SCIENTIFIC ARTICLE

Controversies in Poland Syndrome: Alternative Diagnoses in Patients With Congenital Pectoral Muscle Deficiency

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Purpose Poland syndrome was first described as a deficiency of the pectoral muscle with ipsilateral symbrachydactyly. Currently, numerous case reports describe variations of Poland syndrome in which pectoral muscle deficiency is often used as the only defining criterion. However, more syndromes can present with pectoral muscle deficiency. The aim of this review is to illustrate the diversity of the phenotypic spectrum of Poland syndrome and to create more awareness for alternative diagnoses in pectoral muscle deficiency.

Methods A systematic literature search was performed. Articles containing phenotypical descriptions of Poland syndrome were included. Data extraction included number of patients, sex, familial occurrence, and the definition of Poland syndrome used. In addition, hand deformities, thoracic deformities, and other deformities in each patient were recorded. Alternative syndrome diagnoses were identified in patients with a combination of hand, thorax, and other deformities.

Results One hundred-and-thirty-six articles were included, describing 627 patients. Ten different definitions of Poland syndrome were utilized. In 58% of the cases, an upper extremity deformity was found and 43% of the cases had an associated deformity. Classic Poland syndrome was seen in 29%. Fifty-seven percent of the patients with a pectoral malformation, a hand malformation, and another deformity had at least 1feature that matched an alternative syndrome.

Conclusions Pectoral muscle hypoplasia is not distinctive for Poland syndrome alone but is also present in syndromes with other associated anomalies with a recognized genetic cause. Therefore, in patients with an atypical phenotype, we recommend considering other diagnoses and/or syndromes before diagnosing a patient with Poland syndrome. This can prevent diagnostic and prognostic errors.

Clinical relevance Differentiating Poland syndrome from the alternative diagnoses has serious consequences for the patient and their family in terms of inheritance and possible related anomalies. *(J Hand Surg Am. 2017;■(■):1.e1-e14. Copyright © 2017 by the American Society for Surgery of the Hand. All rights reserved.)*

Key words Congenital upper limb anomalies, differential diagnoses, Poland syndrome.

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The use of the term "Poland syndrome" has a long, controversial history in the literature. In 1841, Alfred Poland[1] described a cadaver with deficiency of the pectoral muscles and ipsilateral symbrachydactyly. In 1895, Thomson was the first to document that syndactyly and deficiency of the pectoral muscles often accompany each other, which led to the suggestion of a new syndrome by Furst in 1900, characterized by deficiency of the pectoral muscles and ipsilateral syndactyly.[2] Two years later, Bing was the first to publish a case series of patients with deficiency of the pectoral muscles and syndactyly.[3] Nevertheless, it was 60 years later that Poland's name was used by Clarkson, a plastic and hand surgeon, as an eponym for the combination of deficiency of the pectoral muscle and syndactyly (Poland syndactyly).[2] Unfortunately, the original phenotypic description of the patient of Alfred Poland was thereby abandoned. Subsequently, "Poland syndactyly" was transformed into "Poland syndrome" and its equivalents "Poland sequence" and "Poland anomaly." These terms have been used in the scientific literature ever since[2].

Currently, the eponym "Poland syndrome" has become a universal term for clinicians to describe all disturbances of pectoral development, with or without symbrachydactyly. This is illustrated by Yiyit et al[4] who reported 113 patients with Poland syndrome of whom only 25 had various upper limb anomalies. Moreover, Catena et al[5] described 8 different types of hand anomalies related to Poland syndrome. The diversity of these reports raises the question of whether Poland syndrome is 1 entity or a group of separate subentities sharing only 1 phenotypic feature, namely pectoral deficiency.

Poland syndrome is not the only syndrome in which disturbances of pectoral development can be observed. For example, Holt-Oram and Duane radial-ray syndrome both can present with absence of the pectoral major muscle together with upper limb anomalies.[6,7] Misdiagnosing patients with pectoral muscle deficiencies as Poland syndrome instead of 1 of the alternative diagnoses might lead to false assumptions about etiology, resulting in a failure to identify associated anomalies or genetic diagnoses.

To create more awareness of the alternative diagnoses in patients with pectoral muscle deficiency, we sought to illustrate the phenotypical spectrum of Poland syndrome in the literature by conducting a systematic review on its presentation. From this review, we identified all atypical Poland cases and defined the phenotypic features that should alert the clinician for a possible alternative diagnosis. We hypothesized that the incorrect use of the eponym Poland syndrome might result in misdiagnosis of some patients.

METHODS

For this systematic review, the PRISMA (Preferred Reporting Items for Systematic Reviews and Meta-Analyses) guidelines were followed and the checklist is available in the online supplements to this article (Appendix A; available on the *Journal*'s Web site at www.jhandsurg.org). The systematic review protocol was registered in PROSPERO (CRD42015016679).

Search strategy

Seven different databases (Embase, Medline [OvidSP], Web-of-science, Scopus, Pubmed publisher, Cochrane, and Google scholar) were searched for eligible articles. The search strategies used are listed in Appendix B (available on the *Journal*'s Web site at www.jhandsurg.org) and the search was performed in May 2015. Original research articles and case reports containing a phenotypical description of Poland syndrome in the Dutch or English language were included. Articles exclusively about treatment or surgery in Poland syndrome, Möbius syndrome, and general thoracic deformities were excluded. Moreover, reviews, letters to the editor, and articles not available in full text in the medical library of the Erasmus University Medical Center, were also excluded.

Inclusion of articles was done by at least 2 out of 3 reviewers (M.B., E.B.B., and D.S.) and was based on screening of title and abstract. All differences between reviewers in the selection of articles were resolved by consensus. A subsequent exclusion of articles was done during full-text reading, when articles did not fulfill the inclusion criteria.

Data-extraction

Two reviewers (M.B. and E.B.B.) independently extracted data regarding study characteristics and outcomes with the use of a standardized extraction table. The included studies were scored based on number of patients, sex, familial cases, and side of deformity. Furthermore, journal type, definition of Poland syndrome used in the paper, and causal hypothesis supported by the authors were extracted from the articles. A second database was created that included all separate patients described in the included studies. Specific hand and thoracic deformities were extracted, together with other reported anomalies and genetic outcomes. Other reported anomalies were classified in groups by cardiovascular, respiratory,