Journal of Pakistan Association of Dermatologists 2005; **15**: 288-289.

PhotoDermDiagnosis

Sparse scalp hair and erythematous rash in a girl.

Section Editor

Amor Khachemoune, **MD CSW,**
Suny Down State Medical Center, Brooklyn, NY, USA

Asher Ahmed Mashhood

Department of Dermatology, Combined Military Hospital, Multan.

My patient a 6-years-old girl was born with scanty scalp hairs. As she grew old she developed light-coloured sparse, soft, uncombable hairs which broke down easily. The patients also gave history of itchy, erythematous lesions over her body since last 4 years. She had been developing infection in the lesions off and on. There was no history of any growth retardation, mental abnormality, teeth or nail defects. My patient was fourth among 4 sisters and 2 brothers. One of her elder brother, who was 10 years of age, was a patient of cerebral atrophy. Her parents were first cousins.

On examination the patient was a young girl of average build, well-oriented in time and space. Her vital signs were stable. There was no anaemia or jaundice. Rest of the general physical, systemic and mental state examination was unremarkable.

The examination of the scalp skin was unremarkable. The hair was; however, scanty, brownish, soft, dry and curly (**Figure 1**). The hair of eyebrows and lashes was apparently normal. On the examination of rest of the skin, there was a diffuse and at places discrete erythematous and scaly rash over the chest, back and upper limbs (**Figure 2**). There were several excoriation marks on the skin. The nails and teeth of the patient were completely normal.

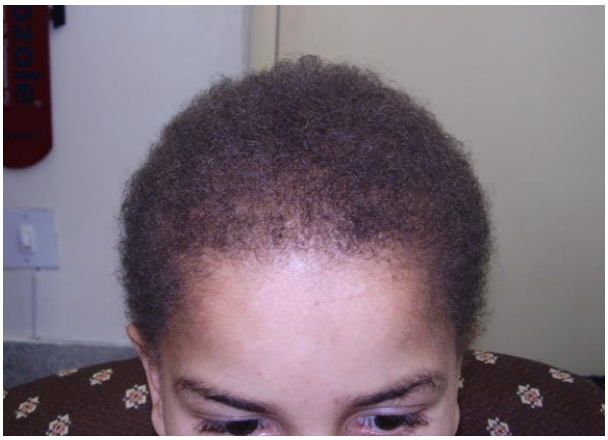

Figure 1

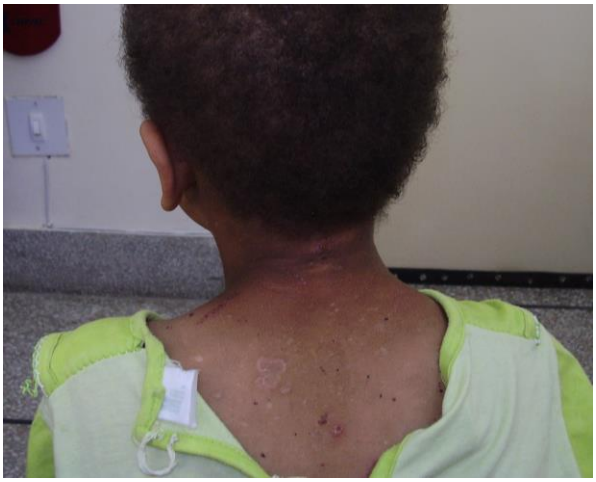

Figure 2

The microscopic examination of several hairs cut from the scalp, revealed nodular swellings over the shaft of a few hairs. The histopathological examination of the skin revealed mild hyperkeratosis, spongiosis and perivascular mononuclear cell infiltrate.

Diagnosis

Netherton's syndrome

Discussion

Netherton's syndrome[1] is an autosomal recessive condition of variable expressivity. It is common in girls. The syndrome is characterized by two features; bamboo-like nodes (trichorrhexis invaginata) on the delicate and fragile hairs and a scaly erythematous dermatitis (ichthyosis linearis circumflex). The underlying defect is localized to a gene on chromosome 5q32, known as SPINK5.[2] Examination of SPINK5 gene in 13 families revealed 11 mutations, 9 of which were associated with RNA instability.[3] The diagnosis is confirmed by light microscopic examination of several hairs cut from the scalp surface, as pulling of the hairs may cause breakage of the shaft. Detection of even a single node (trichorrhexis invaginata) in a single hair is diagnostic.[4]

The patient usually a girl presents primarily in young age with either complaints of fragile and sparse hairs or by an erythematous and scaly rash over the body since birth.[5]The hair defect is sometime so subtle that it has to be deliberately sought and sometimes the hairs are short, dry, lusterless and brittle and very apparently abnormal. The hairs of eye brows and lashes may also be less or absent. The skin rash mimics atopic dermatitis. There is generalized erythema or scaling since birth but the severity varies from patient to patient. On trunk, fine dry scales are associated with polycyclic and serpiginous outlines.[5]

Although the rash mimics atopic eczema the response to topical steroids is disappointing and may lead to more side effects.[4] Use of any bland emollients is usually sufficient. Topical tacrolimus 0.1% ointment is used with some success. Nagata described some response to phototherapy.[6] The use of systemic retinoids does not produce significant benefit.[5] As far as the hairs are concerned, the only advice is to avoid chemical and physical trauma.[4]

References

1. Netherton GW. A unique case of trichorrhexis nodosa: 'bamboo-hairs'. *Arch Dermatol* 1958; **78**: 482-90.
2. Chavanas S, Garner C, Bodemer C *et al*. Localization of Netherton syndrome gene to chromosome 5q32, by linkage analysis and homozygosity mapping. *Am J Hum Genet* 2000; **66**: 914-21.
3. Chavanas S, Bodemer C, Rochat A *et al*. Mutations in SPINK5, encoding a serine protease inhibitor, causes Netherton syndrome. *Nat Genet* 2000; **25**: 141-2.
4. de Berker DAR, Messenger AG, Sinclair RD. Disorders of Hair. In: Burns T, Breathnach S, Cox N, Griffith C, eds. *Rook's Textbook of Dermatology 7th edn.* Oxford:. Blackwell Science: 2004. p. 63.1-63.120.
5. Judge MR, Morgan G, Harper JL. A clinical and immunological study of Netherton's syndrome. *Br J Dermatol* 1994; **131**: 615-9.
6. Nagata T. Neterton's syndrome which responded to photochemotherapy. *Dermatologica* 1980; **161**: 51-60.

Journal of Pakistan Association of Dermatologists 2005; **15**: 288-289.