DuchenneConnect: Self-Report Registry for Duchenne and Becker Muscular Dystrophy

Holly Peay, PhD
RTI International
PI, DuchenneConnect
Disclosure

- Holly Peay received research support from Santhera Pharmaceuticals
DuchenneConnect

- Online self-report registry and educational resource for individuals with DBMD and carrier females
- 3,000+ registrants
- Established by Parent Project Muscular Dystrophy in 2007
Decode Duchenne

A new genetic testing program that allows patients with Duchenne or Becker muscular dystrophy to access to genetic testing. Decode Duchenne is administered by DuchenneConnect and Parent Project Muscular Dystrophy and is funded by Sarepta Therapeutics.

How Your Data Is Being Used

Have you ever wondered how your data in DuchenneConnect is used? Learn about studies and clinical trials that have utilized the DuchenneConnect data and/or recruited DuchenneConnect registrants.

Registry Data

See how you or your child compare to others in the registry. Families, clinicians and researchers can learn from the registry. Login to your account today to explore the newest information from more than 3,000 participants.
Ann Test 1

MEDICAL SURVEYS
- Researcher Questions
- GI/GU Module
- Heart Module
- Corticosteroid Module
- Muscle Function Module

ACCOUNT INFORMATION
- Update

Diagnosis: Confirmed carrier (I am a carrier of Duchenne or Becker and I do not have any symptoms)
Age: 44
Country: UNITED STATES

TESTING RESULTS
++ Add
- Genetic Results (1)
  1431657063.Ann_Emory_Genetic_results.pdf
  Created: 2015-05-15

- Cardiac Testing Results (1)
  1431657090.Ann_Echo_report.pdf
  Created: 2015-05-15

- Clinical Notes or Records (1)

HEALTHCARE PROVIDERS
- Add
  - Brenda Wong
    Neurology
  - Linda Cripe
    Cardiology

INVITATIONS
- Invite Others
  Invite friends or extended family members with Duchenne or Becker to join.

FAMILY MEMBERS
++ Add
- Ann Test 1
- John Martin

GROWTH MEASUREMENTS
++ View Chart
- Height
  2014-06-16 - 170.00 centimeters
- Weight
Registry Services

- Prep-to-research data, protocol advising, and recruitment for clinical research
- Collaborative studies with academic researchers/clinicians
- Expanding source of longitudinal natural history data
- Education and services to Registrant community
- Source of research priorities, advisors, and participants for DuchenneConnect/PPMD research studies
EDUCATION AND SERVICES FOR REGISTRANT COMMUNITY
Decide Duchenne: A Genetic Testing Program for Patients with Duchenne or Becker

DuchenneConnect, a program of Parent Project Muscular Dystrophy, announces a genetic testing program for patients with Duchenne or Becker muscular dystrophy. The program, called Decode Duchenne, is supported by Sarepta Therapeutics. It provides genetic testing at no cost to eligible patients who are unable to access testing due to barriers such as a lack of insurance or insufficient insurance coverage.

To participate in Decode Duchenne, patients must:

- Have a confirmed diagnosis or be suspected of having Duchenne or Becker muscular dystrophy based on clinical symptoms, as assessed by their treating physician, and have a positive creatine kinase (CK) test.
- Have not previously had genetic testing, or must require additional genetic testing to identify a causative mutation.
- Provide documentation to confirm the patient’s lack of insurance coverage, insufficient insurance coverage, or a denial of coverage for genetic testing.
- Be citizens or legal residents of the United States or Canada.
- Register on DuchenneConnect.

If you or your child need genetic testing, please contact the DuchenneConnect Coordinators at coordinator@duchenneconnect.org or (201) 937-1408 (Mon – Fri, 8am – 5pm EST). You will be sent an Application Form that must be completed and signed by your healthcare provider. You can also download and print the Application Form by clicking here.

Thank you to Sarepta Therapeutics for supporting this important initiative!
# Research FAQs

The following clinical trial or research FAQ sheets are family-friendly summaries of actively recruiting clinical trials and research studies. Many are for pre-clinical research that is soon to be in clinical trial. These FAQ sheets are written for the program book of the PPMD Annual Connect Conference (held every June). We house them on the DuchenneConnect site since they are a useful resource for families and professionals. Although we try to include the majority of studies in the United States, this is not intended to be a comprehensive list.

If you have any questions about the FAQ sheets, please email coordinator@duchenneconnect.org.

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<tr>
<td>ARM210 - Using ARM210 to Improve Muscle Strength &amp; Function in Duchenne</td>
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<tr>
<td>AT-300 - Akashi’s Novel Modulator of Stretch-Activated Calcium Channels</td>
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<td>BECKER MUSCULAR DYSTROPHY - A Natural History Study to Predict Efficacy of Exon Skipping</td>
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<td>BIGLYCAN - A Unique Urophin Upregulator</td>
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<td>BMS-986089 - Bristol-Myers Squibb’s Candidate for Myostatin Inhibition</td>
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<td>CARMSEAL-MD - Poloxamer 188 NF</td>
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<td>CAT-1004 - Catabasis’ Upcoming MoveDMD Trial</td>
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<td>CATENA®/RAXONE® - Phase 3 Study of Idebenone in Duchenne (DELOS)</td>
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Direct Access Webinars

DuchenneConnect and Parent Project Muscular Dystrophy present a webinar series that allows you direct access to experts in the fight to end Duchenne. Whether it's a researcher offering updates on a developing therapy, or industry providing clinical trial information, or experts in care and management, this series will be your resource to find out what is going on in the community from the people making it happen.

Upcoming Webinars

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<th>Date</th>
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<tr>
<td>TBA</td>
<td>PPMD Webinar on Caregiver Wellness</td>
<td>Amy Napoli, RDH</td>
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<td>June 10, 2015</td>
<td>Pfizer Webinar on Anti-Myostatin (PF-06252616)</td>
<td>Dr. Michael Binks, VP and Head of Clinical Research for Pfizer's Rare Disease Research Unit</td>
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<tr>
<td>May 13, 2015</td>
<td>Catabasis Webinar - Upcoming MoveDMD Phase 1/2 Clinical Trial with CAT-1004 in Boys with Duchenne</td>
<td>Dr. Joanne Donovan, Chief Medical Officer at Catabasis Pharmaceuticals</td>
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COMMUNITY-FOCUSED RESEARCH & ADVOCACY
Clinical Trial Decision Making

• Response to anecdotal reports of high expectations that were not met in trials
• Series of studies related to clinical trial expectations, hopes, decision influences, trial experiences
• Decision aid in development
Patient Focused Drug Development

- Food and Drug Administration Safety and Innovation Act (FDASIA) and Prescription Drug User Fee Act (PDUFA V)- 2012
  - Committed FDA to Patient-Focused Drug Development Program
  - Aims to inform FDA’s benefit/risk assessments and systematically obtain patient perspective on disease impact and treatment benefits
- Anticipating that Duchenne/Becker would not be chosen for an engagement forum...

(http://www.fda.gov/forindustry/userfees/prescriptiondruguserfee/ucm326192.htm)
Importance of Measuring Preferences

The FDA:

• Wants to improve their benefit-risk framework
• Is mandated to better understand the patient experience and preferences
• Is interested in testimony but… deals in data

Assessing meaningful benefit; tolerance for risk, burden and uncertainty informs drug development
Beyond Testimony: Advocacy-Led Stated Preference Research

Beyond Testimony: “Share Your Story”

“I understand the need for caution and care, but I also know that our children are dying. Parents should be able to decide the risk/benefit of a drug that has gone through and passed preliminary testing. I would rather my son die trying and fighting than waiting and wondering and wishing….I am one parent willing to take an educated risk!”
LEADING THE FIGHT TO END DUCHENNE

June 25, 2014

Guidance Document Submission
Division of Dockets Management (HFA-305)
5650 Fishers Lane, Rm. 1061
Rockville, MD 20852

Dr. Janet Woodcock,
Center for Drug Evaluation and Research
Food and Drug Administration
10903 New Hampshire Ave
Silver Spring, MD 20993-0002

Dear Dr. Janet Woodcock and colleagues at the FDA,

This correspondence constitutes a formal submission of a draft guidance authored by a consortium of stakeholders, under the coordination of Parent Project Muscular Dystrophy (PFMD), for consideration by the Food and Drug Administration (FDA). This material is intended as a submission to the docket as provided under the advice from the FDA’s guidances practice work group, with the expectation that FDA will seriously consider adoption of all or significant sections of this submission.

When FDA, PFMD and other interested parties met on December 11, 2013 in the spirit of public-private partnership to convene a Duchenne policy forum, we discussed the challenges designing and implementing clinical trials for rare diseases like Duchenne muscular dystrophy and the need to develop guidance to help accelerate development and the review of potential therapies for Duchenne muscular dystrophy (Duchenne). The forum culminated with an agreement that the Duchenne community, led by PFMD, would develop the first draft guidance on Duchenne for industry.

After an intensive five month long process, overseen by a steering committee, developed by working groups composed of clinical experts, developers and patients, and further reviewed by a community advisory board, we are pleased to hereby present to you the Duchenne muscular dystrophy community’s draft of the Guidance for Industry: Duchenne Muscular Dystrophy, Developing Drugs for Treatment Over the Spectrum of Disease, the first-ever patient advocacy-initiated draft guidance for a rare disease, written to help accelerate the development and review of potential therapies for Duchenne muscular dystrophy.

Our submission is prefaced by the Duchenne Imperatives, which begins with a few case studies, summarizes the document’s key points, and explains the Duchenne community’s key imperatives — what we hope will be the take home messages from the community for the sponsors, the Duchenne community and for the FDA, and to strive to frame the importance of the development of guidance for the community. We understand that the FDA may choose not to formally adopt this preface, though it is hoped that such information will inform FDA’s deliberations regarding adoption of the formal draft guidance, which follows.

Parent Project Muscular Dystrophy


DuchenneConnect.org
FDA Draft Guidance

Duchenne Muscular Dystrophy and Related Dystrophinopathies: Developing Drugs for Treatment
Guidance for Industry

DRAFT GUIDANCE

This guidance document is being distributed for comment purposes only.

Comments and suggestions regarding this draft document should be submitted within 60 days of publication in the Federal Register of the notice announcing the availability of the draft guidance. Submit electronic comments to http://www.regulations.gov. Submit written comments to the Division of Dockets Management (HFA-305), Food and Drug Administration, 5630 Fishers Lane, rm. 1061, Rockville, MD 20852. All comments should be identified with the docket number listed in the notice of availability that publishes in the Federal Register.

For questions regarding this draft document contact Colleen Locicero 301-796-1114.

U.S. Department of Health and Human Services
Food and Drug Administration
Center for Drug Evaluation and Research (CDER)

June 2015
Clinical/Medical
Impact and Collaboration

• Self-report data informs natural history and endpoint development
• Identification of research priorities that matter to patients and families
• Understand needs, priorities and preferences of patients and families
• Registry encourages and facilitates collaboration
  – Biopharmaceutical
  – Academic researcher
  – Clinician
  – Regulatory
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www.duchenneconnect.org