Cleft lip and palate, pili torti, malformed ears, partial syndactyly of fingers and toes, and mental retardation: a new syndrome?

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SUMMARY  Two sibs with a syndrome including cleft lip and palate, sparse scalp hair, malformed protruding ears, and partial syndactyly of the fingers and toes are reported. The older child also has mental retardation and pili torti. This syndrome is most probably inherited as an autosomal recessive disorder.

Cleft lip and palate or both is found in more than 150 syndromes, but many patients with multiple malformations including cleft lip/palate cannot at present be classified. Recently, we examined two sibs with a syndrome including cleft lip and palate, sparse scalp hair, malformed protruding ears, and partial syndactyly of the fingers and toes.

Case reports

The parents were first cousin Moslem Arabs. The two children reported here were born of the seventh and eighth pregnancies when the mother was 35 and 37 years old respectively and the father 37 and 39 years old. Both pregnancies were normal and the mother did not take any medication. Seven children, born before the two reported here had no malformations. There were two males and five females and one of them, a male twin of an unlike sexed pair, died suddenly in infancy.

The proband was first examined at the age of four months when referred for correction of his cleft lip. The child appeared retarded for his age and there was complete head lag. The head circumference was normal for age, the hair was sparse, the ears were protruding and malformed, and micrognathia and bilateral cleft lip and palate were evident (figs 1 and 2). There was partial syndactyly of the left third and fourth fingers and the third and fourth toes on both feet. A simian line was present on the left palm and a Sydney line on the right. The karyotype was normal 46,XY.

The mother became pregnant again at the age of 37 years but did not contact us. The child, a girl, was referred at the age of four months for correction of a cleft lip. The resemblance of the girl to the proband was striking (table). She had the same type of hair, malformed protruding ears, left cleft lip, and cleft palate. In both hands partial syndactyly of the third and fourth fingers as well as Sydney lines were noted. Partial syndactyly of the third and fourth toes was also present. The child died suddenly at the age...
circumference (10th centile) were normal. The inner and outer canthal distances were normal for age (80th centile) but relatively large for the head circumference. There was an antimongoloid slant to the eyes. The ears were protruding and malformed. An additional finding was widely spaced nipples (>97th centile) with a normal chest circumference (10th centile). The scrotum was hypoplastic and the testes were in the inguinal canal. A transverse crease was present on both palms, simian on the left and Sydney on the right. Partial syndactyly of the third and fourth fingers of the left hand and toes of both feet was also found. The hair was sparse and the outer part of the eyebrows was almost absent. The lashes were normal. On microscopical examination (fig 3) the hair was twisted and pili torti was diagnosed. The teeth were widely spaced and much caries was present. The skin and the nails were normal.

**Discussion**

Both children were affected with a similar syndrome. Their parents are first cousins and healthy.
A new syndrome

The karyotype of the boy was normal and therefore this syndrome is likely to be inherited as an autosomal recessive disorder. From a review of published reports on syndromes including cleft lip or cleft palate or both, we were unable to find a syndrome similar to the one described here.

Pili torti is characterised by twisted hairs and is often present with other ectodermal signs, mainly involvement of the teeth. Pili torti has been reported as an autosomal dominant entity or as part of many different syndromes.\(^2\) Wiedemann\(^3\) reported an inbred family where three out of five children presented with pili torti, sparse eyebrows and lashes, tooth anomalies including severe crown hypoplasia and delayed atypical eruption of permanent teeth, as well as thick yellowish nails. The skin was dry and hyperkeratosis was present. In addition, syndactyly of both fingers and toes to variable degrees was also present in the children as well as transverse creases on both palms. The palate was highly arched and growth and psychomotor development were normal. Many of the signs in the children reported by Wiedemann\(^3\) were similar to those found in the children in this report, including pili torti, syndactyly, and transverse crease on the palms, but other signs differ, including mental development, lip and palate involvement, and other ectodermal signs in nails, teeth, and skin. Both syndromes are probably inherited as autosomal recessive disorders.

Recognition of further affected children as well as follow up of the child presented here will help to delineate this syndrome better.

References


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