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Posterior Shoulder Girdle Abnormalities With Absence of Pectoralis Major Muscle

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We have reviewed 12 consecutively ascertained individuals with unilateral absence of the sterno-costal head of the pectoralis major muscle and have studied associated abnormalities of the posterior shoulder girdle and their frequency and extent as contrasted with that of the symbrachydactly. We found certain previously unreported associations of skeletal and renal anomalies in these individuals. We review various speculations regarding the possible causes of the Poland anomaly.

Key words: Poland anomaly, Sprengel shoulder deformity, brachydactyly, renal anomalies, homovertebral anomalies

INTRODUCTION

Congenital absence of the pectoralis major muscle is recognized as a common musculo-skeletal abnormality [Bing, 1902] and its association with other thoracic [Smith, 1976] limb, and musculo-skeletal [Mace et al, 1972] abnormalities has been well described in the literature. Although associated abnormalities of the posterior chest wall have been observed in the past [Bing, 1902; Beals and Crawford, 1976; Ireland, Takayama, and Flatt, 1976; Frias and Felman, 1974], their extent and frequency has not been defined. The purpose of this communication is to a) outline the extent and frequency of posterior shoulder girdle abnormalities associated with absence of pectoralis major muscle, b) attempt to shed some light on the cause of the Poland "syndrome", and c) describe certain previously unreported skeletal and renal abnormalities that are associated with absence of pectoralis major muscle.

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CLINICAL DATA

Twelve individuals with unilateral absence of the sterno-costal head of the pectoralis major muscle were examined. Whether the ipsilateral pectoralis minor was hypoplastic or absent could not be determined. We found abnormalities in the posterior shoulder girdle in eight individuals. Table I outlines these eight cases and the observed morphological abnormalities.

Posterior Shoulder Girdle Anomalies

Clinically detectable hypoplasia of the scapula was the commonest abnormality observed in the posterior shoulder region (6/12). Winging of the medial border, elevation, and omovertebral anomaly were the other scapular abnormalities encountered in this order of frequency. These abnormalities often led to asymmetry in posterior chest wall and occasionally caused ipsilateral elevation of the shoulder. Hypoplasia varied from 1 to 3 cm on the medial border of the scapula, when compared to its contralateral counterpart. Most of these patients (4/6) lacked the upper portion of the serratus anterior muscle and consequently also had winging of the medial border of the scapula. Two patients had absence of the infraspinatus with resultant flattening of the ipsilateral posterior shoulder. Another had an elevated hypoplastic scapula with omovertebral bone (Sprengel deformity). In one case, scapular hypoplasia was not associated with any gross muscle abnormality. Conversely, in two instances, hypoplasia of the attaching muscles was not associated with scapular abnormality. This suggests that the scapular hypoplasia may occur independently of the abnormalities of muscles attached to it.

Muscle Abnormalities

The commonest muscle abnormality observed in the posterior shoulder girdle was the absence of the upper portion of the serratus anterior (5/12). All of these patients had winging of the scapula with slight elevation that caused significant asymmetry in the back. Hypoplasia of the infraspinatus was noted in two cases and these patients had flattening of the scapular region. The only patient with absent supra spinatus had a noticeable depression in the supraspinatus region and had slight difficulty in abducting the arm at the shoulder joint.

Klippel-Feil Anomaly

One girl had a cervical encephalocele that was repaired in the neonatal period and a Sprengel

deformity with omovertebral bone. She also had a Klippel-Feil anomaly with fusion of the cervical vertebrae and shortness of the neck (Figs. 1 and 2). Although the absence of the pectoralis major muscle with Sprengel anomaly has been observed in the past [Brown and McDowell, 1940l, its association with the Klippel-Feil anomaly, to our knowledge, has not yet been reported.

Renal Abnormalities

Temtamy and McKusick [1969] reported a child with Poland defect and ipsilateral renal hypoplasia. Subsequently, Mace et al [1972] found renal agenesis in one of their seven cases of Poland anomaly. This was also found by Miller and Miller [1975] in a child with absent pectoralis major muscle, without digital abnormality, who developed acute lymphoblastic leukemia. We found renal abnormalities in two of the six specifically investigated cases. Six other patients were either unavailable or refused to provide consent for such investigation. The frequency of the renal anomalies might well have been higher had all our patients undergone intravenous urography or realtime ultrasonography. One patient had absence of the ipsilateral kidney, while the other had duplication of the ipsilateral collecting system. To date, the latter abnormality in association with absence of the pectoralis major muscle or Poland anomaly has not been reported. Both of these patients had abnormalities in the posterior shoulder girdle but did not have any gross digital abnormality. In view of the high frequency of associated renal malformations and the potential seriousness of such anomalies, it is probably prudent to recommend routine renal ultrasonography and/or excretory urography to individuals with absence of the pectoralis major muscle with or without other manifestations of the Poland anomaly.

DISCUSSION

Although a clinical observation of the absence of the pectoralis major muscle was made earlier by others [Ravitch, 1977], Poland [1841] described its absence in a cadaver, which, in addition to the absence of the sterno-costal head of the pectoralis major muscle, had other muscle, nerve, and vessel abnormalities in the anterior chest wall, and ipsilateral symbrachydactyly. Patrick Clarkson [1962] called syndactyly associated with absence of pectoralis major muscle "Poland's syndactyly." Baudinne, Bovy and Wasterlain [1967] labeled this combination of deformities "Poland's syndrome." After observing a consistent pattern, Beals and Crawford [1976] suggested that the midphalangeal hypoplasia is the criterion for "Poland's anomaly."

Occurrence of abnormalities in the posterior shoulder region in association with absent pectoralis major muscle was known to Bing [1902]. He observed scapular hypoplasia in 9 of his 102 cases of absent pectoralis major muscle reported in 1902. Among them, some had a Sprengel deformity as well. Beals and Crawford [1976] reported that the absence of the upper portion of serratus anterior led to varying degrees of hypoplasia, elevation, and winging of the scapula in some of their cases of Poland's syndrome. Similar minor scapular abnormalities in individuals with Poland's anomaly have also been noted by other observers [Ireland, Takayama, and Flatt, 1976; Frias and Felman, 1974].

Abnormalities in the posterior shoulder girdle are common in individuals with congenital absence of pectoralis major muscle. In our experience, they are twice as frequent as the digital abnormalities and seem to occur independently of symbrachydactyly. A higher frequency of these abnormalities observed in our cases, as compared to the previously reported cases, can be explained on the basis that a) most cases with absent pectoralis major muscle reported in the literature are those that are associated with symbrachydactyly (Poland's Syndrome), and only 13.5% of such patients have this combination of anomalies [Ireland, Takayama, and Flatt, 1976], and b) the posterior shoulder girdle in these patients is frequently not examined.

The cause of the absent pectoralis major muscle, and of the Poland's anomaly remains obscure. David observed potentially noxious antenatal factors preceding the birth of the infants with the Poland anomaly [David, 1972]. Several of the mothers in his series had reportedly attempted abortion. This was not corroborated by subsequent investigators and on careful questioning of the mothers we could not confirm the presence of this factor in our study. Goldberg and Mazzei [1977] refuted the theory of nerve root dysgenesis and postulated a concept of local injury occurring in the 6-7 week old embryo, at a time when the immature hand is juxta-positioned next to the differentiating pectoris major muscle mass. Although a consistent pattern of digital abnormalities and the absence of sterno-costal head of the pectoralis major muscle may conceivably arise from such an injury, the frequently associated abnormalities in the posterior shoulder region can not be explained on this basis. The widespread abnormalities in the anterior chest wall, posterior shoulder girdle and the upper limb may be more convincingly attributed to a vascular injury which compromises the circulation in the region, during limb bud formation. In his report of the findings in a cadaver, Poland mentioned that the vessels in the affected region were small. Whether these changes in the vessels were primary or secondary to another lesion which caused the various other abnormalities, is not known.

The multiplicity of sites involved suggests that this anomaly may well be an example of polytopic field defect of the contiguous mesoderm during human embryogenesis, perhaps due to vascular insult, resulting in topically disparate anomalies in the fully developed individual. Though this has been recognized in animal experiments, only few instances have been convincingly demonstrated in man. In view of the involvement of the kidney, on the ipsilateral side, together with shoulder and upper limb abnormalities, the reported anomaly may constitute yet another example of the acrorenal polytopic field defect.

SUMMARY

Poland anomaly may be an example of a polytopic field defect with the manifestations ranging from isolated absence of the sterno-costal head of the pectoralis major muscle to varying degrees of abnormalities of the anterior chest wall and the posterior shoulder girdle. It is occasionally associated with hypoplasia of the hand and brachydactyly. Renal abnormalities are not uncommon in individuals with absence of pectoralis major muscle with or without other changes of Poland anomaly. Routine ultrasonography and analysis of urine in all such individuals would help the early detection of these abnormalities so appropriate measures may be taken to prevent further damage.

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