
DuchenneConnect 2013 Annual Report

Introduction:

[DuchenneConnect](#) is a robust and cutting-edge registry and resource that serves the needs of the Duchenne and Becker community. The goal of DuchenneConnect is to connect Duchenne and Becker patients with actively recruiting clinical trials and research studies, and to educate patients and families about Duchenne and Becker research. At the same time, DuchenneConnect is a valuable resource for researchers in academia and industry, allowing access to a database of information provided by patients and their families—information that is vital to advances in the care and treatment of Duchenne.

DuchenneConnect was created in 2007 by [Parent Project Muscular Dystrophy](#) (PPMD), with assistance from the NIH, the CDC, and Emory Genetics. In early 2011, PPMD alone began financing the registry's operation and maintenance, and is the sole guardian of DuchenneConnect and its material.

This 2013 Annual Report will illustrate how we have grown and expanded our services over the past year. **Highlights from 2013 include:** 1) securing \$970,000 in PCORI funding, 2) establishing an innovative genetic testing program called Decode Duchenne, and 3) recruiting for double the number of clinical trials and research studies as the previous year.

Accomplishments from 2013 include:

I. Registry growth and enhancement

- a. [Awarded \\$970,000 in PCORI Funding in December, 2013](#) - DuchenneConnect is honored to be one of 29 members of [PCORnet, the National Patient-Centered Outcomes Research Network](#). PCORI envisions PCORnet to be a secure, national data network that improves the speed, efficiency, and use of patient-centered comparative effectiveness research (CER). We are collaborating with PatientCrossroads, Geisinger Health Systems and UCLA for this project, which will bring many significant and positive changes to the registry throughout the next 18 months. Our main goals during the PCORI project include:
 - i. Improve research capacity and speed
 - ii. Empower registrants to set the agenda
 - iii. Increase diversity of registrants
 - iv. Improve the registrant experience
 - v. Decrease registrant burden

- b. [DuchenneCentral Mobile App for Smartphones](#) - Created in collaboration with Siren Interactive, the app enables the Duchenne community to learn about and locate clinical trials and clinics. Users can view status details on trials, get contact information for clinics, and stay up-to-date with the latest Duchenne news.

II. Industry collaborations

a. Sarepta Therapeutics:

- i. Developed the [Decode Duchenne Genetic Testing Program](#) with Kate Haviland at [Sarepta](#). Program provides genetic testing at no cost to patients who have been unable to access genetic testing due to financial barriers.
- ii. Partnered with [Emory Genetics Laboratory](#) to provide all genetic testing through Decode. Emory is providing testing for Decode patients at significantly reduced prices.
- iii. Created materials for Decode program, including Application form and Emory documents. Created logo and postcard for marketing of program.
- iv. Decode Duchenne went live in early December and will continue through 2014.

III. Recruitment Efforts

- a. **Clinical Trials:** Assisted with recruitment for 12 clinical trials (up from 7 in 2012). This accounted for approximately 90% of all clinical trials in the US that were recruiting Duchenne and Becker patients in 2013. For each trial, an announcement was posted on the website, a general email was sent to all registrants, and a targeted email was sent to registrants who appeared to match inclusion criteria.

| Name of Clinical Trial | Number Clicking Study Link | Number Needed for Study |
|--|----------------------------|-------------------------|
| Early Treatment for Cardiomyopathy (To determine if eplerenone, an approved medication for high blood pressure, can actually prevent heart muscle damage in patients with Duchenne.) | 78 | 40 |
| Genetic Modifier Study (To find rare variants in genes other than the dystrophin gene that may be playing a role in disease severity.) | 36 | 40 |

| Name of Clinical Trial | Number Clicking Study Link | Number Needed for Study |
|--|---|-------------------------|
| FOR-DMD (To determine which corticosteroid regimen increases muscle strength the most and causes the fewest side effects.) | (Could not be tracked due to multiple trial sites and coordinators) | 300 |
| Becker Epicatechin (To provide initial evidence of biological activity of (-)-epicatechin in Becker patients, by improving strength and skeletal muscle exercise response.) | 2 | 10 |
| DMD Natural History Study (CINRG) (To establish the largest long-term assessment of patients with Duchenne.) | 21 | 810 |
| Becker Limb Perfusion 2 nd recruitment effort (1 st in 2012) (To determine the safety and feasibility of a particular delivery method for gene therapy that could be used in the future to treat people with muscular dystrophies.) | (Study link not created for this trial) | 36 |
| Becker Follistatin Gene Transfer (To perform a gene therapy trial to deliver the follistatin gene to muscle cells in order to build muscle size and strength.) | 8 | 15 |
| Prosensa Natural History Study (To characterize the natural history and progression of Duchenne to help inform the design of future studies, to capture biomarkers of safety and disease progression and to provide comparative data.) | Targeted emails not sent (only newsletters and website postings) | 250 |
| Becker Sodium Nitrate (To learn whether patients with Becker will benefit from the supplement sodium nitrate by investigating blood flow during exercise.) | (Study link not created for this trial) | Unk |
| ACT DMD Confirmatory Trial (To evaluate the effect of ataluren on walking ability, as well as the effect of ataluren on physical function, quality of life, and activities of daily living.) | Targeted emails not sent until 2014 (only newsletters and website postings in 2013) | 220 |
| DP ARF Ultrasound 2 nd recruitment effort (1 st in 2012) (To assess the ability of a new ultrasound-based imaging method to monitor muscle degeneration and disease progression in Duchenne.) | 42 | 30 |

| Name of Clinical Trial | Number Clicking Study Link | Number Needed for Study |
|---|---|-------------------------|
| Eli Lilly Tadalafil Trial (To determine if tadalafil can slow the decline in walking ability of boys with Duchenne, and to assess the safety of the drug and any potential side effects.) | (Could not be tracked due to multiple trial sites and coordinators; however, Lilly 877# received 32 phone calls from DC.) | 306 |

- b. **Research Studies:** Assisted with recruitment for 10 research studies (up from 5 in 2012):
- i. **University of Minnesota Interview Study for Young Adults with Duchenne:** The purpose of this study was to learn more about how families communicate to their children about Duchenne.
 - ii. **Georgia Robotics and Computing Study:** WizKidz Science & Technology Centers partnered with Georgia Institute of Technology to conduct workshops to gauge participation of youth with disabilities in the world of robotics and computing.
 - iii. **Northwestern Interview Study of Parents:** Illinois parents were recruited for a study entitled “Parental Decision Making Regarding the Disclosure of Disease Morbidity, Mortality and Inheritance Patterns with Children with Duchenne”.
 - iv. **Risk/Benefit in Duchenne Therapies:** PPMD conducted this study to inform the FDA and other government agencies as well as biopharmaceutical companies about the treatment priorities and risk tolerance of the Duchenne community.
 - v. **University of South Carolina Transition to Adulthood Study:** A CDC funded research study of the transition from adolescence to young adulthood for people with rare conditions.
 - vi. **Expectations for Clinical Trials Study:** A PPMD study to understand the community’s expectations, hopes, experiences and motivations about clinical trials, with the goal of improving family wellbeing when participating in a trial.
 - vii. **Parental Communication to their Child about their Diagnosis of Duchenne:** A graduate student in genetic counseling at Northwestern University conducted this study on communication between parents and their child with Duchenne.
 - viii. **Finding the Silver Lining: How Families Cope with the Diagnosis of Duchenne:** University of Central Arkansas study to analyze the most common coping mechanisms of guardians of children with Duchenne.
 - ix. **Decision Making, Social Support and Privacy in Prenatal Diagnosis:** A genetic counseling graduate student at University of Alabama at Birmingham examined the decision making process for carriers considering prenatal diagnosis.
 - x. **Development of a Motor Function Questionnaire for Young Children:** The National Institute of Nursing Research of the NIH recruited parents or guardians of young children with neuromuscular disease to participate in this study.

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- c. **Feasibility Data:** Provided feasibility data for planning clinical trials and/or research studies to 3 institutions (1 academic and 2 industry). Feasibility data includes statistics from the registry regarding the number of patients matching specific criteria. This allows for optimal planning of clinical trials and research studies, including enrollment criteria and site location.

IV. Outcome Data from DuchenneConnect

- a. TREAT-NMD: Large data export in July, 2013 of all US registrants in DuchenneConnect, provided to TREAT-NMD global registry for manuscript to be published in 2014.
- b. TREAT-NMD Human Mutation Publication: [*The TREAT-NMD Duchenne muscular dystrophy registries: conception, design and utilisation by industry and academia*](#) describes in detail how the TREAT-NMD national patient registries for Duchenne were established and how the registries have grown since their inception in 2007. Article highlights the registries success in fostering collaboration between academia, patient organizations and industry, ultimately leading to increased clinical trial opportunities for patients and translating directly to improvements in patient care and treatment options.
- c. GlaxoSmithKline Study: “Multi-National, Cross-Sectional, Observational Study of Patient and Caregiver Burden of Duchenne Muscular Dystrophy” that TREAT-NMD and the University of Newcastle carried out in collaboration with OptumInsight. Manuscript completed in 2013 and to be published in 2014.
- d. Stan Nelson, MD, David Geffen School of Medicine, UCLA: Dr. Nelson and his research team used the DuchenneConnect data to investigate associations between corticosteroid use, medication and supplement use, and ambulation in Duchenne patients. Ongoing work on the publication of this data occurred in 2013.

V. Presentations/Publicity

- a. *Updates from DuchenneConnect*, presented by Holly Peay at UPPMD Conference in Rome, Italy (February, 2013).
- b. *Review of the Clinic Survey Data*, presented by Holly Peay at Care-NMD International Conference in Budapest, Hungary (April, 2013).
- c. *DuchenneConnect: The Power of the Registry*, presented by Ann Martin at North Carolina Medical Genetics Association (NCMGA) meeting in Charlotte, NC (April, 2013).

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- d. *Genetics and Proteomics: DuchenneConnect, Biomarkers and Modifiers*, presented by Stan Nelson, MD at PPMD Annual Connect Conference in Baltimore, MD (June, 2013)
 - e. *The Clinic Services Resource Survey in DuchenneConnect*, poster presentation by Ann Martin at World Muscle Society in Asilomar, CA (October, 2013).

VI. Educational Resources

- a. Contributed to the monthly [Direct Access Webinar Series](#), which provides the community with direct access to experts in the field of Duchenne.
- b. Wrote 30 [Clinical Trial/Research FAQ Sheets](#) for PPMD Annual Connect Conference Program and DuchenneConnect website. These are family-friendly summaries of active clinical trials and research in the pipeline.
- c. Sent 6 bimonthly DC Newsletters to all patient and professional registrants. The newsletters provide a summary of all current research and clinical trial news.
- d. Answered approximately 50 contact requests (“Ask an Expert”) through the registry website, and responded to numerous PPMD site comments/questions. Also responded to hundreds of phone calls and emails regarding the registry, clinical trials, genetic testing and genetic counseling issues.

VII. Registration

- a. New patient registrants for 2013: 423 (up from 405 in 2012)
 - i. Total patient registrants as of 12/31/13: 3,098 (2,567 completed profiles)
 - ii. Each new registrant account is curated by the DC Coordinator, which includes 1) reviewing Profile Survey (medical history) to confirm that answers are complete and consistent with age, sex and diagnosis of registrant, 2) emailing each new registrant to thank them for registering and to request any missing information, 3) analyzing all reports and records (genetic test report, echocardiogram, spirometry, muscle biopsy and clinic notes) and entering data into registry, and 4) assigning a Clinical Verification and Genetic Verification Status as outlined by [TREAT-NMD](#).
- b. New professional registrants (clinicians & researchers): 22
 - i. Total professional registrants as of 12/31/13: 367
- c. Continued the **DuchenneConnect Pilot Project** to increase registration of underserved patients and families, by providing tablets to neuromuscular clinics in the US. Several

interested clinics were identified, and 3 clinics signed the Letter of Agreement (University of Michigan, SUNY-Brooklyn, Nemours-Orlando). Site coordinators were trained at these 3 clinics, to assist patients and families with registering in DuchenneConnect and updating their accounts while in clinic.

DuchenneConnect Priorities for 2014:

1. Through the PCORnet project, we plan several enhancements to the registry. To start, we will be revising the Profile Survey, revising the informed consent, and translating the core elements of the registry into Spanish.
2. As part of PCORnet, we will be expanding our outreach to underserved patients and families and increasing our total number of registrants and the diversity of our registrants.
3. In addition, we plan to add validated PRO (patient reported outcome) measures, such as surveys involving quality of life and pain.
4. We plan to decrease registrant burden and increase the rate of updating, by providing incentives for registrants to update and by implementing new technologies, such as a mobile app.
5. We will continue marketing the registry and the Decode Duchenne program, providing free genetic testing to patients in need.

Questions?

Please contact the DuchenneConnect Team at coordinator@duchenneconnect.org or 201-937-1408.