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Dermatoglyphic features in Prader-Willi syndrome with respect to chromosomal findings

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Abstract

Dermatoglyphic findings were compared in 38 Prader-Willi syndrome (PWS) patients and 270 normal controls. Twenty-one of the PWS patients had an interstitial deletion of the proximal long arm of chromosome 15 and seventeen PWS cases had normal chromosomes. Findings in PWS are not diagnostic but do show some consistent deviations that can be used in the clinical evaluation of PWS patients. These include a displacement of the axial triradius away from the normal proximal position, an excess of whorls primarily on the thumbs, radial termination of the palmar A mainline, and lack of arches on the big toe. Deletion PWS patients were much more homogeneous than non-deletion cases with respect to plantar patterns. The previously reported deficit of plantar pattern intensity was restricted only to deletion PWS and was characterized by a lack of plantar interdigital II–IV patterns with almost exclusively hallucal distal loops.

Keywords

Chromosome 15 deletion; dermatoglyphics; Prader-Willi syndrome

The syndrome consisting of hypotonia, hypogonadism, obesity, short stature and mental retardation was first described by Prader, Labhart and Willi (Prader et al. 1956). Ledbetter et al. (1980) first reported an interstitial deletion of the proximal long arm of chromosome 15 (bands q11 to q13) in the Prader-Willi syndrome (PWS) and more recent studies indicate that approximately 50% of such patients have the chromosomal abnormality (Ledbetter et al. 1982, Butler et al. 1982a, Butler & Palmer 1983).

Previous studies from this laboratory (Butler et al. 1982a) revealed that PWS patients with the chromosome 15 deletion had more homogeneity in anthropometric measurements, including radiographic measurements of the bones of the hand as reflected in the metacarpophalangeal pattern profile (MCPP) (Butler et al. 1982b), than those PWS cases with apparently normal chromosomes. Dermatoglyphic studies in PWS patients have not been strikingly abnormal, but there has been enough similarity in dermatoglyphics of

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reported cases to suggest that comparison of PWS patients with respect to the chromosome 15 deletion may be more fruitful.

Material and Methods

Dermatoglyphics were collected from thirty-eight PWS cases (17 females and 21 males ranging in age from 2 weeks to 38 years). All patients had clinical features [neonatal hypotonia, neonatal feeding problems, delayed developmental milestones, mental impairment, obesity (in 83% of our cases), small hands and feet, hypogonadism (males), and chromosome 15 deletions (in 55% of our cases)] consistent with the diagnosis of PWS. The diagnosis was made by at least two physicians on more than one occasion.

High resolution chromosome studies revealed that 21 PWS patients had an interstitial deletion of chromosome 15 (10 ♀, 11 ♂) and the remaining 17 cases (7 ♀, 10 ♂) had normal chromosomes. Detailed chromosome analysis will be published elsewhere. The patients dermatoglyphics were scored prior to the knowledge of the chromosome results. The control group consisted of 270 normal individuals (123 ♀, 147 ♂) collected over the past ten years by students and staff of the Department of Medical Genetics.

Statistical comparison between the different PWS groups and controls was via the standard χ^2 with continuity correction or t-test for qualitative (pattern type) and quantitative variables, respectively. On visual inspection there were no obvious differences between male and female PWS cases and in all analyses the sexes were combined. This approach was employed for two reasons; to avoid further reduction in sample size, and more importantly if a feature is of any clinical significance it should be useful irrespective of the sex of the patient. For dermatoglyphic nomenclature the reader is referred to Schaumann & Alter (1976) or Reed (1981). We have prepared in tabular form, the complete dermatoglyphic findings of all 38 PWS patients. These are available upon request. We will only discuss those features significantly different ($P < 0.05$) in PWS versus controls or in comparisons between the two PWS groups.

Results

PWS Versus Controls

In our PWS patients (Table 1), there was a deficit of proximal (t) positioned axial triradii. This is reflected by both an increase in intermediate (t′) and distal (t″) axial triradii. As a result of this shifting, the mean percent displacements, atd angles, and to some degree the presence of large hypothenar patterns are increased in PWS patients. Although reaching significance in the deletion PWS cases, the latter trends were similar in those cases without the chromosome 15 deletion. Combined 28 of 38 PWS patients (73.7%) lacked a proximal triradius compared with 108/264 (40.9%) of normal controls.

A non-significant excess of fingertip whorls was noted (3.4 per individual in PWS versus 3.0 in controls). Table 2 indicates that most of this excess was found on the thumbs in PWS. Although both deletion and non-deletion PWS showed the excess of whorls on the thumbs, it is interesting to note that the only arch pattern was in a non-deletion case. Paralleling the

increase in whorls on the thumb was an increase in ridge count which attained significance on the left thumb in the deletion PWS (18.25 versus 15.17; $P < 0.05$).

In our PWS there was no arch on any great toe and 8.8% (44/498) of the big toes had arch pattern in controls ($P < 0.025$). In part reflecting the lack of arches, the big toe ridge counts were significantly higher in PWS patients (left, 14.36 versus 11.04, $P < 0.01$; right, 15.06 versus 11.50, $P < 0.01$). The deletion PWS group also had significantly higher big toe ridge counts (15.37 right and 14.53 left), and the non-deletion PWS displayed a very similar non-significant trend for higher big toe ridge counts.

At least one mainline A terminating in position 1 at the base of the thumb (radially) was found in 12 of 38 (31.6%) of PWS patients. This is at least six times more frequent than in normal controls (Schaumann & Alter 1976). As in normal controls radial A mainlines were more common on the left than the right palms (in our series 28.6% of deletion PWS and 23.5% of non-deletion PWS on the left palm and less than 10% in both groups on the right). It is our impression that when the A mainline ends radially in position 1 it is not uncommon to have a distal loop in the palmar interdigital IV area originating from the d triradius and the C mainline terminates ulnarly in the 5″ or 5′ position. Indeed in the four cases illustrated in Holt's review (1975), two of four of the left palms had the combination of termination of A in position 1 and the termination of C in 5.

There was a significant excess of transitional simian creases on the left palm (23.7% versus 3.7%). Only two cases had unilateral true simian creases (one deletion and one non-deletion) and only one non-deletion case had an unequivocal unilateral Sydney line. Due to the very non-specific nature of palmar crease variants, their relative infrequent occurrence in PWS, and the subjective difficulties in classification of transitional forms, palmar creases are unlikely to provide any useful clinical information.

There were a few other statistically significant differences in the PWS groups versus normal. We attribute these to be most likely due to chance. There were no thenar patterns on the left palm in any of the 21 deletion PWS cases. In contrast, thenar patterns or vestiges were increased in frequency on the right palms in non-deletion PWS (17.7% versus 3.7%) and a similar non-significant trend was indicated on the left palm (23.3% versus 13.3%). There was an increase in absent c triradii on the right palm in the non-deletion PWS (23.5% versus 6.8%). There was a non-significant similar trend on the left palm (17.6% versus 5.3%).

Deletion PWS Versus Non-Deletion PWS

All of the preceding findings were not significantly different when compared between the deletion and non-deletion PWS cases. In the plantar pattern areas there was a striking difference between deletion and non-deletion PWS. A decrease in plantar pattern intensity was exclusive to the deletion PWS patients and reflected a decline in pattern frequency in interdigital II, III and IV with an excess of hallucal distal loops. The data are summarized in Table 3.

Despite the fact that hallucal distal loops are the most common pattern in controls, there is a striking increase in deletion PWS patients with a frequency of over 80%. Even though the

frequency in non-deletion PWS was normal, the combined deletion and non-deletion PWS patients had a significant increase in hallucal distal loops (68.4% left, 70.3% right).

More than half of the soles in the deletion PWS cases had an absence of patterns in plantar interdigital areas II–IV and only 4 of 21 deletion PWS had more than one pattern in any of these three interdigital areas. In some instances individual interdigital pattern areas also displayed significantly lower frequency of patterns (Table 3). In contrast, non-deletion PWS in most cases had at least one interdigital pattern, particularly on the left foot. Combining the hallucal distal loop and absent interdigital II– IV patterns resulted in over a three fold excess of such a combination in PWS deletion cases and significant increase even in the combined PWS groups versus normal controls.

Discussion

Both Holt (1975) and Smith & Simpson (1982) who studied the dermatoglyphics in 14 and 24 PWS cases, respectively, concluded that there is no distinct pattern or outstanding dermatoglyphic feature in PWS. Our results tend to substantiate this conclusion. The dermatoglyphics in PWS are not diagnostic. However, there appear to be some consistent findings. These include a tendency for distal displacement of the axial triradius (Laurance 1967, Holt 1975); a tendency toward increased fingertip whorls (Cohen & Gorlin 1969, Holt 1975, Smith & Simpson 1982); radial termination of the palmar A mainline (Smith & Simpson 1982); increased frequency of transitional simian creases and variants (Dunn et al. 1981, Smith & Simpson 1982); and a decrease in plantar pattern intensity (Penrose, in Jancar 1971, Holt 1975). Except for the latter finding, the differences for both the deletion and non-deletion PWS were similar versus normal controls.

The decline in plantar pattern intensity was exclusive to the deletion PWS patients. The plantar combination of hallucal distal loop and open fields in interdigital areas II, III and IV was found on at least one foot in over 60% of deletion PWS compared to only a single non-deletion patient. It is of interest that in the 25 soles enumerated in the topological formulation used by Holt (1975), twelve correspond to hallucal distal loops with no interdigital II, III or IV patterns (If4, Ifz3, and IV̂fh4). Of these twelve they occurred in 7 of the 13 patients presented individually which is very close to the frequency of chromosome 15 deletions found in PWS.

Our plantar results re-inforce the impression that PWS patients with a deletion of chromosome 15 are a more homogeneous group than their non-deletion counterparts. Similar conclusions have been reached with anthropometric measurements and hand X-ray profiles (Butler et al. 1982a, 1982b). In addition, deletion PWS have lighter hair color (blond to light brown), fairer complexion and are more sun sensitive than the PWS individuals with normal chromosomes (Butler 1984). High resolution chromosome analysis should be done on all patients with features suggestive of PWS. The plantar dermatoglyphic patterns along with the above features may be used to support the cytogenetic diagnosis, particularly in those few instances where the cytogeneticist may be uncertain whether or not a subtle deletion of chromosome 15 is present.

The dermatoglyphic features in PWS may be used as another aid in the evaluation of suspected PWS cases. Individuals with proximally placed axial triradii, an arch or small loop on the big toe, and several arches on the fingers are less likely to have PWS. In contrast, cases with distal axial triradii, radial termination of the palmar A mainline, and large whòrls on the thumbs are compatible with the diagnosis of PWS. If the latter individual also lacked plantar interdigital patterns one would expect the chances for a chromosomal deletion to be increased. Anthropometric data and dermatoglyphics may be useful in differentiating PWS from normal in some questionable PWS patients with normal chromosomes, but additional quantitative markers are needed.

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Table 1

Axial triradius and hypothenar patterns

	Control	Total PWS	Non-deletion PWS	Deletion PWS
Left				
%t	45.9	21.1*	17.6*	23.8
%t′	47.7	65.8	70.6	61.9
%t″	6.4	13.2	11.8	14.3
mean % displacement	19.7	24.6*	23.3	25.6*
mean atd angle	44.2	48.2*	47.0	49.2*
% large hypothenar patterns!	10.4	21.1	23.5	19.0
Right				
%t	49.6	15.8**	17.6*	14.3**
%t′	41.0	57.9	70.6*	47.6
%t″	7.9	23.7**	11.8	33.7**
mean % displacement	20.0	28.1**	25.1	30.6**
mean atd angle	44.0	49.6**	47.6	51.4**
% large hypothenar patterns†	14.1	28.9*	23.5	33.3*

† (ulnar loop, whorl, carpal loop, and complexes)

* P<0.05 versus control.

** P<0.01 j

Table 2

Thumb whorl patterns (%)

	Control	Total PWS	Non-deletion PWS	Deletion PWS
Left	33.2	60.6**	59.2*	61.9*
Right	41.9	73.7**	58.8	85.7**

* $P < 0.05$ versus control.

** $P < 0.01$ versus control.

Table 3

Plantar patterns

	Deletion PWS	Non-deletion PWS	Control
I. Hallucal			
Left distal loop (L^d)	85.7 *††	47.1	42.8
whorl	14.3	29.4	39.6
other	–	23.5	17.6
Right distal loop	85.7 *†	50.1	50.2
whorl	14.3	37.5	33.5
other	–	12.4	16.4
II. Interdigital II–IV			
L II Open	81.0	82.4	69.8
L III Open	57.1 **	5.9 †	37.1
L IV Open	95.2	76.5	87.0
R II Open	95.2 *†	62.5	71.3
R III Open	52.4	25.0	27.8
R IV Open	90.5	75.0	79.9
L Open II–IV	52.4 **†	0.0 †	28.3
R Open II–IV	47.6 ††	12.5	19.2
Hallucal L^d + Open ll–IV (at least one)	61.9 **††	6.3	17.8

* P<0.05 versus non-deletion.

** P<0.01 versus non-deletion.

† P<0.05 versus control.

†† P<0.01 versus control.