Distal brachyphalangy of the thumb in mental retardation

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Summary. Distal brachyphalangy (DBP) of the thumbs is a hereditary autosomal anomaly found in 1 to 1.5% of whites and in less than 1% of blacks. It was found in 26 of 852 mentally retarded patients in the present study; a frequency of over 3%. The male:female ratio was 70:30. There was bilateral DBP in 69.5% and unilateral DBP on the right hand in 19% and on the left in 11.5%. The 26 cases (100%) showed a subsyndromic aggregate of distal brachyphalangy of the thumb (DBT), short stature and mental retardation, 19 of them presented an abnormal head and 17 presented abnormal feet (nine of them also had convulsions). Six cases are discussed in detail; it is suggested that they belong to particular clinical categories.

In addition to gene abnormalities (which is the most likely cause) a possible explanation of the aetiological factors could be found in some sort of anomaly of the hypopituitary-hypothalmic area, since in the five cases where it was investigated, the sella turcica was small in four cases and large in one. Almost all known syndromes of which the subsyndrome discussed here is a part also show abnormal skulls, and this may add to the aetiology.

Shortness of the distal phalanx of the thumb is a relatively frequent hand anomaly. It may appear as an isolated trait or may accompany a more complex symptomatology. In this paper, we consider it in relation to oligophrenia, since we noted that the anomaly was found frequently among mentally retarded individuals.

The use of many different names has made it difficult to understand precisely what this anomaly is. Of course, shortness being the main feature in these cases, the prefix 'brachy-' seems most appropriate. Because we are referring to the distal phalanx of the thumb, distal or terminal brachyphalangy of the thumb (DBT) seems to be the most appropriate designation (Villaverde, 1940).

A short, distal phalanx may be found in one or both thumbs or big toes, and in one or more extremities. It may be found in varied combinations, with or without bone anomalies, include other fingers or toes and also other phalanges. Measurement of these phalanges has been done (Breitenbecher, 1923; Thomsen, 1927; Burrows, 1938; Poznanski et al, 1971) and the findings give the ratio between the proximal and distal ones as about 1:0.75. In the case of DBT we have found a ratio between 1:0.35 and 1:0.4, but this may vary; there are reports of a normal ratio (1:0.68) and a short ratio (1:0.44) in the same patient (Villaverde, 1943). Our findings are in general agreement with others that DBT consists of a normal first phalanx to which a distal phalanx of about two-thirds of its regular length is attached. The transversal diameters are practically within adequate limits, though at times the thumb has broader proportions particularly in the anteroposterior diameter, an increase due to the re-arrangement of fleshy parts. Diagnosis can be difficult when the abnormality is not too marked, but film measurements will solve such problems. Traumatic or infectious lesions to the distal portion of the thumb or big toe should not be mistakenly diagnosed as brachyphalangism. The criterion followed by us for the diagnosis of DBT has been the relative shortness of the distal
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phalanx when compared with the normal phalanx; the ratio being 1:0.4 or less.

**Classification and epidemiology**

Several attempts have been made to present a workable classification of abnormal digits, but the enormous amount of possible combinations of abnormalities have wasted these interesting attempts. A classification could be presented by grouping the most frequent findings of brachyphalangy corresponding to phalanges of any finger or toe. This has been the approach of Pfizter (1898) and Villaverde (1943), and more recently of Bell (1951), who made a modification of the former classifications, in which group D 'stub thumbs', includes DBT.

The global incidence can be estimated to be 1 to 1.5% of living persons. Pfizter et al (1898) found six patients (1.5%) with DBT in a series of 400; Villaverde (1943) recorded 16 (1.06%) among 1500 patients. DBT appeared more frequently in females (70%), and in people of short stature (75%); the incidence of bilateral or unilateral involvement was about the same. There is only one report (Villaverde 1943) of a coloured subject showing the trait. According to Stecher (1957), the incidence in the United States is 0.41% in Caucasians and 0.1% in Negroes and also a larger incidence among females. Manabe (1938) may have described cases among people in Japan. In Israel, Goodman et al (1965) found 1.6% among Jews and 3% among Arabs; however, the Arabs in this study belonged to 15 large kinships, thus decreasing the value of the percentage. The anomaly was unilateral in 50% of the cases and both sexes were equally affected. A significant increase of whorls on short thumbs was noted, and the anomaly was frequently accompanied with a short fourth toe; in Villaverde’s report (1943) clinodactyly of the fifth finger was more frequently found.

Many cases are familial and found even among large families. The transmission has been considered to be hereditary and autosomal recessive (Villaverde, 1943; Stecher, 1957). But other authors have stated that it is a dominant trait; recently, it has been presented as autosomal dominant with a low penetration of about 40% (Goodman et al, 1965).

**Subjects**

Eight hundred and fifty-two residents in an institution for the mentally retarded were screened for the anomaly;

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* The only black subject in the series.
cases of Down's syndrome were excluded. Twenty-six
presented one or both thumbs, and sometimes also big
toes, with distal brachyphalangism. This incidence, 3.12%, is substantially greater than that found among
other, non-retarded populations. There were 14 males
(54%) and 12 females (46%); a higher proportion of
males to females in contrast with other series, but adjusting
for the male:female ratio this gives 70:40 which is
close to the average.

Unilateral shortness was prevalent on the left side
(5:3). One case was coloured. In the institution there
are 856 whites and 145 blacks, thus the incidence for the
black population in only 0.73%. With this correction, the incidence for the white population increases to
3.6%.

A summary of the cases is presented in the Table.

Comments

Seventy-five per cent of cases with distal brachy-
phalangism of the thumb were also of short stature in a
previous, non-retarded, series (Villaverde, 1943). In the present series of retarded patients all
are of short stature (below the 10th centile); 19
(73%) of them are below the 3rd centile. In the institution there are residents of average height, and
even tall individuals, but none of these showed the anomaly. However, it must be remembered that
short stature is a common finding among mentally
retarded persons.

After 3 years of age, a head circumference of less
than 50 cm is small; about 42 cm is microcephalic.
In this series there are nine small heads; two of them
are microcephalic. A flat occiput was noted in four
cases. Only seven heads were considered normal.

Radiological investigations for skull and sella
 turcica were carried out in only five cases; there
were two small heads, two heads of average size, and
one large head. Four of the sellae were relatively
small; and the sella of one of the heads of average
size was somewhat larger than usual. In case 552,
both head and sella were rather small and there was
evidence of hyperostosis frontalis interna. It has
been previously stated (Villaverde et al, 1972) that
in most mentally retarded patients the sella are
rather smaller than for other populations.

Defective ears were noted in six cases, and one
half of all the cases showed some sort of symptomat-
ology from the visual organs: strabismus in nine,
blindness in two, microphthalmia and hypotelorism
in one, and one case each of microphthalmia or hypo-
telorism.

Eight patients showed some anomalies of the
mouth, high arched palate being the prevalent one.
Scoliosis or kyphoscoliosis was present also in eight
cases and abnormality of the chest in seven. Hoffman
(1924) mentioned hypertrichosis as a frequent
finding among his patients with DBT; but there
were only three instances in the present series.

Abnormalities of the feet, other than distal
brachyphalangism, were noted in 17 cases (65.5%);
abnormalities of the hands were not so frequent be-
ing present in eight (30.8%). Spasticity of the
limbs was present in 13 (50%) of the affected
patients. Twelve cases (seven females, five males)
were epileptic (46.2%).

Chromosome studies were performed in nine of
the cases; seven showed normal karyotypes. In
one case there was a pericentric inversion of a C-
group chromosome and in another a marker G
chromosome.

Diagnosis of the basic condition of the patients,
and the family histories were of no help in adding
substantial information about the aetiology of the
malformation. However, many accompanying
symptoms could add to our knowledge of the aeti-
ology. We would like to call attention to a few of
our cases.

Case 699 was a 19-year-old male, 1.18 m tall, with a
right short thumb. He had a short, small head, the left
side of the face was lower than the right, low set ears with
deficient lobes and stenosis of the canals, microphth-
amos, hypotelorism, proptosis of the eyelids, epicanthi,
high arched palate, irregular teeth, micrognathia, scoliosis,
possible amyotonia congenita. The chest was narrow
and excavatus. He had a simian crease of the right
hand, polydactyly, small third toe, syndactyly of the
second and third toes, talipes varus, spasticity of the ex-
tremities, right inguinal hernia, cryptorchidia with
atrophy of the testes, phymosis, large joints, and a para-
centric inversion of a C-group chromosome. His re-
tardation was not attributed to any known cause, until
the chromosome study was done. There are other
family members with mental retardation, and also poly-
dactyly and polythelia.

Case 974, a 15-year-old male, 1.26 m tall with both
thumbs short. He had a normal head, big ears, broad
hands and feet; no other striking signs or symptoms were
recorded. There are other cases of mental retardation in
his family. He had a marker G chromosome.

Case 552, a 39-year-old female, 1.28 m tall, 53.25 kg,
had both thumbs short. She had a very small brachy-
cephalic head, low set ears with absent antitragus, cata-
ract and strabismus, small mouth, beaked nose, short
distal phalanges of all fingers and toes, syndactyly of
fingers and second and third toes, pes cavus, atrophic
legs, normal karyotype, normal family. Her retarda-
tion is due to unknown cause.

Cases 264, 984, and 1071 also presented a wide range
of clinical symptoms.

Discussion

Distal brachyphalangy of the thumbs (and big
toes) is regarded by some authors as a minor con-
genital anomaly consistent with otherwise normal development and health. It is also a frequent finding accompanying well-known clinical complexes, such as Leri's pleonostosis, pycnody sostosis, Mohr's syndrome, Taybi's oto-pal digital syndrome, and Ellis-van Crevel, Smith-Lemli-Opitz, and other syndromes. Distal brachyphalangy is an autosomal, possibly recessive, hereditary anomaly of the skeleton; if it is dominant there is low penetration.

The cases presented here show very varied clinical pictures; each case in the series has four or more anomalies. The symptoms are more various than those reported in non-transported carriers of DBT. The basic finding, present in all cases (100%), is distal brachyphalangy of the thumb, short stature, and mental retardation. This dominant combination may well constitute a subsyndrome (da Silva and Villaverde, 1973), as this group of anomalies appear regularly as a part of more complex aggregates. Other frequent findings in the present series are: abnormal head (73%), abnormal feet (65.5%), and abnormal eyes (50%). Reports by other authors on anomalies found together with DBT were poorly substantiated in this series: hirsutism was present in only three cases (two males and one female); clinodactyly, in only one case; and shortness of the fourth toe was absent, although shortness of the third toe was found in one instance. DBT is an abnormality of the linear growth, we must therefore consider how frequently it appears with equivalent anomalies, particularly abnormal heads (73% in our series) including abnormal hypophyso-hypothalamic areas (as was assumed in the five examples in present series). In most of the syndromes or diseases showing DBT, there is also an abnormal head, eg, in pycnodysostosis, oto-palato-digital syndrome, Smith-Lemli-Opitz and other syndromes. Therefore, it seems possible to assume that some sort of injury to the growth and trophic centres in the hypothalamic area may help to delineate the subsyndrome independently of the most likely basic aetiologic factor of an abnormal gene.

In this series, the subsyndrome of DBT, short stature, and mental retardation also presents larger complexes. It is associated with an abnormal head in 19 cases (73%); abnormal feet in 17 cases (65.5%); and abnormal eyes in 13 cases (50%). The few cases which are described here in detail are so rich in clinical findings as to deserve further investigation. A search for similar patients would make it possible to accept or reject the existence of special clinical categories.

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Distal brachyphalangy of the thumb in mental retardation.

M M Villaverde and J A da Silva

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