# Prenatal Ultrasound Diagnosis of Poland Syndrome

Dario Paladini, MD,
Maria Rosaria D'Armiento, MD, and
Pasquale Martinelli, MD

*Fetal Cardiology Unit, Department of Gynecology and Obstetrics, and Department of Pathology, University Federico II of Naples, Italy*

**BACKGROUND: Poland syndrome is a congenital nongenetic anomaly characterized by unilateral chest wall hypoplasia, ipsilateral hand abnormalities, and hemivertebrae. It has not ben described so far in the fetus.**

**CASE: The patient was referred for suspected left-arm hypomelia at 22 weeks of gestation. On ultrasonography, we confirmed the presence of severe left-sided hypomelia and detected an asymmetry of the rib cage and 3 thoracic hemivertebrae. The absence of heart defects led us to make the putative diagnosis of Poland syndrome. After termination of pregnancy, the diagnosis was confirmed by the pathologist.**

**CONCLUSION: The possibility of diagnosing Poland syndrome in utero is important for proper management and counseling. If the syndrome is suspected in a fetus, counselors may refer to specific postnatal data to provide the couple with survival rates, treatment options, and results and morbidity figures. If the pregnancy is terminated, a detailed necropsy is warranted to confirm the diagnosis because familial transmission has been reported. (Obstet Gynecol 2004;104:00–00. © 2004 by The American College of Obstetricians and Gynecologists.)**

Poland syndrome is a rare congenital nongenetic anomaly with an incidence of 1:7,000 to 1:100,000 live births.[1] It represents a sporadic condition, but familial transmission has been reported in some cases.[2] Poland syndrome is characterized by unilateral chest wall hypoplasia, ipsilateral hand abnormalities, and hemivertebrae and affects the right side of the body in 60–75% of the cases.[1] According to the prevailing pathogenetic theory, it is the interruption of the embryological blood supply to the subclavian artery at the end of the 6th week of gestation that causes the syndrome, with the severity of the anomalies depending on the site and the degree of flow impairment.[3] In fact, Poland syndrome covers a spectrum of anomalies ranging from complete absence of pectoralis major and minor muscles, amastia, severe rib anomalies, and absence of the hand to minor hand abnormalities and isolated pectoralis minor muscle aplasia.[4] We report here the prenatal ultrasound diagnosis of Poland syndrome made at 22 weeks of gestation in a fetus with severe left hypomelia and rib cage asymmetry.

Address reprint requests to: Dario Paladini, MD, Via Petrarca, 72, 80122 - Naples, Italy; e-mail: paladini@unina.it.

## CASE

A gravida 2 was referred to our attention at 22 weeks of gestation for suspicion of left hypomelia in the fetus. By ultrasonography we confirmed the presence of severe hypoplasia of the whole left arm and hand (Fig. 1A). In particular, the clavicle, humerus, ulna, and metacarpals were highly hypoplastic, and the radius was completely absent. In addition, we detected an evident asymmetry of the rib cage (Fig. 1B), with at least 4 hypoplastic ribs on the right side and the presence of 3 thoracic hemivertebrae (Fig. 1C). The absence of heart defects and the detection of at least 3 hemivertebrae led us to make the putative diagnosis of Poland syndrome, considering the presence of CHILD (Congenital Hemidysplasia with Ichthyosiform erythroderma and Limb Defects) syndrome less likely, although not completely excludible. An attempt was made to visualize the left subclavian artery by color Doppler, but the extent of the lesion and the fixed position of the left hypoplastic arm on the thorax impaired proper visualization. After counseling, the couple opted for termination of pregnancy. AQ: 1 F1

The necropsy of the 426-g female fetus confirmed the diagnosis of Poland syndrome. In particular, the following abnormalities were detected on the babygram: the left arm was highly hypoplastic, with the hypoplasia involving the scapulo-humeral joint, clavicle, humerus, and ulna. The radius was absent. The hand presented oligo-brachy-syndactyly, with 2 metacarpal rays, agenesis of the 5th digit, and syndactyly of the 2nd and the 3rd ones (Fig. 2). The ipsilateral foot had 3 metatarsal rays. As for the spine, 4 thoracic hemivertebrae were present. At dissection of the specimen, a pterygium-like attachment of the malformed left arm to the thorax was noted, with athelia (complete absence of the nipple and areola) and amastia. The sterno-costal head of the pectoralis major muscle and the whole of the pectoralis minor muscle were absent. The rib cage was asymmetrical, with rib abnormalities on the right side. The heart was unremarkable. F2 AQ: 2

VOL. 104, NO. 2, AUGUST 2004

0029-7844/04/$30.00
doi:10.1097/01.AOG.0000128115.55051.90

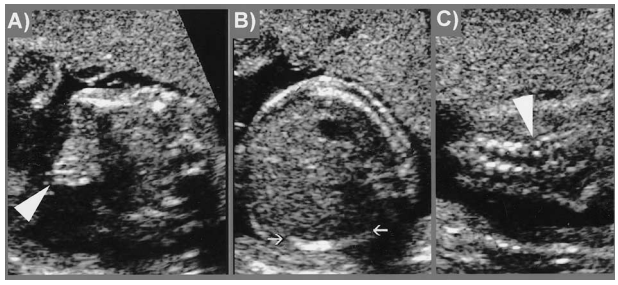


**Figure 1. A.** Oblique scan of the fetal thorax showing the severe hypomelia. The arm is flexed at the elbow. Note that no radial and ulnar ossification nuclei are visible in the forearm and that the single digits cannot be distinguished because of the oligo-brachy-syndactyly *(arrowhead)*. **B.** Transverse view of the fetal upper abdomen, showing the hypoplasia of the ribs on the right side *(arrows)*. The sonolucent area regularly positioned in the left hemi-abdomen is the stomach. **C.** Coronal view of the upper thoracic spine demonstrating one of the hemivertebrae *(arrowhead)*.

*Paladini. Prenatal Diagnosis of Poland Syndrome. Obstet Gynecol 2004.*

## COMMENT

Poland syndrome is an extremely rare condition characterized by hypoplasia of the breast and nipple, absence of the costo-sternal head of the pectoralis major muscle, absence of the pectoralis minor muscle, rib abnormalities, and unilateral brachysyndactyly.[3] Although these abnormalities, when severe, are potentially detectable by ultrasonography in utero, this condition has not been described in the fetus before. In particular, we could not find any article in the English literature describing the occurrence of Poland syndrome in a human fetus in a MEDLINE search (http://www3.ncbi.nlm.nih.gov/Entrez) conducted with the terms "Poland syndrome" or "Poland sequence" and "fetus," with no time limits.

Differential diagnosis in the fetus is limited to those conditions characterized by body asymmetry, such as CHILD syndrome, and to the cluster of anomalies caused by the occurrence of thoracic hemivertebrae, such as chest asymmetry and rib hypoplasia.[5] However, AQ: 3 in cases of isolated thoracic hemivertebrae, upper limb malformations are usually absent. The real problem is represented by CHILD syndrome. This rare X-linked condition, recognized for the first time in 1968,[6] has been demonstrated to be caused by mutations in the glutathione reductase (NAD[P]H) steroid dehydrogenase-like AQ: 4 protein gene.[7] It is characterized by unilateral hypomelia, skin hypoplasia, and, often but not always, major heart defects. Because the erythroderma of CHILD syndrome and abnormalities of the chest muscles of Poland syndrome are not detectable by ultrasonography in utero, the sonographic appearance of the 2 entities is similar, both featuring a unilateral limb hypomelia with a variable degree of arm and hand involvement. The differential diagnosis is made even more difficult by the possible occurrence in CHILD syndrome of vertebral anomalies and rib hypoplasia. A possible hint in the differential diagnosis issue is the common occurrence in CHILD syndrome of major heart defects, which are indeed detectable in utero.[8] However, the final diagnosis can be reached only at autopsy, when anomalies of the pectoralis muscles and the athelia/amastia are found in Poland syndrome (as in the index case), and the skin defects (erythroderma, regional alopecia, etc) become evident in CHILD syndrome. In conclusion, if unilateral rib, chest, and arm anomalies are found at routine midtrimester ultrasonography, both Poland and CHILD syndromes represent possible diagnoses. In such an occurrence, the detection of congenital heart disease might indicate the likely presence of CHILD syndrome, whereas the disclosure of thoracic hemivertebrae would support a diagnosis of Poland syndrome.

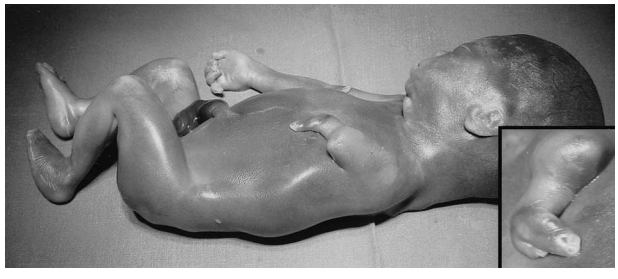

**Figure 2.** The specimen at birth. Note the hypomelia, the deformation of the chest, and the oligo-brachy-syndactyly (magnification of the hand in the lower right-end panel).

*Paladini. Prenatal Diagnosis of Poland Syndrome. Obstet Gynecol 2004.*

The possibility of diagnosing Poland syndrome in utero is important for proper management and counseling. Should the syndrome be suspected in a fetus, the counselors may refer to specific postnatal data to provide the couple with survival rates, treatment options, and results and morbidity figures. In case of termination of pregnancy, the prenatal recognition of the syndrome strengthens the need for a detailed necropsy to confirm the diagnosis because familial transmission has been reported in some instances.[2] In these families, the ability to reassure the couple that the reoccurrence of Poland syndrome can be excluded during the second trimester of pregnancy is of the utmost importance. However, taking into consideration the variable expression of the syndrome and the ultrasound findings in the index case, 2 further points need be addressed: 1) It is likely that, in high-risk families, full-blown forms of the syndrome could be detected by ultrasonography as early as the 12th–13th gestational weeks, whereas mild forms, fea-

turing only pectoralis minor muscle hypoplasia/aplasia and subtle digit anomalies, may escape prenatal ultrasound diagnosis completely. 2) Ultrasound differential diagnosis between Poland and CHILD syndromes may be impossible in some cases, with the final diagnosis based only upon postmortem findings.

## REFERENCES


1. Freire-Maia N, Chautard EA, Opitz JM, Freire-Maia A, Queice-Salgado A. The Poland syndrome: clinical and genealogical data, dermatoglyphic analysis, and incidence. Hum Hered 1973;23:97–104.
2. Sujansky E, Riccardi VM, Mathew AL. The familial occurrence of Poland syndrome. Birth Defects Orig Artic Ser 1977;13:117–21.
3. Bavinck JN, Weaver DD. Subclavian artery supply disruption sequence: hypothesis of a vascular etiology for Poland, Klippel-Feil, and Möbius anomalies. Am J Med Genet 1986; 23:903–18.
4. Fokin AA, Robicsek F. Poland's syndrome revisited. Ann Thorac Surg 2002;74:2218–25.
5. Fraser FC, Teebi AS, Walsh S, Pinsky L. Poland sequence with dextrocardia: which comes first? Am J Med Genet 1997;73:194–6.
6. Falek A, Heath CW Jr, Ebbin AJ, McLean WR. Unilateral limb and skin deformities with congenital heart disease in two siblings: a lethal syndrome. J Pediatr 1968;73:910–3.
7. Konig A, Happle R, Fink-Puches R, Soyer HP, Bornholdt D, Engel H, et al. A novel missense mutation of NSDHL in an unusual case of CHILD syndrome showing bilateral, almost symmetric involvement. J Am Acad Dermatol 2002; 46:594–6.
8. Paladini D, Russo MA, Teodoro A, Pacileo G, Capozzi G, Martinelli P, et al. Prenatal diagnosis of congenital heart disease in the Naples area during the years 1994–1999: the experience of a joint fetal-pediatric cardiology unit. Prenat Diagn 2002;22:545–52.


*Received November 3, 2003. Received in revised form January 20, 2004. Accepted February 5, 2004.*