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POLYDACTYLY — A STUDY OF VARIATION IN THE EXPRESSION

ABSTRACT. — In the present study three pedigress 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 polydactylous 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 metacarpal 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 polydactylism 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 offsprings.

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. Milles (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. Odioane (1943) reported a striking pedigree of polydactyly in six related families and found considerable va-

riation in the expressivity of the trait. Existence of recessive polydactyly is not unknown, though it is rare in comparison with the dominant form. It has been reported by Snyder (1929) in a large Negro family and by Oliver (1940) in a white family.

According to Gates (1957) — polydactyly is a sliding scale of values from absolute to variable dominance with such low penetrace that it can not be distinguished from recessivity.

MATERIAL AND METHOD

The pedigrees presented here exhibit in addition to the more usual forms, some peculiar polydactylous conditions which are of rare occurrence. In the preparation of pedigree charts, conventions in common use have been followed. The circles stand for females and squares represent males. The lines inside the circles or squares stand for the affected condition. While information for the affected condition in the younger generation was checked by the personal observation as far as possible, the occurrence of the trait in older generations was mostly affirmed on the basis of verbal information. Individuals are referred to in the text by Arabic numerals assigned to them in the pedigree charts, prefixed by the number of generation (given in Roman figures) of which they are members. This procedure has been adopted, as it was desired by some of the informants, that their names should not be disclosed.

OBSERVATIONS

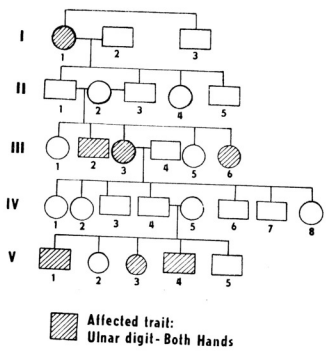
PEDIGREE — I

In this pedigree, the trait is traceable for five generations. The information for the first three generations is limited. The affected individuals show an additional ulnar digit on both hands which is almost of the size of small finger with bones and nail.

ANALYSIS

A special feature of this pedigree is that the trait is found in alternate generations i.e. I, III and V or in other words affected individuals have non-affected parents and hence providing a very good case of irregularly dominant mode of inheritance. The plea for a recessive mode of inheritance, this pedigree is ruled out at the probability of two heterozygotes coming together in every alternate generation is remote, the trait being quite rare in population. The apparent irregularity of the inheritence can best be explained as due to failure of a dominant gene to produce a recognizable effect in certain individuals who appear to have transmitted the trait to their children without themselves being visibly affected.

PEDIGREE - I



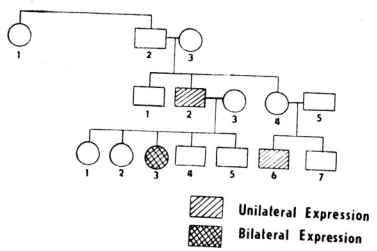
PEDIGREE — II

This is a pedigree with a rare kind of pre-axial (radial) polydactyly. The pedigree could be traced for only three generations, with only three affected individuals. In one of the individuals III-3, the character is expressed bilaterally, while in other two individuals (II-2) and (III-6) the extra thumb appears unilaterally on the left hand and right hand respectively, the additional thumb is fully developed and emerging from the base of normal thumb and is completed with bone and nail.

ANALYSIS

In this pedigree only one of the affected individuals (III-3) has an affected parent, the other two (II-2 and III-6) have non-affected parents. The polydactylous condition in this pedigree seem to behave in an irregularly dominant manner. While the trait first appears in an individual of the second

PEDIGREE - II

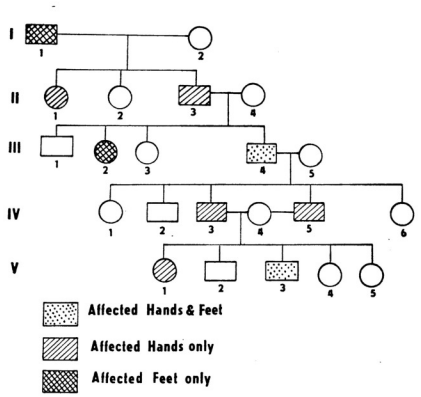


generation (II-2), its recurrence in III-6 makes it very likely that his mother II-5 is genotypically polydactyl who has passed the dominant gene to the next generation without herself giving expression to it on account of its low penetrance.

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 occures 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.

PEDIGREE - III



ANALYSIS

All the affected individuals have been found with one affected parent and no skipping has been observed except of I-1 beyond which no record is available. The ratio of affected individuals to non-affected, born to affected parents is 9:10. In this pedigree the polydactylous 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 (III-4, V-3) additional digits on hand and feet but parents have either extra digits on hand or feet. Similarly III-2 has affected feet only whereas the parent II-3 has affected hands.

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