Implementation of Duchenne Muscular Dystrophy Care Considerations

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abstract

BACKGROUND: Duchenne muscular dystrophy (DMD) is an X-linked disorder characterized by progressive muscle weakness and multisystem involvement. Recent advances in management of individuals with DMD have prolonged survival. Lack of standardized care spurred an international collaboration to develop consensus-based care considerations for diagnosis and management. In this study, we evaluate adherence to considerations at selected sites.

METHODS: We collaborated with the Muscular Dystrophy Surveillance, Tracking, and Research Network. Our sample included males with DMD and Becker muscular dystrophy <21 years as of December 31, 2010, with 1 health care encounter on or after January 1, 2012. We collected data from medical records on encounters occurring January 1, 2012, through December 31, 2014. Adherence was determined when frequency of visits or assessments were at or above recommendations for selected care considerations.

RESULTS: Our analytic sample included 299 individuals, 7% of whom (20/299) were classified as childhood-onset Becker muscular dystrophy. Adherence for neuromuscular and respiratory clinician visits was 65% for the cohort; neuromuscular assessments and corticosteroid side effect monitoring measures ranged from 16% to 68%. Adherence was 83% for forced vital capacity and ≤58% for other respiratory diagnostics. Cardiologist assessments and echocardiograms were found for at least 84%. Transition planning for education or health care was documented for 31% of eligible males.

CONCLUSIONS: Medical records data were used to identify areas in which practice aligns with the care considerations. However, there remains inconsistency across domains and insufficiency in critical areas. More research is needed to explain this variability and identify reliable methods to measure outcomes.

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DOI: https://doi.org/10.1542/peds.2017-4006

Accepted for publication Apr 17, 2018

WHAT'S KNOWN ON THIS SUBJECT: There is marked variability reported in studies in which authors address implementation of care considerations globally. Although the data in these studies include large cohorts from national registries, the generalizability of the data has been called into question.

WHAT THIS STUDY ADDS: Our data are derived from population-based medical record review, which limits bias attributed to registries. This is the most complete reporting of clinical measures related to the implementation of the Duchenne muscular dystrophy care considerations to date and provides US baseline data.

To cite: Andrews JG, Conway K, Westfield C, et al. Implementation of Duchenne Muscular Dystrophy Care Considerations. *Pediatrics*. 2018;142(1):e20174006

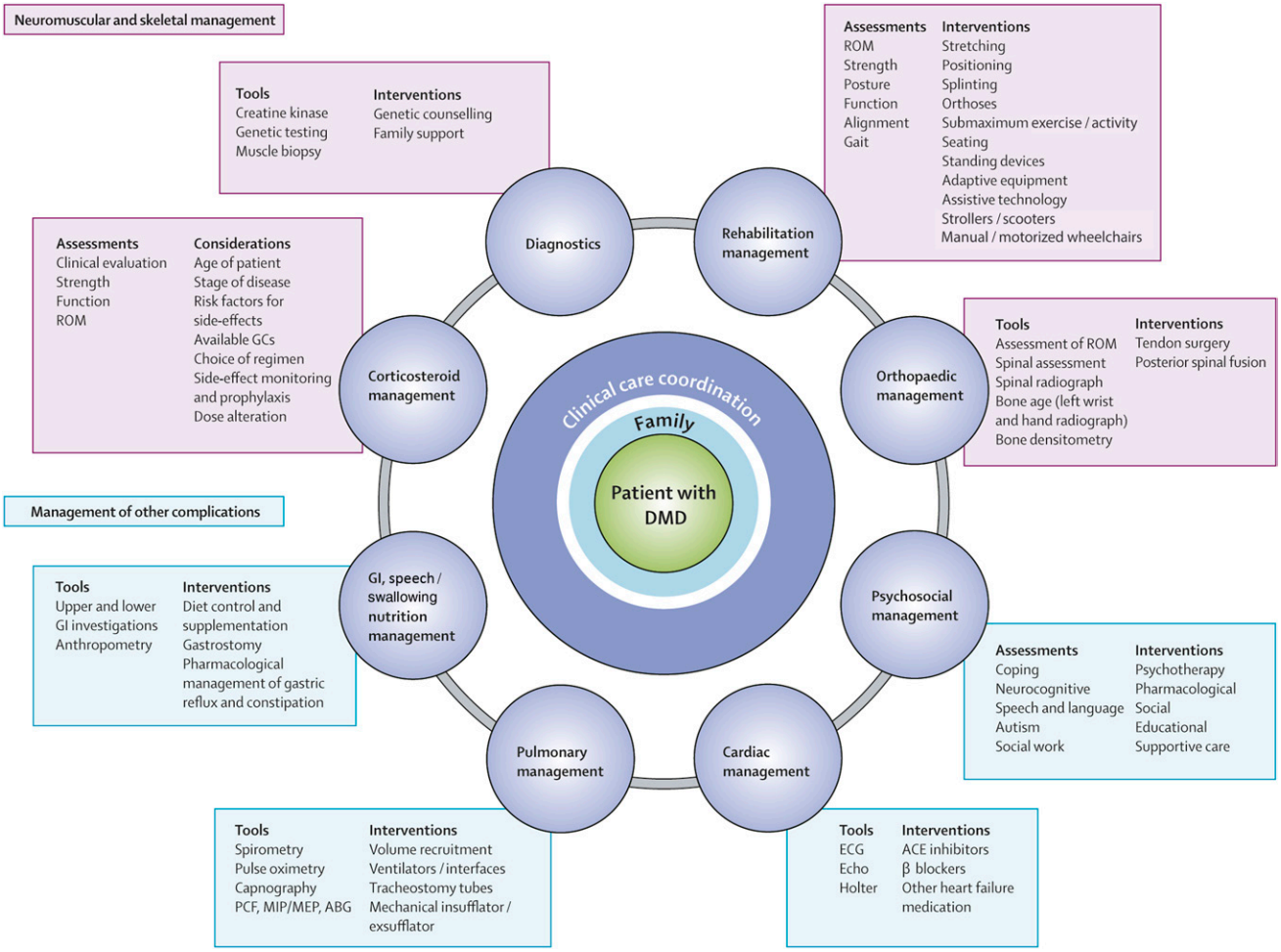

FIGURE 1

Interdisciplinary management of DMD as described by the Care Coordination Working Group. ABG, arterial blood gas; ACE, angiotensinconverting enzyme; DMD, Duchenne muscular dystrophy; Echo, echocardiogram; ECG, electrocardiogram; GC, glucocorticoids; GI, gastrointestina; MEP, maximum expiratory pressure; MIP, maximum inspiratory pressure; PCF, peak cough flow; ROM, range of motion. (Reprinted with permission by Bushby K, Finkel R, Birnkrant DJ, et al; DMD Care Considerations Working Group. Diagnosis and management of Duchenne muscular dystrophy, part 1: diagnosis, and pharmacological and psychosocial management. *Lancet Neurol.* 2010;9(1):77–93 and Bushby K, Finkel R, Birnkrant DJ, et al; DMD Care Considerations Working Group. Diagnosis and management of Duchenne muscular dystrophy, part 2: implementation of multidisciplinary care. *Lancet Neurol.* 2010;9(2):177–189.)

Duchenne muscular dystrophy (DMD) is a childhood-onset, progressive disorder resulting from mutations in the dystrophin gene. DMD is the most common form of childhood muscular dystrophy[1] affecting 1.02 per 10 000 males between 5 and 24 years of age in the United States and 0.478 per 10 000 worldwide.[2,3] Historically, individuals with DMD lost ambulation before 13 years of age and did not survive beyond the second decade.[4] Changes in clinical care to preserve cardiac, pulmonary, and skeletal muscle function[5–8] have changed the natural history, prolonged independent ambulation, and extended survival beyond the second decade among individuals who receive recommended treatments.[8–10]

Lack of standardized management strategies for individuals with DMD spurred an international collaboration to develop consensus-based care considerations for the diagnosis and management of DMD. The effort was funded and coordinated by the Centers for Disease Control and Prevention (CDC) in the United States. Working with a steering committee and panels that included representatives from the European Union or the United Kingdom, recommendations were developed by using modified Rand methodology.[11,12] The panels addressed timing and indication for the use of tests and interventions across a range of clinical areas. In Fig 1, we demonstrate the scope of the recommendations.

In research to date, authors evaluating adherence to the care considerations have yielded conflicting results using registry data with their inherent biases, cross-sectional designs, or limited time periods.[13–16] The purpose of our

study is to evaluate the adherence to care considerations by using medical record abstraction over a 3-year time period in a population-based sample across multiple locations in the United States.

METHODS

We capitalized on the existing surveillance framework of the Muscular Dystrophy Surveillance, Tracking, and Research Network (MD STAR*net*), which was established in 2002 by the CDC to document care and services received by individuals with childhood-onset DMD and Becker muscular dystrophy (BMD) and examine the impact of these services on survival, comorbidities, and quality of life.[17] A cross-sectional cohort of previously screened cases with confirmed DMD and BMD from the MD STAR*net* was selected.[18]

Data elements from the validated MD STAR*net* surveillance abstraction tool were retained, and additional elements were developed to allow evaluation of adherence to care considerations that could be operationalized by using abstraction. Visit rates were based on available data. No adjustments were performed to account for missing data.

The cohort included individuals who had a confirmed diagnosis of DMD or BMD, were ≤21 years of age as of December 31, 2010, were male, and had ≥1 health care encounter on or after January 1, 2012. Data for this project were abstracted from medical records of all clinical visits from January 1, 2012, through December 31, 2014, at facilities located in 1 of 4 MD STAR*net* surveillance regions (Arizona, Colorado, Iowa, western New York) by abstractors with a minimum of 5 years' experience. An electronic database, manuals, and quality control documentation allowed for systematic decisions in the field. In addition, abstractors underwent detailed training for this project. They were required to pass initial interrater reliabilities (IRRs) of 90% in each data tab compared with a gold standard. IRRs with low reliability received subsequent training, testing, and manual revisions. A final IRR was conducted 1 month before the abstraction start date. Follow-up IRR occurred 1 month after field abstraction to ensure systematic abstraction across all raters for actual records and to determine the degree to which information was available in the records.

The MD STAR*net* database includes individuals with both DMD and BMD. Individuals with BMD, as classified by MD STAR*net*, comprised 7% of the total cohort for this analysis. Thus, no distinction for diagnosis was made with regards to care consideration adherence. The study was approved by local institutional review boards or state public health authority. Data collection began in 2015.

Clinical Classification of Variables

Care considerations recommend frequency of visits to providers, timing of assessments, and timing and management of pharmacologic, assistive, or therapeutic interventions.[11,12] Many of the recommendations refer to age (<10 years and ≥10 years), functional status (ambulatory or nonambulatory), or interventions received (actively using corticosteroids or no use or use discontinued). A subset of recommendations refers to specific clinical procedures, such as frequency of lumbar radiographs (curvature <20° or ≥20°) and use of noninvasive ventilation. We created grouping variables to classify individuals into mutually exclusive static groups within these criteria on the basis of the individual's status on or before January 1, 2012. Although some individuals in the analytic sample may change status during the study period (eg, became nonambulatory), it simplified the methodology to retain mutually exclusive groups of cases for the entire study period.

Outcome (Visit) Measures

The calculations for recommended visits, assessments, or interventions (hereafter referred to as "visit"), included several calculations. For each visit, we counted the total number of records and calculated a total duration in years of follow-up between the date of the first and last documented record for each case. These data were then used to calculate an estimated number of visits per year (annual visit rate) by dividing the duration of their follow-up period for that visit by the total number of records documented for the visit. Individuals with 1 record for a visit were assigned a visit duration of 0. Individuals with 0 records for the visit were assigned an annual visit rate of 0.

Implementation of a specific care consideration (percent met) was determined by comparing the annual visit rate to the recommended frequency for that visit. For example, the recommended frequency of assessments by a neuromuscular specialist is 2 times per year; therefore, individuals with annual visit rates ≥2.00 met or exceeded the recommendation, whereas those <2.00 did not. Lastly, we reported if ≥1 record (percent any visit) occurred during the 3-year follow-up period.

Statistical Analysis

Means, medians, and confidence intervals (95% CIs) for the annual visit rate and each component (*n*, count, duration) are reported. Given the diversity of care considerations measures, elements included in each visit are defined as table footnotes. Results and their corresponding tables are discussed in the order discussed in the care considerations, part 1, Fig 2 in Bushby et al.[11]

TABLE 1 Baseline Demographics, Stratification Variables, and Clinical Status as of January 1, 2012, for All Individuals in the Cohort (N = 299 males)

	n (%)
Demographics	
Site	
Arizona	83 (28)
Colorado	106 (36)
Iowa	62 (21)
New York	48 (16)
Race and/or ethnicity	
Non-Hispanic white	183 (61)
Hispanic, any race	71 (24)
Non-Hispanic African American	8 (3)
Other	14 (5)
Unknown	23 (7)
BMD	20 (7)
Clinical classification variables[a]	
Age, y	
<10	98 (33)
≥10	201 (67)
Ambulation status[b]	
Ambulatory	143 (48)
Nonambulatory	151 (51)
Ambulation status unknown	5 (2)
Corticosteroid status[c]	
Not using corticosteroids	157 (53)
Using corticosteroids	139 (46)
Corticosteroid status unknown	3 (1)
Clinical status	
Scoliosis surgery[d]	86 (28)
Noninvasive ventilation[e]	76 (25)
Tracheostomy[f]	5 (2)
Cardiomyopathy[g]	91 (30)

[a] Clinical classification variables were used to analyze recommendations that were qualified by ambulation status, corticosteroid use, or age.
[b] Ambulatory = any capacity for walking; nonambulatory = medical record indicated ambulation ceased or full-time wheel chair use; unknown = no definitive ambulation data were available.
[c] Not using corticosteroids = never used corticosteroids or ceased using corticosteroids before baseline; using corticosteroids = used corticosteroids for a minimum of 1 d with no evidence of having terminated at baseline; corticosteroids status unknown = no definitive data were available.
[d] Scoliosis surgery only included those for whom surgery was documented as completed.
[e] Noninvasive ventilation included all individuals who were advised, prescribed, or actively using a bilevel positive airway pressure machine, continuous positive airway pressure machine, or mechanical ventilator with a mask or nasal piece or sip (mouthpiece).
[f] Tracheostomy included those individuals for whom the procedure was documented as completed.
[g] Cardiomyopathy = shortening fraction was <28%, ejection fraction was <55%, or a calculated shortening fraction using left ventricle measurements was <28%.

RESULTS

Demographics and clinical characteristics of the cohort are reported in Table 1 and include 299 individuals from Arizona (28%), Colorado (36%), Iowa (21%), and western New York (16%). Individuals were predominantly non-Hispanic white, equally distributed across ambulation groups and corticosteroid status, and primarily ≥10 years at baseline (67%). Clinically, 30% or less had cardiomyopathy, tracheostomy placement, noninvasive ventilation, or undergone scoliosis surgery as of January 1, 2012. We did not evaluate genetic testing or counseling because most individuals included in this data set were diagnosed before 2010; therefore, the proportion of individuals who were offered services after 2010 would be a minute subset. Overall, >75% of individuals visited a neuromuscular, cardiology, pulmonology, or physical therapy specialist at least once during the follow-up surveillance period; visits to other types of specialists ranged from 2% to 69% (see Table 2). In Tables 3–8, we report the proportion of individuals meeting the care considerations for neuromuscular management, corticosteroid management, psychosocial and neurocognitive management, orthopedic management, pulmonary management, and cardiac management, respectively.

Briefly, 64% of all individuals met the semiannual neuromuscular visit recommendation and lower extremity range of motion measurement. Functional timed testing occurred in <50% of the cohort, and the proportion of users of durable equipment was variable (data not shown). The most common equipment used by nearly 50% of the cohort included fixed ankle-foot orthoses and motorized wheelchairs. One hundred and thirty-nine (46.4%) individuals in the cohort were on corticosteroids, and monitoring of side effects (eg, weight, height, blood pressure) were collected at the recommended frequency for nearly two-thirds of those individuals (63%, 57%, and 63%, respectively). Sixty-four percent of the 212 school-aged individuals had documentation of an individualized education or 504 plan for school. Formal transition planning for education or health care was identified for 31% when restricted to individuals aged 12 to 18 years (n = 123). Observational monitoring for scoliosis was documented for 45% of individuals who were ambulatory. Among those who were nonambulatory, 75% with curves <20°C were assessed by lumbar radiograph annually, whereas <20% of those with curves ≥20°C were assessed twice per year. Eighty-one percent (81%) of the cohort (both ambulatory and nonambulatory) received ≥1 respiratory clinical consultation, but less than two-thirds met the recommended frequency. Among those ≥10 years, cardiology

consultations occurred twice a year for 87%. Echocardiograms revealed high completion rates, with >84% of younger individuals and 90% of older individuals receiving the recommended assessments.

TABLE 2 Types of Specialist Visited at Least Once Between January 1, 2012, and December 31, 2014, for the Cohort ($N = 299$)

Specialist	*n* With Visit	Any Visit, %
Neuromuscular and/or neurology specialist	291	97
Cardiologist	244	82
Pulmonologist	243	81
Physical therapist	237	79
Nutritionist and/or dietitian	205	69
Social worker	195	65
Orthopedist	120	40
Rehabilitation medicine and/or physical medicine and rehabilitation specialist	112	37
Neuropsychologist	92	31
Nurse practitioner and/or nurse	74	25
Endocrinologist	60	20
Occupational therapist	40	13
Ophthalmologist	34	11
Gastroenterologist	32	11
Behavioral health specialist	24	8
Speech therapist	7	2

DISCUSSION

Overall, in our data, there are several areas in which practice aligns well with the care considerations, but there are several areas in which implementation is lacking. Ninety-seven percent of our cohort had ≥1 visit with a neuromuscular specialist during the study period, and 64% met the semiannual follow-up and monitoring recommendations. Of note, the frequency of neuromuscular assessments was consistent with that of other areas typically managed by the neuromuscular specialist, including corticosteroid monitoring. Respiratory and physical therapy assessments appear to follow the same patterns; however, performance of related measurements in these

TABLE 3 Neuromuscular Management Between January 1, 2012, and December 31, 2014, for Individuals in the Cohort ($N = 299$)

Visit Outcome Measure	*N* or *n*	Any Visit[a], %	Care Considerations		Annual Visit Rate				Cases With ≥2 Visits		
					Mean	Median	95% CI			Mean	
			Recommend[b], per y	Met[c], %			LL	UL	*n*	Count[d]	Duration[e]
Neuromuscular clinical assessment[f]	299	97	2×	64	2.18	2.18	2.03	2.30	291	4.58	2.10
Physical therapy assessment	299	79	2×	50	2.08	2.00	1.78	2.37	237	4.40	1.93
Range of motion[g]											
Lower extremity											
All individuals	299	93	2×	64	2.32	2.22	2.08	2.56	279	4.56	2.00
Ambulatory[h]	143	95	2×	67	2.40	2.31	2.05	2.74	136	4.85	2.05
Nonambulatory[h]	151	93	2×	62	2.29	2·18	1.95	2.63	140	4.31	1.96
Upper extremity											
All individuals	299	88	2×	50	1.89	2.00	1.75	2.03	262	3.81	1.83
Nonambulatory[h]	151	92	2×	58	2.04	2.12	1.85	2.23	139	4.04	1.89
Timed lying to standing											
Ambulatory individuals[h]	143	76	2×	48	1.90	2.00	1.52	2.28	108	3.65	1.69
Timed walk 30 ft											
Ambulatory individuals[h]	143	55	2×	34	1.25	1.00	1.02	1.47	79	3.63	1.69
Motor function assessment scales[i]											
All individuals	299	33	2×	23	0.78	0.00	0.64	0.92	99	3.37	1.53
Ambulatory[h]	143	36	2×	24	0.77	0.00	0.59	0.96	51	4.18	1.92
Nonambulatory[h]	151	32	2×	23	0.82	0.00	0.60	1.03	48	2.52	1.12

LL, lower limit; UL, upper limit. Adapted from Emery AEH. Population frequencies of inherited neuromuscular diseases–a world survey. *Neuromuscul Disord.* 1991;1(1):19–29.

[a] Percent that had ≥1 entry for the visit outcome measure.

[b] Care considerations threshold rate for outcome measure.

[c] Percent that met or exceeded the threshold rate.

[d] Average number of visits for the visit outcome measure during the study period.

[e] Average time in years between first and last reported visit.

[f] Status determined as of January 1, 2012.

[g] All visual and measured (goniometry) assessments were combined into 1 category. Cases with both visual and measured assessments were only counted once.

[h] Assessment by a licensed physical therapist.

[i] Vignos lower extremity scale, North Star Ambulatory Assessment, Brooke upper extremity scale, Egen Klassifikation functional assessment, Hammersmith motor scales, or other motor function measure that asses specific domains and give a composite score.

TABLE 4 Corticosteroid Side Effect Monitoring Between January 1, 2012, and December 31, 2014, for Individuals in the Cohort Using Corticosteroids (n = 139)

Visit Outcome Measure	n	Any Visit[a], %	Care Considerations		Annual Visit Rate				Cases With ≥2 Visits		
					Mean	Median	95% CI			Mean	
			Recommend[b], per y	Met[c], %			LL	UL	n	Count[d]	Duration[e]
Constitutional monitoring											
Obesity (wt)											
All individuals	139	98	2×	63	2.21	2.17	2.06	2.37	136	4.94	2.15
Growth retardation (height)											
All individuals	139	89	2×	57	2.11	2.17	1.91	2.30	124	4.29	1.84
Hypertension (blood pressure)											
All males	139	96	2×	63	2.26	2.22	2.09	2.43	134	5.07	2.19
Glucose intolerance[f]											
All individuals	139	69	NC	NC	1.46	1.20	1.22	1.70	96	3.26	1.70
Cataracts (ophthalmologist)											
All individuals	139	17	1×	16	.21	.00	.12	.29	23	1.74	1.28
Bone demineralization or fracture risk											
Serum 25-hydroxy vitamin D											
All individuals	139	71	1×	68	1.47	1.00	1.09	1.88	99	2.46	1.44
DEXA scan											
All individuals	139	40	1×	40	.93	.00	.54	1.32	56	2.30	1.41

Status determined as of January 1, 2012. DEXA, dual-energy radiograph absorptiometry; LL, lower limit; NC, not calculated; UL, upper limit.
[a] The proportion of cases that had ≥1 entry for the visit outcome measure.
[b] Care considerations threshold rate for outcome measure.
[c] Percent that met or exceeded the threshold rate.
[d] Average number of visits for the visit outcome measure during the study period.
[e] Average time in years between first and last reported visit.
[f] All urine and blood glucose tests were combined into 1 category. Cases with both urine and blood tests were only counted once.

TABLE 5 Psychosocial Management Between January 1, 2012, and December 31, 2014, for School-Aged Individuals in the Cohort (6–18 Years) (n = 212)

Support	n	Any Visit, n	Any Visit[a], %
IEP or 504 plan[b]			
All individuals[c]	212	136	64
Transition planning			
All individuals[c]			
Education	212	39	18.4
Pediatric-adult providers	212	39	18.4
Individuals, 12–18 y[c]			
Education	123	38	30.9
Pediatric-adult providers	123	39	31.7
Neurocognitive testing			
All individuals[c]	212	51	24.1

IEP, individualized education plan.
[a] At least 1 entry for the visit outcome measure.
[b] An IEP is developed for schoolchildren needing special education, and a 504 plan is developed for schoolchildren with a disability to receive accommodations to ensure they have access to learning environments and will succeed academically.
[c] Status determined as of January 1, 2012.

management areas do not adhere to recommendations. Of particular concern is the lower adherence to twice-annual monitoring in nonambulatory individuals for whom increased vigilance is recommended to detect any changes in function. Cardiology clinical assessments and echocardiograms appear to be performed as recommended in most of the cohort. Clinical monitoring of scoliosis and peak cough flow are performed in a low proportion of the cohort despite their importance in directing preventive care, interventions, and improvements in quality of life. Similar to the pulmonary function testing, peak cough flow and spinal radiography adherence to recommendations decrease with age and severity. Equally low are the clinical efforts and involvement in transition to adulthood both for education and health care. With these results, it is suggested that implementation of the recommended multidisciplinary neuromuscular management plan is occurring at each neuromuscular visit, despite the lower-than-recommended frequency.

Additional problems include low rates of formal monitoring for side effects of long-term corticosteroid use. Glucose testing was performed in <70% at any point during the study period. Adherence to recommendations was not evaluated because urine dipsticks were infrequently reported, and blood glucose testing is recommended after a urine dipstick that tests positive. Dual-energy radiograph absorptiometry scans were

TABLE 6 Orthopedic Management Between January 1, 2012, and December 31, 2014, for Individuals in the Cohort Without a History of Scoliosis Surgery (n = 216)

Visit Outcome Measure	n	Any Visit[a], %	Care Considerations		Annual Visit Rate				Cases With ≥2 Visits		
					Mean	Median	95% CI		Mean		
			Recommend[b]	Met[c], %			LL	UL	n	Count[d]	Duration[e]
Clinical observation scoliosis monitoring											
Ambulatory[f]	141	45	1× per y	45	0.89	0.00	0.68	1.10	63	2.83	1.46
Spinal radiography											
Ambulatory[f]	141	40	Indication	NC	0.65	0.00	0.47	0.83	56	1.86	1.26
Nonambulatory[f]											
Curves <20°[g]	20	75	1× per y	75	1.45	1.10	0.83	2.07	15	2.27	1.35
Curves ≥20°[g]	28	54	2× per y	18	0.99	1.00	0.53	1.44	15	2.33	1.30
Degree of curvature unknown[g]	24	58	NC	NC	1.06	1.00	0.53	1.59	14	2.50	1.60

Status determined as of January 1, 2012. LL, lower limit; NC, not calculated; UL, upper limit.
[a] Percents that had ≥1 entry for the visit outcome measure.
[b] Care considerations threshold rate for outcome measure.
[c] Percents that met or exceeded the threshold rate.
[d] Average number of visits for the visit outcome measure during the study period.
[e] Average time in years between first and last reported visit.
[f] Status determined as of January 1, 2012.
[g] Curve severity determined by measures collected through January 2012. Cases with completed scoliosis surgery at any time through December 31, 2014, were excluded from table (n = 83, 28%).

TABLE 7 Respiratory Management Between January 1, 2012, and December 31, 2014, for Individuals in the Cohort Without a History of Tracheostomy Placement (n = 294)

Visit Outcome Measure	n	Any Visit[a], %	Care Considerations		Annual Visit Rate				Cases With ≥2 Visits		
					Mean	Median	95% CI		Mean		
			Recommend[b], per y	Met[c], %			LL	UL	n	Count[d]	Duration[e]
Respiratory clinical assessment[f]											
All individuals	299	81	NC	NC	1.92	2.00	1.71	2.12	243	4.34	1.95
Ambulatory[g]	143	68	1×	65	1.37	1.50	1.18	1.55	97	3.90	1.98
Nonambulatory[g]	146	93	2×	66	2.39	2.32	2.05	2.73	141	4.67	1.95
Forced vital capacity											
Ambulatory[g]	143	85	1×	83	1.57	1.55	1.41	1.73	121	3.50	1.89
Nonambulatory[g]	146	89	2×	58	2·16	2.12	1.80	2.52	130	4.12	1.93
Peak cough flow											
Nonambulatory[g]	146	56	2×	33	1.19	1.00	.98	1.40	81	3.14	1.54
Oxygen saturation											
Nonambulatory[g]	146	72	2×	40	1.98	1.44	1.52	2.45	105	3.52	1.57
Carbon dioxide[h]											
Nonambulatory + NIV use[g]	70	43	1×	26	0.94	0.00	0.59	1.29	30	3.03	1.46
Flu vaccine											
All individuals	294	76	1×	75	1.26	1.00	1.14	1.37	224	2.08	1.32

LL, lower limit; NIV, noninvasive ventilation; UL, upper limit.
[a] Percents that had ≥1 entry for the visit outcome measure.
[b] Care considerations threshold rate for outcome measure.
[c] Percents that met or exceeded the threshold rate.
[d] Average number of visits for the visit outcome measure during the study period.
[e] Average time in years between first and last reported visit.
[f] Performed by a pulmonologist or respirologist.
[g] Status determined as of January 1, 2012.
[h] Includes either capnography (end tidal) or blood gas (partial pressure) carbon dioxide measurements.

performed in only 40% of the cohort annually, and cataracts assessments were performed in 17%. If these assessments are performed in external clinics, it is imperative that results are obtained and included in specialty center semiannual notes. This ensures consistent monitoring is tracked by the neuromuscular specialists and shared with their managing primary care provider. It also alerts the neuromuscular specialist of the need to obtain these clinical assessments if they are not being performed. This is true for

TABLE 8 Cardiac Management Between January 1, 2012, and December 31, 2014, for All Individuals in the Cohort ($N = 299$)

Visit Outcome Measure	*N* or *n*	Any Visit[a], %	Care Considerations		Annual Visit Rate				Cases With ≥2 Visits		
					Mean	Median	95% CI		Mean		
			Recommend[b]	Met[c], %			LL	UL	*n*	Count[d]	Duration[e]
Cardiology clinical assessment[f]											
All individuals[g], age in y	299	82	NC	NC	1.42	1.20	1.27	1.56	244	2.34	1.46
<10	98	66	Once baseline		1.02	1.00	0.79	1.25	65	1.74	1.23
≥10	201	89	2× per y	87	1.61	1.50	1.43	1.78	179	2.56	1.55
Electrocardiogram[g], age in y											
<10	98	50	1× per 2 y	50	0.83	0.36	0.56	1.11	49	1.84	1.32
≥10	201	63	1× per y	60	1.15	1.00	0.95	1.35	126	2.61	1.57
Echocardiogram[g], age in y											
<10	98	84	1× per 2 y	84	1.28	1.00	1.10	1.45	82	2.21	1.45
≥10	201	93	1× per y	90	1.67	1.50	1.47	1.86	186	2.62	1.61

LL, lower limit; NC, not calculated; UL, upper limit.
[a] Percents that had ≥1 entry for the visit outcome measure.
[b] Care considerations threshold rate for outcome measure.
[c] The proportion of cases that met or exceeded the threshold rate.
[d] Average number of visits for the visit outcome measure during the study period.
[e] Average time in years between first and last reported visit.
[f] Performed by a cardiologist.
[g] Status determined as of January 1, 2012.

all measures demonstrating low adherence.

Few studies in which researchers describe the implementation of care considerations for DMD have been performed. Of those published, evaluation relied on self-report from families of individuals affected with DMD[13–15] or survey data reported from neuromuscular centers.[16] To our knowledge, this is the first attempt to conduct retrospective medical record review on a population-based sample to evaluate implementation of the care considerations. In addition, our study evaluated care practices over a more extended observation period (3 years) than other studies, which included cross-sectional responses[13,15,16] and reports for the previous 6 months.[14] The longer observation period allowed for a more robust estimate of visit frequency.

Neuromuscular management includes skeletal function assessments, documentation of functional performance, and monitoring of potential side effects from the long-term use of corticosteroids. These assessments are critical to the health, well-being, and decisions about disease management by the physician and family. Despite the case management aspect of the neuromuscular team, twice-annual visits to neuromuscular specialists vary greatly from country to country.[13–15] In the majority of reported survey data as well as our retrospective study, it is indicated that <70% of individuals attend 2 times per year.

The care considerations list numerous types of neuromuscular assessments to be conducted during the neuromuscular visits in addition to steroid monitoring. These assessments include range of motion, timed testing, motor function assessment scales, spinal assessments, and equipment recommendations or adjustments. Lower extremity range of motion was measured semiannually in 64% of our population, whereas all other neuromuscular assessments occurred semiannually for <50% of the cohort. Many of these tasks are typically performed by physical therapists. We found that only 50% of our sample saw a physical therapist twice a year, with other authors reporting between 48% and 93%.[13–16]

Additional assessments include monitoring for scoliosis, pulmonary function, and cardiac function. In our data, it is demonstrated that 75% of individuals with curves <20° received radiographic assessments; however, other authors report <50%.[13,15] Initial respiratory and cardiac care recommendations for individuals with DMD were released in 2004 and 2005 and were addressed again in the 2010 care considerations. Overall, the proportion that had forced vital capacity and echocardiogram assessments at or above the recommendations of the care considerations was 80% in our study. Other authors report similar proportions of adherence in younger patients, but these proportions drop dramatically in older patients, with <46% of adults meeting assessment recommendations, which is in direct contrast to recommendations for increased frequency in pulmonary function testing and monitoring.[5,14,15] Many patients who use wheelchairs full-time require transfers from their wheelchair, which may not be possible in all clinical settings. It is possible that some assessments (eg, height, weight, scoliosis monitoring) are not performed because of logistical difficulties in certain clinical settings without the facilities to assist

persons who rely on technology such as power wheelchairs.

Psychosocial and neurocognitive assessment is difficult to evaluate by using medical record data because records of specialists directly involved in such assessments (eg, psychiatrists, psychologists) are not available, and educational information is rarely recorded. Nonetheless, we attempted to ascertain the degree to which neuromuscular clinics manage the psychosocial needs of individuals under their care. We found >64% of our cohort received specialized services in the school system for either psychosocial or physical accommodations. This corresponds to estimated proportions of individuals with DMD who are predicted to have an intellectual disability, learning disabilities, or comorbid neurodevelopmental conditions such as autism, attention-deficit/hyperactivity disorder, anxiety, and obsessive-compulsive disorder.[19–21] Interestingly, Landfeldt et al[14] indicate diagnoses for neurodevelopmental comorbidities appear to be more frequently documented in the United States (51%) than in other countries (11%–17%). Transition planning was infrequently documented in the clinical setting, noted in only 32% of our cohort of individuals between the ages of 12 and 18 years. Numerous researchers document the shortcomings of existing transition programs or the lack thereof in meeting the needs of individuals with DMD.[22–27]

In several studies on other chronic pediatric conditions, researchers have found similar variability in adherence to care across centers and have documented improvement in adherence when using interventions.[28,29] An important component of any consensus-based clinical care recommendation is follow-up to demonstrate clinical benefits from each recommendation. In a 2013 review in which various surveillance and self-enrollment patient registries are described, authors called for the creation of a unified registry that would provide publicly available patient outcomes that could be stratified by clinic.[30] Although this model could be used to measure outcomes and adherence to performance of the care considerations, our experience and that of the literature to date has shown that patient registries often have biased samples that are not representative of the total population unless they are clinically implemented. One exception has been the approach to the cystic fibrosis data registries, which include data points consistently measured across all sites.[28] The Muscular Dystrophy Association initiated a similarly designed clinical registry in 2013.[31] The CDC is spearheading a collaborative effort with Parent Project Muscular Dystrophy, Muscular Dystrophy Association Registry, and MD STAR*net* to develop a coordinated evaluation of the updated care considerations (N. Street, MS, personal communication, 2017). Multiple modalities for evaluating the care considerations are necessary to determine what elements are essential to achieve the best possible quality of life and longevity while reducing the time and financial impact on the system, providers, and patients. The limitations of our study are common to retrospective cohort study chart review designs. Measurement error present from incomplete, missing, or poorly documented information in medical records could lead to missing data that creates the appearance of low adherence. Although some individuals may have left the surveillance area during the study, we made efforts to reduce these effects by creating individual durations using visit dates rather than a 3-year denominator for all cases. Although our study represents population-based ascertainment, our data were collected primarily from neuromuscular specialty centers. This is a practical approach when dealing with rare and complex conditions but limits the generalizability of findings to only those care centers who should be providing up-to-date care according to current clinical guidelines. Individuals with DMD may access certain providers in private clinics that were not accessed by the network and therefore not included in this analysis. In this article, we focused on evaluating adherence to the care considerations, and we had no means of determining the impact these variations may have on actual patient outcomes. Authors of future studies should evaluate the relationship between adherence and outcomes related to function, quality of life, morbidities, mortality, and health care costs. Low adherence could partly reflect variability in practice patterns among the MD STAR*net* sites as evidenced by authors of previous network articles reporting on variation in use of corticosteroids and some services and assistive devices.[32–34] Taken as a whole, with the results to date, we support the need for ongoing, uniform data collection on a national basis and strengthening the proposed effort by MD STAR*net* to measure outcomes. Finally, because of potential for loss to follow-up, our calculated visit rates were based on the first and last documented visits. This resulted in variable duration estimates and the assumption that calculated visit rates extended beyond the observed follow-up period. Despite these limitations, our study has several strengths compared with previous studies. In our study, we used a population-based cohort of individuals with DMD and BMD from multiple sites. Data presented were gathered directly from medical records through expert abstraction and are used to demonstrate objective assessments of current practice and a representative

summation of information documented by practicing physicians in the surveillance area. Lastly, most of our findings reveal agreement with estimates provided by authors of other studies using self-report methods but provide improved precision with respect to recommendations used in the care considerations.

CONCLUSIONS

In our data, we identify several areas in which practice aligns well with recommendations in the care considerations, but also several areas in which implementation is lacking. Specifically, as individuals with DMD move from late childhood to adolescence and adulthood and require more complex and intensive medical care, there are important assessments and interventions that are either not being documented or not being performed at the recommended frequency. Routine functional assessments and transition planning are increasingly important to assure optimal care and improved longevity and quality of life. Many of these screening, treatment, and transition elements can be supported by pediatricians caring for individuals with DMD. Primary care physicians can play a significant role in managing the care of these individuals by keeping abreast of current recommendations, maintaining communication with specialists, and creating support plans when families live a significant distance from specialized care centers. Pediatricians are aware of their medical community and are best able to make the appropriate referrals to adult primary care providers and subspecialists. The 2018 revision of the care considerations addresses the primary care physician's role.[35] Multiple modalities for evaluating the care considerations are necessary to determine what elements are essential to achieve the best possible quality of life and longevity while reducing the time and financial impact on the families, providers, and the health care system. In our study, we provide precise benchmark data garnered from a population-based sample, which serves as a historical control. These data can be used for future studies of adherence to the care considerations.

ACKNOWLEDGMENTS

We thank the staff and investigators from the MD STAR*net* sites participating in this project. We also thank Kathi Kinnett from Parent Project Muscular Dystrophy and Jodi Wolff from the Muscular Dystrophy Association for their contributions to this project.

ABBREVIATIONS

BMD: Becker muscular dystrophy
CDC: Centers for Disease Control and Prevention
CI: confidence interval
DMD: Duchenne muscular dystrophy
IRR: initial interrater reliability
MD STAR*net*: Muscular Dystrophy Surveillance, Tracking, and Research Network

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PEDIATRICS (ISSN Numbers: Print, 0031-4005; Online, 1098-4275).

FINANCIAL DISCLOSURE: The authors have indicated they have no financial relationships relevant to this article to disclose.

FUNDING: Supported by the Centers for Disease Control and Prevention (cooperative agreement number U01DD001041).

POTENTIAL CONFLICT OF INTEREST: The authors have indicated they have no potential conflicts of interest to disclose.

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Pediatrics 2018;142;
DOI: 10.1542/peds.2017-4006 originally published online June 20, 2018;

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