Dermatoglyphs in carriers of a balanced 15;21 translocation

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SUMMARY Cytogenetic and dermatoglyphic features were studied in a large family with an inherited 15;21 translocation. Of 35 healthy members of the family, 21 carried the translocation chromosome and 14 were chromosomally normal. There were six members with Down's syndrome who had the translocation. Dermatoglyphic studies showed that carriers of this balanced translocation had the following peculiarities significantly more often than the general population. On the hands, they had ulnar loops on the fingertips, symmetrical high terminations of the A line, symmetrical ulnar loops on the hypothenar areas, distal loops in the 3rd interdigital areas, open fields in the 4th interdigital areas, axial triradii in the distal position, and single transverse palmar creases (Sydney lines). On the feet, they had small distal loops on the hallucal area and distal loops in the 4th interdigital areas.

The translocation carriers also had significantly more often than non-carrier relatives symmetrical high terminations of the A line, open fields in the 4th interdigital areas, distal axial triradii, and Sydney lines. On the feet, they had small distal loops on the hallucal areas, distal loops in the 4th interdigital areas, and tibial loops on the proximal hypothenar areas. The data obtained from this study, and especially the values of the Walker and general indices, indicate that some of the dermatoglyphic stigmata of Down's syndrome are directly associated with the 15;21 translocation carrier state and can therefore be used for predicting that state.

Several diagnostic indices based on the relative frequencies of various dermatoglyphic patterns on the fingertips, palms, and soles have been proposed for the dermatoglyphic diagnosis of Down's syndrome.^{1–9} The dermatoglyphs in translocation types of Down's syndrome were found to resemble closely those in free trisomy cases.^{10–12} However, the dermatoglyphic data in phenotypically normal carriers of balanced 13–15;21 translocations have been conflicting.

We have tried to clarify this question by making a detailed description of all the dermatoglyphic features on the fingertips, palms, and soles in a large family with an inherited 15;21 translocation.

Cytogenetic and dermatoglyphic findings

We studied the cytogenetic and dermatoglyphic data for a large family spread over four generations, in which six members had features of Down's syndrome

and 21 phenotypically normal members had the same 15;21 translocation with a chromosome count of 45. Normal karyotypes were found in 14 other phenotypically normal close relatives. The pedigree data are given in fig 1. The dermatoglyphic findings in this family are compared with those in 150 patients with free trisomy 21 and in a control series of 552 phenotypically and chromosomally normal persons from southern Germany. The statistical evaluation of the general index included 30 features of the palms and soles (Walker index = 16) as described earlier.^{8,9}

FINGERTIP PATTERNS (TABLE 1)

The translocation carriers had similar relative frequencies of the various fingertip patterns to the controls and the normal relatives. However, there was a high frequency of ulnar loops on the right 2nd finger (43%, compared to 33.3% in the controls and 21.4% in the normal relatives) and of radial loops on both 4th fingers (4.8, 0.2, and 0%, respectively), and a decrease of radial loops on the 2nd fingers

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FIG 1 Pedigree of family with inherited 15;21 translocation.

TABLE 1a Fingertip patterns (%) in trisomy 21 patients, carriers of balanced 15;21 translocation, their normal relatives, and normal controls

Fingertip patterns	Trisomy 21 (n=1500)	t(15;21) (n=210)	Normal relatives (n=140)	Controls (n=5520)
Whorls (W)	16.1	35.7	28.6	30.7
Ulnar loops (U)	79.7	58.6	65.0	58.6
Radial loops (R)	2.8	3.8	4.3	4.5
Archs (A)	1.5	1.9	2.1	6.2

TABLE 1b Fingertip patterns (%) on the 2nd and 4th fingers in trisomy 21 patients, carriers of balanced 15;21 translocation, their normal relatives, and normal controls

Fingertip patterns	Trisomy 21	t(15;21)	Normal relatives	Controls
U, 2nd finger	91.0	43.0	21.4	33.3
R, 4th finger	7.8	4.8	0.0	0.2
R, 2nd finger	0.5	4.8	21.4	20.5

(4.8, 20.5, and 20.4% respectively). These differences were statistically significant ($p < 0.05$).

PALMAR PATTERNS (TABLES 2 AND 3)

Higher frequencies of right-left symmetry in the terminations of the main palmar lines were found in the translocation carriers than in the controls and normal relatives: the A line in five, ($p < 0.01$ and 0.05 , respectively); the B line in seven ($p < 0.05$ for both); the C line in nine ($p < 0.05$ for both); and the D line in 11 ($p < 0.05$ for both).

The mean value of the combined right and left *atd* angles of the translocation carriers (90.18 ± 23.91) was higher than in the non-carriers (74.07 ± 18.81). The mean value for the control series was 88.07 ± 18.70 . The surprisingly low mean value of the *atd* angle of the chromosomally normal family members was the result of a familial dislocation of the *a* triradii to the ulnar side, a trait that also appeared in the translocation carriers. The high mean value of the *atd* angles in carriers of the balanced translocation could be explained by the frequent occurrence of distally placed axial triradii *t''* which was found in 19.0% of the palms. The controls had

TABLE 2 Frequency of right/left symmetry of main line terminations in trisomy 21 patients, carriers of balanced 15;21 translocation, their normal relatives, and normal controls (%)

Main lines	Trisomy 21 (n=150)	t(15;21) (n=21)	Normal relatives (n=14)	Controls (n=552)
A5'/A5'	35.3	33.3	21.4	15.8
B7/B7	66.0	28.6	21.4	23.7
C9/C9	48.0	33.3	21.4	20.5
D11/D11	74.0	33.3	21.4	26.6

TABLE 3 Frequency (%) of palmar patterns in trisomy 21 patients, carriers of balanced translocation, their normal relatives, and normal controls*

Palmar patterns	Trisomy 21 (n=150)	t(15;21) (n=21)	Normal relatives (n=14)	Controls (n=552)
Hypothenar				
L ^u /L ^u	38.7	14.3	7.1	3.4
L ^r /L ^r	2.0	4.8	0.0	15.2
Thenar				
O/O	83.7	85.7	71.4	65.9
W+L/				
W+L	1.4	7.1	21.4	9.8
III IDR				
L ^a /L ^a	48.0	38.1	28.6	18.2
IV IDR				
L ^a +D/				
L ^a +D	9.3	23.8	64.2	44.2
V+O/				
V+O	60.7	38.1	7.1	12.5

*Nomenclature from Penrose LS. Memorandum on dermatoglyphic nomenclature. *Birth Defects* 1968;6:No 3.

such t'' triradii only in 8.5% and the normal relatives in 0%. These differences were statistically significant ($p < 0.05$).

The incidence of symmetrical ulnar loops on the hypothenar area was significantly higher in the balanced carriers than in the controls ($p < 0.001$) or in the normal relatives ($p = 0.05$). The incidence of radial loops, however, was clearly decreased ($p < 0.05$). There were fewer true thenar patterns (loops and whorls) in the balanced carriers than in the controls ($p > 0.05$) or the normal relatives ($p < 0.05$), and a markedly higher frequency of symmetrical open fields ($p < 0.05$).

The balanced carrier group had a much higher frequency of symmetrical distal loops in the 3rd interdigital areas than did the controls or the normal relatives (both $p = 0.05$), and a significantly lower frequency of symmetrical true patterns in the 4th interdigital areas ($p < 0.05$ for both). No family member showed a typical simian crease. The incidence of symmetrical atypical forms was clearly higher (28.6% had a Sydney line and 14.3% an abortive form) in the balanced translocation carriers than in the controls (0.4% had a Sydney line and 2.3% had abortive forms) or in the normal relatives (0%). The differences were highly significant for both ($p < 0.001$).

PLANTAR PATTERNS (TABLE 4)

The hallux in the balanced carriers had a markedly higher frequency of symmetrical small distal loops (< 20) than that in the controls ($p < 0.001$) or the normal relatives ($p = 0.05$), and fewer symmetrical whorls ($p < 0.05$ for both). There were also higher frequencies of distal loops in the 4th interdigital area ($p < 0.05$), and of tibial loops on the distal hypothenar area ($p < 0.05$).

Discussion

Some authors, such as Penrose and Delhanty,¹⁰

TABLE 4 Frequency (%) of plantar patterns in trisomy 21 patients, carriers of balanced translocation, their normal relatives, and normal controls

Plantar patterns	Trisomy 21 (n=150)	t(15;21) (n=21)	Normal relatives (n=14)	Controls (n=552)
Hallux				
l^d/l^d	37.0	28.6	7.1	3.4
W/W	3.5	4.8	14.3	19.0
II IDR				
O/O	57.0	38.1	35.7	27.2
LP/LP	7.0	9.5	14.3	12.3
IV IDR				
L^d/L^d	38.0	42.9	21.4	8.2
O/O	0.0	23.8	50.0	41.5
Hypothenar distal L^t/L^t	58.5	71.4	28.6	38.8

Walker *et al.*,¹³ and Fung and Zavattone,¹⁴ found in carriers of the balanced 13-15;21 translocation a combination of dermatoglyphic patterns quite similar to those in Down's syndrome. The combination included increased frequencies of ulnar loops on the fingertips, bilateral transverse single creases, distally placed axial triradii, high atd angle values, and large hypothenar patterns on the palms. Based on their results, Penrose and Delhanty¹⁰ postulated that "the major genetical determinants of the hypothenar patterns and height of the axial triradius are located on chromosome 21 and that the presence of an allele upon the fused D/G chromosomes tends to centralize the triradius t in distal position t' and/or t'' ." In contrast, Sergovich *et al.*¹⁵ found no difference between the mean atd angles in eight carriers of balanced 13-15;21 translocations and 13 non-carrier relatives. However, these authors did not study any other dermatoglyphic patterns and furthermore it seems possible that they were studying a different type of translocation because the acrocentrics could not be identified by banding techniques.

In our material the following atypical dermatoglyphic patterns were found in the carriers of the balanced 15;21 translocation compared with the controls and the chromosomally normal relatives. On the hands, there were frequent ulnar loops and fewer radial loops on the 2nd finger; a greater frequency of radial loops on the 4th finger; more frequent symmetry of the A line ending in five, the B line in seven, the C line in nine, and the D line in 11; and increased frequency of all the following: ulnar loops on the hypothenar area, open fields in the thenar area, distal loops in the 3rd interdigital area, rudimentary patterns (vestigial) in the 2nd and 4th interdigital areas, distal axial triradii, Sydney lines, and abortive forms of simian creases. On the soles, there were small loops on the hallux; open fields in the 2nd and 3rd interdigital areas; distal loops in the 4th interdigital areas; and tibial loops in the hypothenar areas were increased. These combinations of dermatoglyphic patterns found in translocation carriers resembled those found in Down's syndrome. Great symmetry of the dermatoglyphic patterns on the right and left palms in patients with Down's syndrome has been reported.^{6,8,9} This strong tendency to bilateral symmetry of the patterns in Down's syndrome patients accounts for the statistical significance of differences from the balanced carrier group in the frequencies of fingertip patterns: B line termination in seven, C line termination in nine, D line termination in 11, distal axial triradii, and distal loops in the 3rd and open fields in the 4th interdigital areas (all on the palms), and small loops on the hallux. No significant differences were found in the terminations of the A line or in the

hypothenar pattern on the hands, or in the patterns of the 2nd and 4th interdigital areas and hypothenar areas on the feet. Generally, the group of balanced carriers was intermediate between the Down's syndrome patients and the controls with regard to the courses of the main lines and the patterning on the fingertips, palms, and soles.

The combination of a distally placed axial triradius and large hypothenar patterns was found bilaterally in ten and unilaterally in two of the 21 translocation carriers. An unusual finding was the high incidence of a single transverse crease (especially a Sydney line) on the palm (11 bilaterally and three unilaterally) and a small distal loop on the hallux (six bilaterally and three unilaterally). It is therefore interesting in this context to examine how often these dermatoglyphic peculiarities were combined in the same translocation carrier. Twelve carriers had a simian crease, distally placed axial triradius, and hypothenar pattern, but only three carriers had the combination of small distal loops on the hallux, a distal axial triradius, and hypothenar pattern.

The combination of dermatoglyphic patterns and the degree of the stigmata of Down's syndrome can be mathematically expressed by the 'Walker' and the 'general' indices.^{1 2 8 9} The calculation of the Walker index values in the carriers showed seven values characteristic of the normal population (range -3 to -9) and 14 in the region of overlap (-3 to +3) between normal and Down's syndrome populations (fig 2). Eight normal relatives had index

FIG 2 Histograms of the Walker indices based on digital, palmar, and plantar patterns.

FIG 3 Histograms of general indices based on symmetrical/asymmetrical relationship of palmar and plantar patterns.

values of the normal population and six of the overlap region. The values of the general index in the balanced translocation carriers agreed better with those in Down's syndrome patients: eight carriers had values typical of Down's syndrome (+0.5 to +19), six typical of the overlap region (+0.5 to -2), and only seven typical of the normal population (-2 to -15) (fig 3). The data presented from this family indicate that the state of having a balanced 15;21 translocation is often associated with distinctive combinations of dermatoglyphic patterns. Further investigations will be necessary to elucidate whether translocations between other acrocentric chromosomes (for example, 13;21 and 14;21) cause similar dermatoglyphic patterns in the balanced carriers.

The existence of special genes on chromosome 21, which cause t' or t'' triradii, or both, in the case of a translocation, as postulated by Penrose and Delhanty,¹⁰ seems rather improbable, because the theoretical interpretation of the effect of such a changed gene position is very difficult. Furthermore, the unusual combination of hypothenar patterns with t'' triradii is not present in all carriers of balanced 13-15;21 translocation but can be found in almost all other autosomal aberrations. The same feature can also be observed in 5% of normal subjects.⁸ A more probable explanation for the increased occurrence of the dermatoglyphic peculiarities in carriers of 13-15;21 translocations described here seems to be a general disturbance of regulatory mechanisms resulting from the loss of genes on the

fused acrocentric chromosomes. Similar effects are caused by other genetic derangements, especially chromosomal aberrations.

Since dermatoglyphic stigmata typical of Down's syndrome appeared in two-thirds of the 15;21 translocation carriers in this family, we suggest that dermatoglyphic examination might detect carriers in other 15;21 translocation families. Furthermore, we have some evidence that similar stigmata also appear in carriers of other translocations involving a chromosome 21. We therefore feel that dermatoglyphic examination should be used to detect carriers of such balanced translocations in the normal population.

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