Stub thumbs in Israel revisited

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SUMMARY A three generation family with stub thumbs and short fourth toes is reported. This combination of anomalies has been observed in the past and the question is raised whether this association could be genetically distinct from that of stub thumbs alone.

In 1965 one of us (RMG) called attention to the relative commonness of stub thumbs (brachydactyly type D) among the various Jewish ethnic communities in Israel. At that time it was noted that the most frequent anomaly associated with this trait was a short fourth toe resulting from brachydactyly of the fourth metatarsal. Recently, we had the opportunity to see a family with stub thumbs in which this associated malformation also occurred. Since three generations were affected the full range of expressivity became apparent. The purpose of this brief report is to emphasise this association and to comment upon its possible genetic significance.

Family studies

The proband (fig 1) was a 35 year old Sephardi Jewish woman who presented to our dermatology clinic because of a fungus infection of the feet. At the time of examination it was noted that she had bilateral brachydactyly of the fourth toes and stub thumbs. After treatment she was referred to the genetic clinic where she and all her available family members were invited for examination. A summary of their physical findings is shown in the table and

selected anomalies can be seen in fig 2. No other physical abnormalities were noted. When possible x-ray studies of the hands and feet were done (fig 3).

FIG 1 Family pedigree.

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TABLE Clinical findings in affected family members.

Pedigree No	Sex	Age	Examined	Not Examined	Brachydactyly			
					Thumb		4th toe	
					Right	Left	Right	Left
II.12	F	72	+		-	-	+	+
III.2	M	42		+	-Hx	-Hx	+	+
III.3	M	45		+	+Hx	-Hx	+	+
III.5	F	38	+		+	+	+	+
IV.4	M	25	+		+	-	-	-
IV.13	F	13	+		+	+	+	-

+ = involved, - = not involved, Hx = affected or unaffected by history.

FIG 2 Normal thumbs and bilateral short fourth toes in II.12. III.5 shows bilateral stub thumbs and short fourth toes, while IV.13 shows bilateral stub thumbs and unilateral short fourth toe.

Discussion

Stub thumbs, which is a form of brachydactyly type D, is transmitted as an autosomal dominant trait.² Among the various Jewish ethnic groups in Israel this trait occurs in approximately 1.6% of all Jews irrespective of their communal origin. Although a

complete survey was not done in our original study,¹ four of 29 (13.8%) unrelated Jewish persons were noted to have a short fourth toe. Stetcher,³ in his survey of thumbs among US whites (0.41% affected) and blacks (0.1% affected), observed that one in 95 had a short fourth toe. Despite these observations on the association of a short fourth toe

FIG 3 X-rays showing brachydactyly of the fourth metatarsal bilaterally in II.12 and III.5 and unilateral involvement in IV.13. IV.13 shows brachydactyly of the distal phalanx of the thumb.

with stub thumbs, we believe that a proper survey on the incidence of this finding with stub thumbs should be done. It can be stated that such an association has been noted in several Jewish families from Israel. The wide range of expressivity observed in our family (table), varying from a single thumb affected (IV.4) to both thumbs and both fourth toes (III.5), is compatible with that seen in most autosomal dominant disorders. The question could be raised as to whether this combination of findings, stub thumbs and short fourth toes, is a distinct entity resulting from a different gene from that causing just stub thumbs. Linkage studies may be helpful in answering this question. In conclusion, from our clinical experience it would seem that there is a definite association between the trait of stub thumbs and short fourth toes, and whether this is a distinct genetic entity from that of stub thumbs alone remains to be answered. Population studies on the incidence of these two traits among Jews and other ethnic groups may be rewarding in terms of use as a genetic marker.

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References

- 1 Goodman RM, Adam A, Sheba C. A genetic study of stub thumbs among various ethnic groups in Israel. *J Med Genet* 1965;2:116-21.
- 2 McKusick VA. *Mendelian inheritance in man*. 6th ed. Baltimore: Johns Hopkins University Press, 1983:77.
- 3 Stecher RM. The physical characteristics and heredity of short thumbs. *Acta Genet (Basel)* 1957;7:216-22.

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