KERATOSIS FOLLICULARIS SPINULOSA DECALVANS: REPORT OF A CASE AND LITERATURE REVIEW

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Abstract - Keratosis follicularis spinulosa decalvans (KFSD) represents a rare, probably X-linked recessive genodermatosis, characterized by keratosis pilaris of face, trunk and extremities, followed by atrophy, cicatricial alopecia of the scalp, eyebrows and eyelashes, photophobia and corneal abnormalities. We report a rare case of KFSD and review the literature.

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Key Words: *Keratosis pilaris atrophicans, keratosis follicularis spinulosa decalvans, ulerythema ophryogenes, atrophoderma vermiculara, ichthyosis follicularis with photophobia and alopecia, keratitis, ichthyosis, deafness (KID) syndrome*

INTRODUCTION

There is a group of related disorders that are characterized by inflammatory, keratotic, follicular papules and later by atrophy. These rare diseases can be grouped under the encompassing term "keratosis pilaris atrophicans" (KPA) (1,4).

Differences in the degree of inflammation and atrophy as well as location have been used to categorize these syndromes. Many confusing terms are scattered throughout the literature, but they probably represent different stages of a single process. The accepted categories include: keratosis pilaris atrophicans faciei (KPAF), atrophoderma vermiculata (AV) and keratosis follicularis spinulosa decalvans (KFSD).

The three features that distinguish these three disorders from many other cutaneous diseases that involve the follicles are: onset in infancy, follicular hyperkeratosis, and scarring (1,4). Other entities that have features similar to KFSD are "ichthyosis follicularis with alopecia and photophobia" (IF), and KID syndromes (5).

CASE REPORT

A 22 year - old Iranian man was referred to Tehran Razi Hospital in August 1994 with pruritic scaly skin lesions, progressive loss of scalp hair and visual loss. He had developed horny papules on the face and other parts of the body during infancy, with progressive loss of scalp, eyebrow, eyelash and body hair, diffuse pruritic scaly lesions and photophobia during childhood and adolescence. His parents and siblings (4 brothers and 2 sisters) were all healthy. No other member of his family had similar skin problems. In May 1987, when he was 16 years old, he was admitted to Razi Hospital with multiple hyperkeratotic, infected and crusted lesions of the face and genitalia and numerous hyperkeratotic papules and plaques on the abdomen, chest and limbs. At that time, other significant physical findings included diffuse scalp and body hair loss, absence of eyelashes and scanty eyebrows, hyperkeratosis of knees, elbows, palms and soles; subungual hyperkeratosis; congestion and keratitis of both eyes; and decreased visual and auditory acuities. Biopsies of different hyperkeratotic, scaly and vegetating lesions were performed with the impression of epidermolytic hyperkeratosis, psoriasis or pemphigus vegetans. The successive histopathological examinations were suggestive but not completely compatible with this diagnosis. The patient received etretinate (Tigason) and

then prednisolone, with partial improvement. He also received systemic antibiotics and ketoconazole to control secondary bacterial infection and candidiasis.

The following laboratory study values were either negative or normal: CBC, liver function tests, urinalysis, BUN, creatinine, the percentage of B and T lymphocytes, serum IgG, IgA levels, serum C3, C4 and CH50, and response of cells to phytohemagglutinin (PHA). Ophthalmologic examination showed dry eyes with severe corneal neovascularization and pannus formation, making corneal graft impossible. ENT examination revealed perforation of the right tympanic membrane due to chronic otitis media, and thickly compacted cerumen in the left ear canal. Mastoid radiography showed changes compatible with chronic mastoiditis. The patient was discharged with the presumptive diagnosis of ichthyosis, complicated by candidiasis. In August 1994, he was admitted again with the following findings; diffuse cicatricial alopecia more evident at the vertex (Fig. 1) and frontal areas of scalp (Fig. 2), dry hair with follicular hyperkeratosis, total loss of eyebrows and eyelashes, thick hyperkeratotic and confluent papules on the face (Fig. 2), hyperkeratotic lesions of the trunk and limbs with yellow white scales, hyperkeratotic and vegetating lesions of limbs, especially the posterior part of the legs with oozing and a foul odor (Fig. 3),

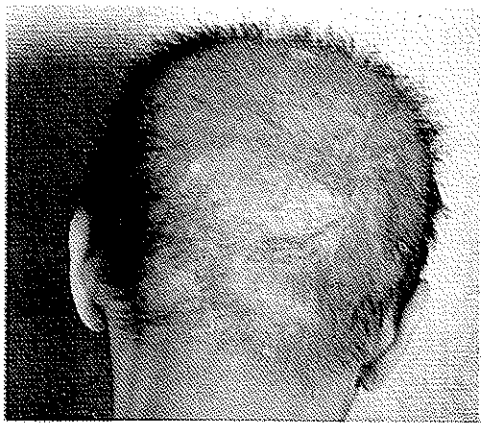

Fig. 1. Diffuse cicatrical alopecia at the vertex

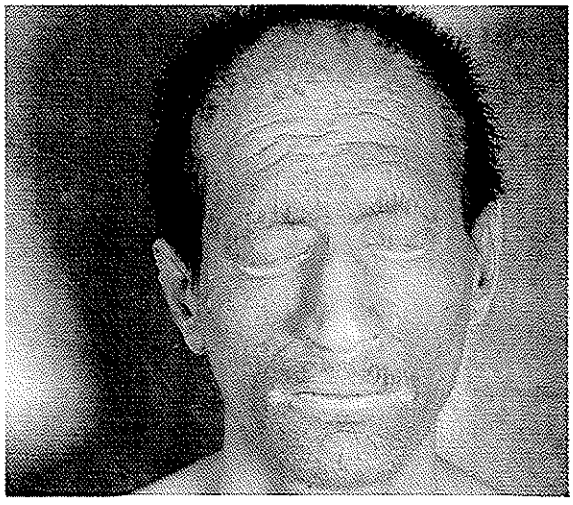

Fig. 2. Diffuse cicatrical alopecia at the frontal area

hyperkeratotic palms and soles, subungual hyperkeratosis, onycholysis and nail dystrophy.

Involvement of the upper limbs and back area were less prominent. There were scars and atrophic lesions on the trunk and buttocks, from previous hyperkeratotic lesions (Fig. 4). Ophthalmologic examination showed complete opacity (Fig. 2) and severe vascularization of both eyes, no light perception, and bilateral pendular nystagmus. ENT examination revealed chronic otitis media of the right ear, and audiometry was compatible with conductive hearing loss. The teeth and oral mucosal membranes were normal. Other physical examinations and all laboratory studies were within normal limits. Microscopic hair shaft examination was normal. The biopsy specimen from trunk and arm showed severe lamellate orthokeratosis, moderate hypergranulosis, slight spotty parakeratosis and irregular acanthosis of the epidermis. An inflammatory reaction was evident by the presence of lymphocytes, PMNs, and a few eosinophils in the dermis. In PAS staining, superficial fungal (mycelial) elements were observed in the trunk specimen.

Biopsy specimen of the scalp revealed mild thickening of the epidermis and hyperkeratosis-parakeratosis mostly in the infundibular portion of the hair follicles. There was also diffuse fibrosis and marked atrophy of

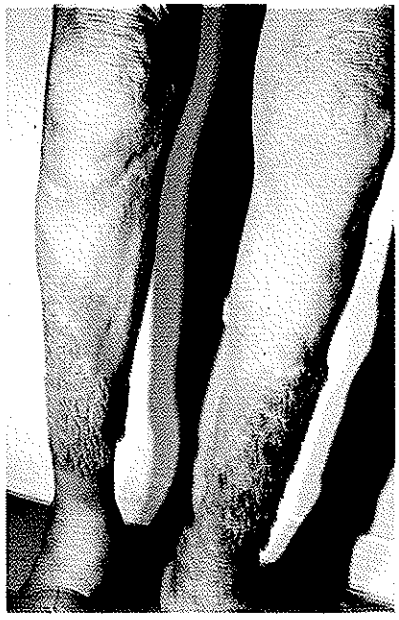

Fig. 3. Hyperkeratotic and vegetating lesions.

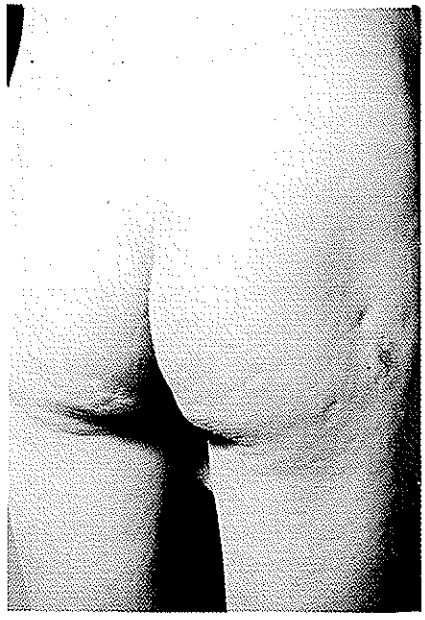

Fig. 4. Back area of case 4

the hair follicles in the dermis.

The patient received etretinate (1 mg/kg/day) for 12 weeks with the diagnosis of keratosis follicularis spinulosa decalvans (KFSD) and improved partially.

DISCUSSION

In all of the syndromes categorized as keratosis pilaris atrophicans (KPA), the primary defect is abnormal keratinization in and around the pilo-sebaceous follicle (1,4). When keratosis pilaris is followed by atrophy one speaks of KPS. Inflammation may or may not precede the atrophy. Familial incidence has been documented in all types of KPA. These syndromes should therefore be considered as genodermatoses. The classification is based on inflammation, atrophy and location: there is exclusive involvement of the follicles in keratosis pilaris atophicans faciei (KPAF), ulerythema ophryogenes, (UO) and KFSD and involvement of the epidermis and dermis in atrophoderma vermiculata (AV), resulting in pit-like depressions (1,4).

KFSD is a syndrome consisting of diffuse keratosis pilaris associated with scarring alopecia of the scalp (1,2). Other features may include: atrophy, hyperkeratosis of the palms and soles, photophobia and corneal abnormalities. The latter may result from primary affection of the cornea, or may be due to keratotic spines of the eyelids or hardened secretions of the meibomian glands. Corneal biopsy shows scattered subepithelial opacities (15) in the Bowman's membrane or its total absence (7,14). The end result may lead to corneal opacification and vascularization. The most severe manifestations are found in males (1,4), and X-linked recessive inheritance has been proposed (3,5). Females tend to be less severely affected than males (3,12).

DNA linkage analysis has shown significant linkage to X p 21.2-p 22.2. Other reports indicated genetic heterogeneity in KFSD (16). Several sporadic cases are also reported (9).

KPAF (and ulerythema ophryogenes) refers to an inflammatory process that begins in the lateral third of the eyebrows, with an onset of a few months after birth (4) resulting in hair loss of the involved follicles (UO). The process subsequently involves the cheeks and forehead (KPAF) (4). There is no photophobia or scarring alopecia of scalp (5). Association of PAF with Noonan's syndrome (10) and woolly hair (11) have been described.

AV begins between ages of 5 and 12, often symmetrically in the periauricular region of the cheeks. Erythema and pinhead follicular plugs are soon followed by reticulate atrophy as the plugs are shed ("worm-eaten" appearance) (3). Photophobia, alopecia, and follicular papules are absent (4). The two other entities most similar to KFSD are KID syndrome and "ichthyosis follicularis with alopecia and photophobia" (IF) (5). Neither IF nor KFSD are real inflammatory processes, but KFSD has as a late feature, atrophy and scarring of the follicles; whereas IV does not. Also, in KFSD the alopecia may be quite minimal and patchy, but in IF it is extensive or total. Hyperkeratosis of palms and soles is present in the original reports of KFSD, but is not a feature of IF. Follicular hyperkeratosis is seen histology of both IF sebaceous glands are atrophic or absent (5).

Deafness with keratitis and ichthyosis constitute the triad of features that define KID syndrome. It is a specific oculocutaneous syndrome, characterized by a distinctive congenital ichthyosis with generalized fine dry scaling, follicular keratotic spines, and a reticulated pattern of palmoplantar hyperkeratosis; a vascularizing keratitis that begins in early childhood and results in severe visual impairment; and congenital nonprogressive neurosensory deafness (13). There may be some other associated features including dystrophic nails, absent or scanty scalp, eyebrow and eyelash hair, and increased susceptibility to cutaeneous infections (5). An autosomal dominant mode of inheritance is probable (17).

The typical cutaneous abnormality of papillomatous hyperkeratosis which gives the skin a grainy, rugose, spiculated appearance, together with neurosensory hearing defect, which is a necessary element for diagnosis (17), and other associations, differentiate KID syndrome from KFSD. The scalp alopecia of KID syndrome may (1) or may not (17) accur. A variety of agents including systemic and topical antibiotics, corticosteroids, keratolytics, topical tretinoin, oral isotretinoin and etretinate are used in the treatment of patients of KPA and KFSD, but response to therapy has been limited and unsatisfactory (1,6).

This case of keratosis follicularis spinulosa decalvans, presented with a full picture of the disease: widespread keratosis pilaris, followed by atrophy, cicatricial alopecia, photophobia and bilateral corneal opacity resulting in blindness. An exact search and history taking is mandatory to find the pattern of heredity in this genodermatosis. In our reported case, family history for similar illness was negative.

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