Brachydactyly in an Indian Family*

JAG MOHAN

From the International Laboratory of Genetics and Biophysics, Naples, Italy

Brachydactyly was the first case of Mendelian inheritance shown in man and was reported to be a simple dominant (Farabee, 1905). Since then, pedigrees have been published showing this pattern of inheritance.

The term brachydactyly is generally used to describe a number of abnormal conditions which result in the shortening of one or more digits, and these have been grouped into five main types by Bell (1951). Variations of all these types have been noted both within and between different pedigrees in the literature. It is not known whether these various types might be due either to variable expressivity of the same gene depending upon the milieu; or to different alleles of the same gene, or to the existence of two or more different mutant genes.

Dwarfism of the stature, differing from the conventional type, has been associated with one type of brachydactyly (Farabee, 1905; Drinkwater, 1908, 1913, 1914, 1915; Liebenam, 1938; Grebe, 1943; Iltis, 1944) classified as A₁ by Bell (1951): in this

TABLE
SOME ANTHROPOMETRIC MEASUREMENTS (cm.)
OF THREE SIBS AS INDICATED IN FIG. 1

Age in years	IV.11 (normal) 43		IV.12 (affected) 34		IV.17 (affected) 23	
	R	L	R	L	R	L
Measurements						
Height vertex	166.7		164.55		160.9	
Upper arm length	30.1	30.3	27.9	28.2	27.5	27.8
Lower arm length	25.1	25.3	24.7	25.05	24.9	25.1
Hand length	18.3	18.5	16.8	17.0	16.65	16.7
Hand breadth	7.8	8.0	8.85	9.0	8.15	8.25
Hand index		42.9		52.8		49.2
Fingers' lengths						
I	6.4	6.4	5.1	5.0	5.5	5.6
II	7.95	8.1	6.8	6.8	7.0	7.1
III	9.45	9.60	7.35	7.45	7.6	7.5
IV	8.75	8.90	6.45	6.55	7.1	7.1
V	7.0	7.0	5.2	5.3	5.7	5.6
Length of thigh*	43.3	43.5	39.9	40.2	38.4	39.6
Lower leg length	39.9	40.0	38.8	38.8	36.7	36.2
Foot length	25.5	25.7	23.9	24.4	22.45	22.4
Foot breadth	10.1	10.3	10.8	11.0	10.35	10.3
Foot index		39.8		45.1		46.0
Toes' lengths						
I	6.40	6.55	5.6	5.1	5.05	5.0
II	5.8	5.9	4.5	4.6	4.5	4.3
III	5.0	5.15	4.4	4.1	3.9	3.7
IV	4.9	4.85	4.0	3.8	3.7	4.0
V	4.35	4.45	3.6	3.1	3.3	3.3
Span		170.0		160.8		160.7

Received November 4, 1968.
* This work was carried out, in part, under the Euratom-C.N.R.-C.N.E.N. Contract No. 012-61-12 BIAI.

* Calculated from the formula (height iliospinale - height tibiale) - 7%.
Note: The landmarks for measurements are those described by Montagu (1960).

FIG. 1. Pedigree of family.
349

FIG. 2. The brachydactylous hands and feet of IV.12. The length of the right big toe is larger than the left one.

FIG. 3. Brachydactylous hands and feet of IV.17. Note the length of the fourth toe of the left foot.

case the middle phalanx of all digits is rudimentary or short and may or may not be fused with the terminal phalanx; the proximal phalanges of thumbs and toes are also shortened.

This paper describes a pedigree of brachydactyly in a Punjabi Hindu family, domiciled in Delhi and Muradabad (U.P.). Except for the abnormal hands and feet, all the affected subjects were apparently normal. The external appearance of the affected digits was like that of the normal relatives (except for their reduced size).

Case Report

The distribution of the defect in the family has been traced through four generations (Fig. 1). Information about the deceased was obtained by separate interviews from II.5, III.4, III.6, and IV.12, as indicated in Fig. 1. No consanguineous marriages were reported.

Fig. 2 and 3 illustrate the phenotype of the anomaly.

Both hands and feet of IV.12 and IV.17 show the effect. The presence of the trait is primarily due to the involvement of the middle phalanx in all the fingers and toes. The little finger in both subjects does not show the middle phalanx. The fusion of the middle phalanx with the terminal phalanx is observed in IV.12, and this fusion is symmetrical in the case of the ring finger, while the index finger exhibits fusion only in the right hand. The remaining fingers of this individual (IV.12) show middle-phalanx shortening, as do the rest of the fingers of subject IV.17. The abnormality in the toes, where only two phalanges are present in all digits, appears to be more consistent at first glance in both subjects. However, closer scrutiny reveals these differences in quantitative terms, as shown in the Table.

One or more affected subjects were observed in each of the first four generations (Fig. 1). The abnormality regularly appeared in the progeny of a marriage between a brachydactyl and a normal person. The observed proportion in the progeny was 5 brachydactyls to 7 normals. In the affected, 2 were males and 3 were females.

From these observations, it appears that the abnormality is due to an autosomal dominant.

Anthropometric measurements taken on a sibship of three of generation IV are listed in the Table. Except for the measurements of the extremities, observed differences in most of the other comparative measurements could be within the range of normal variation. The length of all fingers and toes is shorter in the affected, and thus the indices for hands and feet are increased. Nissen (1932-33) has described a brachydactylous pedigree with similar features to those presented here, and likewise found no association with dwarfism.

Summary

A new pedigree from India of brachydactyly primarily due to involvement of the middle phalanx in the fingers and toes is reported. It shows autosomal dominant inheritance. Anthropometric measurements of 3 sibs are also presented.

REFERENCES

- Bell, J. (1951). On hereditary digital anomalies. Part I. On brachydactyly and symphalangism. In *The Treasury of Human Inheritance*, Vol. 5, p. 1. Ed. by L. S. Penrose. Cambridge University Press, London.
- Drinkwater, H. (1908). An account of a brachydactylous family. *Proc. roy. Soc. Edinb.*, **28**, 35.
- (1913). Account of a family showing minor brachydactyly. *J. Genet.*, **2**, 21.
- (1914). Minor brachydactyly. *ibid.*, **3**, 217.
- (1915). A second brachydactylous family. *ibid.*, **4**, 323.
- Farabee, W. C. (1905). Inheritance of digital malformations in man. *Papers Peabody Mus. Harvard Univ.*, **3**, 65.
- Grebe, H. (1943). Der Nachweis der Heterozygoten bei rezessiven Erbweisen. *Der Erbarzt*, **11**, 1. (Quoted by Bell, 1951.)
- Iltis, H. (1944). A new case of typical brachydactyly. *J. Hered.*, **35**, 145.
- Liebenam, L. (1938). Beitrag zum familiären Auftreten der Brachydactylie. *Z. menschl. Vererb.-u. Konstit.-Lehre*, **22**, 418.
- Montagu, M. F. A. (1960). *An Introduction to Physical Anthropology*. Charles C. Thomas, Springfield, Illinois.
- Nissen, K. I. (1932-33). A study in inherited brachydactyly. *Ann. Eugen. (Lond.)*, **5**, 281.