WELCOME!!!!!!!!!!!!!
Overview of Comprehensive Duchenne Care

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About Duchenne
**Duchenne & Rare Disease**

Approximately 50% of the people affected by rare diseases are children.

- Rare diseases affect about **30 million Americans**, or 1 in 10 people.
- It is estimated that **350 million people worldwide** suffer from rare diseases.
- About **30% of children** with these debilitating diseases **will not live** to see their 5th birthday.
- About **80% of rare diseases** are genetic.
- Rare diseases **impact more people** than AIDS or cancer combined.
- **95% of rare diseases** do not have a single FDA-approved drug treatment.

Source: Global Genes
About Duchenne Muscular Dystrophy

- X-linked, pediatric neuromuscular disease, with onset in early childhood
- Incidence rate: 1:5000 boys (30% spontaneous), 20,000 new dx/year
- Diagnosis: 3-5 years of age
- Predictable course, variable timeline
- Multi-systemic condition
- Progressive loss of function
- 100% lethal, typically by late 20’s
Duchenne affects more than just muscle

Dystrophin found in:
- Skeletal muscle
- Cardiac muscle
- Smooth muscle
- GI/GU
- Brain
- Retinas
- Kidney

- POSSIBLE LEARNING AND COGNITIVE DIFFICULTIES (speech, learning, behavior, sensory, psychosocial)
- DECREASED HEART FUNCTION
- CARDIOMYOPATHY
- LEADS TO HEART FAILURE
- WEAKENS DIAPHRAGM
- REQUIRES VENTILATION IN TEENS
- LEADS TO PNEUMONIA
- LOSS OF MUSCLE MASS
- WEAKNESS
- INFLAMMATION
- FIBROSIS
- BRITTLE AND WEAK
About PPMD
PPMD fights every single battle necessary to End Duchenne.
Parent Project Muscular Dystrophy fights to end Duchenne. We accelerate research, raise our voices to impact policy, demand optimal care for every single family, and strive to ensure access to approved therapies.
PPMD has played a vital role in every single victory in the fight to end Duchenne since 1994.

1994
PPMD founded and 1st Annual Conference

1997
PPMD opens 1st Duchenne Muscular Dystrophy Research Center

2000
1st Annual Advocacy Conference held

2001
MD CARE Act signed into law

2003
PPMD funds 1st Duchenne Drug Discovery Program

2006
PPMD and CDC convene community through it leaders to develop Standards of Care

2009
Standards of Care published

2012
PPMD registered the way for FASDA to be passed by Congress, mandating regulatory flexibility for rare diseases

2014
PPMD leads a community effort to draft Guidance for the FDA around Duchenne

2015
PPMD launches the 1st Duchenne-specific registry

2016
1st spot in Duchenne – Evenroses

2017
PPMD announces largest grant to date – $3.5 million – and launches Gene Therapy Initiative

2018
Expand Gene Therapy Initiative
- Certify additional Duchenne Care Centers (LB have been established)
- Launch Global Certified Duchenne Care Center Program with first international certification
- Expand Duchenne registry to more countries (Australian portal added this year)
- Continue leading efforts to obtain an ICD code for Duchenne
- Continue leading Newborn Screening effort to ensure early diagnosis
- PPMD End Duchenne Tour to visit 8 more states, both served/engaged and underserved populations
PPMD TEAM!

- Pat Furlong, President, CEO
- Amanda Weiglein, Care
- Jody Gabbard, Care
- Annie Kennedy, Legislation and Public Policy
- Ryan Fischer, Community Engagement
- Abby Bronson, Research Strategy
- Ann Martin, DuchenneConnect
The Muscular Dystrophy Community Assistance, Research & Education (MD-CARE Act)

**MD-CARE Act 2001**
Centers of Excellence
MD STARnet tracking and surveillance
MD Coordinating Committee – Action Plan for Muscular Dystrophies

**MDCA Reauthorized 2008 – (MD-CARE Act 2)**
Added the National Heart, Lung, and Blood Institute to MDCC
Enhancement of clinical research
Expansion of MD-STARnet
Duchenne Care Considerations – Develop and Disseminate

**MDCA Amended in 2014 – (MD-CARE Act 3)**
Expanded research to focus on Endocrine, Pulmonary and Cardiac, Transitions
Additional federal agencies added as members of Coordinating committee
Sharing of data from MD-STARnet
Expansion of care considerations to include Duchenne adult population & reflect updates in care
Update of MD Action Plan
Results of 3 iterations of MDCA

Care
Care considerations published
Care much more standardized (clinics)

Research
Wellstone Centers of Excellence
Animal Studies
Basic and Translational Research grants

Federal Coordination and funding
Coordinating Committee grows
Research Plan for MD’s
500+million in Duchenne Funding

Data collection
Outcome measures
Natural History Studies
MD-STARnet Surveillance

Drug Development
45+ companies
Pipeline full of hope
Care Considerations

• Originally published in 2010
  – 8 subspecialty areas of care/recommendations

• Update began in 2014 (published!)
  – 11 subspecialty areas of care
  – Algorithm for surveillance/management of each subspecialty area across the lifespan
  – Evaluation tool
  – Checklist for parents/patients/providers
  – 3 volumes of Lancet (core guidelines)
  – Pediatrics (subspecialty articles) – coming soon!
### Original vs. Updated Care Considerations

<table>
<thead>
<tr>
<th>Original Care Considerations</th>
<th>Updated Care Considerations</th>
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<tbody>
<tr>
<td>8 subspecialty areas of care</td>
<td>11 subspecialty areas of care</td>
</tr>
<tr>
<td>• Diagnosis</td>
<td>• Primary and emergency care</td>
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<tr>
<td>• NM management</td>
<td>• Endocrine management</td>
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<tr>
<td>• Rehabilitation management</td>
<td>• Transition of care/adult care</td>
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<td>• GI/nutrition</td>
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<td>• Respiratory</td>
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<td>• Cardiac</td>
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<td>• Orthopedic/surgical management</td>
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<tr>
<td>• Psychosocial</td>
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<td>Pediatric focused</td>
<td>Across the lifespan</td>
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Development of the Care Considerations

- Organization of the CC Working Group Steering Committee
- Identification of 11 subspecialty areas of care
- Organization of subspecialty committees
- Guidance based on evidence (where it existed) and expert opinion (where evidence did not exist)
Certified Duchenne Care Center Program

– Create a network of centers that:

• Provide standardized care in alignment with the Standards of the Certified Duchenne Care Center Program, which are in agreement with the CDC Care Considerations
Goals of CDCC

- Improved access to comprehensive Duchenne care and services
- Standardized care across network of institutions
  - Many major clinical trial sites
  - Strengthened clinical trial outcomes
  - Speed novel therapies from bench to bedside
Process of Certification

Center Clinical and Subspecialty Services,
Duchenne Care Surveys

Parent/patient report:
DuchenneConnect and Clinical Experiences Survey

Site visit, faculty/staff Interviews, chart review
Comprehensive Duchenne Care Team