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POLAND ANOMALY, A SPORADIC SYNDROME: A REVIEW.

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Poland Anomaly, Congenital sporadic syndrome, Subclavian Artery Supply Disruption Sequence

Abstract
Poland Anomaly, one of the sporadic congenital disease involves the congenital unilateral absence or underdevelopment of Pectoralis major muscle with ipsilateral cutaneous syndactyly. It is thought to be due to Subclavian Artery Supply Disruption Sequence. There are evidences which suggest it due to genetic factors or teratogenic. The child has structural abnormalities involving absence or underdevelopment of muscles, or bones like ribs. This is also associated with malignancies, dextrocardia, atrial septal defects, renal impairment, ophthalmic abnormalities. These are diagnosed by CT, MRI and X- Ray along with thorough clinical examination. The chest reconstruction therapy is the treatment available for the Poland Anomaly.

Introduction

Poland Anomaly syndrome (Poland Syndrome, Poland Syndactyly and Poland Sequence) is one of the rarest birth defect characterized by congenital unilateral absence or underdevelopment of pectoralis major with ipsilateral cutaneous syndactyly [1]. In some cases additional muscles in the chest wall, side, shoulder and the sternum are missing or underdeveloped. It is associated with leukemia, carcinoma of hypoplastic breast, dextrocardia etc. in some cases [2]. According to National Genome Research Institute, Poland Syndrome, which occurs once in 1 in 10,000 to 1 in 1,00,000 , is more common in boys than girls and right side being affected twice as often as the left [3]. For the present review, many online search engines were utilized for extraction of relevant electronic data from internet like PUBMED, Google Scholar, Medline, and Central on the Cochrane Library etc. Well used and well related key terms were included in the search. The article focusses on the Etiology, Pathogenesis, Clinical Features, Diagnosis and Treatment options available for Poland Syndrome.

Etiology and Pathogenesis:

Poland anomaly is rarely inherited and generally sporadic. Familial occurrence has been observed in some cases. Different types of transmission include: transmission from parent to offspring, in siblings born to unaffected parents and in distant family members like cousins.

The exact cause of Poland Anomaly is not discovered. Different possible causes of PS can be taken into consideration, in particular genetic factors (multifactorial or single gene defect) and teratogenic agents (maternal exposure to drugs, smoking, and others) [26]. According to many investigators, primary defect may be impaired development of certain artery or mechanical factors that may result in diminished or interrupted blood flow during sixth week of fetal development. The term Subclavian Artery Supply Disruption Sequence is suggested for this birth defect [6]. There are two theories which suggest the probable pathogenesis of the disease. One of the Theory states that there is an interruption of embryonic blood supply of subclavian arteries that lie under the clavicle. This may be due to the forward growth of the ribs reducing the flow of blood. The other theory affirms that a malformation of subclavian arteries may cause a reduced blood delivered to the developing tissues. This reduced blood flow due to any of the cause reported by the two theories results in the underdevelopment pectoralis muscles, additional muscles in the chest wall, side, shoulder and the sternum depending on where the restriction of blood flow occurs [4]. No animal support has provided the support the above theories.

Poland is also explained as a paradominant trait [8]. Another proposed concept for Poland Syndrome is the presence of an autosomal lethal gene by means of mosaicism [10]. A de novo heterozygous 126 Kbp deletion at chromosome 11q12.3 involving 5 genes, four of which namely HRASLS5, RARRES3, HRASLS2, PLA2G16, encode proteins

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that regulate cellular growth, differentiation, and apoptosis, mainly through Ras- mediated signaling pathways, were detected in identical twins with Poland Syndrome and they prove that some genetic basis exist for this syndrome [28].

Clinical Features:

Peoples born with Poland Anomaly have physical and cosmetic disabilities which are mostly unilateral. They are even bilateral [17]. All the developmental mile stones are normal in the children. This does not cause any mental abnormalities and their intelligence quotient is normal.

Absence of the some of the chest muscles mainly pectoralis major is seen [18]. Sometimes only a part of the pectoralis major may be absent or underdeveloped. The most common variation is the absence of sternocostal head of pectoralis major muscle with concomitant hypertrophy of clavicular head and absence of pectoralis minor. There may also be unilateral absence of other regional muscles such as certain large muscles of the back like latissimus dorsi or muscles of chest wall like serratus anterior. Hypoplasia or aplasia of external oblique, infraspinatus and supraspinatus muscles may be seen. The nipple and areola may be underdeveloped or absent. In female breast and the underlying tissues may be absent [13]. It may be associated with breast cancer [23].

Abnormally short and webbed fingers called syndactyly may be present unilaterally. Certain fingers may be underdeveloped or absent. Abnormally short fingers called as bradydactyly are reported. Hypoplasia or aplasia of middle phalanges, Bradymesophalangy and ectrodactyly may be visualized [9]. Bilateral palmar hyperhydrosis may be seen [15].

Axillary hair may be missing. Skin is hypoplastic with thinned subcutaneous fat layer. The upper ribcage may be underdeveloped or missing. Sometimes the shoulder blade or bones of the arm may be even absent.

Rarely spine or kidney problems may be present. Dextrocardia is also associated with Poland syndrome [11, 15]. Atrial septal defects are detected. Many vascular malformations of diseased thorax like the hypoplastic or missing vessels are being associated [19]. Herniation of lung due to absence of anterolateral ribs is reported. It is sometimes associated with lymphoreticular malignancies and some solid tumors along with lung cancer [12]. Vertebral defects, renal aplasia or hypoplasia and lower limb anomalies have been noted [7]. Renal Structural anomaly may be an intergral part of this syndrome [7]. Eye abnormalities may be associated. It is rarely associated with undescended testes, cleft palate and clubfoot [20]. A large heterogenous mass, Neuroblastoma in a right hemithorax in a patient with Poland syndrome was reported [24]. It is associated with leukemia and non Hodkin's lymphomas [25]. It is also associated with congenital hemangioma [29].

It is not necessary that all the above mentioned features are present in each and every child diagnosed with the syndrome. The severity of the disease is variable. If the disease is mild, the anomaly may not be detected till puberty in most cases especially in girls who may notice a difference in the development of breasts. The disease may be severe such that it is associated with systemic anomalies too.

Diagnosis:

Poland anomaly can be detected at birth or at adolescence depending on the severity of the disease. This can be detected by clinical evaluation and specific tests which include Magnetic Resonance Imaging (MRI) techniques, Computerized Tomography (CT) scans and X Rays. CT scans help determine the extent to which the muscles may be affected by showing cross sectional images of particular structures within the body. Mammary hypoplasia and further muscle abnormalities can be accuarately defined by sonography and MRI [16]. X Rays help to identify and characterize the abnormality in hand, forearm, ribs and shoulder blades. The evaluation of vascular status can be performed using color coded duplex sonography for peripheral arteries and contrast enhanced MR angiography for supra aortic arteries [16]. Cross sectional imaging with CT or MRI will give useful information with this syndrome requesting anterior axillary reconstruction and breast mound formation [14].

Treatment:

The most viable treatment for Poland Anomaly available is reconstructive surgery. Existing chest muscles are used to rebuild the chest. It they are not sufficient for reconstructing the chest, muscles from other parts of the body can be taken. For males with Poland Anomaly, reconstructive surgery can be done as early as 13 years of age. But for females, surgery may have to be postponed until the breast development is complete. Reconstructive surgery may be performed to correct the abnormality in different sizes of two breasts in females. Differences in nipple and areola can be treated by therapeutic tattooing. Simple deformity can be effectively repaired with latissimus dorsi muscle transfer if male and latissimus dorsi with sublatissimus mammary prosthesis if female [22]. Repair of complex deformity, in addition to latissimus transfer, selectively includes musculoskeletal chest wall realignment [22].

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In case of sternal anomalies, bilateral subperichondrial costal cartilage resection and sternal osterotomy can be performed for allowing anterior displacement and orthorotation of sternum[21]. An autologous rib transplantation and terylene patch can be used for repair of chest wall defect [27].

Conclusion

Poland Anomaly is characterized by the congenital absence or underdevelopment of the pectoral muslces, shoulder group muscles with unilateral syndactyly. The cause of the disease is unknown. There are many theories proposed for this disorder which includes subclavian Artery Disruption Syndrome, genetic and teratogenic factors. It is clinically diagnosed and MRI, CT Scans and X Rays are used for identifying the abnormalities. It is treated surgically by chest reconstruction.

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