Use of Pediatric Outcomes Data Collection Instrument to Evaluate Functional Outcomes in Multiple Hereditary Exostoses

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Background: The Pediatric Outcomes Data Collection Instrument (PODCI) is a validated quality-of-life questionnaire with 6 domains designed to provide a standardized method of measuring outcomes in pediatric musculoskeletal conditions. To our knowledge there are no reports on its use in children with multiple hereditary exostosis (MHE).

Questions/Purposes: Most published studies on MHE patients have described the efficacy of specific surgical techniques or the specification of deformities. Little is known about the general health status of pediatric patients, the severity of pain, loss of function, and how MHE influences the activities of daily life. We aim to assess the functional levels of MHE pediatric patients with PODCI questionnaire.

Patients and Methods: As a cross-sectional study, we prospectively administered PODCI to 34 pediatric patients diagnosed with MHE and their families. The score distributions were compared with values published earlier for children and adolescents without musculoskeletal disorders using the Student and Welch *t* tests. Parents and adolescents' reports were compared using Wilcoxon signed rank test. Physical examination and PODCI score relation were evaluated by Spearman test.

Results: Children with MHE have significantly lower scores ($P < 0,05$) in comparison with unaffected children in all domains using the Student and Welch *t* test. Parents score differs from children score with statistically relevance in pain and comfort domain ($P < 0,5$). The Spearman test showed a negative correlation between physical examination and PODCI score with statistical significance.

Conclusions: These results point towards PODCI's capacity in evaluating functional outcomes of pediatric patients with MHE.

Level of Evidence: Diagnostic Study, Level III.

Key Words: multiple hereditary exostosis, osteochondromatosis, Health-Related quality of life, patient health questionnaire, patient reported outcome measures, PODCI

(*J Pediatr Orthop* 2023;43:332–336)

The main goal of orthopaedic interventions is to improve health and physical function of patients, and thus their quality of life. It is difficult to measure and quantify functional health in children as functional performance increases with normal neurological maturation.[1] The patient is the most important source of information regarding its health status.[2] With young or intellectually impaired children, the parent is the proxy, and in many instances, they do not accurately reflect the child or adolescent perceptions.[1]

The Pediatric Outcomes Data Collection Instrument (PODCI) was developed in 1994 by the American Academy of Orthopaedic Surgeons and the Pediatric Orthopaedic Society of North America (POSNA).[1] It is a self-report scale designed to provide a standardized method of evaluation of musculoskeletal conditions in pediatric population. It consists of 6 domains: (1) upper extremity function, (2) transfers and mobility, (3) sports participation, (4) pain and comfort, (5) global function, and (6) happiness with physical condition. Each domain is given a numerical score from 0 to 100, which 100 represents the highest level of function.[1] The advantages of using PODCI includes an easy administration to patients, parents and caretakers, and, relevance to their interest. The PODCI has been validated and shown to be reliable and sensitive to changes.[1] Because of its properties, the PODCI was translated and validated in multiple

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C.B.R.d.M.: Interpretation of data; critical revising for intellectual content; approval and agreement of the version to be published. N.S.P.d.O.: Conception and design of the work; drafting the work, interpretation of data, and critical revising for intellectual content; approval and agreement of the version to be published. M.R.D.d.S.: Drafting the work, interpretation of data, and critical revising for intellectual content; approval and agreement of the version to be published.

The authors state that the views expressed in the submitted article are their own and not an official position of the institutions where the study took place.

No financial support was given to the conduction of this study nor the authors.

The authors declare no conflicts of interest.

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DOI: 10.1097/BPO.0000000000002372

languages.[3–7] It has earlier been evaluated in children with many conditions, such as cerebral palsy,[8,9] scoliosis and kyphosis,[10,11] arthrogryposis,[12] obstetric brachial palsy,[3,13–15] type 1 neurofibromatosis,[16] mucopolysaccharidosis,[17] idiopathic juvenile arthritis, and obesity.[6,7,18]

The Brazilian version was translated and validated in 2013 by Do Monte FA et al[7] with 57 patients. To our knowledge, PODCI was never used to evaluate children with multiple hereditary exostoses (MHE).

MHE is a rare disorder with an autosomal dominant inheritance though to arise in around 1 in 50.000 individuals. Tumor suppressor genes EXT1 and EXT2 are involved with the etiology of the characteristic non-malignant bone tumors covered by cartilage that appear in the metaphyseal region, close to the growth zone, of long bones, ribs, hips, and vertebraes.[19,20] The symptoms are frequently related to the size and location of the exostoses, varying from totally asymptomatic to severe pain and deformities, potentially interfering in daily activities and psychosocial well-being.[19–22]

METHODS

Cross-sectional prospective research was performed in 2 Institutions by the rules and authorization of the Brazilian Ethical Research Committee (Comitê de Ética em Pesquisa – CEP). Recommendations and approval of both institutions' Ethical Research Committees were respected before the beginning of the study.

Children and adolescents with MHE diagnosed by the oncological service in our Institutions with follow-up or first evaluation between October 2017 and August 2018 were candidates for inclusion. Patients under completion of 2 years old or aged over 18 years old were excluded. The diagnosis of solitary osteochondroma was an exclusion criterion regardless of musculoskeletal secondary disorders. The non-agreement or consent by the subject or its legal guardians was also an exclusion criterion at any time of the study.

After PODCI's terminology, were considered children patients between 2 and 10 years old, meanwhile patients between 11 and 18 years old were considered adolescents. Children and adolescents will be referred collectively as patients in this paper.

The PODCI questionnaire was administered to parents of patients between 2 and 18 years old and to adolescents (11 to 18 y old). All patients were evaluated by physical examination which assessed lower extremities coronal alignment, leg length discrepancy, pain complain, and upper extremities mobility based on Leung and Peterson Classification System.[23]

The main objective of this study was to evaluate the results of PODCI for children and adolescents with MHE. Secondary objectives were to correlate physical examination with PODCI results, and to assess differences between parents' report and adolescents' self-report outcomes.

The Student *t* test and Welch *t* test were used to compare mean scores between children and adolescents without musculoskeletal disorders and patients with MHE.[1] Wilcoxon signed rank test was used to compare parent-reports and adolescents self-reports. The assessment

TABLE 1. Descriptive Statistical Analysis of MHE Patients Data

	N	Min	Max	Mean	Std*	CI 95	Skewness z-score	*t* test *P*-value	Welch test *P*-value
Global functioning scale									
A†	23	41	100	83,83	13,90	(77,8-89,8)	−2,64	< 0,05	< 0,05
PA‡	23	49	100	78,96	15,70	(72,2-85,7)	−1,00	< 0,05	< 0,05
PC§	11	50	98	80,55	15,88	(69,9-91,1)	−1,33	< 0,05	< 0,05
Happiness scale									
A†	23	30	100	84,35	15,54	(77,6-91,1)	−4,60	0,49	0,45
PA‡	23	5	100	72,61	29,19	(60,0-85,2)	−2,24	< 0,05	0,16
PC§	11	40	100	77,73	20,54	(63,9-91,5)	−1,51	< 0,05	0,08
Pain/comfort scale									
A†	23	8	100	68,65	27,35	(56,8-80,5)	−1,11	< 0,05	< 0,05
PA‡	23	0	100	58,09	29,59	(45,3-70,9)	−0,14	< 0,05	< 0,05
PC§	11	35	100	70,27	21,91	(55,6-85,0)	−0,26	< 0,05	< 0,05
Sports and physical functioning scale									
A†	23	21	100	80,65	20,41	(71,8-89,5)	−2,98	< 0,05	< 0,05
PA‡	23	17	100	74,78	22,86	(64,9-84,7)	−2,12	< 0,05	< 0,05
PC§	11	40	95	78,73	19,85	(65,4-92,1)	−2,16	< 0,05	0,08
Transfer and basic mobility scale									
A†	23	60	100	92,65	12,06	(87,4-97,9)	−3,92	< 0,05	< 0,05
PA‡	23	73	100	92,09	7,88	(88,7-95,5)	−2,02	< 0,05	< 0,05
PC§	11	74	100	91,36	10,77	(84,1-98,6)	−1,03	< 0,05	0,06
Upper extremity scale									
A†	23	67	100	93,22	9,35	(89,2-97,3)	−3,37	< 0,05	< 0,05
PA‡	23	38	100	91,26	14,70	(84,9-97,6)	−5,43	< 0,05	< 0,05
PC§	11	48	100	81,64	20,61	(67,8-95,5)	−1,46	< 0,05	0,13

*Standard deviation.
†Adolescents self-report.
‡Parents of Adolescents report.
§Parents of Children report.
Max indicates maximum; Min, minimum; N, number.

of physical examination was related to PODCI scores using the Spearman test. The skewness z-score was used to evaluate the discriminative property between individuals ("ceiling/floor effect").

Statistical significance was defined as P-value <0.05 in a 2-tailed test. Data analysis was performed using SciPy 1.1.0 open-source software.

RESULTS

A total of 34 patients met the criteria to be included in this study. There were 21 male and 13 female, of those, 11 children and 23 adolescents. All questionnaires were sufficiently complete to calculate a score for all domains. Thirty-three patients had full assessment of physical examination.

The descriptive statistics of PODCI responses for children and adolescents with MHE is shown in Table 1. The mean scores for patients with MHE were significantly lower than the mean aggregated scores for children and adolescents without musculoskeletal disorders (Table 2) in all domains using Student and Welch t tests (Fig. 1).

Wilcoxon test shows no significant difference among all pairs of comparison between pediatric self-reports and parent proxy-reports in all domains, except Pain, and Comfort Scale. (Table 3). In all domains, the parent mean score (PA) was lower than the adolescent mean score (A). The skewness z-score for children with MHE was below zero (Table 1).

TABLE 2. Posna Normative Data

	N	Mean	Std*
Global functioning scale			
A†	1834	95.88	5.38
PA‡	1834	95.15	7.24
PC§	1791	93.31	7.77
Happiness scale			
A†	1834	81.83	17.59
PA‡	1834	81.47	18.01
PC§	1791	89.80	14.10
Pain/comfort scale			
A†	1834	89.31	14.79
PA‡	1834	88.96	16.67
PC§	1791	92.43	13.75
Sports and physical functioning scale			
A†	1834	95.51	9.74
PA‡	1835	93.66	10.99
PC§	1791	90.22	12.32
Transfer and basic mobility scale			
A†	1834	99.05	4.70
PA‡	1834	99.22	4.56
PC§	1791	98.35	5.68
Upper extremity scale			
A†	1834	98.71	4.73
PA‡	1834	98.82	5.08
PC§	1791	91.97	11.49

*Standard deviation.
†Adolescents self-report.
‡Parents of Adolescents report.
§Parents of Children report.
N indicates number.

Pain, range of motion of upper extremities, and coronal alignment and dysmetria of lower extremities were assessed by physical examination and compared with PODCI score using Spearman test. On the basis of Leung and Peterson Classification System,[23] alterations on physical examination in the upper extremities were indicated on a binary score that ranged from 0 to 15 (from less impairment to great impairment). It was shown a distribution range of physical examination from 0 to 10. The comparison between physical examination and parent based reports (N = 33) showed a negative correlation (−0,43) with statistical significance and confidence ($P < 0,05$; CI 95%) which reflects that disabilities objectively measured by physicians have a negative impact on HRQL perception with lower PODCI scores as a result. Physical examination data were also compared with adolescents self-report (N = 23) which maintained the same tendency (correlation −0,42 ; $P < 0,05$, CI 95%).

DISCUSSION

MHE is a genetic driven musculoskeletal disorder with a wide variety of clinical presentation, from totally asymptomatic to great disability.[19,20,22,24] Decision making in orthopaedic practice can be challenging as patient, parents, and physician perception about functional condition and complains may differ and impact on children and adolescents' treatment.[1,2,25]

Patient-based outcomes are used to evaluate a patient's subjective health experience and the consequences of the patient's disease and medical interventions. The self-assessment with self-administered questionnaire is considered the main standard method of quality-of-life evaluation, removing influence or interpretation from a clinical or investigator.[2,25] This approach to outcome assessment has particularities as functional performance and neurological maturation changes over time in a child/adolescent under either typical or atypical development.[25]

The PODCI was developed to assess musculoskeletal functional outcomes in children and adolescents and has not been replicated in all areas of pediatric orthopaedics.[1,25] The purpose of this study was to evaluate its use in MHE patients.

When PODCI was evaluated in children and adolescents without musculoskeletal disorders, a "ceiling effect" was noted, where the distribution was heavily skewed with scores clustered near the maximum score (no mean score under 85), resulting in poor ability to differentiate between individuals.[26] As MHE can be presented as totally asymptomatic condition we were concerned about the reproduction of this "ceiling effect". Although PODCI scores in MHE patients were left skewed, it was shown a tendency of distribution proximate to normative values yet with an absence of "ceiling effect" (means range: 70-93). It reflects a good discriminative property on our sample although we believe selection bias may interfere on this result, as *pauci* or asymptomatic patients often do not have diagnosis or clinical follow-up on Institutions.[19]

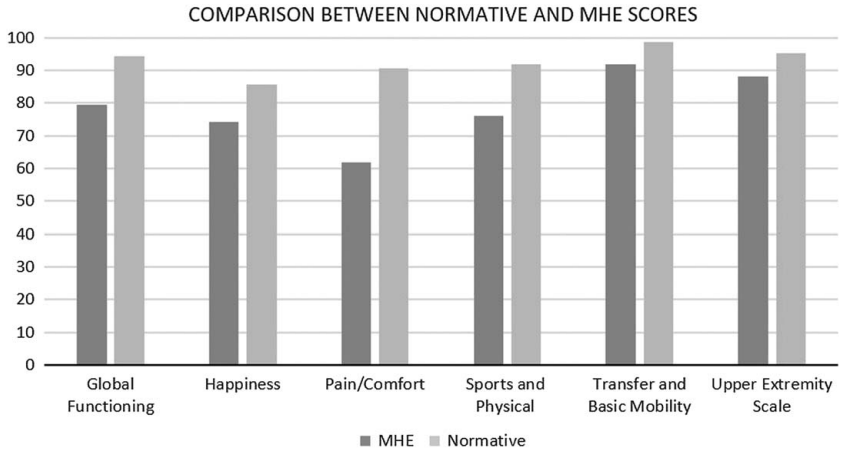

FIGURE 1. Comparison between normative and MHE scores. MHE indicates multiple hereditary exostosis.

Physical examination evaluation showed a statistical relevant correlation with PODCI score assessing its capacity of revealing quality-of-life impairment related to physical disabilities. The negative correlation between physical examination and reports (parents and adolescents self-reports) points toward a negative correlation between alteration on physical examinations and quality-of-life, where higher scores in the Leung and Peterson classification, and thus greater impairment in the upper extremities, in addition to lower extremity deformities are correlated to lower PODCI scores, as expected.

The perception of pain and comfort by the parents was worse than the patient's own perception in our data. Daltry et al[1] reported that the correlation among parents and adolescents in that scale was also low at follow-up. We agree with those authors' speculation were the health and well-being of the parents might influence their perceptions on how well the child is doing, as well as the expectations of 2 different individuals can similarly vary greatly.[1]

Our literature review reveled only a few studies dedicated specifically to evaluate quality of life of pediatric MHE patients. Chhina H and colleagues assessed health-related quality of life (HRQL) of MHE patients using The Child Health Questionnaire (CHQ PF-50). The CHQ PF-50 is a generic pediatric HRQL questionnaire that was applied to patients (n = 43; 5 to 18 y old) in the mentioned study by parent proxy. Chhina H et al[21] concluded that MHE population had lower HRQL than the general population. Goud et al[24] also conducted a HRQL study about MHE patients but, as highlighted by the authors, no validated pediatric quality-of-life assessment tool was used.

Caino and colleagues were the first authors to conduct a MHE evaluation study with a specific HRQL pediatric instrument. The population assessment with the Pediatric Quality of Life Inventory (PedsQL) was made by self-administration by children aged 8 to 18 years and by the parents of children aged 2 to 18 years. The PedsQL is a 23-item instrument with 4 domains: (1) physical functioning (8 items); (2) emotional functioning (5 items); (3) social functioning (5 items); and (4) school functioning (5 items). Although no statistically significant difference was observed between children and caregivers responses, children reported school domain as the most affected, whereas parents referred to the emotional aspect. Caino et al[22] concluded that pain and disease severity had a negative impact on HRQL.

PODCI is a validated HRQL instrument specific to musculoskeletal conditions in pediatric population. Considering the importance of self-administered questionnaires and its properties, adolescent's responses were evaluated in separate and compared with its parents' counterparts on this study. In all domains, the parent mean score was lower than the adolescent mean score. Although Wilcoxon test showed statistically significant difference only in Pain and Comfort domain ($P < 0,05$), a tendency of over estimating limitation by parents can be seen. A bigger sample would be more reliable evaluating this behavior in all domains.

TABLE 3. diference Significance between Self (A) and Parent Report (PA)

	Wilcoxon *P*-value
Global functioning scale	0,067
Happiness scale	0,081
Pain/comfort scale	0,004
Sports and physical functioning scale	0,191
Transfer and basic mobility scale	0,534
Upper extremity scale	0,511

CONCLUSION

This study was designed to evaluate the results of PODCI for children and adolescents with MHE as a sufficiently sensitive HRQL instrument. Hence, analysis of parent-reported scores and adolescents self-report achieved its purpose and it is in concordance with other studies found in literature.[21,22] PODCI adds to orthopaedic practice the patient perspective, in a pediatric scenario, about its own quality-of-life related specifically to

musculoskeletal conditions. We believe PODCI questionnaire can be used as an assistant managing tool for physicians concerning MHE follow-up and treatment decision making. Prospective studies with bigger samples focused on assessment of its psychometric properties specifically for MHE pediatric population may be of greater value.

ACKNOWLEDGMENTS

The authors thank Lucas Pierezan Magalhães, MSs Computer Science and Bsc Mathematics, for its great assistance with statistical methodology and application. This paper would not be possible without his efforts and patience. The authors also acknowledge their former institutions, Instituto Nacional de Traumatologia e Ortopedia (INTO), Rio de Janeiro, RJ—Brazil and Hospital Estadual da Criança (HEC), Rio de Janeiro, RJ—Brazil that made this research project possible.

REFERENCES

1. Dantroy L, Liang M, Fossel A, et al. The POSNA Pediatric Musculoskeletal Functional Health Questionnaire: Report on Reliability, Validity, and Sensitivity to Change. *J Pediatr Orthop*. 1998;18:561–571.
2. Meadows KA. Patient-reported outcome measures: an overview. *Br J Community Nurs*. 2011;16:146–151.
3. Holst M, Vlieland T, Sande M, et al. Translation and adaptation of the Pediatric Outcome Data Collecting Instrument (PODCI) into the Dutch language and preliminary validation in children with Neonatal Brachial Plexus Palsy. *J Pediatr Rehabil Med*. 2015;8:219–226.
4. Kwon DG, Chung CY, Lee KM, et al. Transcultural adaptation and validation of the Korean version of the pediatric outcomes data collection instrument (PODCI) in children and adolescents. *J Pediatr Orthop*. 2011;31:102–106.
5. Merder-Coskun D, Kenis-Coskun O, Celenlioğlu AE, et al. Reliability of cross-cultural adapted Turkish version of the Pediatric Outcomes Data Collection Instrument (PODCI). *J Pediatr Rehabil Med*. 2016;9:101–105.
6. Misterska E, Adamczak K, Kaminiarczyk-Pyzałka D, et al. The Pediatric Outcomes Data Collection Instrument for a Polish sample with juvenile idiopathic arthritis: psychometric properties of proxy version. *Int J Rheum Dis*. 2017;20:2077–2085.
7. Do Monte FA, Ferreira MNL, Petribu KCL, et al. Validation of the Brazilian version of the pediatric outcomes data collection instrument: a cross-sectional evaluation in children and adolescents with juvenile idiopathic arthritis. *BMC Pediatr*. 2013;13:1–6.
8. McMulkin ML, Baird GO, Gordon AB, et al. The pediatric outcomes data collection instrument detects improvements for children with ambulatory cerebral palsy after orthopaedic intervention. *J Pediatr Orthop*. 2007;27:1–6.
9. Seok Park M, Youb Chung C, Min Lee K, et al. Rasch analysis of the pediatric outcomes data collection instrument in 720 patients with cerebral palsy. *J Pediatr Orthop*. 2012;32:423–431.
10. Lerman JA, Sullivan E, Haynes RJ. The Pediatric Outcomes Data Collection Instrument (PODCI) and functional assessment in patients with adolescent or juvenile idiopathic scoliosis and congenital scoliosis or kyphosis. *Spine (Phila Pa 1976)*. 2002;27:2052–2057.
11. Ugwonali OF, Lomas G, Choe JC, et al. Effect of bracing on the quality of life of adolescents with idiopathic scoliosis. *Spine J*. 2004;4:254–260.
12. Amor CJ, Spaeth MC, Chafey DH, et al. Use of the pediatric outcomes data collection instrument to evaluate functional outcomes in arthrogryposis. *J Pediatr Orthop*. 2011;31:293–296.
13. Dedini RD, Bagley AM, Molitor F, et al. Comparison of pediatric outcomes data collection instrument scores and range of motion before and after shoulder tendon transfers for children with brachial plexus birth palsy. *J Pediatr Orthop*. 2008;28:259–264.
14. Huffman GR, Bagley AM, James MA, et al. Assessment of children with brachial plexus birth palsy using the pediatric outcomes data collection instrument. *J Pediatr Orthop*. 2005;25:400–404.
15. Lerman J, Sullivan E, Barnes D, et al. The Pediatric Outcomes Data Collection Instrument (PODCI) and functional assessment of patients with unilateral upper extremity deficiencies. *J Pediatr Orthop*. 2005;25:405–407.
16. Johnson BA, Sheng X, Perry AS, et al. Activity and participation in children with neurofibromatosis type 1. *Res Dev Disabil [Internet]*. 2015;36:213–221.
17. O'Brien A, Bompadre V, Hale S, et al. Musculoskeletal function in patients with mucopolysaccharidosis using the pediatric outcomes data collection instrument. *J Pediatr Orthop*. 2014;34:650–654.
18. Podeszwa D, Stanko K, Mooney J, et al. An analysis of the functional health of obese children and adolescents utilizing the PODC instrument. *J Pediatr Orthop*. 2006;26:140–143.
19. WHO Classification of Tumours Editorial Board. *Soft Tissue and Bone Tumours*. International Agency for Research on Cancer (IARC); 2020:437–439.
20. Ryckx A, Somers J, Allaert L. Hereditary multiple exostosis. *Acta Orthop Belg*. 2013;79:597–607.
21. Chhina H, Davis JC, Alvarez CM. Health-related quality of life in people with hereditary multiple exostoses. *J Pediatr Orthop*. 2012;32:210–214.
22. Caino S, Alba R, Bevilacqua S, et al. Osteochondromatosis: clinical variability and factors related to quality of life in children and adults. *Arch Argent Pediatr*. 2022;120:180–186.
23. Leung AG, Peterson HA. Fractures of the proximal radial head and neck in children with emphasis on those that involve the articular cartilage. *J Pediatr Orthop*. 2000;20:7.
24. Goud AL, De Lange J, Scholtes VAB, et al. Pain, physical and social functioning, and quality of life in individuals with multiple hereditary exostoses in the Netherlands: a national cohort study. *J Bone Jt Surg - Ser A*. 2012;94:1013–1020.
25. Phillips L, Carsen S, Vasireddi A, et al. Use of patient-reported outcome measures in pediatric orthopaedic literature. *J Pediatr Orthop*. 2018;38:393–397.
26. Haynes RJ, Sullivan E. The Pediatric Orthopaedic Society of North America pediatric orthopaedic functional health questionnaire: an analysis of normals. *J Pediatr Orthop*. 2001;21:619–621.