CASE REPORT

Left-sided Poland's syndrome in a girl with rare associations like spina bifida and diaphragmatic hernia

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SUMMARY

Poland's syndrome is a rare congenital anomaly characterised by partial or complete absence of sternocostal head of pectoralis major muscle and anomalies of ipsilateral hand and digits. Other associated anomalies involving anterior thoracic wall, breast, diaphragm and vertebrae have also been reported in various cases. We report a case of a 10-year-old girl, with features of left-sided Poland's syndrome associated with spina bifida, dextroposition of the heart and left-sided diaphragmatic hernia. These are rare associations of Poland's syndrome. She was investigated with chest X-ray, contrast-enhanced CT of the thorax, ultrasonography of abdomen and echocardiography which helped in arriving at an accurate diagnosis and assessing all the associated abnormalities.

BACKGROUND

Poland's syndrome is a congenital anomaly characterised by partial or complete absence of sternocostal head of pectoralis major muscle and anomalies of ipsilateral hand and digits.^{1–3} This case is reported because Poland's syndrome as such is a rare entity and familiarity with its clinical features and various associations is important for making an accurate diagnosis. Our patient is a girl, pathology was on left side and she showed features like spina bifida, dextroposition of heart and diaphragmatic hernia. All these are rare among the reported cases of Poland's syndrome.

CASE PRESENTATION

A 10-year-old girl, a first born to non-consanguineous parents presented with flattening of left chest wall since birth. Except for cosmetic disfigurement, patient was asymptomatic. She was born through normal vaginal delivery and weighed 2.5 kg at birth. Neonatal period was uneventful. There was no history of maternal drug intake or exposure to radiation during antenatal period. There was no family history of similar disorder. Her developmental milestones were normal.

On clinical examination, her chest wall was asymmetric with hypoplasia of the left side and underdeveloped left nipple (figure 1A). She had a shapeless left shoulder with hypoplastic left upper limb. Apex beat was located in the right hemithorax close to midline.

INVESTIGATIONS

The chest X-ray showed hyperlucency of left hemithorax, mediastinal shift to right side,

dextroposition of heart, hypoplasia involving the anterior portions of left fourth to sixth ribs and elevated left hemidiaphragm (figure 1B). X-ray of the left forearm showed radio ulnar synostosis (figure 1C). The hand X-ray showed no abnormality. CT of the thorax with and without intravenous contrast showed, hypoplasia of the pectoralis major and minor muscles, intercostal muscles and serratus anterior muscle on left side (figure 2A). CT also revealed hypoplasia of the left lung with maintained bronchovascular as well as lobar and segmental pulmonary anatomy (figure 2B). Heart was located in the right hemithorax with the apex normally directed towards left (figure 2C). Left hemidiaphragm was indistinct and there was herniation of part of left lobe of the liver and fundus of the stomach into the left hemithorax (figure 2D). Left fourth to sixth ribs were hypoplastic. Sternal rotational anomaly was present and there was spina bifida of D2–D4 vertebrae (figures 3A,B). Ultrasonography of abdomen and echocardiography were normal except for dextroposition of the heart. Correlating the clinical and imaging findings, the diagnosis of Poland's syndrome was made.

DIFFERENTIAL DIAGNOSIS

Anterior thoracic hypoplasia: Hypoplasia of the ipsilateral breast, superior location of the nipple–areola complex compared with the contralateral side with normal pectoralis muscles.

Amazone Syndrome: If Poland's syndrome is associated with ipsilateral hypo or aplasia of the breast as a leading feature in a female patient, it is called Amazone syndrome. Aetiology of the Amazone syndrome is simultaneous damage to an extremity bud and the lacteal ridge.

Klippel–Feil anomaly: It is characterised by abnormal union or fusion of two or more bones of the cervical vertebrae. Characteristic clinical triad includes abnormally short neck, restricted movement of the head and neck, and a low posterior hairline. Anomalies of the aortic arch, carotid arteries, subclavian artery, vertebral artery and a persistent trigeminal artery are known in association with KFS. The arches of the vertebrae may fail to unite posteriorly, resulting in spina bifida occulta.

TREATMENT

She was referred to a plastic surgeon for further management. Chest wall reconstruction was planned after completion of her growth.

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Figure 1 (A) Depressed left thoracic wall and hypoplastic left nipple and left upper limb. (B) Chest X-ray reveals hyperlucent left hemithorax, dextroposition of heart, hypoplastic left ribs and elevated left hemidiaphragm. (C). X-ray of the left forearm showing proximal radioulnar synostosis (arrow).

DISCUSSION

In our case, the patient is a girl and the affected side is left, both of which are less common among the reported cases of Poland's syndrome. There were hypoplasia of the left lung and dextroposition of the heart and both these can be attributed to reduced volume of the left hemithorax caused by hypoplasia of thoracic wall muscles and fourth to sixth ribs. Dextroposition of the heart in left-sided Poland's syndrome is not a coincidence but secondary to reduced volume of the left hemithorax.⁴ Lacorte *et al*³ also supports the same view. Our patient also had left-sided diaphragmatic hernia with herniation of part of left lobe of the liver and fundus of the stomach into the left hemithorax which is also a rare but documented association of Poland's syndrome.⁵ Her left upper limb was hypoplastic and there was radio ulnar synostosis, which are common associations with classical Poland's syndrome. No obvious digit malformations were noted in our case. Vertebral defects in previously reported cases of Poland's syndrome include hemivertebra and scoliosis. But in our case, the patient had spina bifida involving three consecutive upper thoracic vertebrae which is very rare.

Poland's syndrome is a rare congenital anomaly first described by Sir Alfred Poland in 1841. It is also known as Hand and ipsilateral thorax syndrome, subclavian artery supply disruption sequence and Fissure thoracis lateralis.⁴⁻⁷ The original description of Poland's syndrome included absence of the sternal head

of pectoralis major, pectoralis minor, serratus anterior, and external oblique muscles associated with brachy syndactyly. The definitive diagnostic criterion for Poland's syndrome is aplasia or hypoplasia of the pectoralis major muscle with at least one associated abnormality.¹ Other abnormalities frequently associated with Poland's syndrome include costal aplasia / hypoplasia, atelia, amastia, absence of axillary hair, hypoplasia of subcutaneous fat, radius abnormalities and hand malformations.¹ Other major systemic anomalies reported in Poland's syndrome are central nervous system anomalies like microcephaly, cerebral atrophy, myelination disorders, paralysis of cranial nerves and psychosocial retardation, cardio vascular system anomalies like situs inversus or dextrocardia, skeletal anomalies like hemivertebra and scoliosis, gastrointestinal tract anomalies like gastroschisis and urinary system anomalies.⁸⁻⁹ Platelet disorders, malignancies like leukaemia and lymphoma are also reported associations.¹ It is more common in boys with a male to female ratio of 3:1 and more commonly present on right side (75%). The incidence of Poland syndrome is 1:20 000 to 1:50 000.¹⁻² Most cases are sporadic although autosomal dominant inheritance, multifactorial inheritance and para-dominant inheritance have also been reported.¹⁰ The exact aetiology of Poland's syndrome is unknown. Many theories including mechanical factors, genetic factors, teratogenic effects and interruption of the subclavian artery during fetal development have

Figure 2 (A) Axial CT of the thorax showing hypoplastic left pectoralis muscles (arrow). (B). Axial CT of the thorax showing hypoplastic left lung. (C) Axial contrast-enhanced CT of the thorax revealing dextroposition of the heart. (D) Axial contrast-enhanced CT of the thorax revealing herniation of left lobe of liver and fundus of stomach (arrows) into left hemithorax.

been suggested as probable aetiologies for Poland's syndrome.⁷ A defect in the blood supply by the subclavian or vertebral arteries or their branches during the sixth week of embryonic development is the most widely accepted hypothesis. According to this hypothesis, disruption of the subclavian artery supply would lead to maldevelopment of the pectoralis major muscle and breast, rib defects, limb deformities and hypoplasia of subcutaneous tissue on the chest wall.⁵ The degree of chest wall and limb abnormalities depends on the duration and intensity of vascular interruption.¹ Besides Poland's syndrome, the subclavian artery disruption sequence can also cause anomalies like Klippel Fiel anomaly, Mobius syndrome, Sprengel's anomaly and transverse limb defects.²

The diagnosis is generally postnatal. There are only two case reports in which the diagnosis is made prenatally.¹¹ Since the functional disability will be minimal in most of the cases of Poland's syndrome, patients usually present late to the clinician. Various surgical reconstructive procedures are available to correct the functional disabilities and cosmetic disfigurement associated with this syndrome. Treatment by surgical correction of the chest wall deformity is usually carried out after completion of growth. Customised silicone prosthesis and transposition of the latissimus dorsi muscle for soft-tissue reconstruction have been used with acceptable results. Emergency surgery is reserved for those with compromised respiratory function.¹²

Figure 3 (A) Axial CT of the thorax showing hypoplastic left fourth rib (thin arrow) and spina bifida (thick arrow) of the D4 vertebra. (B) Three-dimensional volume rendered reconstruction of thoracic CT showing hypoplastic ribs on left side and sternal rotation.

Learning points

- ▶ Poland's syndrome is a rare congenital anomaly characterised by hypoplasia/agenesis of thoracic wall muscles, anomalies of ipsilateral ribs and upper limb, vertebrae as well as anomalies affecting various organ systems.
- ▶ Probable aetiology is disruption of the subclavian artery during embryonic life.
- ▶ Familiarity with this syndrome is important in order to make accurate diagnosis and should be followed by detailed clinical examination and relevant investigations to find out other associated major and minor anomalies.
- ▶ CT has got an important role in diagnosing this syndrome as well as assessing the extent of involvement of thoracic wall and internal organs of the thorax and abdomen.

Contributors SVC acquired data drafted the article, analysed and interpreted the data, revised the article critically for important intellectual content, and finally approved the version for publishing. SN acquired data, drafted the article, and finally approved the version for publishing. PPN analysed and interpreted data, revised the article critically for important intellectual content, finally approved the version for publishing. MLP analysed and interpreted data, revised the article critically for important intellectual content, and finally approved the version for publishing.

Competing interests None.

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