Concurrent Poland anomaly and idiopathic hirsutism

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ABSTRACT

Poland syndrome is characterized by congenital and unilateral absence of the pectoralis major muscle and ipsilateral upper limb anomalies. Identified patients also may include other disorders. We report a 15-year-old Caucasian woman with a unilateral hypoplasia of the breast and nipple, ipsilateral chest wall depression deformity, pectoralis major muscle agenesis, and severe hirsutism (Ferriman-Gallwey score: 21) without extremity anomaly. She had regular menses, and no hormonal abnormality and family history of hirsutism. Therefore, she was considered as a case of idiopathic hirsutism. This is the first case report of hirsutism in a patient with Poland syndrome.

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Case Report. A 15-year-old female was admitted to our clinic with complaints of hirsutism and unilateral breast and nipple hypoplasia with ipsilateral chest wall deformity. From her history, it was learned that her birth had been normal and that the chest wall deformity was congenital. She had had excessive body hair since birth. Menstruation had begun at the age of 13, and continued regularly since then. At puberty, the patient became aware of the retarded development of her left breast and resultant asymmetry. On physical examination, an excessive amount of male-type terminal hair was present on the face, back, hips, arms, legs and around the umbilicus (Ferriman-Gallwey score: 21). Depression deformity in the left chest wall, total absence of the pectoralis major muscle and ipsilateral hypoplasia of the breast and nipple were observed (Figures 1-4). No upper extremity deformities or acne or evidence of increased masculinization were determined. Examination of her family members (2 brothers and 3 sisters) showed no evidence of structural deformity or excessive hair. Routine biochemical and hematological parameters were found within normal limits.

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limits in laboratory examination. No abnormality was observed in hormonal parameters, namely, Luteinizing hormone, follicle stimulating hormone, prolactin, 17α-hydroxy-progesterone (17αOH-PRG), total and free testosterone, estradiol, dehydroepiandrostrosterone-sulphate, sex hormone binding globulin, ACTH - adrenocorticotrophic hormone. *value for women at follicular phase of menstrual cycle **value for women, ***value for women at premenopase state.

**Discussion.** The classic definition of Poland syndrome includes hypoplasia or aplasia of the pectoralis major muscle and anomalies of the upper extremities. Our patient had aplasia of the pectoralis major muscle, but no upper extremity anomaly. Although anomaly of the upper extremities is a
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component of Poland syndrome, it is not a constant finding for the diagnosis as reported by Shamberger et al. The chest wall anomalies reported in Poland syndrome are scoliosis, axillary bands, aplasia or hypoplasia of the breast or nipple, hypoplastic scapula or ribs, and hypoplasia or anomalies of other shoulder girdle muscles. Shamberger et al reported depression deformity in 16 patients. In our patient, we observed advanced depression deformity in the chest wall. Particularly in patients affected on the left side, dextrocardia and certain heart anomalies had been reported to accompany Poland syndrome. Thus, we performed echocardiography to our patient, but found no anomaly other than minimal mitral and tricuspid regurgitation.

Although it has been reported to occur in 2 or more family members, Poland syndrome is generally of sporadic occurrence. We examined the other members of our patient’s family and found that they were normal.

To our knowledge, in the more than 400 cases of Poland syndrome that appear in the literature, the concurrence of hirsutism has not been reported. This may be because Poland syndrome patients are usually male (male/female ratio: 3/1), and because many of the female patients have been reported during childhood and not at the age at which hirsutism most frequently occurs. A “subclavian artery supply disruption sequence” hypothesis has been suggested for the pathogenesis of Poland syndrome. It proposes that interference with the early embryonic blood supply by the subclavian and vertebral arteries or their branches, or both, could give rise to the Poland phenotype and to the limb defects. According to this theory, the hirsutism in our patient may be coincidental. Because hirsutism generally begins at puberty, whereas in our patient it is congenital, it is possible that this condition is related to other embryonic pathological factors.

In conclusion, we suggest that further description of various clinical features in Poland syndrome provides better understanding of the pathogenesis of this syndrome.

References