

and Charcot-Marie-Tooth disease. Scoliosis may also be seen secondary to neurofibromatosis. Neurofibromatosis is a tempting explanation for the scoliosis and eye findings in these two brothers, especially in the light of the mental retardation, nerve deafness, and seizure disorder in the older of the two brothers. There were no skin manifestations in the propositi and no family history to support this diagnosis.

X-linked recessive inheritance is a possible, but unlikely, explanation of the findings in these two brothers, in view of the absence of the condition in their mother's three brothers and her sisters' 12 sons.

The close parental consanguinity and the strikingly similar clinical presentation in these two brothers suggest that this is an autosomal recessive condition. The absence of scoliosis or gaze palsy in any of the remaining nine sibs suggests that the scoliosis and gaze palsy are expressions of the same single autosomal recessive gene.

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## Re-evaluation of CHANDS

**SUMMARY** A rare ectodermal dysplasia with the acronym CHANDS (Curly Hair, Ankyloblepharon, Nail Dysplasia Syndrome) was described by Baughman (1971) as being a new autosomal dominant condition. Additional pedigree data obtained after the original report indicate that the mode of inheritance is more likely to be autosomal recessive, with an instance of quasi-dominant transmission as a result of multiple consanguineous matings in the family. These data are provided in this report.

In 1971, Baughman reported a 'new' autosomal dominant trait characterised by curly hair, ankyloblepharon, and nail dysplasia, which was given the acronym CHANDS. The assumption that this trait was inherited as an autosomal dominant syndrome was based on a pedigree showing direct transmission from the mother to 3 of her 9 offspring. Both male and female children were affected, thus ruling out sex linkage of a recessive trait. In addition, ataxia was reported in 3 of the 9 sibs, including 2 of the CHANDS-affected children. The aetiology of the ataxia was not known at the time of the original report, but it was postulated that it could be part of the syndrome.

A more extensive pedigree has subsequently been obtained from this family, and it is the purpose of this report to present evidence that CHANDS is inherited as a recessive, and not as a dominant, trait.

#### Discussion

The original pedigree showed that V.1 and V.2 are first cousins and have 9 children. V.2 has 7 sibs and is herself the product of a consanguineous mating of second cousins. The family members exhibiting the CHANDS trait are V.3, 4, and 7 of her sibs, and 3 of her children (Fig.). However, when this family was re-evaluated in 1976, it was discovered that V.3 and V.4 have a total of 11 normal children, but none with the CHANDS trait. This was verified by examining recent photographs of all 11 children and noting that all have long, straight hair and normal nails. There is no historical evidence of any of the children having had ankyloblepharon at birth. The CHANDS trait is manifested by curly hair that apparently does not grow past shoulder length, dysplastic nails, and ankyloblepharon at birth. In addition, the ataxia has been shown to be ataxia-telangiectasia based on clinical, phenotypic, and immunological features present in the affected children. Therefore, the CHANDS trait can reasonably be assumed to be a separate entity from the ataxia.

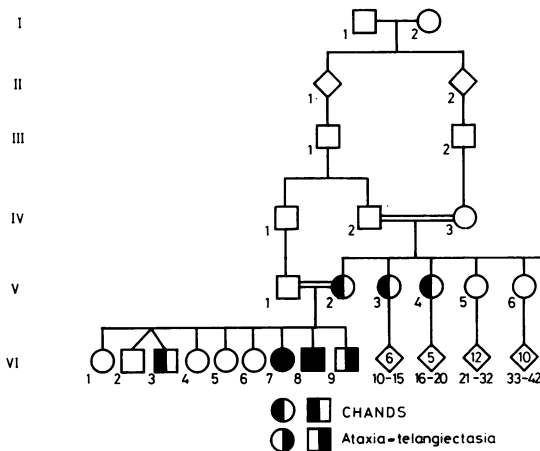


Fig. Pedigree of consanguineous family in which 2 traits, CHANDS and ataxia-telangiectasia, are segregating.

Given the more complete pedigree data, it is our belief that this trait is actually an autosomal recessive trait showing quasi-dominant inheritance in this family because of the multiple consanguinity. We have no means of verifying this hypothesis directly because of the extreme rarity of the gene. As far as we are aware, there are no other reported families with this constellation of defects. However, using a means of estimation in which the relative likelihoods of autosomal dominant versus recessive inheritance are weighed, it is discovered that autosomal recessive inheritance is by far the more likely. If we let  $P_1$  equal the probability that IV.2 and IV.3 are both carriers of the gene, given that the gene is an autosomal recessive, and  $P_2$  equal the probability that IV.2 or IV.3 carry the gene, given that the gene is an autosomal dominant, then segregation analysis yields the ratio  $162(P_1):P_2$  with the odds being dependent on  $P_1$  and  $P_2$  (Table). If we assume that the ratio of  $P_1$  to  $P_2$  does not differ too drastically from 1:1, then the evidence is greatly in favour of  $P_1$ , and consequently of the gene being an autosomal recessive.

Although a number of other possibilities exist for the occurrence of this rare trait in this family, for

example, polygenic inheritance, greatly reduced penetrance accounting for non-expression in VI.10-20 because of the presence of a modifier gene (which had to be present in IV.2 or IV.3, absent in V.3, 4, and 7, and present again in VI.10-20), and environmental factors, we feel, at least intuitively, that autosomal recessive inheritance is the most likely explanation for the genetics of this rare condition.

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Table Conditional probabilities of gene

Condition	Conditional probabilities if gene is	
	Autosomal recessive	Autosomal dominant
IV.2 and IV.3 have 4 affected children V.1 is a carrier of the CHANDS gene	$(3/4)^4 (1/4)^4$ $(1/2)^2$	$(1/2)^8$ Not applicable
V.1 and V.2 have 3 affected children and 6 normal children	$(1/2)^9$	$(1/2)^9$
V.3 has 5 normal children	1	$(1/2)^5$
V.4 has 6 normal children	1	$(1/2)^6$
Total conditional probability	$3^4 (1/2)^{27}$	$(1/2)^{28}$

## Trisomy 16q arising from a maternal 15p;16q translocation

SUMMARY Trisomy 16q is reported in a malformed infant who died at 12 days of age. The karyotype was 46,XX,der(15)t(15;16)(p11;q11)mat. A balanced translocation was found in the mother. The consequences of various types of aneuploidy of chromosome 16 are discussed.