On the inheritance and development of preaxial and postaxial types of polydactylism

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The various skeletal defects of the extremities in man stem from a combination of factors. The intricate architectural design imposed on the extremities makes them more prone to a greater number of genetically controlled developmental errors. These morphological errors are seldom lethal and in no way do they interfere with the general physiological processes of the body. Moreover, they do not interfere substantially with the survival value of modern man, creating no particular problem in the propagation of the species. In addition, these traits are generally exposed and easily detected. Many such defects have been summarized by Bell (1951, 1953) and Werthemann (1952). X-ray studies have contributed more accurate knowledge to the genetics of these skeletal parts, especially to the field of human developmental genetics (Hersh et al. 1953).

Polydactylism is not a rare skeletal anomaly in man. This condition is generally defined as having more than the usual number of digits in hands and feet. If the extra digit occurs on the radial or tibial side the anomaly is usually referred to as preaxial polydactylism; if it occurs on the ulnar or fibular side it is referred to as postaxial polydactyly. It is indeed a very rare occurrence to have a duplication of the middle fingers and toes. Polydactylism may occur in one, two, three or all four extremities. The duplication of a digit may range from a complete functional finger, containing all the normal bony elements, to mere expression of a finger, without any signs of extra phalanges.

The data

The data collected and described here has been chiefly derived from gross inspection of the hands, X-ray examination, and the gathering of pedigree, supplemented by a brief family history.

Preaxial Polydactyl-Thumb duplication. This came to our attention when X-ray examinations were made of a « new born » baby girl who externally displayed a
partial duplication of the left thumb. Consequent inquiry led to the following information: the affected child is an only child; the mother of the child claims that her father had affected thumbs (see pedigree in figure 6A). Roentgenograms were made of the hands of the child, III-5 and both parents, II-4 and II-5. The «new born» child, III-5 has a broad left thumb, slightly flared at the distal end with a soft tissue appearance suggesting partial duplication.

Figure 1 shows X-rays of the left and right hands of this «new born» child. The left thumb shows an extra bone bifurcation of the distal phalanx (shown at the point of the arrow). Although the right thumb appears normal externally, the X-ray view shows a partial duplication of the terminal phalanx. It may be noted that it is opened at its distal end and fused at its base. No other abnormal ossifications are in view at this age. The hands were X-rayed once more 5½ months later. These are shown in figure 2. Here of course, the terminal phalanges of both thumbs are more fully developed. The two terminal phalanges of the left thumb show no sign of fusion and the terminal phalanx of the right show no sign of further separation. A new ossification center, however appears at this stage in the left thumb. It appears just below the distal phalanx of the medial side. The growth and shape of these...
duplicated portions of the thumb at the two different ages are best compared in figure 3, a, b, c, d and e. Figure 3a and b shows the condition of the left and right terminal phalanges at the time of birth. Figure 3c and d (e is another view of d from a slightly different angle) shows the condition of these terminal phalanges 5½ months later. In figure 3c may be seen (at the point of the arrow) what appears to be the beginning of an ossification center, for the distal epiphysis. The ossification of this epiphysis seems to occur unusually early. Generally, in girls, it begins at about 15 months after birth, according to Gruelich and Pyle (1950), and perhaps slightly earlier according to Caffey (1956). The extra distal bone, in this case, may act to induce an earlier ossification of this epiphysis.

The mother shows no outward sign of duplication of the thumbs, except that the distal portion of the right thumb is angled medially. X-ray study of her hands shows no sign of the duplication of any of the phalanges. A slight deformity occurs in the right thumb. The base of the medial portion of the distal phalanx seems shorter, thus causing it to tilt medially at an angle of about 24°. Figure 4a and b shows lateral views of the right and left thumbs of the mother respectively, while figure 4c and d shows AP views of the same. Figure 4c, marked with an arrow, points to the approximate location of the deformity which is causing it to angle medially. The left thumb appears normal. In itself this slight deformity would have escaped our attention, if it had not been X-rayed and observed in connection with the affected child. It is
Fig. 3 - X-ray views of the thumbs, comparing their growth and shape at two different ages. $a$ and $b$ are left and right thumbs, respectively at birth. $c$ and $d$ are left and right thumbs, respectively as they appear $5\frac{1}{2}$ months later. The arrow points at a new ossification center appearing since birth. $e$ is another view of the right thumb from its lateral position.

Fig. 4 - X-ray views of the thumbs of the mother. $a$ and $b$ are lateral views of the right and left thumbs, respectively. $c$ and $d$ show A-P view of the same. The arrow points at the approximate location of the slight deformity. The distal phalanx tilts medially about $24^\circ$. 
this type of weakly expressed trait which may easily be missed, that oftentimes leads
to errors in determining the degree of penetrance or the mode of inheritance of
the trait.

Roentgenograms of the father's hands revealed no noticeable variations with
respect to this trait.

Although the duplication of the thumb is a rare trait, it has been previously de­
scribed in the literature. Duschl (1917), Reeves (1920), Penhallow (1928), and Wein­
grow (1930) have described this form of polydactylyism but have not attached any he­
reditary significance to it. Sinha (1918) briefly described this condition in a Hindu
family and believed it to be hereditary but did not make any attempt to analyze
the pedigree. Manoiloff (1931) described a pedigree which might indicate that this
trait is dominant. Rudert (1938) and Callan (1942) concluded from their studies that
this form of polydactylyism is due to an irregular dominant gene. Hefner (1940) con­
cluded it to be due to a simple regular dominant factor. There is no mention in

Fig. 5 - X-ray view of the left and right hands showing postaxial polydactylyism. Arrows point at the extra
digits arising at the site of the metacarpal-phalangeal joints (Age of woman, 44)
the literature that might indicate the possibility that this form of polydactylism is inherited as a recessive. Figure 6A gives the pedigree for this type of polydactyl. An inspection of this pedigree seems to indicate that this trait may be inherited as a simple dominant. If the mother, II-4, given in this figure had not been X-rayed she would have been passed off as normal. Hefner (1940) likewise discovered these slight defects of the thumb among the normal-appearing individuals in the pedigree. From the best available data to date we tentatively conclude that preaxial polydactylism is inherited as a simple autosomal dominant having a highly variable expression.

Postaxial Polydactylism—a 5th finger duplication. This condition was discovered in a 44-year old coloured woman (figure 6B, III-2). She had a sixth miniature finger arising at the base of the little finger (5th) in both hands. A similar condition occurred on the feet but here the extra toes had been removed by surgery at birth. X-ray examination of the hands reveals the presence of only two phalanges in each of these extra digits. They arise at the site of the metacarpal-phalangeal joint (see figure 5). Figure 7 gives a detailed view of these extra digits. Further inquiry of this case revealed that this woman has two daughters. The older is unaffected, and the other had a similar extra digit on both hands like the mother but in a modified form. These had been removed surgically at birth. No polydactylism was indicated in the feet. It is of interest to record that this younger daughter is presently suffering from leukemia and arthritis. The pedigree chart given below (figure 6B) was constructed partly from information supplied by the mother, III-2. It is of interest to note that both of her parents, II-4 and II-5, had this trait on both hands and feet. Yet her two sisters were unaffected (III-3 and III-5). Such a condition indicates that this trait may be inherited as a dominant.

Many cases of postaxial polydactylism were described in medical literature previous to 1915. These, however, are too incomplete to be properly evaluated from a genetic point of view. After this period some advances were made in this field. Brandeis (1915) studied this trait and concluded that it is inherited as an irregular dominant factor. Koehler (1923) more extensively showed that this trait is inherited as a dominant factor, but very much influenced by other genetic factors. Snyder (1929), on the other hand, analyzed a group of Negro families from Pamlico

Figure 6. Two pedigrees of polydactylism. A, preaxial polydactylism involving a duplication of the thumb in a white family. B, postaxial polydactylism involving a duplication of the little finger (5th) in a Negro family

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County N.C., who were more or less related to each other, and concluded that this trait is inherited as a recessive. Koehler (1930) reanalyzed Snyder’s data and concluded that it could be also explained on the basis of an irregular dominant factor, which he (Koehler, 1923) had used to explain his data. More recent workers in this field, McClintic (1935), Jackson (1937), Ordiorne (1943), and Johnston and Davis (1953) are all in agreement that this trait is due to an irregular dominant factor. Our data as presented in figure 6 B, seems likewise to be in harmony with this point of view. The non-affected children, III-3 and III-5, derived from the two affected parents, II-4 and II-5, excludes the possibility that the parents are homozygous recessive. The probability is more favorable that both parents are heterozygous dominant for this trait.

A distinction is made here between preaxial and postaxial polydactyly. From our
observed and our survey of the literature we have never come across a pedigree in which the two forms occurred together, that is, one is not a variant of the other. This points to the possibility that each type may be due to a different dominant gene.

**Summary**

Two types of polydactyly are described. Preaxial polydactylism is a relatively rare trait, involving a varying degree of duplication of the thumb. X-ray evidence is presented for this condition in a baby girl when first born and 5½ months later, showing the nature of its development. X-ray of the parents revealed that the mother was affected and the father was normal. Evidence points to the possibility that this trait follows a simple autosomal type of inheritance. Postaxial polydactylism, the more common type, generally involves a duplication of the little finger (5th) with a varying degree of expression. A pedigree of a Negro family is described and X-ray evidence for this trait is given. Each duplicated miniature finger consists of two small but well formed phalanges. The trait seems to be transmitted as an incomplete dominant.

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SOMMARIO
L'autore descrive due tipi di polidattilia. La polidattilia preaxiale è una caratteristica relativamente rara che riguarda la duplicazione del pollice in grado variabile. Con radiografie d'una bambina appena nata, e ancora ai cinque mesi e mezzo, si mostra la natura dello sviluppo di questa condizione. Radiografie dei genitori mostrano che anche la madre ne era affetta, ma il padre era normale. È possibile che questa condizione segua un tipo semplice autosomal di eredità. La polidattilia postaxiale di tipo più comune consiste d'una duplicazione del mignolo (5°) che si mostra in grado variabile. L'autore presenta anche una genealogia d'una famiglia negra e con radiografie mostra questa condizione. Ogni mignolo duplicato consiste di due falangi piccolissime ma ben formate. Sembra che la caratteristica si trasmetta come un dominante incompleto.

RÉSUMÉ
L'auteur décrit deux formes de polydactylie. Polydactylie préaxiale, un type relativement rare, comprend des degrés variables de duplication du pouce. Par des photographies de rayons-X, faites d’une enfant, nouvelle née et encore à l’âge de cinq mois et demi, on montre la nature du développement de cette condition. Le Radiographie des parents révèle que la mère était aussi affectée mais que le père était normal. Les témoignages indiquent qu’il est possible que ce trait suive un simple type autosomal d’hérédité. Polydactylie postaxiale, le type plus commun, comprend généralement une duplication du petit doigt (5ème doigt) laquelle se montre en divers degrés. On décrit la généalogie d’une famille noire et l’on donne pour ce trait le témoignage des radiographies. Chaque doigt miniature et doublé consiste en deux phalanges, petites mais bien formées. Il paraît que le trait se transmet comme un dominant incomplet.

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