

Case report

ASSOCIATION OF POLAND SYNDROME AND
HYPERTRICHOSIS IN PUBERTAL GIRL - CASE REPORT

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Abstract

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Poland syndrome (PS) is a rare congenital malformation, most commonly characterized by absence of chest wall muscles on one side of the body. It may be accompanied with other deformities of the extremities. We present the case of a 10-year-old girl with Poland syndrome and hypertrichosis of the back of the trunk and extremities. The clinical examination did not reveal the etiology of the syndrome, such as familial predisposition or some event that led to interrupted blood flow during the early embryonic growth. The pregnancy was conceived with in vitro fertilization (IVF); triplets were born and our patient is one of these three girls. The hypertrichosis appeared at 8 years of life, without evidence of previous familial occurrence, medications or hormonal imbalance. Other malformations that were found were: a mild form of kyphoscoliosis and mitral valve prolapse. The child was evaluated using a multidisciplinary approach, with further follow-up planned with surgical correction of the chest wall and breast augmentation.

Приказ на случај

ПОВРЗАНОСТ НА POLAND-ОВ СИНДРОМ И ХИПЕРТИРЕОЗА КАЈ
ЖЕНСКО ДЕТЕ ВО ПУБЕРТЕТ – ПРИКАЗ НА СЛУЧАЈ

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Извадок

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Клучни зборови: Поландов синдром, хипертиреоза, пубертет, женско дете

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Печатарски права: ©2021 Марина Крстевска-Константинова; Констандина Кузевска-Манева; Христијан Несторов; Даниела Георгиева. Оваа статија е со отворен пристап дистрибуирана под условите на нелокализирана лиценца, која овозможува неограничена употреба, дистрибуција и репродукција на било кој медиум, доколку се цитираат оригиналниот(ите) автор(и) и изворот.

Конкурентски интереси: Авторот изјавува дека нема конкурентски интереси.

Поландовиот синдром е ретка конгенитална малформација, која најчесто се карактеризира со отсуство на мускули на градниот кош на една страна на телото. Може да биде придружена и со други деформитети на екстремитетите. Нашиот приказ на случај е женско дете старо 10 години со Поландов синдром, здружен со појава на хипертрихоза на грбот и екстремитетите. Направените иследувања кај детето не ја открија етиологијата на синдромот, како генетска предиспозиција или настан за време на бременоста на мајката кој довел до намалена циркулација за време на раниот ембрионален раст на плодот. Кај мајката е спроведена in vitro фертилизација (IVF) и ова е едно од трите женски деца кои се родени. Хипертрихозата се појавила на 8-годишна возраст, без податок за претходна фамилијарна појава, медикаментозна терапија или хормонален дисбаланс. Додатни малформации кои се најдени беа: лесна форма на кифосколиоза и пролапс на митрална валвула. Детето е водено мултидисциплинарно со план за понатамошна хируршка корекција на градниот кош и неразвиената града.

Introduction

Poland syndrome (PS) is a rare congenital malformation, most commonly characterized by absence of chest wall muscles on one side of the body. The pectoralis major muscle is absent or underdeveloped, and may be associated with thoracic anomalies, upper limb anomalies, anomalies of fingers (syndactyly and brachydactyly), ipsilateral underdeveloped breast and subcutaneous fat tissue, and rarely other organ involvement¹.

Hypertrichosis, which may be congenital or acquired, is characterized by excessive hair growth anywhere across the body².

Case presentation

A 10-year-old girl presented at the Pediatric Endocrinology Department due to unilateral breast development and hypertrichosis. The child was born from in vitro fertilization (IVF); the pregnancy was uneventful

and resulted in triplets, all of them were female. The clinical examination did not reveal the etiology of the syndrome, such as familial patterns or some event leading to interrupted blood flow during the early embryonic growth. The birth parameters were as follow: birth weight was 2,250 gr, birth length was 48 cm. The other two babies, also girls had a normal appearance.

At the time of admission, the puberty has started and left breast was M33, while the right breast was absent, although a small nipple was present. Chest wall deformity of the right thoracic cage was noted as well as a mild kyphoscoliosis (Figure 1). The deformity had been noticed at birth, although further clinical examinations or hospital visits were not performed. The hair was pigmented, long and soft. Teeth and nails were normal without gingival hypertrophy. There was excessive hair growth on the extremities and the back of the trunk (Figure 2).

Figure 1. Chest wall deformity in our patient



Figure 2. Hypertrichosis in the arm



Hypertrichosis started at the age of 8 years and it became more noticeable, leading to a conclusion it was acquired.

The height was 135 cm (50% on growth chart) and the weight was 29 kg (25%). Pubertal stage was M33 (on the left side), P2A13 and the menstrual cycle was still absent (Pubertal

stage by Tanner and Whitehouse).

From the laboratory findings, sex chromatin was 5%. Routine blood investigations were normal. Hormonal study was within normal limits for

her age. Chest X-ray showed absent rib deformities, and normal lung transparency. Medistinal structures were centrally placed (Figure 3).



Figure 3. Chest X-ray

Echocardiography revealed mitral valve prolapse without mitral regurgitation.

Evaluation was made by plastic and orthopedic surgeons revealing a mild kyphoscoliosis and physiotherapy was started. Clinical follow-up was suggested until reconstruction and breast augmentation can be performed in late adolescence or adulthood.

Discussion

This is the second case of Poland syndrome in our practice (although 4 more were seen by colleagues). The first was a 3-year-old girl who also had unilateral premature thelarche. Unfortunately, the patient was lost from follow-up, due to migration abroad.

The incidence of PS has been estimated from 1:30 000 to 1:80 000 live births⁴ and it is more frequent in males, more commonly seen on the right side of the thorax, as in our case. Familial cases have been described, with different inheritance

patterns, suggesting a genetic background including gene mutations on different chromosomes⁵. Yet, the most common theory is impairment of embryonic blood supply induced by hypoplasia of the ipsilateral subclavian artery, due to prenatal exposure to teratogens that interfere with vascular development⁶. Other theories include disruption of the lateral mesodermal plate after fertilization⁷. Also, there have been alternative diagnoses proposed in patients with congenital pectoral muscle deficiency. In a large study by Baas et al., 627 patients were described, using 136 articles, where ten different definitions of PS were utilized. In 58% of the cases, an upper extremity deformity was found, 43% had an associated deformity, 57% had a pectoral malformation, hand malformation and another deformity that matched an alternative syndrome. Twenty-nine percentage had classical PS. The authors concluded that differentiating PS from other syndromes had serious consequences for the patients and their families

in terms of inheritance and possible related anomalies^{8,9}.

The clinical presentation of PS may be subtle, so the diagnosis can be made by radiological studies using standard mammograms in adulthood^{10,11}. Identifying PS is important for better care and avoiding potential clinical mismanagement and possible litigation in cases of incidental trauma¹².

A study of 72 patients with PS in Italy revealed the need of specific national diagnostic and therapeutic guidelines, which will guarantee patients complete and appropriate health services that will improve the quality of life of people living with rare diseases¹³.

The treatment of PS is surgical, meaning surgical reconstruction of the thoracic wall anomaly and hand anomaly if present. Surgical treatment options include reconstruction of the chest wall with myocutaneous flap coverage and breast augmentation procedure as needed. Preoperative angiography and chest computed tomography or MRI should be performed to evaluate the blood supply of potential flaps and the extent of the anomaly^{14,15}. For children with more severe thoracic deformities and if the patient is more than 3 years old, Nuss procedure should be performed as early as possible, as it is done without high rate of complications^{16,17}. Other methods include muscle transfer with or without implants. Young patients with PS in their adolescence may be psychologically concerned regarding their body image and early surgical procedures may have positive benefits on their quality of life.

Hypertrichosis, which was also present in our patient, did not point to any specific syndrome associated with this condition, medication, hormonal etiology or inheritance. Low birth

weight may have been a contributing factor. Some authors have described premature adrenarcha in babies born small for gestational age 18, although the girls pubic and axillary hair was absent. The association between PS and hypertrichosis was not found.

Conclusion

PS is a rare syndrome which needs early recognition, correct diagnosis and multidisciplinary approach. Surgical intervention should be planned on basis of the severity of disease and/or accompanying malformations. The finding of hypertrichosis in our patient is also interesting, but seems to be sporadic and not associated with the syndrome.

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