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Polydactyly — A Study of Variation
In the Expression

Abstract. — In the present study three pedigrees of polydactyly were studied. It is found that the character appears in different forms and in different combinations. Variations in expression of the trait are also found to occur in some pedigrees. It is observed that the factor responsible for polydactyly is a dominant gene the expression of which is highly variable.

Introduction
Inherited abnormalities of hands and feet generally known as polydactyly have attracted attention from early times. The phenomenon has received the scrutiny of many investigations. Pedigrees showing familial occurrence of the trait have frequently been described in the genetic and medical literature. Marked variations in the expression of polydactyly in man have come to be described with respect to number of extra digits and their position on extremities. The most frequently observed condition is hexadactyly. In this, the additional digit may occur on radial (pre-axial) or ulnar (post-axial) side, on either or both hands, together with either or both the feet. Variations in the expression of trait are further accentuated due to various degrees of development of the character in different persons. Gates (1957) envisages three modes of polydactyly condition representing three stages of development:

a) Most commonly the extra digit is a small attachment not adherent to the skeleton and often without bones or cartilage, muscle or tendons.

b) The extra digit is often more or less like an ordinary finger or toe, containing bones connected with the skeleton of the finger.

c) Very seldom extra digit is complete and may have even its own metacarpol or metatarsal bone.

All possible intergrades occur between the stages described above.

Evidently different hereditary factors appear to be responsible for various forms and grades of polydactyly in man. With such wide range of expression of the condition it is not surprising to see different modes of transmission reported in different pedigrees. The wide range of variation in the expression of polydactyly has led to varied interpretation as to the mode of transmission of the condition. In many pedigrees of polydactyly reported in the past the manifestation of the trait has been attributed to a dominant gene with complete or incomplete penetrance. In the latter case the inheritance is irregular due to failure of the dominant gene to express itself phenotypically in some individuals who can indirectly be shown to have transmitted the condition to their offspring.

Atwood and Pond (1917) described a family with radial polydactyly in which thumbs and great toes were generally double and the mode of inheritance was found to be dominant. Miles (1928) likewise observed strictly dominant inheritance in a number of pedigrees he studied. Oral (1928) reported a pedigree of double thumb strictly dominant with 18 cases appearing in five generations. Osburne (1943) reported a striking pedigree of polydactyly in six related families and found considerable va-
nogeneity (II-2), its recurrence in III-6 makes it very likely that his mother II-5 is genotypically polydactyl who has passed the dominant gene to the next generation without herself giving expression to it on account of its low penetrance.

PEDIGREE – III

It is a large pedigree with a high incidence of polydactyly which could be traced up to the five generations. The propositus III-4 in this pedigree is head of a Rajput family and most of its affected members were available at one place. There are many variations in the expression of the trait in this pedigree. In some individuals the additional digit occurs on hands as well as feet, in others it is confined to hands or feet alone. The extra digits containing bones and nail are of normal size.

ANALYSIS

All the affected individuals have been found with one affected parent and no skipping has been observed except of I-5 beyond which no record is available. The ratio of affected individuals to unaffected, born to affected parents is 9:10. In this pedigree the polydactylyous condition is inherited in a dominant fashion, though the expression of the trait is variable. The affected condition in children is not necessarily similar to one in the parents. In some cases the individuals have (II-3, IV-3, V-3) additional digits on both hands and feet but parents have either extra digits on hand or foot. Similarly II-2 has affected feet only whereas the parent II-3 has affected hands.

REFERENCES


