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Case Report

Spinular follicular keratosis of Siemens associated with wooly hair in two sisters: Trichoscopic description and anatomopathological correlation

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ABSTRACT

Spinular follicular keratosis (SFK) is a rare X-linked inherited disorder of keratinization characterized by diffuse follicular hyperkeratosis and progressive scarring alopecia of the scalp, eyebrows, and eyelashes. Trichoscopy is a non-invasive dermatological tool that has improved the differential diagnosis of hair pathologies. In fact, dermoscopic models have been developed for several entities of scalp diseases, allowing early diagnostic confirmation and rapid action without the need for histopathology, as well as good follow-up for patients and signs of pathological activity. To date, the specific trichoscopic signs of KFSD have not been developed. Herein, we report the case of KFSD of Siemens in two sisters associated with wooly hair. We employed trichoscopy as a highly useful tool for diagnosis, then performed an anatomopathological study to confirm the diagnosis and explain the signs found on trichoscopy.

Key words: Spinular follicular keratosis; Trichoscopy; Anatomopathology; Wooly hair; Isotretinoin

INTRODUCTION

Spinular follicular keratosis (SFK) is a rare X-linked hereditary disease. It is a keratinization abnormality involving the hair follicle, characterized by progressive scarring of the scalp, eyebrows, and eyelashes associated with skin involvement. Other organ involvement has been reported, such as ocular involvement [1]. Approx. fifty cases have been reported in the literature. Although Siemens syndrome is highly rare and its clinical manifestations are peculiar, anatomopathological confirmation remains mandatory. Trichoscopy is a non-invasive dermatological tool that has improved the differential diagnosis of hair pathologies. In fact, dermoscopic models have been developed for several entities of scalp diseases, allowing early diagnostic confirmation and rapid action without the need for histopathology, as well as good follow-up for patients and signs of pathological activity. To date, the specific trichoscopic signs of KFSD have not been developed.

Herein, we describe a case of KFSD proven by pathology in a patient in whom trichoscopy served an important role in helping to achieve a differential diagnosis with respect to non-scarring alopecia, showing the typical signs of scarring alopecia and selecting the most appropriate place to perform a scalp biopsy.

CASE REPORT

This case was a six-year-old female from a second-degree consanguineous marriage who presented with progressive alopecia and generalized asymptomatic skin lesions present since birth. The prenatal and postnatal histories were normal. Her younger, four-year-old sister presented the same symptomatology, involving scalp

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hair loss without eyebrow involvement, yet at a less advanced stage (Figs. 1a and 1b).

A scalp examination revealed obvious hypotrichosis composed of a diffuse rarefaction of the density of scalp hair, follicular prominence, short, thinned hair giving an aspect of uncombable wooly hair (Figs. 1b and 1c). There was also hypotrichosis, follicular prominence, and scaling of the eyelashes (Fig. 1d). The traction sign was negative in the eyebrows, lashes, and scalp. The trunk, arms, and thighs showed extensive, tiny follicular keratotic papules of normal skin color, typical of keratosis pilaris (Figs. 2a and 2b). A dermoscopic examination revealed an inflammatory background, dilated white spots, diffuse disappearance of the pilar orifices, with localized, homogeneous peri-pilar pigmentation (Fig. 2c). The remaining hair holes gave rise to highly fine, scattered hairs, with the emergence of a single hair per hole, leukotrichial hairs, and broken or bent hairs. The nails, teeth, and mucous membranes were normal. She also had bilateral fissural plantar hyperkeratosis, predominantly next to the calcaneum (Fig. 2d). A skin biopsy of the scalp showed follicular hyperkeratosis with follicular ostia dilated by keratotic plugs protruding on the surface and a significant reduction in the number of hair follicles. The remaining hair follicles had atrophic hair sheaths, with their superficial parts surrounded by lymphocytic lichenoid inflammatory infiltrate with vacuolation of the basal cells of the hair sheaths. The sebaceous glands were hypoplastic (Figs. 3a and 3b). This appearance was in favor of keratosis follicularis spinulosa decalvans of Siemens. The patient was initiated on isotretinoin with a significant improvement of the keratosis pilaris and the disappearance of the plantar keratoderma two months later and the attenuation of the inflammation.

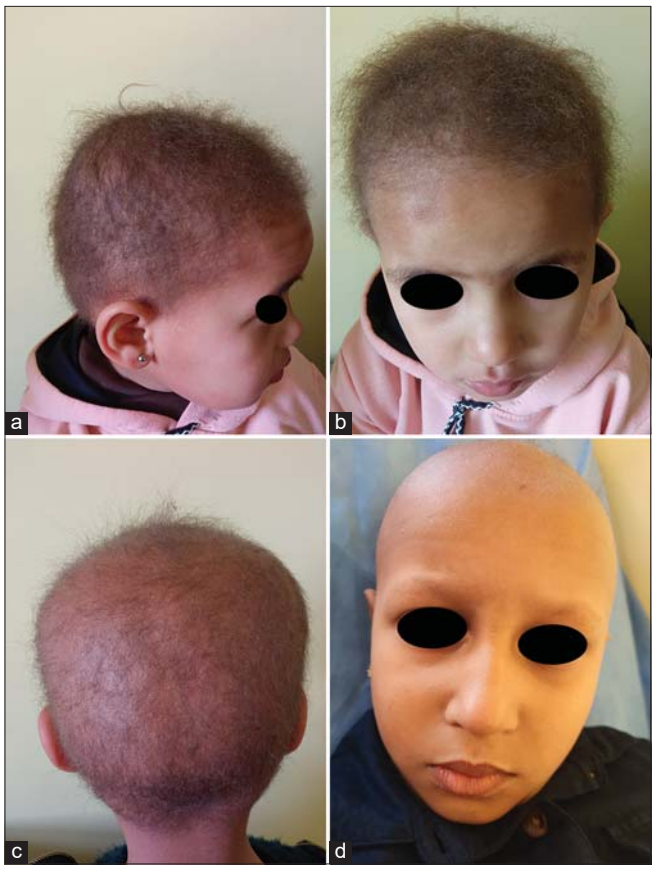

Figure 1: (a and b) Photos of the sister with diffuse hypotrichosis with follicular prominence of the scalp, without the involvement of the eyebrows. (c) Obvious hypotrichosis consisting of a diffuse rarefaction of the density of scalp hair, follicular prominence, short, thinned hair giving an aspect of uncombable woolly hair. (d) Hypotrichosis, follicular prominence, and scaling of the eyelashes.

DISCUSSION

Spinular follicular keratosis of Siemens (KFSD) is a rare genodermatosis with an autosomal dominant X-linked mode of inheritance. The condition was first described by Macleod, yet the term KFSD was employed by Siemens in 1926. The gene was mapped to Xp21.2-p22.2. Sporadic cases have also been described [1].

The main clinical features of the disease are diffuse cutaneous follicular hyperkeratosis and progressive scarring alopecia of the scalp, eyebrows, and eyelashes. The alopecia may be patchy or generalized. The less frequently reported features are atopy, palmoplantar hyperkeratosis with the predominance of the calcaneal region, clumpy hair folliculitis or wooly hair among hair abnormalities, and the unusual sign of high cuticles (or long cuticles) [2].

Severe pruritus may be present and may be related to increased levels of substance P, which has been reported in numerous inflammatory skin conditions, including alopecia, atopic dermatitis, and psoriasis [2].

Ocular abnormalities, such as photophobia, blepharitis, keratitis, corneal dystrophy, conjunctivitis, congenital glaucoma, and lenticular cataract, have been described in several patients with KFSD [3].

Nowadays, dermoscopy is an essential tool for the dermatologist to more effectively reconcile the diagnosis of several pathologies with clinical similarities, especially hair pathologies and alopecia. Since the trichoscopic model of KFSD has never been developed before, the role of trichoscopy is not diagnostic for KFSD, yet it is useful, at the beginning, to differentiate

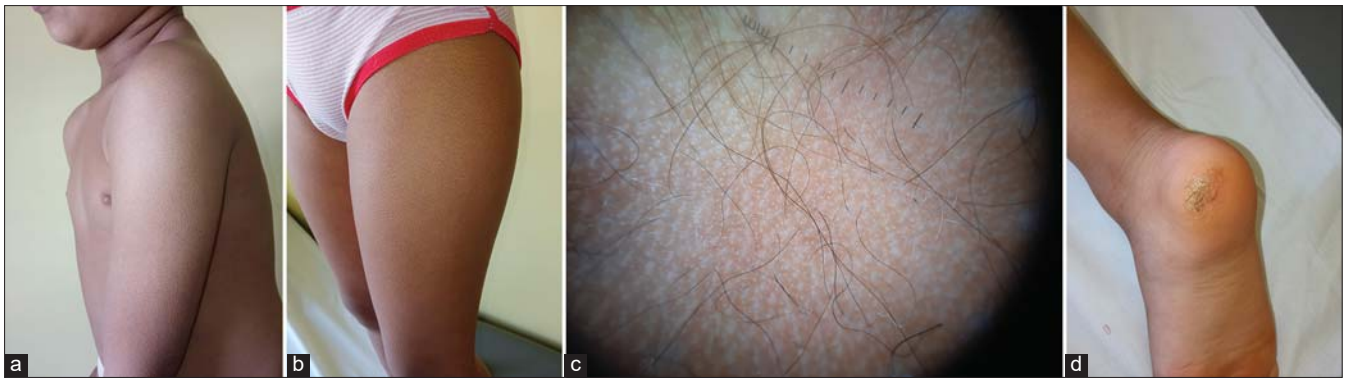

Figure 2: (a) Keratosis pilaris of the trunk and arms. (b) Keratosis pilaris of the thighs. (c) Trichoscopy showing ilated white spots, diffuse disappearance of the pilar orifices, the remaining hair holes giving rise to highly fine, scattered hairs with the emergence of a single hair per hole, leukotrychial hairs, and broken or bent hairs. (d) Fissural plantar hyperkeratosis of the calcaneum.

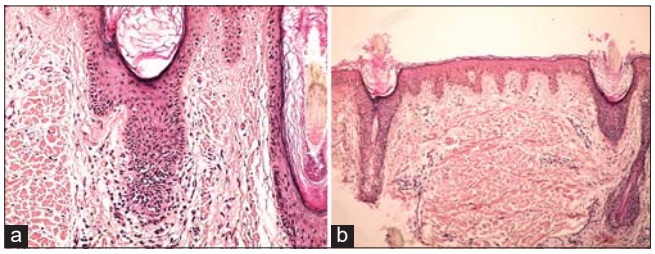

Figure 3: (a) Keratotic plugs protruding above the dilated follicular ostia, rarefaction of the hair follicles, and atrophic hair sheaths. (b) Slight lymphocytic infiltrate around an atrophic hair sheath and loose, concentric fibrosis.

between scarring alopecia and non-scarring alopecia. In 2020, Alessandrini et al. suggested the first trichoscopic description in a 26-year-old female, in whom clumpy hair, dystrophic hair, yellow spots, and follicular ostium loss were noted [4]. In our case, trichoscopy suggested the diagnosis, with the discovery of dystrophic hair composed of broken or bent hair, leukotrychial hair, loss of follicular ostia homogeneously scattered over the entire scalp and eyebrows, an erythematous background testifying dermal inflammation, and the presence of yellow dots expressing ostial dilatation with the secretion of the sebaceous glands.

The evaluation of hair loss is a diagnostic challenge for both the dermatologist and the pathologist. KFSD is a rare type of primary scarring alopecia with lymphocytic predominance. A good clinicopathological correlation is quite essential [4].

We decided to perform a biopsy to differentiate the disease from other scarring types of alopecia. Trichoscopy is crucial to select the most appropriate location for the scalp biopsy, as in other hair disorders.

Histopathologically, in advanced KFSD, the alopecia is lymphocytic [4]. However, in the early stages, the infiltrate is more neutrophilic [5]. A keratinization defect is usually observed, resulting in hypergranulosis and compact hyperkeratosis in the upper part of the infundibulum [5], which correlate clinically with follicular plugs. In the next phase of acute inflammation, spongiosis appears with a neutrophilic infiltrate in the infundibulum and the adjacent epidermis. The evolution includes the appearance of a lymphocytic infiltrate associated with perifollicular fibrosis, predominantly in the upper part of the follicle. In the terminal stages, fibrosis is observed with the presence of foreign-body granulomas surrounding the hair shafts [6].

In our case, the follicular ostia dilated by keratotic plugs protruding on the surface corresponded to keratotic papules and yellow dots on dermoscopy. The atrophic follicular sheaths surrounded in their superficial part by a lymphocytic inflammatory infiltrate in slightly lichenoid places with vacuolation of the basal cells of hair sheaths may explain both the hair dystrophy responsible for fine, brittle, wooly hair and the inflammatory background of the scalp. The loose, concentric fibrosis surrounding the hair follicles explains the white spots found on dermoscopy, representing fibrosed follicles in the terminal stage unable to produce hair.

Evidence for the treatment of keratosis follicularis spinulosa decalvans is anecdotal. Keratolytic agents and topical emollients only offer a symptomatic improvement in skin texture [6]. A combination of topical and/or intralesional corticosteroids may be employed for symptomatic relief [6]. Antibiotics such as tetracyclines, sulfonamides, macrolides, penicillins, and rifampin may be required during pustular flare-ups of the disease. Etretinol and isotretinoin may be useful,

yet have been reported to produce variable results. Oral retinoids are effective in the early phase of the disease when an active perifollicular infiltrate is present and should be continued for six to twelve months for an optimal response. Retinoids decrease epidermal proliferation and cytokine production, thereby reducing hyperkeratosis and inflammation [7].

Non-Q-switched, long-pulse ruby laser may be useful in recalcitrant KSFD.

Cases of end-stage KSFD treated with 800 nm diode laser have been reported with a striking response, with skin smoothing and the complete resolution of inflammation without the recurrence of symptoms within 2.5-year follow-up [7].

Our patient was treated with oral isotretinoin with smoothing of the skin surface and scalp and the disappearance of hon plantar hyperkeratosis over three months. Emollient creams based on 10% urea were combined with the oral treatment.

CONCLUSION

KFSD is a rare genodermatosis of the X chromosome. It consists of follicular hyperkeratosis of the skin, scarring alopecia of the scalp, the absence of the eyebrows, and corneal degeneration. Clinical and genetic heterogeneity has been described, yet the trichoscopic characteristics of this pathology have never been elaborated. The gold standard for diagnosis remains histopathology, yet trichoscopy may be highly useful for scalp biopsy site selection and patient follow-up. Through our clinical case, a novel trichoscopic description was established, as well as a correlation between dermoscopic signs and histopathology. Analytical studies of large series and novel cases are necessary to unravel the trichoscopic signs of this entity for more effective management and a more favorable aesthetic prognosis.

Consent

The examination of the patient was conducted according to the principles of the Declaration of Helsinki.

The authors certify that they have obtained all appropriate patient consent forms, in which the patients gave their consent for images and other clinical information to be included in the journal. The patients understand that their names and initials will not be published and due effort will be made to conceal their identity, but that anonymity cannot be guaranteed.

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