Case Report

Poland Syndrome with a Rare Association

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Abstract

Poland syndrome is a rare congenital anomaly typically characterized by unilateral chest wall hypoplasia and ipsilateral hand abnormalities. Occasionally functionally debilitating, the disorder is mostly benign in nature with the severe deformities requiring surgical correction. A need to watch for possible malignant associations is also warranted. We present a case of a young female with the quintessential features of Poland syndrome with a relatively rare association of ipsilateral renal hypoplasia. We review the literature and discuss the possible renal complications that may arise and their management.

Introduction

Poland syndrome, first described by Sir Alfred Poland in 1941, is a rare entity characterized by a variable incidence of hypoplasia of some of the elements of the thoracic cage and the corresponding upper extremity with absent/underdeveloped breast, pectoralis muscle, axillary web, syndactyly and brachydactyly. It is sporadic in nature with a reported incidence of 1 in 7,000 to 1 in 100,000 live births with a male preponderance (M:F ratio of 3:1). There are very few cases of Poland syndrome with renal anomaly reported and the structural abnormality (ipsilateral hypoplasia or aplasia) may be an integral part of this syndrome as described in our patient.

Case Report

We report a case of a 24 year old female who presented with under-developed left breast, inability to draw her left hand across the chest, asymmetric upper limbs from birth. Examination revealed hypoplastic/aplastic left breast with absence of the nipple-areola complex, complex, lack of subcutaneous fat and axillary hair, absent anterior axillary fold and the left pectoralis major muscle were absent with flattened chest on the left. She also had short left upper arm, forearm and fingers (with syndactyly and absent middle phalanges). Her BP was 140/90 mm of Hg (Stage 1-JNC VII) right upper limb with no significant variations in the other limbs. Other vitals and systemic examination (excluding the deformities) were unremarkable. A chest radiograph was taken which showed absent mammary and pectoralis major shadows with relative lucency of the left hemithorax and dextroposition of the heart (Figure 1). Dextrocardia was ruled out from the normal findings on ECG (Figure 2) and 2d-Echo. Ultrasonography revealed a hypoplastic kidney on the left measuring 2.3 x 2 x 1 (cm) and the right kidney measuring 14 x 7 x 6.5 (cm). CT thorax, Doppler Ultrasonography, MR and CT angiography were not done owing to the financial constraints of the patient. Urinalysis showed proteinuria (2+) and no active sediment. Serum Creatinine was 0.9 mg/dl. Her 24 hr urinary protein was 1200 mg and her estimated GFR (MDRD formula) was 90 ml/min. She was started on ramipril 2.5 mg/day. Other lab parameters were normal. Final diagnosis of Poland syndrome was made. She was advised reconstruction surgery and regular follow up to monitor her kidney function but was lost to follow up.

Discussion

Poland syndrome is a rare congenital anomaly typically characterized by unilateral chest wall hypoplasia and ipsilateral hand abnormalities.1 The absence of pectoralis major on one side constitutes the hallmark although underdevelopment or absence of the breast and nipple-areola complex, abnormalities of the anterior ribs, clavicle, and scapula, axillary bands or webs and lung herniation can also be seen. The limb abnormalities may include brachydactyly, syndactyly, absent phalanges or digits, and hypoplasia of the forearm, wrist, or hand. Other features that may accompany the entity are hemivertebrae, renal anomalies (hypoplasia in our case), dextrocardia (dextroposition in our case), Sprengel deformity, clubfoot and submucous cleft palate.2 About 10% of patients with syndactyly demonstrate features of this syndrome.3 More than 75% of the aforementioned defects are present on the right side (unlike our case). It has been postulated that various factors like congenital vascular maldevelopment (may include an inheritable component), intrauterine insults and drugs could ultimately result in hypoxia to one side of the fetus as the limb bud develops adjacent to the chest wall leading to the development of the syndrome. Impedance plethysmography (rheography) shows marked decrease of the velocity of the systolic increase in the arterial volume in the affected arms, and with significant difference between the two arms compared to control group pointing toward the hypoplasia of the ipsilateral subclavian artery as the origin of the malformation. Associations have been found with leukemia, lymphoma, carcinoma of the hypoplastic breast, carcinoma lung, neuroblastoma, arterial septal defects and Wilms’s tumour demanding heightened oncological awareness in these settings.4

Few renal anomalies in conjunction with Poland syndrome have been reported some of these cases including undescended testes and pylonephritis. The mechanism postulated attributes the renal anomalies to the maldevelopment of the metanephros affecting the ureteral bud and the mesonephric duct early on in the development at about 4th or 5th week of gestation.5 A familial form has also been described with megacalycosis of the ipsilateral kidney.6 Aplasia of pectoralis major with renal anomalies, (unilateral renal agenesis or duplication of the renal collecting system, etc) is termed acro-pectoral-renal field defect, and in some cases may cause renal hypertension (as shown in our case).7 Owing to the asymptomatic nature of these patients, routine renal investigation is usually not performed and hence the under reporting of the cases. Renal ultrasonography, 99mTc-DTPA radionuclide scan, and/or any other appropriate imaging study help improve the detection rate. A mild degree of proteinuria with modest BP elevations can be seen in some prompting therapy with ACE inhibitors/ARBs but most patients have good prognosis unless complicated by other comorbid factors.

Most patients do not require surgical correction but in amastia and hypoplastic breasts repair with a multi-layered approach, may be attempted (two stages in children and in a single stage...
Surgical treatment often improves functionality and cosmetic appearance although the inherent skeletal anomalies may limit the degree of the same.

Poland syndrome (along with the constellation of associations with various organ systems) requires clinicians to have adequate knowledge about the entity, its complications and the management aspects. Our case highlights the need to probe for renal anomalies apart from the other abnormalities as early diagnosis, prevention and treatment of renal disorders in these patients may promise a better outcome.

References