Highlights from 2014

DuchenneConnect, a registry for those with Duchenne and Becker Muscular Dystrophy, is a program of Parent Project Muscular Dystrophy (PPMD). The overall goal of the Registry is to learn as much as possible about Duchenne and Becker muscular dystrophy, connect patients with actively recruiting clinical trials and research studies, and educate patients and families about Duchenne and Becker research. At the same time, DuchenneConnect is a valuable resource for researchers, allowing access to data provided by patients and their families—information that is vital to advances in the care and treatment of Duchenne.

We have grown and expanded our services over the past year. While we still focus on speeding the clinical trial processes, we have expanded our work to address other important clinical areas. **Highlights from 2014 include:**

- Being chosen to participate in PCORnet, a national learning network that prioritizes patient and family preferences in research
- Offering free genetic testing through our Decode Duchenne program
- Providing data or recruiting registrants for three journal articles that were published in 2014

Decode Duchenne Program Offers Free Testing

The Decode Duchenne Genetic Testing Program, with funding from Sarepta Therapeutics, provides genetic testing at no cost to patients who have been unable to access genetic testing. **Emory Genetics Laboratory** performs the testing and to date we have received over 100 applications for this program.

Numerous families have reported how grateful they are for this service, as they would not have been able to have genetic testing otherwise. In addition, all providers have been appreciative of the Decode program, since they can offer genetic testing to all of their Duchenne and Becker patients regardless of their financial situation.

“We are very thankful for you. You will never know how much your work means to us. It is a lifeline to hope.”

- Ruth
  Parent of a child in the DuchenneConnect Registry

---

**Contents**

**Special Interest Articles**
- Highlights from 2014 ........................................... 1
- Decode Duchenne ............................................. 1
- DuchenneConnect and PCORnet ..................... 2
- Recruitment for Clinical Trials ....................... 2
- Recruitment for Research Studies ................. 3
- Educational Resources.................................... 4
- Publications .................................................. 4
- DuchenneConnect Priorities.............................. 4

**Individual Highlights**
- Feasability Data ............................................. 3
- Reaching out to the Professional DBMD Community .............................................. 3
- Registrations ............................................... 4
DuchenneConnect and PCORnet

DuchenneConnect is honored to be one of 29 members of PCORnet, the National Patient-Centered Outcomes Research Network. PCORnet is a national data network that aims to improve the speed and efficiency of patient-centered clinical research. We are collaborating with PatientCrossroads, Geisinger Health Systems, and UCLA for this project, which brings many significant and positive changes to the registry. Some of the enhancements made possible by our PCORI funding include:

- Adding a project coordinator and genetic counsel, Ann Lucas
- Developing a Family Advisory Committee for the PCORnet project
- Creating a new informed consent
- Modifying the past Medical Profile Survey into a new and improved format
- Making the website mobile-friendly
- Working toward a longer-term goal of integrating electronic health record (EHR) data into the registry
- Striving to increase diversity in the Registry
- Providing incentives for participation in the Registry

Recruitment for Clinical Trials by DuchenneConnect

In 2014, DuchenneConnect was asked to recruit for seven clinical trials. This was the majority of clinical trials in the US that were recruiting Duchenne and Becker patients in 2014. Methods of recruitment include targeted emails to registrants who appear to match inclusion criteria, newsletter articles, website postings on homepage, FAQ Sheet (family-friendly summary for website and Annual Connect Conference program book), webinars, Facebook posts, and PPMD social media ads. The following trials were recruited for in 2014:

- **Tadalafil Trial (Eli Lilly)** – 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.
- **ACT DMD Confirmatory Trial (PTC Therapeutics)** – To evaluate the effect of ataluren/Translarna on walking ability, as well as the effect of ataluren on physical function, quality of life, and activities of daily living.
- **FOR-DMD** – To determine which corticosteroid regimen increases muscle strength the most and causes the fewest side effects.
- **HT-100 (Halo Therapeutics)** – To test the safety and tolerability of increasing doses of HT-100, a drug candidate intended to reduce fibrosis and inflammation and promote healthy muscle regeneration.
- **Myostatin Inhibitor Phase II Trial (Pfizer)** – To evaluate the safety, tolerability, and efficacy of PF-06252616, an antibody-based myostatin inhibitor, in ambulatory boys with Duchenne.
- **Confirmatory Trial of Eteplirsen in Duchenne (Sarepta)** – To provide confirmatory evidence of efficacy of eteplirsen in ambulatory boys with Duchenne who are amenable to skipping exon 51.
- **Safety Study of Eteplirsen to Treat Advanced Stage Duchenne (Sarepta)** – To explore safety and tolerability of eteplirsen in patients with advanced stage Duchenne who are amenable to exon 51 skipping and non-ambulatory.

“*I couldn’t keep connected to progress and clinical trials, and know the fight continues to help our boys, without DuchenneConnect. We need everything we can get, and we need it FAST!*”

- Judy
  Parent of a child registered in DuchenneConnect

Feasibility Data

DuchenneConnect provided feasibility data for planning clinical trials and/or research studies to **8 institutions** (3 academic, 3 industry, and 2 consulting firms) in 2014. 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.
Reaching out to the DBMD Professional Community

The DuchenneConnect Team presented about DuchenneConnect and DuchenneConnect programs in professional forums more than 10 times in 2014 to ensure that clinicians, researchers, sponsors, and the broader community are aware of the capabilities of the Registry. Activities included presentations, webinars, scientific posters, and booths at professional meetings.

DuchenneConnect has a Professional Portal, and in 2014 we had 48 new professional registrations. The total professional registrations as of 12/31/14 were 408.

Recruitment for Research Studies by DuchenneConnect

DuchenneConnect assisted with recruitment for 12 research studies in 2014:

• **Expectations for Clinical Trials Study** – PPMD’s study to understand the community’s needs, experiences, and motivations about clinical trials, with the goal of improving family wellbeing when participating in a trial.

• **New York Stem Cell Foundation (NYSCF) Study** – study to make stem cells for current and future research projects from skin cells.

• **Study of Sleep Patterns in Duchenne Teens** – study to determine if there is a relationship between sleep patterns and health related quality of life (HRQoL).

• **Adult Disease Burden Study** – study on how genetic neuromuscular disease affects patient’s lives, particularly around issues such as pain, depression, fatigue, sleep, and cognitive problems.

• **Barriers in the African-American Population** – study to understand the barriers to healthcare, clinical trials, and registries among African American patients with Duchenne.

• **Sudden Cardiac Death Registry at Nationwide Children’s Hospital (NCH)** – study to learn more about cardiac natural history, to determine the rate of sudden cardiac death and the benefit of implantable devices.

• **Robotic Arm Study at New Jersey Institute of Technology** – prototype robot arm designed to ‘float’ a person’s arm and let remaining muscle forces move their arm.

• **Aquatic Therapy Study at New Jersey Institute of Technology** – study on range of motion data on non-ambulatory boys with Duchenne.

• **DuchenneConnect Needs Assessment Survey** – study to determine the needs of our registrants and to learn what we can do better for our community.

• **Carrier Survey regarding Genetic Counseling and Health Risk** – study to determine if women at risk of being carriers for Duchenne or Becker were receiving appropriate genetic counseling.

• **Mothers Wellbeing Study** – PPMD’s study to determine how the experiences and wellbeing of mothers caring for a child with Duchenne change over time, with the goal of obtaining information to support the development of interventions for mothers.

• **Duchenne and Epilepsy Study** – study to determine whether there are an increased number of people with Duchenne who have epilepsy.
Registrations in DuchenneConnect

In 2014, we had 515 new patient registrations. The total patient registrations at the end of 2014 was 3,150 (2,812 completed profiles).

To help increase the registration of underserved patients and families, DuchenneConnect provides tablets to neuromuscular clinics in the US. Five clinics are now participating (University of Michigan, SUNY-Brooklyn, Nemours-Orlando, Children’s Hospital Colorado, and Kennedy Krieger Institute). We plan to provide a tablet to every Certified Duchenne Care Center (CDCC). Site coordinators are trained at each clinic, to assist patients and families with registering in DuchenneConnect and updating their accounts while in clinic.

Questions?
Please contact the DuchenneConnect Team, Ann Martin and Ann Lucas (Certified Genetic Counselors), at coordinator@duchenneconnect.org or 201-937-1408.

Publications in the Scientific Literature with Data from DuchenneConnect

PPMD Benefit/Risk Study Results, Clinical Therapeutics (May 2014) – Study concludes that caregivers are willing to accept serious risk when balanced with noncurative slowing or stopping of the progression of muscle weakness, even absent improvement in lifespan. The study has implications for the Food and Drug Administration’s (FDA’s) benefit-risk assessment of emerging Duchenne therapies.

Duchenne Burden of Illness Study, Neurology (July 2014) – This study revealed the different costs accompanying living with Duchenne and the overall financial burden of affected families. The research was carried out in collaboration with TREAT-NMD Duchenne registries in Germany, Italy, the UK, and the US (DuchenneConnect). A total of 770 patients/caregivers completed the survey, with the US having the largest number of participants (284).

Ambulation Study Using DuchenneConnect Data, PLoS Currents Muscular Dystrophy (October 2014) – “Online Self-Report Data for Duchenne Muscular Dystrophy Confirms Natural History and Can Be Used to Assess for Therapeutic Benefits” – Dr. Stanley Nelson and his team found that the data in DuchenneConnect is very similar to the data from natural history studies. This is important because using registry data is much easier and less expensive than a traditional clinical trial. Dr. Nelson’s team also looked at corticosteroids as well as commonly used supplements to determine which ones, either alone or in combination, may help to prolong ambulation.

Educational Resources
Educational information is available on the DuchenneConnect website and through our mailing list. As part of our educational outreach we:

- Send a bimonthly newsletter to all patient and professional registrants
- Create Clinical Trial/Research FAQ Sheets for PPMD’s Annual Connect Conference and DuchenneConnect website
- Contribute to the monthly Direct Access Webinar Series
- Respond to phone calls, emails, and “Ask an Expert” contact requests.

“I joined DuchenneConnect because I wanted to make sure my son has the best opportunities for studies, information, and care available.”

- Donna
Parent of a child registered in DuchenneConnect

DuchenneConnect Priorities for 2015

- Support the development of robust clinical trials and translate that information to the community.
- Better understand the Duchenne and Becker communities’ priorities for research and care/management.
- Help with research that leads to improvements in care.
- Make the Registry easier to use and find ways to make the Registry more useful to the Duchenne and Becker community.
- Increase our outreach to underserved patients and families and increase the diversity in our Registry. This will include translating the core elements of the registry into Spanish, which has already begun.
- Provide all the Certified Duchenne Care Centers (CDCCs) with a tablet that can be used in clinic for patients to register in DuchenneConnect or update their account. Joining forces with the CDCCs will bring added exposure to the Registry.
- Continue to provide incentives for registrants, to encourage registration, and updating of accounts.
- Continue our efforts to provide genetic testing, genetic education, and support for decision making about research participation.