ORTHODACTYLY

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IN 1901 G. Walker described a family marked by inherited stiffness of finger joints, and in 1915 Harvey Cushing compiled the genealogy, through seven generations, of another such family comprising 452 individuals.[1] These were all descendants of one William B., who had come to Virginia from Scotland about 1700.

I have recently found a family which shows the same peculiarity. It is not included in Cushing's chart, but probably derives the trait from the same source, since family tradition traces it to ancestors in Scotland.

It is not the purpose of this paper to give the anatomy of the stiff joints, but only to point out their inheritance. The joints affected are the second ones of the fingers and frequently the corresponding ones of the toes. The joints are absolutely rigid. That the bones are not always fused is evidenced by the fact that one case is known where the joint was accidentally disjointed. The abnormality may occur in two, three or four of the fingers.

This trait behaves as a Mendelian dominant. Whenever the trait does not occur in an individual that person is unable to transmit it, that is, the normal hand is recessive to this trait and cannot transmit it.

Inspection of the chart shows the following facts. If we discard the D generation where the size of the family is small and consider only the B and C generations, we find that the number of affected individuals is eleven and the number of normal is four.

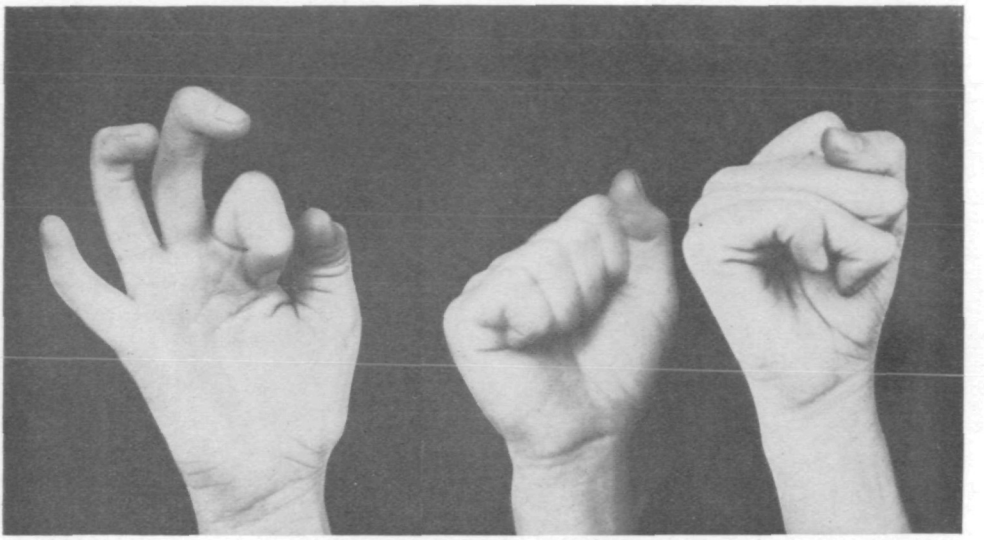
As only part of generation B shows the trait, it evidently was not homozygous in generation A. Further, as it appears to be a dominant trait, it could not have been carried by the mother in generation A, since she would have shown it if she had it. This leaves only one possible type of mating to fit the fact—a mating of heterozygous dominant with homozygous recessive. From such a mating it would be expected that affected and normal offspring would be produced in equal numbers, or 7.5 : 7.5, whereas the figures actually found are 11 : 4. This is very poor agreement, although not impossible when the numbers involved are so small.

Cushing charts 150 children from such matings, of whom 78, or 52%, were affected. This is so near the expected 50% as to make it practically certain that the abnormality is due to a single Mendelian factor.

Recent work of Cushing, Marie and others makes it seem probable that the trait here studied is related to some prenatal interference with the normal function of the pituitary body. Its apparent relation with brachydactyly, a well-established Mendelian character, is evident.

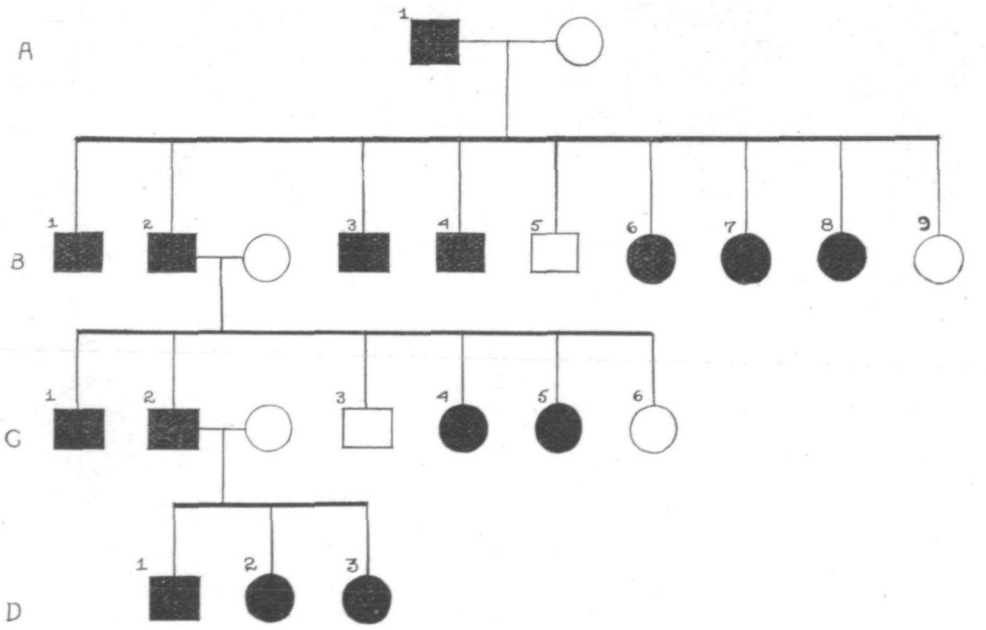
Cushing has called this trait symphalangism, a good descriptive term. Before I knew of his work I called the trait orthodactyly, or straight finger. As this trait is so similar in character and inheritance to other digital abnormalities it seems that it would be better to give it a similar name; however, the name is of little consequence.

[1] Cushing, Harvey. Heredity anchylosis of the proximal phalangeal joints (symphalangism). Genetics, i, pp. 90–106, January. 1916.



THE EFFECT OF ORTHODACTYLY

At the left is a hand with the third, fourth and fifth fingers affected. The middle joints of these fingers are stiff and cannot be bent. At the right the same hand is shown, closed. A normal hand in the middle serves to illustrate by contrast the nature of the abnormality, which appears in every generation of the family. (Fig. 8.)



A FAMILY WITH ORTHODACTYLY

Squares denote males, circles females. Black squares or circles denote affected individuals. A1 had all fingers affected; B2 had all but one finger affected; C2 had all but one finger affected; D2 had all fingers affected; D3 has all but forefingers affected. (Fig. 9.)