



Patient Reported Outcomes

Cooperative International Neuromuscular
Disease Research Group

University of California, Davis



Relationship between Impairment, Activity Limitation, Participation and Quality of Life in DMD

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Goal: Develop a program for multi-center natural history profile for rehabilitation research



Purpose

- Establish outcome measurements for impairment, activity limitation, participation and health-related quality of life
- Characterize the longitudinal profile of the impairment, secondary conditions, and health-related quality of life in DMD
- Examine the relationship between impairment and secondary conditions, activity limitations, participation, and life satisfaction--using the ICF Classification of Functioning, Disability and Health



Subjects

- Enroll 295 boys with DMD from the 10 domestic CINRG centers. Boys will be followed for 5 years yielding 1475 clinic visits.
- Perform measurements once each year for the next four years.
- Goal is to have approximately 60 boys at each age ranging from 6 – 25 years



Components of Study

- Patient-reported health–related quality of life outcomes (HR-QOL)
- Patient reported review of systems medical history—reviewed by physician
- Clinical evaluation (MMT, QMT, PFT, TMP, ROM, FE)
- Diagnostic evaluation (molecular genetics and muscle biopsy)
- Medical Record Abstracting



Dimensions of Quality of Life

Physical

Health
Physical functioning
Ambulation
Exercise tolerance
Energy/stamina
Adequate sleep/rest
Nutrition
Absence of pain
Control of symptoms
Somatic comfort
Physical independence
Freedom from illness
Sexuality

Psychological

Control of ones life
Level of stress
Life satisfaction
Body image
Self-acceptance
Absence of negative moods
Self esteem/self-worth
Psychological well-being
Achievement of life goals
Intellectual functioning
Illness concerns, &
prognosis
Adjustment to
illness/disability

Social

Ability to communicate
Role functioning
Social support resources
Usefulness to others
Recreation
Social interaction
Marital & Family
Standard of living
Financial independence
Neighborhood
Sexual relations
Employment
Spiritual support



HR-QOL Measurements

- **PedsQL**
- **POSNA Functional Assessment**
- **NMD Sleep Study**
- **Life Satisfaction Index for Adolescents**
- **WHO Quality of Life Bref**



PedsQL

- Generic measure of HR-QOL
- Child self-report for ages 5-18 & parent proxy-report for ages 2-18
- Extensive normative databases provide comparisons across chronic disease groups, changes in time, and with healthy populations
- Measure 4 domains:
 - Physical/health function (8), Emotional function (5), Social function (5), School function (6)



PedsQL

- *In the past one month, how much of a problem has your child had with...*

	Never	Almost never	Some-times	Often	Almost Always
Having hurts or aches	0	1	2	3	4

Walking more than one block

Running

Participating in sports activities or exercise

Lifting something heavy

Taking a bath or shower by himself

Low energy level

Doing chores around the house



Pediatric Outcomes Data Collection Instrument

- No specific HR-QOL instrument for DMD
- Domains of interest. We want an instrument responsive to treatments that may ameliorate the disease progression.
 - changes in strength
 - functional status & ADLs
 - pulmonary function
- Pediatric Orthopedic Society of North America (POSNA) Measures functional levels in pediatric orthopedic subjects (Pediatric Outcomes Data Collection Instrument – PODCI)
 - Existing database of able-bodied controls and individuals with disabilities
 - Internal reliability > 0.80
 - Test-retest > 0.80
 - Sensitivity is good, especially for severe physical disabilities



POSNA/PODCI Domains

- Global function & comfort
- Upper extremity function
- Physical function & sports
- Transfers & mobility
- Comfort (pain-free)
- Happiness and satisfaction



POSNA/PODCI

Upper extremity function

During the <u>last week</u> was it easy or hard for you to...	Easy	A little hard	Very hard	Can't do at all
Lift heavy books?	1	2	3	4
Pour a half gallon of milk?	1	2	3	4
Open a jar that has been opened before?	1	2	3	4
Use a fork and spoon?	1	2	3	4
Comb your hair?	1	2	3	4
Button buttons?	1	2	3	4



POSNA/PODCI

Physical function & sports

During the <u>last week</u> was it easy or hard for you to...	Easy	A little hard	Very hard	Can't do at all
Run short distances?	1	2	3	4
Bicycle or tricycle?	1	2	3	4
Climb one flight of stairs?	1	2	3	4
Walk one block?	1	2	3	4
Walk one block?	1	2	3	4
Participate in sports or recreational activities?	1	2	3	4



POSNA/PODCI

Transfers & mobility

During the <u>last week</u> was it easy or hard for you to...	Easy	A little hard	Very hard	Can't do at all
Stand while washing your hands and face at a sink?	1	2	3	4
Sit in a regular chair without holding on?	1	2	3	4
Get on and off a toilet or chair?	1	2	3	4
Get in and out of bed?	1	2	3	4
Turn doorknobs?	1	2	3	4
Open a door?	1	2	3	4



POSNA/PODCI

Happiness and satisfaction

During the last week, how happy have you been with...	Very happy	Some-what happy	Not sure	Some-what unhappy	Very unhappy
How you look?	1	2	3	4	5
Your body?	1	2	3	4	5
What clothes or shoes you can wear?	1	2	3	4	5
Your ability to do the same things your friends do?	1	2	3	4	5
Your health in general?	1	2	3	4	5



Review of Systems

- Medical history, surgeries, illness & hospitalizations (pneumonias, fractures, spinal surgeries, surgery to improve range of motion, problems with anesthesia)
- Family history of DMD (symptoms – age of loss of ambulation, weakness, hypertrophy, cardiomyopathy, etc.)
- Neuromuscular symptoms (Age of diagnosis, Age in manual wheelchair, Age in power wheelchair)
- Cognitive & education issues (IQ tests, special ed, services)
- Steroid use and dose
- Nutritional supplements (modified food, PEG tube, NG tube)

Review of Systems: Gastrointestinal Issues

- Difficulty swallowing
- Rapid weight loss
- Difficulty chewing
- Rapid weight gain
- Problems with aspiration
- Weight gain with puffy face
- Poor appetite
- Poor growth
- Indigestion
- Blood in stool
- Diarrhea
- Excessive appetite
- Kidney stones
- Bloating
- Excessive thirst
- Problematic soiling
- Nausea
- Early satiety (gets full fast)
- Bowel obstruction



Review of Systems cont'd

- Cataracts
- Clinical Trial participation
- DME usage (splints, braces, standing board)
- Pulmonary/cardiovascular issues
 - (oximetry & sleep studies
 - Usage of cough assist, non-invasive breathing, suctioning, supplemental oxygen, suctioning
 - breathing exercises, intubated, tracheostomy)
 - Difficulties breathing,

Review of Systems: Cardiopulmonary Issues

- Weak cough
- Hay fever, allergies, rhinitis
- Chronic cough
- Gasp for air (air hunger)
- Pneumonia or bronchitis
- Shortness of breath
- Snoring
- Night sweats
- Early morning headaches
- Asthma
- Nightmares
- Frequent awakening or turning in sleep
- Difficulty with breathing when lying on back
- Palpitations
- Difficulty breathing when exercising
- Excessive sleepiness
- Difficulty concentrating (or) difficulty with school studies
- Chest pains



Review of Systems cont'd

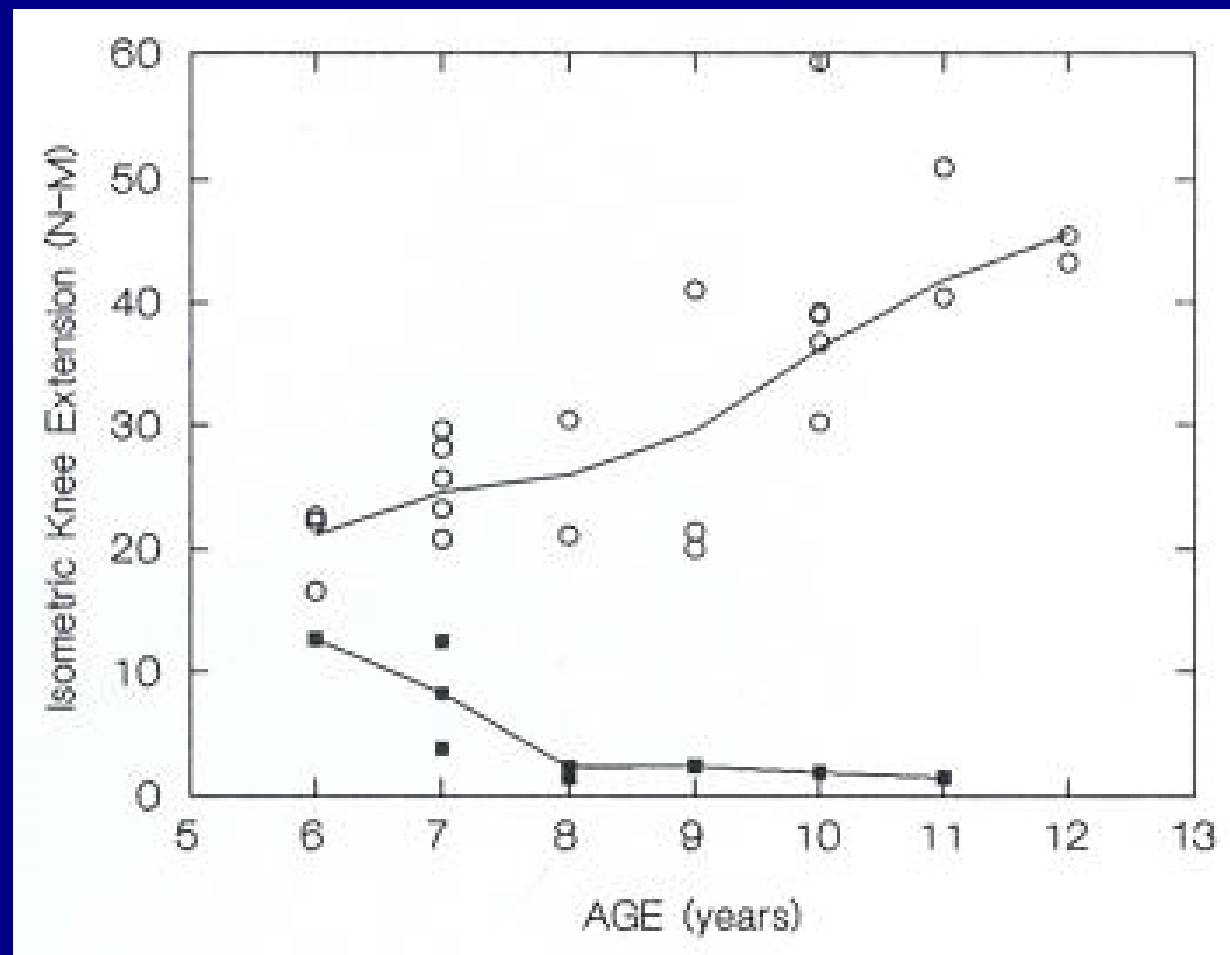
- Medication Usage
- Physicians seen
- Allied health usages
- Participation in recreation, aquatics, support groups
- Sleep questionnaire. During the past month, how often have you had trouble sleeping because you...
 - a. Cannot get to sleep within 30 minutes
 - b. Wake up in the middle of the night or early morning
 - c. Have bad dreams
 - i. Have pain



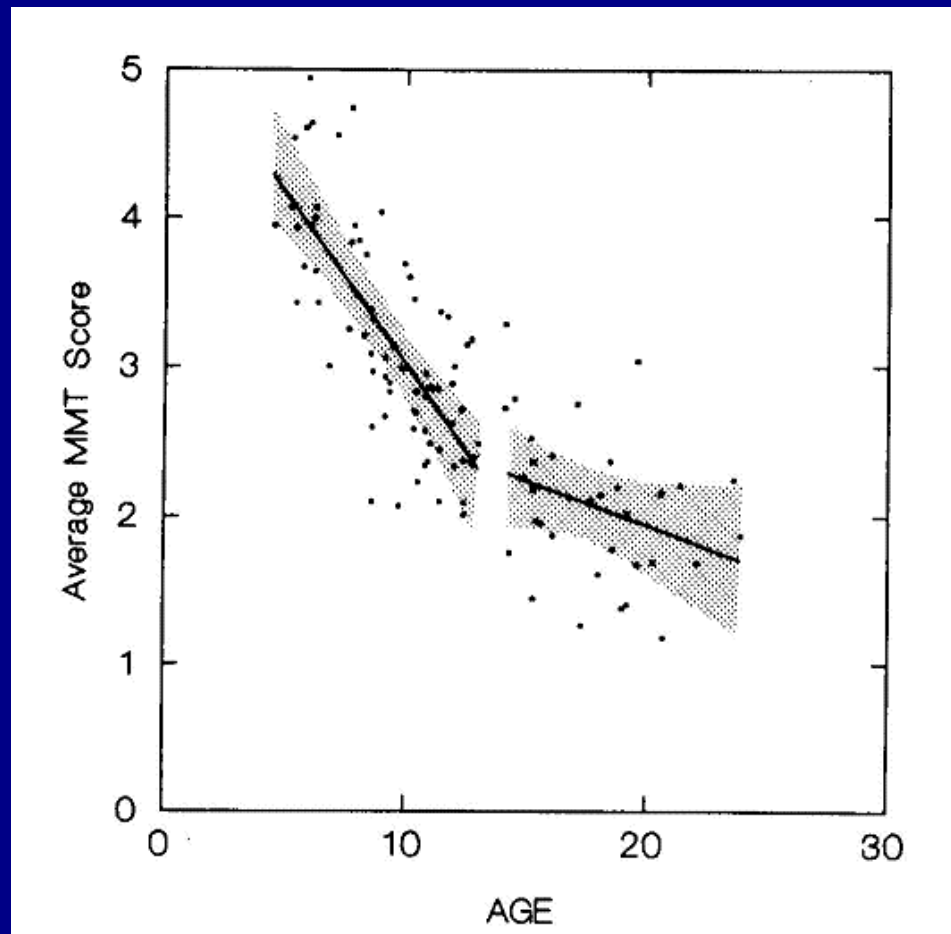
Clinical evaluation: Strength & Pulmonary Function

- Quantitative Muscle Testing (QMT)
- Manual Muscle Tests (MMT)
- Pulmonary Function Tests (PFT)
- Brooke/Vignos Functional Classification
- Timed Motor Performance
- Anthropometrics

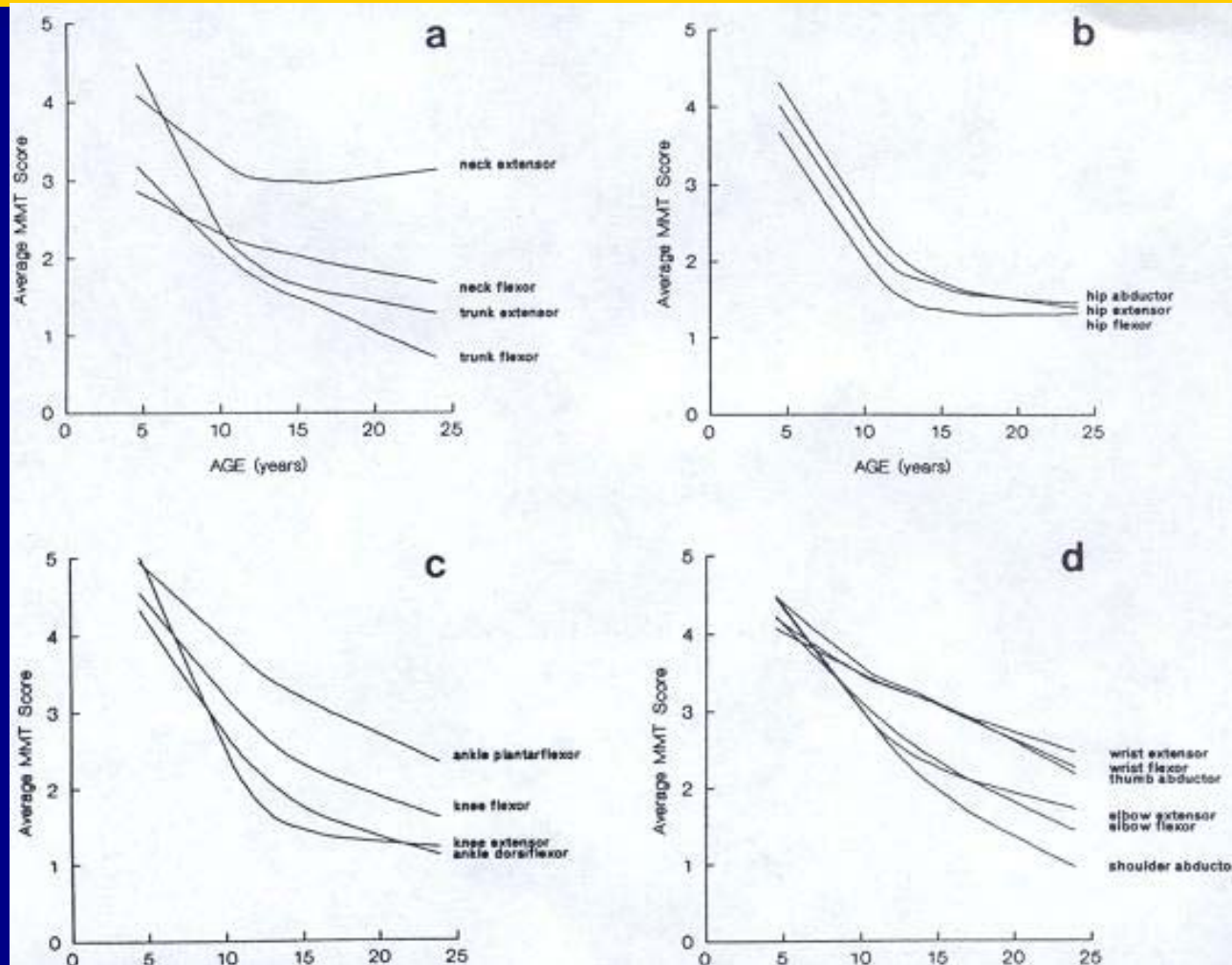
Quantitative strength



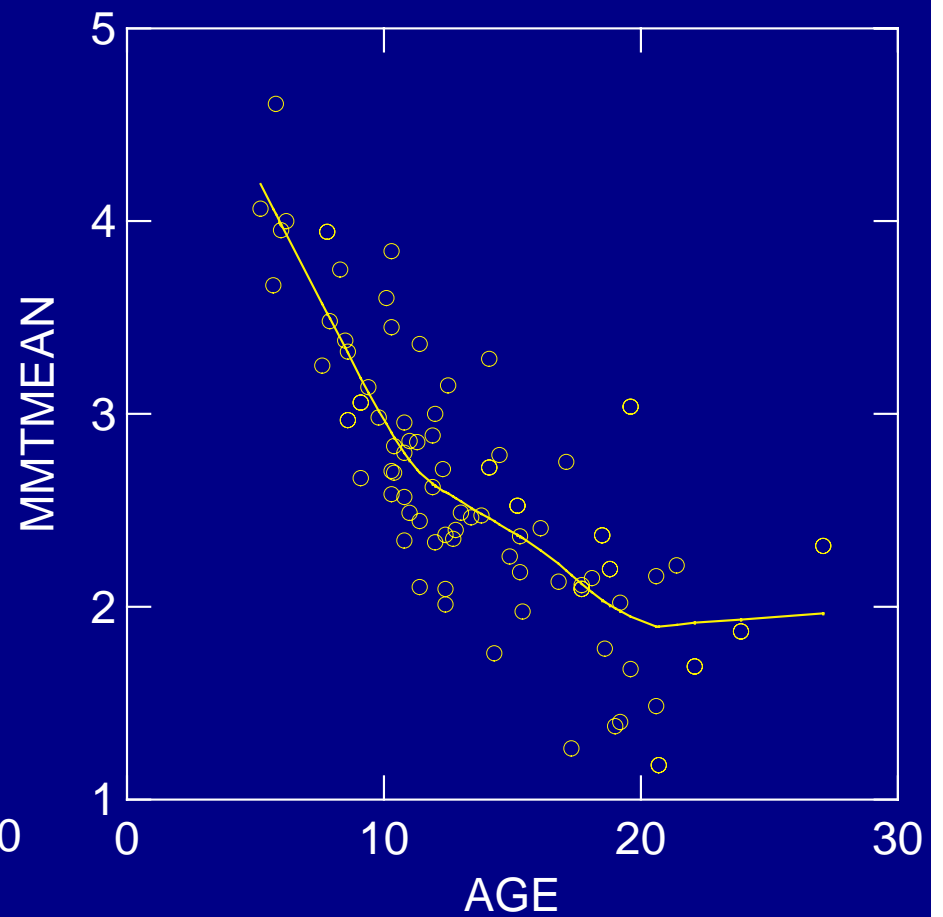
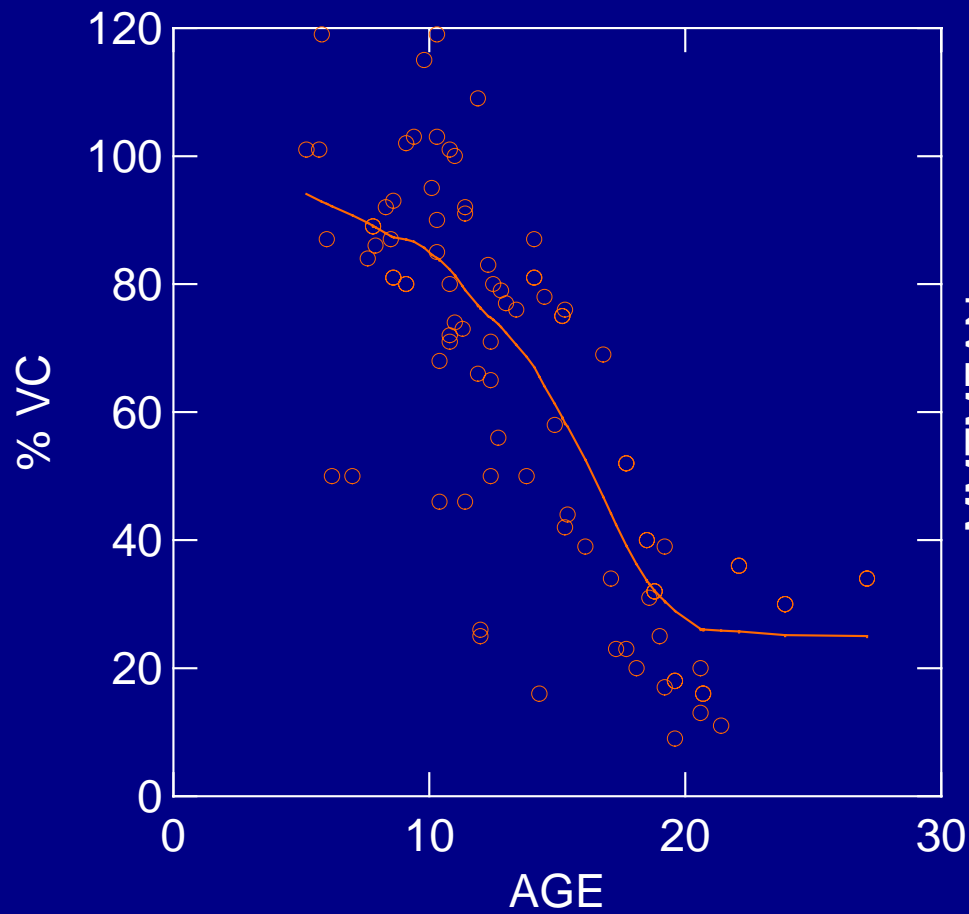
Manual Muscle Tests



Manual Muscle Tests



Pulmonary vs. MMT Measures





Assess participation, life satisfaction, service utilization and health-related quality of life

Describe characteristics of impairment, activities, participation and quality of life using self-report questionnaires from affected children and their parents.

- Explore the relationship between impairment, activities, participation, & health-related quality of life.
- Correlate demographic variables, illness related variables, and psychosocial variables.



Develop outcome measurement tools for future studies

- Determine the most robust impairment, activity, participation and quality of life outcome measures from this study
- Reliable data to formulate clinically relevant research questions.
 - 1) clinical features of disease pathology
 - 2) rates of disease progression and impairment
 - 3) development of secondary pathologies
 - 4) effects of prednisone



Outcomes of Spine Surgery

- Assess the immediate and long term impact of surgery to correct spine deformity in DMD on:
 - Strength
 - Pain
 - Nutritional status
 - Activity levels
 - Self-reported health and wellness
- Identify and assess the risk factors for complications associated with spine surgery
 - pulmonary complications
 - hospitalization



Aggressive pulmonary intervention

- Design an aggressive perioperative intervention consisting of
 - pulmonary technologies to assist cough
 - pulmonary technologies to assist ventilation
- Assess the effectiveness of these interventions to reduce associated co-morbidity



24 hour/day Noninvasive Ventilation Program NPPV Treatment algorithm

- Nocturnal NPPV
 - 1) Symptoms of sleep disturbance and abnormal sleep study nocturnal hypoventilation and/or obstructive sleep apnea, or
 - 2) FVC < 50%, or
 - 3) PaCO₂ > 50 mm Hg, or
 - 4) nocturnal oxyhemoglobin desaturation (< 88%) followed by abnormal sleep study
- Daytime mouthpiece NPPV
 - 1) PaCO₂ > 48 despite adequate nocturnal NPPV
 - 2) Peak cough flow < 200 LPM (for auto-breathstacking)Diurnal sx of dyspnea.



Outcomes

This study will lay the groundwork for one of two possible future studies:

- a randomized controlled trial of 24 hour/day NPPV; or
- a pre/post quasi-experimental design study in the CINRG group where 24 hour/day ventilation is introduced into a population previously naïve to this therapy.