Dermatoglyphs in congenital heart disease

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SUMMARY The palmar dermatoglyphs of 800 patients with anatomically proven congenital heart disease were compared with prints from 1000 controls. A review of the previous studies revealed major technical deficiencies, and the present study failed to confirm most of the previously reported positive findings.

An overall increase in the incidence of hypothenar patterns was found, probably explaining the previous suggestion of increased atd angle in congenital heart disease. A large number of statistical comparisons inevitably produced a few ‘significant’ results, most of which were inconsistent in various ways.

Two percent of cases were found to have a rare epidermal ridge malformation, ridge dissociation. The nature of the relationship between this and congenital heart disease is obscure.

Claims that there are diagnostically useful dermatoglyphic changes in congenital heart disease can be disregarded.

Although dermatoglyphs have attracted much interest in recent years, the quality of much of the work is abysmal. Publications on dermatoglyphs in congenital heart disease are no exception, and all previous studies1–34 suffer from at least one of the major deficiencies listed below. These faults are to be found in many other dermatoglyphic studies.

1. An accurate cardiac diagnosis was often not made, and cases were often not subdivided into different anatomical groups.1, 5, 16 There are important embryological and genetic differences between different cardiac lesions and it is pointless to lump them all together.

2. Even when a group of apparently similar cases are studied, there will be considerable aetiological heterogeneity. For example, a group of persistent ductus arteriosus (PDA) may include a few cases resulting from rubella, which itself may effect the dermatoglyphs.35

3. Very small numbers were often used.5, 14, 15 With the enormous natural dermatoglyphic variation occurring in normal people, small numbers lead to serious sampling errors.

4. Inadequate or inappropriate statistical tests were often used, for example when examining the TRC.35

5. Some studies only examined fingerprints,2, 7, 14 and others only palm prints.1, 4, 5 Two studies examined only the right palm.8, 9

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Dermatoglyphs differ from other patterns. It was later suggested that dermatoglyphs, by detecting this genetic fraction of cases, might be useful for genetic counselling. Another study suggested an inverse relationship between the TRC and the systolic pulmonary artery pressure. Most studies, however, merely claimed an increased number of certain dermatoglyphic traits in patients with congenital heart disease, sometimes suggesting that this might be diagnostically useful. Despite these claims of diagnostic use, no single study has ever pursued the subject to its logical conclusion by seeing whether possession of the dermatoglyphic characters that are apparently distinctive of a particular cardiac lesion in fact allows diagnosis to be made. In fact this approach has only ever been taken once, and for the other diseases studied, diabetes, schizophrenia, duodenal ulcer, asthma, and various cancers, it was a failure.

One report even claimed that by analysing a baby’s palm prints it is possible “to detect serious heart defects in time for life-saving surgery”. This, and other less dramatic claims, will be examined in the present study, which will also attempt to overcome the problems and deficiencies of previous reports.

Patients and methods

Everyone studied lived in the south and west of England. The controls, 500 males and 500 females, consisted of healthy unrelated subjects representing as far as possible a cross section of social classes.

A total of 800 patients with cardiovascular malformations was studied. Only those who had undergone cardiac catheterisation or cardiac surgery were studied, thus excluding patients with undiagnosed heart lesions. The aim was to study as many accurately diagnosed patients as possible, and no attempt was made to select patients with a particular defect. Patients with chromosome abnormalities or inherited or non-inherited recognisable syndromes, or those in whom there was a known cause for the defect (for example, rubella), were not studied. The mere presence of a non-cardiac malformation did not exclude a case from the study.

Only patients over 6 months of age were studied because of the difficulties of obtaining good prints in babies. The prints were recorded on specially prepared fingerprint forms using black fingerprint ink. Rolled and plain impressions were obtained from all fingers, and two sets of palm prints were recorded, one of which was taken with the fingers fully adducted (for atd angle measurement).

The dermatoglyphic features studied were the same as in a previous study of tuberous sclerosis, which used the same controls.

The 425 female and 375 male patients were divided into separate diagnostic groups. Groups of very small numbers of cases were excluded. Where more than one cardiovascular lesion was present (for example, aortic stenosis and atrial septal defect) the case was included in both diagnostic groups.

In each diagnostic group the following were compared between patients and controls.

1. Fingerprint patterns on individual digits: sexes separate.
2. Total ridge count: sexes separate.
3. Thenar patterns: sexes separate.
5. Interdigital patterns: sexes separate.
7. Mean ridge breadth (MRB): matched age groups, sexes separate.
8. Summed (right and left) corrected atd angle: matched age groups, sexes combined.

The a-b ridge count was used to calculate the mean ridge breadth but was not used for comparisons between patients and controls.

For analysis and comparison of fingerprint patterns, no group of fewer than ten subjects was examined. For analysis of palm patterns and for quantitative analyses, no group of fewer than 20 subjects was examined.

Results

The results for the 1000 healthy controls have been previously published. The positive results for patients are summarised as follows.

GROUP A. ENDOCARDIAL CUSHION DEFECTS (12 MALES, 12 FEMALES)
1. Significant (by Fisher’s test, p = 0.02) deficit of ulnar loops on right index finger of females. Same trend on females’ left index finger, but not in males.
2. Significant (by Fisher’s test, p = 0.03) excess of whorls on right middle finger of females. Reverse trend on left middle finger of females and right middle finger of males.

GROUP B. SECUNDUM ATRIAL SEPTAL DEFECT (37 MALES, 64 FEMALES)
1. Significant ($\chi^2 = 5.9$, p <0.05) excess of arches on right index finger of males. Same trend on left index finger of males and right index finger of females.
2. Significant ($\chi^2 = 4.9$, p <0.05) excess of left hypothenar patterns in males, mainly distal and
outer loops. Similar trend in right palm of males, but not females.

(3) Significant (by Fisher's test, p = 0.025) excess of patients with distally displaced triradii (R plus L corrected atd angle over 110 degrees) in the age group 8 years and over. Same trend in the age group 4 to 7 years.

GROUP C. VENTRICULAR SEPTAL DEFECT (83 MALES, 82 FEMALES)
This, the largest group of patients, showed no significant differences from the controls.

GROUP D. PERSISTENT DUCTUS ARTERIOSUS (20 MALES, 64 FEMALES)
(1) Significant ($\chi^2 = 6.9$, p<0.01) excess of whorls on left index finger of females. Same trend present on right index finger of females. Males showed the reverse trend.
(2) Significant ($\chi^2 = 4.0$, p<0.05) deficit of whorls on left ring finger of males. No similar trend on right ring finger of males or females.
(3) Significantly (Wilcoxon-White test quotient 2.20, p<0.05) higher TRC in females, males showing the reverse trend.

GROUP E. FALLOT'S TETRALOGY (60 MALES, 64 FEMALES)
(1) Significant ($\chi^2 = 5.6$, p<0.05) deficit of thenar patterns in males. Similar trends in females.
(2) Significantly (Wilcoxon-White test quotient 3.67, p<0.001) lower TRC in males. Females showed an almost significant (test quotient 1.89, p = 0.059) reverse trend (that is, higher TRC).
(3) Significant ($\chi^2 = 4.5$, p<0.05) excess of right hypothenar outer loops in males. Similar trend in females, but not on the left either sex.
(4) Significant deficit of right ($\chi^2 = 14.4$, p<0.001) and left ($\chi^2 = 4.7$, p<0.05) hypothenar patterns in females.

GROUP F. PULMONARY STENOSIS (51 MALES, 49 FEMALES)
There were no significant differences between patients and controls.

GROUP G. AORTIC VALVE STENOSIS (40 MALES, 13 FEMALES)
(1) Significant ($\chi^2 = 7.8$, p<0.01) increase in right hypothenar patterns in males, the same trend being present on the left. Females did not show this trend.

GROUP H. TRANPOSITION OF GREAT VEESELS (29 MALES, 14 FEMALES)
(1) Significant ($\chi^2 = 4.0$, p<0.05) deficit of patterns on the right third interdigital area in females, the same trend being present on the left. This trend was not shown in males.
(2) Significant (Fisher's test, p = 0.001) excess of left hypothenar outer loops in females.
(3) Significant excess of unusual hypothenar patterns (arch, composite, double proximal axial triradius) on (a) left palm of females (Fisher's test, p = 0.023), (b) right palm of females (Fisher's test, p = 0.027), and (c) right palm of males (Fisher's test, p = 0.004).

GROUP I. COARCTATION OF AORTA (36 MALES, 21 FEMALES)
(1) Significant (Fisher's test, p = 0.008) excess of radial loops on the right index finger of female patients, the same trend being present on the left index finger of females but not in males on either index finger.
(2) Significant excess of hypothenar twinned loops on both palms of males (Fisher's test; right p = 0.03, left p = 0.003).
(3) Significant excess of hypothenar outer loops on both palms of females (Fisher's test; right p = 0.005, left p = 0.002).

PALMAR CREASES
The results for all 800 patients not subdivided up are shown in table 1. As expected, a greater proportion of patients had a single transverse crease than the controls. An unexpected sex difference was found, the proportion of male patients having a single transverse crease being significantly higher ($\chi^2 = 4.0$, p<0.05).

HYPOTHENAR PATTERNS
The results for individual diagnostic groups are given

<p>| TABLE 1 Palmar creases in 800 subjects with congenital heart disease and 1000 controls |
|----------------------------------------|-----------------|-----------------|-----------------|---------------|</p>
<table>
<thead>
<tr>
<th>Normal creases</th>
<th>Left single crease only</th>
<th>Right single crease only</th>
<th>Bilateral single creases</th>
<th>Total with single crease</th>
</tr>
</thead>
<tbody>
<tr>
<td>Males</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Controls</td>
<td>497 (99.4%)</td>
<td>3 (0.6%)</td>
<td>—</td>
<td>3 (0.6%)</td>
</tr>
<tr>
<td>Patients</td>
<td>359 (95.7%)</td>
<td>9 (2.4%)</td>
<td>2 (0.5%)</td>
<td>16 (4.3%)</td>
</tr>
<tr>
<td>Females</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Controls</td>
<td>498 (99.6%)</td>
<td>2 (0.4%)</td>
<td>—</td>
<td>2 (0.4%)</td>
</tr>
<tr>
<td>Patients</td>
<td>418 (98.4%)</td>
<td>3 (0.7%)</td>
<td>1 (0.2%)</td>
<td>7 (1.6%)</td>
</tr>
</tbody>
</table>
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TABLE 2  Hypothenar patterns in 800 subjects with congenital heart disease and 1000 controls

<table>
<thead>
<tr>
<th>Pattern</th>
<th>No pattern</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Males</strong></td>
<td></td>
</tr>
<tr>
<td>Controls</td>
<td>320 (32.0%)</td>
</tr>
<tr>
<td>Patients</td>
<td>297 (39.6%)</td>
</tr>
<tr>
<td>( \chi^2 = 10.5, p &lt; 0.01 )</td>
<td></td>
</tr>
<tr>
<td><strong>Females</strong></td>
<td></td>
</tr>
<tr>
<td>Controls</td>
<td>368 (36.8%)</td>
</tr>
<tr>
<td>Patients</td>
<td>375 (44.1%)</td>
</tr>
<tr>
<td>( \chi^2 = 9.9, p &lt; 0.01 )</td>
<td></td>
</tr>
</tbody>
</table>

above. However, when all 800 patients were combined, they were found to have a significant excess of hypothenar patterns (table 2). This excess was spread fairly evenly among all hypothenar patterns, with the largest excess being of outer loops, proximal loops, and double proximal axial triradii.

RIDGE DISSOCIATION
None of the 1000 controls showed ridge dissociation, but this was present in 19 (2.4%) of the 800 patients. Eight of these had Fallot's tetralogy, but otherwise no particular cardiac lesion appeared in this group. Where ridge dissociation was present on the palm this was usually in the region of the t triradius, but also sometimes in the hypothenar and interdigital areas.

Discussion

CONTROLS
It is difficult to know what would comprise a perfect control group. Ideally such subjects would differ from patients only by the absence of cardiac malformation. To obtain subjects with the same genotype and with the same experience of intrauterine events as the patients is clearly impossible. Like-sexed unaffected sibs might be useful in some situations, but are still far from ideal.

Both patients and controls were living in the south and west of England, a large area. This may seem satisfactory, but findings such as those of Roberts and Coope in the South Midlands suggest quite important local geographical dermatoglyphic variation. Clearly a study such as the present one could not be done in a small village, and one is left with the choice of a regional or national study, the aim being to cancel out the effect of local variation by drawing from a large area.

PATIENT SELECTION
Although previous investigators have not commented on this, it is important to recognise that patients in a series such as this are a selected group, and this is essential if one is to extrapolate the findings to the general population.

By not studying cases under 6 months, certain severe disorders such as the hypoplastic left heart syndrome will escape study. Severe fatal cases of other lesions will also be missed. However, by insisting on only cases where an accurate diagnosis had been made, many mild cases not warranting cardiac catheterisation will also be missed.

In short, it is thought that the groups of secundum ASD, VSD, pulmonary stenosis, and aortic stenosis tend to be the more severe cases. The group of endocardial cushion defects tends to contain the milder cases, and, in terms of severity of the lesion, the groups of PDA, Fallot's tetralogy, coarctation, and transposition are thought to be fairly representative.

FINGERPRINT PATTERNS
Of a very large number of statistical comparisons, there were only six significant results. In several the trends were not consistent in both hands or both sexes, and it is unlikely that these findings would occur in another series. It appears that fingerprint patterns are not affected in any of the major groups of cardiac defects studied here.

TOTAL FINGER RIDGE COUNT
The only two significant results were accompanied by opposite trends in the other sex studied, and again it rather appears that the TRC is not altered in congenital heart disease.

MEAN RIDGE BREADTH
Comparisons were only possible in three groups because of the need to subdivide by age and sex. Females over the age of 20 with ASD, males aged 4 to 7 years with VSD, and males aged 4 to 7 with Fallot's tetralogy constituted the three groups. In none was there any significant departure from the figures for controls.

SUMMED CORRECTED atd ANGLE
Subjects with congenital heart disease were notable for having a normally placed t triradius. The only possible departure from this was the group of 78 subjects over the age of 8 years with ASD: eight of these (10%) had a summed corrected atd angle over 110°. The same trend, not reaching significance, was found in the 17 subjects aged 4 to 7 years: two of them (12%) had a summed corrected atd angle over 120° (the arbitrary cut-off point for this age group). The control figures are 4.2% (those over 8 years of age) and 3% (those aged 4 to 7 years). Similar
trends, though not statistically significant, have been noted previously, though the methodology was unsatisfactory. In general, it appears that significant changes in the adt angle are not found in congenital heart disease. Previous reports suggesting the reverse are probably partly attributable to the use of the maximal adt angle and the failure to recognise the presence of hypothenar patterns.

HYPOTHENAR PATTERNS
It is here that the results are most interesting, for there does appear to be a significant increase in the proportion of subjects with hypothenar patterns of all sorts. Some trends are so strong that significant results are found even when single patterns are studied within individual groups of subjects.

A previous report suggested a specific association between a double proximal axial triradius (DPAT) and septal defects. The present study has shown a significant overall increase in DPAT, significant in both sexes, but not confined to septal defects. However, another study has failed to confirm this, finding that if anything a DPAT is associated with pulmonary stenosis.

PALMAR CREASES
Several authors report no significant increase in the frequency of a single transverse palmar crease in congenital heart disease. Nevertheless it is clinical experience (and the finding of others) that a single crease is a non-specific sign that is sometimes found in patients with malformations, and one might therefore have expected to find a small increase in the incidence of a single crease in this study.

As the incidence of a single crease was unusually low in the controls it is difficult to come to any firm conclusion in this study. If there is a true increased incidence of single crease in congenital heart disease then it is small and of no clinical significance.

RIDGE DISSOCIATION
Ridge dissociation is an extremely rare congenital malformation of epidermal ridges, sometimes inherited as an autosomal dominant trait. A previous report found only two cases among 430 patients with congenital heart disease, but this was an underestimate and failed to detect some examples of ridge dissociation. It is not clear at present why a malformation of epidermal ridges should be associated with cardiovascular malformations.

Conclusions
Claims that congenital heart disease is associated with characteristic dermatoglyphic patterns are probably false. The few associations that reached statistical significance in this large study were generally internally inconsistent and must be viewed with caution in view of the very large number of statistical comparisons that were made. An overall increase in hypothenar patterns is probably genuine, and may explain previous reports of supposedly displaced t triradii in congenital heart disease.

It appears that a rare congenital malformation of epidermal ridges, dissociation, is found in about 2% of cases of congenital heart disease. The specific association with Fallot's tetralogy may be a little stronger than this.

Suggestions that dermatoglyphs are diagnostically useful in congenital heart disease can be completely dismissed.

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References
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