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Case Report

Imaging findings in Möbius-Poland syndrome

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ABSTRACT

Poland syndrome refers to a chest wall disorder in which there is a deficiency of the pectoral musculature. Möbius syndrome is a rare disorder in which there is absence or hypoplasia of the facial or abducens nerve, either unilaterally or bilaterally. Described here is a case in a newborn male in which both conditions manifest simultaneously as Poland-Möbius syndrome. The imaging findings here serve as a useful guide for the radiologist and ordering providers by reinforcing the need for dedicated cranial nerve imaging in patients who have deficiencies in anterior chest wall musculature.

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Introduction

The Poland and Möbius syndromes are rare congenital disorders of musculoskeletal and nervous system anatomy, respectively, that can be associated with one another. Poland syndrome is characterized by unilateral aplasia or hypoplasia of the chest wall muscles, often with symbrachydactyly or other anatomical malformations of the hand, arm, or chest [1]. Alfred Poland first described the syndrome in 1841. He documented a patient at autopsy with aplastic left pectoralis muscles and ipsilateral symbrachydactyly [2]. The syndrome

tends to have a higher incidence among males and occurs more often on the right side of the body [3]. Cases are generally believed to arise sporadically, although familial inheritance of Poland syndrome has been described [4]. The etiology of Poland syndrome is unknown, but a dominant theory posits that chest wall anatomical malformations are caused by compromised subclavian artery blood supply during *in utero* development [5].

Möbius syndrome is a congenital cranial nerve palsy typified by hypoplastic or aplastic facial and abducens cranial nerves with bilateral or unilateral involvement. The syndrome was first described by Von Graefe in 1880 and subsequently

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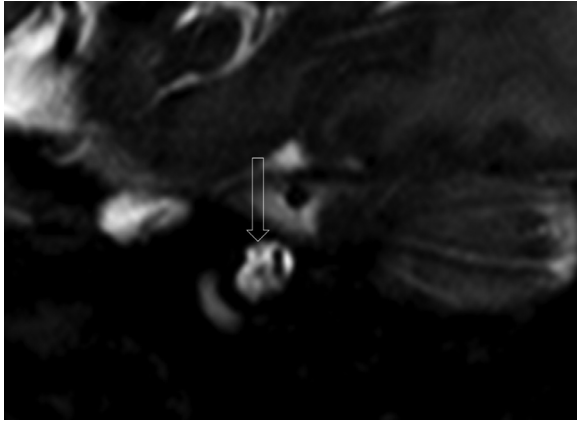


Fig. 1 – Sagittal reformatted image of the right internal auditory canal acquired at 3 days old shows a hypoplastic right facial nerve (arrow).

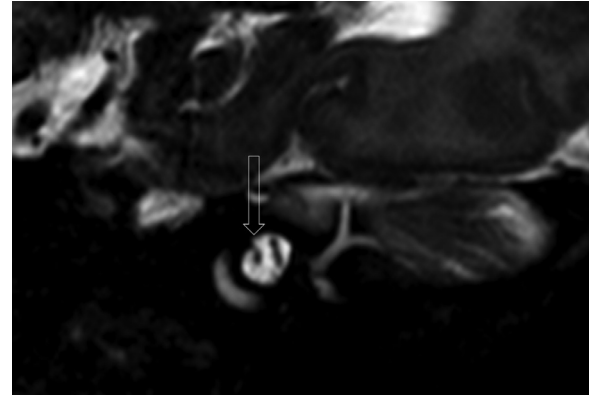


Fig. 2 – Sagittal reformatted image of the left internal auditory canal obtained at 3 days old demonstrates an absent left facial nerve (arrow).

by Paul Julius Möbius in 1888 [6,7]. Möbius syndrome is uncommon with a reported incidence in Holland in 1996 of 4 in 189,000 live births [8]. The majority of Möbius syndrome cases are believed to be sporadic with some attributed to teratogenic exposure or chromosomal abnormalities. Only a minority of cases have been shown to be familial [7,9]. The etiology of Möbius syndrome is also not fully elucidated. Similar to Poland syndrome, a pathogenesis for Möbius syndrome has been proposed which attributes the congenital anomalies to deficient arterial supply during early embryogenesis [5].

In this report, we present the case of a child who demonstrated features consistent with both Poland and Möbius syndromes. The role of the radiologist in recommending appropriate imaging studies in order to evaluate fully the extent of congenital abnormalities in patients with suspicion for Poland syndrome is discussed.

Case report

A newborn male exhibited facial paresis with mouth droop, mask-like face, and absent gag reflex. Physical exam showed asymmetry of the chest. The diagnosis of Möbius syndrome was suggested by the genetics service, and the patient underwent magnetic resonance imaging (MRI) of the brain with dedicated high-resolution images through the internal auditory canals to confirm the diagnosis. MRI demonstrated hypoplasia of the right facial nerve (Fig. 1) and absence of the left facial nerve (Fig. 2).

Chest radiographs obtained throughout the first few months of life showed opacities throughout both lungs which masked radiographic findings of Poland syndrome. A chest radiograph obtained at 2 years of age showed a hyperlucent left hemithorax (Fig. 3). Subsequent CT of the chest confirmed severe hypoplasia of the left pectoral musculature (Fig. 4) thus confirming Poland syndrome.

At the age of 9 years, the patient continues to experience complete paralysis of the left side of his face and cannot close his left eye while sleeping.

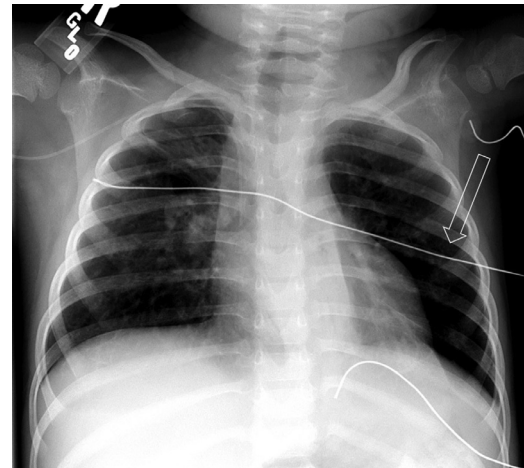


Fig. 3 – Radiograph of the chest performed at 2 years of age depicts a relatively hyperlucent left hemithorax (arrow).

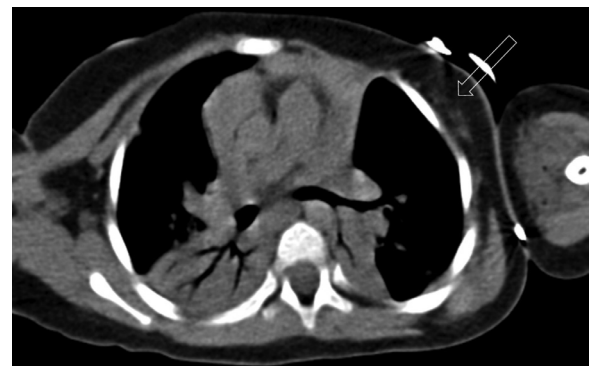


Fig. 4 – CT of the chest attained at 2 years of age shows absence of the left pectoral musculature (arrow).

Discussion

Poland syndrome is often suspected after the physical exam of a patient, yet subtle anomalies may be missed and ultimately discovered on routine imaging studies. Although rare, Poland syndrome should be included on the differential diagnosis for a unilaterally hyperlucent hemithorax on chest radiography, especially in the absence of cardiopulmonary symptoms. Romanini et al proposed an algorithm for the imaging evaluation of suspected Poland syndrome that begins with ultrasound to confirm unilateral deficiency in chest wall musculoskeletal anatomy. The authors recommend a dedicated chest radiograph if there is suspicion for rib agenesis. Furthermore, a computed tomography scan or MRI is indicated for patients with chest wall malformations requiring plastic or thoracic surgery intervention [10].

Radiologists working in pediatrics should be aware of the association between Poland and Möbius syndromes. McClure et al found that 20% of the 44 patients with Möbius syndrome had Poland syndrome [9]. A brain MRI should be considered to evaluate for hypoplasia/aplasia of cranial nerves if clinical suspicion exists for concurrent Möbius syndrome in a patient with chest wall asymmetry. The radiologist must protocol the study with sequences that permit the focused assessment of the abducens and facial cranial nerves. Specifically, the brain MRI should include high resolution heavily T2-weighted sequences through the brainstem and internal auditory canals with sagittal reformats for the seventh and eighth cranial nerves. Additionally, the brainstem should also be thoroughly investigated, as 32% of patients with Möbius syndrome have been shown to have brainstem hypoplasia [11].

Poland syndrome and Möbius syndrome have a variety of other associations that may require additional imaging, although the strength of association with these other disorders is currently not well elucidated. For example, Poland syndrome is associated with congenital anomalies which include, but are not limited to, “morning glory” syndrome, dextrocardia, and renal malformations [12–14]. Furthermore, Möbius syndrome is associated with a range of orthopedic abnormalities including clubfoot and scoliosis [9]. Additional work up for these other conditions should be considered as clinically appropriate.

In summary, Poland and Möbius syndromes are associated rare congenital disorders of anatomy with a possible common underlying pathophysiology during embryonic development which involve the musculoskeletal and nervous systems,

respectively. The radiologist plays an integral role in the diagnosis of both syndromes and can help recommend additional imaging and/or clinical evaluation given the known association of the 2 syndromes.

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