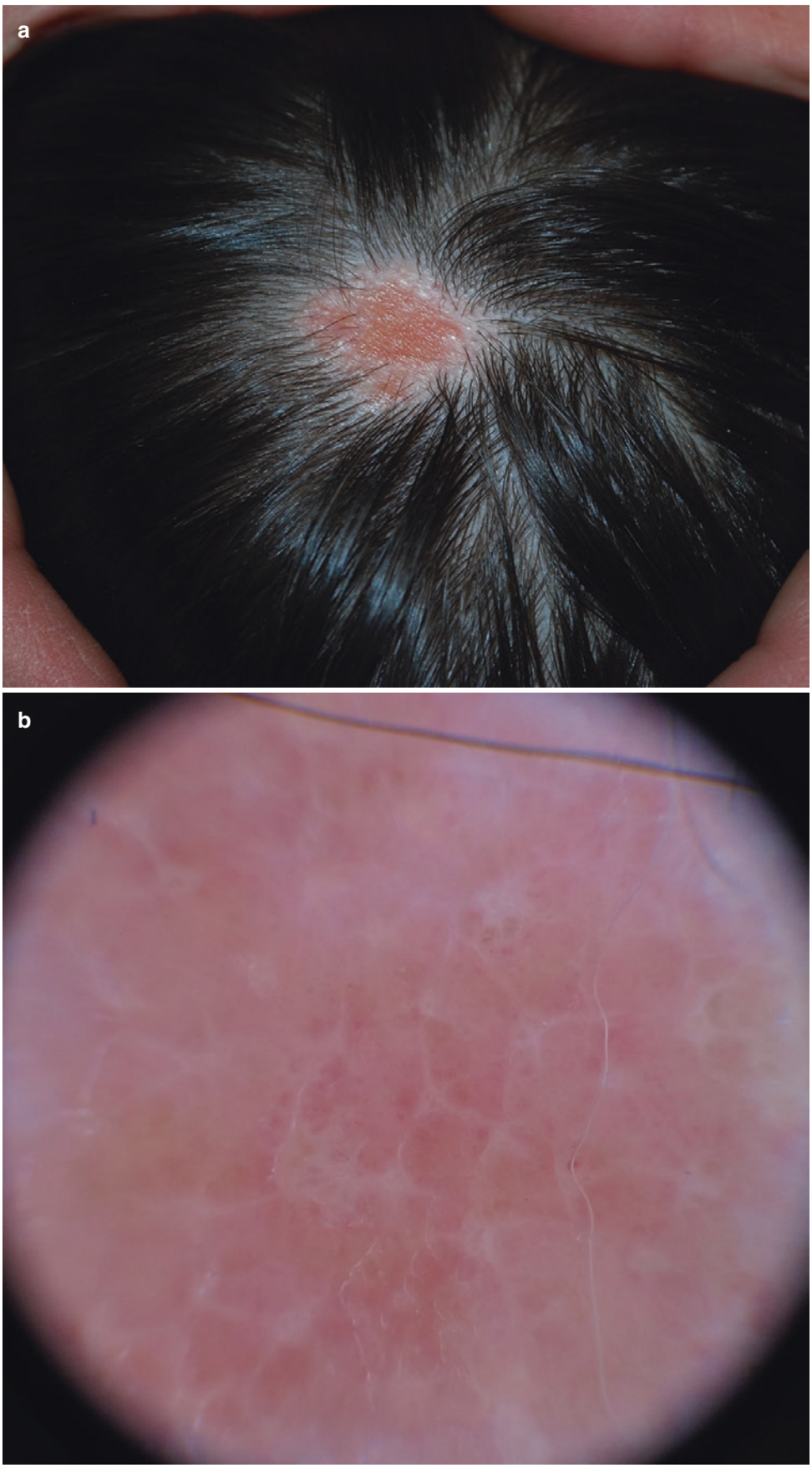

Fig. 29.3 (**a**) Sebaceous nevus. Flat lesion of the scalp with a velvety surface. (**b**) Dermatoscopy (×10): pink orange structures associated with a whitish network

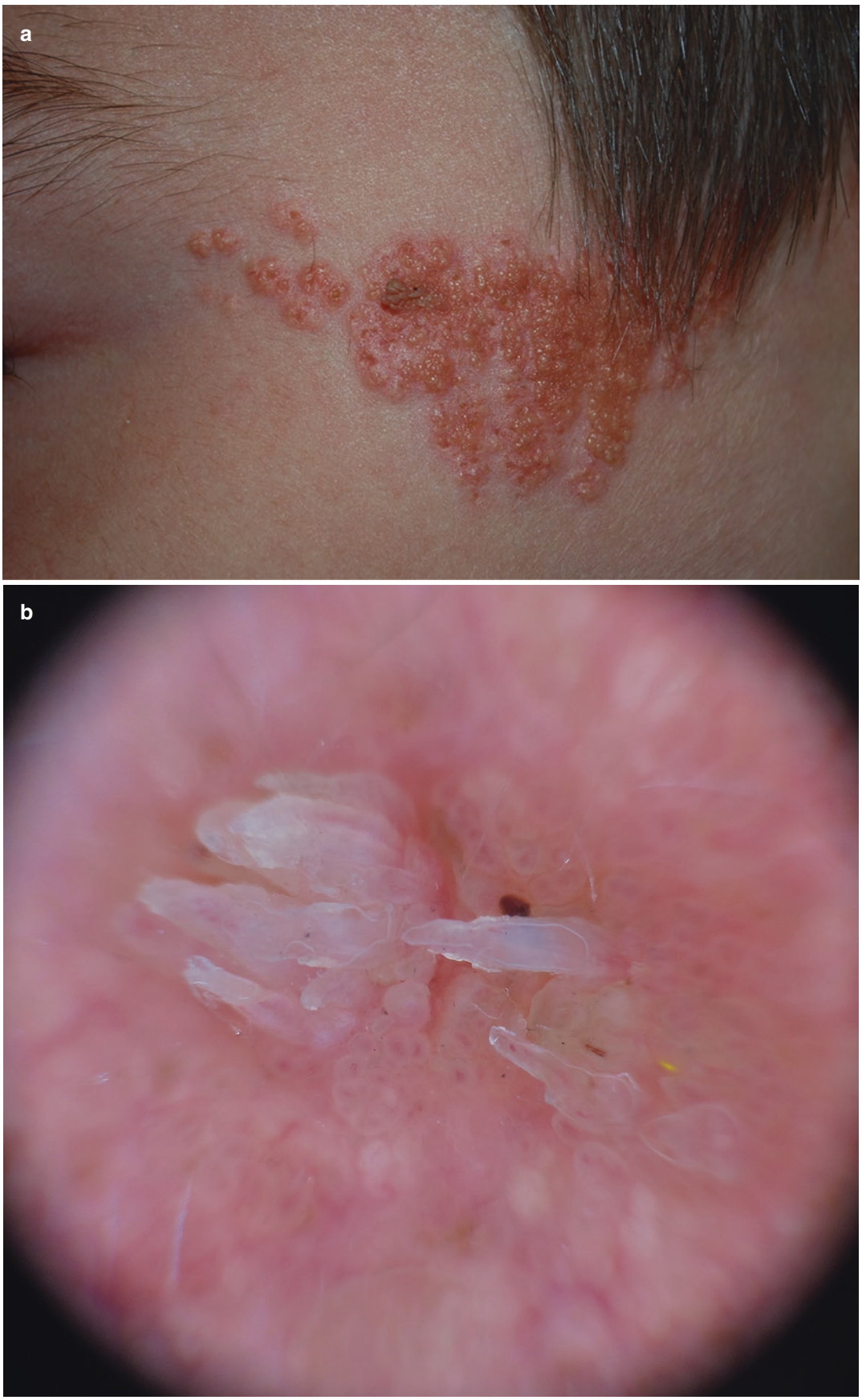

Fig. 29.4 (**a**) Sebaceous nevus. Yellowish, verrucous lesion of the face in an 8-year-old child. (**b**) Dermatoscopy (×10): whitish exophytic papillary structures surrounded by hairpin vessels with a white halo. The final diagnosis, after punch biopsy and PCR DNA, was verrucous proliferation of sebaceous nevus

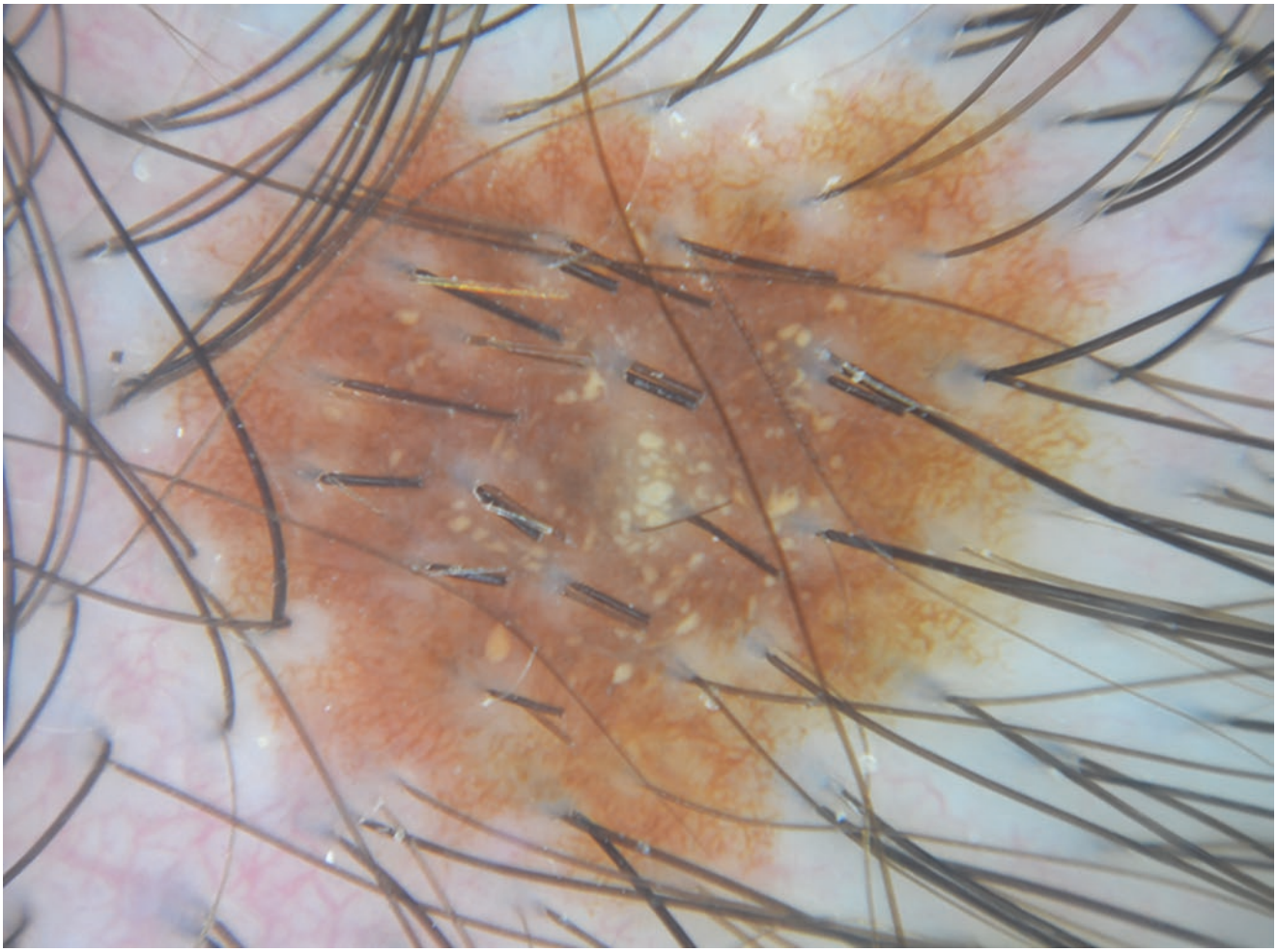

Fig. 29.5 Balloon cell nevus of the scalp in a 11-year-old girl. Brownish reticular pattern associated with roundish yellow-colored structures (Courtesy of Giuseppe Micali, MD)

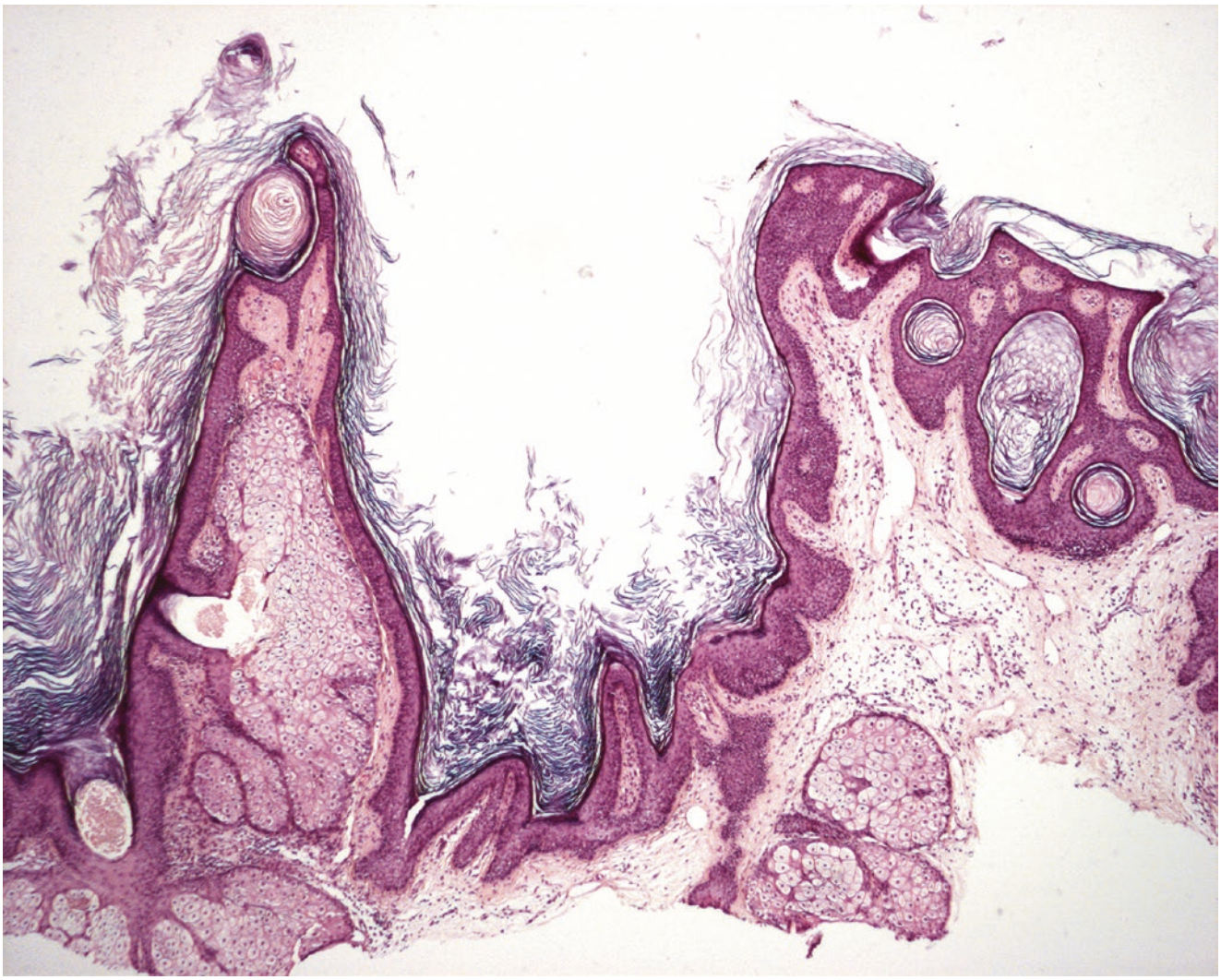

Fig. 29.6 Sebaceous nevus. Histopathology: epithelial hyperplasia, papillomatosis, and large sebaceous glands [H&E staining; magnification, ×40]

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Basal Cell Nevus Syndrome

30

Iria Neri

30.1 Definition

The basal cell nevus syndrome (BCCS), also known as Gorlin-Goltz syndrome, is an autosomal-dominant disease with complete penetrance and variable expressivity causing the development of multiple basal cell carcinomas (BCCs) in childhood, together with other multiorgan abnormalities (OMIM 109400) [1].

30.2 Epidemiology

The reported prevalence of NBCCS ranges between 1 and 9/100,000, with a male-to-female ratio of 1:1.

30.3 Etiology

BCCS is determined by mutations in the genes PTCH1, PTCH2, and SUFU related to hedgehog signaling pathway. PTCH1 encodes a transmembrane receptor acting as a tumor suppressor. Its mutation determines a carcinogenesis dysregulation [2].

30.4 Clinical Features

Multiple BCCs usually manifest early in life, up to several hundreds of elements. BCCs appear as 1- to 10-mm hyperpigmented or skin-colored, dome-shaped papules, soft nodules, or flat plaques (Fig. 30.1a) [3]. In children acrochordon-like BCCs have been described as a polypoid and exophytic variant of BCC in BCCS (Fig. 30.2a) [4]. Recently, early onset acral BCCs have been reported, presenting as multiple pearly papules of a few millimeters in diameter, located on the dorsum and lateral aspects of the hands and feet (Fig. 30.3a) [5].

BCCs may be associated with odontogenic jaw cysts, bifid ribs, intracranial calcifications, and palmoplantar pits (Fig. 30.4a). Other features include hypertelorism, broad nasal root, and macrocephaly. Mental retardation, seizures, deafness, or medulloblastoma can occur.

30.5 Differential Diagnosis

The BBCs observed in BCCS may resemble melanocytic nevi, acrochordons, and molluscum contagiosum.

30.6 Diagnosis

The diagnosis of BCCS is made clinically (concomitance of two major criteria or one major and two minor criteria). Major criteria include multiple BCCs (>2 or 1 under 20 years), odontogenic jaw cysts, ≥3 palmoplantar pits, lamellar calcifications in the cerebral falx, medulloblastoma typically desmoplastic, and a first-degree relative affected by BCCS. Minor criteria include macrocephaly, ocular abnormalities, cardiac/ovarian fibroma, cleft lip/palate, rib/vertebral abnormalities, pleural/lymphomesenteric cysts, and polydactyly. A genetic test can be performed [2].

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30.7 Dermatoscopy

As regards BCCs, dermatoscopy of pigmented lesions reveals leaf-like and spoke-wheel areas (superficial BCC) or blue-gray globules and large ovoid nests (nodular-cystic BCC). Ulceration or small erosions may be present [3, 4]. In nonpigmented BCCs, the main feature is represented by the presence of branching arborizing (tree-like) vessels (Fig. 30.1b). Blue-gray globules can be observed in small acrochordon-like lesions (Fig. 30.2b), whereas larger lesions present small blue-gray ovoid nests and/or telangiectases over their surface [4]. A pinpoint papular variant has been even reported, mainly localized on the extremities (Fig. 30.3b) [5].

Dermatoscopy is also useful to detect palmar pits (Fig. 30.4b) [6, 7], showing flesh-colored depressed lesions with both blue structure and arborizing vessels or red-dotted vessels (more frequent in adults). These diagnostic elements are not detected in other rare conditions showing palmar pits, such as Darier's disease and generalized follicular hamartoma.

30.8 Histopathological Correlation

In BCCs, blue-gray globules, blue-gray ovoid nests, leaf-like structures, and spoke-wheel areas correspond to large aggregates of melanin pigment and melanophages in the stroma of the basaloid cells [3].

Histopathological examination of pits is characterized by a sharply circumscribed zone of hypokeratosis, hypogranulosis, parakeratosis, and basal cell hyperplasia with crowding and palisading of basal keratinocytes. In some cases, buds of basaloid cells may be present under the zone of hypokeratosis [4].

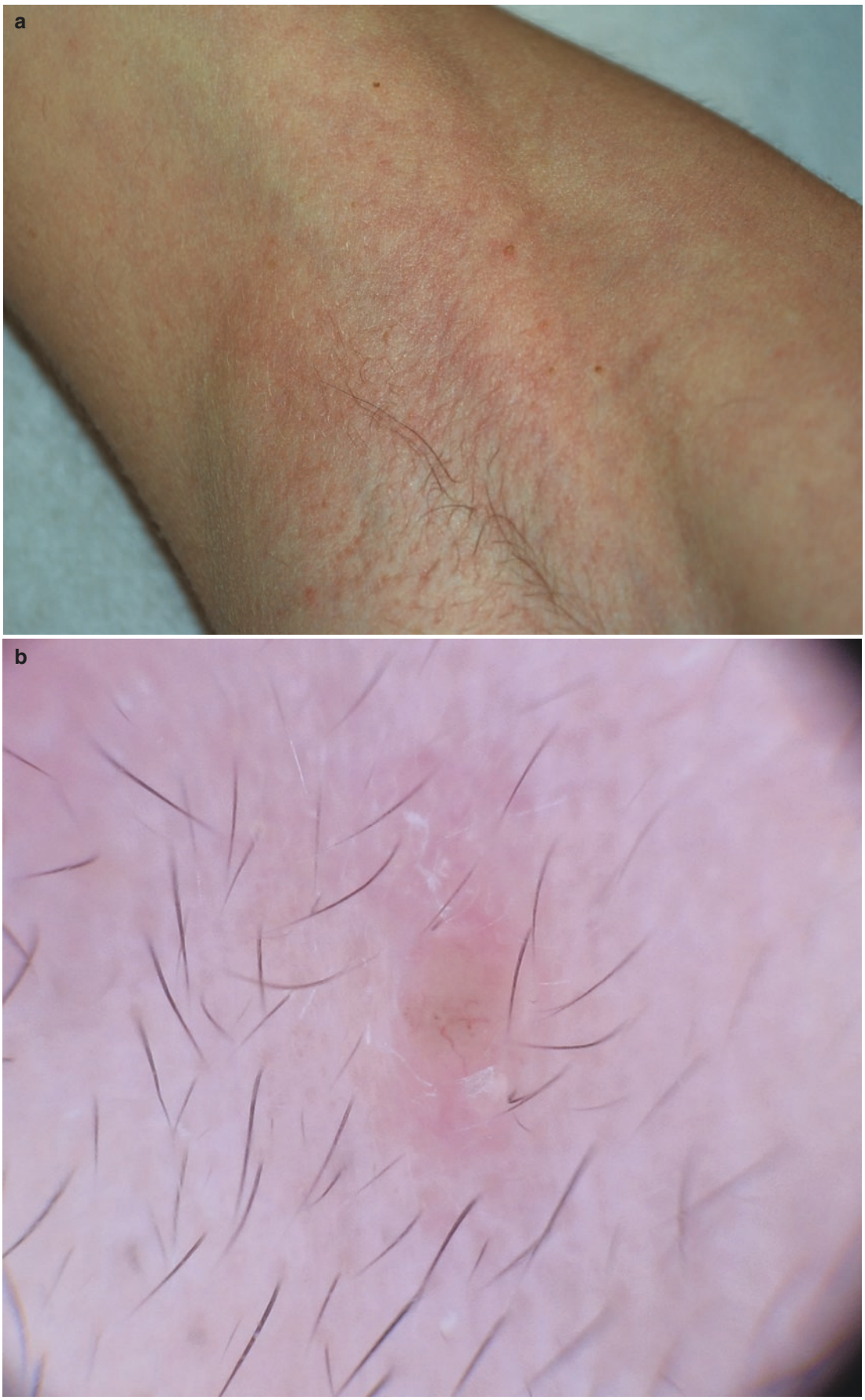

Fig. 30.1 (**a**) Basal cell nevus syndrome. Small nonpigmented basal cell carcinomas in a young patient. (**b**) Dermatoscopy (×10): arborizing vessels and few brown globules

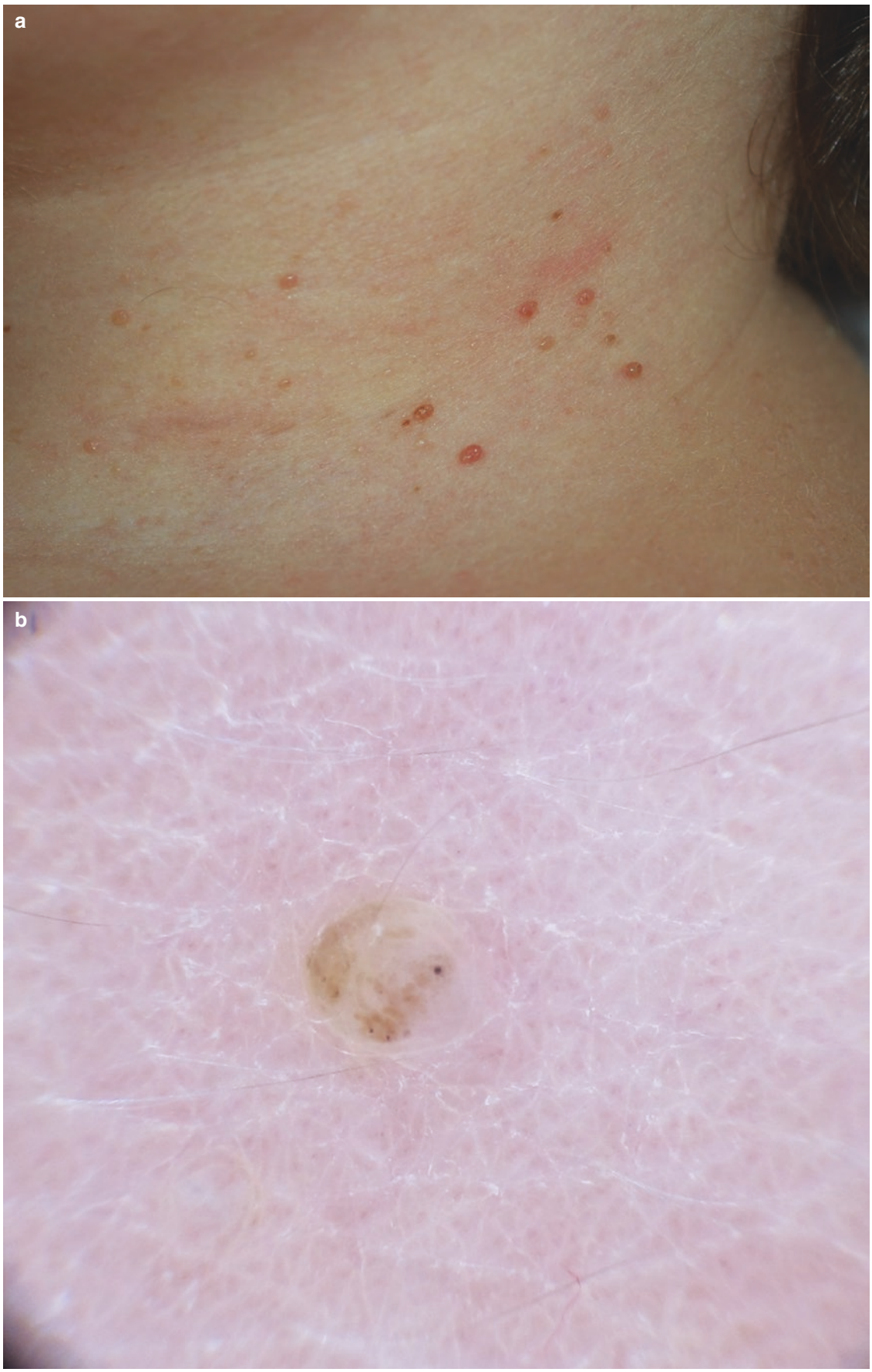

Fig. 30.2 (**a**) Basal cell nevus syndrome. Acrochordon-like basal cell carcinoma in a young patient. (**b**) Dermatoscopy (×10): blue-gray dots

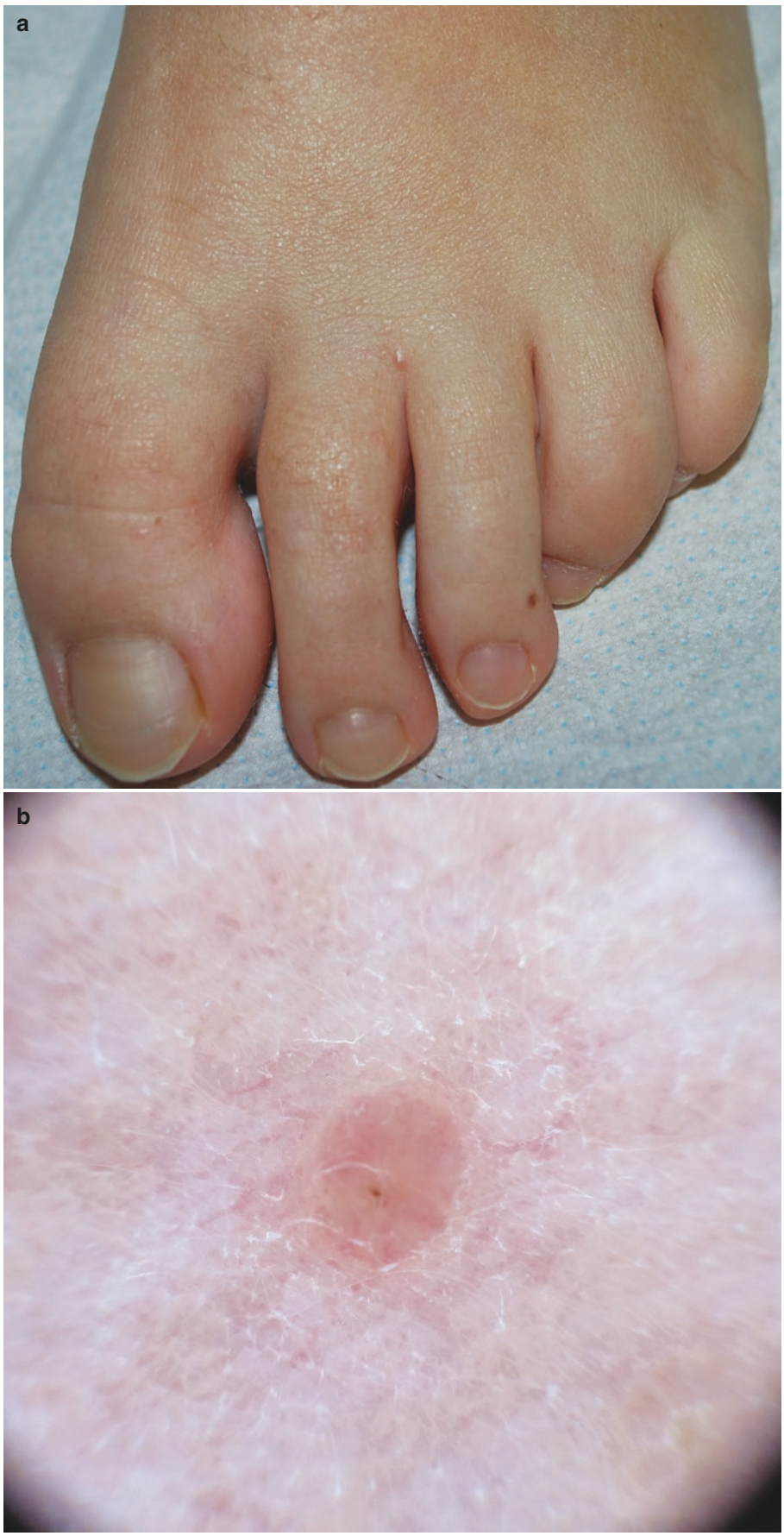

Fig. 30.3 (**a**) Basal cell nevus syndrome. Pinpoint papular variant of basal cell carcinoma localized on the extremities. (**b**) Dermatoscopy (×10): few blue-gray dots and fine superficial fine telangiectasia

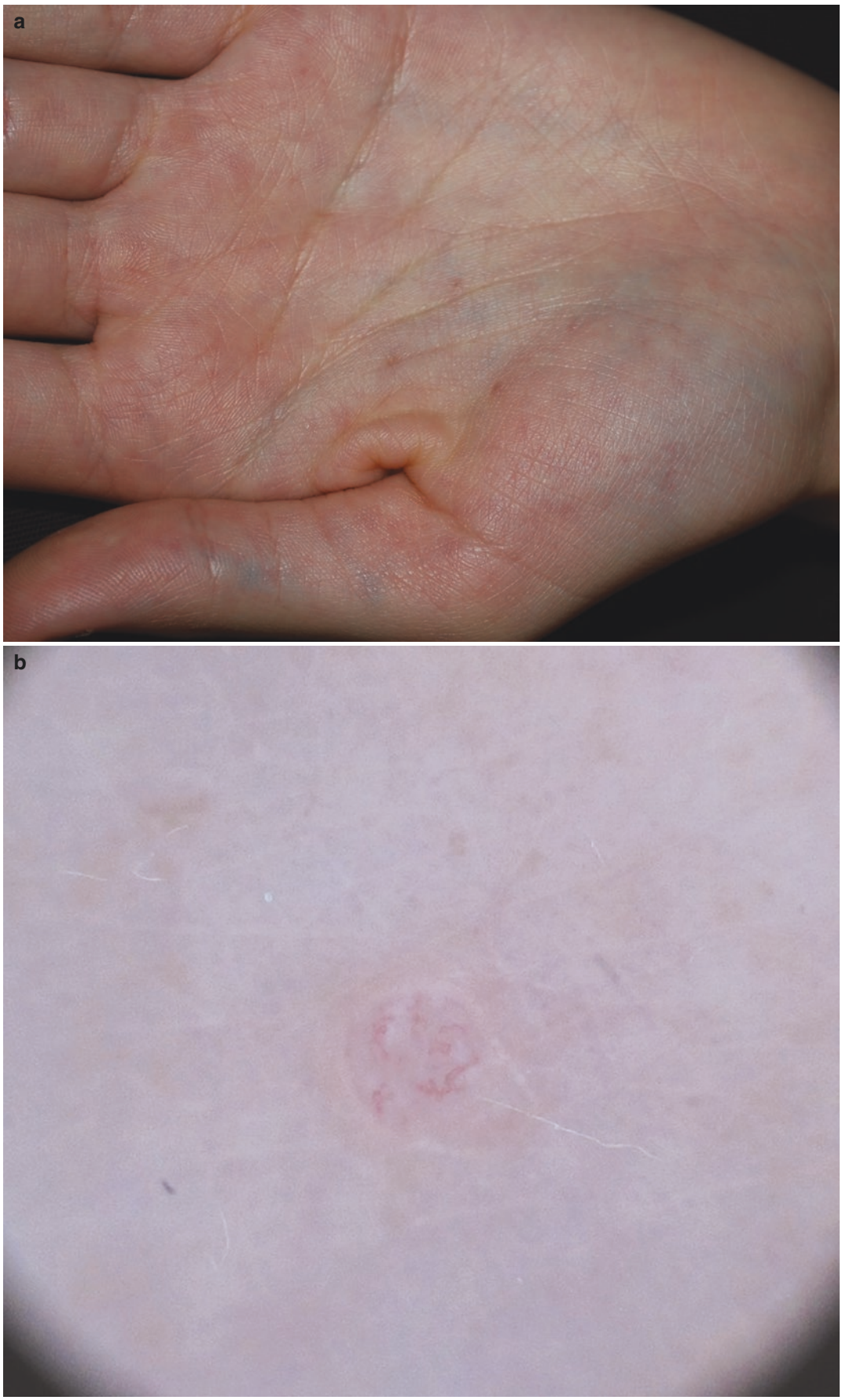

Fig. 30.4 (**a**) Basal cell nevus syndrome. Palmar pits: flesh-colored depressed lesions. (**b**) Dermatoscopy (×10): arborizing vessels

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Vascular Anomalies

31

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31.1 Definition

Vascular anomalies are a heterogeneous group of congenital disorders that are subcategorized based on their histology, biological behavior, and clinical presentation into vascular tumors, e.g., hemangiomas, and vascular malformations [1].

31.2 Epidemiology

Vascular anomalies are frequently seen in the pediatric age occurring in about 4.5% of all children [2, 3].

31.2.1 Hemangiomas

Hemangiomas are the most common tumor occurring in infancy. In a recent retrospective study, they occurred in about 1.97 per 100 person-years [4] although their incidence was previously estimated up to 10% [5]. Hemangiomas have been related to decrease in gestational age at birth and to a lower birth weight, along with an increase in pregnancy complications [4].

31.2.2 Vascular Malformations

Vascular malformations are very heterogeneous and occur in approximately 0.3% of the general population, excluding the salmon patch which shows a prevalence up to 50% [6, 7].

31.3 Etiology

31.3.1 Hemangiomas

Hemangiomas are localized or regional areas of abnormal vascular development and proliferation. Their pathogenesis remains unclear, but several factors may take part in the development of hemangiomas, including endothelial cells arising from disrupted placental tissue embedded in fetal soft tissues during gestation or birth, endothelial progenitor and stem cells in the circulation, and cytokines and genetic alterations in growth factor receptors [5].

31.3.2 Vascular Malformations

Vascular malformations are localized defects of vascular morphogenesis and structure without evidence of endothelial cell proliferation, involving capillaries, veins, lymphatic vessels, arteries, or a combination of these. Dysfunction in signaling processes that regulate migration, differentiation, maturation, adhesion, and survival of the cells of vascular walls is thought to play a pathogenic role.

31.4 Clinical Features

31.4.1 Hemangiomas

Most hemangiomas have a typical presentation and growth pattern developing in three clearly defined stages: proliferation, quiescence, and involution. They can be fully developed

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at birth (congenital hemangiomas) or present shortly after birth and grow rapidly (infantile hemangiomas). Infantile hemangiomas are classified as focal, multifocal, segmental, and indeterminate depending on their morphology, extent, or distribution and as superficial, deep, and mixed depending on their location in the dermis and/or hypodermis. Superficial lesions appear as early telangiectatic patches that after a few weeks begin to turn red and swell reaching variable sizes (Figs. 31.1a and 31.2a). Sometimes, they can be destructive and life-threatening. During proliferation, ischemia, necrosis, ulceration, bleeding, and secondary infection may occur. Deep lesions appear as warm, ill-defined, light blue-purple masses with minimal or no overlying skin changes, sometimes making the diagnosis difficult. Mixed hemangiomas have both superficial and deep components.

31.4.2 Vascular Malformations

In contrast to hemangiomas, vascular malformations are present at birth and never completely regress. They are commonly classified depending on flow characteristics (high/low flow) and which type of anomalous vessel is involved in capillary malformation (i.e., salmon patches, port-wine stain), venous malformation, lymphatic malformation (i.e., lymphangioma circumscriptum), and arteriovenous malformation.

The most common types of capillary malformations are salmon patches. They usually appear as irregular, pinkish, or reddish macules or patches located on the midface (forehead, glabella, eyelids, nose, philtrum) and occiput and may lighten and disappear with time. Another variant of capillary malformations is the port-wine stain, which usually appears as red-violaceous patches and, over years, may darken in color and thicken (Figs. 31.3a and 31.4a). The involvement of maxillary and/or ophthalmic dermatome may be signs of a complex syndrome (Sturge-Weber syndrome).

31.5 Differential Diagnosis

31.5.1 Hemangiomas

Hemangiomas may sometimes mimic many other types of vascular anomalies such as pyogenic granuloma, tufted angioma, infantile hemangiopericytoma, diffuse neonatal hemangiomatosis, as well as neuroblastoma, myofibromatosis, and lipoblastoma. Moreover, early lesions may sometimes be misdiagnosed as vascular malformations.

31.5.2 Vascular Malformations

The main differential diagnosis of vascular malformations is with hemangiomas.

31.6 Diagnosis

Most hemangiomas and vascular malformations can be diagnosed clinically based on patient history and lesion appearance. Histopathology and imaging techniques, such as magnetic resonance imaging and ultrasound, can be helpful to better evaluate deep lesions [8].

31.7 Dermatoscopy

31.7.1 Hemangiomas

Dermatoscopy reveals the presence of sharply demarcated and red round-oval structures resembling lacunae (Figs. 31.1b, c and 31.2b). A polymorphous vascular pattern consisting of red globular/circulated/comma-like/wavy/dilated/linear vessels has been described. Blue-whitish septa usually separate the lacunae from each other. If any of these lacunae develops a thrombus, they will appear black [9–12].

31.7.2 Vascular Malformations

Few studies reported dermatoscopic features of vascular malformation. Lesions may reveal three dermatoscopic patterns: *type 1* or superficial, characterized by a vascular structure of red-dotted, globular vessels (Fig. 31.3b); *type 2* or deep, revealing red dilated, linear vessels forming irregular networks (Fig. 31.4b, c); and *type 3*, showing round/sacular/glomerular structures [13, 14]. Nonvascular structures have also been described, including a perifollicular pale halo and a gray-whitish veil [15, 16]. Some authors have highlighted the importance of dermatoscopy in the prognostic evaluation of treatment response. Superficial lesions have been related to laser therapy to a better response than deeply located vessels [12, 15–17].

31.8 Histopathological Correlation

31.8.1 Hemangiomas

The vascular pattern seen at dermatoscopy correlates with ectatic vascular structures in the papillary and subpapillary dermis.

31.8.2 Vascular Malformations

These lesions are characterized by the presence of ectatic vessels at different depth. The red round globular vessels seen at dermatoscopy are related to superficial ectatic vertical papillary vessels, whereas the red linear vessels correspond to deeper dilated horizontal subpapillary capillaries. Enlargement of the horizontal plexus over time correlates to the roundish vascular structures seen at dermatoscopy.

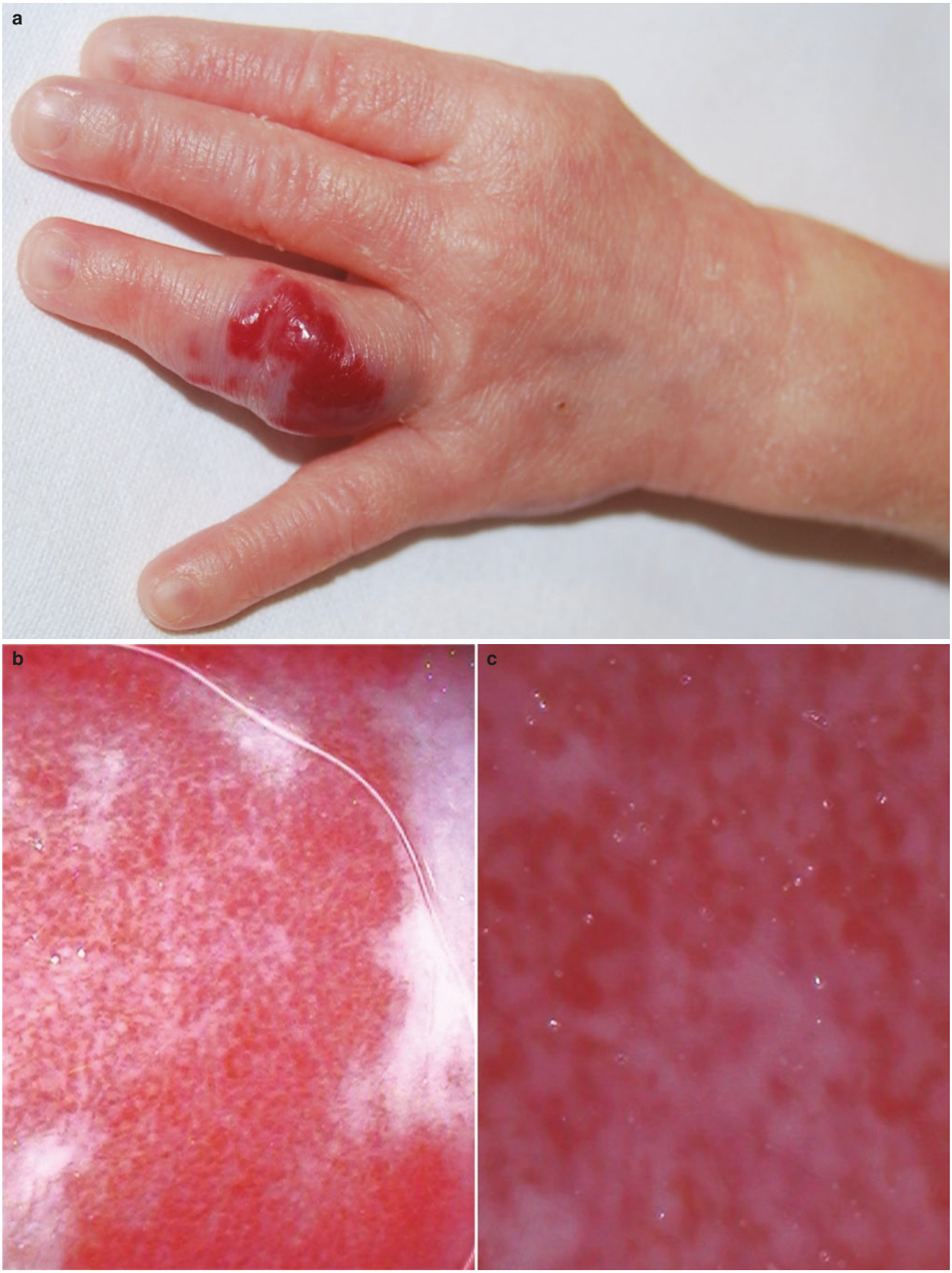

Fig. 31.1 (**a**) Hemangioma. Reddish tumor involving the forth finger in a 1-month-old girl. (**b**) Dermatoscopy (×10): diffuse, red round-oval structures resembling lacunae with interposed whitish septa. (**c**) High-magnification dermatoscopy (×100): enhanced visualization of dilated vessels

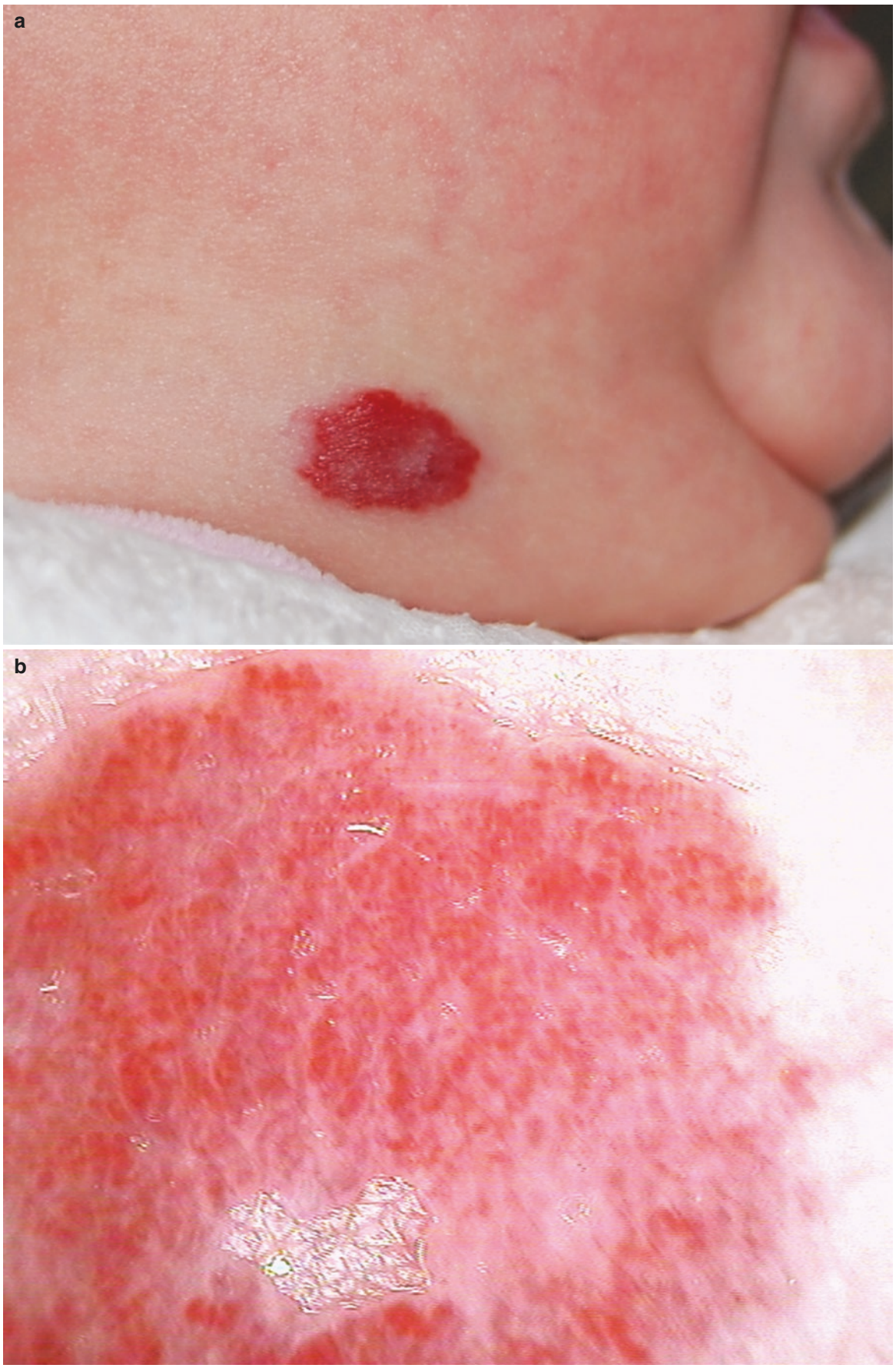

Fig. 31.2 (**a**) Hemangioma. Reddish plaque on the face in a 1-year-old girl. (**b**) Dermatoscopy (×10): red round-oval structures resembling lacunae and whitish septa

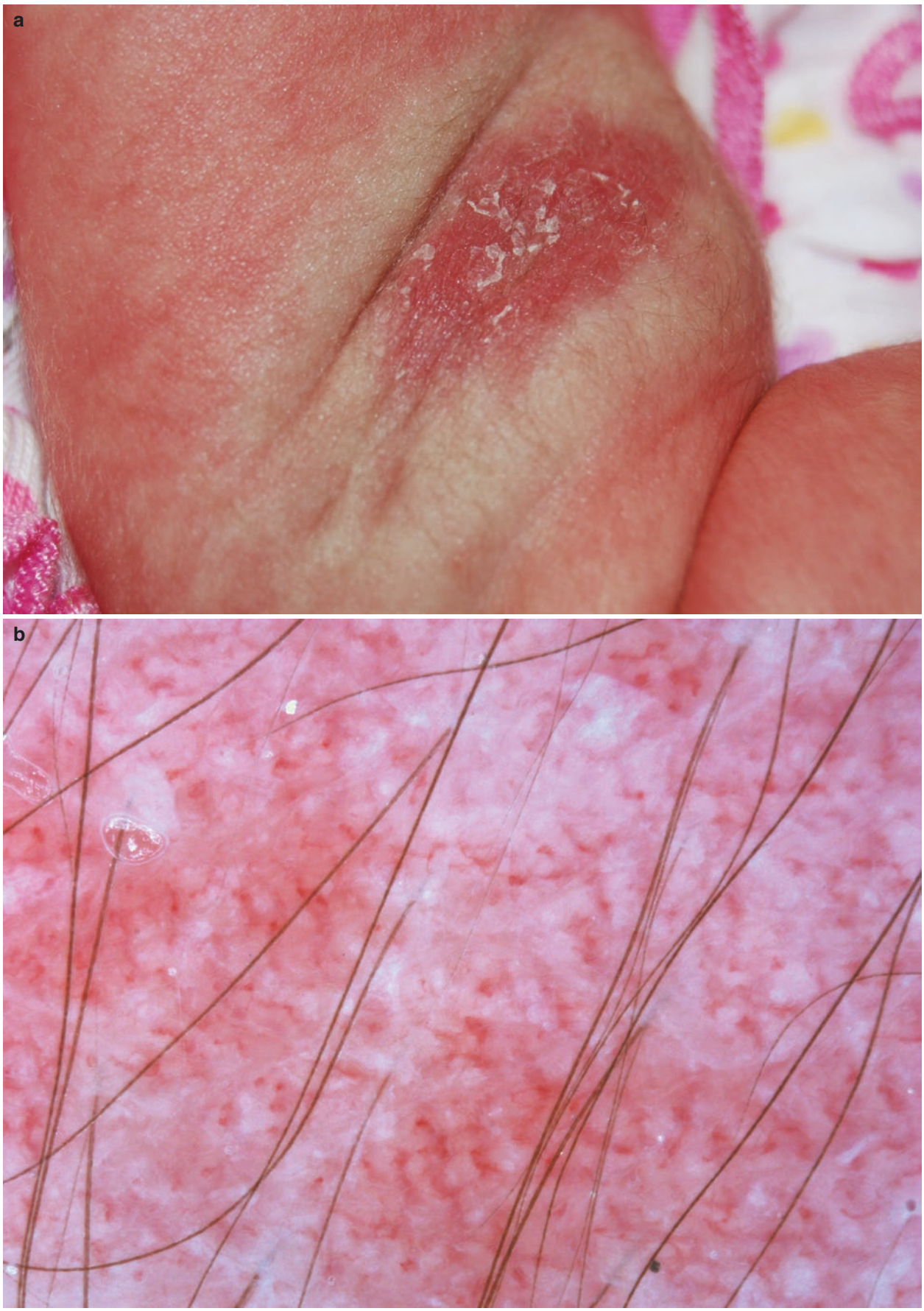

Fig. 31.3 (**a**) Vascular malformation. Reddish macule of the arm in a 2-month-old girl. (**b**) High-magnification dermatoscopy (×100): red-dotted and globular vessels (*type 1* pattern)

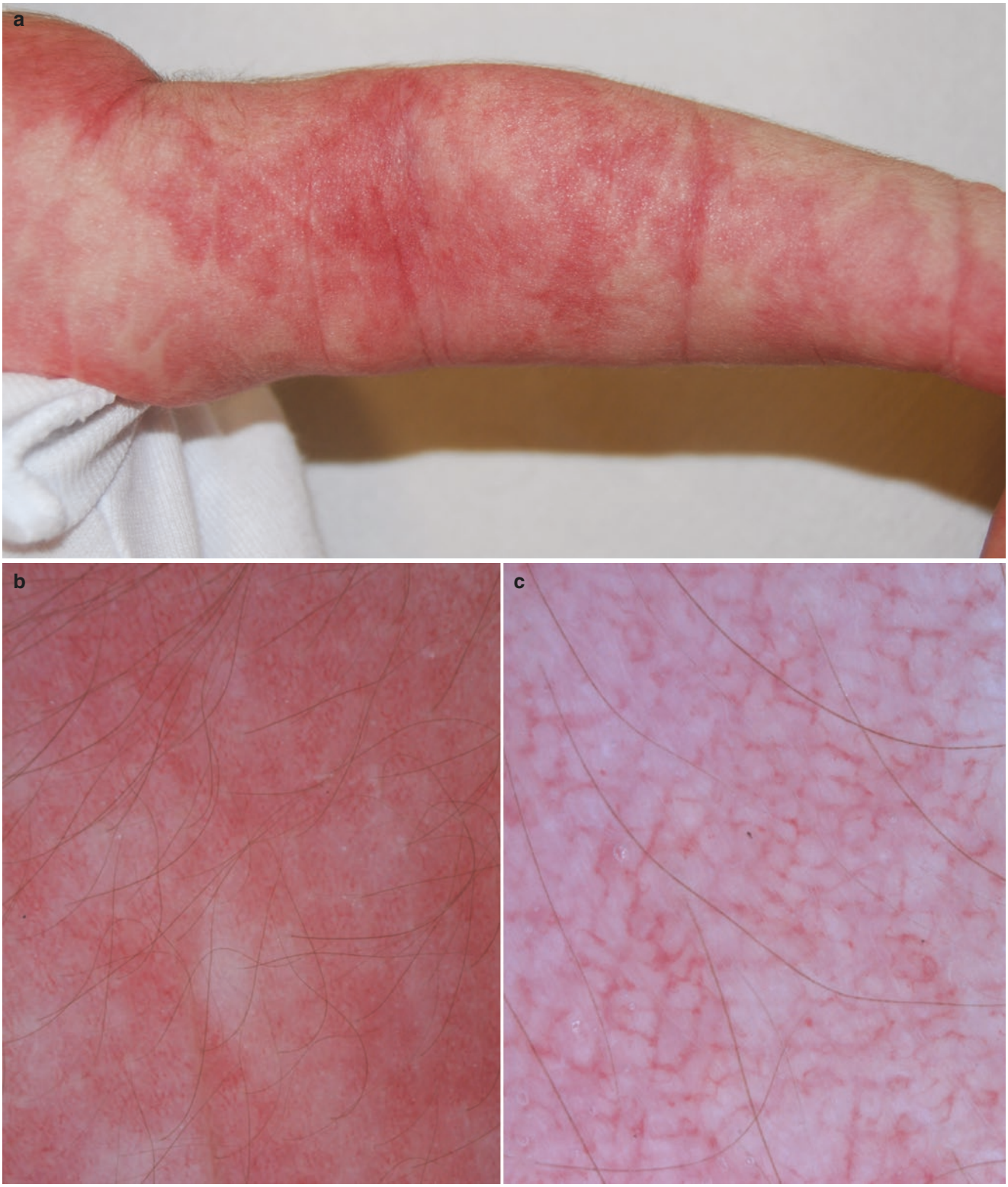

Fig. 31.4 (**a**) Vascular malformation. Reddish macule involving almost the entire arm in a 3-month-old boy. (**b**) Dermatoscopy (×10): diffuse dilated, linear vessels and some perifollicular pale halos (*type 2* pattern). (**c**) High-magnification dermatoscopy (×100): linear vessels forming an irregular network

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Pyogenic Granuloma

32

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32.1 Definition

Pyogenic granuloma (PG), also known as lobular capillary hemangioma, is a benign vascular lesion that can occur on the skin and superficial mucosae.

32.2 Epidemiology

PG is a frequent lesion of the childhood and adolescence, representing about 0.5% of all skin nodules in this age [1, 2]. In childhood, the ratio between male and female is 3:2, while in adulthood this ratio is inverted, because of the high number of women affected during pregnancy [3].

32.3 Etiology

The exact etiology of PG is unknown. An inflammatory, hyperproliferative vascular response to different triggering factors, such as infections, trauma, or burns, has been supposed. Moreover, an important role might be played by estrogens. Its onset on a preexisting nevus flammeus or spider nevus has been described [3, 4].

32.4 Clinical Features

PG is usually characterized by a rapid growth in a period of few weeks. It appears as a soft reddish papule, nodule, or polypoid lesion, often bleeding following minor traumas (Figs. 32.1a, 32.2a, and 32.3a). In children, the most affected sites are the head, in particular the face, lips, oral and nasal mucosa, and the upper extremities (fingers) [4].

32.5 Differential Diagnosis

Differential diagnosis includes amelanotic melanoma and other nonpigmented lesions, such as basal cell carcinoma, squamous cell carcinoma, keratoacanthoma, inflamed seborrheic keratosis, common wart, Spitz nevus, Kaposi sarcoma, and true hemangioma [3].

32.6 Diagnosis

A correct diagnosis is based on an accurate collection of clinical history and physical examination, although histopathology is sometimes needed [5].

32.7 Dermatoscopy

Five main dermatoscopy findings have been recognized in PG: reddish homogeneous area, white collarette, white rail lines, vascular structures, and ulceration [1]. The reddish homogeneous area (Fig. 32.1b) represents the most common dermatoscopic structure related with PG, but has low specificity, as it may be present in other lesions, especially amelanotic melanoma. The white collarette is a whitish ring that completely or partially surrounds the PG, while the

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white rail lines are whitish bands that divide the lesion in lobules (Figs. 32.2b and 32.3b). As regards the vascular structures, different types of vessels have been described, such as telangiectasias, hairpin vessels, dotted vessels, etc. Although none of these single signs is specific for PG, the combination of three or more of them is very indicative for the diagnosis [1].

Dermatoscopy may be also useful for the evaluation of response to treatment [6].

32.8 Histopathological Correlation

Histopathologically, PG shows clustered of hyperplastic vessels arranged in lobules separated by fibrous septa (Fig. 32.4) [7]. In particular, the red homogeneous area observed at dermatoscopy corresponds to small capillaries or proliferating vessels, the white collarette correlates to epidermal hyperplasia, and the "white rail" lines correspond to the fibrous septa that surround the capillary lobules [1].

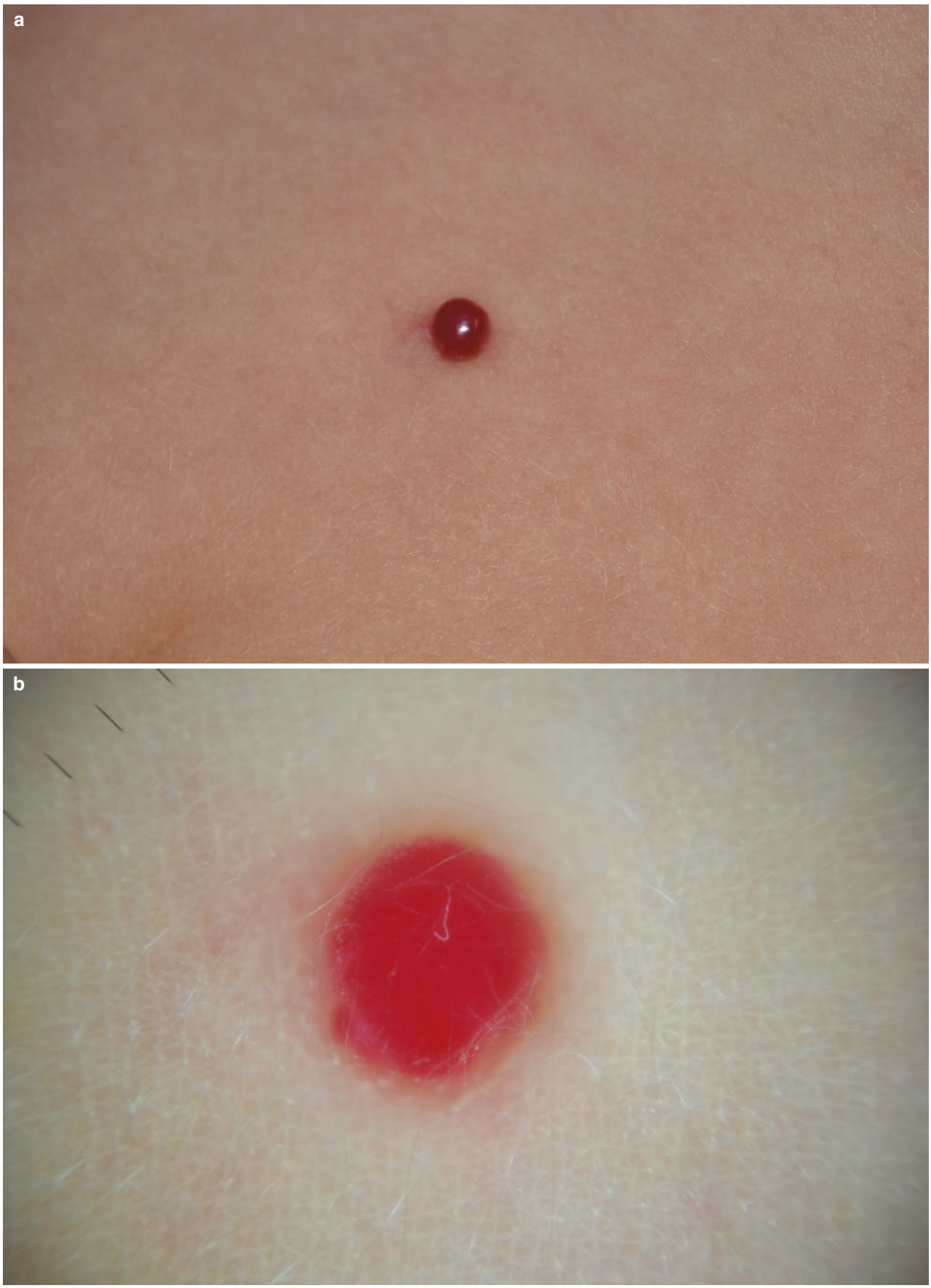

Fig. 32.1 (**a**) Pyogenic granuloma. Reddish papule located on the back of a 5-year-old girl. (**b**) Dermatoscopy (×10): red homogeneous color all over the lesion (*reddish homogeneous area*)

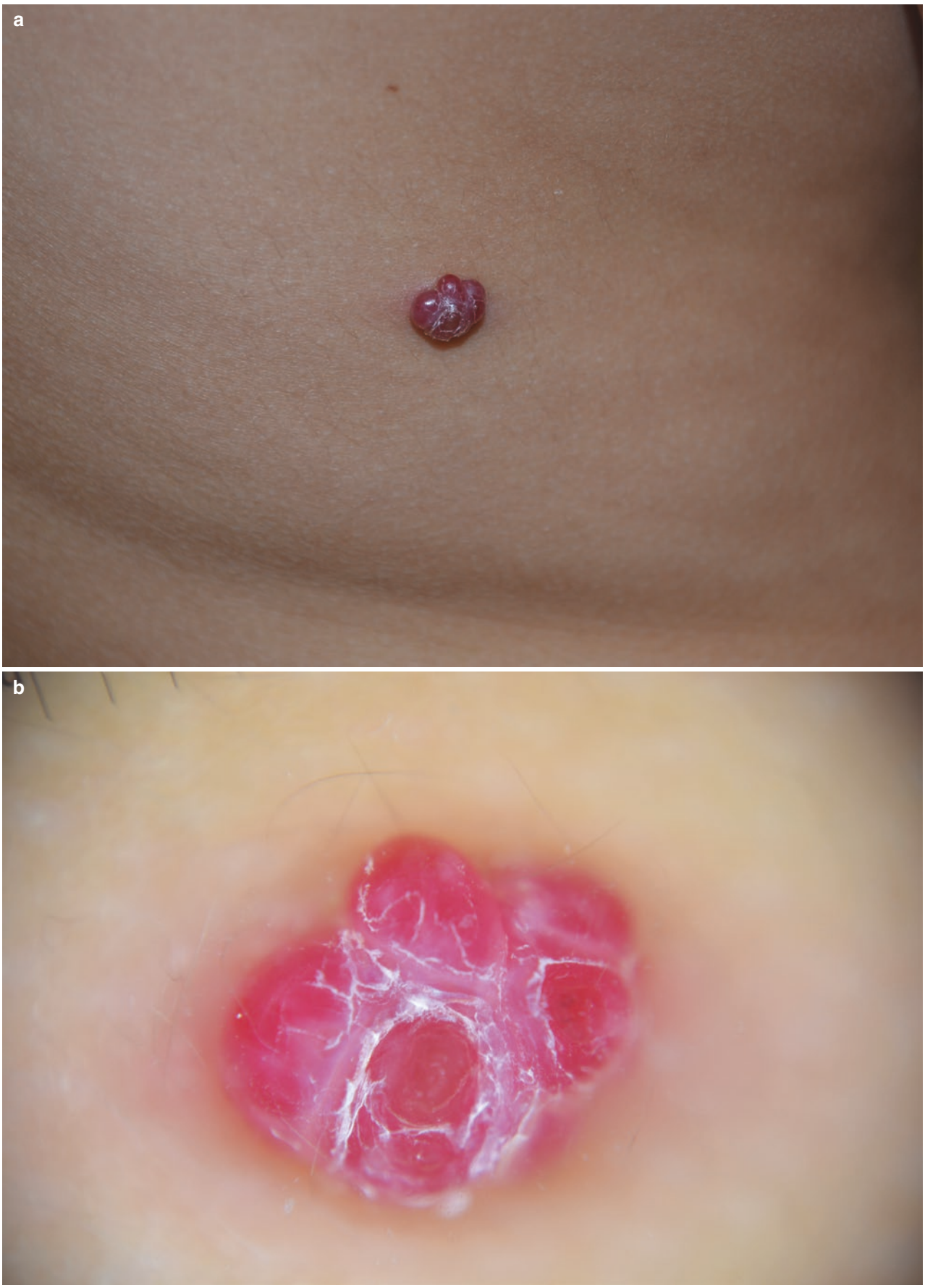

Fig. 32.2 (**a**) Pyogenic granuloma. Reddish, polylobulated, and irregular-shaped nodule on the trunk of a 9-year-old boy. (**b**) Dermatoscopy (×10): multiple reddish areas separated by white rail lines

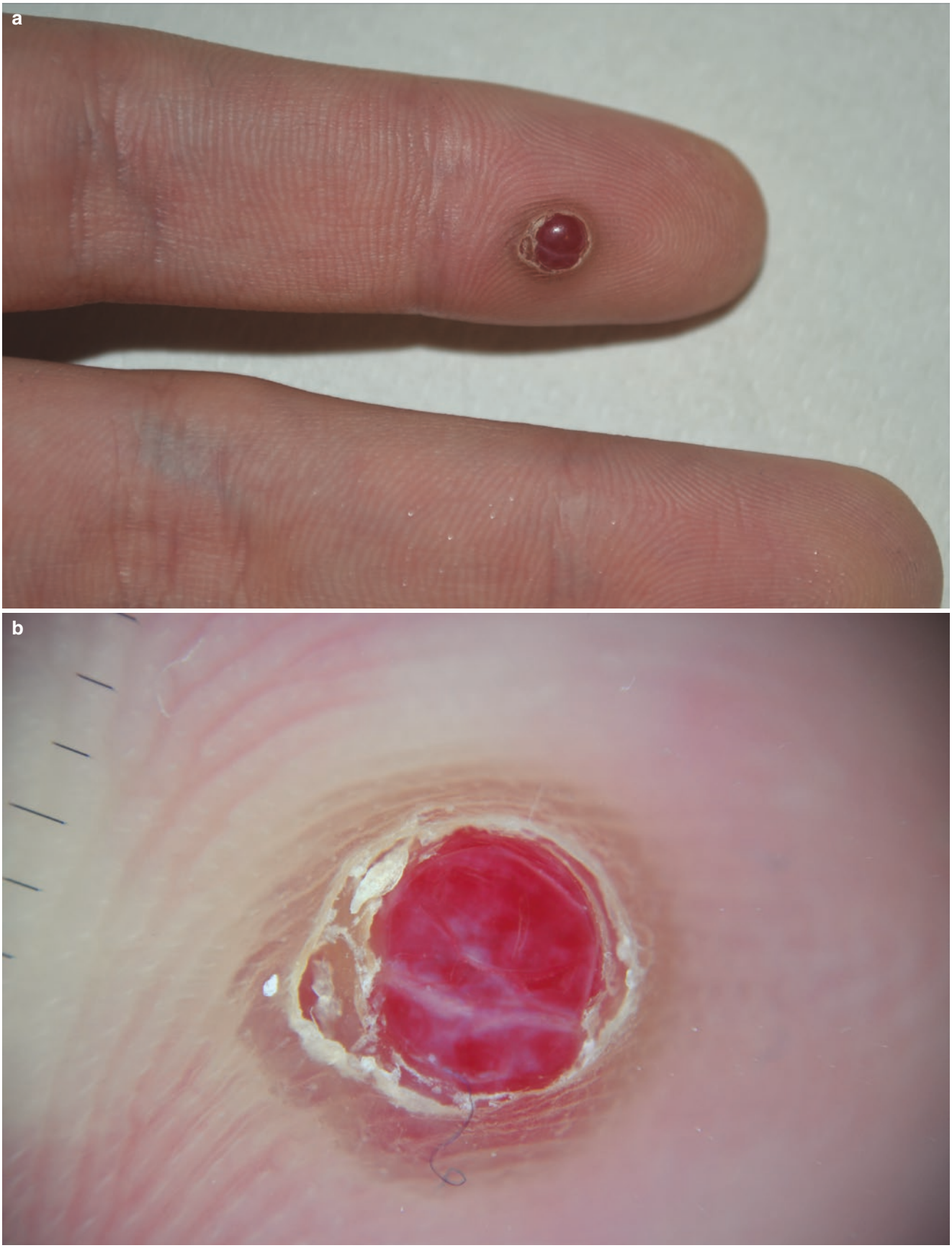

Fig. 32.3 (**a**) Pyogenic granuloma. Reddish papule demarcated by a white collarette located on a fingertip. (**b**) Dermatoscopy (×10): multiple reddish areas separated by white rail lines with a marked collarette surrounding the lesion

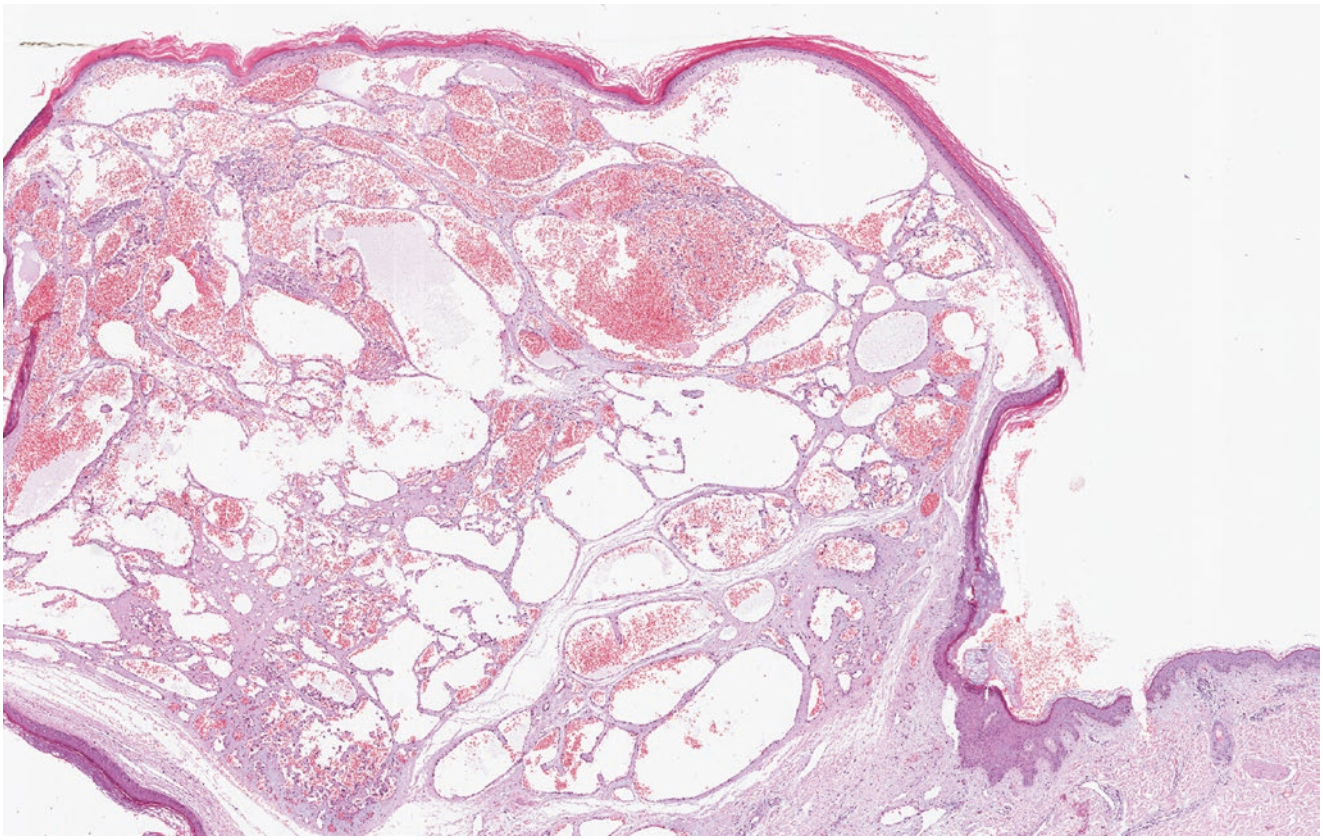

Fig. 32.4 Pyogenic granuloma. Histopathology: hyperplastic vessels arranged in lobules separated by fibrous septa [H&E staining; magnification, ×100]

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Lymphatic Malformations

33

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33.1 Definition

Superficial lymphatic malformations (LMs) include a group of vascular malformations of the lymphatic network involving the skin and the subcutaneous tissue. A recent reclassification of International Society for the Study of Vascular Anomalies (ISSVA) in 2014 divides common cystic lymphatic malformation in microcysts, macrocysts, and mixed forms according to the vessels size [1].

33.2 Epidemiology

Microcystic LM accounts for 4% of all vascular tumors, representing the 25% of all benign vascular tumors in children. About 50% of them are observed at birth or manifests by 2 years of age.

33.3 Etiology

Anomalous lymphatic cisterns grow apart from the regular vascular network, probably arising from a primitive lymphatic sac.

33.4 Clinical Features

Microcystic LM clinically presents as a cluster of translucent and clear vesicles (compared to frog spawn) that tend to increase in number and size (Fig. 33.1a). Such lesions may turn purplish or black colored as a result of blood leakage or frank hemorrhage (Figs. 33.2a and 33.3a). The most involved sites are the extremities, trunk, axillae, and tongue.

Macrocystic LM manifests as palpable tender subcutaneous masses with a superimposed thickened skin. Superinfection is not uncommon [1].

33.5 Differential Diagnosis

Differential diagnosis mainly includes angiokeratoma, warts, molluscum contagiosum, epidermal naevi, and herpes zoster.

33.6 Diagnosis

Diagnosis is usually made clinically although confirmation by ultrasound or magnetic resonance imaging can be necessary in order to evaluate the depth and extension of the lesion.

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33.7 Dermatoscopy

Dermatoscopy reveals a whitish or yellowish lacunar pattern (Fig. 33.1b). Frequently, based on the possible presence of blood, scattered reddish areas within the yellowish lacunae (Fig. 33.2b) or sole reddish lacunae (Fig. 33.3b) may be observed. In some cases, the variable quantity of blood content within the lacunae generates a wide spectrum of color transition from yellow to purple, which is described as hypopyon-like pattern [2–5].

33.8 Histopathological Correlation

The yellowish lacunae histopathologically correspond to irregular lymphatic channels proliferating under a thinned epidermis (Fig. 33.4) [6]. The reddish areas are related to blood cells extravasation [5].

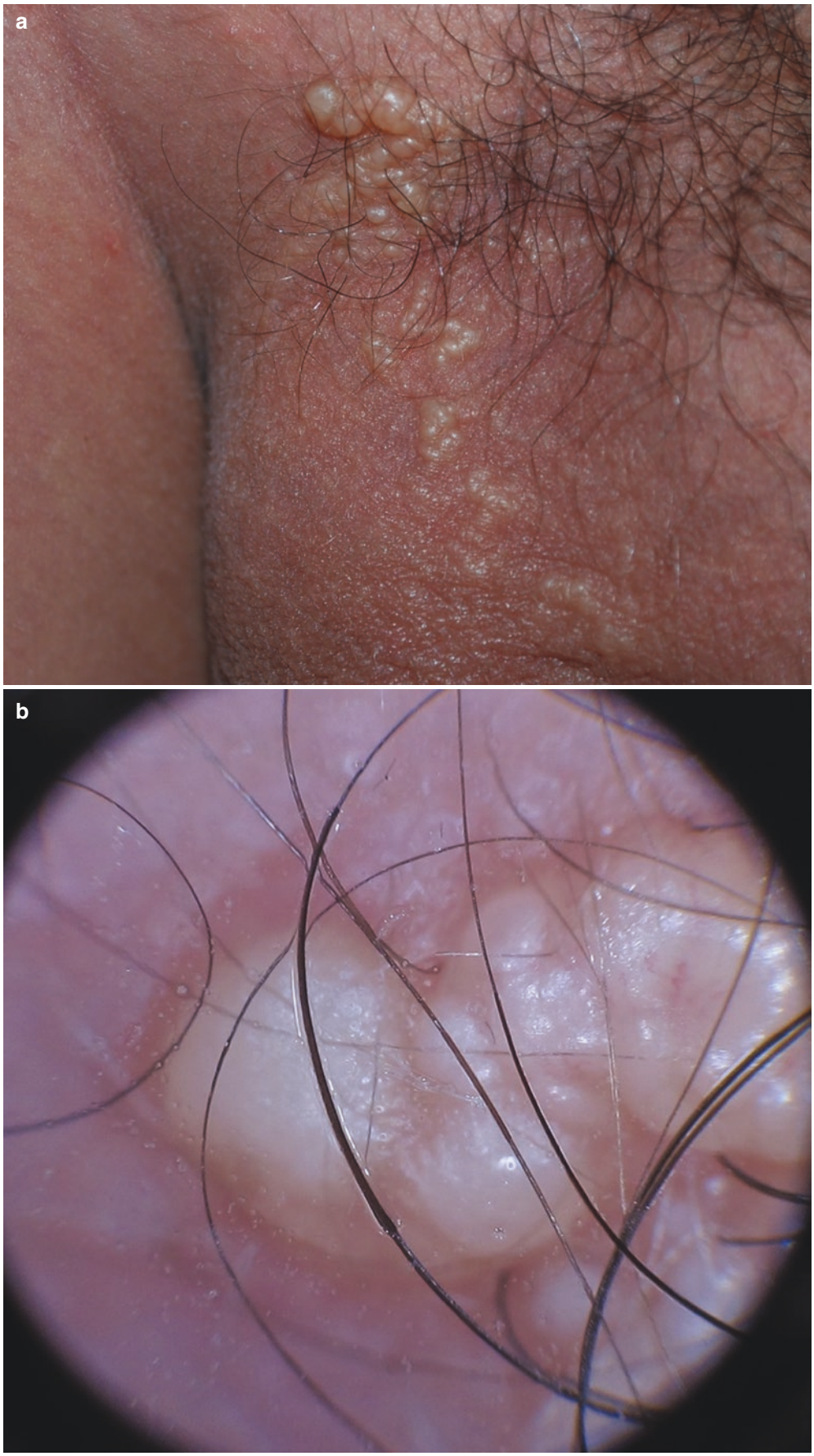

Fig. 33.1 (**a**) Microcystic lymphatic malformation in a boy. (**b**) Dermatoscopy (×10): whitish lacunae surrounded by pale septa

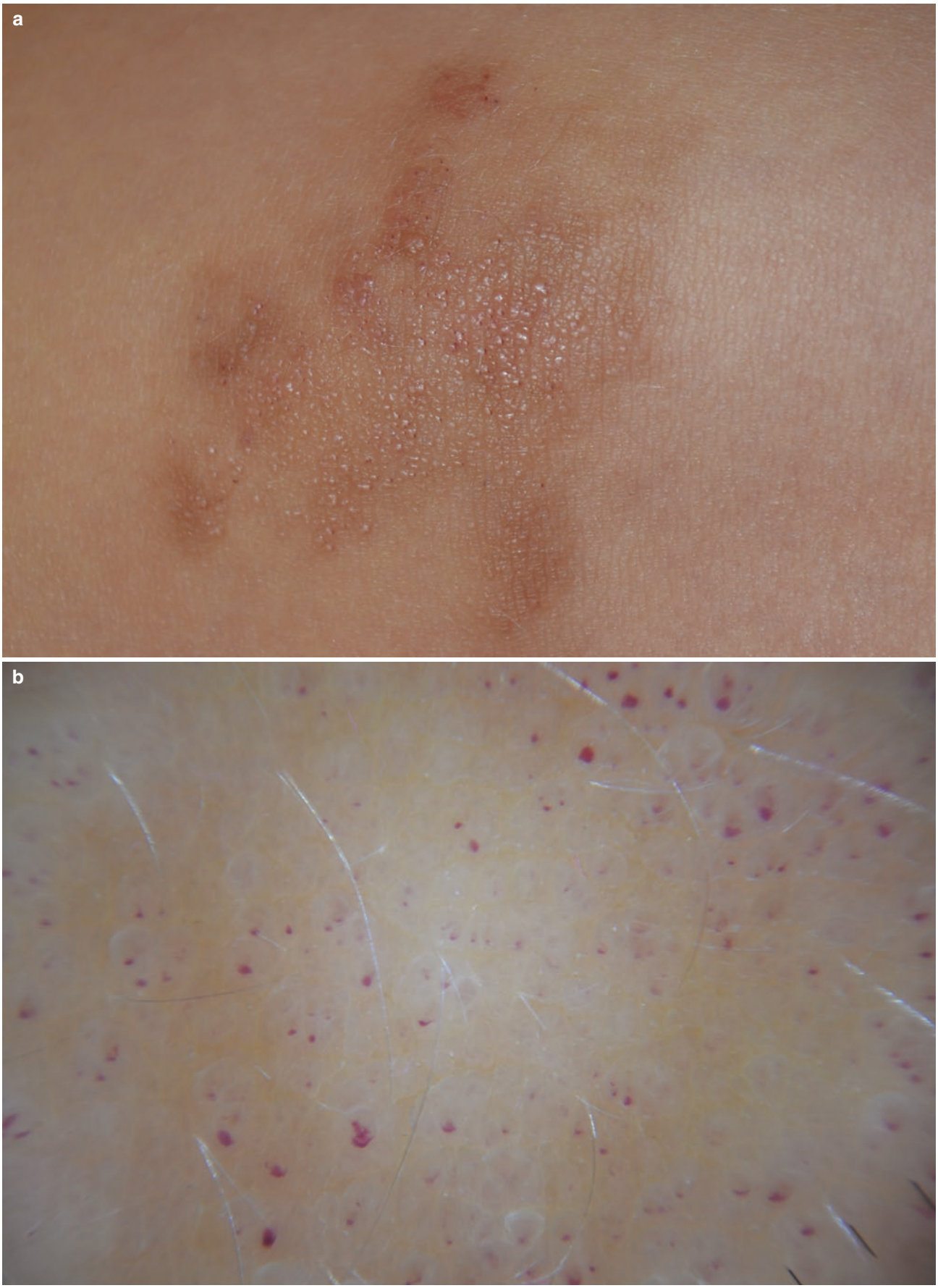

Fig. 33.2 (**a**) Microcystic lymphatic malformation in a 4-year-old girl. (**b**) Dermatoscopy (×10): yellowish lacunae associated with scattered red areas (Courtesy of Giuseppe Micali, MD)

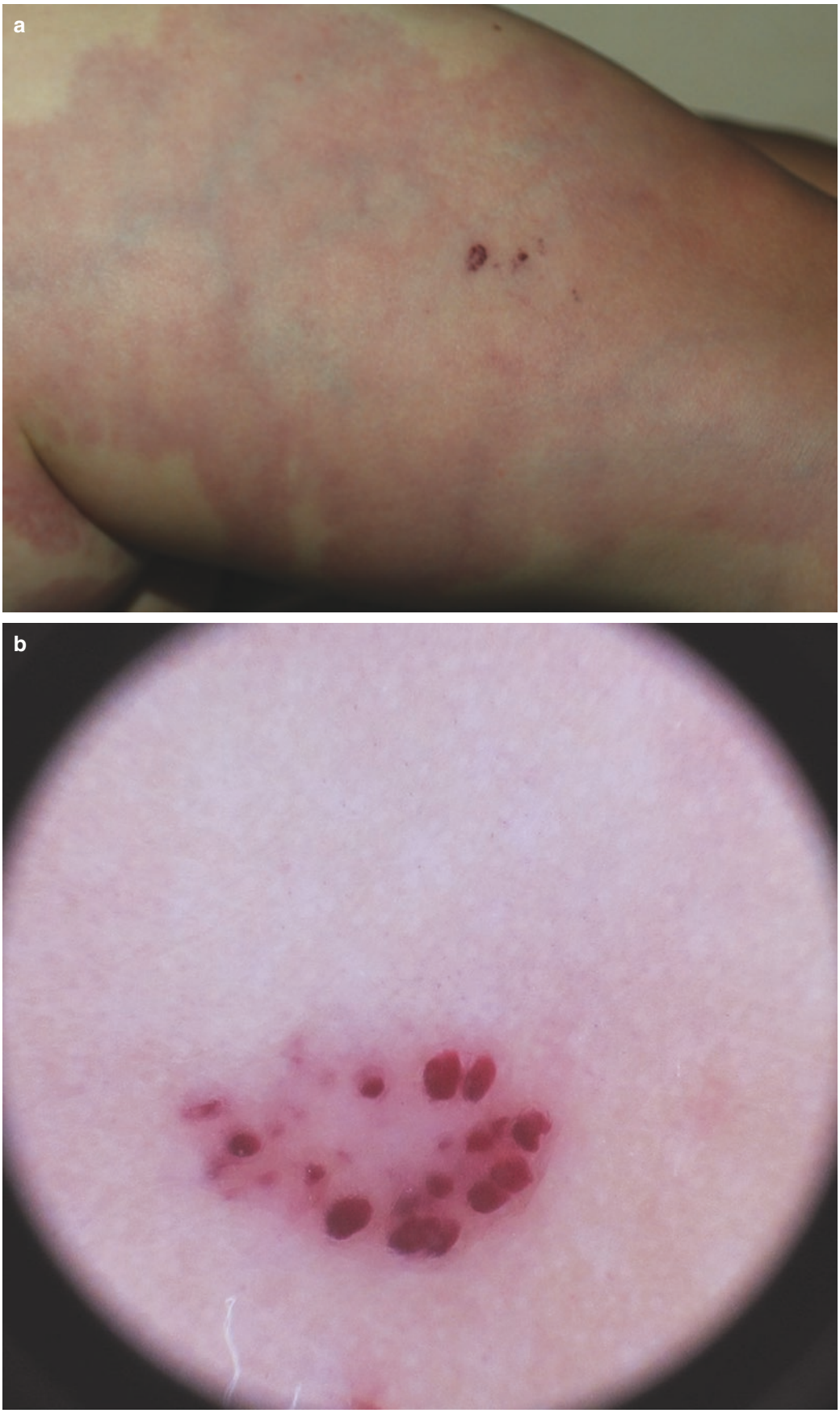

Fig. 33.3 (**a**) Combined vascular malformation: superficial lymphatic malformation in the context of a capillary malformation. (**b**) Dermatoscopy (×10): reddish lacunae

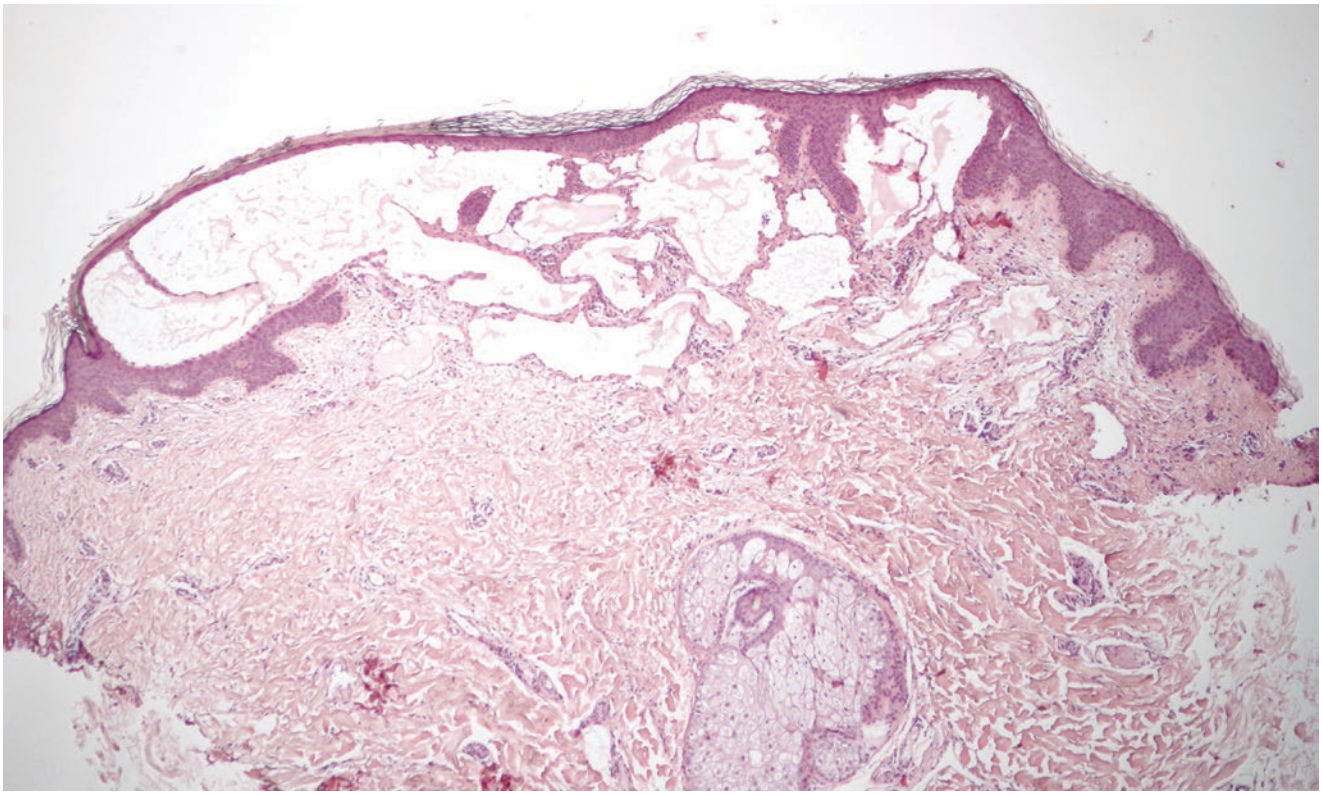

Fig. 33.4 Microcystic lymphatic malformation. Histopathology showing thin epidermis with underlying proliferating and irregular lymphatic channels [H&E staining; magnification, ×40]

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Developmental Defects

34

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34.1 Definition

Developmental defects are errors of embryologic development arising during fetal life. Among these, median raphe cyst, also known as mucous cysts of the penis, genitoperineal cysts of the medium raphe, parameatal cysts, hydrocystoma, and apocrine cystadenoma of the penile shaft, and aplasia cutis congenita of the scalp have been reported to show diagnostic dermatoscopic patterns.

34.2 Median Raphe Cyst

34.2.1 Epidemiology

It is considered an uncommon developmental defect, although it is probably underreported as most cases are asymptomatic [1].

34.2.2 Etiology

The etiopathogenesis is not well understood. Three main theories have been proposed to explain its occurrence: incomplete ventral fusion of the urethral or genital folds, ectopic periurethral glands of Littré (mucoid cysts), or separated outgrowths of urethral endoderm (urethroid cysts) [2].

34.2.3 Clinical Features

It can occur anywhere along the median raphe, located on the ventral side of the male genital area between the urethral meatus and the anus, and usually appears as a translucent, yellowish, tender nodule [3]. Multiple linearly arranged lesions may be observed. When two or more cystic lesions are connected, the term of median raphe canal may be used (Fig. 34.1a), although this connection may sometimes be clinically difficult to visualize. Sometimes, an indurated cord-like lesion may occur. Generally, both cysts and canals are asymptomatic and unrecognized during childhood, but they may enlarge and become symptomatic, especially following traumas and bacterial infections, during adolescence or adulthood.

34.2.4 Differential Diagnosis

The differential diagnosis of median raphe cyst mainly includes steatocystoma, molluscum contagiosum, pilonidal cyst, dermoid cyst, epidermoid cyst, and urethral diverticulum.

34.2.5 Diagnosis

The diagnosis is usually incidental. The history and physical examination are in most cases suggestive although the histopathology examination is sometimes necessary.

34.2.6 Dermatoscopy

Dermatoscopy may confirm the clinical diagnosis by showing the presence of a homogeneous, white/yellowish pattern. Moreover, by revealing a translucent tract between two cysts, it may highlight the presence of a clinically undetected connection so to address to the final diagnosis of median raphe canal (Figs. 34.1b, c) and, consequently, to a more appropriate therapeutic approach [4, 5].

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34.2.7 Histopathological Correlation

Histopathologically, both cysts and canals are typically lined by stratified squamous or pseudostratified columnar epithelium with no communication to the urethra.

34.3 Aplasia Cutis Congenita of the Scalp

34.3.1 Epidemiology

Aplasia cutis congenita is a rare malformation with an incidence of approximately 1–2.8/10,000 births [6]. It is usually located on the scalp (85% of cases) but can occur anywhere on the body including the face, trunk, and extremities [7]. Female are more commonly affected than male [8].

34.3.2 Etiology

The exact etiology and pathogenesis of aplasia cutis congenita remain unclear although many abnormalities have been suggested, including neural tube defects (encephalocele/meningocele), focal pressure, necrosis, paucity or impaired vascular development in the cranial vertex, incomplete healing and fusion of the mesoderm, ischemic or thrombotic episodes, teratogen drugs (methimazole and carbimazole), and congenital infections [8].

34.3.3 Clinical Features

In 1986, Frieden proposed a classification of aplasia cutis congenita into nine groups according to the location and pattern of skin absence, causes, associated anomalies, and the type of inheritance [9]. In 2014, a treatment-oriented classification of aplasia cutis congenita of the scalp has been suggested in order to estimate the severity/prognosis of the disease and provide management guidelines [6]. Nonsyndromic scalp aplasia cutis congenita is usually a solitary lesion, located predominately in the midline vertex (Fig. 34.2). Although rarely it can present at birth as an ulcerative lesion, most commonly it appears as a noninflammatory, well-demarcated, alopecic patch covered, especially in the first years of life, with a thin, fragile, transparent membrane. Moreover, the "hair collar sign," consisting of a ring of dark, coarse, thick hairs surrounding the lesion, may be clinically detected [10].

34.3.4 Differential Diagnosis

Aplasia cutis congenita must not be confused with alopecia areata, which is the most common cause of patchy alopecia in children. Other differential diagnoses include nevus sebaceous, tinea capitis, congenital triangular alopecia, trichotillomania, and epidermolysis bullosa.

34.3.5 Diagnosis

Diagnosis is usually based on clinical presentation. The presence of an ulcer at birth as well as the existence of the hair collar sign may be helpful.

34.3.6 Dermatoscopy

Some studies reported the dermatoscopy pattern of aplasia cutis congenita of the scalp [11–13]. Lesions show a central area of scarring alopecia with no follicular openings and some telangiectatic vessels. At the hair-bearing margin, hair bulbs are visible through a semitranslucent epidermis (Fig. 34.2). These hair bulbs are horizontally and radially oriented, configuring the *starburst hair follicles pattern* [11], probably due to aberrant shearing forces occurring early at some point of follicle development forcing them to bend [10]. The recognition of this pattern is essential to definitively rule out other forms of localized hair patches of alopecia that show different dermatoscopic aspects, such as sebaceous nevus (Fig. 34.3) [14, 15].

34.3.7 Histopathological Correlation

Histopathologic examination reveals the presence of flattened epidermis and cicatricial alopecia.

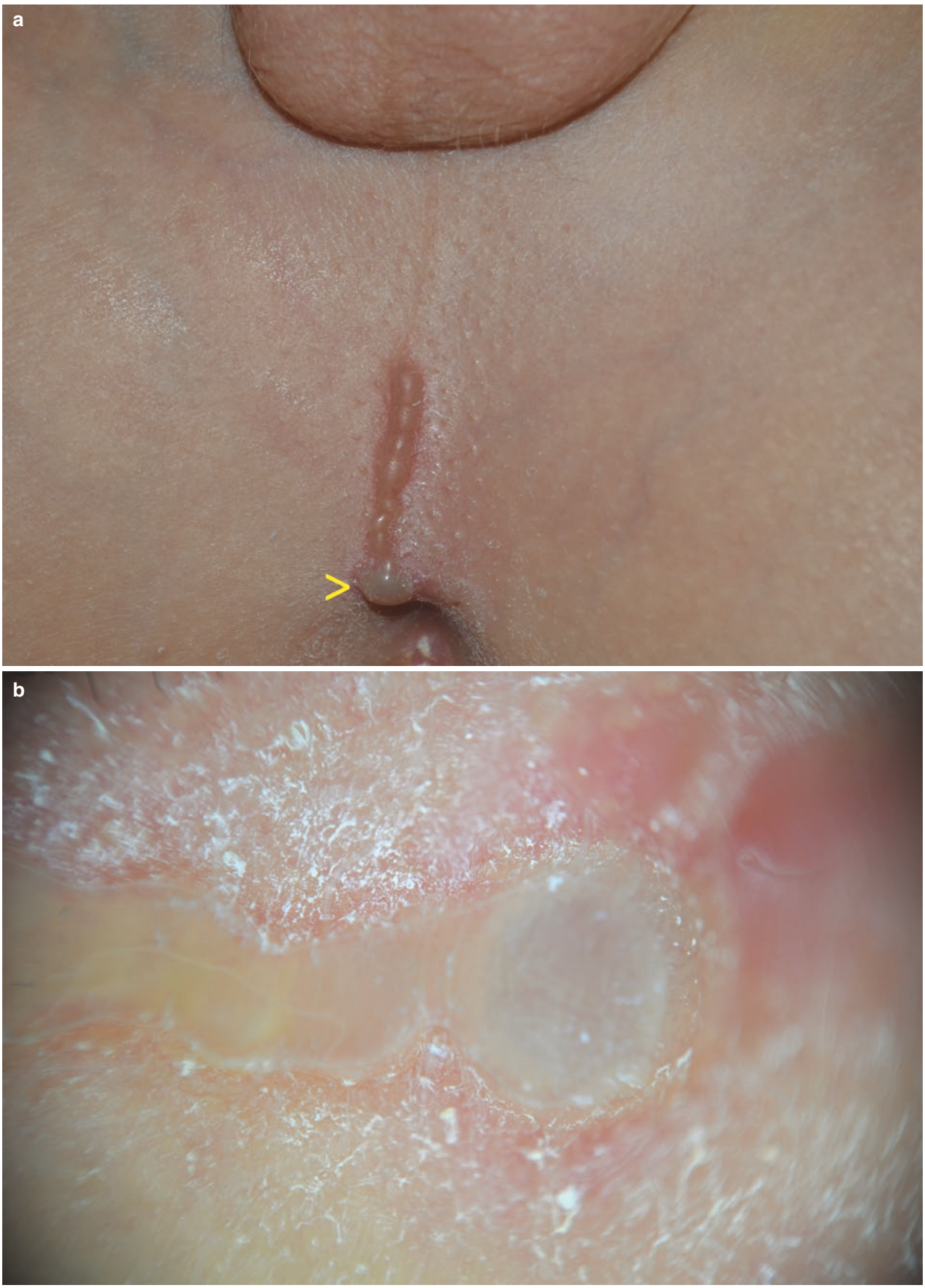

Fig. 34.1 (**a**) Median raphe canal. Translucent nodule of the perianal area (arrowhead) contiguous to a linear raised tract along the median raphe in a 3-year-old boy. (**b**) Dermatoscopy (×30) of the translucent nodule: homogeneous, whitish pattern. The nodule appears to be connected to the adjacent linear tract which shows the same dermatoscopic features. (**c**) Dermatoscopy (×30) of the linear raised tract (median raphe canal): homogeneous, whitish pattern

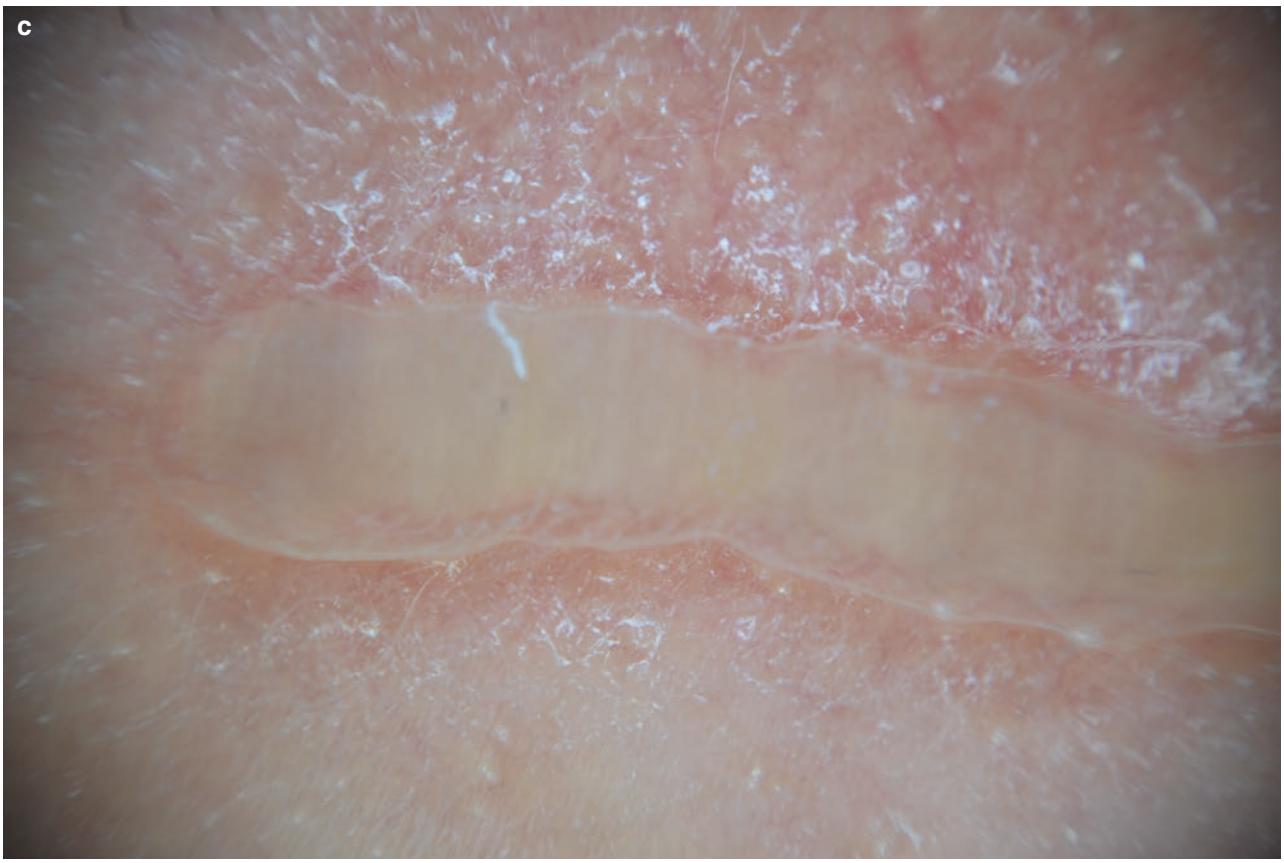

Fig. 34.1 (continued)

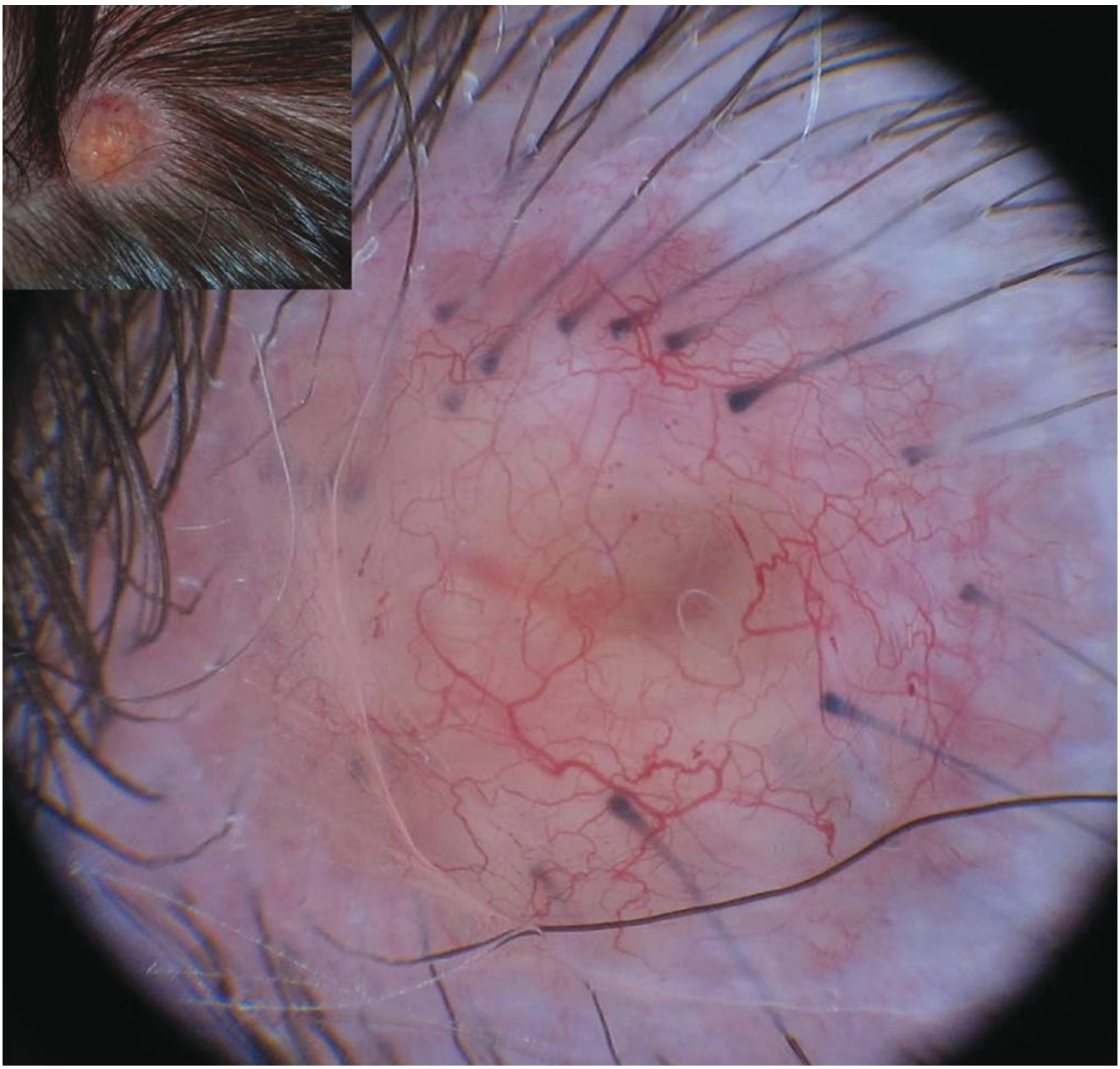

Fig. 34.2 Aplasia cutis congenita of the scalp. Dermatoscopy (×10) of a well-demarcated alopecic patch of the vertex (box) in a 9-year-old girl: central area of scarring alopecia with no follicular openings and telangiectatic vessels. At the hair-bearing margin, hair bulbs that are visible through a semitranslucent epidermis are horizontally and radially oriented (*starburst hair follicles pattern*) (Courtesy of Iria Neri, MD)

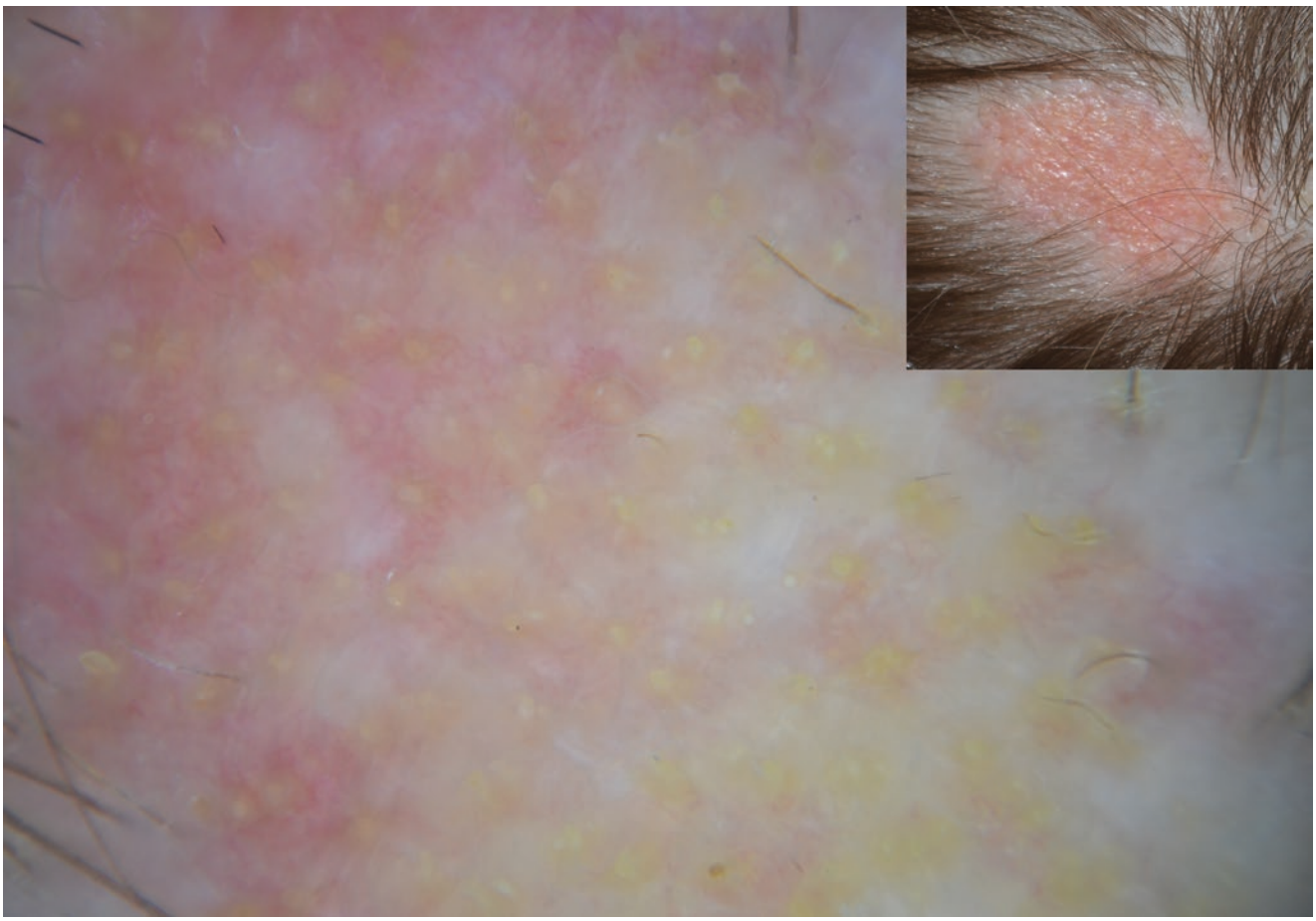

Fig. 34.3 Sebaceous nevus. Smooth yellowish patch of hair loss of the vertex in a 9-year-old girl (insert). Dermatoscopy (×10): bright yellow dots not associated with hair follicles

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