An autosomal dominant syndrome of acromegaloïd facial appearance and generalised hypertrichosis terminalis

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Abstract
We report a family in which a phenotype of acromegaloïd facial appearance (AFA) and generalised hypertrichosis terminalis segregates through three generations. Congenital hypertrichosis terminalis and AFA have been previously reported as independent autosomal dominant traits. This is the first report to delineate an autosomal dominant transmission of the combined phenotype.

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Key words: terminal hypertrichosis; acromegaloïd facies; autosomal dominant.

Case report
The proband (fig 1, III.2), a term product of healthy, non-consanguineous parents, was initially seen aged 2 years, when a dermatological opinion was sought regarding striking generalised terminal hypertrichosis (fig 2). The hypertrichosis was first noted shortly after birth and had become progressively more marked. There was a coarsening of the facial appearance with broadening of the nose and swelling of the lower lip (fig 3). The intraoral mucosa and the tongue were normal. General physical examination, including the genitalia, was normal; height and weight were just below the 97th centiles. Endocrine assessment, including mucopolysaccharide analysis, was normal and radiological survey was normal, with bone age commensurate with chronological age. There was no digital "tufting".

Family history
The pedigree is shown in fig 1. The syndrome is inherited as an autosomal dominant trait with both sexes affected and male to male transmission. One affected subject (fig 1, I.1) was not examined by the authors; the affected status was assigned following a collaborative history from the affected offspring. In generation II, the proband’s paternal aunt (fig 1, II.3) and father (fig 1, II.7) showed an acromegaloïd facial appearance (figs 4 and 5) and generalised terminal hypertrichosis. Facial hypertrichosis (fig 4) was seen in the paternal aunt. Physical examination was normal and the intraoral mucosa and the gingivae were normal in both patients. Both II.3 and II.7 were referred to the Endocrinology Department because of a clinical suspicion of acromegaly.

Figure 1 Family pedigree. *Personally examined by the authors.

Figure 2 Proband aged 2 years: marked generalised terminal hypertrichosis. (All photographs reproduced with permission.)
Acromegaloid facial appearance and generalised hypertrichosis terminalis

Figure 3  Proband aged 2 years: coarsening of facial features and thickening of the lower lip.

Figure 4  The paternal aunt (patient II.3, Fig 1): acromegaloid facial appearance and facial hypertrichosis.

Figure 5  The proband’s father (patient II.7, fig 1), aged 28: marked facial coarsening with “acromegaloid” appearance.

The endocrine opinion did not support this diagnosis; there were no typical changes of excess growth hormone secretion, such as enlargement of jaw or tongue, and hand volumes were normal. II.3 had initial investigations which showed persistently high levels of serum growth hormone on a number of tests (oral glucose tolerance, insulin hypoglycaemia), and she had been treated briefly on the basis of these results. Subsequent tests remained fully normal and no subsequent treatment has been given. The most recent measurement (1994) in II.3 are growth hormone 0.5 mU/l, IGF1 22.7 nmol/l, and in II.7 growth hormone 1.5 mU/l, IGF1 14.3 nmol/l, all of which are within a normal adult range. There was no evidence of insulin resistance, and serum insulin levels during oral glucose tolerance tests in both were normal. The father of the proband (fig 1, II.7) had a history of recurrent idiopathic cardiac effusions, necessitating pericardial fenestration. No other family member had pericardial effusions.

Discussion
This family has four affected members in three generations, confirming an autosomal dominant pattern of inheritance. The most striking features of the affected phenotype are an “acromegaloid” facial appearance and generalised hypertrichosis terminalis. All affected subjects had normal intraoral mucosa and gingivae. An acromegaloid facial appearance is associated with several autosomal dominant conditions, such as pachydermoperostosis and Ascher syndrome. Our family does not have any other clinical features suggestive of these disorders nor, to our knowledge, has hypertrichosis been a feature of these conditions. Our family has hypertrichosis and a coarsened facial appearance in common with the Gorlin–Chaudray–Moss syndrome (GCM). They lack, however, the characteristic stocky build, genital hypoplasia, phalangeal hypoplasia, conductive hearing loss, and midface hypoplasia considered characteristic of GCM. The acromegaloid facial appearance (AFA) syndrome (MIM No 102150) described by Hughes et
hyperplasia of variant and perplasia, The with gingival hyperplasia generalised hypertrichosis, of this condition may represent a new syndrome, discrete from AFA syndrome and congenital hypertrichosis/gingival hyperplasia.12

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