

Brachydactyly and Pseudo-Pseudohypoparathyroidism *

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Arkless and Graham (1967), in reviewing a case which had been previously described as pseudo-pseudohypoparathyroidism by Miles and Elrick (1955), discussed the roentgenographic relationships and semantic problems involving brachydactyly, cone epiphyses, peripheral dysostosis and pseudo-pseudohypoparathyroidism. The present study will present the results of a roentgenographic examination of the hands of a sample of normal school children and will discuss these results with respect to each of the four above-mentioned conditions.

Observations

The roentgenograms of the left hands of 296 Philadelphia children were examined. The sample was composed of 96 Chinese, between the ages of 4 and 16, from the files of the Philadelphia Center for Research in Child Growth and Development, and 200 Negroes, between the ages of 7 and 11. Five of the Chinese, aged 4 to 10, were found to have fifth middle phalanges with cone-shaped epiphyses projecting into a diaphyseal crater (Fig. 1) similar to that illustrated by Laurent and Brombart (1953). In the oldest of these children, the epiphysis was beginning to show union at the apex of the cone (Fig. 2). Three others, between 10 and 12 years of age, had complete epiphyseal union of the fifth middle phalanx, without any other epiphyseal unions in the hand (Fig. 3). Thus it appears that the cone epiphysis and early union types are simply age variants of the same phenomenon.

In all eight of these cases the phalanges were unusually short but were well proportioned without having the radial side shorter than the ulnar, as is characteristic of clinodactyly. Measurements were made in the midline of the diaphyses of the fifth and fourth middle phalanges (or the entire phalanx in case of total epiphyseal union), and a ratio of the lengths of the fifth to the fourth was utilized in assessing fifth middle phalanx length variation. All of the Chinese cone epiphysis-early union type had a $5/4$ mesophalangeal ratio of less than 0.50, while of the 200 Negroes, none had a ratio of less than 0.50, none had cone epiphyses and none had early union. Four

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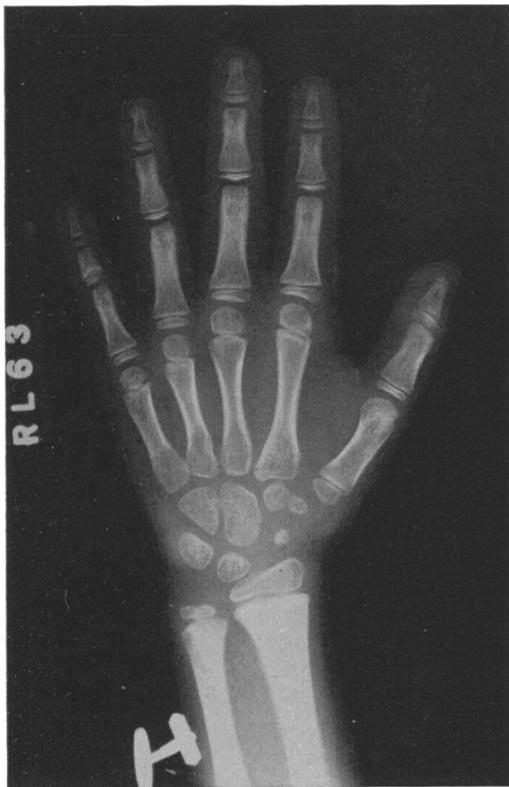


Fig. 1. Cone epiphysis and short cratered diaphysis of the fifth middle phalanx (Male, age 9)



Fig. 2. Cone epiphysis of the fifth middle phalanx beginning union at its apex (Female, age 10)

other Chinese also had ratios of less than 0.50. One of these individuals, 16 years old, had union of all epiphyses and doubtless represents the adult variant of the cone epiphysis type. Another, 10 years old, with the smallest diaphysis of the entire sample, had no epiphysis. The remaining two, with ratios just under 0.50, had the radial side shorter than the ulnar and perhaps represent cases of clinodactyly. None of the 84 Chinese with a ratio above 0.50 had a cone-shaped epiphysis or early union. Two of the 296 individuals in the sample showed other anomalies of the fifth digit, and these were two of the three Chinese early union group. One of these had no fifth distal epiphysis, while the other had a very short fifth metacarpal (Fig. 3).

Shortened fifth middle phalanges are a very common feature of Down's syndrome (trisomy 21 or mongolism) (Hefke, 1940; Roche, 1961), and the shortening of this bone to the exclusion or virtual exclusion of all other hand bones is also occasionally found with cleidocranial dysostosis (Kohlmann, 1955), osteogenesis imperfecta (Brailsford, 1948), ovarian dysgenesis (Acheson, 1966), myositis ossificans progressiva (Singleton and Holt, 1954) and congenital heart disease (Schmid and Junker, 1950).

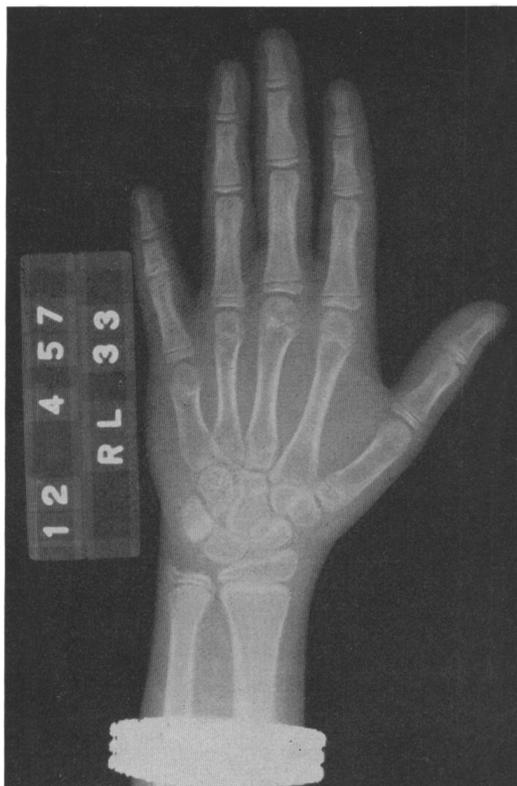


Fig. 3. Fifth middle phalanx showing epiphyseal union prior to other phalanges (Female, age 10)

Schmid and Junker feel that the presence of this trait is generally indicative of some more serious clinical condition (1950). Medical summaries of the twelve Chinese children having short (i. e. with a ratio of < 0.50) fifth middle phalanges did not indicate anything clinically remarkable, and the sole suggestion of the presence of any type of health impairment was provided by an apparent growth failure in two sisters, both being of the early union group. Applying Wetzel's method of utilizing growth measurements in the assessment of a child's physical condition (1941), it was found that both fell below the 98% auxodrome, and thus on the basis of Wetzel's standards, these girls were quite small. However, both of the sisters' parents were short, and Chinese children are, on the average, smaller than white children, which comprised the majority of Wetzel's material, so that their growth status can be explained by genetic considerations and is not necessarily indicative of any disease or nutritional factors. The absence of any indications of such factors, and the presence of shortened phalanges in three pairs of Chinese siblings, suggests that this is a form of inherited brachydactyly, brachymesophalangy 5, which reaches a high frequency in some populations. Anthropological discussion of this variation is found in Garn *et al.* (1967) and Hertzog (1967).

Discussion

Cone epiphyses in the proximal phalanges of the feet are common, being present in about 15% of a large British sample (Venning, 1961), and it is likely that some of the associations of cone epiphyses with other conditions (e.g., aseptic necrosis of the head of the second metatarsal or Freiberg's disease) are merely chance associations. In the hand, which is the concern here, cone epiphyses are associated with several distinct entities, specifically the Ellis van Creveld syndrome (chondroectodermal dysplasia) (Caffey, 1952), and cleido-cranial dysostosis (Brailsford, 1948), in addition to other uncertain associations with more nebulous conditions. Not only are cone epiphyses in the hand not diagnostic, they are not necessarily indicative of a pathological condition, being a feature of several types of genetic brachydactyly (Birkenfield, 1928; Haws, 1963). However, those types are quite rare, while the frequency of the cone epiphyses reported here is very high.

Cone epiphyses are currently thought to be the result of a differential vascular supply to the central and peripheral portions of the growth plate (Arkless and Graham, 1967), which in this case could represent some type of local genetic arterial deficiency. The apparent association of cone epiphyses with short diaphyses raises the question of whether some type of vascular consideration could also account for the latter. The only clue to a possible mechanism producing these short diaphyses is the finding that the time of onset of fifth middle diaphyseal ossification is considerably more variable than any other bone in the hand and on occasion is greatly retarded (Noback and Robertson, 1951; O'Rahilly and Meyer, 1956; Gray *et al.* 1957), with a possibility being that perhaps a postnatally short diaphysis is the result of a prenataally retarded center which is in turn due to a late vascular invasion of the cartilaginous shaft. In passing it is interesting to note that, on the basis of present meager knowledge, the neighboring phalangeal variants would not necessarily be contradictory to a suggestion concerning the primacy of vascular variation in the production of genetic osseous variation in this region. Dystelephalangy (Kirner's deformity or bilateral incurving of the terminal phalanges of the fifth fingers) is probably due to aseptic necrosis (McAfee and Donner, 1963) and McNutt (1962) has proposed underlying genetic arterial variation as the basic cause of the aseptic necrosis of the capital epiphysis of the femur (Legg-Perthes disease). Clinodactyly involves deficient ossification in the upper radial region of the fifth middle phalanx, and it has been suggested that this arrest may be caused by a local failure in the supply of the bone minerals (Hersh *et al.*, 1953). This vascular discussion has been presented largely because nothing is known concerning the etiology of human brachydactylies, and even if factors of arterial variation could apply in some instances, where the osseous involvement is restricted to a single bone, for example, clearly there are many forms in which such a factor could not.

As previously mentioned, there are numerous conditions where either the fifth middle phalanx can almost exclusively be shortened, or where cone epiphyses are seen in the hand, but most of these are easily differentiated by means other than

hand roentgenograms. However, there are three conditions in which shortened bones in hand roentgenograms are of primary diagnostic importance and in which other specific features may be absent; these three are peripheral dysostosis, brachydactyly and pseudo-pseudohypoparathyroidism (PPH). Peripheral dysostosis is a form of chondroplasia in which the abnormalities are mainly restricted to the tubular bones of the hands and feet, with the characteristic feature being cone-shaped phalangeal epiphyses. Mild cases can involve only one or two bones in the hand (Brailsford, 1948), and thus Fig. 1 could justifiably be considered a mild case of peripheral dysostosis. The distinction between peripheral dysostosis and brachydactyly is mainly semantic, and the first reported case of peripheral dysostosis could just as easily have been considered a new type of brachydactyly. If the phalangeal variation reported here is a distinct genetic entity and if there are no associated clinical abnormalities, then brachymesophalangy 5 is definitely the more desirable term.

The distinction between the remaining two conditions should not be merely semantic, because PPH is the incomplete manifestation of Albright's hereditary osteodystrophy (Mann *et al.*, 1962). The understanding of the difference between PPH and brachydactyly is important, if the shortened fifth metacarpal in Fig. 3 is indeed associated with the shortened fifth middle phalanx, and it would seem improbable that it is merely a chance association. The sibship involving the girl in Fig. 3 and her four-year-old sister, who exhibited just a shortened phalanx, could be placed in the first pedigree of brachydactyly presented by Birkenfield (1928), in which the roentgenogram of a child, age 7, exhibited shortened second to fifth metacarpals and shortened second and fifth middle phalanges with cone epiphyses. Arkless and Graham (1967) have pointed out that this pedigree in turn presents hand roentgenograms similar to PPH. McKusick (1966) considers that such similarity has resulted in confusing metacarpal brachydactyly with PPH, where shortened metacarpals are the most constant feature (Goeminne, 1965).

Despite the large literature on PPH and confusion regarding its diagnostic parameters, there has been little attempt to review or analyze the literature concerning types of brachydactyly which involve short metacarpals, in an effort to gain some insight into PPH. Allusions to this literature have been restricted to citations of Brailsford's familial brachydactyly, and Boorstein's and Birkenfield's types; however, all three presented two different pedigrees involving short metacarpals and/or metatarsals, and this type of nomenclature is imprecise and should be avoided. The definitive work on the subject is Bell's review of 113 pedigrees (1951), and her system of classification and terminology, although based on material published prior to 1949, is still the best and most widely used (McKusick, 1966). Of interest here is Bell's type E, in which the primary feature is the shortening of one or more metacarpals. Bell recognized that this is a mixed and probably heterogeneous group, and if metacarpal brachydactyly is to yield any information bearing on PPH, then some refinement of her type E is necessary.

At least two distinct subgroups can be recognized. In the first of these, comprising at least six of Bell's pedigrees, to which that presented by Hortling (1960)

may be added, the brachydactyly is limited to the fourth metacarpal and/or metatarsal, with no other metacarpal or phalangeal involvement. Of this subtype, the pedigrees of Boorstein (1926), Hortling (1960), and one of the numerous isolated cases not covered by Bell (where the relatives or offspring have not been sufficiently investigated), that of Fischer and Vandemark (1945), have been mentioned in the literature on PPH. For present purposes, this subtype may be provisionally designated type E1 brachydactyly. It should not be confused with the so-called "positive metacarpal sign" (Archibald *et al.*, 1959; Vartio and Meronen, 1961), although this sign could well be some type of intermediate manifestation of the same genetic entity.

The second subtype, which can be separated out of Bell's type E, includes Birkenfield's first pedigree (1928) and Brailsford's initial pedigree of familial brachydactyly (1945), to which the pedigree presented by McKusick and Milch (1964) can be added. An examination of the 17 individuals of these three pedigrees, for whom hand roentgenograms are presented, indicates that this subtype is characterized by involvement of a variable combination of metacarpals, plus a variable but characteristic combination of certain middle and distal phalanges, almost exclusively restricted to the first and third distal and the second and fifth middle phalanges. The important features seem to be (with a very few exceptions) involvement of both metacarpals and phalanges, considerable variation within pedigrees regarding the combination of affected bones in any given individual, cone epiphyses for the involved phalanges (Birkenfield, 1928), and an associated reduction in stature and a round face (McKusick and Milch, 1964; McKusick, 1966); it is this subtype which is discussed by McKusick (1966) under type E brachydactyly. Other pedigrees which Bell did not include in her type E (e.g., Klaussner's and one of Jeanselme's) probably belong to this subtype. Regarding terminology, the term familial brachydactyly has become entrenched in the literature, but it is devoid of any discriminating descriptive meaning, in that all brachydactylies are familial, and Brailsford used the same term for a second pedigree not having shortened metacarpals (1946). Brachymetacarpus (or brachymetapody) is not appropriate because one of Birkenfield's individuals had shortened second and fifth middle and first and third distal phalanges with no affected metacarpals. The ideal descriptive term is brachyphalangometacarpus, but it would seem more advisable to avoid additional confusion and simply designate this subtype by type E2 brachydactyly; Bell's system of terminology has the great advantage of being flexible while minimizing eponymous terminological confusion.

The characteristic pattern of phalangeal involvement in type E2 merits some further discussion. Fig. 3 suggested that shortened fifth metacarpals may occasionally be associated with shortened fifth middle phalanges. In the same manner, shortened fourth metacarpals and/or metatarsals also occasionally occur in pedigrees of shortened first distal phalanges (stub thumbs or brachytelophalangy 1) (Hefner, 1924; Stecher, 1957; Goodman *et al.*, 1965), while Temtamy reported a pedigree of brachydactyly involving only the second and fifth middle phalanges (McKusick, 1966). It is of interest to note that there are four bones in the hand which can and do exhibit independent inherited shortening without other phalangeal or metacarpal involve-

ment: the fifth middle phalanx, in clinodactyly (Bell's type A₃) and in the cone epiphyses-early union variation reported here (which can be provisionally designated type A₄ pending an investigation of its relationship to clinodactyly), the second middle phalanx (Bell's type A₂), the first distal phalanx (Bell's type D), and the fourth metacarpal (type E₁). One of the hand roentgenograms of type E₂ presented in McKusick and Milch (1964) has exactly these four bones involved as a group. It is this type of occasional association which confounds systems of typology, but which may provide important information concerning the nature of the developmental process in brachydactyly.

The residue of Bell's type E presents some difficulties. The problem is whether there is a valid category involving the shortening of a variable combination of metacarpals (as in E₂) without any phalangeal involvement (as in E₁). There are only three pedigrees which might be placed in such a group, for which hand roentgenograms are presented for two affected individuals. In one of these families, one individual clearly has shortened third and first distal phalanges (Boorstein, 1926), and is probably E₂, leaving the reports of Miskolczy (1929) and Friedlaender (1916) as a shaky basis for such a subtype, which could be designated type E₃.

The question now to be examined is whether this subclassification of metacarpal brachydactyly can provide any information bearing on the problem of pseudo-pseudohypoparathyroidism (PPH). PPH, the incomplete manifestation of Albright's hereditary osteodystrophy (AHO) (Mann *et al.*, 1962), exhibits most of the features of the fully developed pseudo-hypoparathyroidism (PH), except for the hypocalcemia and its resultant complications. The principal features of PPH, in approximately decreasing incidence, are shortened metacarpals, short stature, round face, relative obesity, slight mental retardation, and ectopic calcifications (Goeminne, 1965; Minozzi *et al.*, 1963; Cruz and Barnett, 1962). Mann *et al.*, (1962) reviewing the reported cases of PH and PPH to 1962, concluded that AHO is a sex-linked trait. Since that time, several cases of male-to-male transmission of PPH have been reported (Hermans *et al.*, 1964; Goeminne, 1965; Minozzi *et al.*, 1963), which, if true, would indicate that AHO cannot be sex linked but rather is an autosomal dominant trait. However, several workers (McKusick, 1966; Bartter, 1966) feel that these recent cases are not AHO, because they lack mental retardation, ectopic calcifications, and any case of PH in the family, and maintain that AHO is indeed sex linked.

The hand roentgenograms of those cases with reports of male-to-male transmission and of those with reported phalangeal involvement, as listed in the reviews of Goeminne (1965) and Minozzi *et al.* (1963), were examined. In a review of approximately 100 cases of PPH reported to 1965, Goeminne found some phalangeal involvement in about 10% of the cases (1965). Hermans *et al.* (1964) reported a large pedigree of PPH, having metacarpal involvement as the hand feature. They report that the ratio of females to males in their pedigree is 2 : 1 so that it is AHO and not Brailsford's familial brachydactyly (type E₂ here), but that there is an example of male-to-male inheritance, so that AHO cannot be sex linked. However, the sex ratio is largely due to the fact that 27 females and 16 males were examined, and the

hand roentgenograms of two of these individuals which are presented elsewhere (Gorman *et al.*, 1962) show the first, third, and fourth distal phalanges, and in the female, the second middle phalanx, to be shortened in addition to various combinations of metacarpals. The best evidence of male-to-male transmission is provided by the studies of Goeminne (1965) and Minozzi *et al.* (1963). The proband of Goeminne and his daughter have shortened second and fifth middle phalanges in addition to short metacarpals. Examination of the six individuals for whom Minozzi presents hand roentgenograms shows that four also have assorted terminal and middle phalangeal involvement, primarily the second and fifth middle and first and third distal (in two of these, all of the middle phalanges are reported to be affected, but the third and fourth appear normal). The case of McNeeley *et al.* (1956) is interesting because in addition to the standard pattern of shortened second and fifth middle and first and third distal phalanges, plus metacarpals, the fifth proximal phalanx is also shortened.

It would seem that those cases with reported male-to-male transmission have phalangeal involvement. Such involvement is rare in PPH (Goeminne, 1965), and indeed Mann *et al.* (1962) in their discussion of the differential diagnosis of AHO and myositis ossificans progressiva cited the lack of phalangeal abnormalities in AHO. This involvement follows the characteristic pattern described for type E2 brachydactyly, which is also associated with reduced stature and round faces. It is therefore felt that this discussion of hand roentgenograms supports the contention of McKusick (1966) that some of the cases reported in the literature as PPH are in fact examples of metacarpal brachydactyly, specifically type E2 as defined in this paper, and that the comments of Arkless and Graham concerning the restriction of the term PPH (1967) are definitely well taken. The possibility that genuine cases of PPH can be distinguished by the presence of roentgenograms of the type E3 variety deserves further attention.

Summary

Shortened fifth middle phalanges, defined as being less than half the length of the fourth middle phalanx, were found in 12 of 96 Philadelphia Chinese and none of 200 Philadelphia Negro children. At least nine of these can be characterized as having stunted cratered diaphyses, with precocious cone epiphyses, which underwent very early epiphyseal union.

The possible extension of a current vascular hypothesis for cone epiphyses, to include inherited osseous variation in this region of the little finger, is discussed.

A classification of metacarpal brachydactyly is presented, with the intention of investigating the syndrome of pseudo-pseudohypoparathyroidism. Reports of cases which have been held to negate the theory that Albright's hereditary osteodystrophy is sex linked were found to have the pattern of phalangeal shortening exhibited by one of the forms of metacarpal brachydactyly. This finding lends support to the contention of others that these reported cases are not pseudo-pseudohypoparathyroidism.

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RIASSUNTO

Un accorciamento della quinta falange mediale, corrispondente a meno della metà della lunghezza della quarta falange mediale, è stato riscontrato in 12/96 bambini cinesi ed in 0/200 bambini negri, tutti di Philadelphia. Almeno 9 di essi presentavano diafisi malformate, con epifisi precoci, e ad unione precoce.

Viene discussa la possibile applicazione di una corrente ipotesi vascolare sulle epifisi, alle variazioni ossee ereditarie di questa regione del mignolo. Viene presentata una classificazione della brachidattilia metacarpica, al fine di studiare la sindrome dello pseudo-pseudoipoparatiroidismo. I casi presentati in letteratura contro la teoria dell'eredità legata al sesso della osteodistrofia ereditaria di Albright comportavano l'accorciamento tipico di una delle forme di brachidattilia metacarpica, il che farebbe concludere che non si tratti di pseudo-pseudoipoparatiroidismo.

RÉSUMÉ

Un raccourcissement de la 5^{ème} phalange médiale, correspondant à moins de la moitié de la longueur de la 4^{ème} phalange médiale, a été remarqué chez 12/96 enfants chinois et 0/200 enfants nègres, tous de Philadelphia. Au moins 9 présentaient une malformation des diaphyses, avec précocité des épiphyses et des soudures.

La possibilité d'appliquer une hypothèse vasculaire courante sur les épiphyses aux variations osseuses héréditaires de cette région de l'auriculaire est discutée. Une classification de la brachydactylie métacarpienne est présentée, afin d'étudier le syndrome du pseudo-pseudo-hypoparathyroïdisme.

Les cas présentés dans la littérature, contrastant avec la théorie de l'hérédité liée au sexe de l'ostéodystrophie héréditaire de Albright entraînaient le raccourcissement typique d'une des formes de brachydactylie métacarpienne, ce qui ferait conclure qu'il ne s'agissait pas de cas de pseudo-pseudohypoparathyroïdisme.

ZUSAMMENFASSUNG

Bei 12 von 96 Chinesen— und 0 von 200 Negerkindern in Philadelphia wurde festgestellt, dass das fünfte Mittelglied der Hand auf weniger als die Hälfte der Länge des vierten Mittelglieds verkürzt war. Wenigstens 9 davon wiesen missgebildete Diaphysen mit vorzeitiger Epiphysen und vorzeitiger Verbindung auf.

Es wird Möglichkeit erwogen, eine bezüglich der Epiphysen verbreitete Gefäss-Hypothese auch auf die erblichen Knochenveränderungen an diesem Teil des Kleinfingers auszudehnen. Es wird eine Einteilung der Mittelhand-Kurzfingerigkeit vorgeschlagen, um das Syndrom des Pseudo-Pseudohypoparathyroidismus untersuchen zu können. Die in der Literatur beschriebenen Fälle gegen die geschlechtsgebundene Vererbungstheorie der erblichen Knochendystrophie nach Albright wiesen die typische Verkürzung einer der Mittelhand-Kurzfingerigkeitsformen auf, was darauf schliessen liesse, dass es sich nicht um Pseudo-Pseudohypoparathyroidismus handelt.