

Recessive aplasia cutis congenita of the limbs*

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SUMMARY Six inbred persons (five males and one female) in three generations of a single family are reported as having simple congenital absence of skin on the upper or lower limbs or both. The data suggest an autosomal recessive pattern of inheritance for this apparently new clinical entity.

Aplasia cutis congenita (ACC) is a clinically and aetiologically heterogeneous group of conditions usually involving the scalp (over 80% of cases) as well as the lower limbs, trunk, neck, upper limbs, and face (with or without scalp involvement). A number of other congenital anomalies have been reported in association with ACC: absence of ear lobes¹; congenital heart disease²; cleft lip and palate, anophthalmos, and deformed ears³; and neurological changes.⁴ ACC may also be seen in the 4p— syndrome, the 13 trisomy syndrome, Johanson-Blizzard syndrome, focal dermal hypoplasia (also known as Goltz-Gorlin syndrome, an ectodermal dysplasia of the tricho-odonto-onycho-dyshidrotic subgroup), acrocephalopolysyndactyly, and a syndrome of reduction deformities of the limbs and aplasia of the skull and scalp.^{5,6} For reviews and case reports, see also Abt,⁷ Adair and Stewart,⁸ Dowler,⁹ Rauschkolb and Enriquez,¹⁰ Sirol *et al*,¹¹ Bart,¹² and Fisher and Schneider.¹³

This paper describes and discusses the occurrence of congenital absence of skin in the upper or lower limbs or both in six members (five males and one female) of three inbred sibships of the same kindred.

Case reports

The pedigree (fig 1) comprises seven generations of 238 Caucasians (six with ACC) from the north-eastern Brazilian state of Ceará. Three males, not personally examined, (VI.8, VI.17, and VI.20) from a consanguineous marriage (F = 3/64) were reported as having had ACC only in the lower limbs. One (VI.8) had the right foot (dorsum, sole, and toes) affected, and two (VI.17 and VI.20) had both legs and

feet (dorsum, sole, and toes) affected. ACC in the upper and lower limbs (dorsum of the hands, and dorsum of the feet and toes) was reported in IV.14, an inbred man (F = 1/8) who died at the age of 30 years.

The following is a description of two cases, the only ones we were able to examine.

CASE VII.4 (INDEX CASE)

A 5-year-old female from a consanguineous marriage (F = 1/27) had ACC in the right leg and foot (fig 2). Labour was normal and the baby was otherwise normal. The treatment consisted of covering the red areas with vitamin A; sterile gauze dressings were applied to avoid trauma and infection. A few months after birth the lesions healed completely and spontaneously (fig 2). The clinical appearance of the defect is now similar to the hypotrichotic scar of an old burn.

CASE VI.7

This case was an adult when the diagnosis of ACC was made. A 34-year-old male from a consanguineous marriage (F = 3/64) had a scarred area on the dorsum of his right hand (fig 3). No treatment was given, but precautions to avoid trauma and infection were taken by his parents.

Discussion

Intrauterine disturbances and genetic factors have been considered in the aetiology of ACC (reviews by Rogatz and Davidson,¹ Farmer and Maxmen,³ Mardini *et al*,⁶ Dowler,⁹ Montgomery¹⁴). The 'scalp' form is reported as having an autosomal dominant pattern of inheritance.^{2,10,13,15} Isolated cases of 'different' forms of ACC have also been reported, at least one of them being the offspring of a consanguineous mating.^{3,7,8,9,11,16-18} These isolated

*This study was supported by grants from the National Council for Scientific and Technological Development (Brazil) and the World Health Organization.
Received for publication 31 May 1979

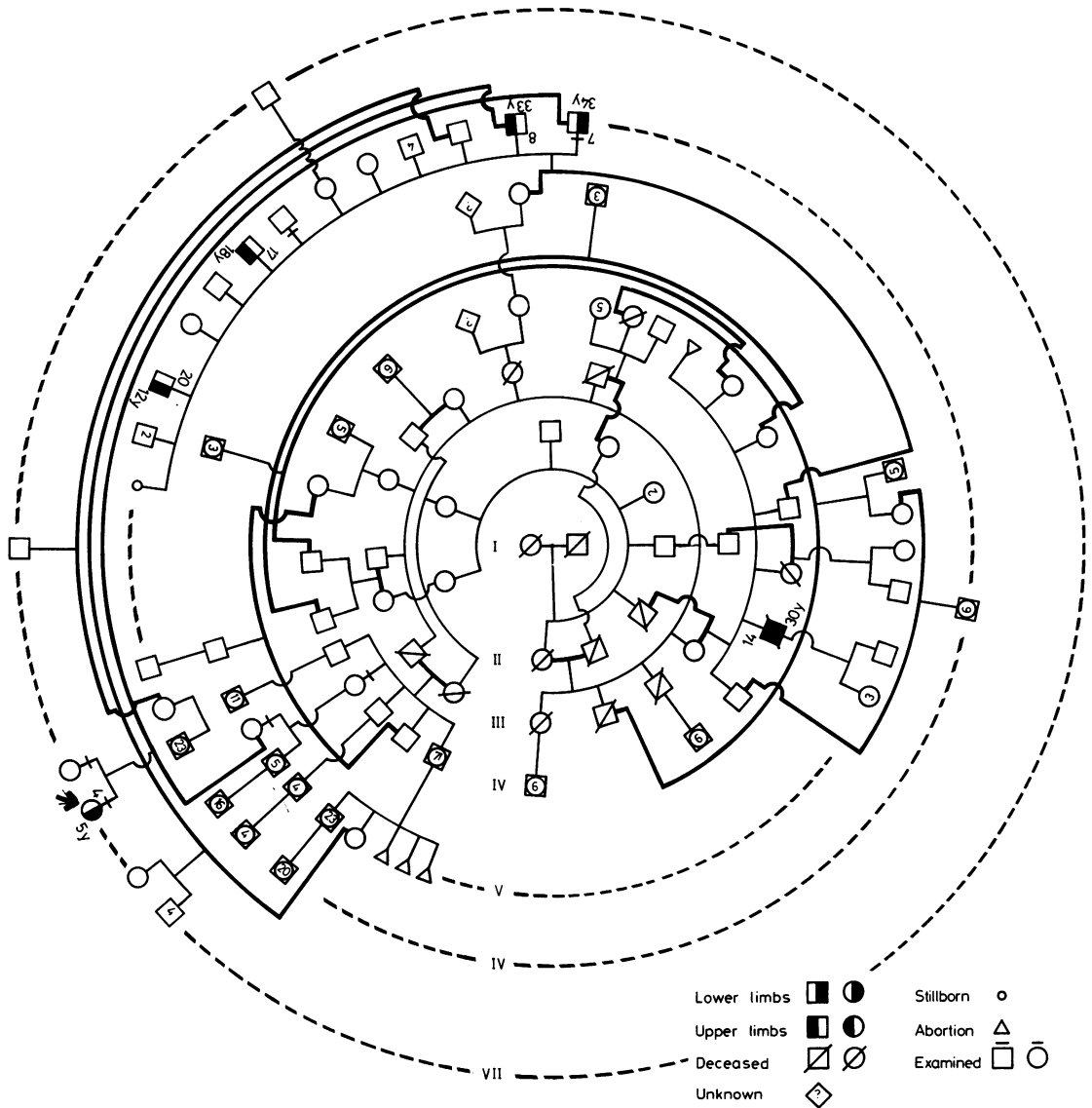


FIG 1 Pedigree of family.

cases involved (1) the scalp with or without bone defect; (2) the scalp and the trunk; (3) the scalp and congenital malformations not related to the skin; (4) the trunk and the upper limbs; (5) the knees only; and (6) the lower limbs only.

Our data show that (1) all affected persons are inbred; (2) the father of one of them, who married a relative, is also affected; (3) the segregation ratio among the children of normal parents is 5/22, and among the children of the affected father is 1/2; (4)

there are five males and one female among the affected. These facts indicate an autosomal recessive pattern of inheritance.

This condition is not listed in McKusick's catalogue.⁵ The condition referred to by Rauschkolb and Enriquez¹⁰ in item 20770 of this catalogue is also characterised by skin aplasia of the limbs, but, as mentioned before, is the result of a dominant gene with incomplete penetrance. Gedda *et al.*,¹⁹ also cited in that item, reported 'cases of aseptic gangrene of

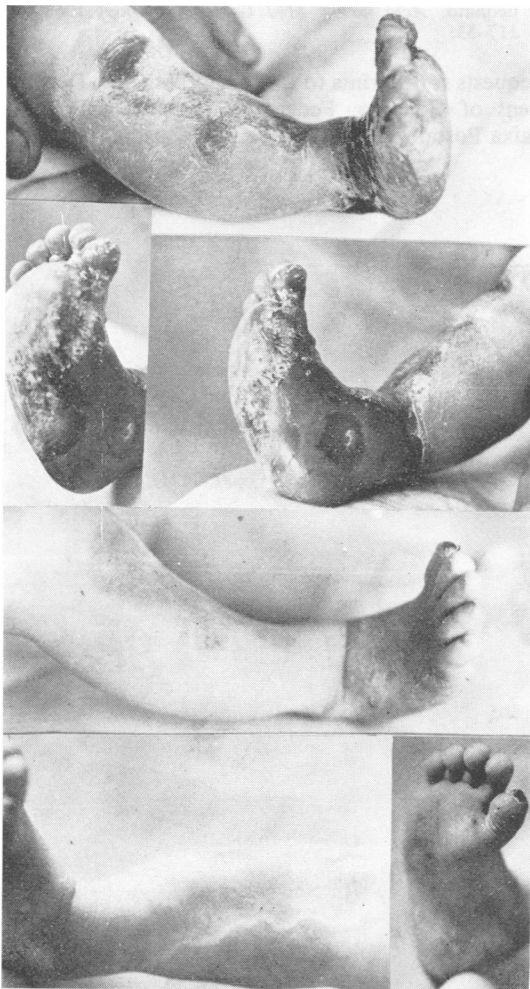


FIG 2 Right leg and foot of index case VII.4. Upper photos: at birth, defect covered by glistening, translucent membrane. Lower photos: cicatrization after a few months.



FIG 3 Right hand of case VI.7 when 31 years old. Note the healed scar tissue.

the skull', also called 'localised skull aplasia'. This again has nothing to do with the condition seen in our patients who had normal skulls.

The form of ACC we describe, involving regions of both upper and lower limbs (without scalp and trunk involvement), seems to be a 'new' condition with an unquestionable autosomal recessive pattern of inheritance.

We are very grateful to Dr Rita Maria Patribu Faria for her collaboration during the treatment of the index case.

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