SCIENTIFIC ARTICLE

Functional Impact of Congenital Hand Differences: Early Results From the Congenital Upper Limb Differences (CoULD) Registry

Donald S. Bae, MD,* Maria F. Canizares, MD,* Patricia E. Miller, MS,* Peter M. Waters, MD,* Charles A. Goldfarb, MD†‡

Purpose To characterize the functional, emotional, and social impact of congenital upper limb differences on affected children and families before treatment, using validated functional outcome instruments.

Methods From June 2014 to March 2016, 586 children with congenital upper limb differences from 2 pediatric hospitals were enrolled in the Congenital Upper Limb Differences registry. Demographic, clinical, and radiographic data were collected, and diagnoses categorized according to the Oberg-Manske-Tonkin classification. Functional outcomes were assessed in 301 patients using the Pediatric Outcomes Data Collection Instrument (PODCI) and Patient-Reported Outcomes Measurement Information System (PROMIS) upper extremity (UE) function, pain, anxiety, depression, and peer relationships modules.

Results The cohort had high median PODCI scores in all domains, ranging from 83 to 100 in children and adolescents. Patients had decreased PROMIS UE scores compared with population norms; however, they showed low scores for pain, anxiety, depression and higher scores in the peer relationship domain, respectively. Patients with entire limb involvement had higher PROMIS pain scores and lower PODCI UE and global functioning than those with differences limited only to the hand. Compared with those with bilateral involvement, patients with unilateral differences reported higher scores for PODCI sports global functioning, better PROMIS UE function, and lower pain scores. Additional orthopedic conditions and medical comorbidities negatively influenced all PODCI scores and PROMIS pain and UE function domains.

Conclusions Children with congenital hand differences report decreased upper limb function but better peer relationships and positive emotional states compared with population norms.

Clinical relevance The Congenital Upper Limb Differences registry is a valid source of information related to congenital upper limb differences in clinical practice. With continuous enrollment and longitudinal follow-up, the registry will increase the understanding of UE function and psychosocial aspects of health in pediatric population. *(J Hand Surg Am. 2017;■(■):■−■. Copyright © 2017 by the American Society for Surgery of the Hand. All rights reserved.)*

Key words Congenital, hand differences, function, outcomes, registry.

*From the *Department of Orthopaedic Surgery, Children's Hospital Boston, Boston, MA; the †Department of Orthopaedic Surgery, Shriners Hospitals for Children—St. Louis; and the ‡Department of Orthopaedic Surgery, Washington University School of Medicine, St. Louis Children's Hospital, St. Louis, MO.*

Received for publication December 2, 2016; accepted in revised form October 2, 2017.

No benefits in any form have been received or will be received related directly or indirectly to the subject of this article.

Corresponding author: Donald S. Bae, MD, Department of Orthopaedic Surgery, Boston Children's Hospital, 300 Longwood Avenue, Hunnewell 2, Boston, MA 02115; e-mail: donald.bae@childrens.harvard.edu.

0363-5023/17/■ ■-0001$36.00/0
https://doi.org/10.1016/j.jhsa.2017.10.006

CONGENITAL UPPER LIMB DIFFERENCES affect approximately 1 in 1,000 children,[1–3] and the goal of treatment continues to be maximization of overall function. After the World Health Organization's[4] (WHO) proposal of standard language for the description of health states and disability (the International Classification of Function, Disability, and Health, or ICF), there have been increased efforts to assess pediatric patients in a holistic manner, including physical, psychological, and social perspectives. To this end, patient-reported outcome (PRO) tools have been advocated to assess the impact of a condition on the patient's function and well-being; additionally, these tools help providers better understand the outcomes of treatment.[5]

Prior studies have used PROs to assess the impact of congenital upper limb differences in children.[6–17] Overall, these studies have reported a wide spectrum of outcomes depending on the type of congenital difference and the level of involvement. For example, patients with unilateral below-the-elbow deficiency exhibit deficits in upper extremity function but better emotional and social health than population controls.[6,11] In contrast, bilateral involvement in amyoplasia results in lower physical and emotional health states than unaffected children.[13] Interestingly, differences are seen in how affected children view their condition compared with their parents; parents commonly overestimate emotional domains yet perceive worse physical function and quality of life compared with their affected children.[8,14]

Although these preliminary findings are enlightening, prospective longitudinal investigation is needed in larger numbers of patients throughout the spectrum of congenital hand differences. In addition, the advent of newer PROs has created opportunities for an additional study using validated instruments. The purpose of this investigation was to assess the functional, emotional, and social impact of congenital upper limb differences in children enrolled in the Congenital Upper Limb Differences (CoULD) registry before treatment, using the Pediatric Outcomes Data Collection Instrument (PODCI) and the Patient-Reported Outcomes Measurement Information System (PROMIS).

METHODS

Data source

The analysis was based on patient information collected for the CoULD registry. This ongoing prospective cohort study was implemented in June 2014 by 2 US tertiary-care pediatric centers and has now been expanded to include multiple centers across the United States. The purpose of the registry is to describe the epidemiology, clinical characteristics, function, and health status of children with congenital upper limb differences, and the quantification of improvements with nonoperative and surgical care. Patients below 18 years of age are only considered eligible to enroll in CoULD if a diagnosis of a congenital difference has been established and no surgical treatment has been performed previously. At baseline, pertinent clinical and radiographic data are collected, and diagnoses are categorized according to the Oberg-Manske-Tonkin classification system.[18] In addition, age-appropriate PODCI and PROMIS questionnaires are administered to patients and parents during the initial enrollment and preoperative visit, for ages 2 to 18 when applicable. As part of the longitudinal follow-up, clinical, operative, and functional outcome information is collected during clinic visits or electronically. The Institutional Review Board at the Washington University School of Medicine in St. Louis approved the study, and parental consent and pediatric assent were obtained in accordance with institutional guidelines.

Patient population

From June 2014 to March 2016, 586 patients from 2 pediatric hospitals were enrolled in the CoULD registry. Of those, 285 subjects did not have available questionnaire data, including 252 who were not able to complete the first questionnaire because the PROMIS and PODCI questionnaires were not validated for patients younger than 2 years of age; 33 patients older than 2 years of age had incomplete forms, leaving 301 subjects with available preoperative questionnaires data.

Outcome instruments

The PODCI was designed by a joint effort between the American Academy of Orthopaedic Surgeons and Pediatric Orthopaedic Society of North America to assess the overall health, pain, and ability to participate in daily activities and sports in patients from 2 to 18 years of age. It was designed to be completed by parents with knowledge of their child's condition, as well as by patients capable of independent self-reporting.[19] PODCI questionnaires included the following domains: upper extremity and physical function; transfer and basic mobility; sports and physical functioning; pain and comfort; happiness and global functioning. PODCI scores are presented