Sir,

A case of familial trichomegaly in association with oculocutaneous albinism type 1

A case of abnormally long eyelashes was first described by Reiter[1] in 1926, which he attributed to adrenal dysfunction as the patient also had excessive facial hair. Bab[2] in 1931 reported a second case, a 9-year-old girl who also had abnormally long eyelashes, but in the absence of other pathology. In 1944, Gray[3] described a third example of this condition in a 64-year-old man and assigned the term trichomegaly or 'movie lashes'.

We describe a case of familial trichomegaly with coexistent oculocutaneous albinism type 1.

Case Report

A 3-year-old boy with severe nystagmus, photophobia, and reduced visual acuity was referred to the paediatric eye clinic. On examination, he had pendular nystagmus and iris transillumination. Visual acuity was 6/60 in each eye, and cycloplegic refraction revealed a small hypermetropic astigmatic refractive error. Ocular media were clear and dilated fundoscopy revealed hypopigmented fundi with bilateral foveal hypoplasia. He also had complete absence of pigment in his hair and skin. These characteristic features led to the diagnosis of oculocutaneous albinism type 1. The boy's parents were second cousins, and both had normal ocular examination.

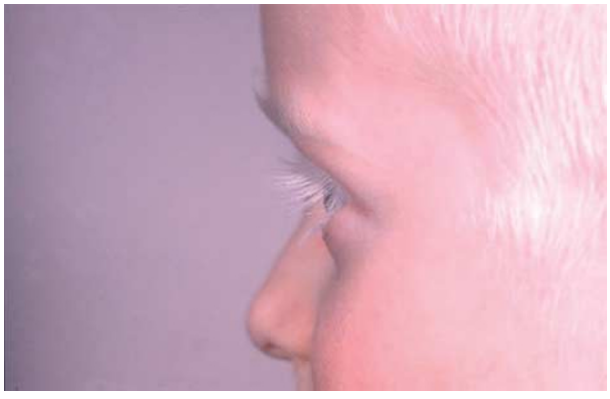

Figure 1 Oculocutaneous albinism with abnormally long eyelashes.

It was also noted at presentation that both the patient and his father had abnormally long eyelashes (Figure 1). On further questioning and construction of a pedigree, the high incidence of trichomegaly was noted in other family members on the paternal side, with a total of eight affected individuals (Figure 2). No other family members had oculocutaneous albinism. The family is of Indian origin.

Comment

Since its first description, trichomegaly has been associated with several different entities. Acquired forms of trichomegaly have been reported with many conditions but with no unifying aetiological explanation. These include malnutrition, use of various medications, acquired immunodeficiency syndrome, porphyria, anorexia nervosa, pregnancy, hypothyroidism, dermatomyositis, and pretibial myxoedema.[4,5]

Congenital trichomegaly has been reported as a part of many syndromes. Oliver–McFarlane syndrome describes congenital trichomegaly in association with pigmentary retinal degeneration, dwarfism, and mental retardation.[6] De Lange syndrome (also known as Cornelia de Lange or Brachmann de Lange syndrome) describes trichomegaly, synophrys, and a low hairline in association with

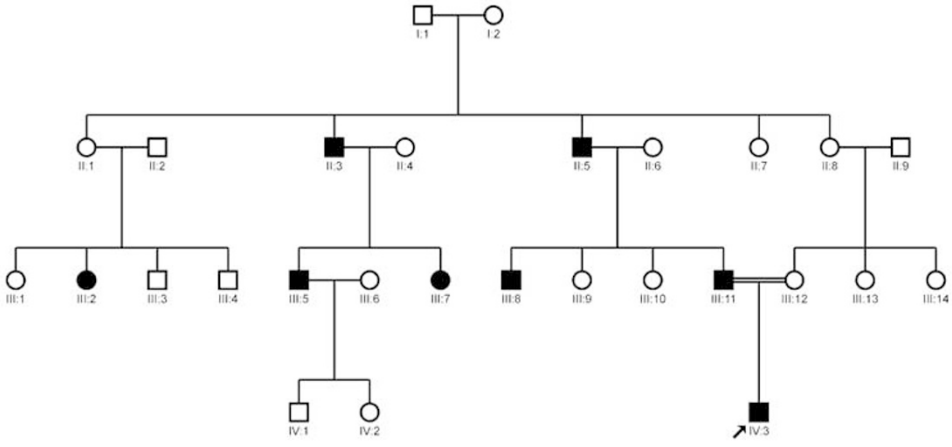

Figure 2 Pedigree of the family with trichomegaly. Black squares (men) and circles (women) indicate affected persons; white squares and circles unaffected persons; double lines, consanguineous marriages; single lines, nonconsanguineous marriages; and arrow, the index case.

developmental and musculoskeletal abnormalities.[7] Goldstein–Hutt syndrome describes congenital trichomegaly in association with cataract and hereditary spherocytosis.[8] Cone-rod dystrophy associated with congenital hypertrichosis, including trichomegaly, has also been reported.[9]

Interestingly, trichomegaly has recently been commonly identified in patients with Hermansky–Pudlak syndrome (HPS).[10] These patients, with HPS from northwest Puerto Rico, were found to be homozygous for a 16-base pair duplication in exon 15 of *HPS1*, a gene on chromosome 10q23 known to cause the disorder.[10] However, the subject we report here did not have a history of easy bruising, epistaxis, or prolonged bleeding following injury or surgery.

In 1997 Harrison *et al*[11] described what they believed was the first reported case of familial trichomegaly. This was manifest in three healthy siblings, but was not present in other generations. They felt that the consanguinity of two generations of first-cousin marriages played a causative role in the trichomegaly.

We report a case of trichomegaly with oculocutaneous albinism type 1. In view of the lack of co-segregation of these two phenotypes in this family, it seems that they are inherited as separate traits, with independent genetic loci. Nevertheless, to the best of our knowledge, this is the first report of familial trichomegaly with a documented pedigree showing the presence of the disorder in three generations. This is also the first report of trichomegaly in association with non-syndromic oculocutaneous albinism.

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Eye (2004) **18,** 863–864. doi:10.1038/sj.eye.6701326
Published online 30 January 2004