PPMD End Duchenne Tour
Parent Project Muscular Dystrophy

Overview

Annie Kennedy
Senior Vice President
Legislation & Policy
Research
Advocacy
Care
Education
Grassroots Advocacy

Supporting every single diagnosis
Amplifying every single voice
Caring for every single person
Connecting every single family
Investing in every single therapy

Parent Project Muscular Dystrophy

PARENT PROJECT MUSCULAR DYSTROPHY | ENDDUCHENNE.ORG
1994 - Questions and Missing Pieces

- What was the current state of research and care in Duchenne?
- What was the federal government investment in Duchenne?
- What data existed about the Duchenne population?
- How do we move the needle toward better care and treatments for Duchenne?

Critical Missing Pieces

- Care standards
- Natural history data
- Outcome measures
- Federal investment & coordination
- Companies
- Mobilized grassroots community
Results 2001 - Present

**Care**
- Care considerations published
- Care much more standardized (clinics)

**Research**
- Wellstone Centers of Excellence
- Animal Studies
- Basic and Translational Research Grants

**Data collection**
- Outcome measures
- Natural History Studies
- MD-STARnet Surveillance
- ICD-10 Code

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

**Drug Development & Access**
- 2 Approved Therapies
- 45+ Companies
- Pipeline Full of Hope
- Pre-competitive, collaborative consortium (PPMD Duchenne Drug Development Roundtable)
- Payer Engagement
The Muscular Dystrophy Community Assistance, Research & Education Act (MD-CARE Act)

**MD-CARE Act 2001**
Established:
- Centers of Excellence
- MD STARnet tracking and surveillance
- MD Coordinating Committee
- MDCC to develop ‘MD Action Plan’

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

**Paul D Wellstone MD-CARE Act Amendment 2014**
- Expanded research to focus on Endocrine, Pulmonary, Cardiac & Transition into Adulthood
- Additional federal agencies added as members of Coordinating committee
- Sharing of data from MD-STARnet with community researchers
- Update of DBMD Care Considerations to include adults with Duchenne & reflect care advances since 2010
- Update of initial ‘MD Action Plan’ by MDCC
The PPMD community has led the passage of 5 Congressional Bills!

Muscular Dystrophy Community Assistance Research & Education Act (MD-CARE Act) 2001

Paul D. Wellstone MD-CARE Amendments 2008

Paul D. Wellstone MD-CARE Amendments 2014

Patient Focused Impact Assessment (PFIA) Act (2016) (became a key provision of 21 CC)

Better Empowerment Now to Enhance Framework and Improve Treatments Act of 2017 (BENEFIT Act)
Federal Agencies We Currently Collaborate With

- Department of Defense
- FDA
- CDC
- NIH
- Administration for Community Living
- Social Security Administration
- U.S. Department of Health and Human Services
- Health Resources and Services Administration
- Department of Education
- CMS

END DUCHEENNE.ORG
PPMD Hill Days
Building Congressional Champions

Appropriations Bill & Annual Report Language
• The only annual Advocacy Conference focused on Duchenne

• Focused on advancing MD-CARE Act as well as regulatory policy

• Prioritize impacting:
  • Food and Drug Administration (FDA)
  • Center for Disease Control (CDC)
  • National Institutes of Health (NIH)
  • Department of Defense (DOD)
• Advocating and participating in the development of a Standard of Care (recently updated)

• Established the Duchenne Certified Care Center Program

• Focusing on gaps that exist in care sub-specialties
Duchenne Care Considerations – Published January 2018

Duchenne Muscular Dystrophy Care Considerations

Duchenne muscular dystrophy (DMD) is a rare genetic (inherited) disease defined by muscle weakness that gets worse over time and ultimately affects the heart and lungs. People born with DMD will see many healthcare providers throughout their lives.

The updated care considerations for DMD are contained in a set of three articles published in *Lancet Neurology*. They offer the latest clinical considerations to improve care and quality of life for people living with DMD. Clinicians who specialize in treating people with DMD developed these care considerations based on current evidence for optimal care reported in the literature and their own clinical experiences.

As science and medicine are advancing, people with DMD are living longer; therefore, their care throughout life is evolving as well. These care considerations, funded by the Centers for Disease Control and Prevention (CDC), are intended to raise the standards of care, help clinicians provide the best possible care to people with DMD, and give families and caregivers the necessary information to manage their care.

Read the articles here:

- [Diagnosis and management of Duchenne muscular dystrophy. part 1: Diagnosis, neuromuscular, rehabilitation, endocrine, and gastrointestinal and nutritional management](#)
- [Diagnosis and management of Duchenne muscular dystrophy. part 2: Respiratory, cardiac, bone health, and orthopaedic management](#)
- [Diagnosis and management of Duchenne muscular dystrophy. part 3: Primary care, emergency management, psychosocial care, and transitions of care across the lifespan](#)

- All sections updated to reflect progress in care & research
- All sections updated to reflect care through the lifespan of Duchenne, not just pediatric care
- New sections within 2018 CDC Care Considerations:
  - Primary/Emergency Care
  - Endocrine Management
  - Transition of Care for Teens & Adults
An Introduction to the Updated 2018 Clinical Care Guidance on the Diagnosis and Management of Duchenne

Neurology Care, Diagnostics, and Emerging Therapies of the Patient with Duchenne

Rehabilitation Management of the Patient with Duchenne

Bone Health and Osteoporosis Management of the Patient with Duchenne

Obesity and Endocrine Management of the Patient with Duchenne

Nutritional and Gastrointestinal Management of the Patient with Duchenne

Respiratory Management of the Patient with Duchenne

Cardiac Management of the Patient with Duchenne

Orthopedic and Surgical Management of the Patient with Duchenne

Primary Care and Emergency Department Management of the Patient with Duchenne

Psychosocial Management of the Patient with Duchenne

A Transition Toolkit for Duchenne

Evaluating Implementation of the Updated Care Considerations for Duchenne
# A Transition Toolkit for Duchenne Muscular Dystrophy: Supplementary Materials

## Supplementary Material 1: Transition Readiness Assessment for Young Adults with Duchenne Muscular Dystrophy

Please fill out this form to help us see what you already know about your health, how to use health care and the areas you need to learn more about. If you need help completing this form, please ask your parent/caretaker. Then we’ll review the form together to develop a plan.

<table>
<thead>
<tr>
<th>Date: ______________</th>
<th>Participants in today’s discussion:</th>
</tr>
</thead>
<tbody>
<tr>
<td>Name: ______________</td>
<td></td>
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<tr>
<td>DOB: _______________ Age: _______</td>
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### Transition Importance and Confidence

On a scale of 0 to 10, circle the number that best describes how you feel right now.

| How important is it to you to prepare for/change to an adult doctor before age 22? |
|---------------------------------|-----------------|-----------------|-----------------|-----------------|-----------------|-----------------|-----------------|-----------------|-----------------|
| 0 (not)                         | 1               | 2               | 3               | 4               | 5               | 6               | 7               | 8               | 9               | 10 (very)       |

| How confident do you feel about your ability to prepare for/change to an adult doctor? |
|---------------------------------|-----------------|-----------------|-----------------|-----------------|-----------------|-----------------|-----------------|-----------------|-----------------|
| 0 (not)                         | 1               | 2               | 3               | 4               | 5               | 6               | 7               | 8               | 9               | 10 (very)       |

*Publication pending; Shared with permission Pediatrics*
ADVANCES IN RESEARCH

• Invested over 50 million in research, leveraging $500 million in federal dollars and billions in private dollars

• Have worked to establish a research infrastructure to enable drug development

• 2 approved therapies with over 25 active treatment trials and over 40 companies in the space
Treating Duchenne

- Dystrophin Restoration/Replacement
- Steroid Replacement
- Gene Therapy
- CRISPR/Cas9
- Stop-Codon Readthrough
- Anti-Fibrotics
- Inflammation & Fibrosis
- Calcium Regulation
- Ryanodine Receptors
- Calcium Homeostasis
- Myostatin Inhibition
- Follistatin Upregulation via Gene Therapy
- Muscle Growth and Protection
- Selective Androgen Receptor Modulators
- Utophin Upregulation
- Stem Cells
- Cardiac
- Blood Flow
- Mitochondria
- Mitochondrial Biogenesis
- Mitochondrial Enhancers
- nNOS Upregulation
- Traditional Cardiac Drugs

Parent Project Muscular Dystrophy | EndDuchenne.org
For over two decades, Parent Project Muscular Dystrophy (PPMD) has contributed to each stage of the drug development pipeline, awarding grants, filling in critical gaps, convening stakeholders, and redefining the clinical trial landscape.
FDA Guidance on Duchenne Muscular Dystrophy

Finalized February 2018

“The newly finalized Guidance ... was preceded by a pioneering effort from Parent Project Muscular Dystrophy who, in 2014, submitted their own independent proposed draft guidance that provided important scientific and patient input from the DMD community.

It helped inform the FDA’s development of both our own draft guidance and the final version issued today.”

-Commissioner Scott Gottlieb
• All federal partners invited
• White paper published prior to the meeting
• Webcast & live polling
• Compass Report published June 2018
Goals:

– To identify current policy, care, and clinical trial priorities among our Duchenne community members (by sub-population).

– To begin to identify measures of impact not currently captured in health economic models or value frameworks.

The Audience: Included federal agency & pharmaceutical industry partners

More than 400 members of our Duchenne community participated
It’s About Access
Our Path to Access?

Photo credit: Conrad Reynolds
ACCESS & COVERAGE RESOURCES

With the first two FDA approvals of Duchenne therapies, the Duchenne community has entered in a new environment for access and reimbursement. With this brave new world at play, PPMD has been leading the effort to educate and guide the community through these unchartered waters.

OVERVIEW OF THE FUNDAMENTALS OF ACCESS

- What are the major differences with Medicaid vs private insurance?
- What is the process for appealing a denial, what are the layers of appeal?
- What are Medical vs. Drug Benefits?
- What is the role of a PBM’s or Pharmacy Benefit Managers?
- Types of Insurance Coverage?
- What are formularies?

These webinars are non-product specific, but can provide you with a basic understanding about the path to access.
Roadmap for Navigating Path to Access

**STAGE 1**
Prescription written by Doctor and submitted to Insurer

**STAGE 2**
Patient, Drug Company, and Prescribing Doctor work with Insurer to process prescription

**STAGE 3**
Drug is provided to Patient
### Stage 1 Resources:

<table>
<thead>
<tr>
<th>Drug</th>
<th>Resources</th>
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| **EMFLAZA**| - Emflaza is a corticosteroid that demonstrates anti-inflammatory and immunosuppressant effects. Emflaza is available in an immediate-release tablet formulation at multiple dosages (6 mg, 18 mg, 30 mg, 36 mg) as well as in an oral suspension formulation (22.75 mg/mL). Emflaza label and prescribing information  
  - Please contact EmflazaCares to be connected with a case manager who can help you begin the process for access. EmflazaCares will contact your physician for the start form and the prescription.  
  - Emflaza FAQs  
  - Learn about the AssistanceFund |
| **EXONDYS 51** | - EXONDYS 51 is an antisense oligonucleotide indicated for the treatment of Duchenne muscular dystrophy (Duchenne) in patients who have a confirmed mutation of the Duchenne gene that is amenable to exon 51 skipping. FDA label document  
  - Unsure if you are amenable to this therapy? Try our Deletion tool to perform a search on your mutation |

**For U.S. Residents:**

- For those amenable to exon 51 skipping please contact SareptAssist to be connected with a case manager who can help you begin the process for access. SareptAssist will contact your physician for the start form and the prescription.  
- Learn about the AssistanceFund  
- Sample Letter of Medical Necessity (for Clinicians/Doctors)  
- SareptAssist Patient Services Overview [PPMD Webinar Recording]

**If you live outside of the U.S.:**

- Sarepta supports a Managed Access Program (Expanded Access Program) for those residing in numerous countries, including: Argentina, Brazil, Canada, Colombia, France, Germany, Greece, Iceland, Italy, Mexico, Spain, Turkey, and the United Kingdom.  
- For more information about eligibility and access, visit Sarepta’s Managed Access Program.  

![STAGE 1 Prescription written by Doctor and submitted to Insurer](red Emblem)
WHAT HAPPENS IF ACCESS IS DENIED BY MY INSURER?

It is at this stage a complication could occur.

If you receive a denial, there are steps you can take in order to appeal the decision. Your clinician will be a key advocate in this process. The denial letter should have specific information about the appeals process.

The clinician or doctor can request what is called a “peer to peer” where your clinician will interact directly with the insurer one-on-one to provide justification for access and coverage.

Things to Consider:

- Ask your clinician and the insurer about whether a “peer to peer” can occur and how to initiate.
- Write your own Patient Appeal Letter (sample below)
- Has your clinician submitted a Letter of Medical Necessity? (sample below)

Tips for Advocating for Access:

- Track all communications with the drug company, health insurer, and clinician/doctor. Save all letters and emails and write down who you speak with and when. Request letters sent by your physician.
- Consider contacting your state Consumer Assistance Program (CAP) or Department of Insurance during an appeal process. Visit localhelp.healthcare.gov.
• **PPMD Convened Meeting of Stakeholders to Discuss Model of Pilot:** October 2017; Pilot launched October 2018

• **Complete CK Assay Validation Studies:** FDA approval of PKI kit expected Q1 2019

Contact Annie@parentprojectmd.org
State Specific Resources –
22 PPMD Connect Groups & Growing
Tracking payer determinations by state & payer and integrating them into our website...
PPMD School Advocacy Resources

Brain Pop Video
PPMD’s 4-minute animated video about Duchenne for kids of all ages.
https://www.youtube.com/watch?v=6wLnR7GJakY

Education Matters
Helpful tips on how to talk about Duchenne
Issues to be aware of at school
An Individualized Education Plan (IEP) overview
Sample scenarios and suggested ways to respond
A Parents Guide
A Teacher’s Guide
Adaptive PE
Learning & Behavior in Duchenne

School Presentation Materials
‘Chocolate Chip Cookie’ class presentation

IEP, 504 Plan, and School Accommodation Considerations
IEP vs 504 Plan side by side comparison
Tips & tools for creating IEPs and 504 for students with Duchenne & Becker

https://www.parentprojectmd.org/care/for-families/classroom-resources-for-teaching-about-duchenne/
PPMD School Webinars

Navigating School: An Ongoing Journey
https://www.youtube.com/watch?v=M1py6M9ce0I

From Where We Sit (Adults with Duchenne):
‘PCA’ – 3 Little Letters that Mean So Much
https://www.youtube.com/watch?v=5lBEdb4IBIk

Promoting Independence In Children with Duchenne:
Panel of Parents Whose Sons Live Away from Home Reflect on How they Navigated the Delicate Balance of Protecting Their Sons, While Fostering Independence
https://www.youtube.com/watch?v=vtWbyt-j-lA
PPMD Adult Advisory Committee

Nominations Being Accepted Through Early November
Transitions Initiative

PPMD’s recent National leadership –

• MD CARE Act 2014
  • CDC MD STARnet – Transitions surveys & data elements
  • MDCC Action Plan ‘Living With’ section
  • Care Considerations, Lancet January 2018
  • Transitions Tool, Pediatrics supplement – published October 2018 Care Considerations Transitions section

PPMD Adult Advisory Committee (PAAC)

Transition to Care (T to C) federal coalition established –
PPMD among 4 advocacy partner leads, with CDC, NICHD Coalition meeting convened, June 2018
Flextension

Innovator:
Arjen Bergsma & Micha Paalman

Community Member:
Justus Kuijer

X-Ar Arm

Innovators:
Blake Mathie & NJIT

Community Member:
Zach Smith
PPMD conducted community survey of preferences related to robotics in 2015

Clear priorities among the highly valued activities include:
- standing from a seated position
- picking up an object from the floor
- repositioning oneself at night, and
- the ability to bring one’s hands to mouth
Thank you!