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Poland syndrome with retinochoroidal coloboma: A rare association

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Abstract
A 58 year aged man with the Poland syndrome is presented with left pectoralis major muscle, hypoplasia of the left arm, symbrachydactyly, and ipsilateral coloboma of the optic disc.

Keywords: Poland syndrome, symbrachydactyly, hypoplasia, brachydactyly

Introduction

The Poland's Anomaly was first described in 1841 by Sir Alfred Poland as a syndrome [1]. It consists of anatomic anomalies that include the absence of the sternocostal head of the pectoralis major muscle with other varied manifestations that include hypoplasia or absence of the pectoralis minor muscle as well as digital anomalies such as syndactyly [2]. Other anomalies include the absence or hypoplasia of the ipsilateral breast, excavatum deformities, and rib aplasia. Poland syndrome affects about 1 in 36,000 to 50,000 newborns, with higher frequency among males (ratio: 2:1- 3:1). In 75% of the cases, it is located on the right hemithorax in the unilateral form [3, 4].

Upper limb anomaly includes underdeveloped hand with abnormally short fingers (brachydactyly), small and underdeveloped (vestigial) fingers and some fingers are fused together (syndactyly). The combination of hand anomalies is called symbrachydactyly. Affected individuals may have variable associated features, such as underdevelopment or absence of one nipple including the areola and/or patchy absence of hair in the axilla [4]. In females, there may be underdevelopment or aplasia of one breast and underlying (subcutaneous) tissues [5]. In some cases, associated skeletal abnormalities may also be present, such as underdevelopment or absence of upper ribs; elevation of the shoulder blade (Sprengel deformity); and/or shortening of the arm, with underdevelopment of the forearm bones (i.e., ulna and radius) [6, 7]. Thoracic anomaly includes ipsilateral breast and nipple hypoplasia and/or aplasia, deficiency of subcutaneous fat and axillary hair, absence of sternal head of pectoralis major muscle, hypoplasia of rib cage second to fifth rib.

Retinochoroidal coloboma results from the failure of the optic fissure to close during the fifth to seventh week of gestation, when the embryo corresponds to 7 mm to 14 mm stage of development. The timing of the defect explains the multiple association with systemic malformations. In this article we report a combination of Poland Syndrome with ipsilateral Retinochoroidal coloboma.

Case Report

We examined a 58 years old man with hypoplasia of left side pectoralis major muscle, along with abnormally short fingers in left side (brachydactyly), fused phalanges (syndactyly) [Figure 1]. Patient has history of diabetes mellitus since last 5 years and hypertension since last 2 years on treatment for both the conditions. The patient was picked up as a case of Poland Syndrome when he came for routine diabetic retinopathy screening. On examination BCVA RE was 20/30 and BCVA LE was 20/20. Anterior segment examination of right eye showed early nuclear sclerosis changes. Anterior segment of left eye was essentially normal.

On fundus examination right eye showed normal fundus and there was presence of retinochoroidal coloboma on the left side in the inferior quadrant not involving the optic disc or macula. No other ocular abnormality was noted [Figure 2].

Discussion

Poland Syndrome is a rare congenital anomaly characterised by hypoplasia or aplasia of pectoralis major muscle, variably associated with same side upper limb anomalies. The etiology of Poland anomaly is unknown with an ipsilateral vascular alteration (of unknown origin) to the subclavian artery in early embryogenesis being the currently accepted theory [7, 8].

Most evidence support the idea that malformation of subclavian arteries cause reduced amount of blood delivered to the developing tissues of one side of body "subclavian artery supply disruption sequence". This happens during the sixth week of gestation. This is around the same time at which retinochoroidal coloboma develops i.e around fifth to seventh week of gestation [9, 10].

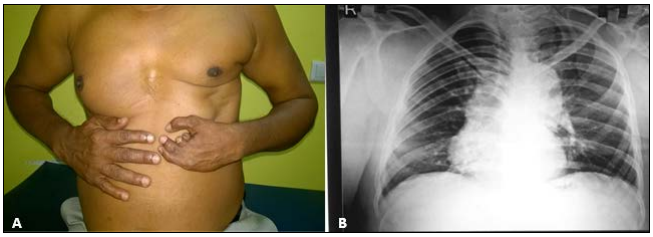

Fig 1: A. Image showing left sided thoracic and upper limb anomally. B. Image showing radiological finding of rib abnormality

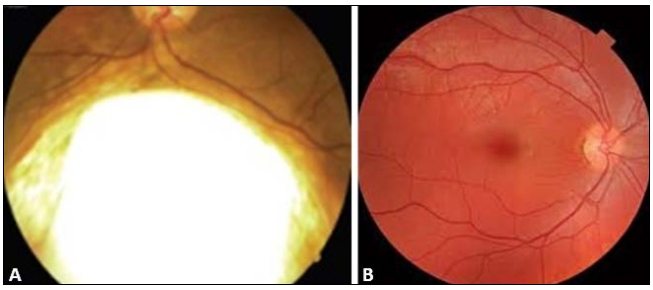

Fig 2: A 58 year aged man with the Poland syndrome is described, presented with left pectoralis major muscle, hypoplasia of the left arm, symbrachydactyly, and ipsilateral coloboma of the optic disc

Conclusion

A mechanical external factor is not likely to be the causative factor in our case. Multiple abnormality of musculoscleletal system as a result of vascular anomaly and failure of choroidal fissure closure which occur around the same time in embryogenesis may point towards common etiology. In our case report the possibility of coincidental presentation of Poland Syndrome and retinochoroidal coloboma is worth considering, and we look forward for more case reports which may establish a common etiology of both presentations.

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