

# Poland Syndrome: A curious anomaly

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## ABSTRACT

Poland syndrome also referred to as Poland Anomaly or sequence is a rare congenital anomaly characterized by absence or under development of the pectoralis major and sometimes minor muscle with or without limb asymmetry and a combination of other physical anomalies. No cases have been published from Ghana. A sixteen-year-old girl presented with gross asymmetry of the breasts. Further examination revealed asymmetry of the upper limbs with shortened digits on the left. Anomaly only became noticeable during puberty. Though defects in Poland syndrome are present at birth, some individuals with relatively minor defects only present during puberty when the asymmetry becomes more glaring.

## INTRODUCTION

Congenital malformations are diverse in form and presentation. Some are obvious and hence easily recognized at birth thus bringing patients to early medical attention. Others such as Poland syndrome apart from being rare and thus not considered by most doctors, usually become obvious only during puberty.

## CASE DESCRIPTION

A sixteen-year-old girl had 'normal' physical development throughout childhood but developed gross asymmetry of her breasts during puberty. Her right breast was about ten times the size of the left which seemed to have virtually no breast tissue [Figure 1]. Further examination by a dermatologist revealed shortening of the fingers on the left [Figure 2]. Her radius, ulna and all other bones appeared symmetrical. She had no other physical anomaly besides the asymmetry of her breasts and fingers.

Chest CT scan revealed absence of the left pectoralis major muscle with hypoplastic pectoralis minor muscle and left breast tissue. The right pectoralis major and minor muscles as well as both lungs appeared normal.



Figure 1: Gross asymmetry of breasts with the absence of the left axillary fold pointed



Figure 3: Shortened digits of the left hand



Figure 2: Chest CT showing absent left pectoralis major and left hypoplastic pectoralis minor muscle. (Thin and long arrow- normal right pectoralis major muscle, thin short arrow- normal right pectoralis minor muscle, thick and short arrow -hypoplastic left pectoralis minor muscle with absent pectoralis major).



Reconstructive surgery with implantation of an inflatable silicon balloon was recommended. Expansion of the skin over the left breast was done in preparation for silicone implant insertion. Patient in the meantime uses padded brassiere to conceal the defect because she cannot afford the implant.

## DISCUSSION

Embryonal cell migration and differentiation should result in a symmetrical body. Some individuals for unknown reasons develop absence of the pectoralis major muscle or underdevelopment of the muscle with various limb abnormalities and other physical defects. Disruption of blood flow through the subclavian artery during the sixth embryonal week and abnormalities of the apical ectodermal ridge have been postulated as possible causes.<sup>1</sup> The hallmark of Poland syndrome is the unilateral absence or underdevelopment of the sternal portion of the pectoralis major muscle with or without pectoralis minor involvement. It occurs sporadically though there are few instances where individuals of the same family are affected.<sup>2,3</sup>

Similar disorders involving both sides of the body have been described but there is no general consensus as to whether such cases can be classified as Poland syndrome or not.<sup>4,5</sup>

Limb anomalies such as brachydactyly, shortening of the radius and ulnar, cutaneous syndactyly especially of the middle and index finger and rhizomelic micromelia have also been described in association with the syndrome. In a few instances, Sprengel deformity, vestigial fingers, simian crease, dextrocardia, duplex renal system and absence of the latissimus dorsi and serratus anterior muscle have been reported.<sup>6</sup> There are also reports suggesting an increased risk of tumors most especially lympho-proliferative disorders and leukaemia in affected individuals.<sup>2</sup> One case report also found multiple ipsilateral cavernous sinus malformations in association with the classic features of Poland syndrome.<sup>4</sup> Our patient only had absence of the left pectoralis major and hypoplastic breast tissue with associated shortening of the fingers on the same side. Poland syndrome is three times commoner in males and usually affects the right side of the body for reasons that are still unclear.<sup>6</sup> Our patient was female and had the left side of her body affected. There are also other reports of the syndrome involving the left side.<sup>7,8</sup>

Though Poland syndrome is congenital, only severe cases tend to be noticed at birth. Most patients with a mild form of the anomaly like our patient may only present during

puberty.<sup>9</sup> This is attributable to the pubertal growth spurt with its attendant increase in muscle bulk and breast development especially in females. Scanty or abnormally positioned axillary hair occurs in some patients but this was absent in our patient.<sup>1</sup>

Individuals with Poland syndrome may have some weakness of the affected limb as the pectoralis major muscle is responsible for swinging of the arm across the chest. Physiotherapy may help to improve muscle strength and range of motion. Our patient however had no limitation of movement in her left arm and had normal muscle strength. This is not unusual as individuals with this defect have been involved in sports like boxing and golfing. This may be due to compensatory hypertrophy of the pectoralis minor muscle and any remaining portion of the pectoralis major muscle<sup>10</sup> Treatment of the chest wall deformities in this syndrome is mainly for cosmetic reasons. Factors considered include the sex, age, degree of severity- which may increase risk of heart or lung damage- and its psychosocial impact on the patient. Adolescent females especially find the breast asymmetry distressing. Breast remodelling is the commonest procedure performed. It usually involves computer aided design and implantation of a silicone implant with or without lipomodeling. Depending on the nature of malformation, other specialized procedures such as laparoscopic reconstruction using the omentum flap technique, reconstruction of the chest wall and microsurgical or pedicled transfer of the rectus abdominis muscle may be necessary.<sup>3,10</sup>

The timing of surgery is very important as a hasty surgery can result in unnecessary revision surgeries. It is generally recommended to correct the chest wall deformities after puberty in order to get an accurate measurement of the disparity. In the case of females, gradual skin expansion on the affected side can be necessary prior to definitive surgery in order to accommodate the implant. The unaffected breast may also require remodelling to ensure symmetry.<sup>11</sup> Other anomalies such as syndactyly and vestigial digits are usually easily corrected surgically during childhood.



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**CONCLUSION**

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Though Poland syndrome is a congenital anomaly, some individuals with milder anomalies are only diagnosed during puberty. Early detection is possible through careful physical examination at birth to avoid psychotrauma associated with late diagnosis. The chest wall asymmetry though distressing to most patients does not limit physical activity in most affected persons. Most cases involve the right side but the left can also be involved. Reconstructive surgery of the chest wall deformities though possible is not readily available or affordable especially in third world countries. The syndrome may be under reported due to its varying degrees of severity and a general lack of awareness about the condition.

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