Dermatoglyphs and chromosome mosaicism in parents of children with trisomy 18

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SUMMARY Parents of patients with Down's syndrome show dermatoglyphic features intermediate between the affected and the normal population. Dermatoglyphs were studied on the parents of 19 cases of trisomy 18, but no similar 'intermediate' traits were discovered. The number of cases studied needs to be enlarged before it can definitely be stated that trisomies 18 and 21 differ in this respect.

Certain dermatoglyphic patterns are highly characteristic of infants with trisomy 18 (Edwards's syndrome),¹ but we know of no published data on the dermatoglyphic patterns of their parents. It seemed appropriate to look at the dermatoglyphs of such a group. It is known that, in another chromosome anomaly, trisomy 21 (Down's syndrome), parents of such children can occasionally be shown to be mosaic for trisomy 21.^{2,3} Children with this condition also have characteristic dermatoglyphic patterns¹ and children who are mosaic for Down's syndrome have patterns which are 'intermediate' between those of regular trisomy 21 and those of the general population.⁴ Some parents of regular

trisomy 21 patients also have these 'intermediate' dermatoglyphic patterns.⁵ The relationship between these facts is not clear but there is definitely an increased risk of a second Down's syndrome infant occurring after the birth of one with regular trisomy 21.⁶ We have examined trisomy 18 families for dermal ridge patterns and for chromosome mosaicism in the parents; we have analysed the sibships in these families for information on recurrence risks.

Subjects, methods, and results

Since 1969, 46 families with regular trisomy 18 have been identified in the Oxford region. Of those whom we could contact, 19 agreed to co-operate in the study. Fingertip and palmar patterns were examined

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TABLE *Finger and palm patterns of parents of trisomy 18 infants compared with controls and trisomic infants*

	Controls ^{1 7-11}	Parents of trisomy 18 infants	Trisomy 18 infants ¹ (+ our data)
<i>Fingers</i>			
Arches (mean No per 10 digits)	0.54	0.43	7.61
Whorls (mean No per 10 digits)	2.69	2.16	0.05
Radial loops	0.15%	0%	4.84%
thumb			
index finger	22.25%	36.49%	1.41%
middle finger	3.55%	1.35%	4.23%
ring finger	0.65%	2.70%	1.35%
little finger	0.20%	0%	2.86%
Total finger ridge count			
M	139.04	139.61	very low
F	126.30	130.79	very low
<i>Palms</i>			
Presence of t'	22.5%	21.6%	38.5%
t''	10.0%	8.1%	23.1%
H	16.2%	13.5%	13.3%
H ⁺	29.7%	18.9%	10.3%
H ⁺	2.5%	1.35%	0%
III/IV	94.6%	94.5%	87.1%
Thenar exit of A line	3.5%	0%	26.0%

A 5% level of significance was not reached in any comparison of parents with controls.

and 29 parents gave blood samples for chromosome analysis.

The dermatoglyphic features of trisomy 18 and our findings in controls and parents of trisomy 18 children are listed in the table.

The parents did not show an increased incidence of arches but they did show fewer whorls compared to controls. The distribution of radial loop patterns was very similar to that of the general population. Total finger ridge counts were also close to the population mean. Palmar patterns did not show a significant difference from controls.

We counted an average of 27 metaphases per parent (range 8 to 70). A single 47,+18 cell was found among 70 in only one subject; the other 69 cells had counts of 46 with normal 18s. This man had arch patterns on four of ten fingertips and had a low total finger ridge count (73). He had no other finger or palm characteristics of trisomy 18.

Mean maternal age (at birth of the trisomy 18 infant) was 32.34 years (SD 6.54) and mean paternal age was 35.17 years (SD 6.10 years); 11 of the 19 mothers were aged less than 35 years at the birth of the affected infant. Pregnancies before the trisomic child comprised 15 normal infants and seven spontaneous abortuses. Eight subsequent pregnancies produced normal infants. Amniocentesis was carried out in all.

Comment

We have been looking for 'intermediate dermal patterns' in parents of regular trisomy 18 children and for evidence that some parents could be mosaic for this trisomy. In these respects our findings to date are negative, and there were no recurrences of trisomy 18 within sibships of the affected child. No recurrences of trisomy 18 were found in a previously reported amniocentesis survey.⁶ However,

our sample is small and dermatoglyphic and chromosome abnormalities might be detected in a larger survey. Since guidance on recurrence risk to sibs would be useful in counselling, we plan to extend our survey and would welcome information and co-operation from other centres.

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