Simple Anonychia

Further Evidence for Autosomal Recessive Inheritance

MOHSEN MAHLOUDI and M. AMIDI

From the Departments of Neuropsychiatry and Medicine, Pahlavi University School of Medicine and Nemazee Hospital, Shiraz, Iran

Complete or partial absence of nails is a feature of a number of familial disorders. There may be additional anomalies such as absence of patella, ectrodactyly (Lees *et al*, 1957), minor deformities of the fingers (Strandskov, 1939), and lymphoedema (Maisels, 1966), or the defect might be confined to the nails—simple anonychia—(Charteris, 1918; Hobbs, 1935; Timerman, Museteanu, and Simionescu, 1969). The majority of these anomalies appear to be manifest in the heterozygous state and there is good evidence, at least in some cases, that the mutations are at different loci (Lees *et al*, 1957).

In 1933, Cockayne reviewed the literature and suggested that there might be a recessively inherited variety of simple anonychia. More recently, Littman and Levin (1964) have put forward the same suggestion but this mode of inheritance has not yet gained general acceptance (McKusick, 1968). The purpose of the present communication is to report a family in which the distribution of the affected members gives strong support in favour of autosomal recessive inheritance.

Case Reports

Case 1. The proband was a 30-year-old woman who had come to hospital for a 'check-up'. She had minor complaints such as discomfort over the precordium and general aches and pains. Complete absence of all nails (Figs. 1 and 2) had been noticed since birth but did not bother her. Physical examination and a number of laboratory tests revealed no other abnormality and she was reassured and sent home without medication. In particular her hair, teeth, and skeletal system appeared to be normal.

Case 2 was a 60-year-old woman (III.4) who was persuaded to attend the clinic by case 1. She did so reluctantly and refused even simple photography. She too had no nails on her fingers or toes and no other abnormality.

FIG. 1. Case 1: the hands showing total absence of nails.

FIG. 2. Case 1: the feet showing total absence of nails.

Family History

The patients came from a small town in southern Iran with a population of about 5000. They belonged to a well-known family some of whom, like case 1, had emigrated to Bahrain. Fig. 3 shows the pedigree. It was constructed from information given by the 2 patients

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Familial aggregation in the absence of abnormality in the parents has been reported only 3 times previously. Jacob quoted by Heidingsfeld (1913) and Cockayne (1933) described a Russian girl with almost total anonychia who had 2 similarly affected sibs; the teeth in the only patient examined were abnormal. From Britain, O'Neill (1916) reported complete absence of all nails in 3 sibs. Finally, Littman and Levin (1964) described partial

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anonychia of the fingers but normal toes in a girl of German extraction who was said to have a similarly affected brother.

Consanguinity has been reported in two isolated cases. From Cincinnati, Heidingsfeld (1913) described a man, whose parents were first cousins, with complete absence of nails. Timmer and Wildervanck (1969) reported a Dutch girl with total anonychia whose parents were second cousins. The present family is the largest on record and the only one with both multiple cases and consanguinity.

Cases falling in the category of recessively inherited simple anonychia have been reported in individuals from Russia, Great Britain, America, Holland, and now Iran. In all except the cases of Littman and Levin (1964) the anonychia appears to have been total; the fingers as well as toes being affected. It is still not possible to say if all these cases are due to mutations at the same loci or that there is further heterogeneity.

Summary

A family is described in which several members suffered from total absence of all nails on the fingers

Recessively inherited simple anonychia appears to be extremely rare. A number of isolated cases are on record but as Cockayne pointed out, these may be examples of new dominant mutations.

and toes. The normal parents, the mixed sexes of the affected individuals, and the frequent consanguinity are strongly in favour of autosomal recessive inheritance. The literature is reviewed and it is suggested that there is an extremely rare variety of simple anonychia that is recessively inherited.

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