Distal brachyphalangy of the thumb in mental retardation

MANUEL M. VILLAYERDE and JACYNTHO A. DA SILVA

From Rutgers University School of Medicine, New Jersey, USA

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-hypothalamic 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 apposite. 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 (Breiten-

becher, 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 infective 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 Pfitzner (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. Pfitzner *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;

TABLE

Case	Sex	Small Frame	Right Thumb	Left Thumb	Abnormal Head	Abnormal Occiput	Abnormal Ears	Abnormal Eyes	Abnormal Mouth	Abnormal Nose	Abnormal Spine	Abnormal Chest	Abnormal Hands	Abnormal Feet	Abnormal Chromosomes	Spasticity	Epilepsy	Abnormal Relatives	Additional Findings
153	M	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	
178	F	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	
263	F	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	
284	F	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	
332	M	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	
397	F	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	
489	M	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	
505	M	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	
551	F	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	
552	F	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	Atrophic legs
592	F	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	Hirsutism
607	M	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	Hirsutism; congenital pulmonary stenosis
640	M	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	Cryptorchidia; atrophic testes; large joints
699	M	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	Diplegia
927	M	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	
974	M	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	
980	M	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	
984	F	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	Diplegia; hypotonia
1008	F	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	Hirsutism
1047*	M	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	Diplegia; muscle atrophy
1071	F	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	Diplegia
1086	M	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	Diplegia
1177	M	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	Short left leg
1255	F	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	
1303	F	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	
1320	M	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	Diplegia

* 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 brachyphalangism 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 symptomatology from the visual organs: strabismus in nine, blindness in two, microphthalmia and hypotelorism in one, and one case each of microphthalmia or hypotelorism.

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 being 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 aetiology. 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, microphthalmos, hypotelorism, ptosis of the eyelids, epicanthi, high arched palate, irregular teething, 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 extremities, right inguinal hernia, cryptorchidia with atrophy of the testes, phymosis, large joints, and a paracentric inversion of a C-group chromosome. His retardation was not attributed to any known cause, until the chromosome study was done. There are other family members with mental retardation, and also polydactyly 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 brachycephalic head, low set ears with absent antitragus, cataract 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 retardation 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, pycnodysostosis, Mohr's syndrome, Taybi's oto-palat digital syndrome, and Ellis-van Creveld, 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-retarded 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); clynodactyly, 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 aetiological 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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