KERATOSIS FOLLICULARIS SPINULOSA DECALVANS

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The case findings in a 22-year-old male patient of keratosis follicularis spinulosa decalvans are described. In addition to the characteristic cutaneous, occular and histological features, he had striking angular stomatitis and fissuring of the tongue simulating vitamin B-complex deficiency. This is an unreported feature to our knowledge. The mode of inheritance suggested X-linked trait.

Key Words: Keratosis follicularis spinulosa decalvans, X-linked trait

Introduction

Keratosis follicularis spinulosa decalvans (KFSD) is a rare hereditary disorder of keratinization characterized by the involvement of skin and eyes. The prominent cutaneous features include generalized spiny follicular hyperkeratosis resulting in wide spread scarring alopecia especially in the region of the scalp, face and neck.[1] Loss of eyebrows and eyelashes, inflammation of the cornea and conjunctiva with photophobia are the main occular manifestations.[2] Siemens[3] in 1925 first reported this entity in a Bavarian family and also assigned its present name. Since then occasional reports of this disorder have been published in literature from time to time.[4,5] We describe this rare entity in an young male patient with some unusual findings.

Care Report

A 22-year-old man presented with the complaints of generalized follicular skin eruptions since his second year of life , starting initially over the legs and gradually extending all over the body. The lesions were spiny to start with and some of them especially over the scalp turned pustular which ruptured spontaneously resulting in scarring alopecia. At the age of 10 years, the patient lost most of the scalp hair. Subsequently in a span of few years, he lost his axillary and pubic hair totally. Around this period similar process occurred over the eyebrows and eyelashes. He also had redness and watering of both eyes with photophobia. The patient was fourth child of non-consanguinous parents and born after a full-term normal delivery. First child is a female maintaining good health. Second and third were male who developed similar skin lesions at the age of 1 year and deceased after 3 years, because of chronic infections.

Examination revealed small, discrete, spiny papules with follicular plugging present on a normal looking skin all over the body. In the natal cleft and pubic region, the lesions were grouped to form plaques with occasional pustules. Cicatricial alopecia was noted all over the scalp (Fig. 1), face, axilla, groin and extremities. Hyperkeratosis with fissuring was seen over the palms and soles. Nails were normal. Redness of the conjuntiva was seen in both eyes with prominent loss of eyelashes and eyebrows. Chronic meibominitis of all eyelids and ulcerative blepheritis ever the upper eyelids were prominent. Cornea and anterior chamber were normal. Schirmer's test for tear secretion was normal in both eyes. Oral mucosa showed angular stomatitis. Tongue

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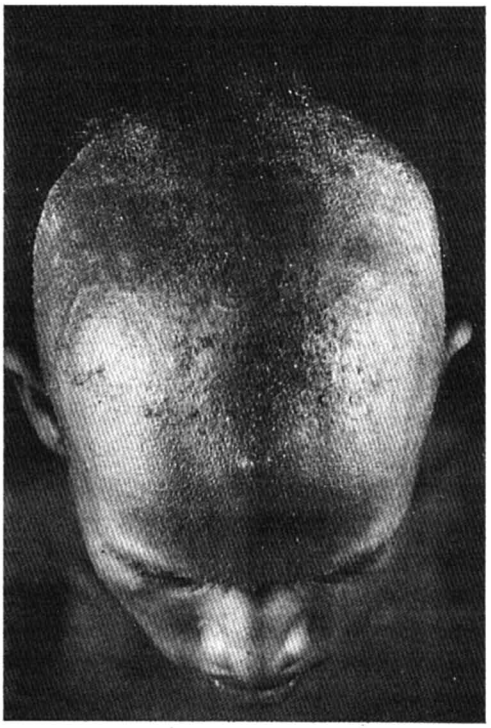

Fig. 1. Close up view of the scalp showing follicular keratotic papules and cicatricial alopecia.

was inflammed with longitudinal fissures (Fig. 2). Rest of the systemic examination was normal but for the tender lymphadenopathy over the occipital, submandibular and cervical regions.

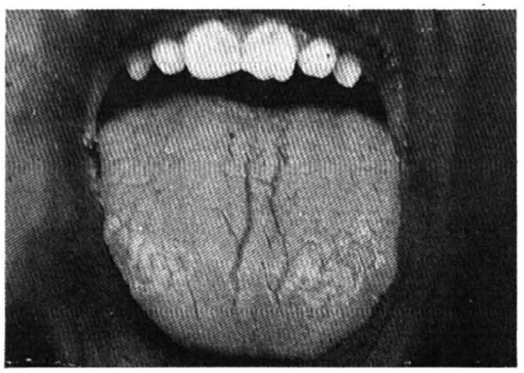

Fig. 1. Close up view of tongue showing fissuring.

Routine haematological and other laboratory studies were normal. Swab for candida from the tongue and angular cheilitis was negative on microscopic examination. Pus culture from the skin lesions showed *S. aureus*. Histopathological examination of skin biopsies obtained from the trunk and extremities showed prominent hyperkeratosis and follicular plugging with focal parakeratosis. Mild non-specific inflammatory infiltrate was seen in the dermis. Hair follicles showed absence of normally formed hair. Biopsy of the oral lesions could not be obtained because of non-compliance of the patient. Based on these clinical and histological findings the patient was diagnosed as a case of KFSD and the oral lesions were thought to be due to vitamin B-complex deficiency. However intensive treatment with injectable B-complex (2cc IM once daily) for one month and topical application of gentian violet 1% paint resulted in little improvement of his oral lesions.

Discussion

Our patient had characteristic cutaneous, occular and histological features of KFSD. However. We were intrigued to note the striking abnormalities in the oral mucosa in the form of angular stomatitis, redness and fissuring or the tongue simulating vitamin B-complex deficiency. The later condition was excluded by the fact that the lesions persisted despite intensive treatment with injectable vitamin B-complex. Hence, we believe that these features represent KFSD that have not been recorded in literature to our knowledge.

KFSD is thought to be an inherited X-linked disorder with distinct manifestations in males and apparently sparing the females.[1] This is true in our patient also, where the disease manifested in all the male siblings, out of which two died at an early age because of recurrent infections and totally spared the

female child who enjoys good health. Britton[4] also described an isolated case of KFSD with failure to thrive and recurrent infections. Less commonly associated abnormalities with this disorder include deafness, physical and mental retardation and hypoplastic nails.[5] These features were not seen in our patient. At present the treatment of this disorder is frustrating. Only symptomatic measures are available. Continous oral tetracycline (500 mg daily) was found to prevent hair loss and promote regrowth of hair in one case.[2] Systemic retinoids may be useful and deserve a trial.

References

1. Kukkanen K. Keratosis follicularis spinulosa decalvans in a family from Northern Finland. Acta Derm Venereol 1971; 51: 146-50.
2. Dawber RPR, Ebling FJG, Wojnarawska FT. Disorders of Hair. In : Textbook of Dermatology (Champion RH, Burton JL, Ebling FJG, eds). Oxford: Blackwell Scientific Publications, 1992; 2603-3604.
3. Siemens HW. Keratosis follicularis spinulosa declavans. Arch Derm Syph (Berlin) 1926; 151 : 384
4. Britton H, Lustigo J, Thompson BJ. Keratosis pilaris decalvans. An infant with failure to thrive, deafness and recurrent infections. Arch Dermatol 1975; 114: 761-4.
5. Eramo LR, Esterly NB, Zielseri ES. Ichthyosis follicularis with alopecia and photophobia Arch Dermatol 1975;121:1167-74.