DuchenneConnect 2012 Year-End Report

Introduction:

In 2007, a group of thought leaders in the Duchenne muscular dystrophy community began discussing the need for a new kind of resource that would connect and serve the needs of the entire community. Today, the result of this endeavor is DuchenneConnect, a robust and cutting-edge registry and website.

The purpose 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 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. A 2011 Year-End report was published one year ago, and this 2012 Year-End Report will highlight how we have grown and expanded our services over the past year.

Accomplishments from 2012 include:

I. Industry collaborations
   a. GlaxoSmithKline Study: “Multi-National, Cross-Sectional, Observational Study of Patient and Caregiver Burden of Duchenne Muscular Dystrophy (DMD)” that TREAT-NMD and the University of Newcastle carried out in collaboration with OptumInsight, a contract research company appointed by GSK. This was the first time in Duchenne history that industry has collaborated with multiple registries from different countries.
   At DuchenneConnect, we:
      i. Obtained IRB approval through Western Institutional Review Board.
      ii. Worked with OptumInsight to revise the survey and study materials.
      iii. Organized an extensive recruitment effort using both email and traditional mail.
      iv. Recruited 286 US registrants to complete the survey, surpassing the goal of 250.
         We were the first country to reach goal.

II. Recruitment Efforts
   a. Assisted with recruitment for 7 clinical trials (same number that we recruited for in 2011). This accounted for 85% of all clinical trials in the US that were recruiting Duchenne and Becker patients in 2012.
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.

Trials recruited for are listed in the table below, with the number of patients who clicked the link (to directly email the study coordinator at each site) listed after each trial:

<table>
<thead>
<tr>
<th>Name of Clinical Trial</th>
<th>Number Clicking Study Link</th>
<th>Number Needed for Study</th>
</tr>
</thead>
<tbody>
<tr>
<td>GSK2402968/DMD114876</td>
<td>4</td>
<td>54</td>
</tr>
<tr>
<td>(To determine if GSK2402968 is effective in the treatment of ambulant boys with DMD resulting from a mutation correctable by exon 51 skipping.)</td>
<td></td>
<td></td>
</tr>
<tr>
<td>DART EIM</td>
<td>33</td>
<td>90</td>
</tr>
<tr>
<td>(To determine if Electrical Impedance Myography (EIM) has the potential to serve as a non-invasive, quantifiable, diagnostic tool for neuromuscular disease.)</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Becker Limb Perfusion</td>
<td>3</td>
<td>36 (only 2 more needed when we were contacted)</td>
</tr>
<tr>
<td>(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.)</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Early Treatment for Cardiomyopathy</td>
<td>69</td>
<td>40</td>
</tr>
<tr>
<td>(To determine if eplerenone, an approved medication for high blood pressure, can actually prevent heart muscle damage in patients with Duchenne.)</td>
<td></td>
<td></td>
</tr>
<tr>
<td>DP ARF Ultrasound</td>
<td>31</td>
<td>30</td>
</tr>
<tr>
<td>(To assess the ability of a new ultrasound-based imaging method to monitor muscle degeneration and disease progression in Duchenne.)</td>
<td></td>
<td></td>
</tr>
<tr>
<td>PDE Inhibitors</td>
<td>17</td>
<td>12</td>
</tr>
<tr>
<td>(An acute dosing study to inform the design of a larger, randomized, multicenter trial of PDE inhibitors for clinical skeletal muscle and cardiac endpoints.)</td>
<td></td>
<td></td>
</tr>
<tr>
<td>DELOS/idebenone</td>
<td>(N/A – could not be tracked due to multiple trial sites &amp; coordinators)</td>
<td>80</td>
</tr>
<tr>
<td>(To determine if idebenone, as compared to placebo, shows an improvement or delays the decline in respiratory function (PEF) in patients with Duchenne.)</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>
b. Assisted with recruitment for 5 research studies:
   1. **GSK Patient and Caregiver Burden in Duchenne**: See details in section I.a. above. Results from study to be published in 2013/2014.
   2. **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 identifying targets for interventions to improve family wellbeing when participating in a trial.
   3. **Adoption Study**: A study conducted by Cincinnati Children’s Hospital Medical Center and the University of Cincinnati to better learn the needs of parents who have adopted a child with Duchenne/Becker.
   4. **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.
   5. **Mother’s Survey**: A PPMD study of mothers of children with Duchenne and Becker, to assess their needs, strengths, and wellbeing, with the goal of developing new interventions to improve mothers’ wellbeing.

c. Provided feasibility data for planning clinical trials and/or research studies to 3 institutions (2 academic and 1 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.

III. **Presentations/Publicity**
   a. **Clinical and Molecular Basis of Duchenne and Becker Muscular Dystrophy**, presented by Holly Peay at UPPMD Conference in Rome, Italy (February, 2012).
   c. **Review of the Clinic Survey Data**, presented by Holly Peay at Transforming Duchenne Care Conference in Ft. Lauderdale, FL (June, 2012).
   d. **DuchenneConnect: The Power of the Registry**, presented by Holly Peay at PPMD Annual Connect Conference in Ft. Lauderdale, FL (June, 2012).
   e. **Duchenne Carrier Moms Breakout Session**, presented by Ann Martin and Dr. Linda Cripe at PPMD Annual Connect Conference in Ft. Lauderdale, FL (June, 2012).
   f. **Updates from DuchenneConnect**, presented by Holly Peay at TREAT-NMD Curators’ Meeting in Istanbul, Turkey (September, 2012).
IV. Outcome Data from DuchenneConnect
   a. Stanley Nelson, MD, Professor of Human Genetics, 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.
      i. Dr. Nelson presented the data at the UPPMD Italy meeting in February 2012, the PPMD Annual Connect Conference in June 2012, and presented a PPMD/DuchenneConnect Direct Access Webinar on April 25, 2012.
         1. 147 people listened live to the webinar.
         2. At the end of 2012, more than 1,000 people have listened to the recorded webinar on YouTube.
      ii. Dr. Nelson’s publication of outcome data is expected in 2013.

V. 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 26 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.
   d. Answered 35 contact requests (“Ask an Expert”) through the registry website, and responded to numerous PPMD site comments/questions. Also responded to approximately 50 phone calls and hundreds of emails regarding the registry, clinical trials, and genetic testing and counseling issues.

VI. Registration
   a. New patient registrants for 2012: 405
      i. Total patient registrants: 2,675 (2,187 completed profiles)
      ii. Each new registrant account is curated by the DC Coordinator, which includes 1) reviewing the Profile Survey (medical history) to confirm that the answers are complete and consistent with the age, sex and diagnosis of the 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 the data into the registry, and 4) assigning a Clinical Verification and Genetic Verification Status as outlined by the TREAT-NMD guidelines.
b. New professional registrants (clinicians & researchers): 24
   i. Total professional registrants: 345

c. Designed the DuchenneConnect Pilot Project to increase registration of underserved patients and families, by providing tablets to 5 neuromuscular clinics in the US. The 5 sites were identified and the tablets were purchased. Training of a site coordinator at each clinic will begin in early 2013. Once trained, each site coordinator will assist patients and families with registering in DuchenneConnect and updating their accounts while in clinic.

VII. Website enhancement

a. New Profile Survey went live in June, 2012, with the goal of obtaining a more robust dataset containing the key elements needed by both industry and academic researchers. The New Profile Survey went through multiple rounds of revisions with the DuchenneConnect advisory board.

b. New “Donate to DuchenneConnect” button on homepage (directed donation to PPMD).

c. New, interactive patient dashboard to go live in 2013.

DuchenneConnect Priorities for 2013:

1. Train all 5 site coordinators in the DuchenneConnect Pilot Project

2. Launch New Patient Dashboard

3. Add a validated Quality of Life Survey to DuchenneConnect

4. Increase awareness of and participation in the Clinic Survey

5. Submit for publication at least two scientific articles, in collaboration with academic researchers

6. Data cleaning to improve the accuracy of the dataset

Questions? Please contact the DuchenneConnect Coordinator, Ann Martin, MS, CGC: coordinator@duchenneconnect.org or 201-937-1408.