

*East African Medical Journal Vol. 87 No. 11 November 2010*

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## POLAND SYNDROME WITH DEXTROCARDIA: CASE REPORT

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### SUMMARY

**Poland syndrome is a rare congenital condition presenting with typical features including an absent costosternal head of pectoralis major and ipsilateral brachysyndactyly. There are many clinical variations of the syndrome including rib defects, absence of shoulder girdle muscle and breast hypoplasia or agenesis. Dextrocardia is rarely associated with Poland Syndrome with only 22 cases being previously reported in the worldwide literature. Whereas 'classical' Poland syndrome is predominantly right sided, all cases associated with dextrocardia have been left sided. We report a further case of left sided Poland syndrome with dextrocardia which might have important implications for the understanding of the pathogenesis of this unusual condition.**

### INTRODUCTION

The eponymous syndrome was first described by Alfred Poland of Guy's Hospital, London, in 1841 (1). It is a rare congenital condition occurring in approximately one in every 30,000 to 100,000 live births. Classical Poland Syndrome has the typical features of hypoplasia or absence of the costosternal portion of the pectoralis major muscle and ipsilateral brachysyndactyly (2). Numerous clinical variations of the syndrome exist including rib defects, absence of shoulder girdle muscles, and breast hypoplasia or agenesis athelia, vertebral anomalies and renal malformations (3). The association of dextrocardia with the condition is extremely rare with only 22 cases being previously reported in the worldwide literature (4,5). Whereas 'classical' Poland syndrome is predominantly right sided (approximately 70% of cases) and not normally associated with rib defects, all reported cases associated with dextrocardia have been left sided with coexisting rib anomalies (4,6). We present the case of Poland syndrome and dextrocardia in a young child and proceed to discuss how this supports the hypothesis that the cardiac anomaly is probably a consequence of the 'Poland sequence'.

### CASE REPORT

A three month-old male baby was referred to the Reconstructive Plastic Surgery Clinic at CoRSU Hospital as the parents were concerned about a mobile bulge on the left side of the chest wall. Following an

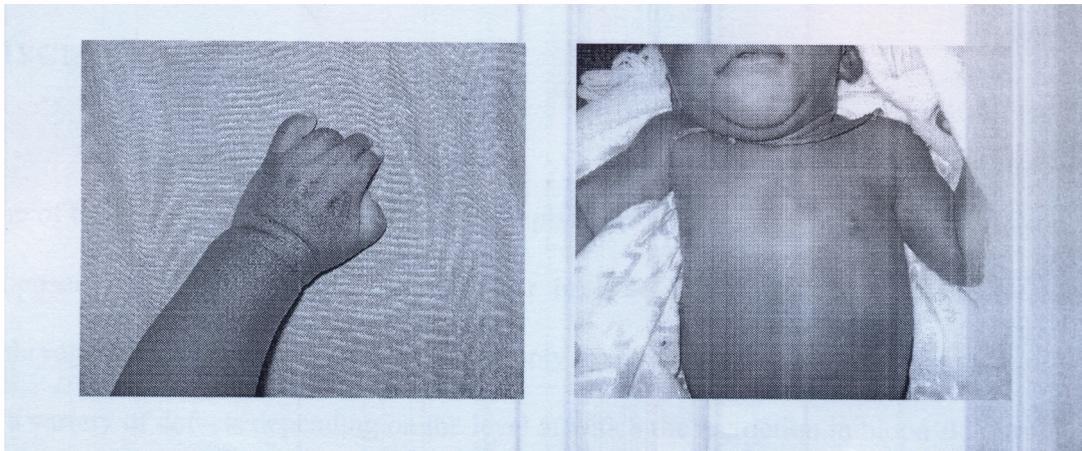
uneventful pregnancy, the child was born at term by spontaneous vaginal delivery. The parents were non-consanguineous with no family history of Poland Syndrome, cardiac or limb anomalies.

On examination, the otherwise healthy infant was found to have brachysyndactyly of the second webspace of the left hand, an absent left pectoralis muscle (and thus a deficient ipsilateral anterior axillary fold), and an obvious dynamic bulging and retraction of the skin during respiration at the level of the left anterior fourth and fifth ribs (Figures 1 and 2). The left nipple was sited higher compared to the contralateral side but was otherwise normal. The apex beat was palpable just medial to the right nipple within the fourth intercostal space cardiovascular examination was otherwise unremarkable. There was no evidence of Moebius, syndrome (congenital bilateral absence of the VI and VII cranial nerves) or Sprengel's deformity (an abnormally elevated, often hypoplastic, scapula, which may be associated with Klippel-Feil syndrome - a congenital disorder characterised by fusion of the cervical vertebrae).

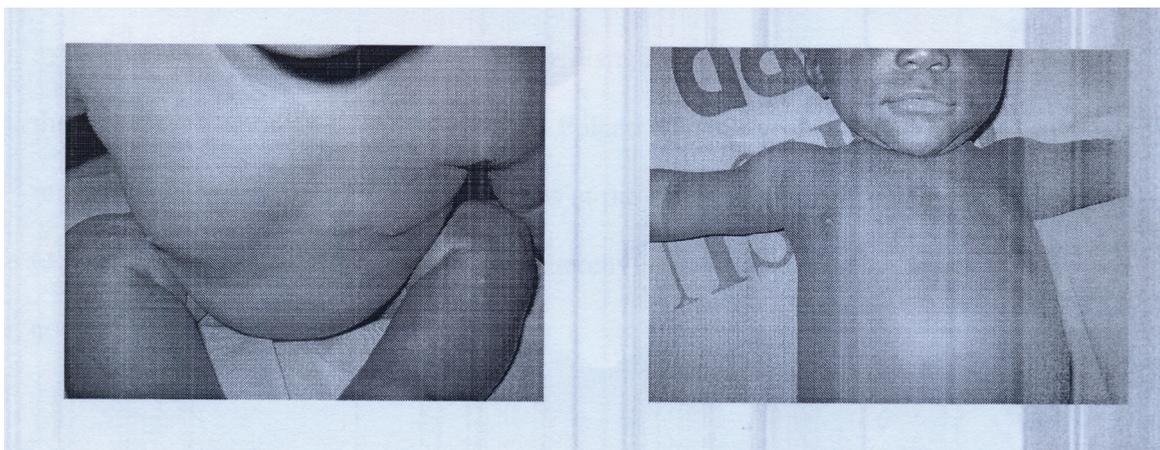
Radiological examination (Figure 3) and echocardiography demonstrated dextrocardia with hypoplasia of the left fourth and fifth ribs. The cervical vertebrae and scapulae were normal. There was no evidence of any intracardiac anomaly and the great vessels appeared normal. Abdominal examination was unremarkable with no ultrasonographic evidence of situs invertus. The kidneys were normal. Karyotyping was not performed.

**Figure 1**

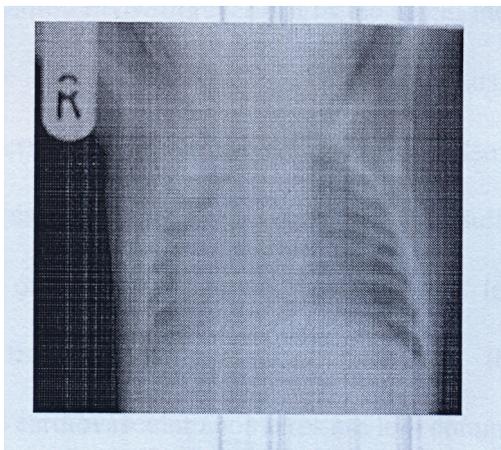
*Left sided branchysyndactyly (left) with an absent left anterior axillary fold (right)*

**Figure 2**

*Left sided chest wall hypoplasia with lung herniation at the level of the nipple*

**Figure 3**

*Dextrocardia and hypoplasia of the sternal ends of the left fourth and fifth ribs*



## DISCUSSION

One of the most convincing hypotheses to explain the pathogenesis of Poland Syndrome was proposed by Bouwes Bavinck and Weaver in 1986 (7). It postulates that during early embryogenesis, a disruption of the blood supply in the subclavian or vertebral arteries results in a variety of defects depending on the level at which the restriction in blood flow occurred. This could provide a common pathogenesis to Poland, Klippel-Feil, Moebius and Sprengel syndromes. This 'subclavian artery supply disruption sequence' could, for example, explain the chest wall anomalies encountered in Poland Syndrome due to the involvement of the internal mammary artery (which originates at the origin of the subclavian artery) in addition to the Moebius syndrome phenotype whereby flow in the basilar artery is presumably compromised.

Dextrocardia is a rare embryologic malformation in which the major axis (base to apex) of the heart lies in the right hemithorax with reversion of the apical inclination. If the abdominal viscera are transposed, the condition is known as situs inversus, however in the case of isolated dextrocardia, the term situs solitus may be used (6,8). Isolated dextrocardia may be due to a dextroposition (essentially cardiac displacement resulting from either reduced volume of the right lung or a space occupying lesion in the left hemithorax) or a true dextroversion (resulting from a rotational abnormality of the cardiac loop during organogenesis). Associated cardiovascular anomalies are less commonly seen in the case of a 'mechanical' dextroposition than a dextroversion (4).

However the clinical evidence, as seen in our case, might actually support the view that the dextrocardia is secondary to the thoracic anomalies in Poland Syndrome. That is to say that the dextrocardia is a dextroposition resulting from the thoracic pathology caused by the presumptive vascular event. This perspective is supported by the observation that associated cardiovascular anomalies in Poland Syndrome with dextrocardia is extremely rare, whereas cardiovascular anomalies are almost always seen in isolated dextrocardia. Furthermore isolated dextrocardia is not associated with rib anomalies, whereas in isolated Poland Syndrome the incidence of a rib anomaly is approximately 20%. However in Poland Syndrome with dextrocardia, the incidence of rib defects is 100%, as seen in our proband (4). Indeed dextrocardia with Poland Syndrome has not been reported in cases with a solitary affected rib; perhaps the mechanical influence of an isolated rib defect is insufficient to manipulate cardiac migration. This supports the hypothesis first proposed by Fraser in 1997 that the thoracic wall defect in Poland Syndrome is the driving mechanical force for the secondary dextrocardia - that is to say that it is a dextroposition whereby a structurally normal heart is displaced towards the right hemithorax (6).

Interestingly a case was reported of a male child born with Poland Syndrome including multiple rib agenesis) and dextrocardia with a documented normal cardiac position in early pregnancy (9). Due to a complex maternal obstetric history and the pregnancy being achieved by *in vitro* fertilisation, regular antenatal ultrasound surveillance scans were undertaken. The 12-week scan revealed a singleton foetus with a low risk of aneuploidy based on normal nuchal translucency thickness, The 21-week scan demonstrated a normal cardiac position, whereas a dextrocardia was noted on the 31-week scan. Thus whereas the presumptive vascular event which lead to the Poland phenotype would have occurred early in the first trimester, the abnormal cardiac position was proven to occur in the third trimester, thus supporting the view that the thoracic defect of Poland Syndrome

precedes, and is possibly responsible for, the cardiac malpositioning.

In conclusion, dextrocardia is only associated with left sided Poland Syndrome when two or more ribs are affected with accompanying lung herniation. Patients with Poland Syndrome should undergo a thorough physical examination with appropriate radiological investigations in order to exclude cardiac, renal or other anomalies. This case, although rare, provides a valuable insight into the mechanistic nature of this fascinating condition and support the view that the cardiac anomaly is a dextroposition resulting from mechanical intrauterine displacement of a structurally normal heart into the contralateral hemithorax as the abnormal left sided rib cage would be prone to compression *in utero*. It is anticipated that this child will undergo correction of the brachysyndactyly at the age of approximately 12 months whilst further reconstruction of the left anterior axillary fold or chest wall defect would usually be deferred until adulthood. The parents are counselled that the recurrence rate for Poland Syndrome is extremely low.

'Isolated' dextrocardia occurs more frequently in patients with Poland Syndrome than one would expect by chance, with a concurrence rate of up to 11.5% (4,6). Due to the rarity of both conditions (the incidence of Poland Syndrome and isolated dextrocardia are similar at approximately one in 30,000 live births) this relatively high concurrence suggests that the two conditions are related in some manner, but which comes first?

In all 23 reported cases of Poland Syndrome with dextrocardia (including this case), the left side has been affected, whereas isolated Poland Syndrome normally lateralises to the right. Indeed not a single case of right-sided Poland Syndrome with dextrocardia has been reported, in the literature. In the majority of the reported cases the dextrocardia appeared to be a dextroposition, rather than a dextroversion. Whereas patients with isolated dextrocardia almost always have an associated cardiac anomaly, the heart is usually structurally) and functionally normal when the condition is associated with Poland Syndrome.

The question arises as to whether dextrocardia precedes or follows the thoracic wall and upper limb anomalies in the developmental sequence that represents Poland Syndrome with dextrocardia. One might suppose that if dextrocardia were to occur first as a true dextroversion, then the left subclavian artery (a direct branch off the aorta) might be more susceptible to disruption than the right subclavian artery (which is a branch of the brachiocephalic artery). The spatial rotation of the more distal thoracic aorta branches about the cardiac axis would therefore be greatest which might predispose them to the higher risk of vascular injury. This might account for the Poland Syndrome with dextrocardia phenotype being exclusively left-sided.

## REFERENCES

1. Poland, A. Deficiency of the pectoralmuscles, *Guy's Hospital Reports*. 1841; **6**: 191.
2. Ram, A. and Chung, K. Poland's syndrome: current thoughts in the setting of a controversy. *Plast. Reconstr. Surg.* 2009; **123**: 949-953.
3. Fokin, A.A. and Robicsek, F. Poland's syndrome revisited. *Ann. Thorac. Surg.* 2002; **74**: 2218-2225.
4. Torre, M., Baban, A., Buluggiu, A., et al. Dextrocardia in patients with Poland syndrome: phenotypic characterization provides insight into the pathogenesis: *J. Thorac. Cardiovasc. Surg.* 2010; **139**: 1177-1182.
5. Lacorte, D., Marsella, M. and Guerrini, P. A case of Poland syndrome associated with dextroposition. *Ital. J. Pediatr.* 2010; **36**: 21.
6. Fraser, F.C., Teebi, A.S., Walsh, S. and Pinsky, L. Poland sequence with dextrocardia: which comes first? *Am. J. Med. Genet.* 1997; **73**: 194-196.
7. Bouwes Bavinck, J.N. and Weaver, D. Subclavian artery supply disruption sequence: hypothesis of a vascular aetiology for Poland, Klippel-Feil, and Mobius anomalies. *Am. J. Med. Genet.* 1986; **23**: 903-918.
8. Mutlu, M., Sildiroglu, O., Basekim, C.C. and Kizilkaya, E. A variant of Poland syndrome associated with dextroposition. *J. Thome. Imaging.* 2007; **22**: 341-342.
9. Sepulveda, W. Poland syndrome: a rare cause of cardiac dextroposition in the fetus. *Prenar. Diagn.* 2009; **29**: 903-905.