JACKSON - LAWLER SYNDROME

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A 36-year-old woman presented with multiple yellowish cutaneous cysts of 5 years duration, over the scalp, trunk and upper limbs. She had pachyonychia, keratoderma of hands and feet, eyebrows which stood straight out and a single cafe-au-lait macule.

Key words : Jackson-Lawler syndrome, Pachyonychia congenita

Introduction

Jackson-Lawler syndrome or Schonfeld's type-2 pachyonychia congenita (PC-2) is an autosomal dominant disorder characterised by hypertrophic nail dystrophy, mild keratoderma of hands and feet, later development of multiple pilosebaceous cysts (steatocystoma multiplex) and other features of ectodermal dysplasia. There can be autosomal recessive inheritance and spontaneous mutations. This can be associated with neurofibromatosis, polyneuropathy,[1] tuberous sclerosis, mottled hyperpigmentation and amyloid deposits, Kyrle's disease, hidradenitis suppurativa and squamous cell carcinoma in previously ulcerated area. We report a case of this rare syndrome associated with cafe-au-lait macule and eye brow abnormalities.

Case Report

A 36-year-old woman presented with cluster of yellowish papules and nodules distributed on scalp, forehead, neck, trunk and upper limbs of five years duration.Their size varied from 0.5 to 3cm, and were mobile within the skin. On puncturing, yellowish creamy material was expressed from the lesions. Almost all nails of fingers and toes were wedge shaped. Nail plates were hard, lusterless and thickened. Thickening was mainly at the free border, and there was subungual hyperkeratosis. A case-au-lait macule of 4cm diameter was noticed on upper medial aspect of right arm. Her eye brows were thickened and stood straight out. She had moderate palmoplantar keratoderma. No bony lesions were detected by skiagrams. Other systems including gastrointestinal system were normal. No other family member was affected. All baseline investigations were normal. Histopathological examination of a nodule was consistent with steatocystoma multiplex.

Her keratoderma responded partially to 6% salicylic acid ointment. Larger nodules were excised and wounds repaired by plastic surgery.

Discussion

Jackson-Lawler syndrome (PC-2) is an autosomal dominant disrorder. But sporadic cases due to spontaneous mutations have been described. In our case there was no family history. In addition to the features of classical pachyonychia congenita type 1 (PC-1), PC-2 may have

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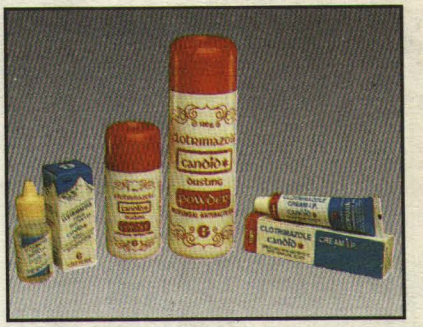

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neonatal teeth and multiple epidermoid follicular cysts. But in this case natal teeth was absent.

It has been shown that mutations in either keratin 16 (K-16) or keratin 6a (K6a) genes, which form a keratin expression pair; produce the PC-1 variant. They are expressed in mucosae, follicular keratinocytes and palmoplantar skin. Missense mutations in keratin 17 (K17) gene alone, an unpaired accessory keratin (a differentiation specific keratin), which are expressed in the pilosebaceous unit, nail bed and basal appendage keratinocytes with some basal expression in palmoplantar skin, results in PC-2. There have been identical mutations of K17 in steatocystoma multiplex, where predominantly sebaceous and keratin cysts reflects expression in sebaceous gland as well as hair follicle. Steatocystoma may therefore be considered a limited form of PC-2.[2] It has been reported that missense mutations in keratin 6b (K6b), a previously unknown expression partner of K17 analogous to K6a/K16 pair, produce a phenocopy of the K17 disease PC-2, revealing genetic heterogenicity in PC-2.[3]

It is a progressive condition. Emollients and keratolytics are usually prescribed for mild keratodermas. Our patient improved with salicylic acid (6%) ointment. Her cosmeic disfigurement improved with excision of cystic lesions. Acitretin (25-35mg/day) may produce a reasonable degree of flattening of nails if given for prolonged period.

References

1. Bensa AF, Dalac S, Beer F, et al. Congenital pachyonychia, neurofibromatosis and sensory-motor polyneuropathy. Ann Dermato Venereol 1995; 122: 428-431.

2. Smith FJD, Corden LD, Rugg EL, et al. Missense mutations in keratin 17 cause either pachyonychia congenita type 2 or a phenotype resembling steatocystoma multiplex. J Invest Dermatol 1997; 108: 220-223.

3. Smith FJD, Jonkman MF, VanGoor H, et al. A mutation in human keratin K6b produce a phenocopy of the K17 disorder pachyonychia congenita type 2. Hum Mol Genet 1998; 7 : 1143-1148.