SRIDHAR R. ALLAM, MD, MPH
Department of Internal Medicine, Cleveland Clinic

RUCHI YADAV, MD
Department of Diagnostic Radiology, Cleveland Clinic

MOULAY MEZIANE, MD
Head, Section of Chest Radiology, Department of Diagnostic Radiology, and Department of Pulmonary, Allergy, and Critical Care Medicine, Cleveland Clinic

ATUL C. MEHTA, MD
Vice Chairman, Department of Pulmonary, Allergy, and Critical Care Medicine; Head, Section of Bronchology, Transplant Center, Cleveland Clinic

CME CREDIT

The Clinical Picture

A middle-aged man with asymptomatic chest wall asymmetry

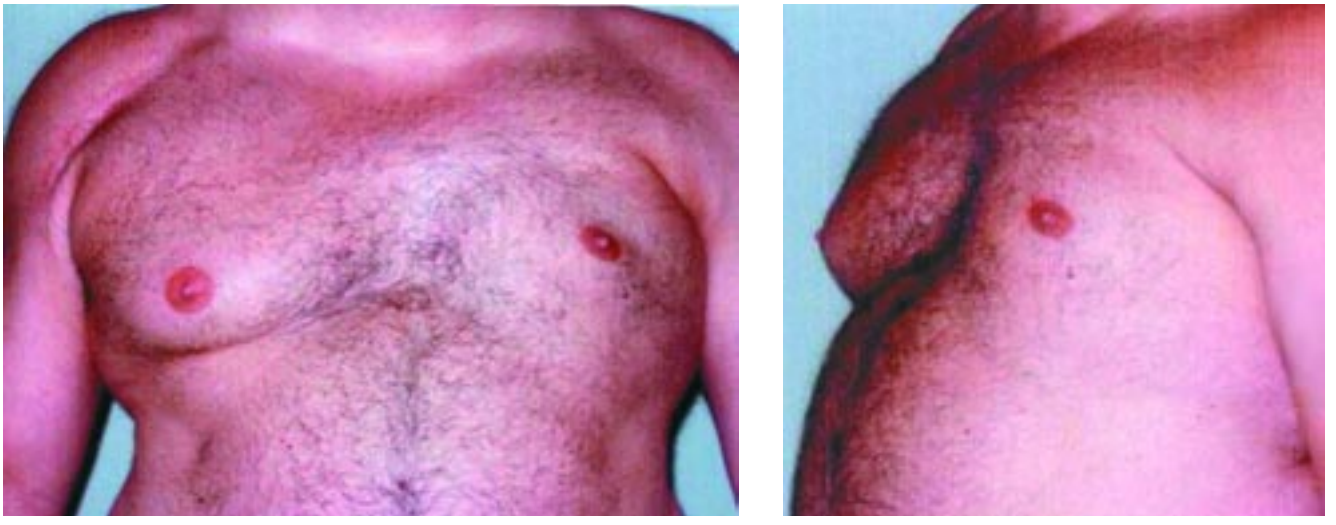

FIGURE 1

A 52-YEAR-OLD MAN presents with intermittent symptoms of bronchial asthma. Physical examination reveals reduced prominence of the rib cage on the left side (**FIGURE 1**). The patient has had no history of trauma or chest wall surgery, and he has been aware of the asymmetry since early childhood. His lungs are clear to auscultation. A frontal chest radiograph shows hyperlucency of the left hemithorax (**FIGURE 2**).

What is your diagnosis?

A CONGENITAL DISORDER

The differential diagnosis[1] of one-sided hyperlucency seen on chest radiography is shown in **TABLE 1.**

The patient is missing the sternal head of the pectoralis major muscle. This is considered the minimal expression of Poland syndrome, also known as Poland anomaly or Poland syndactyly.

Disease characteristics

Poland syndrome, described in a cadaver by Sir Alfred Poland in 1841 after a few earlier reports,[2] is characterized by unilateral absence of the sternal head of the pectoralis major muscle; hypoplasia of the rib cage and upper extremity; breast and nipple hypoplasia or aplasia; reduced subcutaneous fat, sweat glands, and chest and axillary hair; and scoliosis.[3–5]

Although Poland syndrome is traditionally described as unilateral, a bilateral case has been reported.[6]

Adjacent muscles, including the pectoralis minor, serratus, latissimus dorsi, and

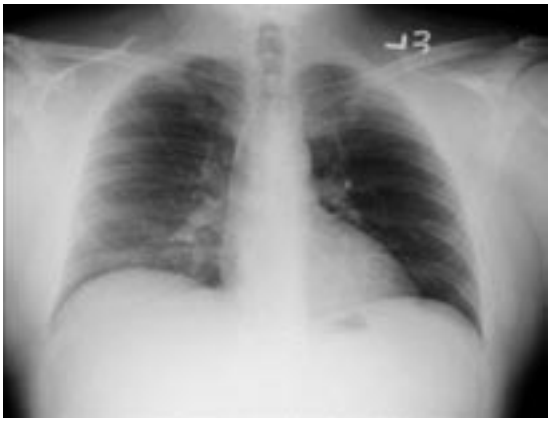

FIGURE 2. The patient's chest radiograph, posteroanterior view.

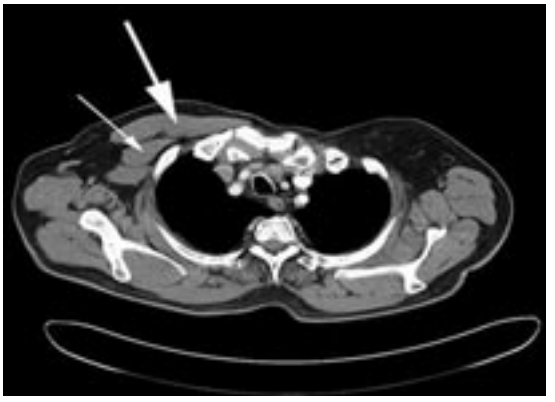

FIGURE 3. Axial computed tomographic scan through the upper chest demonstrates the absence of both left pectoralis major and minor muscles. Note the normal right pectoralis major (large arrow) and minor (small arrow).

external oblique, may also be involved. The nipple on the affected side is small and highly placed in many patients of either sex, and in many women the breast is hypoplastic. Skeletal deformities may include absence of the ribs or costal cartilages anteriorly. In severe cases, the lung may herniate anteriorly. The Sprengel deformity (congenital nondescent and hypoplasia of the scapula) may also be present.

The upper arm, forearm, and fingers may be shortened (brachysymphalangism), and simple, complete, or incomplete syndactyly with hypoplasia or aplasia of the middle phalanges may be found.

Patients may have abnormalities involving the gastrointestinal tract (diaphragmatic hernia), liver and biliary tract, kidney (agenesis or hypoplasia), testes, and heart (dextrocardia).

The syndrome has also been reported in association with neoplasias such as leukemia, non-Hodgkin lymphoma, cervical cancer, leiomyosarcoma, breast cancer, and lung cancer.

Frequency

Poland syndrome, a congenital developmental disorder, has a reported incidence of 1 in 7,000 to 100,000 persons.[2] Although most cases arise sporadically, genetic transmission also occurs. Sporadic cases tend to occur more often in males and commonly involve the right side. Familial cases have a more equal distribution between the sexes and do not have a right-sided predominance.

Poland syndrome has also been described in association with Möbius syndrome (unilateral or bilateral facial paralysis and defective extraocular eye movements secondary to congenital paresis of the facial [VII] and abducens [VI] cranial nerves) and Klippel-Feil syndrome (fused vertebrae, especially in the cervical spine, Sprengel deformity, and sometimes genitourinary abnormalities and deafness).

CT or MRI easily detects the absence of the pectoralis muscle

Etiology

Poland syndrome most likely develops from a critical vascular event, interruption of the embryonic blood supply in the subclavian artery (known as "subclavian artery supply disruption sequence" or SASDS) during the sixth week of gestation.[4] The specific region of subclavian artery involvement determines the clinical manifestation (ie, Poland syndrome, Möbius syndrome, or Klippel-Feil syndrome). A "disorganization mutation" has also been suggested as a cause of Poland syndrome.[7]

Clinical presentation

Although Poland syndrome may involve the muscular, skeletal, integumentary, nervous, and respiratory systems, it is associated with little disability. Many patients present in their late teens to inquire about cosmetic options for chest deformity and breast asymmetry.

Evaluation should include assessment of the following:

- Stage of breast development

TABLE 1

Causes of unilateral diffuse hyperlucency of the hemithorax on chest radiography

Technical factors
- Grid cutoff
- Patient rotation

Extrapulmonary conditions
- Soft tissue abnormalities such as Poland syndrome (absent pectoralis muscle), mastectomy and chest wall surgery
- Contralateral pleural effusion or thickening
- Thickening of soft tissues in the contralateral chest wall soft tissues

Pulmonary conditions
- Pneumothorax
- Diminished pulmonary blood flow, as in hypoplasia of lung or pulmonary artery, obstruction of pulmonary artery caused by embolism, mediastinal or hilar tumor, or fibrosing mediastinitis
- Diminished pulmonary blood flow or hyperinflation, as in lobar atelectasis, lobar resection, Swyer-James syndrome, endobronchial tumor, or foreign body with subsequent air trapping and increased lucency (ball-valve phenomenon)

ADAPTED FROM KLEIN JS. RADIOGRAPHIC FINDINGS IN CHEST DISEASE. IN: BRANT WE, HELMS CA, EDS. FUNDAMENTALS OF DIAGNOSTIC RADIOLOGY. 2ND ED. BALTIMORE: WILLIAMS & WILKINS; 1999:343–356.

- Status of the latissimus dorsi muscle; computed tomography (CT) is often required to help determine the surgical approach
- Extraocular eye movements to determine if Möbius syndrome is present
- Lymph nodes and complete blood count to determine if leukemia or non-Hodgkin lymphoma is present
- Renal function.

Imaging

The chest anomalies of Poland syndrome are usually incidentally detected on chest radiography, CT, or magnetic resonance imaging (MRI). On a frontal chest radiograph, the reduction of chest musculature is reflected as a hypodense hemithorax compared with the contralateral side. Cross-sectional CT or MRI of the chest makes the absence of the pectoralis muscle easily and readily detectable (FIGURE 3).

CT and MRI can also reveal the spectrum of anomalies that can occur in Poland syndrome, ranging from hypoplasia of the minor and major pectoralis muscles to their total absence. Skeletal deformities and nipple, cardiac, subdiaphragmatic, and contralateral upper limb anomalies can also be identified. CT or MRI can also help differentiate Poland syndrome from asymmetrical pectus excavatum or breast asymmetry.

Treatment

The treatment of Poland syndrome is guided by the severity of the deformity, age and sex of the patient, and the status of the latissimus dorsi muscle.[2] Indications for surgery are unilateral chest wall depression, lack of adequate covering to the heart and lungs, paradoxical respiration, or cosmetic defects.

In children with severe deformity, correction is made in two stages: rib aplasia is repaired initially, and muscle flap transposition is carried out after puberty. For children with mild deformity, treatment is deferred until after puberty.

In adults, stabilization and reconstruction of the chest wall with a simultaneous muscle transposition is carried out in a single procedure. If it is healthy, the ipsilateral latissimus dorsi muscle is used for muscle flap transposition; otherwise, microsurgical transfer of the contralateral muscle is used. If augmentation mammoplasty is desired, it should be performed as an additional procedure.

Separation of webbed fingers is usually started by 1 year of age and is completed by the time of school entry to prevent abnormal function patterns and progression of the deformity.

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ADDRESS: *Sridhar R. Allam, MD, MPH, Department of Internal Medicine, Box 445, Cleveland Clinic, 9500 Euclid Avenue, Cleveland, OH 44195; e-mail allams@ccf.org.*