

Unilateral Gynecomastia in an Adolescent with Poland Syndrome: A Clue for Diagnosis

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Children affected with Poland syndrome are born with missing or underdeveloped muscles (typically pectoralis major) on one side of the body. Breast abnormalities such as unilateral hypoplasia or agenesis of the breast and nipple may also occur. Other muscles on the affected side, including other muscles in the chest wall, shoulder, arm, and hand, may be missing or underdeveloped [1]. Ribs may be noticeable due to the loss of subcutaneous fat. Sparse or absent axillary and pectoral hairs are a common manifestation of this syndrome.

The mechanism underlying these defects is not known [1]. Familial recurrence of the syndrome with higher prevalence in males suggests a genetic, hereditary basis. However, the genetic basis of the Poland syndrome has not been determined [1]. Pubertal idiopathic gynecomastia is observed in 50–60% of adolescents and is usually bilateral [2]. Rarely, pathological conditions such as Klinefelter syndrome, hypogonadism, androgen resistance, and medications may also result in bilateral gynecomastia in less than

5% of adolescents [2]. In this report, we present a combination of unilateral pubertal gynecomastia with Poland syndrome that has not been described previously in adolescence.

PATIENT DESCRIPTION

A 12-year-old boy was referred to our clinic after breast enlargement for a year. The mother noted an axillary odor only on his right axilla. He complained of weakness in his left arm, especially during his recent workout in a fitness club. His past medical history was otherwise uneventful. His parents and a 14.5-year-old sister are healthy. On physical examination, right gynecomastia was noted. The right shoulder and arm muscles were well developed [Figure 1A]. The left chest was flat, and no breast tissue was palpated [Figure 1A]. His height was 157.8 cm and his height was standard deviation score (SDS) +1 (87 percentile). He weighed 41.4 kg with a weight SDS 0.15 and body mass index (BMI) SDS of -0.40 (BMI 16.6). No axillary hairs were noted on either side. Testicular volume was 15 ml. At the age of 12 3/12 years, bone age was compatible with 13 years.

In laboratory investigations, gonadotropins, testosterone, prolactin, IGF-1 concentrations, and thyroid function tests were all within the normal range: luteinizing hormone 1.2 IU/L, (normal range 0.2–4.9 IU/L), follicle-stimulating hormone 1.5 IU/L (normal range 0.3–3.5

IU/L), testosterone 6.3 nmol/L (normal range 3.4–11.1 nmol/L), prolactin 51 mIU/L (normal range 43–375 mIU/L), insulin-like growth factor-1 108 ng/ml (normal range 143–506 ng/ml), normal thyroid stimulating hormone 2.1 mIU/L (normal range 0.5–4.8 mIU/L), FT4 13.2 pmol/L (normal range 10.3–19.7 pmol/L).

The right breast ultrasound was consistent with benign gynecomastia tissue. Abdominal ultrasound excluded kidney abnormalities. These features were consistent with Poland syndrome associated with pubertal gynecomastia. In the absence of internal organ involvement, we assured him of the benign nature of gynecomastia as well as of the Poland syndrome. On follow-up, the gynecomastia peaked at the age of 13 2/12 years (8 × 10 cm) and became smaller (7 × 6 cm) by the age of 14 8/12 years [Figure 1B]. He continued working out at a fitness club, which positively affected his muscle mass over the right chest and torso. His left chest, shoulder, arm, and trapezius muscles were underdeveloped. Mild scoliosis and prominent ribs due to reduced subcutaneous fat over the left chest were also noted [Figure 1C]. Testicular volume was 20 ml with a Tanner stage of 3 axillary hair (much less on the left side) and Tanner 4 pubic hair. At the age of 14.5 years, he was 178.7 cm tall (percentile 96) and weighed 56 kg (BMI 17.6).

Figure 1. Right pubertal gynecomastia at the age of age 12 years

[A] Underdeveloped chest, shoulder, and arm muscles of the contralateral side



[B] Decreased size of right gynecomastia with prominent ribs, loss of subcutaneous fat of the left chest, and sparse right axillary hair at 14 8/12 years of age



[C] Scoliosis, underdeveloped muscles of left torso, and shoulder



COMMENT

This boy presented with a unique combination of Poland syndrome with unilateral pubertal gynecomastia. He was referred due to enlarged unilateral breast tissue and not due to the manifestations of the Poland syndrome. The examination clearly showed underdeveloped muscles on the left side of the chest characteristic of Poland syndrome and torso [1].

Anhidrosis due to the absence of sweat glands may also occur. Rarely, severely affected individuals may have abnormalities of internal organs such as lung or kidney [1]. In our case, ultrasonographic evaluation excluded kidney abnormalities. In most cases of Poland syndrome, the abnormalities do not cause health problems or affect movement. Therefore, multiple radiological and laboratory investigations should be avoided [1]. Co-existence of unilateral gynecomastia and Poland syndrome has been previously reported in two adult patients aged 29 and 26 years of age [3,4]. To the best of our knowledge, this is the first case of unilateral gynecomastia in an adolescent with Poland syndrome. Many of the manifestations such as lack of odor, sparse or deficiency of axillary hair lack of hairs over the pectoral region, anhidrosis due to the absence of sweat glands, underdeveloped muscles, and breast may suggest androgen resistance due to underlying tissue damage in tissues expressing androgen receptors [5].

CONCLUSIONS

To the best of our knowledge, this case is the first report of pubertal gynecomastia associated with Poland syndrome. In

most cases, pubertal gynecomastia is bilateral. Since mild cases of Poland syndrome may remain undiagnosed, a high index of suspicion is required for correct diagnosis in adolescence, especially in cases with unilateral gynecomastia. Unilateral gynecomastia in a boy may provide a clue for the diagnosis of Poland syndrome.

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That is what learning is. You suddenly understand something you’ve understood all your life, but in a new way.

Doris Lessing (1919–2013), novelist, poet, playwright, Nobel laureate

If any man seeks for greatness, let him forget greatness and ask for truth, and he will find both.

Horace Mann (1796–1859), educational reformer