An unusual case report showing overlapping Features of Poland and Moebius syndrome

Abstract
Moebius and Poland syndromes are rare congenital anomalies. Poland syndrome (PS) is characterized by unilateral aplasia/hypoplasia of the breast and pectoralis muscle and ipsilateral deformities in the extremities. However, Moebius-Poland syndrome is rarer than the isolated syndromic conditions. Moebius syndrome is characterized by unilateral or bilateral loss of ocular abduction, unilateral or bilateral complete or incomplete facial weakness or unilateral/ bilateral facial paralysis. However, Moebius-Poland syndrome is rarer than the isolated syndromic conditions reported very recently by Diego López de Lara et al. We hereby report an rare case of 16 year old male of Asians ethnic showing overlapping features of Poland and Moebius syndrome.

Key words: Poland syndrome, Moebius syndrome, syndactyly, Pectoral hypoplasia, facial paralysis.
**Introduction**

Poland syndrome (PS) (OMIM 173800) is an uncommon congenital anomaly characterized by unilateral aplasia/hypoplasia of the breast and pectoralis muscle and ipsilateral deformities in the extremities with a missing anterior axillary fold in its most common form [1,2]. Currently the prevailing theory in the etiology of Poland Syndrome is that, at the end of the sixth week of gestation, when the upper limb bud adjacent to the chest wall is still in a stage of development, the interruption of the embryonic blood supply causes hypoplasia of the ipsilateral subclavian artery or one of its branches. Additionally, hypoplasia of the internal thoracic artery could cause the absence of the sterno-costal portion of the pectoralis major muscle, whereas hypoplasia of the brachial artery may lead to hand abnormalities. Poland syndrome affects the right side in 67%–75% of cases [3]. Moebius syndrome is characterized by unilateral or bilateral loss of ocular abduction, unilateral or bilateral complete or incomplete facial weakness or unilateral/bilateral facial paralysis and primary or secondary congenital anomalies of the extremities. It also causes breast hypoplasia with an incidence of 1:3000 up to 1:10000 in children [4]. Moebius syndrome is also known by other names such as congenital nuclear aplasia, child like nuclear aplasia, oculofacial congenital paralysis and facial diplegia [5]. The causes of Moebius syndrome are poorly understood. Moebius syndrome is thought to result from a vascular disruption of the features of differing designations, and would also address the problem of identifying the syndrome in cases of partial expression of the phenotypes.

**Case report**

16 year old male patient third among five children of consanguineous and normally developed parents presented with asymmetric chest, with mammary hypoplasia of the right side (Figure-1). He had a shapeless right shoulder, flattening of the right chest wall and absence of the right axillary fold. The pectoralis major muscle of right side was absent but the pectoralis minor muscle and anterior serratus muscle were present (Figure-1). Hypoplasia of scapular bone was also observed. (Figure-2) Right hand showed syndactyly of 2nd and 3rd finger whereas fingers of contralateral hand was normal. (Figure-3) Physical examination of face showed facial paralysis of right side. It also showed ptosis, lack of facial expression, strabismus and lip was cornered downwards and mouth was deviated towards right side. (Figure-4) Examination revealed an adolescent with normal growth parameters and normal motor and intellectual development. There was no history of recurrent respiratory infections, sinusitis, or bronchiectasis. Familial history was unremarkable. There were no unusual cranio-facial findings. He had normal heart sounds, and respiration and breath pattern were normal as well. Radiological examination of the chest showed no abnormalities of the ribs or heart. After a thorough search and symptomatic analysis the diagnosis was narrowed down to two syndromes.

1. Poland syndrome
2. Moebius syndrome

So, final diagnosis of Moebius-Poland syndrome was given.

**Discussion:**

The Moebius syndrome (OMIM 157900) is an infrequent symptomology in which the sixth and seventh cranial nerves are involved, this results in facial paralysis. Moebius syndrome is also known by other names such as congenital nuclear aplasia, child like nuclear aplasia, oculofacial congenital paralysis and facial diplegia [5]. The causes of Moebius syndrome are poorly understood. Moebius syndrome is thought to result from a vascular disruption

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**Figure-1: Asymmetric chest, with mammary hypoplasia of the right side, flattening of the right chest wall and absence of the right axillary fold and pectoralis major muscle.**
(temporary loss of blood flow) in the brain during prenatal development[6]. Other studies demonstrated that it has a genetic origin (locus 13q12.2), associated to autosomic dominant inheritance, autosomic recessive and including cases with recessive inheritance linked to the X chromosome [7].

Figure-2: Photograph showing hypoplasia of scapular bone.

Figure-3: Syndactyly of 2nd and 3rd finger of right hand.

Figure-4: Deviation of mouth towards right side.

The syndrome is usually diagnosed during the first few days of life, because of the incomplete closure of the eyelids during sleep and the difficulty in lactation or can also be noticed while the infant is crying or laughing, associated with dribbling saliva [8]. Present case showed facial palsy of right side with deviation of mouth towards affected side. Other features of restricted eye movements and limb deformity directed towards diagnosis of Moebius syndrome. But additional features of absence of scapula, hypoplasia of pectoralis major muscle arose the ambiguity regarding diagnosis. (Table-1) Poland syndrome (OMIM 173800) is a rare congenital abnormality of the chest wall. It was described for the first time by Alfred Poland in 1841[9]. The incidence of this syndrome has been estimated to be 1:32000 with higher frequency among males (ratio 2: 1 and 3: 1) and affects the right side of the body twice as often as the left [10]. It was also reported in 20 patients with deletion of a dominant autosomic gene [11,12]. Currently the prevailing theory in the etiology of Poland Syndrome is that, at the end of the sixth week of gestation, when the upper limb bud adjacent to the chest wall is still in a stage of development, the interruption of the embryonic blood supply causes hypoplasia of the ipsilateral subclavian artery or one of its branches. Additionally, hypoplasia of the internal thoracic artery could cause the absence of the sterno-costal portion of the pectoralis major muscle, whereas hypoplasia of the brachial artery may lead to hand abnormalities [13]. Geneticists currently hold the view that Poland syndrome is rarely inherited and generally is a sporadic event. There are rare instances where more than one individual has been identified with Poland syndrome either in
the immediate or extended family. Therefore, some authors believe that an inherited abnormal vasculature formation may be the central underlying mechanism for this condition [14]. The most common clinical features seen in Poland syndrome is described in table-1 in comparison with Moebius syndrome. In present case, we found many features of Poland syndrome overlapped with those associated with Moebius syndrome. This possibly indicates incomplete penetrance and partial expression of the phenotypic characteristics. Moebius and Poland syndromes are rare congenital anomalies. However, Moebius-Poland syndrome is rarer than the isolated syndromic conditions. Its first case was recently reported in 2007 by Diego López de Lara et al [5]. On account of the consanguinity of the pedigree and the parents being healthy, a recessive pattern of inheritance is highly probable in the present case.

<table>
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<tr>
<th>Characteristic features</th>
<th>Poland Syndrome</th>
<th>Moebius Syndrome</th>
<th>Present case</th>
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<tbody>
<tr>
<td>Abnormal Chest wall</td>
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<tr>
<td>unilateral absence of the major pectoral muscle</td>
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<td>Breast hypoplasia</td>
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<tr>
<td>Unilateral or bilateral complete or incomplete facial weakness</td>
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<td>Scapula anomaly</td>
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<td>Limitation of eye movement</td>
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<td>Abnormal limb craniofacial malformations</td>
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<td>Syndactyly</td>
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<td>Auricular anomalies</td>
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<td>Difficulty in breathing/ Swallowing</td>
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Table-1 : Comparative evaluation of features in the present case with Poland syndrome and Moebius syndrome

Conclusion
Proper systematic methodology is needed in classifying or categorizing the syndromes discussed. The authors suggest the need for further gene analysis that considers the possibility of mutation at different gene loci in syndromes showing overlapping features.

Reference