Parent Project Muscular Dystrophy

JOIN THE FIGHT. END DUCHENNE.

PPMD: YOUR STRENGTH, YOUR FAMILY

I had no intention of being part of the Duchenne community this long.

When we started PPMD in 1994, I naively thought we would have a cure and I would be spending these years with my sons and daughters and a ton of grandkids I would spoil rotten.

And here we are 25 years later. My sons are no longer here, I worry about my daughters being carriers, I worry about my granddaughter being a carrier, and our community has never had to work harder and the losses have never felt more profound.

But, once we dove in and began to understand Duchenne — while simultaneously educating those around us — we understood that progress wasn't going to come in the form of a magic wand or silver bullet. With time, we have had to redefine what progress is. And while it may be too late for my boys and all of the children we have lost along the way, *there is progress*. With every victory, large or small, we honor our brave loved ones.

PPMD started with two basic ideas in mind:

- We must give parents a community to turn to and a voice to advocate for their child's healthcare.
- We must end Duchenne.

Both of these goals have gone through their own redefinition.

More than a Parent Project

When my sons were diagnosed in 1984, I went to the library to find everything I could on Duchenne. This was before the internet. We're talking microfiche and a card catalog! My husband and I are both medically trained — he's a retired family doc and I was in nursing — but Duchenne was something we maybe covered in a half-day course on neurological diseases if that. It was hard to find a community, to build an army to fight this horrible disease. We needed to unite all of the families that, like ours, felt alone.

The Duchenne community has never been stronger. It continues to grow and strengthen and is a force of nature in the international rare disease community. We have become a model for other diseases and a case study in how parent advocacy can unite and get things done.

But we also realized that this isn't just a 'parents' project.' There are many voices joining us in this fight — entire families, whole communities, friends from work, our child's school, not to mention regulatory agencies, pharmaceutical companies, biotechs, universities, and clinics.

PPMD's Pat Furlong with her sons, Chris & Patrick

But perhaps the most important voice we hear today is that of the people living with Duchenne. A voice that for years had been left out of the conversation and today is often leading the narrative.

Parent Project Muscular Dystrophy is a community-wide effort that we strive to lead, incorporating the priorities and passion of every stakeholder.

Redefining an End to Duchenne

To me, success was going to be when we could turn off the lights at PPMD, close the door, and not look back. Because Duchenne would be done. Maybe it would be a vaccination you got as a child, a quick shot to guarantee you don't 'catch' Duchenne, like the Polio vaccine. It would maybe take 5, 10 years at the most because nothing happens quickly with government agencies. That was my plan in 1994.

What we have learned is that Duchenne is complicated. I know, sounds simplistic and it almost feels ridiculous to write. But it's true — if it was easy, PPMD wouldn't be turning 25.

Duchenne is a progressive disorder that needs to be treated with a progressive attack plan. We didn't realize in those early days that it was going to take a combination of therapies to treat Duchenne. We know that now. We also know that the fight won't end when we have treatments. We have to make sure that our loved ones can access these treatments and that insurance companies will cover them.

In my 25 years at PPMD, I realize that this fight is an evolution: as our understanding of Duchenne evolves, so does our strategy to end it.

Pat Furlong, *Founding President & CEO*

IMPERATIVES FOR DUCHENNE MD

A Guide for Providers

D — DIAGNOSIS

- If developmental delay or elevated liver enzymes, do a creatine kinase (CK) (ChildMuscleWeakness.org)
- If male patients have a high CK (CK>800), order full genetic testing for Duchenne Muscular Dystrophy
- Discuss carrier testing/reproductive options for mother and testing for other family members

U — UNDERSTAND BREATHING PROBLEMS

- Pulmonary function test at least every year starting as young as possible
- Discuss cough assist when cough peak flow is < 270 liters per minute or if cough becomes weaker (use during respiratory illnesses while ambulatory and daily and as needed after loss of ambulation)
- Discuss nighttime Bi-PAP as needed or when forced vital capacity (FVC) < 30%
- Keep immunizations (including pneumonia and annual flu) up to date
- Treat respiratory infections promptly and aggressively
- Do NOT give supplemental oxygen without monitoring CO2

C — CORTICOSTEROIDS

- Start early!
- Discuss the benefits and possible side effects of corticosteroids by age 3 years, or as young as possible
- Evaluate efficacy and manage side effects of corticosteroids at each neuromuscular visit
- Discuss the rationale for lifelong steroid management
- Never stop taking steroids abruptly
- Discuss the need for stress dosing of steroids for illnesses or surgeries

H — HEART

- Cardiology visit with imaging (cardiac MRI preferred; echocardiogram if cardiac MRI not available) every year from diagnosis or more often if needed
- Discuss cardiac medications if fibrosis is seen on cardiac MRI, for any decrease in cardiac function decreases from baseline, or for heart failure (SF or shortening fraction <28% or ejection fraction <55%) or by age 10 even if findings are normal

E — ENDOCRINE

- If taking steroids, check 25-OH vitamin D prior to starting steroids, then annually
- Supplement vitamin D as needed
- Encourage sun exposure
- Nutrition discussions of adequate calcium and vitamin D intake
- Discuss measurement of bone density and use of bisphosphonates
- Assess spine for scoliosis at each visit
- Monitor for puberty starting at 9 years of age for need for testosterone therapy

N — NEVER FORGET PHYSICAL & OCCUPATIONAL THERAPY

- Specialized PT evaluations every 4-6 months
- Stretching every day
- Discuss and encourage contracture prevention (splints, stretches), appropriate exercise, assistive mobility devices (strollers, scooters, wheelchairs) and other assistive devices (beds, arm assistance, lifts, etc.) from the time of diagnosis

N — NUTRITION & GASTROINTESTINAL

- Monitor weight
- Assess/discuss diet (healthy eating, calcium, vitamin D)
- Evaluate swallowing/need for intervention
- Treat GERD and constipation as necessary
- See your dentist every 6 months

E — EMERGENCY

- Have patients/parents carry a copy of their last visit/note summary (including medications and neuromuscular contact information) and a Duchenne emergency card with them at all times
- Use caution with all anesthesia; avoid inhaled anesthesia
- Never use succinylcholine

M — MENTAL HEALTH

- Assess adjustment, coping, behavioral and emotional disorder and social isolation for the patient and family at each visit
- Screen for learning disability (reading and math), language problems, attention deficit disorder (ADD), attention deficit and hyperactivity disorder (ADHD), autism and obsessive compulsive disorder (OCD)
- Neurocognitive evaluation done at diagnosis and prior to formal schooling; screening/management as needed
- Discuss the need for individualized/special educational plan

D — DON'T DO IT ALONE

- Direct to trustworthy, reliable online resources
- Organize follow up via a comprehensive neuromuscular center with expertise in caring for people living with Duchenne
- Offer contact with organizations (**ParentProjectMD.org**, **TREAT-NMD.eu**, **WorldDuchenne.org**)

Care for Duchenne
parentprojectmd.org/care

Emergency Care Considerations
parentprojectmd.org/emergencycare

Center for Disease Control and Prevention Care Considerations
parentprojectmd.org/careguidelines

Parent Project Muscular Dystrophy

TREAT-NMD Neuromuscular Network

WORLD DUCHENNE ORGANIZATION UPPMD

ADULT IMPERATIVES FOR DUCHENNE MD

A Guide for Providers

D — DISCUSS GOALS

■ Provider should spend time alone with patient at every visit ■ Monitor transition to adult care ■ Assess independence at every visit ■ Discuss educational and/or employment goals at every visit ■ Facilitate discussions around advanced care planning including advanced directives and having an emergency care plan in place

U — UNDERSTAND BREATHING PROBLEMS

■ Pulmonary function evaluation every 6 months or as recommended by a pulmonologist ■ Discuss cough assist when cough peak flow is < 270 liters per minute or if cough becomes weaker ■ Discuss sleep study and nighttime non-invasive ventilation (Bi-PAP) when forced vital capacity (FVC) < 50% or with symptoms of hypoventilation (frequent awakenings, morning headaches, and behavioral changes) ■ Discuss daytime non-invasive ventilation when exhaled CO_2 > 45 mmHg ■ Keep immunizations (including pneumonia and annual flu) up to date ■ Treat respiratory infections promptly and aggressively ■ Do NOT give supplemental oxygen without monitoring CO_2

C — CORTICOSTEROIDS

■ Discuss the rationale for lifelong steroid management ■ Evaluate efficacy and manage side effects of corticosteroids at each neuromuscular visit ■ Never stop taking steroids abruptly ■ Discuss the need for stress dosing of steroids for illnesses or surgeries

H — HEART

■ Cardiology visit with imaging (cardiac MRI preferred; echocardiogram if cardiac MRI not available) every 6 months or as recommended by a cardiologist ■ Prescribe first line cardiac medications (ACEi or ARB) as tolerated ■ Standard heart failure medications should be initiated with evidence of heart failure (SF or shortening fraction <28% or ejection fraction <55%)

E — ENDOCRINE

■ If taking steroids, check 25-OH vitamin D annually ■ Supplement vitamin D as needed ■ Nutrition discussions of adequate calcium and vitamin D intake ■ Discuss measurement of bone density and use of bisphosphonates ■ Assess for back pain or signs of vertebral compression fractures ■ Evaluate sexual maturity at each clinic visit until progression through puberty is established for need for testosterone therapy

N — NEVER FORGET PHYSICAL & OCCUPATIONAL THERAPY

■ Specialized PT evaluations every 4-6 months using standardized strength and function measures ■ Stretching every day if comfortable ■ Discuss and encourage contracture prevention (splints, stretches), appropriate exercise, assistive mobility devices (strollers, scooters, wheelchairs) and other assistive devices (beds, arm assistance, lifts, etc.) ■ Assess positioning and posture and the need for supports at each visit to prevent scoliosis

N — NUTRITION & GASTROINTESTINAL

■ Monitor weight ■ Assess/discuss diet (healthy eating, calcium, vitamin D) ■ Evaluate swallowing/need for intervention ■ Treat GERD, constipation and gastroparesis as necessary ■ See your dentist every 6 months

E — EMERGENCY

■ Have patients, parents and/or caregivers carry a copy of their last visit/note summary (including medications and neuromuscular contact information) and a Duchenne emergency card with them at all times ■ Use caution with all anesthesia; avoid inhaled anesthesia ■ Never use succinylcholine

M — MENTAL HEALTH

■ Assess adjustment, coping, behavioral and emotional disorder and social isolation for the patient and family at each visit ■ Discuss the need for individualized personal care/support ■ Assess relationships, friendships, activities and community engagement at each visit

D — DON'T DO IT ALONE

■ Direct to trustworthy, reliable online resources ■ Organize follow up via a comprehensive neuromuscular center with expertise in caring for people living with Duchenne ■ Offer contact with organizations (**ParentProjectMD.org, WorldDuchenne.org**) ■ Encourage connection to other adults living with Duchenne locally or through social media

Care for Duchenne
parentprojectmd.org/care

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Parent Project Muscular Dystrophy

WORLD DUCHENNE ORGANIZATION UPPMD

RANGE OF MOTION MANAGEMENT IN DUCHENNE MUSCULAR DYSTROPHY – TREATMENT OPTIONS

Parent Project Muscular Dystrophy
JOIN THE FIGHT. END DUCHENNE.

General Guidelines – Getting Started at Home

Included in this document are general guidelines for methods for management of range of motion in children and adults with Duchenne muscular dystrophy. These are guidelines based on currently available research and information known about muscle and the natural history of Duchenne.

**Please consult an experienced physical therapist in Duchenne for individualized recommendations.

Not everything in this document will apply to every person with Duchenne. Individualized plans are best carried out by an experienced physical therapist who is familiar with your loved one.

Find a routine that fits for your family and child.
Be reasonable in what will work into your daily schedule.
Prioritize based on your child's needs and your physical therapist recommendations.

SUMMARY OF GUIDELINES AND DEFINITIONS OF TREATMENT OPTIONS

ANKLE NIGHT SPLINTS

A brace is used to provide a long (maintained) stretch by holding the foot in a better position over the course of several hours. Most often recommended for wearing overnight, if tolerated. They are often made of plastic and are made specifically to fit an individual's foot & ankle by an experienced orthotist. They are not recommended for wear during walking activities.

RESTING HAND SPLINTS

A splint is intended to support the hand, wrist, and fingers in a neutral or flat position when at rest. Resting hand splints are typically molded from a plastic material with Velcro straps and padding specifically to fit an individual's hand & wrist. They should be custom-fitted by an experienced occupational therapist, physical therapist, or orthotist.

POSITIONING

Positioning consists of using bolsters, rolls, or pillows to position or support a body part in a good alignment. When individuals are spending increasing amounts of time in a seated position or lying down, muscles and joints become stiff more quickly. It is important to be sure they are in a good position to help maintain their range of motion and comfort.

STRETCHING

Stretching involves moving an individual's body part (ex: arm or leg) to their comfortable, available end range of motion with a slow, gentle movement and holding the position for a specific length of time.

STANDER

Standers are custom-fitted equipment designed to help an individual stand up for a prolonged amount of time.

SERIAL CASTING

Serial casting describes the process used to place a series of rigid casts over an individual's joint (most often used for heel cords at the ankle). The casts are used to slowly (with frequent cast changes over several weeks) increase range of motion at a specific joint. The joint is stretched "slightly" further with each new cast placement.

GUIDELINE SPECIFICS

ANKLE NIGHT SPLINTS

- Starting at a young age may improve tolerance & wear time.
- Most useful before your child gets tight or stiff muscles.
- If NOT tolerated at night, splints may be used for daytime positioning, when seated or lying.
- Custom-made braces from an experienced orthotist are recommended. Off-the-shelf night splints may not fit as well, may be less tolerated, uncomfortable, and are not made specific to your child.

Good to Know
Your PT or orthotist will know the right recommendations for your child.

Caution
Night splints may not work for everyone. The position of the foot and ankle will determine if your child is a candidate for night splints – always check with an experienced PT in Duchenne that is familiar with your child.

Examples of commonly used orthoses:

Not all night splints will look like these, however the design should be similar with the idea to hold the ankle for a maintained period of time.

Photo images courtesy of Cascade DAFO

RESTING HAND SPLINTS

(not intended to be used during functional activities)

- Considered for older children or adolescents when signs of finger and wrist tightness is noted.
- More likely to be tolerated if resting splints are worn on one arm at a time.

Good to Know
- Consult your experienced OT/PT, and orthotist for individual recommendations. A custom fitting is suggested.
- Resting hand splints are not for everyone.

Caution
Not recommended for adolescents and adults where the hand and fingers are fixed in fisted position.

Fingers and wrists tend to get tight over time as seen in this photo:

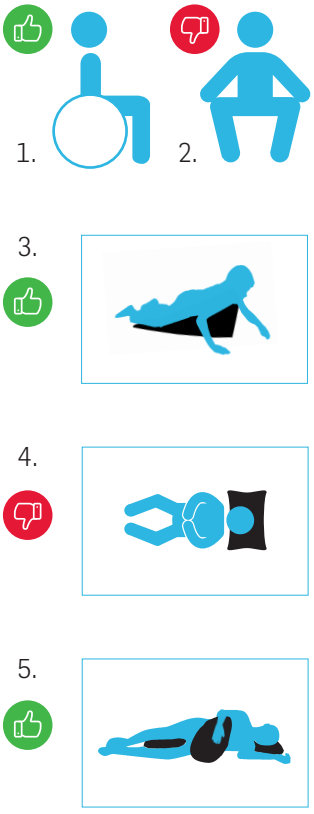
A typical wrist and finger splint used when wrist and fingers are tight:

POSITIONING

- Proper positioning may assist in maintaining range of motion.
- **Ideal seated positions:**
  1. Both feet flat on the floor or footrest (or box step for younger boys), with hips & knees at a 90° position and back & pelvis aligned with knees in line with hips.
  2. Avoid long periods of time with legs in frog position (splayed out). Knees should be in line with feet & hips.

Good to Know
- Changing positions regularly is recommended.
- Positioning may help decrease complaints of pain and discomfort for those who have muscle and joint stiffness in the arms, legs, and back.
- Your PT will know what is best for your child in your own environment.

Caution
- Prolonged periods of time in any position may increase the risk of skin breakdown, discomfort or pain, and further tightness. (Example: lying on back with the knees propped with pillows)
- Limit the amount of time the knees are positioned apart. Your PT will have suggestions for your child.

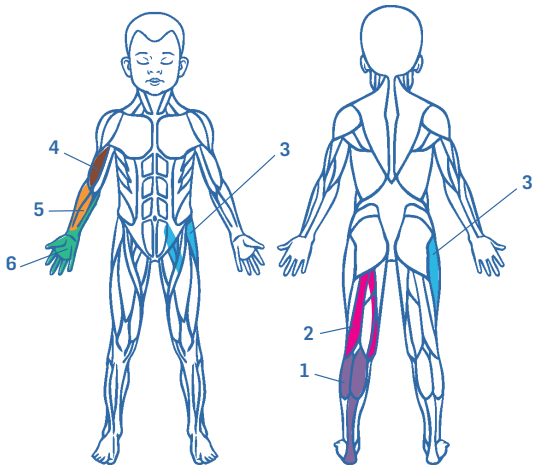


Limit amount of time with legs open wide as seen in examples 2 and 4 above.

- **Ideal positioning when lying down:**
  3. Lying on your stomach can provide a long stretch to hip muscles if comfortable & tolerated –placing a pillow or wedge under the chest may be helpful.
  4. When lying on back: position legs as straight as possible but comfortable (do not force the legs straight), support tight hip and knee muscles with pillows or blanket or towel rolls for comfort. A pillow under the knees may relieve pressure on the back. Be sure to alternate positions.
  5. When lying on side, support the body with pillows or towel rolls. Support between the thighs can reduce strain on the back & hips. Supporting an arm may provide additional comfort.

STRETCHING

– **Potential benefits of doing stretching may include:**
  – Temporary improvement in blood flow to the muscle
  – Decrease report of discomfort
  – A feeling of wellbeing
  – Temporary increase in tolerance to stretch
  – Temporary decrease in muscle stiffness
  – Continued movement through the joint's full available range of motion



– **Muscles most commonly developing tightness:**

Legs

1. Calf muscles (often referred to as heel cords):
   – tightness limits ability to move ankles up or stand with the feet flat
2. Hamstrings (located behind the hip and knee):
   – tightness limits ability to straighten knee
3. Hip muscles (often referred to as hip flexors and/or hip abductors):
   – tightness limits ability to lie flat on stomach or move leg towards the middle of body bringing the legs together

Arms

4. Elbow flexors:
   – tightness limits ability to straighten elbow
5. Forearm pronators:
   – tightness limits ability to turn hand/palm up
6. Long finger flexors:
   – tightness limits ability to straighten fingers

– **General guidelines for stretching:**
  – Hold the position of the arm or leg when you feel tightness or resistance of the joint or muscle.
  – General recommendations are a 30 second hold. Let your PT guide you on repetitions and frequency.

Good to Know

– There may be benefits to stretching.
– Prioritize your routine based on your PT's recommendations and your child's condition.
– Stretching may not prevent muscle tightness from developing. Individuals will continue to develop tightness over time. The progression of tightness varies from one person to the next with Duchenne.
– Stretching may not fit into your daily routine. THAT'S OK! Talk to your PT to help you prioritize.

Caution

– Bones, muscles, and tendons can be fragile so stretching should be slow, gentle, ensuring proper alignment. NEVER go beyond point of resistance, tightness, or tolerance. AVOID fast or forceful movements.
– Pushing too far during stretching may not provide added benefit.

STANDING DEVICES

– Walking activities are more beneficial than supported standing, if safe.
– Standers could be considered in children who are unable to stand by themselves, have difficulty standing, or have lost the ability to walk.
– **Options for standing devices:**
  1. Sit-to-stand standers – allow a person to go from sitting to a standing position
  2. Supine standers – provide support along the back of the body. The child lies down and then is brought to standing.
     **These standers can be more difficult for the caregiver.*
  3. Wheelchairs that transition to standing

Good to Know

– Active standing may be beneficial for bones and muscles.
– Standing can make people feel good.
– There may be a delay in equipment arrival after ordering, so begin the process prior to loss of range of motion and ability to stand and walk.

Caution

– The effects of standing on bone mineral density is not well understood in this population.
– Standing when there is a loss of range of motion (ROM) – i.e. unable to straighten the knees or hips, unable to get a good foot position, or to keep the back flat (i.e. arched away from surface) – may be painful. DO NOT push through pain.
– Standing with muscles and joints in a poor alignment is not considered beneficial.

SERIAL CASTING

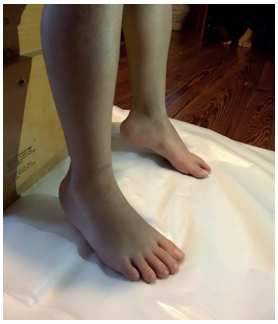
- **Serial casting may improve ankle range of motion (ROM) in select individuals with Duchenne who:**
  1. Have mild ankle tightness at the heel cord
  2. Have the ability to straighten their leg when sitting
- Serial casting must be applied & managed by a PT experienced in serial casting in Duchenne.
- Frequency of cast change may vary by site but should generally include cast changes 1-2 times per week.
- Demonstration of improved ankle range of motion between cast changes should be present to continue serial casting – consult your expert.
- Total time of casting should be about 4 weeks.

Good to Know

- Serial casting should only be done by an experienced PT.
- ROM gained with this option is usually temporary but may assist a child who is still walking.
- Night braces are commonly prescribed after serial casting.
- This option is considered when certain criteria mentioned above is met.
- Consult a therapist or orthotist familiar with serial casting.

Caution

- Assess for signs of skin irritation (redness that doesn't go away within 20 minutes) or blisters noted with cast changes.
- NOT recommended for individuals with severe muscle tightness.
- NOT recommended for individuals who can't walk in the cast.
- NOT recommended for individuals who can't get up from the floor without a chair or help from someone.
- NOT recommended for an individual that cannot straighten their knee while sitting in a chair or on the edge of a table.
- Limit total duration of casting to about 4 weeks since long periods of immobilization may impact strength.



Prior to casting

Preparing to cast

This boy is wearing his shoes over the casts and demonstrates he can stand and walk with the casts on. Not all casts will fit in regular shoes. In that case a walking surface can be added to the bottom of the cast.