



# Duchenne Muscular Dystrophy... An Update

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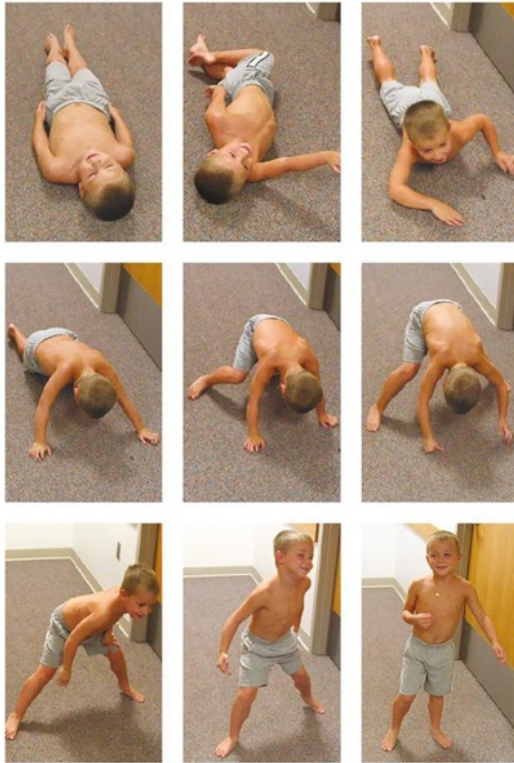
# Faculty/Presenter Disclosure

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We have nothing to disclose

# Duchenne Muscular Dystrophy



Dystrophin gene mutation

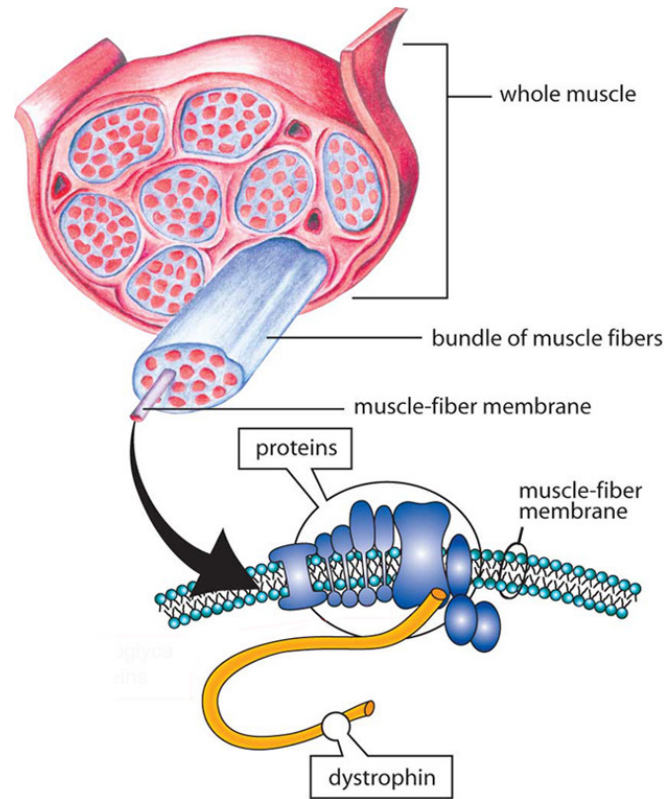
- ↓ Dystrophin protein = ↓ muscle repair = progressive muscle loss

Lack of Dystrophin: brain, lungs and heart

Early Signs & symptoms - Gower's, motor & speech delays, calf hypertrophy

Impacts: ↓ muscle strength, ↓ mobility, cardiac & respiratory, musculoskeletal, bone density, behavior, learning, language, communication, mental health

Phases: pre-symptomatic/diagnosis, early ambulatory, late ambulatory, transition/early non-ambulatory, non-ambulatory



# Advances in DMD Care

Genetic sequencing → Phenotype specific treatment

Not all DMD is the same – different trajectories & Treatment

• Standard - steroid treatment: (start early) Deflazacort, Prednisone (may extended amb 2-3 yrs, preserve cardio-resp function)

On going studies, new treatments, e.g.:

- Vamorolone (dissociative steroid) – extension studies
- Ginivistat (enzyme blocker) – may ↑ muscle repair – phase 3 trial & open label extension)
- Gene therapy coming.... Viral vectors, exon skipping.....

<https://www.parentprojectmd.org/duchenne-drug-development-pipeline/>

<https://musculardystrophynews.com/2021/03/24/mda2021-long-term-vamorolone-disease-progression-dmd-boys-phase-2-trial/>

# Clinical Implications



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1. Increased longevity, (Fa)
  2. Longer ambulation, some may be walking when 'graduating' from our services,
  3. Fewer and/or delayed orthopedic surgeries
  4. Improved Cardio-Respiratory status.
- Importance of transition to adult services (Patricia)
    - Referral to specialized adult clinics (neurologist, physiatrist, PT and OT, cardiologist, respirologist, endocrino/bone health, orthopedist, dietician, psychosocial support)
    - Vocational, education and employment/career

# Therapy Implications



# Therapy Implications (Fa)

1. Predicted trajectory/individual progression of symptoms
2. Importance of basics:
  - Start stretch early – establish daily routine
  - Safe exercise: prescribed and specific,
  - Start overnight AFOs early,
  - Healthy lifestyle and good nutrition,
  - Early power mobility,
  - Recreation (adapted and regular),
  - Future planning mobility supports and adaptations, home renovations, learning supports



# Team Approach

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## Community & Tertiary Care Links

- Early intervention: (Fa)
  - Establishing early therapy routines
  - Help child understand/deal with diagnosis according to age/cognition
  - Supporting developmental, behavioural needs
- School Age: (Patricia)
  - Education of peers/principal/teachers/SEAs
  - Supplement school therapy with private PT/OT services with AHP
  - Mobility supports
  - Home accessibility & care equipment
  - Ongoing learning & behavioural supports
  - Fun & Friends – access to recreation and community participation
  - Future - early planning to transition to adult, taking charge of their health/well-being

# Resources



## Neuromuscular and DMD Organizations

- Parent project <https://www.parentprojectmd.org/>
- Muscular Dystrophy Canada <https://muscle.ca/>
- Jesse's Journey <https://www.jessesjourney.com/>

Signs & Symptoms: <https://www.parentprojectmd.org/about-duchenne/is-it-duchenne/signs-and-symptoms/>

Neuromuscular Disease Network for Canada <https://neuromuscularnetwork.ca/>

# Resources



## Clinical practice guidelines

Treat NMD <https://treat-nmd.org/care-overview/>

Care Guidelines Parent Project <https://www.parentprojectmd.org/care/care-guidelines/Rehab Management of DMD.pdf>

## Learning & Behaviour Guides

<https://www.parentprojectmd.org/care/care-guidelines/by-area/learning-and-behavior/>

<https://www.musculardystrophyuk.org/wp-content/uploads/2015/02/DMD-Learning-and-behaviour.pdf>

[https://media.gosh.nhs.uk/documents/Learnin\\_and\\_behavioural\\_toolkit\\_final.pdf](https://media.gosh.nhs.uk/documents/Learnin_and_behavioural_toolkit_final.pdf)

<https://muscle.ca/access-help/personal-support-program/educational-resources/>

# Resources



## Community Resource Contacts

- BC Children's Hospital – OT/PT department: 604-875-2123
- Sunny Hill Centre – Neuromotor Program
- GF Strong – Adolescent and Young Adult Program
- Jooay app – for adapted recreation: <https://jooay.com/>