Your Data at Work: The Duchenne Registry

Ann Martin, MS, CGC
June 28, 2018
New Name / New Website!

THE DUCHENNE REGISTRY

The strength of the registry is you

Your data is critical in the fight to end Duchenne
OUR IMPACT

4336 Total Registrants

113 Countries Represented

60 Trials Recruited

17 Industry Partners

Join
The Duchenne Registry

• Online self-report registry for individuals with Duchenne or Becker, as well as carrier females
• Established by PPMD in 2007, in collaboration with the NIH, CDC and Emory Genetics
Goal of Duchenne Registry

• To educate and connect the entire Duchenne and Becker community:
  – Individuals with Duchenne and Becker
  – Families and caregivers
  – Healthcare providers
  – Researchers
  – Industry
What We Do

**Education**
Website, newsletters, email blasts, social media

**Recruitment**
Recruited for 8 clinical trials & 11 research studies in 2017

**The Duchenne Registry**

**Genetic Testing**
Decode Duchenne genetic testing program

**Advance Research**
Exported data to 9 institutions (5 industry) in 2017
Screenshots from the New Registry Dashboard
Welcome back, Bruce!

Your newest survey, the Corticosteroid Module is now available. Take it today and view the Survey Data to see how others have responded.

Health Surveys (3 of 12 Complete)

<table>
<thead>
<tr>
<th>INCOMPLETE SURVEYS</th>
<th>START SURVEY</th>
</tr>
</thead>
<tbody>
<tr>
<td>Bone/Orthopedic Module</td>
<td>START SURVEY</td>
</tr>
<tr>
<td>A moderate length survey covering spinal X-rays, scoliosis, bone fractures, ...</td>
<td></td>
</tr>
<tr>
<td>Behavior &amp; Learning Module</td>
<td>START SURVEY</td>
</tr>
<tr>
<td>A moderate length survey covering any diagnosed behavior or learning problems ...</td>
<td></td>
</tr>
<tr>
<td>Clinical Trials &amp; Registry Participation Module</td>
<td>START SURVEY</td>
</tr>
<tr>
<td>A moderate length survey about your participation in any research studies, ...</td>
<td></td>
</tr>
<tr>
<td>Insurance Module</td>
<td>START SURVEY</td>
</tr>
</tbody>
</table>

Medical Records

Upload relevant medical records to share more detailed data while protecting your privacy.

ADD MEDICAL RECORDS

Medications
Highlights from 10 Year Registry Report
Diagnosis

- Manifesting carrier (I am a carrier of Duchenne or Becker and I have symptoms)
- Possible carrier (I do not know for sure if I am a carrier)
- Confirmed carrier (I am a carrier of Duchenne or Becker and I do not have any symptoms)
- Duchenne or Becker (not clear yet)
- Becker
- Duchenne

- 76%
- 7%
### Genetic Categories of Registrants with Verified Results

<table>
<thead>
<tr>
<th>Category</th>
<th>Count</th>
<th>Percent (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Deletion</td>
<td>868</td>
<td>64.6</td>
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<tr>
<td>Nonsense</td>
<td>168</td>
<td>12.5</td>
</tr>
<tr>
<td>Duplication</td>
<td>129</td>
<td>9.6</td>
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<tr>
<td>Deletion - Small mutation</td>
<td>73</td>
<td>5.4</td>
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<tr>
<td>Splice site</td>
<td>53</td>
<td>3.9</td>
</tr>
<tr>
<td>Duplication - Small mutation</td>
<td>29</td>
<td>2.2</td>
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<tr>
<td>Insertion</td>
<td>11</td>
<td>0.8</td>
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<tr>
<td>Missense</td>
<td>7</td>
<td>0.5</td>
</tr>
<tr>
<td>Other</td>
<td>4</td>
<td>0.3</td>
</tr>
<tr>
<td>In/del</td>
<td>2</td>
<td>0.1</td>
</tr>
</tbody>
</table>

Please submit a copy of your child’s genetic test report!
Age at First Corticosteroid Use: Duchenne

5 years old – Duchenne
11 years old – Becker
Steroid Use and Age at Loss of Ambulation

Duchenne boys in TDR

- Never on steroids: Age 10
- Prednisone: Age 11
- Deflazacort: Age 12
The Duchenne Registry: Working Together to Advance Research

You

We review and curate data

We notify you about research opportunities

We can provide support specific to you

Our Team

Researchers

Data shared with qualified researchers and global registry

New research ideas generated

New trials begin

Industry

Data Provided for trial planning

Your Registry Data
Your de-identified data is shared with:

- TREAT-NMD Global Duchenne Registry
- Duchenne Regulatory Science Consortium
Recent Publications

• *Human Mutation*, June 2018, Wang RT, et al:
  “*DMD* genotype correlations from DuchenneConnect: Endogenous exon skipping is a factor in prolonged ambulation for individuals with a defined mutation sub-type”

• *Journal of Neuromuscular Diseases*, Nov 2017, Koeks Z, et al:
  “Clinical Outcomes in Duchenne Muscular Dystrophy: A Study of 5345 Patients from the TREAT-NMD DMD Global Database”
Your Data Makes a Difference
Register Today!
www.duchenneregistry.org
You Could Win an iPad!

#PPMD
Free genetic testing and counseling for Duchenne and Becker muscular dystrophy
DECODE DUCHENNE

Criteria for Free Testing:

☑ Male with a confirmed or suspected diagnosis of Duchenne or Becker,
   OR
   Female with a relative with Duchenne or Becker with a known mutation
☑ Never had genetic testing OR previous genetic testing has not confirmed a mutation
☑ Any financial barrier
☑ Legal resident of US or Canada

All testing performed at:

PerkinElmer
Genetics
Over 1,000 participants to date in the Decode Duchenne program!
Thank you!
Please visit us at the Registry table!
coordinator@duchenneregistry.org
888-520-8675