HHS Public Access

Author manuscript

Disabil Rehabil. Author manuscript; available in PMC 2019 July 01.

Published in final edited form as:

Disabil Rehabil. 2018 July ; 40(15): 1773–1780. doi:10.1080/09638288.2017.1312567.

Application of the International Classification of Functioning, Disability and Health System to symptoms of Duchenne and Becker muscular dystrophies

Kristin M Conway[1], **Emma Ciafaloni**[2], **Dennis Matthews**[3], **Chris Westfield**[4], **Kathy James**[5], **Pangaja Paramsothy**[6], and **Paul A Romitti**[1]

[1]Department of Epidemiology, The University of Iowa, Iowa City, USA

[2]Departments of Neurology and Pediatrics, University of Rochester Medical Center, Rochester, USA

[3]Department of Physical Medicine & Rehabilitation, University of Colorado School of Medicine and Children's Hospital Colorado, Aurora, USA

[4]Congenital Malformations Registry (CMR), New York State Department of Public Health, Albany, USA

[5]Department of Family Medicine, Colorado School of Public Health, Aurora, USA

[6]National Center on Birth Defects and Developmental Disabilities, Centers for Disease Control and Prevention, Atlanta, USA

Abstract

Purpose—Duchenne and Becker muscular dystrophies, collectively referred to as dystrophinopathies, are X-linked recessive diseases that affect dystrophin production resulting in compromised muscle function across multiple systems. The International Classification of Functioning, Disability and Health provides a systematic classification scheme from which body functions affected by a dystrophinopathy can be identified and used to examine functional health.

Materials and Methods—The infrastructure of the Muscular Dystrophy Surveillance, Tracking, and Research network was used to identify commonly affected body functions and link selected functions to clinical surveillance data collected through medical record abstraction.

Results—Seventy-one (24 second-, 41 third- and 7 fourth-level) body function categories were selected via clinician review and consensus. Of these, 15 of 24 retained second-level categories were linked to data elements from the Muscular Dystrophy Surveillance, Tracking, and Research network surveillance database.

Corresponding Author: Paul A. Romitti, PhD, The University of Iowa, Department of Epidemiology, S416 CPHB, 145 N Riverside Dr., Iowa City, Iowa 52242, Telephone: 319-384-1549, FAX: 319-335-4095, paul-romitti@uiowa.edu.

Declaration of Interest: Data collection for this publication was supported by the Cooperative Agreement numbers, DD000187, DD000189, DD000190, DD000191, and DD000392 (plus current cooperative agreement numbers for co-authors from original and new sites) funded by the Centers for Disease Control and Prevention. Writing of this publication was supported by Cooperative Agreement numbers, DD001119, DD001123, and DD001126. Its contents are solely the responsibility of the authors and do not necessarily represent the official views of the Centers for Diseases Control and Prevention or the Department of Health and Human Services. The authors report no conflicts of interest.

Conclusions—Our findings support continued development of a core set of body functions from the International Classification of Functioning, Disability and Health system that are representative of disease progression in dystrophinopathies and the incorporation of these functions in standardized evaluations of functional health and implementation of individualized rehabilitation care plans.

Keywords

Duchenne muscular dystrophy; Becker muscular dystrophy; dystrophinopathies; International Classification of Functioning; disability; surveillance

Introduction

Duchenne and Becker muscular dystrophies, herein referred to as dystrophinopathies, are X-linked recessive genetic diseases that affect the production of dystrophin (1). Disease progression affects multiple systems with deterioration of pulmonary, cardiac, and orthopedic function (2). Severity of disease progression can occur along a spectrum. Duchenne muscular dystrophy is the more severe phenotype with earlier onset of initial motor symptoms (typically before the 5th birthday) and loss of ambulation (typically before the 12th birthday), and progressive deterioration in cardiac and pulmonary function. Becker muscular dystrophy is less severe in phenotypic expression with later onset of initial motor symptoms and loss of ambulation (typically after the 16th birthday), and less severe deterioration in other body systems. The severity and progressive nature of dystrophinopathies can have considerable impact on a patient's participation in activities across multiple life domains (3). Educational attainment rarely goes beyond secondary school and employment opportunities may be unavailable due to the need for assistance with activities of daily living, reduced physical accessibility, and the lack of assistive technology. Social relationships can also be affected due to the physical limitations imposed by reduced mobility over the lifespan.

The use of a systematic classification scheme to describe body function among patients with dystrophinopathies is a useful approach for examining physical health and its association with potential limitations in other life domains (4, 5). The International Classification of Functioning, Disability and Health (ICF) [http://www.who.int/classifications/icf/en/] (6) system was constructed, in part, to provide such a framework and develop standard language for describing disease and health-related functioning (7-10). However, the ICF system exceeds 1400 categories all of which may not apply to a particular disease symptomatology. As such, The World Health Organization and the ICF Research branch have instituted a process for identifying disease-specific ICF core sets [http://www.icf-research-branch.org/icf-core-sets-projects-sp-1641024398] consisting of selected categories representative of a disease (11, 12). Existing core sets have been developed for diseases with varying degrees of chronicity (*e.g.*, acute, post-acute, and chronic conditions) and affected systems (*e.g.*, spinal cord, cardiopulmonary, neurological, and musculoskeletal) (11, 13).

To our knowledge, the development of an ICF core set for neuromuscular diseases has been limited. In a study by Wynia et al (14), expert panels were asked to select ICF categories

most relevant to three neurologic disorders: multiple sclerosis, Parkinson's disease, and neuromuscular diseases, such as motor neuron diseases and muscular dystrophies. From this initial list, Bos et al (3) validated the ICF core set by linking the selected categories with health-related quality of life measures designed to evaluate functioning among those affected primarily by neuromuscular disease, specifically the Individualized Neuromuscular Quality of Life Questionnaire, Amyotrophic Lateral Sclerosis Assessment Questionnaire, and the Myasthenia Gravis Quality of Life 60. For this study, we propose to further refine the list of selected ICF categories by selecting those that are representative of a specific neuromuscular disease, childhood-onset dystrophinopathies, and to evaluate the feasibility of retrospectively linking selected ICF categories to medical record data using clinical data collected by the Muscular Dystrophy Surveillance, Tracking, and Research Network (MD STAR*net*).

Materials and Methods

Sample

Methodology for the MD STAR*net*, a population-based surveillance system, has previously been described (15, 16). Briefly, through the 'Muscular Dystrophy Community Assistance, Research and Education Amendments of 2001', the U.S. House of Representatives and Senate enacted legislation to amend the Public Health Service Act to provide for research with respect to various forms of MD, including dystrophinopathies. The overall objectives of the MD STAR*net* were to: 1) conduct active, population-based surveillance and characterize the epidemiology of dystrophinopathies and 2) develop long-term follow-up and tracking of children with dystrophinopathies to describe disease progression and outcomes of treatment and services provided.

Starting in 2004, the MD STAR*net* retrospectively identified and longitudinally followed all cases born since January 1, 1982 that were diagnosed with a childhood-onset dystrophinopathy and resided at some time following diagnosis in one of four states: Arizona, Colorado, Iowa, and the western part of New York State. In 2005 and 2008, Georgia and Hawaii, respectively, joined the surveillance system. Since the start of surveillance, all newly diagnosed cases were identified and prospectively followed. Health and vital status information was systematically abstracted through December 2011, the time of their death, or the time they moved out of a participating state's catchment area. Cases identified from September 2011 through December 2011 were followed through December 2012 to ensure a minimum of one year of follow-up. Public health authority was utilized for medical record abstraction for Colorado, Georgia, Iowa, and western New York. For Arizona, institutional review board approval was obtained at the University of Arizona and at individual healthcare facilities where data was collected.

Trained abstractors identified potential cases through review of medical records, located primarily at neuromuscular clinics. Medical record data abstracted were reviewed by a clinical review committee, composed of clinicians who have experience treating dystrophinopathies, and assigned a case status: definite, probable, possible, asymptomatic, or female (17). Definite cases had a positive genetic test for a dystrophin mutation, a positive muscle biopsy, or an X-linked pedigree and an affected family member with a dystrophin mutation or positive muscle biopsy. Probable cases also had an X-linked pedigree consistent

with a dystrophinopathy but did not have confirmatory genetic testing. Possible cases had recorded clinical symptoms related to a dystrophinopathy and elevated creatine kinase. Individuals who met the criteria for definite but did not show any clinical symptoms were defined as asymptomatic. Females who were diagnosed with a dystrophinopathy before age 21 years and who had a positive genetic test for a dystrophin mutation or positive muscle biopsy were also enumerated.

The MD STAR*net* surveillance methodology included medical record abstraction for key data elements associated with function and disease progression. Indicators of mental function included mentions of cognitive function, psychosocial issues, and education service usage. Mentions of pain, muscle cramps, and medication use for pain were used for indicators of sensory function. Indicators for cardiovascular function were created using results from echocardiograms, electrocardiograms, blood pressure measurements, and medication use. Pulmonary function indicators were created using results from forced vital capacity tests and pulmonary device usage (noninvasive positive airway pressure, cough-assist machine, invasive ventilation, oxygen use). Indicators of digestive function included the use of nasogastric tube, having percutaneous endoscopic gastromy, dietary counseling for weight management, and symptoms of poor weight gain. Neuromusculoskelatal and mobility indicators were created using symptoms noted in the record (Gowers' sign, muscle hypotonia, muscle weakness, trouble climbing stairs, abnormal gait, toe walking, trouble walking, ambulation ceased) and mobility devices (orthotics, wheelchair use). If an individual did not have an indicator/symptom noted in their medical record it was assumed they did not have it.

ICF

The ICF was developed by the World Health Organization to provide a standardized method to document functioning and development among children with disabilities, and to incorporate measures of the impact of functioning on personal and social limitations (6). The ICF consists of four components: body function (b), body structures (s), activity limitations and participation restriction (d), and environmental factors (e). Each component is comprised of multiple ‘chapters’ corresponding to finer categories and qualifiers (*e.g.*, none, mild, moderate, severe, and complete) that are used to indicate the degree of disability. The letters b, s, d, and e represent the different components and are followed by a numeric code that starts with the chapter number (one digit), followed by second- (two digits), third- (one-digit), and fourth- (one digit) level codes. For example, the code ‘b28010′ corresponds to a body function (b28010) in Chapter 2 (b2) for second-level category ‘sensation of pain’ (b28010) in a third-level body part (b28010), specific to the fourth-level category ‘pain in head and neck’ (b28010). There are varying levels of specificity for each function and category with some including only second-level categories and others containing all four levels of categories.

Severity of functional disability is indicated by qualifiers added to the end of the designated ICF code (ICF code.qualifier). There are five levels of severity: no problem (ICF.0=affects function 0-4% of the time), mild (ICF.1=affects function 5-24% of the time), moderate (ICF. 2=affects function 25-49% of the time), severe (ICF.3=affects function 50-95% of the time),

and complete (ICF.4=affects function 96-100% of the time). Additional qualifier codes for 'not specific' (ICF.8) and 'not applicable' (ICF.9) are also used.

Procedure for identifying ICF dystrophinopathy functions

A two-staged review process was developed to identify clinically relevant ICF categories. The first stage involved identifying ICF categories from the Body Function domain representative of dystrophinopathy symptomatology. Two medical doctors (authors DM [Physical Medicine and Rehabilitation] and EC [Neurology]) who specialize in the treatment of patients with childhood-onset dystrophinopathies and serve as clinicians on the MD STAR*net* clinical review committee independently identified clinically relevant ICF categories. Consensus on discordance in selected categories was reached via conference between the clinicians, the lead medical abstraction coordinator (CW), and remaining corresponding authors (KC, KJ, PP). The second stage involved linking surveillance data collected by the MD STAR*net* with the selected ICF categories to identify those functions that could be quantified using these surveillance data. Where possible, the linking rules described by Cieza et al (8, 10) were used to link MD STAR*net* surveillance data with the final set of ICF categories.

Analyses

We calculated the proportion of overall agreement for second-level categories within each ICF chapter. Each category was assigned a 0 (not retained) or 1 (retained) by each of our two clinicians. Agreement was calculated as the number of ICF categories with the same assigned value divided by the total number of ICF categories within a chapter. Due to the variation in the number of third- and fourth-level categories across ICF functions, the agreement between clinicians is only described for second-level categories (11). We provide a description of third-and fourth-level categories selected, where applicable. Frequencies and percentages for MD STAR*net* surveillance data elements linked to the retained second-level ICF categories were estimated for cases who were the oldest affected male in a family, classified as definite or probable childhood-onset dystrophinopathy, and resided in Arizona, Colorado, Iowa, Georgia or western New York State (n=703). HI was excluded from analyses due to limited follow-up

Results

ICF function categories

Overall, 24 of 78 possible second-level categories (excluding other and unspecified) were selected (table 1). For mental functions, 5 of 19 categories were retained; agreement between our clinical reviewers was 74%. Only 1 of 12 sensory and pain categories was retained with 100% agreement. None of the speech function categories were retained after conference; initial reviewer agreement was 50%. Out of 9 possible cardiovascular, haematological, immunological and respiratory functions, 6 categories were retained based on 90% agreement. Of the possible digestive, metabolic, and endocrine functions, 7 of 10 categories were retained based on 90% reviewer agreement. None of the 7 second-level categories representing genitourinary and reproductive functions were retained after conference, despite initial reviewer agreement of 86%. Out of 120 possible neuromuscular

and movement functions, 4 second-level categories were retained based on 75% agreement between reviewers. Finally, 1 out of 5 possible categories was selected from the skin and related structures functions with 80% reviewer agreement.

ICF third- and fourth-level categories that provide additional specificity for retained second-level categories are also presented in table 1. Third-level categories of sustaining attention and expression of language were retained for attention and mental functions of language, respectively. A fourth-level category further clarifying compromised language expression, expression of spoken language, was also retained. Third-level categories corresponding to generalized pain and pain in body part with the latter further clarified by fourth-level categories of pain in back, pain in upper limb, pain in lower limb, and pain in joints, were retained. For heart functions, third-level categories retained were heart rate, heart rhythm, and contraction force of ventricular muscles. Indications of compromised blood pressure functions were represented by the third-level categories of increased, decreased, and maintenance of blood pressure. Respiratory rate, rhythm, and depth were retained to further describe compromised respiratory functions. Muscle functions specific to respiration retained included thoracic, diaphragmatic, and accessory muscles. Categories related to exercise tolerance included general physical endurance, aerobic capacity, and fatigability.

The third-level category swallowing was retained to describe compromised ingestion functions, along with fourth-level categories of pharyngeal and esophageal swallowing. The third-level categories of transport of food through stomach and intestines and elimination of feces provided further clarification of compromised digestive and defecation functions, respectively. For general metabolic functions, third-level categories of basal metabolic rate, and metabolism of carbohydrates, protein, and fat were retained. Finally, the third-level categories, balance of minerals and electrolytes, were retained to further describe water, mineral and electrolyte balance functions. All possible third-level categories were retained to describe mobility of joint functions. Similarly, all third-level categories describing muscle power functions were retained. For muscle endurance functions, the third-level endurance categories retained were isolated muscles, muscle groups, and all muscles of the body. Categories describing gait pattern and protective functions of the skin were not retained.

Linking ICF categories and MD STARnet surveillance data

The final step in our study involved linking the selected ICF categories with available clinical data from the MD STAR*net* surveillance database (table 2). The process of linking differed from that recommended by Cieza et al (8, 10) due to the large volume of data elements contained in the MD STAR*net* surveillance database. Rather than identifying concepts associated with each surveillance data element, the inclusion and exclusion criteria of each selected ICF category was used to identify clinical indicators that would be representative of the intended function. Furthermore, the linkage is described only for second-level categories, because the diagnostic reason for the test is not recorded in the MD STAR*net* surveillance database, a step recommended for the prospective linkage of clinical test results to ICF categories (10).

Of the 24 second-level ICF categories retained, we were unable to link 9 (denoted by nc-hc in the table) to any of the MD STAR*net* surveillance data elements that appropriately

captured the intended functions or related exclusions (table 2). To represent intellectual functions, we selected available categorical data elements indicating the presence of autism, cognitive delay, special education or resource room; a diagnosis of attention-deficit hyperactivity disorder was also selected for attention functions. Documentation of autism, behavior problems, or depression were selected to represent global psychosocial functions. The ICF category, mental functions of language, was linked to evidence of speech delay.

The ICF category, sensations of pain, was linked to evidence of intractable pain, muscle pain/cramps, or documentation of pain medication. Linked heart functions included arrhythmia/dysrhythmia, cardiomyopathy, diastolic dysfunction, and tachycardia. High and low blood pressure, as well as documentation of blood pressure medication, were linked to the blood pressure function category. The use of a cough-assist machine or assistive ventilation (invasive or noninvasive), oxygen use, or reduced (<50% predicted) forced vital capacity were used to link to respiratory muscle functions. Reliance on nasogastric or percutaneous endoscopic gastrostomy tubes for nutrition was linked to ingestion functions. Dietary counseling for weight and poor weight gain in early childhood were linked to weight maintenance functions.

Joint mobility was linked to the use of supportive devices, such as knee-ankle-foot orthosis, ankle-foot orthosis, or night splints or documentation of tendon release surgery. There were several surveillance items linked to muscle power functions that were inclusive of symptoms that occur early in life, such as trouble walking or rising from the floor, and symptoms that occur in later stages of disease progression, such as loss of independent ambulation. Expectedly, nearly all cases had documentation of some measure of muscle power function. Finally, gait pattern functions were linked to abnormal gait and toe walking.

Discussion

The objectives of our study included identifying ICF categories describing the clinical presentation of childhood-onset dystrophinopathies and linking these categories to abstracted medical record data from the MD STAR*net*. Seventy-one ICF (24 second-, 41 third- and 7 fourth-level) categories that were descriptive of dystrophinopathies were selected via clinician review and consensus. Of these, we were able to link 15 of the 24 retained second-level categories to data elements abstracted by the MD STAR*net*. The number of selected categories is consistent with the number of categories enumerated from an analysis of ICF studies published through 2012 (92.5 categories for comprehensive core sets; 26 categories for brief core sets) (13).

Our subset of linked ICF categories overlapped with the ICF neuromuscular disease core set expanded by Bos et al (2013), but also showed substantial differences. For mental functions, 6 categories were retained for the neuromuscular disease core set but only 1 of 5 categories from our study overlapped (b140 *Attention Functions*). Additional mental function categories selected for childhood-onset dystrophinopathies included those related to intellectual, global psychosocial, basic cognitive, and language functions. Sensation of pain was retained in both studies; but voice and speech functions were not retained by our study after conference. In addition to the single category of exercise tolerance functions retained

by both studies, we included functions affecting the heart, blood pressure, respiration, respiratory muscle, and exercise tolerance. Two digestion functions (swallowing and defecation) were common across both studies with additional categories (digestion, weight maintenance, metabolism, balancing water, mineral and electrolytes, and the endocrine glands) retained in our study. The overlap between categories representing neuromusculoskeletal and movement related functions was minimal with categories for muscle power and endurance, and gait pattern retained by both studies; mobility of joint functions retained for childhood-onset dystrophinopathies only; and an additional four categories describing muscle tone, voluntary and involuntary movement, and sensations related to muscle movement retained for the neuromuscular disease core set only. The differences in retained ICF categories reinforce the need to identify functions that are disease specific, even though the disease of interest may be included as part of a broader disease class (*e.g.*, dystrophinopathies are neuromuscular diseases), due to variations in clinical presentation and progression of neuromuscular diseases.

Challenges and Limitations of Linking Secondary Data to ICF Categories

In order to promote systematic translation of the ICF into research and clinical settings, rules for assigning ICF categories to existing health measures were developed and the consistency of applying these rules evaluated (8). Updates to the original linking rules were published with the goal of simplifying and clarifying use when linking to different data modalities (*e.g.*, health measure, technical or clinical measures), and a discussion of promoting transparency in the linking process (9, 10). Instead of linking health measures, we attempted to link MD STAR*net* medical record surveillance data to the selected ICF categories for childhood-onset dystrophinopathies. The linking process was challenging due to the use of retrospectively collected data. As described in the linking rules (8, 10, 12), the process of linking ICF categories involves, in part, an *a priori* determination of the purpose of the test. Without knowing the indication of a test, it is difficult to consistently assign a meaningful concept, especially in circumstances were a single procedure may be used to evaluate multiple functions (10, 18, 19). Additionally, exclusion criteria are included to guide assignment of ICF categories. For example, the category ‘b440 – Respiratory functions’ was not linked to surveillance data despite high rates of respiratory dysfunction among dystrophinopathy patients, because the MD STAR*net* did not abstract clinical information describing respiration. Furthermore, the pathophysiology of compromised respiration is muscular in nature, which is listed as an exclusion for describing respiratory function; therefore, surveillance items describing respiratory function were only linked to ‘b445 Respiratory muscle functions’ despite the possibility that respiration was compromised independent of respiratory muscle functioning. Finally, although we were able to identify surveillance items for most ICF categories, we were unable to reliably assign disability qualifiers to these functions since a given test result may not accurately reflect compromised health functioning (19). Similarly, we were unable to identify categories from the activity and participation sections due to lack of such information collected in the abstraction database. From these limitations, recommendations for future linkage studies of health information to ICF categories would include clear goals of collecting functional health outcomes and rehabilitation therapies associated with these outcomes. However, until the

ICF is implemented in clinical practice in a standardized manner (20, 21), the retrospective linkage of medical record abstraction data to ICF categories may prove difficult (22).

Another challenge to identifying a comprehensive set of ICF categories is the degree of multi-system involvement in dystrophinopathies, which also have variable presentation and course. Thus, the number of ICF categories needed to describe impairment sufficiently among those diagnosed may increase exponentially. For example, categories selected from *Chapter 1: Mental Functions* included intellectual, global psychosocial and attention functions. These categories were linked to surveillance items that collected information about diagnoses of autism (intellectual and psychosocial function) and attention-deficit hyperactivity disorder (attention). ICF core sets are being developed for each of these diagnoses with preliminary reports of 28 and 30 second-level function categories, respectively (23, 24). Another morbidity for which an ICF core set has been developed is cardiopulmonary disease, which consists of 20 second-level functions (25). Of these, only 11 overlap with categories selected in the current paper. Thus, the granularity needed to describe affected functions accurately and the severity of functional impairment requires further consideration.

Implications for Rehabilitation

The ICF may supplement the rehabilitation process as a standardized approach to evaluation of primary problems and degree of impairment resulting from these problems. Assessment of relevant functions and their impact can be achieved by linking patient reports of problems provided during a health interview, or incorporating an ICF checklist comprised of selected ICF core set functions, environmental factors affecting function, and participation and limitations of activity (26). A standardized dystrophinopathy ICF-based documentation form can be created from the ICF Research Branch website (https://www.icf-research-branch.org/component/content/article/120-external-links/456-icf-based-documentation-form.html) for use as a screening tool by rehabilitation professionals and for patient goal setting when developing rehabilitation plans. The ICF checklist may require supplementation with patient reports of perceived functional health. These reports could be obtained from existing (27) or newly developed ICF-based measures of functional health (28-30). Results from the linking of health information to ICF categories, use of ICF checklists, or supplemental health measures can then be used to design and evaluate progress in rehabilitation or as a basis for referral to a specialist for further targeted evaluation and rehabilitation, e.g., neuropsychologist for evaluation of attentional issues.

Next Steps

This paper serves as a starting point from which to develop ICF-based assessments of functioning for those diagnosed with childhood-onset dystrophinopathies. In addition to standardizing data collection for research studies, a brief ICF core set specific to dystrophinopathies can contribute to the development and monitoring of rehabilitation plans to increase participation at varying degrees of functional health (4, 26, 31). Although our study demonstrated overlap with existing ICF core sets that are inclusive of neuromuscular diseases in general, we also identified ICF categories specific to childhood-onset dystrophinopathies that may require specialized interventions and rehabilitation not globally

applicable to all neuromuscular diseases. Our findings support formal development of an ICF core set for childhood-onset dystrophinopathies, using methods approved by the ICF Research Branch and described by Selb et al (11), and incorporation of standardized ICF-based approaches to evaluate functional health, as well as environment and activities and participation, (26, 28, 32) with the goal of developing individualized rehabilitation care plans.

Acknowledgments

The authors thank members of the MD STAR*net* data sharing committee for their manuscript review and input. We also acknowledge the efforts of all the study coordinators, abstractors, and data managers in data collection and cleaning. All the authors contributed to the conception, design, analysis, interpretation of data, and final approval of the manuscript.

References

1. Flanigan KM. Duchenne and Becker muscular dystrophies. Neurol Clin. 2014; 32(3):671–88. viii. [PubMed: 25037084]
2. Bushby K, Finkel R, Birnkrant DJ, Case LE, Clemens PR, Cripe L, et al. Diagnosis and management of Duchenne muscular dystrophy, part 1: diagnosis, and pharmacological and psychosocial management. Lancet neurology. 2010; 9(1):77–93. [PubMed: 19945913]
3. Bos I, Stallinga HA, Middel B, Kuks JB, Wynia K. Validation of the ICF core set for neuromuscular diseases. Eur J Phys Rehabil Med. 2013; 49(2):179–87. [PubMed: 23172408]
4. Stucki G. International Classification of Functioning, Disability, and Health (ICF): a promising framework and classification for rehabilitation medicine. Am J Phys Med Rehabil. 2005; 84(10): 733–40. [PubMed: 16205428]
5. Stucki G, Ewert T, Cieza A. Value and application of the ICF in rehabilitation medicine. Disabil Rehabil. 2002; 24(17):932–8. [PubMed: 12523361]
6. World Health Organization (WHO). International Classification of Functioning, Disability, and Health: Children & Youth Version. 2001
7. Stucki G, Cieza A, Melvin J. The International Classification of Functioning, Disability and Health (ICF): a unifying model for the conceptual description of the rehabilitation strategy. J Rehabil Med. 2007; 39(4):279–85. [PubMed: 17468799]
8. Cieza A, Brockow T, Ewert T, Amman E, Kollerits B, Chatterji S, et al. Linking health-status measurements to the international classification of functioning, disability and health. J Rehabil Med. 2002; 34(5):205–10. [PubMed: 12392234]
9. Cieza A, Fayed N, Bickenbach J, Prodinger B. Refinements of the ICF Linking Rules to strengthen their potential for establishing comparability of health information. Disabil Rehabil. 2016:1–10.
10. Cieza A, Geyh S, Chatterji S, Kostanjsek N, Ustun B, Stucki G. ICF linking rules: an update based on lessons learned. J Rehabil Med. 2005; 37(4):212–8. [PubMed: 16024476]
11. Selb M, Escorpizo R, Kostanjsek N, Stucki G, Ustun B, Cieza A. A guide on how to develop an international classification of functioning, disability and health core set. Eur J Phys Rehabil Med. 2014
12. Cieza A, Ewert T, Ustun TB, Chatterji S, Kostanjsek N, Stucki G. Development of ICF Core Sets for patients with chronic conditions. J Rehabil Med. 2004; (44 Suppl):9–11. [PubMed: 15370742]
13. Yen TH, Liou TH, Chang KH, Wu NN, Chou LC, Chen HC. Systematic review of ICF core set from 2001 to 2012. Disabil Rehabil. 2014; 36(3):177–84. [PubMed: 23651126]
14. Wynia K, Middel B, Van Dijk JP, De Ruiter H, Lok W, Ha De Keyser J, et al. Broadening the scope on health problems among the chronically neurologically ill with the International Classification of Functioning (ICF). Disabil Rehabil. 2006; 28(23):1445–54. [PubMed: 17166807]
15. Romitti PA, Zhu Y, Puzhankara S, James KA, Nabukera SK, Zamba GK, et al. Prevalence of Duchenne and Becker muscular dystrophies in the United States. Pediatrics. 2015; 135(3):513–21. [PubMed: 25687144]

16. Miller LA, Romitti PA, Cunniff C, Druschel C, Mathews KD, Meaney FJ, et al. The muscular Dystrophy Surveillance Tracking and Research Network (MD STARnet): surveillance methodology. Birth Defects Res A Clin Mol Teratol. 2006; 76(11):793–7. [PubMed: 17036307]
17. Mathews KD, Cunniff C, Kantamneni JR, Ciafaloni E, Miller T, Matthews D, et al. Muscular Dystrophy Surveillance Tracking and Research Network (MD STARnet): case definition in surveillance for childhood-onset Duchenne/Becker muscular dystrophy. Journal of child neurology. 2010; 25(9):1098–102. [PubMed: 20817884]
18. Stucki G, Cieza A. The International Classification of Functioning, Disability and Health (ICF) in physical and rehabilitation medicine. Eur J Phys Rehabil Med. 2008; 44(3):299–302. [PubMed: 18762739]
19. Reed G, Lux JB, Bufka LF, Trask C, Peterson DB, Stark S, et al. Operationalizing the International Classification of Functioning, Disability and Health in Clinical Settings. Rehabilitation Psychology. 2005; 50(2):122–31.
20. Stucki G, Zampolini M, Juocevicius A, Negrini S, Christodoulou N. Practice, science and governance in interaction: European effort for the system-wide implementation of the International Classification of Functioning, Disability and Health (ICF) in Physical and Rehabilitation Medicine. Eur J Phys Rehabil Med. 2016
21. Kohler F, Selb M, Escorpizo R, Kostanjsek N, Stucki G, Riberto M. Towards the joint use of ICD and ICF: a call for contribution. J Rehabil Med. 2012; 44(10):805–10. [PubMed: 22990383]
22. Han KY, Kim HJ, Bang HJ. Feasibility of Applying the Extended ICF Core Set for Stroke to Clinical Settings in Rehabilitation: A Preliminary Study. Ann Rehabil Med. 2015; 39(1):56–65. [PubMed: 25750873]
23. de Schipper E, Lundequist A, Coghill D, de Vries PJ, Granlund M, Holtmann M, et al. Ability and Disability in Autism Spectrum Disorder: A Systematic Literature Review Employing the International Classification of Functioning, Disability and Health-Children and Youth Version. Autism research : official journal of the International Society for Autism Research. 2015; 8(6): 782–94. [PubMed: 25820780]
24. de Schipper E, Mahdi S, Coghill D, de Vries PJ, Gau SS, Granlund M, et al. Towards an ICF core set for ADHD: a worldwide expert survey on ability and disability. European child & adolescent psychiatry. 2015; 24(12):1509–21. [PubMed: 26428005]
25. Boldt C, Grill E, Wildner M, Portenier L, Wilke S, Stucki G, et al. ICF Core Set for patients with cardiopulmonary conditions in the acute hospital. Disabil Rehabil. 2005; 27(7-8):375–80. [PubMed: 16040539]
26. Rauch A, Cieza A, Stucki G. How to apply the International Classification of Functioning, Disability and Health (ICF) for rehabilitation management in clinical practice. Eur J Phys Rehabil Med. 2008; 44(3):329–42. [PubMed: 18762742]
27. Cieza A, Stucki G. Content comparison of health-related quality of life (HRQOL) instruments based on the international classification of functioning, disability and health (ICF). Qual Life Res. 2005; 14(5):1225–37. [PubMed: 16047499]
28. Bos I, Kuks JB, Wynia K. Development and testing psychometric properties of an ICF-based health measure: The Neuromuscular Disease Impact Profile. J Rehabil Med. 2015; 47(5):445–53. [PubMed: 25679115]
29. Cerniauskaite M, Quintas R, Boldt C, Raggi A, Cieza A, Bickenbach JE, et al. Systematic literature review on ICF from 2001 to 2009: its use, implementation and operationalisation. Disabil Rehabil. 2011; 33(4):281–309. [PubMed: 21073361]
30. Farin E, Fleitz A, Frey C. Psychometric properties of an International Classification of Functioning, Disability and Health (ICF)-oriented, adaptive questionnaire for the assessment of mobility, self-care and domestic life. J Rehabil Med. 2007; 39(7):537–46. [PubMed: 17724553]
31. Bendixen RM, Senesac C, Lott DJ, Vandenborne K. Participation and quality of life in children with Duchenne muscular dystrophy using the International Classification of Functioning, Disability, and Health. Health Qual Life Outcomes. 2012; 10:43. [PubMed: 22545870]
32. Steiner WA, Ryser L, Huber E, Uebelhart D, Aeschlimann A, Stucki G. Use of the ICF model as a clinical problem-solving tool in physical therapy and rehabilitation medicine. Phys Ther. 2002; 82(11):1098–107. [PubMed: 12405874]

Table 1

International Classification of Functioning and Disability (ICF) dystrophinopathy body functions

ICF categories			
2nd level	3rd level	4th level	ICF Functions
Chapter 1: Mental functions			
b117			**Intellectual functions**
b122			**Global psychosocial functions**
b140			**Attention functions**
	b1400		Sustaining attention
b163			**Basic cognitive functions**
b167			**Mental functions of language**
	b1671		Expression of language
		b16710	Expression of spoken language
Chapter 2: Sensory Functions and Pain			
b280			**Sensation of pain**
	b2800		Generalized pain
	b2801		Pain in body part
		b28013	Pain in back
		b28014	Pain in upper limb
		b28015	Pain in lower limb
		b28016	Pain in joints
Chapter 3: Voice and Speech Functions			
Chapter 4: Functions of the Cardiovascular, Hematological, Immunological and Respiratory Systems			
b410			**Heart functions**
	b4100		Heart rate
	b4101		Heart rhythm
	b4102		Contraction force of ventricular muscles
b420			**Blood pressure functions**
	b4200		Increased blood pressure
	b4201		Decreased blood pressure
	b4202		Maintenance of blood pressure
b440			**Respiratory functions**
	b4400		Respiration rate
	b4401		Respiratory rhythm
	b4402		Depth of respiration
b445			**Respiratory muscle functions**
	b4450		Functions of the thoracic respiratory muscles
	b4451		Functions of the diaphragm
	b4452		Functions of accessory respiratory muscles

ICF categories			
2nd level	3rd level	4th level	ICF Functions
b450			**Additional respiratory functions**
b455			**Exercise tolerance functions**
	b4550		General physical endurance
	b4551		Aerobic capacity
	b4552		Fatigability
Chapter 5: Functions of the Digestive, Metabolic, and Endocrine Systems			
b510			**Ingestion functions**
	b5105		Swallowing
		b51051	Pharyngeal swallowing
		b51052	Esophageal swallowing
b515			**Digestive functions**
	b5150		Transport of food through stomach and intestines
b525			**Defecation functions**
	b5250		Elimination of feces
b530			**Weight maintenance functions**
b540			**General metabolic functions**
	b5400		Basal metabolic rate
	b5401		Carbohydrate metabolism
	b5402		Protein metabolism
	b5403		Fat metabolism
b545			**Water, mineral and electrolyte balance functions**
	b5451		Mineral balance
	b5452		Electrolyte balance
b555			**Endocrine gland functions**
Chapter 6: Genitourinary and reproductive functions			
Chapter 7: Neuromusculoskeletal and Movement Related Functions			
b710			**Mobility of joint functions**
	b7100		Mobility of single joint
	b7101		Mobility of several joints
	b7102		Mobility of joints generalized
b730			**Muscle power functions**
	b7300		Power of isolated muscles and muscle groups
	b7301		Power of muscles of one limb
	b7302		Power of muscles of one side of the body
	b7303		Power of muscles in lower half of the body
	b7304		Power of muscles of all limbs
	b7305		Power of muscles of the trunk
	b7306		Power of all muscles of the body
b740			**Muscle endurance functions**

ICF categories			
2nd level	**3rd level**	**4th level**	**ICF Functions**
	b7400		Endurance of isolated muscles
	b7401		Endurance of muscle groups
	b7402		Endurance of all muscles of the body
b770			**Gait pattern functions**
Chapter 8: Functions of the Skin and Related Structures			
b810			**Protective functions of the skin**

Bolded text identifies second-level function categories.

Table 2

Linking 2nd level International Classification of Functioning and Disability (ICF) categories with Surveillance Data from the Muscular Dystrophy Tracking, Surveillance and Research (MD STARnet) (n=703)[1]

ICF categories		Surveillance Data Elements	*n*	% of total sample[2]
Chapter 1: Mental functions				
b117	**Intellectual functions**	**Any[3] linked intellectual function**	**356**	**51%**
		Autism	29	4%
		Cognitive delay	163	23%
		Special education	246	35%
		Resource room	39	6%
b122	**Global psychosocial functions**	**Any linked global psychosocial function**	**283**	**40%**
		Autism	29	4%
		Behavior problems	198	28%
		Depression	130	18%
b140	**Attention functions**	Attention-deficit hyperactivity disorder	**139**	**20%**
b163	**Basic cognitive functions**	***nc-hc***		
b167	**Mental functions of language**	**Speech delay**	**254**	**36%**
Chapter 2: Sensory Functions and Pain				
b280	**Sensation of pain**	**Any linked sensation of pain**	**344**	**49%**
		Intractable pain	42	6%
		Muscle pain/cramps	202	29%
		Pain medication use	195	28%
Chapter 4: Functions of the Cardiovascular, Hematological, Immunological and Respiratory Systems				
b410	**Heart functions**	**Any linked heart function**	**295**	**42%**
		Arrhythmia/Dysrhythmia	51	7%
		Cardiomyopathy	212	30%
		Diastolic dysfunction	69	10%
		Tachycardia	108	15%
b420	**Blood pressure functions**	**Any linked blood pressure function**	**56**	**8%**
		High blood pressure	39	6%
		Low blood pressure	23	3%
		Blood pressure medication	39	6%
b440	**Respiratory functions**	***nc-hc***		
b445	**Respiratory muscle functions**	**Any linked respiratory muscle function**	**275**	**39%**
		Cough-assist machine	48	7%
		Forced vital capacity (<50%)	213	30%
		Noninvasive positive (bilevel/continuous) airway pressure	175	25%
		Invasive ventilation	53	8%

ICF categories		Surveillance Data Elements	*n*	% of total sample[2]
		Oxygen use	66	9%
b450	**Additional respiratory functions**	***nc-hc***		
b455	**Exercise tolerance functions**	***nc-hc***		
Chapter 5: Functions of the Digestive, Metabolic, and Endocrine Systems				
b510	**Ingestion functions**	**Any linked ingestion function**	**18**	**3%**
		Nasogastric tube	4	<1%
		Percutaneous endoscopic gastrostomy	14	2%
b515	**Digestive functions**	***nc-hc***		
b525	**Defecation functions**	***nc-hc***		
b530	**Weight maintenance functions**	**Any linked weight maintenance function**	**333**	**47%**
		Dietary counseling for weight management	312	44%
		Poor weight gain	44	6%
b540	**General metabolic functions**	***nc-hc***		
b545	**Water, mineral and electrolyte balance functions**	***nc-hc***		
b555	**Endocrine gland functions**	***nc-hc***		
Chapter 7: Neuromusculoskeletal and Movement Related Functions				
b710	**Mobility of joint functions**	**Any linked mobility of joint function**	**482**	**66%**
		KAFO/AFO/night splint	464	66%
		Tendon release	150	32%
b730	**Muscle power functions**	**Any linked muscle power function**	**697**	**99%**
		Gowers' sign	633	90%
		Inability to keep up with peers	316	45%
		Independent ambulation ceased	399	57%
		Muscle hypotonia	335	48%
		Muscle weakness	605	86%
		Transfer assistance	334	48%
		Trouble climbing stairs	515	73%
		Trouble walking	570	81%
		Wheelchair use	513	73%
b740	**Muscle endurance functions**	***nc-hc***		
b770	**Gait pattern functions**	**Any linked gait pattern function**	**602**	**86%**
		Abnormal gait	566	81%
		Toe walking	339	48%
Chapter 8: Functions of the Skin and Related Structures				
b810	**Protective functions of the skin**	***nc-hc***		

nc-hc = not covered health condition. KAFO = knee-ankle-foot orthosis. AFO = ankle-foot orthosis. Bolded text identifies second-level function categories.

[1]The oldest affected male in a family, classified as definite or probable childhood-onset dystrophinopathy, and resident of Arizona, Colorado, Iowa, Georgia or western New York State.

[2]The denominator for all linked surveillance data elements is the total sample size (n=703).

[3]'Any' refers to the presence of at least one of the linked surveillance data elements within a function category.