Poland Syndrome

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Clinical Case: Patient A.M.

- 82 year old female presented in 2004 for a routine mammogram
- Breast asymmetry since adolescence
  Previously on HRT
- Has had routine yearly screening mammogram at BIDMC since 1991
  - difficulty with imaging
  - BIRADS1
Mammogram

- **Technique:** X-Ray generate images of compressed breast
- **Patient Preparation:** no powder, deodorant, or lotion on day of procedure, ideally in early stage of menstrual cycle
- **Standard Views:**
  - Cranio-caudal (CC)
  - Medio-Lateral-Oblique (MLO)
- **ROI:**
  - CC: entire breast, QC: posterior fat pad
  - MLO: entire breast, QC: pectoralis major (extend to nipple)
Mammogram (cont.)

• **Benefits:** high accuracy (77-80%), inexpensive ($80-$100)

• **Relative Contraindications:** pregnant women, <30 y.o., <1 wk. post-surgery, breast reconstruct/mastectomy site

• **Limitations:** patient discomfort, negative mammo does NOT rule out CA, dense breasts and breast tissue adjacent to chest wall/axilla are difficult to evaluate
Breast Imaging Reporting and Data System (BIRADS)

**SCORE**

0  incomplete assessment: need US, additional images, or comparison to prior films
1  negative
2  benign findings
3  probably benign
4  suspicious lesion
5  probably CA
6  known malignancy

**ACTION**

<table>
<thead>
<tr>
<th>Score</th>
<th>Action</th>
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<tbody>
<tr>
<td>0</td>
<td>If 0: Additional information needed-US, more images, compare to prior films</td>
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<tr>
<td>1-2</td>
<td>If 1-2: Image in 1 year</td>
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<tr>
<td>3</td>
<td>If 3: Follow up in 6mo</td>
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<tr>
<td>4-6</td>
<td>If 4-6: Take appropriate action – Bx or Sx</td>
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Valerie Fein-Zachary, M.D. HMS BI heart Radiology lecture 2006
Patient A.M. Mammogram 2004

- Breast Asymmetry
- Absent pec?
Differential Diagnosis

Unilateral breast asymmetry and suspected absence of pectoralis major

- Breast Asymmetry
  - Unilateral decrease in size
    - Breast hypoplasia
    - Amastia
    - Athelia
  - Unilateral increase in size

- Chest Asymmetry
  - Thoracic hemivertebrae
  - CHILD Syndrome: hemihypertrophy

- Combination of Chest and Breast Asymmetry:
  - Poland Syndrome

- Other
  - Progeria
Pectoralis Major: Quick Review

**Origin:** Sternal half of the clavicle, sternum to 7th rib, cartilages of true ribs, aponeurosis of external oblique

**Insertion:** Lateral lip of bicipital groove of the humerus

**Function:** Adducts, extends, internally rotates upper limb

**Innervation:** medial and lateral pectoral nerve

**Vascular supply:** pectoral branch of thoracoacromial artery and the internal mammary artery
Patient A.M.
Significant Past Medical History

- Breast deformity “familial in pattern according to patient”
- Hepatic hemangioma (date N/A)
- Claudication (date N/A)
- 2 weeks lightheadedness, nystagmus, changes in peripheral vision and gait, memory loss → evaluated for vascular abnormalities (2000)
- 3 mo. Hx of LEFT hand/arm swelling → evaluated for subclavian vessel abnormality (2001)
Magnetic Resonance Imaging

- **Technique:** magnetic field is used to align protons (mainly contained in Hydrogen in body), radiofrequency pulse then puts protons in higher energy state, when protons return to equilibrium state coil-detectors capture the radiofrequencies emitted. Different tissue = different $[H]$ = different signal intensity.

- **Patient Preparation:** NPO 2-4hr if w/ contrast, sedation for anxious patients, IM glucagon to decrease intestinal motility.

- **Standard Image Planes:** axial, sagittal, coronal, and oblique planes.

- **Image Acquisition Protocols:**
  1. **T1 W** - CSF = black, discharge of returning to original spatial of spin vector of proton for **ANATOMY**
  2. **T2 W** - CSF = white, discharge as a consequence of loss of coherence of processing protons for **EDEMA/PATH**
  3. **Time of Flight** - FLOW-RELATED enhancement of spins entering into an imaging slice, subtract stationary signal

- **ROI:** depends on region of body imaged.

www.primarycareradiology.com
MRI (cont.)

- **Benefits:** great soft tissue contrast, imaging in many planes
- **Contraindications:** ferromagnetic metallic devices, cochlear implants, pacemakers
- **Limitations:**
  - **Practical**
    - Limited availability of machines
    - Size - obese patients
    - Claustrophobia
  - **Technical**
    - Insensitivity to calcification and bone findings
    - Artifact from dental or other hardware
    - Motion sensitivity

Patient A.M.

Normal

PACS, BIDMC
Patient A.M.: Vessel Hypoplasia

- Right ACA
- Hypoplastic Left ACA
- Right MCA
- Left MCA
- Right ICA
- Left ICA
- Basilic
- Hypoplastic Right Vertebral Artery
- Left Vertebral Artery

PACS, BIDMC
Patient A.M.  
Additional Findings (2000)  

L parotid gland absent
Patient A.M. T1W FS MR Gd+ of Chest (Coronal, 2001): breast asymmetry

Contrast imaging showed no thrombus or occlusion
Review of Findings

- Unilateral hypoplasia of left breast and nipple
- Absence of ipsilateral pectoralis major
- Absence of ipsilateral pectoralis minor
- Vascular hypoplasia
- Absence of ipsilateral parotid gland

Other classical findings related to this syndrome:

- Aplasia or deformity of ipsilateral costal cartilage or ribs II-IV or III-V
- Alopecia of ipsilateral axillary and mammary region
- Ipsilateral brachysyndactyly “mitten hand” (phalanges 1, 3, 4)

Fokin et. Al. (2002)
Diagnosis

Poland Syndrome
Etiology

- **Etiology**: vascular event, subclavian artery supply disruption sequence (SASDS), during the critical 6th week of gestation with hypoplasia of the subclavian artery causing musculoskeletal malformations. This occurs when the medial and forward growth of the ribs forces the subclavian vessel into a U-shaped configuration. More proximal occlusions result in more severe syndromes.

- **Incidence**: 1:7,000 to 1:100,000 births
- **Sex Ratio**: M:F → 2:1 to 3:1 (Folin et. Al. (2002))
- **Environ. Factors**: maternal smoking 2-fold inc. risk (Martinez-Frias et. Al. (1999)). Maternal sex hormone intake & vaginal bleeding in 1st trimester? (Castilla et. Al. (1979))
- **Genetics**: mostly sporadic
  - Some familial cases: multifactorial w/ 2 predisposing factors
    1. aberrant vascular formation
    2. thrombophilia (Shalev and Hall (2003))
Findings with Poland Syndrome
Chest Wall Deformity

http://www.operationrestorehope.org/gallery/images/post-polands-syndrome1.jpg
Companion Patient #1 Chest X-Ray:
left breast abnormality and pectoralis major aplasia
Companion Patient #2 Chest X-Ray: 6th Rib Deformity
Companion Patient #3 CT w/ Contrast: Aplasia of Pectoralis Major and Minor w/ No Chest Wall Deformity
Companion Patient #4 CT w/ contrast: Chest Wall Deformity and Missing Pectoralis Major

Axial
Syndactyly in Poland Syndrome

Ailiwadi et. Al. (2005)
Clinical Consequences of Poland Syndrome

- Paroxysmal movements of the chest wall due to the malformation
- Reduction of lung capacity
- Lack of vital organ protection
- Muscular weakness
- Chest scoliosis
- Hand malfunction
- Psychological problems

Fokin et. Al. (2002)
Vascular Anomalies and Poland Syndrome

- **VASCULAR HYPOPLASIA**: present in diseased side (Beer et. Al. (1996))
  - pre-operative CT suggested for patients undergoing CABG to verify patency of LIMA (left internal mammary artery)
  - Case- Study: apparently normal LIMA lead to unremarkable post-op course (Aliwadi et. Al. (2005))

- **HEMANGIOMA** (Riyaz and Riyaz (2006))
Importance of Diagnosis!

• Patient A.M. also had:
  – ANGINA
  – ELEVATED CHOLESTEROL
  – HTN
  – PERIPHERAL ARTERY DISEASE

Potential future candidate for CABG

• Important to know status of vasculature: pre-op CT angiogram
Treatment Options for Poland Syndrome

• Prenatal screening for Poland synd. by ultrasound (22 wks) (Paladini et. Al. (2004))

• Corrective Surgery
  – Mammoplasty
    • Breast Implants (saline, silicone)
    • Muscle flaps (Fokin et. Al. (2002))
  – Thoracoplasty
Companion Patient #5 Corrective Breast Surgery: 71 y.o. female with Poland Syndrome post- elective contralateral breast reduction, ipsilateral breast implantation (Mammo MLO view)
Fun Facts: Is Poland Syndrome named after the country?
Answer: NO!
It is named after Sir Alfred Poland.

• 1841 Alfred Poland, then a British student-demonstrator in anatomy in London, described a cadaver in the *Guy’s Hospital Gazette*
  – lack of pectoralis major and minor muscles
  – ipsilateral syndactyly of phalanx 1, 3, 4
• 1861 promoted to surgeon status
• 1867 died of pulmonary TB

Fokin, et. al. (2002)
Acknowledgments

- Fabio Komlos, M.D.
- Gillian Lieberman, M.D.
- Pamela Lepkowski
References


- PACS, BIDMC
References (cont.)


• www.primarycareradiology.com

• www.polands-syndrome.com

• www.sindromedipoland.org

• www.thefetus.net/page.php?id=434