THE INHERITANCE OF TARSAL COALITION AND ITS RELATIONSHIP TO SPASTIC FLAT FOOT

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Tarsal coalition is not a modern phenomenon. Archaeological findings have demonstrated this anomaly in a Mayan civilisation in Guatemala (Harris 1965) and in a pre-Columbian Indian civilisation in Ohio (Heiple and Lovejoy 1969), both dated around 900-950 A.D. Buffon (1769) was probably the first to recognise the occurrence of tarsal coalition, although the undated specimen in the Museum of the Royal College of Surgeons of England described by John Hunter probably dates from 1760 to 1770 (Allen 1973). Calcaneo-navigular coalition was first described anatomically by Cruveilhier (1829) and talo-calcanal coalition by Zuckerkandl (1877). Holl (1880) made a tentative suggestion relating peroneal spastic flat foot and intertarsal bars.

Pfistner (1896) proposed that tarsal coalitions were caused by the incorporation of accessory ossicles into major adjacent tarsal bones. This idea has received support from the work of Slomann (1921), Cave (1926), Badgeley (1927), Harris and Beath (1948), Chambers (1950), Outland and Murphy (1953) and Hark (1960).

Leboucq (1890), Solger (1890), Dwight (1907), Trolle (1948) and Jack (1954) proposed that tarsal coalitions were the result of a failure of differentiation and segmentation of primitive mesenchyme. Harris (1955) demonstrated that such anomalies can occur in the foetus and her results confirm the proposal that a mesenchymal defect is the cause. Pfistner’s hypothesis of incorporation of accessory ossicles is not acceptable because it fails to explain the disorder in the foetus.

In spite of Sir Robert Jones’s clinical description of peroneal spastic flat foot in 1897, it was not until the work of Slomann (1921), Badgeley (1927) and Harris and Beath (1948) that tarsal coalitions were conclusively linked with peroneal spastic flat foot. It is now appreciated that with early recognition the treatment of peroneal spastic flat foot due to tarsal coalition may be successful (Jack 1954, Blockey 1955, Harris 1965, Mitchell and Gibson 1967).

Occasional reports have appeared in the literature suggesting that tarsal coalitions are inherited. Rothberg, Feldman and Shuster (1935), Boyd (1944), Webster and Roberts (1951) and Bersani and Samilson (1957) reported the anomaly in members of single families, but although they drew attention to the occurrence of painful feet, they did not comment on the presence of peroneal spastic flat foot.

Wray and Herndon (1963) reported three generations of a family in which calcaneo-navicular coalitions were found in each generation. They concluded that some and perhaps all cases of calcaneo-navicular bar were caused by a specific gene mutation which possibly behaved as an autosomal dominant with reduced penetrance. They suggested that this gene was distinct from each of several genes responsible for other fusion defects in the bones of the hand and foot, including multiple fusions, gross anomalies and varieties of symphalangism. Glessner and Davis (1966) reported monozygotic twins with peroneal spastic flat foot and tarsal coalition and suggested that the condition was of genetic etiology.

Tarsal and carpal coalitions are known to be present in phocomelia, hemimelia and other gross limb anomalies (O’Rahilly 1953). They are also found in other congenital disorders (Nievergelt 1944), and the occurrence of symphalangism with tarsal and carpal coalitions has been documented by Drinkwater (1917) and Austin (1951).
This paper concerns the relationship between tarsal coalitions and peroneal spastic flat foot; cases with associated anomalies have been excluded. However, because a relationship exists between tarsal and carpal development, it was felt that this study should include the carpus.

MATERIAL AND METHOD

An attempt was made to trace all those patients still living in Edinburgh who had been treated over the period 1940-70 for peroneal spastic flat foot in association with tarsal coalition. They and their first degree relatives (parents, sibs and children) were interviewed and examined clinically and radiographically, with emphasis on the tarsus and carpus. Family pedigrees were obtained and attention was paid to any symptoms referable to the hands or feet of the relatives. Thirty-one index patients and ninety-eight of their first degree relatives were traced.

<table>
<thead>
<tr>
<th>Authors</th>
<th>Material</th>
<th>Incidence</th>
</tr>
</thead>
<tbody>
<tr>
<td>Pfitzner (1896)</td>
<td>Post-mortem</td>
<td>2 of 524 (0.38 per cent)</td>
</tr>
<tr>
<td>Harris and Beath (1948)</td>
<td>Potential army recruits</td>
<td>1 of 3,600 (0.03 per cent)</td>
</tr>
<tr>
<td>Vaughan and Segal (1953)</td>
<td>Army personnel</td>
<td>21 of 2,000 (1 per cent)</td>
</tr>
<tr>
<td>Shands and Wentz (1953)</td>
<td>Children’s clinic</td>
<td>11 of 1,232 (0.9 per cent)</td>
</tr>
</tbody>
</table>

The incidence of tarsal coalition—This is not known for certain. Various authors have attempted to show the frequency (Table I), but these studies have been based on selected groups and do not give a true over-all population incidence. However, the figures are of some assistance in that not one of these groups gives an incidence above 1 per cent.
Radiography—Because the survey was based on radiography, rigid criteria were used in the interpretation of possible anomalies. All index patients and relatives had antero-posterior and standing lateral views of the feet, together with oblique hindfoot views (Fig. 1) and axial views of the subtalar joint (Harris and Beath 1948) (Fig. 6). It was thought impracticable in a large family survey to resort to lateral tomography of the subtalar region as suggested by Cowell (1972). Antero-posterior, lateral and oblique views of the carpus were also taken in both index patients and relatives. Because Lamb (1958) reported the occurrence of "ball and socket" ankle joints in some cases of tarsal coalition, radiographs of the ankle joint were also taken.

Seddon (1932) considered that a prolongation of the anterior process of the calcaneus was pathological and related to calcaneo-navicular coalition (Fig. 2). In this series such cases
were counted as negative because it was felt that they do not represent true coalition. Pfitzner (1896) considered that an "os calcaneus secundarius" was related to calcaneo-navicular coalition (Fig. 3). Because a true coalition certainly does not exist here, all such cases were counted as negative.

Figure 4 shows a possibly developing bar in a girl aged eight with a family history of tarsal coalition, but this and similar radiographic appearances in this age group were not regarded as definite coalitions. Figures 5 and 7 show examples of undoubted calcaneo-navicular and talo-calcaneal fusion, which are of course regarded as positive findings.

Figure 6—An axial radiograph of the subtalar region of a normal subject showing both components of the subtalar joint. Figure 7—An axial radiograph of the subtalar region showing typical talo-calcaneal coalition, with some evidence of a rudimentary subtalar joint.

RESULTS

Table II shows the proportion of the first degree relatives with tarsal coalition: 33 per cent of parents and 46 per cent of sibs were affected—that is, 39 per cent of first degree relatives. Of the thirty-one index patients, twenty-seven had calcaneo-navicular coalition and four had talo-calcaneal coalition.

An analysis of the first degree relatives of the twenty-seven index patients with calcaneo-navicular coalition is shown in Table III: 25 per cent also had calcaneo-navicular coalition, but 14 per cent had talo-calcaneal or some other type of tarsal fusion. Of the four index patients with talo-calcaneal coalitions, eleven affected relatives all had similar fusions.

<p>| TABLE II |
| TARSAL COALITION: PROPORTION OF AFFECTED FIRST DEGREE RELATIVES |</p>
<table>
<thead>
<tr>
<th>Index patients</th>
<th>Parents</th>
<th>Siblings</th>
<th>Total</th>
</tr>
</thead>
<tbody>
<tr>
<td>31</td>
<td>18 of 55 (33 per cent)</td>
<td>20 of 43 (46.5 per cent)</td>
<td>38 of 98 (39 per cent)</td>
</tr>
</tbody>
</table>
Table IV shows the age and sex distribution of the first degree relatives in the series. The sex ratio was approximately equal, and there was no significant difference in the ages of the affected parents and sibs with regard to sex.

Table V shows the side of fusion; there was no significant difference between index patients, parents or sibs in regard to whether the fusions were bilateral or unilateral; most (80 per cent or more) were bilateral.

### TABLE III
**Tarsal Coalition: Proportion of Affected First Degree Relatives**

<table>
<thead>
<tr>
<th></th>
<th>Index patients</th>
<th>Parents</th>
<th>Siblings</th>
<th>Total</th>
</tr>
</thead>
<tbody>
<tr>
<td>Calcaneo-navicular only</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>27</td>
<td>23 per cent</td>
<td>27.5 per cent</td>
<td>25 per cent</td>
<td></td>
</tr>
<tr>
<td>Talo-calcaneal and other coalitions</td>
<td>11 per cent</td>
<td>17.5 per cent</td>
<td>14 per cent</td>
<td></td>
</tr>
<tr>
<td>Total</td>
<td>34 per cent</td>
<td>45 per cent</td>
<td>39 per cent</td>
<td></td>
</tr>
</tbody>
</table>

### TABLE IV
**Tarsal Coalition: Age and Sex Ratio of Index and First Degree Relatives**

<table>
<thead>
<tr>
<th></th>
<th>Male</th>
<th>Female</th>
<th>Average age in years at time of survey</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Male</td>
<td>Female</td>
<td></td>
</tr>
<tr>
<td>31 index patients</td>
<td>15</td>
<td>16</td>
<td>20 (20 per cent) 22 (20 per cent)</td>
</tr>
<tr>
<td>18 affected parents</td>
<td>10</td>
<td>8</td>
<td>57 (83 per cent) 55 (83 per cent)</td>
</tr>
<tr>
<td>20 affected siblings</td>
<td>9</td>
<td>11</td>
<td>24.5 (85 per cent) 21 (85 per cent)</td>
</tr>
</tbody>
</table>

### TABLE V
**Tarsal Coalition: Distributions of the Coalitions**

<table>
<thead>
<tr>
<th>Subjects</th>
<th>Unilateral coalition</th>
<th>Bilateral coalition</th>
</tr>
</thead>
<tbody>
<tr>
<td>31 index patients</td>
<td>6 (20 per cent)</td>
<td>25 (80 per cent)</td>
</tr>
<tr>
<td>18 parents</td>
<td>3 (17 per cent)</td>
<td>15 (83 per cent)</td>
</tr>
<tr>
<td>20 siblings</td>
<td>3 (15 per cent)</td>
<td>17 (85 per cent)</td>
</tr>
</tbody>
</table>

Clinical and radiological examination of the carpus in both index patients and first degree relatives failed to show any abnormalities. Radiographs of the ankle were also negative, no case of "ball and socket" joint being found.

Clinical examination of the feet of the first degree relatives was quite remarkable in that those with a tarsal abnormality had no evidence whatever of peroneal spastic flat foot. Indeed, not one of the ninety-eight first degree relatives had ever complained of a painful foot or had received medical treatment for such a condition.
DISCUSSION

The results in this study suggest that tarsal coalitions are inherited, most probably as a unifactorial disorder of autosomal dominant inheritance, very nearly of full penetrance. Furthermore, there does not seem to be any genetic difference in the inheritance of the different coalitions, since 14 per cent of relatives were found to have a different form of tarsal coalition from the index patient (Table III). Because tarsal coalitions in relation to peroneal spastic flat foot appear to be inherited, the concept of foetal maldevelopment as a cause is supported, rather than the sporadic incorporation of accessory ossicles into major tarsal bones.

It seems that the carpus is entirely unaffected in this situation, for sixty-nine patients with tarsal fusion failed to reveal any carpal coalition. It is probable therefore that the tarsal coalitions seen in such conditions as Nievergelt's syndrome or in symphalangism, phocomelia or hemimelia are unrelated to the tarsal coalitions found in peroneal spastic flat foot.

It is difficult to explain the absence of symptoms in all first degree relatives, in spite of tarsal coalition being present in 39 per cent of them. In the whole survey of thirty-one index patients and ninety-eight relatives, only the index patients (thirty-one of 129 or 24 per cent) had symptoms. Jack (1954) noted that five of twenty-three patients (22 per cent) studied with tarsal coalitions were free from symptoms. This is in marked contrast to the results obtained in this study, where 76 per cent of the subjects with tarsal coalition were free from symptoms.

From the results of this survey it would appear that tarsal coalition is not a rare phenomenon and that clinical studies based on symptomatic peroneal spastic flat foot under-estimate the true occurrence of the anomaly. The absence of symptoms in certain circumstances may be related to genetic variability of expression and reflected in minor anatomical variations as yet unknown.

SUMMARY

1. A clinical and radiological survey has been made of the families of thirty-one patients with peroneal spastic flat foot and tarsal coalition (twenty-seven calcaneo-navicular, four talo-calcaneal).
2. Thirty-nine per cent of ninety-eight first degree relatives were found to have some type of tarsal coalition.
3. A surprise finding was that not one of the first degree relatives had ever had symptoms referred to the tarsus.
4. No case of "ball and socket" ankle joint or of carpal coalition was found in this series.

I wish to thank Professor J. I. P. James and Miss R. Wynne-Davies for their help in the preparation of this paper. I am grateful to the members of the staff of the Princess Margaret Rose Orthopaedic Hospital for allowing me access to the records of their cases. I am also indebted to the members of the Clinical Genetic staff, and to Miss V. Lindsay and Mrs E. Ward for secretarial assistance.

REFERENCES


