Appendix: Diagnosis and management of Duchenne muscular dystrophy, an update, part 1: Diagnosis, neuromuscular, rehabilitation, endocrine, and nutritional management

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Activities of Daily Living (ADL) Assessment: ADL, instrumental ADL, access, adaptive equipment, & assistive technology

Falls Risk Assessment

Assessment of learning, attention, and sensory processing

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Supplementary Table 2. Care Considerations on Use of Orthotic, Mobility, and Functional/Safety Devices

Device	Care Considerations
Orthotic Devices	
Ankle-foot orthoses (AFOs)	• Resting or stretching AFOs appropriate for use at night throughout life course • Best tolerated if started preventively at young age • Not indicated for use during ambulation (typically compromise use of compensatory movements needed for function and energy efficiency; with increased difficulty walking, getting up from the floor, and climbing stairs; increased falls; increased fatigue; decreased endurance) • May be appropriate for daytime use among full-time wheelchair users
Knee-ankle-foot orthoses (KAFOs)	• No longer used as often as historically because of availability of other supportive standing devices (e.g., power stand-and-drive wheelchairs) • May be appropriate for therapeutic rather than functional use in some cases depending on individual and team preferences
Serial casting	• Option when stretching and orthotic use do not maintain adequate range of motion or when surgery is not a preferred option • Requires a skilled team to implement successfully
Supported standing devices	• Consider preventive use when standing in optimal alignment and walking become more difficult • Use in late ambulatory and early non-ambulatory stages if contractures are not too severe to restrict positioning and devices are tolerated • May be continued into late non-ambulatory stage if contractures are not too severe to restrict positioning and devices are tolerated • Motorized stand-and-drive wheelchairs obviate the need for transfers to use supported standing, decreasing falls risk and increasing duration of supported standing for increased benefits of supported standing
Knee extension splints	• Can be used at rest to optimize range of knee extension
Wrist/hand splints, oval 8 finger splints, stretching gloves	• Used preventatively with emergence of tightness in wrist/hand/finger musculature
Mobility Devices	

Manual mobility devices	• Ultra-lightweight manual mobility device (e.g., manual wheelchair)
	  ○ Ambulatory stage: use when long-distance mobility demands exceed endurance or terrain is too challenging
	  ○ Non-ambulatory stages: use when lack of access or ability to transport motorized devices
	• Custom seating systems* are critical for preventing deformity
	• Power assist can be added for energy conservation / greater independence

Motorized mobility devices	• Individuals who walk in most situations: Use powered mobility devices as needed for energy conservation and intermittent independent long-distance mobility • When walking becomes more difficult: Recommend use of motorized wheelchair • Motorized stand-and-drive wheelchairs: Recommended for supported standing in addition to mobility • Custom seating systems* and power positioning components** are critical for preventing deformity and maintaining skin integrity
Functional/Safety Devices	
Bathing/toileting equipment: Elevated toilets, toilet safety rails, bidets, tub benches, bath seats, slider bath chairs, roll-in showers, roll-in shower/commode chairs (may need head rest, tilt, recline, leg support)	• For safety and support during bathing and toileting, to decrease risk of falls and support hygiene and skin care, and to maintain skin integrity, equipment should be used early for fall prevention in the bathroom, when ambulation is precarious, and transfers become difficult
Lift and transfer/hoist equipment: Transfer boards, stand-and-pivot aides, free standing lifts, ceiling lifts, slider sheets, turning slings, pivot seats in cars	• For safe and functional lifts and transfers, to minimize risk of falls, equipment should be used early when transfers become difficult, especially in bathroom where fall risk is high
Beds and mattresses: Hospital/motorized beds, pressure relief mattresses	• For pressure relief, positional support, motorized change of position to maintain skin integrity and minimize contracture and deformity, motorized adjustable bed height for safe transfers
Access equipment: Ramps, stair climbers, platform/vertical/porch lifts, elevators, vehicle lifts and modifications, evacuation chairs, beach and outdoor chairs	• For safe and functional access and transport of mobility equipment

Assistive technology for function: "smart" computer/phone/tablet/home access with environmental control, blue tooth, voice and eye gaze activation, electronic feeders, exoskeletons and mobile arm supports, robotics, technology mounts	• For optimizing function, independence, and participation in all aspects of life

*Custom seating system components for manual and mobility devices include solid seat with pressure relief cushion, hip guides, and adductors; solid back with rigid lateral trunk supports; headrest with additional components as needed such as facial components for head support; and elbow supports.

** Power positioning components for motorized wheelchairs include power tilt, power recline, separately elevating power elevating leg rests, power adjustable seat height, and power stand-and drive

Supplementary Table 3. Critical Transition Periods in DMD with Anticipated Weight Gain/Loss

Risk for Overweight/Obesity: Glucocorticoid (Steroid) Initiation and Loss of Ambulation

General Recommendations for Prevention of Overweight/Obesity

- Family-centered healthy eating (individual with DMD should not be singled out)
  - Control portions of meats and starches
  - Increase fruit/vegetables
  - Decrease sugar-containing drinks, increase water intake
  - Choose low-fat or fat-free milk
  - Schedule meals/snacks (no grazing), always at the table (limit meals and snacking in bedrooms/living rooms with TV on/in car)
  - Limit fast food and high-calorie/salty snacks (chips, cookies, etc.)
  - Choose low-sodium versions of processed foods, limit added salt in cooking and at the table, use available salt substitutes
  - Choose whole-grain/low-sugar cereals, breads, etc.
- Family-centered physical activity (adapted as necessary and amount/duration/frequency as recommended by care team to meet the needs of the individual with DMD as his ambulation declines)

Additional Strategies if Weight Gain is Excessive

- Structured meals and snacks
- Minimize consumption of energy dense foods
- Supervise activity/play for at least 60 minutes per day
- Limit screen time to 1 hour/day or less
- Close monitoring of weight, diet and activity by clinical team. Due to the decrease in energy expenditure seen at loss of ambulation, reductions of caloric intake by 250-500kcal/day may be required for weight stabilization.

Risk for Underweight/Malnutrition: Onset of Swallowing Dysfunction

- It is common for patients to lose weight unintentionally both before and at onset of dysphagia.
- When signs and symptoms of dysphagia develop or there is unexplained weight loss, a Speech Language Pathologist (SLP) should be consulted.
- Alter food texture as needed to aid in chewing and swallowing (e.g., soft diet, sips of water with each bite).
- Adapt feeding environment as needed: raised surface (counter-height table) with elbows propped up on surfaces, long straws.
- Individuals with DMD tend to lose their ability to eat independently after a spine fusion because they are no longer able to bend over so that their mouth is close to the table.
- Individuals with DMD struggle with the gradual transition from eating independently to requiring maximum assistance with meals and often will not let others help them eat (especially at school). Patients often stay dehydrated intentionally so that they do not need assistance going to the bathroom at school. If patients are home alone for any extended periods of time, they may go without eating.
- Increase calorie intake by mouth:
  - Add calories to prepared foods using full-fat dairy products, cream sauces, oils/butters, avocado, dips (e.g., nut butters, salad dressing, hummus, sour cream).
  - Offer homemade high-calorie yogurt smoothies and milk shakes as snacks.
  - Offer oral nutrition supplements as snacks (e.g., Boost, Ensure, Carnation Breakfast Essentials, Scandishake or 1.5 kcal/ml formulas).
- When a patient cannot achieve caloric goals by mouth and weight loss is sustained, gastrostomy tube feedings are recommended.
- Patient respiratory status and compliance with assisted ventilation, if present, must be considered in assessing overall energy requirements.

Literature Review Search Strategy

Research Triangle Institute (RTI) updated the literature review from the original eight DMD care consideration panel topics: (1) neuromuscular, (2) cardiac, (3) respiratory, (4) rehab, (5) psychosocial, (6) gastrointestinal and nutrition, (7) diagnostic, (8) orthopedic and surgical, using a similar search strategy as done previously. RTI also conducted a comprehensive literature review on the three new topics: (1) management of disease in adults with DMD and transition, (2) endocrinology, and (3) management of the primary and emergency care of people with DMD.

Articles for the literature review were identified by searching Medline, Embase, Web of Science, and the Cochrane Library databases for peer-reviewed, English-language articles published from 2006 through 2013 for the eight original topics and from 1990 through 2013 for the three new topics. The literature was searched using the key search terms of "Duchenne" or "muscular dystrophy," or both, paired with one of **626 search terms** (combination of the **410 search terms** from the original care consideration and **216 new search terms**).

The literature search identified **1,215** articles after duplicates were removed. The titles and abstracts of the retrieved citations were reviewed to determine which met the inclusion criteria outlined in the search strategy above (Articles published in English, year of publication, published complete articles, human subjects, abstract available). Reviews, meta-analyses, case series, case reports, animal models, and articles on unrelated diseases or Becker muscular dystrophy only were excluded upon further review. RTI provided the Steering Committee members with a list of **672** abstracts from the search. The Steering Committee provided feedback on if the article should be included for further review by indicating "yes/no/maybe" if they felt the reference should be considered when compiling literature to be used in updating the clinical care recommendations**.**

Peer-reviewed articles **(430)** that experts chose yes, maybe, or there was disagreement among experts, were reviewed and summarized into a draft summary report for each topic. The draft summary reports and the associated PDF articles were provided to the experts as background information to review and discuss via teleconference to determine the literature required for the update**.** The final summary reports included the abstract summaries and recommendations from the SC on whether the literature reviewed: (1) was consistent with the existing care considerations, (2) conflicted with the existing care considerations, (3) required an update to the care considerations, or (4) presented promising research. Subject matter experts, with the assistance of RTI, also continually updated the references to identify additional relevant articles.

Articles that were identified as required for the update were used to create clinical scenarios in accordance with the RAND method. Many articles that were identified as supporting or conflicting with the current DMD care considerations or considered promising research were used to update the references in the care considerations.

Before publication, an updated literature search was conducted for articles published between October 2013 and July 2017, which identified 880 articles. Committee chairs reviewed 115 articles potentially relevant to care and updated the references and text as necessary.

Table of Search Terms

Topics		Suggested Search Terms
1. Muscle imaging and evaluation of body composition	1	X-Ray Computed Tomography
	2	Muscle imaging
	3	MRI
	4	DEXA
	5	Spine X-ray
	6	Fractures
	7	X-ray
	8	Bone density
	9	Bone quality
	10	EMG
	11	Biopsy
	12	Dystrophinopathy
	13	Dystrophin and clinical
	14	Histochemistry
	15	Histology
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	18	Muscle volume (and MRI)
	19	Magnetic Reasoning Imaging
2. Strength and muscle functioning/physical therapy/limb contractures	20	Exercise
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	27	Eccentric muscle contraction
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	29	Muscle Injury
	30	Concentric muscle strengthening
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	34	Range of Motion, Articular
	35	Functional assessment
	36	Back bracing
	37	Braces
	38	Spinal bracing
	39	TLSO
	40	Physical activity
	41	Kinematics
	42	Muscle power
	43	Concentric strength
	44	Eccentric strength
	45	Quantitative strength
	46	Gait

	47	Kinesiology
	48	Hydrotherapy
	49	Aquatic therapy
	50	Hippotherapy
	51	Stretching
	52	Wheelchairs
	53	Scooter
	54	Walkers
	55	Assistive devices
	56	Self-Help Devices
	57	Ambulation
	58	Falls
	59	Timed function assessment
	60	Energy expenditure
	61	Total energy expenditure
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	65	AFO
	66	Night splints
	67	Splints
	68	Body jacket
	69	Strengthening
	70	Moments
	71	Locomotion
	72	Mobility
	73	Seating
	74	EZ Stand
	75	Strength Testing
	76	Manual Muscle Testing
	77	Range Of Motion Measurements
	78	Timed Function Tests/Testing
	79	Functional Scales
	80	Vignos Scale
	81	Lower Extremity Scale
	82	Brooke Scale/Upper Extremity Scale
	83	Bayley Scale Of Infant Development
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	100	Pain management
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	106	Malignant hypertonia
	107	Malignant hyperthermia
	108	Hyperthermia
	109	Muscle relaxants
	110	Pulmonary complications
	111	Atelectasis
	112	Retained bronchial secretions
	113	Acute respiratory distress
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	119	Postoperative complication
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	129	Spinal deformity
	130	Spinal curvature
	131	Spinal stabilization
	132	Spinal instrumentation
	133	Fixation
	134	Achilles Tendon
	135	Tendo Achilles
	136	Hamstring
	137	Tenotomy
	138	Walking
	139	Deformity
	140	Correction
	141	Prolongation of Walking
	142	Lower limb

	143	Heel cord lengthening
	144	Iliotibial band
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	146	Spinal function
	147	Spinal fusion
	148	Surgery
	149	Luque Technique
	150	Coagulation factor
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	171	Sunlight
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	182	Osteogenic
	183	Fracture risk
	184	Prevention
	185	Demineralization
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	187	Resistance training
	188	Vertebra or Spine
	189	Stander
	190	Vibration

	191	Osteocalcin
	192	Pyridinoline
	193	Telopeptides
	194	Bone Markers
	195	Quantitative Ct
	196	Hypercalciuria
	197	Other Generic Brand Names Of Bisphosphonate: Actonel (Risedronate); Boniva (Ibandronate); Aredia (Pamidronate); Zometa Or Reclast (Zoledronic Acid)
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	199	Pamidronate
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	237	Scopolamine

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	243	Bilevel Positive Airway Pressure (BiPAP)
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	245	Invasive ventilation
	246	Tracheotomy
	247	Supplemental Oxygen
	248	Sleep Quality
	249	Sleep Apnea Syndromes
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	251	Respiratory function tests
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	260	Shortness Of Breath
7. Cardiac monitoring and management	261	Cardiac Care
	262	Cardiomyopathies
	263	Cardiomegaly
	264	Congestive heart failure
	265	Heart block
	266	Systolic Ejection Fraction
	267	Pacemaker
	268	Heart transplant
	269	Dilated Cardiomyopathy
	270	Conduction therapy
	271	Angiotensin-Converting Enzyme Inhibitors
	272	Adrenergic beta-Antagonists (Beta Blockers)
	273	Echocardiogram
	274	Electrocardiogram
	275	Arrythmia
	276	Rhythm abnormities
	277	Dysrhythmias
	278	Cardiac MRI
	279	Contrast enhanced cardiac MRI
	280	Ambulatory electrocardiography
	281	Diastolic dysfunction
	282	Ischemia
	283	Dobutamine
	284	Dobutamine stress echocardiogram
	285	MUGA (Multi-gated acquisition)

	286	Radionucleotide
	287	Mitral Valve Insufficiency
	288	Doppler Echocardiography
	289	Pulmonary hypertension
	290	Chest pain
	291	Syncope
	292	Congenital heart disease
	293	Structural cardiac disease
	294	Ventricular Dysfunction
	295	Carvedilol
	296	Digoxin
	297	Enalapril
	298	Thrombosis
	299	Embolism
	300	Heart Failure
	301	Myocardial Strain
	302	Lisinopril
	303	Aldactone
	304	Losartan
	305	Intracardiac Defibrillator (Icd)
	306	Left Ventricular Assist Device
	307	Gerd
	308	Reflux
	309	Genitourinary
	310	Encopresis
	311	Enuresis
	312	Sip Vent
	313	Breath Stacking
	314	Pneumonia Vaccine
	315	Icd (Implantable Cardiac Defibrillator)
	316	Vad (Ventricular Assist Device)
	317	Losartan/Angiotensin Receptor Blocker
	318	Aldactone
	319	Eplerenone
	320	Halter Monitor
	321	Tissue Doppler Imaging
8. Gastrointestinal/ Nutritional monitoring and management	322	Cardiac Fibrosis
	323	Transition
	324	Obesity
	325	Deglutition Disorders/Swallowing difficulties
	326	Gastric dilatation
	327	Hypotonia
	328	Dysphasia/Aphasia
	329	Deglutition Disorders/Dysphagia
	330	Constipation
	331	Diarrhea
	332	Modified meals
	333	Nutrition

	334	Gastrointestinal
	335	Wasting/Wasting Syndrome
	336	Stool Softeners
	337	Cathartics/Laxatives
	338	Gastrointestinal Stimulants
	339	Cisapride/Propulsid
	340	Feeding Tube
	341	PEG tube
	342	Dietetic counseling
	343	Gastric emptying
	344	Nutritional needs assessment
	345	Weight monitoring
	346	Formal intake analysis
	347	Swallowing studies
	348	Videofluoroscopy
	349	Sodium phosphate/Phosphate
	350	Sodium biphosphate/Enema
	351	Complementary/alternative medicine
	352	Creatine
	353	Royal jelly/Bee pollen/Propolis
	354	Antacids
	355	Zantac/Ranitidine
	356	Prilosec/Omeprazole
	357	Prevacid
	358	Weight loss
	359	Weight gain
	360	Anorexia
	361	Feeding difficulty
	362	Cachexia
	363	Appetite stimulant
	364	Gastric tube
	365	Failure to thrive
	366	Supplement
	367	Resting energy expenditure/Energy Metabolism
	368	Hypermetabolic
	369	Body Weight
	370	Body Height
	371	Body Mass Index (Bmi)
	372	Growth Linear Height
	373	Growth Charts
	374	Anthropometry
	375	Body Composition
	376	Fat Mass
	377	Fat-Free Mass
	378	Arm Span
	379	Segmental Length
	380	Short Stature
	381	Under-Nutrition

	382	Malnutrition
	383	Overweight
	384	Obese
	385	Gastroesophageal Reflux
	386	Proton Pump Inhibitors
	387	Calories
	388	Nutrients
	389	Proteins
	390	Carbohydrates
	391	Fats
	392	Vitamins
	393	Minerals
	394	Fluid Intake
	395	Corticosteroids
	396	Hyperglycemia
	397	Diabetes
	398	Basal Metabolic Rate
	399	Resting Energy Expenditure
	400	Total Energy Expenditure
	401	Gastrostomy Tube
	402	Fundoplication
	403	Anti-Reflux Surgery
	404	Nissen Fundoplication
	405	Jejunostomy
	406	Dysphagia (As Opposed To Dysphasia)
	407	Miralax (Polyethylene Glycol)
	408	Lactulose
	409	Senna
	410	Dysglycemia
	411	Glucose
	412	Hemoglobin Ac
	413	Delayed Puberty
	414	Growth Failure
	415	Growth Hormone
9. Cognitive and psychological issues	416	Cognitive
	417	Learning disability
	418	Behavior
	419	Memory
	420	Working memory
	421	Body image
	422	Depression
	423	Anxiety
	424	Anger Management
	425	Self esteem/self concept
	426	Sexuality
	427	Antidepressants
	428	Anti-anxiety drug
	429	Mood stabilizers

	430	Sleep
	431	Apnea
	432	Daytime sleepiness
	433	IQ/Intelligence
	434	Verbal skills
	435	Quality of life
	436	Neuropsychological profile / testing
	437	Access
	438	Participation
	439	Poor school performance
	440	Learning disorder/Learning difficulty/Learning difference
	441	Special Education
	442	Mental retardation
	443	Cognitive deficit
	444	Math skills
	445	Developmental delay/disability
	446	Motor delay
	447	Autism
	448	Asperger's
	449	Autism Spectrum Disorders
	450	Brain
	451	Oppositional
	452	Adhd
	453	Attention
	454	Ssri
	455	Stimulant
	456	Transition
	457	Independence
	458	Caregivers
	459	Learning Problems
	460	Planning
	461	Executive Function
	462	Advance Guidance/Advance Decision
	463	Hypokalemia
	464	Potassium
	465	Sleep Study
	466	Megace
10. Pharmacological treatment	467	Drug
	468	Medications
	469	Pharmacology
	470	Adrenal Cortex Hormones
	471	Steroid
	472	CoEnzyme Q/Ubiquinone
	473	Adverse effect
	474	Medical Care
	475	Calcium blockers
	476	Anabolic Agents
	477	Dantrolene

	478	Digitalis Glycosides
	479	Prednisone
	480	Albuterol
	481	Nandrolone
	482	Vitamin E
	483	Dietary supplements/Vitamins
	484	Arginine, Glutamine, and HMB
	485	Juven
	486	Protease inhibitors
	487	L-arginine
	488	Prostaglandin Antagonists
	489	Heparin
	490	Dextrans
	491	Adverse event/Drug Safety
	492	Drug interaction
	493	Polypharmacy
	494	Anti-inflammatory Agents
	495	Deflazacort/Glucocorticoids/Pregnenediones
	496	Testosterone
	497	Viagra
	498	Opioids
	499	(Anticonvulsants Gabapentin, Pregabalin)
11. Social participation and family functioning/recreation	500	Independence
	501	Palliative Care
	502	Hospices
	503	Bereavement
	504	Siblings
	505	Divorce
	506	Coping/Psychological Adaptation
	507	Stress
	508	Social isolation
	509	Social Integration/participation/adjustment
	510	Developmental Screening
	511	Speech Therapy/Speech-Language Pathology
	512	Speech/Communication
	513	Accessibility
	514	Assistive Technology
	515	Communication Aids
	516	School
	517	Family support
	518	Respite Care
	519	Canine Companion
	520	Education
	521	Recreation
	522	Sport
	523	End of life
	524	Terminal Care
	525	Community integration

	526	Employment
	527	Marriage
	528	Sexual partners
	529	Vocation/vocational
	530	Job higher education
	531	Adaptive equipment
	532	Parenting
	533	Independent living
	534	Assisted living
	535	Family functioning
	536	Genetic counseling
	537	Higher education
12. Other	538	Ophthalmology
	539	Dental
	540	Bed sores
	541	Decubitus Ulcer
	542	Deep Vein Thrombosis
	543	Pulmonary embolism
	544	Cataract
	545	Funding (medicaid/medicare)
	546	Urinary Problems- Incontinence
	547	Urgency
	548	Pressure Ulcer
New Topic Panels Added for the DMD Care Considerations Update 13-15 Search Terms		
13. Adult Care	549	Ambulatory Care - Adult age groups
	550	Bone Diseases/etiology/therapy - Duchenne OR Becker
	551	Heart Diseases/etiology/therapy
	552	Language Disorders/etiology/therapy
	553	*Patient Care Team
	554	Physical Therapy Specialty/methods
	555	Treatment Outcome
	556	Catastrophization/prevention & control
	557	Chronic Pain
	558	Cognition*
	559	Pain
	560	Analgesics/therapeutic use
	561	Disease management
	562	Case management
	563	Models, Organizational
	564	Rehabilitation Nursing
	565	Continuity of Patient Care
	566	Activities of Daily Living
	567	Physical Therapy Modalities*
	568	Terminal Care*
	569	Sexual Behavior
	570	Interpersonal Relations
14. Endocrinology	571	Adrenal Cortex Hormones/therapeutic use - Humans
	572	Growth Disorders/physiopathology - Duchenne OR Becker

	573	Metabolic Diseases/physiopathology - Male
	574	Obesity/physiopathology
	575	Osteoporosis/physiopathology
	576	Bone and Bones/physiopathology
	577	Growth/physiology
	578	Puberty/physiology
	579	Weight Gain/physiology
	580	Sperm Injections, Intracytoplasmic
	581	Adrenocorticotropic Hormone/blood*
	582	Hydrocortisone/blood*
	583	Hypothalamo-Hypophyseal System/physiopathology
	584	Pituitary-Adrenal System/physiopathology
	585	Follicle Stimulating Hormone
	586	Anesthesia, General*
	587	Hypothalamo-Hypophyseal System/physiopathology
	588	Diuretics/diagnostic use
	589	Fludrocortisone/therapeutic use
	590	Furosemide/diagnostic use
	591	Hyperkalemia/complications*
	592	Hypoaldosteronism/physiopathology
	593	Mineralocorticoids/therapeutic use
	594	Renin/blood
15. Primary and Emergency Care	595	Transition to Adult Care*
	596	Bronchopneumonia/etiology*
	597	Bronchopneumonia/therapy
	598	Pneumoperitoneum/therapy
	599	Subcutaneous Emphysema/etiology*
	600	Tracheostomy/methods*
	601	Urea/blood
	602	Peroxides/blood
	603	Home Nursing/psychology*
	604	Self Concept
	605	Dependency (Psychology)*
	606	Cardiopulmonary Resuscitation/methods*
	607	Respiration, Artificial/methods*
	608	Chest Wall Oscillation*
	609	Influenza, Human/complications*
	610	Pulmonary Atelectasis/etiology*
	611	Combined Modality Therapy/standards
	612	Resuscitation/instrumentation
	613	Adrenergic beta-Antagonists/therapeutic use
	614	Angiotensin-Converting Enzyme Inhibitors/therapeutic use
	615	cardiac assessment
	616	Positive-Pressure Respiration*
	617	Disease Progression
	618	Blood Gas Analysis
	619	Palliative Care*
	620	Patient-Centered Care*

	621	Physician-Patient Relations
	622	Professional-Family Relations
	623	Resuscitation Orders*/ethics
	624	School Health Services/ethics
	625	Home Care Services
	626	Heart Arrest/therapy*

Literature Review Summary by Topic

Articles that the steering committee indicated as "requires update", "promising research", or "conflicts with existing care considerations" are included in the tables below. Articles that are "consistent with the existing care considerations" are not listed in the following tables, but may have been cited in the updated reference section to support previous guidance.

Gastroenterology/Nutrition

The clinical experts agree that there is sufficient new literature to update to the current recommendations for gastroenterology/nutrition topic. There is also new literature that supports current recommendations and will allow expansion of sections where the current recommendations are vague.

Sub-topics that need to be incorporated or expanded in the update are gastrostomy tubing (i.e. aspiration vs non-aspiration, types of food, timing of placement etc.), creatinine supplement use, weight recommendations, and methods for establishing body composition (fat free vs fat free mass). There is no mention of body composition measures in the previous guidelines. It is recommended that dieticians/nutritionist who sees patients on a daily basis be consulted when preparing the update.

Gastroenterology/Nutrition		
Study	**Sub-Topic**	**Recommendation**
Mok et al. (2006) Estimating body composition in children with Duchenne muscular dystrophy: Comparison of bioelectrical impedance analysis and skinfold-thickness measurement. American Journal of Clinical Nutrition, 83(1), 65–69.	Body Composition	Requires update
Mok et al. (2010) Assessing change in body composition in children with Duchenne muscular dystrophy: Anthropometry and bioelectrical impedance analysis versus dual-energy X-ray absorptiometry. Clinical Nutrition, 29(5), 633–638.	Body Composition	Requires update
Archer et al. (2013) Dysphagia in Duchenne muscular dystrophy assessed by validated questionnaire. International Journal of Language and Communication Disorders, 48(2), 240–246.	Nutritional Assessment Tool	Requires update
van den Engel-Hoek et al. (2013) Oral muscles are progressively affected in Duchenne muscular dystrophy: Implications for dysphagia treatment. Journal of Neurology, 260(5), 1295–1303.	Nutritional Assessment Tool	Requires update
Terzi et al. (2010) Impact of tracheostomy on swallowing performance in Duchenne muscular dystrophy. Neuromuscular Disorders, 20(8), 493–498.	Nutritional Assessment Tool	Promising research
Elliott et al. (2012) Predicting resting energy expenditure in boys with Duchenne muscular dystrophy. European Journal of Paediatric Neurology, 16(6), 631–635.	REE	Promising research
Shimizu-Fujiwara et al. (2012) Decreased resting energy expenditure in patients with Duchenne muscular dystrophy. Brain & Development, 34(3), 206–212.	REE	Promising research
Martigne et al. (2011Natural evolution of weight status in Duchenne muscular dystrophy: A retrospective audit. British Journal of Nutrition, 105(10), 1486–1491.	Growth	Promising research

ten Dam et al. (2012) Normal height and weight in a series of ambulant Duchenne muscular dystrophy patients using the 10 day on/10 day off prednisone regimen. Neuromuscular Disorders, 22(6), 500–504.	Growth	Requires update
West et al. (2013) Patterns of growth in ambulatory males with Duchenne muscular dystrophy. Journal of Pediatrics, 163(6), 1759–1763 e1751.	Growth	Requires update
Sarrazin et al. (2014) Sarrazin, E., Hagen, M. V., Schara, U., von Au, K., & Kaindl, A. M. (2014). Growth and psychomotor development of patients with Duchenne muscular dystrophy. European Journal of Paediatric Neurology, 18(1), 38–44.	Growth	Promising research
Skalsky et al. (2009) Assessment of regional body composition with dual-energy X-ray absorptiometry in Duchenne muscular dystrophy: Correlation of regional lean mass and quantitative strength. Muscle & Nerve, 39(5), 647–651.	Obesity	Promising research
Bayram et al. (2013) Correlation between motor performance scales, body composition, and anthropometry in patients with Duchenne muscular dystrophy. Acta Neurologica Belgica, 113(2), 133–137.	Obesity	Requires update
Bach et al. (2010) Open gastrostomy for noninvasive ventilation users with neuromuscular disease. American Journal of Physical Medicine and Rehabilitation, 89(1), 1–6.	Gastrostomy	Requires update
Martigne et al. (2010) Efficacy and tolerance of gastrostomy feeding in Duchenne muscular dystrophy. Clinical Nutrition, 29(1), 60–64.	Gastrostomy	Requires update
P Mizuno et al. (2012) Efficacy and tolerance of gastrostomy feeding in Japanese muscular dystrophy patients. Brain & Development, 34(9), 756–762.	Gastrostomy	Requires update
Bianchi et al. (2011) Low bone density and bone metabolism alterations in Duchenne muscular dystrophy: response to calcium and vitamin D treatment. Osteoporosis International, 22(2), 529–539.	Pharmacology	Requires update
Rutter et al. (2012) Growth hormone treatment in boys with Duchenne muscular dystrophy and glucocorticoid-induced growth failure. Neuromuscular Disorders, 22(12), 1046–1056.	Pharmacology	Promising research
Banerjee et al. (2010) Effect of creatine monohydrate in improving cellular energetics and muscle strength in ambulatory Duchenne muscular dystrophy patients: A randomized, placebo-controlled 31P MRS study. Magnetic Resonance Imaging, 28(5), 698–707.	Pharmacology	Promising research
Spurney et al. (2011) CINRG pilot trial of coenzyme Q10 in steroid-treated Duchenne muscular dystrophy. Muscle & Nerve, 44(2), 174–178.	Pharmacology	Promising research
Mok et al. (2006) Oral glutamine and amino acid supplementation inhibit whole-body protein degradation in children with Duchenne muscular dystrophy. American Journal of Clinical Nutrition, 83(4), 823–828.	Pharmacology	Promising research
Mok et al. (2009) Lack of functional benefit with glutamine versus placebo in Duchenne muscular dystrophy: a randomized crossover trial. PLoS One, 4(5), e5448.	Pharmacology	Requires update
Casteels et al. (2010) Metformin therapy to reduce weight gain and visceral adiposity in children and adolescents with neurogenic or myogenic motor deficit. Pediatric Diabetes, 11(1), 61–69.	Pharmacology	Promising research
Radley et al. (2007) Duchenne muscular dystrophy: Focus on pharmaceutical and nutritional interventions. International Journal of Biochemistry & Cell Biology, 39(3), 469–477.	Review	Promising research
Tarnopolsky (2007) Clinical use of creatine in neuromuscular and neurometabolic disorders. Subcellular Biochemistry, 46, 183–204. [Abstract Only]	Review	Promising research
Davidson and Truby (2009) A review of nutrition in Duchenne muscular dystrophy. Journal of Human Nutrition and Dietetics, 22(5), 383–393.	Review	Promising research
Kley, R. A., et al. (2013) "Creatine for treating muscle disorders." Cochrane Database Syst Rev 6: Cd004760.	Review	Promising research

Neuromuscular

The neuromuscular section will not require an extensive update. Many of the articles confirm what is already recommended in the care considerations. The reference list will need to be updated with the new supportive literature. There is a lot of promising research around treatments and imaging that may be considered in future updates.
The theme of the glucocorticosteroid articles show heterogeneity in methods of using corticosteroids. They are not specific to dose and age. More information on predominate measures (age and dose) is needed. Several articles discuss the benefits of long term corticosteroid use and the side effects, but there were no new side effects identified. Caregivers may want information on how to effectively manage side effects, therefore it is recommended by the clinical experts to include identified side effects in the care considerations and how to care for those side effects. Recommendations for test, measures, and management in patients younger than 6 should also be included in the care considerations once more data is available.

Neuromuscular		
Study	**Sub-Topic**	**Recommendation**
Banerjee et al. (2010). Effect of creatine monohydrate in improving cellular energetics and muscle strength in ambulatory Duchenne muscular dystrophy patients: A randomized, placebo-controlled 31P MRS study. Magnetic Resonance Imaging, 28(5), 698–707. doi:10.1016/j.mri.2010.03.008	Dietary Supplements	Requires Update
Mok et al. (2009) Lack of functional benefit with glutamine versus placebo in Duchenne muscular dystrophy: A randomized crossover trial. PLoS One, 4(5), e5448. doi:10.1371/journal.pone.0005448	Dietary Supplements	Requires Update
Kinali et al. (2009) Local restoration of dystrophin expression with the morpholino oligomer AVI-4658 in Duchenne muscular dystrophy: A single-blind, placebo-controlled, dose-escalation, proof-of-concept study. Lancet Neurology, 8(10), 918–928. doi:10.1016/s1474-4422(09)70211-x	DMD Treatments	Promising Research
Cirak et al. (2011) Exon skipping and dystrophin restoration in patients with Duchenne muscular dystrophy after systemic phosphorodiamidate morpholino oligomer treatment: An open-label, phase 2, dose-escalation study. Lancet, 378(9791), 595–605. doi:10.1016/s0140-6736(11)60756-3	DMD Treatments	Promising Research
Buyse et al. (2011) Idebenone as a novel, therapeutic approach for Duchenne muscular dystrophy: Results from a 12 month, double-blind, randomized placebo-controlled trial. Neuromuscular Disorders, 21(6), 396–405. doi:10.1016/j.nmd.2011.02.016	DMD Treatments	Promising Research
Kirschner et al. (2010) Treatment of Duchenne muscular dystrophy with ciclosporin A: A randomised, double-blind, placebo-controlled multicentre trial. Lancet Neurology, 9(11), 1053–1059. doi:10.1016/s1474-4422(10)70196-4	DMD Treatments	Promising Research
Kim et al. (2010) Quantitative assessment of the T2 relaxation time of the gluteus muscles in children with Duchenne muscular dystrophy: A comparative study before and after steroid treatment. Korean Journal of Radiology, 11(3), 304–311. doi:10.3348/kjr.2010.11.3.304	Imaging	Promising Research
Mathur et al. (2010) Age-related differences in lower-limb muscle cross-sectional area and torque production in boys with Duchenne muscular dystrophy. Archives of Physical Medicine and Rehabilitation, 91(7), 1051–1058. doi:10.1016/j.apmr.2010.03.024	Imaging	Promising Research
Weber et al. (2011) Sodium (23Na) MRI detects elevated muscular sodium concentration in Duchenne muscular dystrophy. Neurology, 77(23), 2017–2024. doi:10.1212/WNL.0b013e31823b9c78	Imaging	Promising Research
Gaeta et al. (2012) Muscle fat-fraction and mapping in Duchenne muscular dystrophy: evaluation of disease distribution and correlation with clinical assessments. Preliminary experience. Skeletal Radiology, 41(8), 955–961. doi:10.1007/s00256-011-1301-5	Imaging	Promising Research

Torriani et al. (2012) Lower leg muscle involvement in Duchenne muscular dystrophy: An MR imaging and spectroscopy study. Skeletal Radiology, 41(4), 437–445. doi:10.1007/s00256-011-1240-1	Imaging	Promising Research
Weber et al. (2012) Permanent muscular sodium overload and persistent muscle edema in Duchenne muscular dystrophy: A possible contributor of progressive muscle degeneration. Journal of Neurology, 259(11), 2385–2392. doi:10.1007/s00415-012-6512-8	Imaging	Promising Research
Hollingsworth et al. (2013) Magnetic resonance imaging in Duchenne muscular dystrophy: Longitudinal assessment of natural history over 18 months. Muscle & Nerve, 48(4), 586–588. doi:10.1002/mus.23879	Imaging	Promising Research
Jansen et al. (2012) Quantitative muscle ultrasound (QMUS) is a promising longitudinal follow-up tool in Duchenne muscular dystrophy. Neuromuscular Disorders, 22(4), 306–317. doi:10.1016/j.nmd.2011.10.020	Imaging	Promising Research
Zaidman et al. (2010) Quantitative ultrasound using backscatter analysis in Duchenne and Becker muscular dystrophy. Neuromuscular Disorders, 20(12), 805–809. doi:10.1016/j.nmd.2010.06.019	Imaging	Promising Research

Rehabilitation Management

Clinical experts agreed that the rehabilitation management section will only require a summary update. Not all reviews or ratings are needed. Subtopics that require updating include test and measures and assistive upper extremity devices. All supportive references should be updated.

Rehabilitation Management		
Study	**Sub-Topic**	**Recommendation**
Nozaki et al.(2010). Range of motion exercise of temporo-mandibular joint with hot pack increases occlusal force in patients with Duchenne muscular dystrophy. Acta Myologica, 29(3), 392–397.	Therapy	Promising Research
Jansen et al. (2010). Physical training in boys with Duchenne Muscular Dystrophy: The protocol of the No Use is Disuse study. BMC Pediatrics, 10, 55. doi:10.1186/1471-2431-10-55	Exercise/Recreation	Promising Research
Jansen et al. (2013) "Assisted bicycle training delays functional deterioration in boys with Duchenne muscular dystrophy: the randomized controlled trial "no use is disuse"." Neurorehabil Neural Repair 27(9): 816-827.	Exercise/Recreation	Promising Research
Soderpalm et al. (2013). Whole body vibration therapy in patients with Duchenne muscular dystrophy—a prospective observational study. Journal of Musculoskeletal & Neuronal Interactions, 13(1), 13–18.	Exercise/Recreation	Promising Research
Pellegrini et al. (2007). Hand versus mouth for call-bell activation by DMD and Becker patients. Neuromuscular Disorders, 17(7), 532–536. doi:10.1016/j.nmd.2007.03.016	Assistive/Adaptive Devices	Requires Update
Kumar, A., & Phillips, M. F. (2013). Use of powered mobile arm supports by people with neuromuscular conditions. Journal of Rehabilitation Research and Development, 50(1), 61–70.	Assistive/Adaptive Devices	Requires Update
Garralda et al. (2006) Knee-ankle-foot orthosis in children with duchenne muscular dystrophy: User views and adjustment. European Journal of Paediatric Neurology, 10(4), 186–191. doi:10.1016/j.ejpn.2006.07.002	Orthoses	Requires Update
Main et al. (2007) Serial casting of the ankles in Duchenne muscular dystrophy: Can it be an alternative to surgery? Neuromuscular Disorders, 17(3), 227–230. doi:10.1016/j.nmd.2006.12.002	Orthoses	Requires Update
Glanzman, A. M., et al. (2011). "Serial casting for the management of ankle contracture in Duchenne muscular dystrophy." Pediatric Phys Ther 23(3): 275-279.	Orthoses	Requires Update

Bartels et al. (2011) Upper limb function in adults with Duchenne muscular dystrophy. Journal of Rehabilitation Medicine, 43(9), 770–775. doi:10.2340/16501977-0841	Muscle Strength	Requires Update
Wren et al. (2008) Three-point technique of fat quantification of muscle tissue as a marker of disease progression in Duchenne muscular dystrophy: Preliminary study. American Journal of Roentgenology, 190(1), W8–12. doi:10.2214/AJR.07.2732	Muscle Strength	Promising Research
Angelini, C., & Tasca, E. (2012). Fatigue in muscular dystrophies. Neuromuscular Disorders, 22 Suppl 3, S214–220. doi:10.1016/j.nmd.2012.10.010	Muscle Strength	Requires Update
Skalsky et al. (2009) Assessment of regional body composition with dual-energy X-ray absorptiometry in Duchenne muscular dystrophy: Correlation of regional lean mass and quantitative strength. Muscle & Nerve, 39(5), 647–651. doi:10.1002/mus.21212	Muscle Strength	Promising Research
Zebracki, K., & Drotar, D. (2008). Pain and activity limitations in children with Duchenne or Becker muscular dystrophy. Developmental Medicine and Child Neurology, 50(7), 546–552. doi:10.1111/j.1469-8749.2008.03005.x	Functional Assessment	Requires Update
Lue et al. (2009). Measurement of the functional status of patients with different types of muscular dystrophy. The Kaohsiung Journal of Medical Sciences, 25(6), 325–333. doi:10.1016/s1607-551x(09)70523-6	Functional Assessment	Promising Research
Akima et al. (2012) Relationships of thigh muscle contractile and non-contractile tissue with function, strength, and age in boys with Duchenne muscular dystrophy. Neuromuscular Disorders, 22(1), 16–25. doi:10.1016/j.nmd.2011.06.750	Functional Assessment	Promising Research
Vuillerot et al. (2010) Monitoring changes and predicting loss of ambulation in Duchenne muscular dystrophy with the Motor Function Measure. Developmental Medicine and Child Neurology, 52(1), 60–65. doi:10.1111/j.1469-8749.2009.03316	Functional Assessment	Promising Research
Diniz et al. (2012) . Motor assessment in patients with Duchenne muscular dystrophy. Arquivos de Neuro-Psiquiatria, 70(6), 416–421	Functional Assessment	Promising Research
Silva et al. (2012) Motor function measure scale, steroid therapy and patients with Duchenne muscular dystrophy. Arquivos de Neuro-Psiquiatria, 70(3), 191–195.	Functional Assessment	Promising Research
Vuillerot et al. (2012) Responsiveness of the motor function measure in neuromuscular diseases. Archives of Physical Medicine and Rehabilitation, 93(12), 2251–2256 e2251. doi:10.1016/j.apmr.2012.05.025	Functional Assessment	Promising Research
Connolly et al. (2013) Motor and cognitive assessment of infants and young boys with Duchenne Muscular Dystrophy: results from the Muscular Dystrophy Association DMD Clinical Research Network. Neuromuscular Disorders, 23(7), 529-539. doi: 10.1016/j.nmd.2013.04.005	Functional Assessment	Promising Research
Mazzone et al. (2010) North Star Ambulatory Assessment, 6-minute walk test and timed items in ambulant boys with Duchenne muscular dystrophy. Neuromuscular Disorders, 20(11), 712–716. doi:10.1016/j.nmd.2010.06.014	Functional Assessment	Require Update
McDonald et al. (2010) The 6-minute walk test as a new outcome measure in Duchenne muscular dystrophy. Muscle & Nerve, 41(4), 500–510. doi:10.1002/mus.21544	Functional Assessment	Requires Update
McDonald et al. (2010) The 6-minute walk test in Duchenne/Becker muscular dystrophy: Longitudinal observations. Muscle & Nerve, 42(6), 966–974. doi:10.1002/mus.21808	Functional Assessment	Requires Update
McDonald et al. (2013) The 6-minute walk test and other clinical endpoints in duchenne muscular dystrophy: Reliability, concurrent validity, and minimal clinically important differences from a multicenter study. Muscle & Nerve, 48(3), 357–368. doi:10.1002/mus.23905	Functional Assessment	Requires Update
McDonald et al. (2013) The 6-minute walk test and other endpoints in Duchenne muscular dystrophy: Longitudinal natural history	Functional Assessment	Requires Update

observations over 48 weeks from a multicenter study. Muscle & Nerve, 48(3), 343–356. doi:10.1002/mus.23902		
Jansen et al. (2012) The assisted 6-minute cycling tests to assess endurance in children with a neuromuscular disorder. Muscle & Nerve, 46(4), 520–530. doi:10.1002/mus.23369	Functional Assessment	Requires Update
Jansen et al. (2013) Assisted bicycle training delays functional deterioration in boys with Duchenne muscular dystrophy: The randomized controlled trial "No Use Is Disuse." Neurorehabilitation and Neural Repair, 27(9), 816–827. doi: 10.1177/1545968313496326	Functional Assessment	Requires Update

Orthopedic and Surgical

Extensive update is not required for the orthopedic section. However, there was a significant amount of new literature that supports the current care considerations and also some new information for subtopics requiring an update to the orthopedic section. Supportive literature will require updating of the references. Specific areas requiring update are effects of steroids in scoliosis reduction, information on dental care, and literature on spine fusion and pulmonary effects in terms of outcomes is an area of interest. The natural history of development of scoliosis in corticosteroid treated individuals is changing (incidence as well as progression).The recommendation about when to consider surgery for correction of scoliosis are still in flux; this may be in conflict with current recommendations and should be discussed further by clinical experts. The issue of incidence and detection of vertebral fractures in corticosteroid treated individuals remain controversial, and requires further discussion. Anesthesia articles were not rated by the committee and it is recommended that an anesthesiologist is added to the steering committee to provide further guidance on this topic area. It was also recommended that an orthopedic surgeon be added to the steering committee.

Orthopedic/Surgical		
Study	**Sub-Topic**	**Recommendation**
Dooley, J. M., et al. (2010). "Impact of steroids on surgical experiences of patients with Duchenne muscular dystrophy." Pediatric Neurol 43(3): 173-176	Surgical	Promising Research
Mielnik-Blaszczak, M. and B. Malgorzata (2007). "Duchenne muscular dystrophy--a dental healthcare program." Spec Care Dentist 27(1): 23-25.	Dental	Requires Update
Morel-Verdebout, C., et al. (2007) "Dentofacial characteristics of growing patients with Duchenne muscular dystrophy: a morphological study." Eur J Orthod 29(5): 500-507.	Dental	Requires Update
Balasubramaniam, R., et al. (2008). "Oral health considerations in muscular dystrophies." Spec Care Dentist 28(6): 243-253.	Dental	Requires Update
Boyce, A.M., et al. (2014) Treatment for children with Disabling conditions. PMR 2014; 6:425-436.	Review	Promising Research

Psychosocial

The consensus of the clinical experts is that there is not much new literature that would require an extensive update to the psychosocial section. Most literature only confirms/supports what is already in the current care considerations. All new supportive references should be updated. New information on transition and independent living beginning with adolescence should be highlighted in the update as many patients with DMD are living longer. Palliative and respite care should be emphasized and the need for evidence on palliative care and support for families highlighted. Behavior and cognition are also areas of interest that should be discussed further. Further discussion is needed to determine if new information regarding adolescent and parent perception of quality of life and burden on caregiver should be included in the updated care considerations. The psychiatric section may require some modifications in the updated guidelines.

Psychosocial		
Study	**Sub-Topic**	**Recommendation**
Wingeier, K., et al. (2011). "Neuropsychological impairments and the impact of dystrophin mutations on general cognitive functioning of patients with Duchenne muscular dystrophy." J Clin Neurosci 18(1): 90-95.	Neuropsychological/Developmental Assessment	Promising Research
Mento, G., et al. (2011). "The neuropsychological profile of infantile Duchenne muscular dystrophy." Clin Neuropsychol 25(8): 1359-1377.	Neuropsychological/Developmental Assessment	Promising Research
D'Angelo, M. G., et al. (2011). "Neurocognitive profiles in Duchenne muscular dystrophy and gene mutation site." Pediatr Neurol 45(5): 292-299.	Neuropsychological/Developmental Assessment	Promising Research
Taylor, P. J., et al. (2010). "Dystrophin gene mutation location and the risk of cognitive impairment in Duchenne muscular dystrophy." PLoS One 5(1): e8803.	Neuropsychological/Developmental Assessment	Promising Research
Garralda, M. E., et al. (2013). "Emotional impact of genetic trials in progressive pediatric disorders: a dose-ranging exon-skipping trial in Duchenne muscular dystrophy." Child Care Health Dev 39(3): 449-455.	Psychosocial Assessment	Promising Research
Pane, M., et al. (2012). "Attention deficit hyperactivity disorder and cognitive function in Duchenne muscular dystrophy: phenotype-genotype correlation." J Pediatr 161(4): 705-709 e701.	Psychotherapy	Promising Research
Miller, A. R., et al. (2009). "Continuity of care for children with complex chronic health conditions: parents' perspectives." BMC Health Serv Res 9: 242.	Care Intervention	Requires Update
Carter, G. T., et al. (2012). "Using palliative care in progressive neuromuscular disease to maximize quality of life." Phys Med Rehabil Clin N Am 23(4): 903-909.	Review	Requires Update

Diagnosis

The diagnosis section will not require an extensive update. Very little has changed in this area, but the new literature is interesting and presents promising research and should be highlighted in the update for future reference. Other areas identified for further review and potential notation in the recommendations are prenatal testing, next generation sequencing (which will be important to molecular diagnostics in the future), and updates to full characterization mutation references. There will be more to come about creatinine kinase screening in newborns in future years but current literature does not support a change to the current statement in the recommendations.

Diagnosis		
Study	**Sub-Topic**	**Recommendation**
Laing, N. G. (2012). Genetics of neuromuscular disorders. *Critical Reviews in Clinical Laboratory Sciences*, *49*(2), 33–48. doi:10.3109/10408363.2012.658906	Clinical History	Promising Research
Mercier et al. (2013). Genetic and clinical specificity of 26 symptomatic carriers for dystrophinopathies at pediatric age. European Journal of Human Genetics, 21(8), 855–863. doi:10.1038/ejhg.2012.269.	Clinical History	Requires update
Hiraki et al. (2006) Attitudes of genetic counselors towards expanding newborn screening and offering predictive genetic testing to children. American Journal of Medical Genetics. Part A, 140(21), 2312–2319. doi:10.1002/ajmg.a.31485	Newborn Screening	Promising Research
Kemper, A. R., & Wake, M. A. (2007). Duchenne muscular dystrophy: issues in expanding newborn screening. Current Opinion in Pediatrics, 19(6), 700–704. doi:10.1097/MOP.0b013e3282f19f65	Newborn Screening	Promising Research

Taylor et al. (2007) Measurement of the clinical utility of a combined mutation detection protocol in carriers of Duchenne and Becker muscular dystrophy. Journal of Medical Genetics, 44(6), 368–372. doi:10.1136/jmg.2006.047464	Newborn Screening	Promising Research
Mendell et al. (2012) dystrophy. Annals of Neurology, 71(3), 304–313. doi:10.1002/ana.23528	Newborn Screening	Promising Research
Vasli et al. (2012) Next generation sequencing for molecular diagnosis of neuromuscular diseases. Acta Neuropathologica, 124(2), 273–283. doi:10.1007/s00401-012-0982-8	Genetic Testing	Promising Research
Huang et al. (2007) Rapid prenatal diagnosis of Duchenne muscular dystrophy with gene duplications by ion-pair reversed-phase high-performance liquid chromatography coupled with competitive multiplex polymerase chain reaction strategy. Prenatal Diagnosis, 27(7), 653–656. doi:10.1002/pd.173.	Prenatal Testing	Promising Research
Malayeri et al. (2011) Detection of Duchenne/Becker muscular dystrophy carriers in a group of Iranian families by linkage analysis. Acta Medica Iranica, 49(3), 142–148.	Prenatal Testing	Promising Research
Helderman-van den Enden et al. (2013) An urgent need for a change in policy revealed by a study on prenatal testing for Duchenne muscular dystrophy. European Journal of Human Genetics, 21(1), 21–26. doi:10.1038/ejhg.2012.101	Prenatal Testing	Promising Research
Yang et al. (2013) MLPA-based genotype-phenotype analysis in 1053 Chinese patients with DMD/BMD. BMC Medical Genetics, 14, 29. doi:10.1186/1471-2350-14-29	Prenatal Testing	Promising Research

Respiratory

The majority of the new literature agrees with and supports the current care considerations; no studies require a change to the current recommendations but allow subtopics that are vague to be expanded upon. The references will need to be updated for the supportive literature. Several themes and interesting topics are seen in the new literature regarding methods for pulmonary function (i.e. breath-stacking) and respiratory treatments that are not in the original care considerations. Many articles discussing upcoming specialty treatments; a specialty article on this topic is recommended by the clinical experts.

There may be a need to point out new themes in the literature and explain why alternative methods are not included in the primary guidelines or to recommend an update to the specialty article in Pediatric Pulmonology. Discussion could include what happens when a person with DMD gets sick or undergoes surgery and future areas that need more study. It could also speak on the direction the field is taking, as pointed out in some of the articles. Future research and discussion is also needed on articles on cardiopulmonary phenotypic variability before incorporation into the care consideration. This topic will affect multiple sections and should be considered promising research for future review.

Respiratory		
Study	**Sub-Topic**	**Recommendation**
Kang and Kang (2006) . Respiratory muscle strength and cough capacity in patients with Duchenne muscular dystrophy. *Yonsei Medical Journal*, *47*(2), 184–190.	Assisted Cough	Requires Update
Bach et al. (2007). Lung inflation by glossopharyngeal breathing and "air stacking" in Duchenne muscular dystrophy. American Journal of Physical Medicine and Rehabilitation, 86(4), 295–300. doi:10.1097/PHM.0b013e318038d1ce	Volume Recruitment	Requires Update
Ishikawa et al. (2008). Cough augmentation in Duchenne muscular dystrophy. American Journal of Physical Medicine and Rehabilitation, 87(9), 726–730. doi:10.1097/PHM.0b013e31817f99a8	Volume Recruitment	Requires Update
Brito, M. F., et al. (2009). Air stacking and chest compression increase peak cough flow in patients with Duchenne muscular dystrophy. Jornal Brasileiro de Pneumologia, 35(10), 973–979.	Volume Recruitment	Requires Update

Toussaint et al. (2006). Diurnal ventilation via mouthpiece: survival in end-stage Duchenne patients. European Respiratory Journal, 28(3), 549–555. doi:10.1183/09031936.06.00004906	Volume Recruitment	Requires Update
Dohna-Schwake et al. (2006). IPPB-assisted coughing in neuromuscular disorders. Pediatric Pulmonology, 41(6), 551–557. doi:10.1002/ppul.20406	Volume Recruitment	Requires Update
Nicot et al. (2006). Respiratory muscle testing: A valuable tool for children with neuromuscular disorders. American Journal of Respiratory and Critical Care Medicine, 174(1), 67–74. doi:10.1164/rccm.200512-1841OC	Respiratory Muscle Strength	Requires Update
Matsumura et al. (2012). Lung inflation training using a positive end-expiratory pressure valve in neuromuscular disorders. Internal Medicine, 51(7), 711–716.	Respiratory Muscle Strength	Requires Update
Bersanini et al. (2012). Nocturnal hypoxaemia and hypercapnia in children with neuromuscular disorders. European Respiratory Journal, 39(5), 1206–1212. doi:10.1183/09031936.00087511	Daytime Monitoring	Conflict
Romei et al. (2012). Low abdominal contribution to breathing as daytime predictor of nocturnal desaturation in adolescents and young adults with Duchenne muscular dystrophy. Respiratory Medicine, 106(2), 276–283. doi:10.1016/j.rmed.2011.10.010	Daytime Monitoring	Requires Update
Brunherotti et al. (2007). Correlations of Egen Klassifikation and Barthel Index scores with pulmonary function parameters in Duchenne muscular dystrophy. Heart & Lung, 36(2), 132–139. doi:10.1016/j.hrtlng.2006.07.006	Vital Capacity	Requires Update
Hahn et al. (2009). Noninvasive determination of the tension-time index in Duchenne muscular dystrophy. American Journal of Physical Medicine and Rehabilitation, 88(4), 322–327. doi:10.1097/PHM.0b013e3181909dfa	Vital Capacity	Requires Update
Hamada et al. (2011). Indicators for ventilator use in Duchenne muscular dystrophy. Respiratory Medicine, 105(4), 625–629. doi:10.1016/j.rmed.2010.12.005	Vital Capacity	Promising Research
Humbertclaude et al. (2012). Motor and respiratory heterogeneity in Duchenne patients: Implication for clinical trials. European Journal of Paediatric Neurology, 16(2), 149–160. doi:10.1016/j.ejpn.2011.07.001	Vital Capacity	Requires Update
Birnkrant et al. (2010). The respiratory management of patients with duchenne muscular dystrophy: A DMD care considerations working group specialty article. Pediatric Pulmonology, 45(8), 739–748. doi:10.1002/ppul.21254	Review	Requires Update
Dreyer, P. S., et al. (2009). Distanciation in Ricoeur's theory of interpretation: Narrations in a study of life experiences of living with chronic illness and home mechanical ventilation. Nursing Inquiry, 16(1), 64–73. doi:10.1111/j.1440-1800.2009.00433.x	Review	Requires Update
Birnkrant DJ, Ashwath ML, Noritz GH, Merrill MC et a. Cardiac and pulmonary function variability in Duchenne/Becker muscular dystrophy: an initial report. J Child Neurol 2010; 25: 1110-1115.	Phenotypic Variability	Promising Research
Rideau Y, Jankowski LW, Grellet J. Respiratory function in the muscular dystrophies. Muscle and Nerve 1981; 4: 155-164.	Phenotypic Variability	Promising Research
Humbertclaude V, Hamroun D,Bezzou K, Berard C, et al. Motor and respiratory heterogeneity in Duchenne patients: implications for clinical trials. Eur J Paediatr Neurol 2012; 16: 149-160.	Phenotypic Variability	Promising Research
Humbertclaude V, Hamroun D, Picot MC, Bezzou K, Berard C, et al. Phenotypic heterogeneity and phenotype-genotype correlations in dystrophinopathies: Contribution of genetic and clinical databases. Rev Neurol (Paris) 2013; 169: 583-594.	Phenotypic Variability	Promising Research
Swaggart KA, McNally EM. Modifiers of heart and muscle function: where genetics meets physiology. Exp Physio 2014; 99: 621-626.	Phenotypic Variability	Promising Research
Mahi L. Ashwath ML, Jacobs IB, Crowe CA, Ashwath RC, Super DM, Bahler RC. Left Ventricular Dysfunction in Duchenne Muscular	Phenotypic Variability	Promising Research

Dystrophy and Genotype. Am J Cardiol http://dx.doi.org/10.1016/j.amjcard.2014.04.038		
Desguerre K, Christov C, Mayer M, et al. Clinical heterogeneity of Duchenne muscular dystrophy (DMD): definition of sub-phenotypes and predictive criteria by long-term follow-up. PLoS ONE 2009; 4: e4347.	Phenotypic Variability	Promising Research
Hyser CL, Province M, Griggs RC, et al. Genetic heterogeneity in Duchenne dystrophy. Ann Neurol. 1987; 22: 553-555.	Phenotypic Variability	Promising Research
Sifringer M, Uhlenberg B, Lammel S, et al. Identification of transcripts from a subtraction library which might be responsible for the mild phenotype in an intrfamilially variable course of Duchenne muscular dystrophy. Hum Genet 2004; 114: 149-156	Phenotypic Variability	Promising Research
Pettygrove S, Lu Z, Andrews JG, Meaney FJ, Sheehan DW et al. Sibling concordance for clinical features of Duchenne and Becker muscular dystrophies. Muscle and Nerve DOI: 10.1002/mus.24078	Phenotypic Variability	Promising Research

Cardiovascular

The consensus of the cardiovascular experts is that this section will require an update to the recommendations. There were a lot of cardiac MRI articles that require an update to the recommendations. These should also be considered promising research as they show that we can detect cardiac abnormalities, but do not demonstrate an impact on short term or long term outcomes. Additional subtopics that need to be addressed in the care considerations are cardiac MRI, cardiac transplant, devices, and the effects of steroid use.

Cardiovascular		
Study	**Sub-Topic**	**Recommendation**
Kajimoto, H., K. Ishigaki, et al. (2006) Beta-blocker therapy for cardiac dysfunction in patients with muscular dystrophy." Circ J 70(8): 991-994.	Pharmacology	Conflicts
Duboc, D., et al. (2007) Perindopril preventive treatment on mortality in Duchenne muscular dystrophy: 10 years' follow-up." Am Heart J 154(3): 596-602.	Pharmacology	Requires update
Markham, L. W., et al. (2008) Corticosteroid treatment retards development of ventricular dysfunction in Duchenne muscular dystrophy." Neuromuscular Disorder 18(5): 365-370	Pharmacology	Requires update
Connuck, D. M., L. A. Sleeper, et al. (2008) Characteristics and outcomes of cardiomyopathy in children with Duchenne or Becker muscular dystrophy: a comparative study from the Pediatric Cardiomyopathy Registry." Am Heart J 155(6): 998-1005.	Cardiac Transplant	Requires update
Wu, R. S., S. Gupta, et al. (2010) Clinical outcomes after cardiac transplantation in muscular dystrophy patients." J Heart Lung Transplant 29(4): 432-438.	Cardiac Transplant	Requires update
Silva, M. C., Z. M. Meira, et al. (2007) Myocardial delayed enhancement by magnetic resonance imaging in patients with muscular dystrophy." J Am Coll Cardiol 49(18): 1874-1879.	Cardiac MRI	Requires update
Yilmaz, A., H. J. Gdynia, et al. (2008) "Cardiac involvement in patients with Becker muscular dystrophy: new diagnostic and pathophysiological insights by a CMR approach." J Cardiovascular Magn Reson 10: 50.	Cardiac MRI	Requires update
Hor, K. N., J. Wansapura, et al. (2009) Circumferential strain analysis identifies strata of cardiomyopathy in Duchenne muscular dystrophy: a cardiac magnetic resonance tagging study." J Am Coll Cardiol 53(14): 1204-1210.	Cardiac MRI	Requires update
Puchalski, M. D., R. V. Williams, et al. (2009) Late gadolinium enhancement: precursor to cardiomyopathy in Duchenne muscular dystrophy?" Int J Cardiovascular Imaging 25(1): 57-63.	Cardiac MRI	Requires update
Hagenbuch, S. C., W. M. Gottliebson, et al. (2010) Detection of progressive cardiac dysfunction by serial evaluation of circumferential	Cardiac MRI	Requires update

strain in patients with Duchenne muscular dystrophy." Am J Cardiol 105(10): 1451-1455.		
Hor, K. N., W. M. Gottliebson, et al. (2010) Comparison of magnetic resonance feature tracking for strain calculation with harmonic phase imaging analysis." JACC Cardiovascular Imaging 3(2): 144-151.	Cardiac MRI	Requires update
Mavrogeni, S., A. Papavasiliou, et al. (2010) Myocardial inflammation in Duchenne Muscular Dystrophy as a precipitating factor for heart failure: a prospective study." BMC Neurol 10: 33.	Cardiac MRI	Requires update
Wansapura, J. P., K. N. Hor, et al. (2010) "Left ventricular T2 distribution in Duchenne muscular dystrophy." J Cardiovascular Magn Reson 12: 14.	Cardiac MRI	Requires update
Hor, K. N., et al. (2011) Hor, K. N., et al. (2011). "Effects of steroids and angiotensin converting enzyme inhibition on circumferential strain in boys with Duchenne muscular dystrophy: a cross-sectional and longitudinal study utilizing cardiovascular magnetic resonance." J Cardiovascular Magn Reson 13: 60.	Cardiac MRI	Requires update
Bilchick, K. C., M. Salerno, et al. (2011) Prevalence and distribution of regional scar in dysfunctional myocardial segments in Duchenne muscular dystrophy." J Cardiovascular Magn Reson 13: 20.	Cardiac MRI	Requires update
Walcher, T., P. Steinbach, et al. (2011) "Detection of long-term progression of myocardial fibrosis in Duchenne muscular dystrophy in an affected family: a cardiovascular magnetic resonance study." Eur J Radiol 80(1): 115-119.	Cardiac MRI	Requires update
Mazur, W., K. N. Hor, et al. (2012) Patterns of left ventricular remodeling in patients with Duchenne Muscular Dystrophy: a cardiac MRI study of ventricular geometry, global function, and strain." Int J Cardiovascular Imaging 28(1): 99-107.	Cardiac MRI	Requires update

Primary Care and Emergency Care

The primary care and emergency care will require a full extensive update to the care considerations. It was agreed by clinical experts that it should be a standalone section. Recommendation is that more research is needed around preventive care to aid with care considerations. A reference to CDC immunizations guidelines should be included in this section. Renal, urological and dental care possibilities are also important subtopics to address in this section.

Current care considerations have a brief mention of the existence of palliative. Updated DMD care considerations should better define what palliative care is to remove the association with end of life and explain the benefits. Respite care is also a huge topic to consider. There is not a lot of evidence on the continuity of care, but clinical experts recommend that it is discussed in this section as well. Clinical experts also recommend that we include what to look for when DMD patients arrive in ER.

Primary Care and Emergency Care		
Study	**Sub-Topic**	**Recommendation**
PRACTICE, C. O. and B. F. P. S. W. AMBULATORY MEDICINE (2014). "2014 Recommendations for Pediatric Preventive Health Care." Pediatrics 133(3): 568-570.	Preventive Care	Requires Update
McMillan, H. J., et al. (2010). "Duchenne muscular dystrophy: Canadian pediatric neuromuscular physician's survey." Can J Neurol Sci 37(2): 195-205.	Immunization	Requires Update
Singh, M., et al. (2007). "Nephrolithiasis in patients with Duchenne muscular dystrophy." Urology 70(4): 643-645.	Renal	Requires Update
Shumyatcher Y, et al. (2008) Symptomatic nephrolithiasis in prolonged survivors of Duchenne muscular dystrophy. Neuromuscul Disord. 18: 561-4.	Renal	Requires Update
Askeland, E. J., et al. (2013). "Urological manifestations of Duchenne muscular dystrophy." J Urol 190(4 Suppl): 1523-1528.	Renal	Requires Update

Mielnik-Blaszczak, M. and B. Malgorzata (2007). "Duchenne muscular dystrophy--a dental healthcare program." Spec Care Dentist 27(1): 23-25.	Dental	Requires Update
Morel-Verdebout, C., et al. (2007) "Dentofacial characteristics of growing patients with Duchenne muscular dystrophy: a morphological study." Eur J Orthod 29(5): 500-507.	Dental	Requires Update
Balasubramaniam, R., et al. (2008). "Oral health considerations in muscular dystrophies." Spec Care Dentist 28(6): 243-253.	Dental	Requires Update
Arias, R., et al. (2011). "Palliative care services in families of males with Duchenne muscular dystrophy." Muscle Nerve 44(1): 93-101.	Palliative/Respite Care	Requires Update
Fraser, L. K., et al. (2011). "Hospice provision and usage amongst young people with neuromuscular disease in the United Kingdom." Eur J Paediatr Neurol 15(4): 326-330.	Palliative/Respite Care	Requires Update
Carter, G. T., et al. (2012). "Using palliative care in progressive neuromuscular disease to maximize quality of life." Phys Med Rehabil Clin N Am 23(4): 903-909.	Palliative/Respite Care	Requires Update
Fraser, L. K., et al. (2012). "A cohort study of children and young people with progressive neuromuscular disorders: clinical and demographic profiles and changing patterns of referral for palliative care." Palliat Med 26(7): 924-929.	Palliative/Respite Care	Requires Update
Feudtner, C. and P. G. Nathanson (2014). "Pediatric Palliative Care and Pediatric Medical Ethics: Opportunities and Challenges." Pediatrics 133(Supplement 1): S1-S7.	Palliative/Respite Care	Requires Update
Mercurio, M. R., et al. (2014). "Unilateral Pediatric "Do Not Attempt Resuscitation" Orders: The Pros, the Cons, and a Proposed Approach." Pediatrics 133(Supplement 1): S37-S43.	Palliative/Respite Care	Requires Update
Morrison, W. and T. Kang (2014). "Judging the Quality of Mercy: Drawing a Line Between Palliation and Euthanasia." Pediatrics 133(Supplement 1): S31-S36.	Palliative/Respite Care	Requires Update
Beresford, B. A. and P. Sloper (2003). "Chronically ill adolescents' experiences of communicating with doctors: a qualitative study." J Adolesc Health 33(3): 172-179.	Continuity of Care/Communication	Requires Update
Miller, A. R., et al. (2009). "Continuity of care for children with complex chronic health conditions: parents' perspectives." BMC Health Serv Res 9: 242.	Continuity of Care/Communication	Requires Update
Budych, K., et al. (2012). "How do patients with rare diseases experience the medical encounter? Exploring role behavior and its impact on patient-physician interaction." Health Policy 105(2-3): 154-164	Continuity of Care/Communication	Requires Update
Jones, B. L., et al. (2014). "The Duty of the Physician to Care for the Family in Pediatric Palliative Care: Context, Communication, and Caring." Pediatrics 133(Supplement 1): S8-S15.	Continuity of Care/Communication	Requires Update
Mack, J. W. and S. Joffe (2014). "Communicating About Prognosis: Ethical Responsibilities of Pediatricians and Parents." Pediatrics 133(Supplement 1): S24-S30.	Continuity of Care/Communication	Requires Update
Walter, J. K. and L. F. Ross (2014). "Relational Autonomy: Moving Beyond the Limits of Isolated Individualism." Pediatrics 133(Supplement 1): S16-S23.	Continuity of Care/Communication	Requires Update
Weidner, N. J. (2005). "Developing an interdisciplinary palliative care plan for the patient with muscular dystrophy." Pediatr Ann 34(7): 546-552	Review	Requires Update
Birnkrant, D. J. and G. H. Noritz (2008). "Is there a role for palliative care in progressive pediatric neuromuscular diseases? The answer is "Yes!" J Palliat Care 24(4): 265-269.	Review	Requires Update

Endocrine

Clinical experts agreed that endocrine as one of the new topics will require an extensive update to the care consideration and its own section. It was also suggested that bone health needs to be a standalone topic beyond the hormonal aspects in the endocrine section.

Since the prior recommendations were published, attention to spine health in children with steroid-treated disorders has increased. This stems from a number of studies in pediatric steroid-treated chronic illnesses that show the spine is particularly susceptible to the deleterious effects of steroids.
The International Society for Clinical Densitometry has recently updated its guidelines (2014) in recognition of this, to the extent that if a child with risk factors for osteoporosis develops a vertebral fracture in the absence of trauma, this alone fulfills the diagnosis of osteoporosis in this child (i.e. BMD criterion is not needed). The ISCD 2014 guidelines should be included in the updated DMD care guidelines as they are broadly applicable and particularly relevant to boys with DMD given the predilection for steroids to have an adverse effect on the spine.
Routine monitoring of spine health with a lateral spine radiograph is advised and is more clinically useful than bone mineral density, based on current knowledge. This is a change in recommendations compared to current care considerations and should be addressed in the update.

There is not a lot of literature on obesity at this time. Discussions are occurring around how mild obesity in adolescence may revert to underweight status in later years. Further research on this topic is needed, but it is suggested the natural evolution of weight history be noted in the updated care considerations.

Endocrine		
Study	**Sub-Topic**	**Recommendation**
Martigne et al. (2011) Natural evolution of weight status in Duchenne muscular dystrophy: A retrospective audit. British Journal of Nutrition, 105(10), 1486–1491	Growth	Promising Research
West et al. (2013) Patterns of growth in ambulatory males with Duchenne muscular dystrophy. Journal of Pediatrics, 163(6), 1759–1763 e1751.	Growth	Promising Research
Sarrazin et al. (2014) Growth and psychomotor development of patients with Duchenne muscular dystrophy. European Journal of Paediatric Neurology, 18(1), 38–44.	Growth	Requires Update
Rutter et al. (2012) Growth hormone treatment in boys with Duchenne muscular dystrophy and glucocorticoid-induced growth failure. Neuromuscular Disorders, 22(12), 1046–1056.	Growth Hormone	Promising Research
Okada, K., et al. (1992). "Protein and energy metabolism in patients with progressive muscular dystrophy." J Nutr Sci Vitaminol (Tokyo) 38(2): 141-154.	Body Composition	Requires Update
Bedogni, G., et al. (1998). "Use of bioelectric impedance analysis (BIA) in children with alterations of body water distribution." Appl Radiat Isot 49(5-6): 619-620.	Body Composition	Promising Research
Pessolano, F. A., et al. (2003) Nutritional assessment of patients with neuromuscular diseases. American Journal of Physical Medicine & Rehabilation, 82(3), 182-185.	Body Composition	Requires Update
Mok et al. (2006). Estimating body composition in children with Duchenne muscular dystrophy: Comparison of bioelectrical impedance analysis and skinfold-thickness measurement. American Journal of Clinical Nutrition, 83(1), 65–69.	Body Composition	Requires Update
Skalsky et al. (2009) Assessment of regional body composition with dual-energy X-ray absorptiometry in Duchenne muscular dystrophy: Correlation of regional lean mass and quantitative strength. Muscle & Nerve, 39(5), 647–651.	Body Composition	Promising Research
Mok et al. (2010) Assessing change in body composition in children with Duchenne muscular dystrophy: Anthropometry and bioelectrical impedance analysis versus dual-energy X-ray absorptiometry. Clinical Nutrition, 29(5), 633–638.	Body Composition	Requires Update

Arikian et al. (2010) Targeting parents for the treatment of pediatric obesity in boys with Duchenne muscular dystrophy: A case series. Eating and Weight Disorders, 15(3), e161–165	Body Composition	Requires Update
Elliott et al. (2012) Predicting resting energy expenditure in boys with Duchenne muscular dystrophy. European Journal of Paediatric Neurology, 16(6), 631–635.	Body Composition	Requires Update
Shimizu-Fujiwara et al. (2012) Decreased resting energy expenditure in patients with Duchenne muscular dystrophy. Brain & Development, 34(3), 206–212	Body Composition	Requires Update
Bayram et al. (2013) Correlation between motor performance scales, body composition, and anthropometry in patients with Duchenne muscular dystrophy. Acta Neurologica Belgica, 113(2), 133–137.	Body Composition	Promising Research
Connolly, A. M., et al. (2002) High dose weekly oral prednisone improves strength in boys with Duchenne muscular dystrophy. Neuromuscular Disorders, 12(10), 917-925.	Corticosteroid Treatment	Promising Research
Kinali, M., et al. (2002) An effective, low-dosage, intermittent schedule of prednisolone in the long-term treatment of early cases of Duchenne dystrophy. Neuromuscular Disorders, 12, S169-S174.	Corticosteroid Treatment	Requires Update
Orngreen, M. C., et al. (2003). "Patients with severe muscle wasting are prone to develop hypoglycemia during fasting." Neurology 61(7): 997-1000.	Corticosteroid Treatment	Requires Update
Straathof et al. (2009) Prednisone 10 days on/10 days off in patients with Duchenne muscular dystrophy. Journal of Neurology, 256(5), 768–773. doi:10.1007/s00415-009-5012-y	Corticosteroid Treatment	Requires Update
Escolar et al. (2011) Randomized, blinded trial of weekend vs daily prednisone in Duchenne muscular dystrophy. Neurology, 77(5), 444–452. doi:10.1212/WNL.0b013e318227b164	Corticosteroid Treatment	Promising Research
Pegoraro, E., et al. (2011). "SPP1 genotype is a determinant of disease severity in Duchenne muscular dystrophy." Neurology 76(3): 219-226.	Corticosteroid Treatment	Promising Research
Ricotti et al. (2013) Long-term benefits and adverse effects of intermittent versus daily glucocorticoids in boys with Duchenne muscular dystrophy. Journal of Neurology, Neurosurgery and Psychiatry, 84(6), 698–705. doi:10.1136/jnnp-2012-303902	Corticosteroid Treatment	Requires Update
Singh, M., et al. (2007). "Nephrolithiasis in patients with duchenne muscular dystrophy." Urology 70(4): 643-645.	Nephrology	Requires Update
Shumyatcher Y, et al. (2008) Symptomatic nephrolithiasis in prolonged survivors of Duchenne muscular dystrophy. Neuromuscul Disord; 18: 561-4.	Nephrology	Requires Update
Askeland, E. J., et al. (2013). "Urological manifestations of Duchenne muscular dystrophy." J Urol 190(4 Suppl): 1523-1528.	Nephrology	Requires Update
Soderpalm et al. (2013) Whole body vibration therapy in patients with Duchenne muscular dystrophy—a prospective observational study. Journal of Musculoskeletal & Neuronal Interactions, 13(1), 13–18.	Bone Health	Promising Research
Crabtree, N. J., et al. (2010). "Regional changes in bone area and bone mineral content in boys with duchenne muscular dystrophy receiving corticosteroid therapy." J Pediatr 156(3): 450-455.	Bone Health	Requires Update
Bianchi et al. (2011) Low bone density and bone metabolism alterations in Duchenne muscular dystrophy: Response to calcium and vitamin D treatment. Osteoporosis International, 22(2), 529–539. doi:10.1007/s00198-010-1275-5	Bone Health	Promising Research

Castellanos, N. P., et al. (2011). "Improving osteoporosis diagnosis in children using image texture analysis." Conf Proc IEEE Eng Med Biol Soc 2011: 6184-6187.	Bone Health	Promising Research
Rufo, A., et al. (2011). "Mechanisms inducing low bone density in Duchenne muscular dystrophy in mice and humans." J Bone Miner Res 26(8): 1891-1903.	Bone Health	Promising Research
Mayo, A. L., et al. (2012). Bone health in boys with Duchenne Muscular Dystrophy on long-term daily deflazacort therapy. Neuromuscular Disorders, 22(12), 1040–1045. doi:10.1016/j.nmd.2012.06.354	Bone Health	Requires Update
Hawker, G. A., et al. (2005) Alendronate in the treatment of low bone mass in steroid-treated boys with Duchenne's muscular dystrophy. Archives of Physical Medicine and Rehabilitation, 86(2), 284-288.	Bone Health	Promising Research
Gordon et al. (2011) Impact of bisphosphonates on survival for patients with Duchenne muscular dystrophy. Pediatrics, 127(2), e353–358. doi:10.1542/peds.2010-1666	Bone Health	Promising Research
Sbrocchi et al. (2012). The use of intravenous bisphosphonate therapy to treat vertebral fractures due to osteoporosis among boys with Duchenne muscular dystrophy. Osteoporosis International, 23(11), 2703–2711. doi:10.1007/s00198-012-1911-3	Bone Health	Requires Update
Apkon, S. D. (2002). Osteoporosis in children who have disabilities. Physical Medicine & Rehabilitation Clinics of North America, 13, 839-855.	Review	Requires Update
Biggar, W. D., et al. (2005) Bone health in Duchenne muscular dystrophy: a workshop report from the meeting in Cincinnati, Ohio, July 8, 2004. Neuromuscular Disorders, 15(1), 80-85.	Review	Requires Update
Quinlivan, R., et al. (2005) Report of a muscular dystrophy campaign funded workshop Birmingham, UK, January 16th 2004. Osteoporosis in Duchenne muscular dystrophy; its prevalence, treatment and prevention. Neuromuscular Disorders, 15(1), 72-77.	Review	Requires Update

Transition and Adult Care

The literature available specifically for DMD transition is limited, but there is a body of literature related to transition between pediatric and adult care in general. The clinical experts SC members came to the consensus that there is enough literature available in the Transition and Adult Care Summary to develop a section on transition in the update, and that adult care should be incorporated into all sections as appropriate. SC members also recommend there be a separate adult-oriented article to be published in a journal such as the Annals of Internal Medicine.

Guidance developed for other disorders that may benefit DMD patients should be considered. The SC suggests that we summarize what we know about transition based on the literature, clinical expert's experience, and results from transition surveys conducted by nationally recognized organizations such as PPMD, MDSTARnet, and MDA. A list of transition resources that are readily available to DMD community should also be developed.