
Case Report

Poland's Syndrome Associated with Thoracic Spine Scoliosis-A Case Report

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Abstract:

Poland syndrome is a rare genetic condition, which involves the chest. There is no genetic testing. Diagnosis is reliant upon history and examination findings. An overlooked feature of Poland's syndrome is thoracic scoliosis. Spinal involvement may be predisposed by associated genetic muscle and skeletal anomalies affecting the serratus, latissimus dorsi, external oblique, ribs, sternum and spine. This paper reviews the history of a patient referred for progressive postural changes and back pain associated with idiopathic thoracolumbar scoliosis, who on examination actually had Poland's syndrome. Presentation of spinal scoliosis should prompt clinicians to investigate potential genetic causes, which may also impact patient management.

Keywords: Poland's syndrome, Scoliosis, Spine, Congenital Breast, Pectoralis Major, Latisimus Dorsi.

Introduction

Poland's syndrome bears the name of the 19th century Moorfield's and Guys Hospital British surgeon Alfred Poland. His dissection of the convict George Elt's body, described a phenotypical chest condition in an 1841 published paper entitled 'Deficiency of the pectoral muscles.' [Poland A, 1841]

Poland's syndrome is a rare genetic condition of variable penetrance, and is characterized by hypoplasia or absence of the breast or nipple, hypoplasia of subcutaneous breast tissue in females, absence of the costosternal portion of the pectoralis major muscle, absence of the pectoralis minor muscle, and absence of costal cartilages or ribs 2, 3, and 4 or 3, 4, and 5. The chest wall defect is often associated with a lung hernia. Associated traits may also include patchy absence of hair in the axilla. The Clinical manifestations are extremely variable and rarely are all the features recognized in one individual. [Urschel HC Jr, 2009]. Cases have been associated with leukemia, carcinoma of the hypoplastic breast, confirming the relationship between developmental defects and tumors, and require oncologic awareness. [Fokin et al 2002]

There is a 3:1 male to female ratio and the condition is sporadic in nature with a variable presentation. It may also be characterized by and ipsilateral short arm or syndactyly. The prevailing theory of its cause is hypoplasia of the subclavian artery or its branches, which may lead to a range of developmental changes. The incidence of Poland's syndrome varies between groups (male versus female patients, congenital versus familial cases, and so on) and ranges from 1 in 7,000 to 1 in 100,000 live births. Cases of Poland's syndrome associated with leukemia, carcinoma of the hypoplastic breast, and other conditions, confirm the

relationship between developmental defects and tumors, and require oncologic awareness. [Fokin et al 2002] Poland's syndrome may also express traits of spinal musculoskeletal abnormalities predisposing to scoliosis. [Bainbridge et al 1991]

Case Report

A 35 year old right-hand dominant female professional photographer was referred to a Sports and Musculoskeletal medicine outpatient service for the management of a progressively worsening thoracolumbar spine scoliosis and right shoulder pain. She had also reported a 6-month history of thoracic back pain, right scapulothoracic shoulder pain and sleep disturbance. The pain had worsened over several months and was no longer relieved by regular doses of paracetamol or cocodamol. Recommended physiotherapy exercises and Pilates and Yoga sessions 3 times per week had also not shifted the pain. Her upper back pain and shoulder complaints were on a background of chronic thoracolumbar pain of circa 10 years duration. Her concern was that the scoliosis had become progressive and the shoulder pain was affecting her ability to work as a photographer. She lived in a first-floor flat and had noted increasing back pain and difficulties carrying objects and packages up the stairs. She reported that part and parcel of her occupation was the daily wear of heavy shoulder-strapped cameras, lenses and other equipment, which she habitually carried on her right side. These activities also caused occasional arm weakness and pin and needles in the fingertips of her right hand. She had initially been referred to physiotherapy sessions a year prior, but had reportedly been unable to attend due to some struggles with depression. The pain was worse during these periods of low mood. Over the

years she had been helped by occasional chiropractic treatment for back pain and had been reviewed by orthopaedic surgeons for the scoliosis 10 years ago with conservative management advised. She reported that she had over the years learned to live with chronic pain, but her usual coping mechanisms were no longer working. She also perceived that the upper back and right scapular pain she now experienced was of a different character than her usual aches and pains.

On further questioning it was apparent that the progression of the her scoliosis, chest-shoulder pain and breast appearance had contributed to psychological distress and significant depression for many years. She was brought up in Kenya and reported a great deal of bullying while growing up, as her right breast had failed to develop. It was impossible to talk to her mother or other family member as such things were not to be talked about in her community and she never understood what was happening or why her chest had developed in that way. She was one of 5 siblings and has an 8-year-old daughter, with no significant family history of note.

Over time she had come to accept the appearance of her spinal deformities from a cosmetic point of view, but had attempted to improve spinal function and retard scoliosis progression by engaging in various exercise regimes. She had eventually sought breast reconstructive surgery in the private sector, but was greatly distressed by unsatisfactory results, with the right breast remaining grossly asymmetric, and the breast appearance continued to affect her feelings of confidence and self-worth.

She had no prior history of injury for fractures. She recollected undergoing a right breast reconstruction procedure in her 20's due to unequal growth. She also had a history of depression. Her medications included Cocodamol, paracetamol, zopiclone, and citalopram. She was a non-smoker and had no history of drug allergies. Her family history was reportedly unremarkable. She was a professional photographer and videographer, originally from Kenya, and she had a 7-year-old daughter.

On examination she was of normal height and weight. Straie was present on her trunk and bilateral flanks. She presented with a marked 'S' type right convex dexter-rotary thoracic scoliosis curve with the primary apex at T4, and a left convex lumbar scoliosis with the apex at the T12-L1 thoracolumbar junction and a hyperlordotic lumbar curve. Breast exam was limited but there appeared to be a complete absence of the pectoralis major on the right in keeping with Poland's syndrome. Both of her hands were of normal appearance. She wore a special type of bra with right-sided padding. On further questioning she reported being unsatisfied by her appearance, in that the prior reconstruction surgery had been only partially successful, leaving her with residually less right breast tissue and nipple deformity. The thoracolumbar scoliosis partially corrected on Adam's forward flexion with improvement in all planes. She had +15 bilateral genu-recurvum bilaterally. There was marked winging of the right scapula. Right shoulder abduction was limited to 140/170 degrees with equal external rotation. Cervical spine range of motion was full, pain free, and Spurling's test was negative.

Strength testing of the biceps, triceps, rotator cuff and grip were all equal bilaterally. There was +4/5 weakness of the right pectorals, lower trapezius and rhomboids. The left leg was 1.5 cm long, which was managed with a custom right shoe insole. The upper limbs were neurovascularly intact. Hip and knee range of motion was full and essentially pain free with some mild left femoroacetabular impingement signs, right hip painless clicking and bilateral patellofemoral joint crepitus on knee extension. Her Beighton score was 9/9.

Discussion

There is no genetic testing available for Poland's syndrome. It is not clear how Poland's syndrome is caused and the theory goes that it's abnormalities occur during embryological development, possibly as a result of vascular malformation or occlusion.

Adding to the complexity of presentation is the involvement of many of the adjacent muscles including the serratus, latissimus dorsi, and the external oblique. [Jenkins et al 2004]

This abnormal muscle development may relate to the spinal scoliosis described in the literature. However, it is not a very well known trait, as it would appear that most all of the clinical case reports related to Poland's syndrome do not describe scoliosis as a feature. Spinal deformities such as hemi-vertebra may also be present which predispose to structural scoliosis.

[Cobben et al 1989]

Conclusion

The patient was reviewed by a Genetics consultant and was also referred for Schroth-type physiotherapy to manage her scoliosis. She was also advised about activity modifications and job-related repetitive lifting and carrying of bulky and heavy camera lenses and other photography equipment. As there was no indication for spinal surgery, no spinal imaging was recommended. However, if there is any further scoliosis progression or the patient doesn't respond to the above recommended management strategies then full spine x-ray imaging will be considered.

The absence of breast development on the affected side was noted and the Poland's syndrome was confirmed. She seemed to be relieved that there was now an explanation of her appearance which had greatly impacted her feeling of confidence and self-worth over her lifetime. She was referred onward to a combined plastic/breast surgical service for further investigations and appropriate surgery.

More recent studies have reported that scoliosis is an important yet overlooked associated trait of Poland's syndrome. [Nurettin et al 2015]

In this case, spinal scoliosis was the primary reason for presentation. Patients may experience appearance related social stigma, bullying, psychological distress and anxiety as adolescents and as adults. Spinal, postural, and chest anomalies may also have cultural and social impact which may prompt patients to hide their appearance or be non-forthcoming in their history. This creates diagnostic challenges warranting increased clinician awareness of the

link between spinal scoliosis and Poland's syndrome.

Achieving an accurate diagnosis impacts management, alleviates patient worry, uncertainties, biopsychosocial stressors, and may also improve patient compliance with management.

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