Polydactyly: a study of a five generation Indian family

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Abstract

Preaxial polydactyly was observed in up to five generations of an Indian family living in a village in the Rajkot district (Gujarat). Among the 71 affected members, 45 were males and 26 were females. All these affected members showed preaxial polydactyly manifesting as a well formed, articulated extra digit of the hand or foot. Twenty other cases were

also identified with polydactyly involving triphalangeal digits replacing the thumbs or duplication of the big toe(s). To the best of our knowledge, the present family is the largest in which several members have preaxial polydactyly of different types. No other abnormalities were apparent. The present study strongly suggests that preaxial polydactyly with a well formed extra digit,

Figure 1 Pedigree of the family.

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triphalangeal thumbs, and duplication of the big toe can be manifestations of the

same autosomal dominant gene. It is likely that other factors are modifying the expression of this gene. (J Med Genet 1993;30:296-9)

Table 1 Details of affected subjects.

No	Pedigree No	RH	LH	RF	LF	Remarks
1	I-1	6	6	5	5	
2	II-1	6	6	5	5	
3	II-3	5	5	5	5	TT
4	II-5	6	6	5	5	
5	II-7	6	6	5	5	
6	II-9	5	5	5	5	TT
7	II-13	6	6	5	5	
8	II-15	6	6	5	5	
9	II-19	6	6	6	6	
10	III-3	6	6	5	5	
11	III-5	6	6	5	5	
12	III-9	6	6	5	5	
13	III-14	5	5	5	5	TT
14	III-17	5	5	5	5	TT
15	III-19	6	6	5	5	
16	III-20	6	6	5	5	
17	III-21	6	6	6	6	
18	III-22	6	6	5	5	
19	III-26	5	5	5	5	TT
20	III-28	5	5	5	5	TT
21	III-35	6	6	5	5	
22	III-37	6	5	5	5	
23	III-39	6	6	5	5	
24	III-41	5	5	5	5	TT
25	III-45	5	5	5	5	TT & DB
26	III-47	5	5	5	5	TT
27	III-49	6	6	5	5	
28	III-53	6	6	5	5	
29	III-57	6	5	5	5	TT
30	III-59	6	6	5	5	
31	III-70	6	6	6	6	
32	III-72	6	6	6	6	
33	III-74	6	6	5	5	
34	III-76	6	6	5	5	
35	IV-8	6	6	5	5	
36	IV-10	6	7	5	5	
37	IV-12	5	5	5	5	TT
38	IV-13	6	6	5	5	
39	IV-15	5	5	5	5	TT
40	IV-29	6	6	5	5	
41	IV-31	6	6	5	5	
42	IV-32	6	6	5	5	
43	IV-33	6	6	5	5	
44	IV-35	5	5	5	5	TT
45	IV-54	6	6	5	5	
46	IV-55	6	6	5	5	
47	IV-56	6	6	5	5	
48	IV-57	5	5	5	5	TT & DB
49	IV-59	6	6	5	5	
50	IV-60	6	6	5	5	
51	IV-63	5	5	5	5	TT
52	IV-64	6	6	6	5	
53	IV-67	6	6	5	5	
54	IV-68	6	6	5	5	
55	IV-70	6	6	5	5	
56	IV-71	6	6	5	5	
57	IV-72	6	6	5	5	
58	IV-80	6	6	5	5	
59	IV-82	5	5	5	5	TT & DB
60	IV-83	6	7	6	6	
61	IV-85	6	6	5	5	
62	IV-87	6	6	5	5	
63	IV-89	6	5	5	5	
64	IV-103	6	6	5	5	
65	IV-105	5	5	5	5	TT
66	IV-106	6	6	5	5	
67	IV-108	6	6	5	5	
68	IV-110	6	6	5	5	
69	IV-111	6	6	5	5	
70	IV-112	6	6	5	5	
71	IV-114	6	6	5	5	
72	IV-116	6	6	5	5	
73	IV-118	5	5	5	5	TT & DB
74	IV-120	6	6	5	5	
75	IV-121	6	6	5	5	
76	IV-124	6	6	5	5	
77	IV-126	6	6	5	5	
78	V-2	6	6	5	5	
79	V-5	6	6	5	5	
80	V-11	6	7	5	5	
81	V-12	6	6	6	6	
82	V-15	7	5	5	5	
83	V-19	5	7	5	5	
84	V-20	5	5	5	5	TT
85	V-21	6	5	5	5	
86	V-22	6	6	5	5	
87	V-23	5	5	5	5	TT & DB
88	V-27	5	5	5	5	TT
89	V-29	6	6	5	5	
90	V-30	6	6	5	5	
91	V-32	6	6	5	5	

RH=right hand, LH=left hand, RF=right foot, LF=left foot, TT=bilateral triphalangeal thumbs, DB=unilateral duplication of big toe (either side).

This paper reports on the largest known Indian family with preaxial polydactyly consisting of a well formed extra digit, a triphalangeal digit in place of the thumb, duplication of the big toe, or hexadactyly, representing a high degree of genetic variability. All affected subjects are otherwise normal in all respects.

Family report

The family studied were inhabitants of a village in the Rajkot district of Gujarat. Among 265 subjects in five generations of the pedigree (fig 1), there were 45 affected males and 26 affected females who had polydactyly with variable expression (table 1). Figs 2 to 8 show

Figure 2 III-39: bilateral fully developed extra finger on both hands.

Figure 3 III-57: six fingers on the left hand and five fingers with triphalangeal thumb on the right hand.

Figure 4 III-59: fully developed extra fingers on the left and right hands.

Figure 5 IV-83: bilateral polydactyly with seven fingers on the right hand and six fingers on the left hand.

Figure 6 V-5: six fingers on both hands with unilateral duplication of the big toe.

Figure 7 V-20: bilateral triphalangeal thumbs.

Figure 8 V-21: polydactyly with six fingers on the right hand and triphalangeal thumb on the left hand.

the various types of polydactyly in different subjects. Of the 71 members with polydactyly, five were symmetrically affected in both hands and feet, whereas 56 subjects had six digits bilaterally on their hands only. The rest of the affected subjects ($n = 10$) showed asymmetry in the number of digits in their hands or feet or both. In addition to those with a well developed extra digit, 20 other subjects had bilateral triphalangeal thumbs or unilateral duplication of the big toes or both, indicating a high degree of phenotypic variation.

Marriages between affected and normal subjects produced affected and normal progeny in a 1:1 ratio. This clearly suggests that the polydactyly is caused by an autosomal dominant gene (table 2). Further, distribution of polydactyly in the two sexes clearly indicates that the expression of the trait is in no way influenced by or linked to the sex of the subject (table 3). Male to male transmission was observed in several instances.

The majority of the family members were agricultural labourers but this anomaly does not interfere with their work. According to the information available and our observations, there were no other malformations, parental consanguinity, or reduction in reproductive fitness in the family.

The present data support autosomal dominant inheritance with variable expression.

Discussion

The incidence of polydactyly in the general population shows quite wide variation among various racial groups ranging from 0.37 to 1.2 in 1000 live births.¹⁻⁴ The incidence in embryos studied through induced abortions in Japan was found to be 0.35%.⁵ Similar information is not available for the Indian population.

Published reports have suggested that preaxial polydactyly involving duplication of the thumbs/big toes, triphalangeal thumbs/duplication of the big toes, and polydactyly of an index finger are three different types of autosomal dominant preaxial polydactyly. However, all three phenotypes are present in the family reported here. In the family under study, parents with a triphalangeal thumb (fig 7) and duplication of a big toe did not produce offspring with a fully developed extra finger or toe. They produced the same type or normal offspring. However, the parents with preaxial

Table 2 Distribution of preaxial polydactyly in offspring from marriages in four generations.

Generation	Parents	No of marriages	Offspring		Calculated χ^2 value
			Affected	Normal	
I	Affected $\times$ normal	1	8 (6.5)	5 (6.5)	0.6923
II	Affected $\times$ normal	8	25 (25.5)	26 (25.5)	0.0196
III	Affected $\times$ normal	16	43 (42.0)	41 (42.0)	0.0476
IV	Affected $\times$ normal	15	14 (18.0)	22 (18.0)	0.4444

Figures in parentheses indicate expected numbers in each group. All calculated χ^2 values were statistically non-significant ($p < 0.05$).

Table 3 Distribution of preaxial polydactyly in the two sexes.

Generations	Sex		Calculated χ^2 value
	Male	Female	
II	5	3	0.50
III	14	11	0.36
IV	27	16	2.8139
V	7	7	0.0000

All calculated χ^2 values are statistically non-significant ($p < 0.05$).

polydactyly with fully developed triphalangeal digits in addition to the thumb produced children with a similar type and/or triphalangeal thumb and duplication of the big toe polydactyly, perhaps suggesting a degree of genetic anticipation.

Graham *et al*⁷ reported absence of the thenar intrinsic muscles in four unrelated families with preaxial thumb polydactyly, which was associated with inability to flex the thumb across the palm. However, such observations were not made in the present family, since the extra digit was functional and flexed like a normal finger.

Occurrence of preaxial polydactyly of a well formed extra digit, triphalangeal thumbs, and duplication of the big toe in the same pedigree

indicates a common genetic basis responsible for all the varieties. The asymmetry or variation in expression may be the result of involvement of modifying genes, polygenes, or some unrecognised effect on the developmental process. The data presented in table 2 clearly indicate that the mode of inheritance of polydactyly in the present study is autosomal dominant.

The distribution of polydactyly in the two sexes is presented in table 3. These findings indicate an even distribution of polydactyly in both sexes. The number of affected males was found to be higher than affected females in different generations, but on statistical analysis this was found to be non-significant. Furthermore, the pedigree clearly indicates that there is equal transmission of the trait to both sexes from affected male or female parents. This suggests that the expression of polydactyly is independent of sex. Temtamy⁶ also reported equal expression of this trait in the two sexes.

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