Familial Poland anomaly

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SUMMARY  The Poland anomaly is usually a non-genetic malformation syndrome. This paper reports two second cousins who both had a typical left sided Poland anomaly, and this constitutes the first recorded case of this condition affecting more than one member of a family. Despite this, for the purposes of genetic counselling, the Poland anomaly can be regarded as a sporadic condition with an extremely low recurrence risk.

The Poland anomaly comprises congenital unilateral absence of part of the pectoralis major muscle in combination with a widely varying spectrum of ipsilateral upper limb defects. There are, in addition, patients with absence of the pectoralis major in whom the upper limbs are normal, and much confusion has been caused by the careless labelling of this isolated defect as the Poland anomaly. It is possible that the two disorders are part of a single spectrum, though this has never been investigated.

The Poland anomaly is a limb defect which enjoys the unique position of being exclusively unilateral. The aetiology of the condition is unknown, although evidence has accumulated to suggest that first trimester exposure to ergot alkaloids may account for some cases. McKusick lists the Poland anomaly as a dominantly inherited disorder in his catalogue, though there is not in fact a single report of the Poland anomaly occurring in more than one member of a family. This listing is probably because of the fact that there are some reports of the familial occurrence of some of the features of the Poland anomaly.

Greif, in 1891, described a 50-year-old man (his own father) with a left hemiplegia resulting from a stroke at the age of 46. This man was said to have complete absence of the left pectoralis major and minor muscles. It is not clear whether this was a congenital anomaly, although the defect had been noted before the stroke. The limbs were normal. The man’s son had complete absence of the left pectoralis major and minor, the left breast tissue was hypoplastic, and there was partial absence of the left serratus anterior muscle. The left arm was 1.5 cm shorter than the right and its circumference was also slightly reduced. The hands were normal. Another son (Greif himself) said that his own left pectoralis major was weaker than the right. “Although the difference is obvious, the author still had to carry out his military duties”!

Trosev and colleagues have been widely quoted as reporting familial cases of the Poland anomaly. However, this is untrue. They described a mother and child with autosomal dominant radial sided upper limb defects. They stated that the son also had an absent right pectoralis major, but the published photograph fails to confirm this unusual association. In the same paper, a patient with the Poland anomaly is reported whose father had partial cutaneous syndactyly of the second and third toes on both feet. How either of these families came to be regarded as familial cases of the Poland anomaly is obscure.

Another report describes a 56-year-old man who, after the removal of a right sided pericardial cyst, was found to have absence of the sternocostal head of the right pectoralis major. The right breast was hypoplastic and there was less hair in the right axilla, both features suggesting that this was a true congenital malformation and not an acquired defect. The right arm was hypoplastic and 2 cm shorter than the left arm, and there were flexion contractures at the distal interphalangeal joints in all digits of the right hand except the thumb. The middle phalanges of these digits were shortened, but there was no syndactyly. This man had five children and two of them had pertinent congenital abnormalities. A daughter had hypoplasia of the left breast and nipple with normal pectoral muscles and normal hands. A son had a hypoplastic left hand with brachydactyly but no syndactyly, and his pectoral muscles were normal. The authors suggested that this was simple autosomal dominant inheritance of the Poland
anomaly. However, there must be some reservations about this. In none of the three subjects was there any syndactyly, in two of them the pectoral muscles were intact, and only one member of the family had the Poland anomaly. The abnormalities in these three cases are sufficiently atypical to suggest that this interesting family had a different condition.

In one case of the Poland anomaly, the paternal cousin had an absent pectoralis major with breast hypoplasia and some possible minor hand asymmetry. The rather remote family relationship was explained on the grounds of delayed mutation.

There is a brief report from China of a man with an absent right pectoralis major and breast whose father had “the same defect”. No other relevant details were given. There is also a report of two sisters with absence of the pectoralis major, though the authors pointed out that the sisters did not have limb malformations and therefore did not have the Poland anomaly.

Finally, there are three reports suggesting some connection between pectoralis muscle absence and preaxial upper limb defects. A report on triphalangeal thumbs describes a father and son who both had bilateral absence of the pectoralis major and minor muscles and bilateral triphalangeal thumbs. However the reliability of these observations is questionable. There were no illustrations of the pectoralis muscle absence, and the authors do not appear to have appreciated the significance of this family, for bilateral congenital absence of the pectoralis major has never been described. A more recent report describes an infant with duplication of the distal phalanx of the right thumb and a hypoplastic left hand, with “mild hypoplasia of the pectoral muscle and nipple”. (The precise nature of the pectoralis defect is not clear.) The mother had absent or hypoplastic distal interphalangeal flexion creases on all four fingers on both hands. The relationship between the abnormalities of mother and child appears obscure. Lastly, there is the newly described ‘IVIC’ syndrome, inherited as an autosomal dominant trait, comprising bilateral preaxial upper limb defects varying from slightly hypoplastic thumbs to severely malformed upper limbs, external ophthalmoplegia, hearing impairment, thrombocytopenia, and imperforate anus. This syndrome has been found in one Venezuelan family, and of the 19 cases studied two had defects of the pectoral muscles. Both had severe, bilateral, but asymmetrical upper limb defects. In both cases defects of the pectoral muscles were observed on the more severely affected side. In one case the left “pectoral girdle muscles” were described as “atrophic”, and in the other case “severe hypoplasia of the deltoid and pectoralis major muscles” was noted on the right side. Despite the regrettable vagueness of these descriptions it is clear that these two cases did not have the Poland anomaly, the nature and bilaterality of the limb defects placing this disorder into an altogether different category. Whatever the significance of these three reports, none constitutes the occurrence of the Poland anomaly in more than one member of a family.

This paper describes two typical cases of the Poland anomaly occurring in the same family.

Case reports

Case 1

This was a 7-year-old boy, the second child of a mother aged 27 and a father aged 28 at the time of conception. The mother had a normal 28-day
menstrual cycle and had never taken an oral contraceptive. At the time of the second missed menstrual period (that is, at approximately 4 to 6 weeks after conception) the mother took hormonal pregnancy test tablets for 3 days (Amenorone forte: ethisterone 50 mg; ethinylestradiol 0.05 mg). Iron injections were required for anaemia in the second trimester, and at 4 months’ gestation the mother’s father died suddenly. Normal delivery occurred at term plus 6 days, and the birthweight was 3515 g.

The abnormalities, which comprise a left sided Poland anomaly, included: absent sternocostal head and partial absence of the clavicular head of the left pectoralis major muscle; winging of the left scapula with probable absence of the serratus anterior muscle; hypoplastic left forearm and hand; brachydactyly of all four fingers of the left hand (mainly owing to shortening of the middle phalanx except in the index finger where the middle phalanx was absent), with cutaneous syndactyly of these fingers; and a slight flexion deformity of the left index and middle fingers owing to short flexor tendons. The syndactyly has been surgically released. No other malformations were present. Typical dermatoglyphic changes of the Poland anomaly were present (see table). Investigation of the family revealed that the mother’s father’s brother’s son (case 2) was similarly affected.

case 2
This was a 38-year-old man, the third child of a mother aged 33 and a father aged 37 at the time of conception. Severe nausea and vomiting in the first trimester was treated with an unidentified “white medicine”. At 12 weeks’ gestation the mother had two teeth extracted under general anaesthetic. An antepartum haemorrhage occurred at 30 weeks’ gestation. Normal delivery occurred at term and the birthweight was 3799 g.

The abnormalities, which comprise a left sided Poland anomaly, included: absent sternocostal head of the left pectoralis major muscle; winging of the scapula with probable absence of the serratus anterior muscle; absence of the left latissimus dorsi muscle; hypoplasia of the whole left arm and hand; and brachydactyly of all four fingers on the left hand, with extreme shortening of the index finger which was reduced almost to a stump, with cutaneous syndactyly of all four fingers. No other malformations were present. Typical dermatoglyphic changes of the Poland anomaly were present (see table). Case 2 has two sons who are normal. Investigation of the family has not revealed any other relatives either with the Poland anomaly or with individual features of the Poland anomaly (such as a limb defect or a pectoral defect). There is no consanguinity in the family.

**TABLE Dermatoglyphs of two cases of the Poland anomaly**

<table>
<thead>
<tr>
<th></th>
<th>Case 1</th>
<th></th>
<th>Case 2</th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Right</td>
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<td>Right</td>
<td>Left</td>
</tr>
<tr>
<td><strong>Fingerprint pattern</strong></td>
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<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Thumb</td>
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<td>R (15)</td>
<td>U (18)</td>
<td>R (17)</td>
</tr>
<tr>
<td>Index finger</td>
<td>TL (23)</td>
<td>A (0)</td>
<td>U (13)</td>
<td>A (0)</td>
</tr>
<tr>
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<td>U (6)</td>
<td>R (7)</td>
</tr>
<tr>
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<td>U (12)</td>
<td>A (0)</td>
</tr>
<tr>
<td>Little finger</td>
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<td>W (7)</td>
<td>U (3)</td>
<td>A (0)</td>
</tr>
<tr>
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<tr>
<td>Triradii</td>
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<td>b, t*</td>
<td>a, b, c, d, t</td>
<td>d, t*</td>
</tr>
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<td>?</td>
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<td>None</td>
</tr>
<tr>
<td>Flexion creases</td>
<td>Normal</td>
<td>Single</td>
<td>Normal</td>
<td>Single</td>
</tr>
</tbody>
</table>

*Rest of triradii missing. TL = twinned loop; W = whorl; U = ulnar loop; R = radial loop; A = arch. The numbers in parentheses refer to the ridge count.

**FIG 3 Chest wall of case 2. The abdominal scar is from a laparotomy (perforated duodenal ulcer).**

**FIG 4 Hands of case 2.**
Discussion

This constitutes the first report of the Poland anomaly occurring in more than one member of a family. The relationship of the two cases was fairly remote (second cousins). This would be compatible with an incompletely penetrant autosomal dominant gene or with multifactorial inheritance. It could of course be a chance event, but although we have no reliable figures for the population incidence of the Poland anomaly (estimates vary from 1 in 17 000 to 1 in 100 000) the condition is so rare that this hardly seems likely.

Teratogens have been implicated in some cases of the Poland anomaly, but there is little evidence of such exposure in either of these cases. It is probable that the condition is slightly more common on the right side than the left, and slightly more common in males in this connection it is worth noting that both cases were males with a left sided defect. The type of chest and limb defect was quite typical of the Poland anomaly in both cases. The involvement of other shoulder girdle muscles is a well recognised feature of the Poland anomaly, and in the absence of the serratus anterior was noted by Poland in his original case.

Our own studies of the Poland anomaly are incomplete, but of the first 78 cases of congenital absence of the pectoralis major muscle (46 cases of the Poland anomaly, 32 of isolated pectoralis absence) there are no other subjects with any relative affected with any similar or related malformation. Other large series of cases do not include any familial examples of the condition, and it seems unlikely that such cases have been under-reported or missed.

The true genetic situation depends partly on the relation between isolated pectoralis major absence and the full-blown Poland anomaly. At present it is not known whether these are separate entities or parts of a spectrum. If the former is true, then genetic counselling is simple for there is only one recorded familial case (the present report) and the recurrence risk is extremely low. If there is a spectrum of defects, then the recurrence risk, at least for some features of the spectrum, would be a little higher, but even so most cases of the Poland anomaly would still be sporadic. Gorlin's conclusion, "that the recurrence rate is considerably less than 1%", seems reasonable.

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References


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