



TEN YEAR REGISTRY REPORT

Parent **JOIN THE FIGHT.
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Project
Muscular
Dystrophy

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The Duchenne Registry

The Duchenne Registry (previously DuchenneConnect) is the largest, most comprehensive patient-reported registry for Duchenne and Becker muscular dystrophy. The Duchenne Registry began in 2007 as a joint collaboration between Parent Project Muscular Dystrophy (PPMD), the CDC, NIH, PatientCrossroads, and Emory University, with the unified goal of connecting and serving the needs of the Duchenne muscular dystrophy community. PPMD fully supports the Registry and is the sole guardian of the Registry and its material.

The Registry is comprised of data from individuals with Duchenne or Becker muscular dystrophy, and from parents on behalf of their children, as well as from female carriers. As of November 2017, medical and family history information has been collected from nearly 4000 Registry participants. This report represents over ten years of data collected in The Duchenne Registry.

“Since a small group of parents and grandparents formed PPMD almost 25 years ago, we have believed in the importance of innovation and patient data in the fight to end Duchenne,” said Pat Furlong, Founding President & CEO of PPMD. “The Duchenne Registry is now powered with 10 years of robust data thanks to the efforts of our incredible community.”

How the Data Has Been Used

The de-identified information in The Duchenne Registry has been used by researchers and sponsors to advance care and treatments for individuals with Duchenne. Over the past ten years, **data has been exported and shared with researchers nearly 50 times**, including multiple exports to the TREAT-NMD International Duchenne Registry. To date, **eight publications** have used data from The Duchenne Registry, and **two additional publications are currently in press**. Numerous posters and presentations at scientific meetings have referenced the Registry data. Visit our website to learn more about our publications (www.duchenneregistry.org/reports/publications).

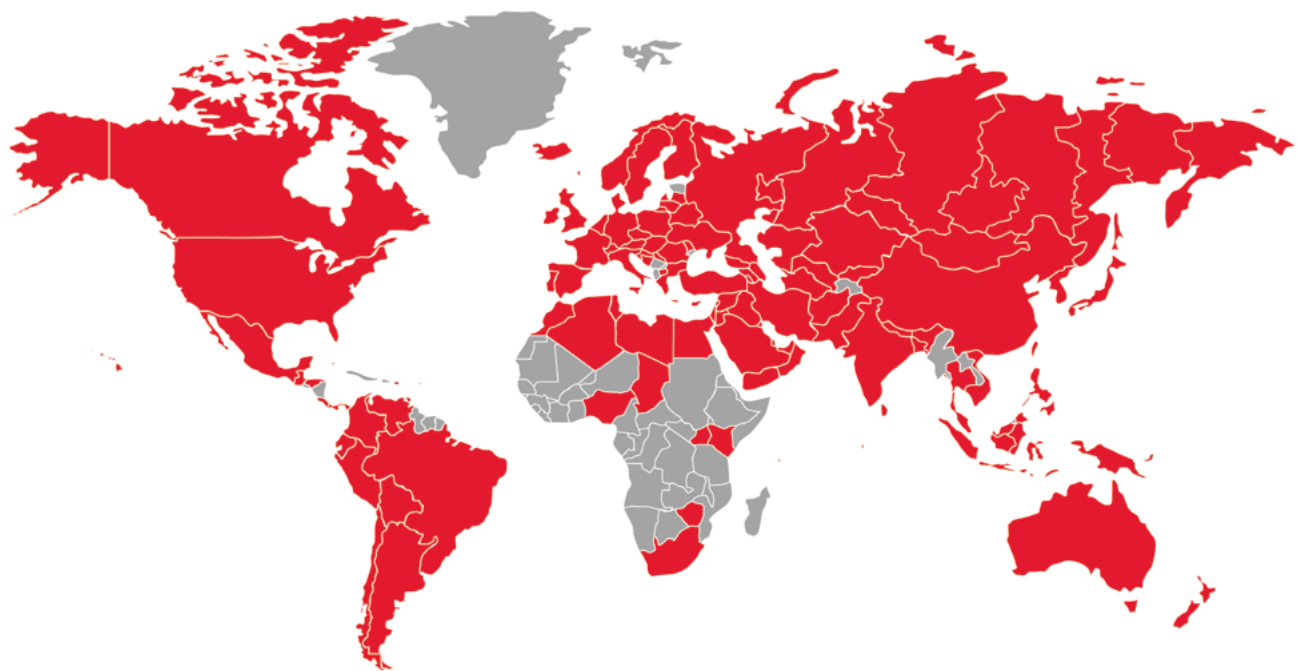
In the past ten years, the Registry team has used the data to identify and connect individuals with Duchenne and Becker to **over 60 actively recruiting clinical trials** and even more non-interventional research studies. In addition, data from the Registry has been utilized by PPMD to identify community needs and priorities.

Our Impact as of November 2018



About the Duchenne and Becker Muscular Dystrophy Registrants

The Registry is comprised of a diverse population from around the world, with over 100 countries represented.



| Top 5 Countries in The Duchenne Registry | |
|--|-----|
| United States | 70% |
| India | 5% |
| Canada | 4% |
| United Kingdom | 2% |
| Australia | 2% |

Most of the registrants (90%) are males with Duchenne or Becker muscular dystrophy. As of November 2017, the Registry included a total of 3997 registrants. Registrants are comprised of a variety of race and ethnicities which includes: 2805 Caucasians (82%), 481 Asians (12%), 73 Native Americans (2%), 69 African Americans (2%), 29 other Pacific Islanders (.7%), 27 Black not African Americans (.7%), 7 Native Hawaiian (.2%), and 10% identified as other. 12% of the registrants are Latino/Hispanic, whereas 82% are not Latino/Hispanic, 4% are unsure, and 2% preferred not to respond to this question.

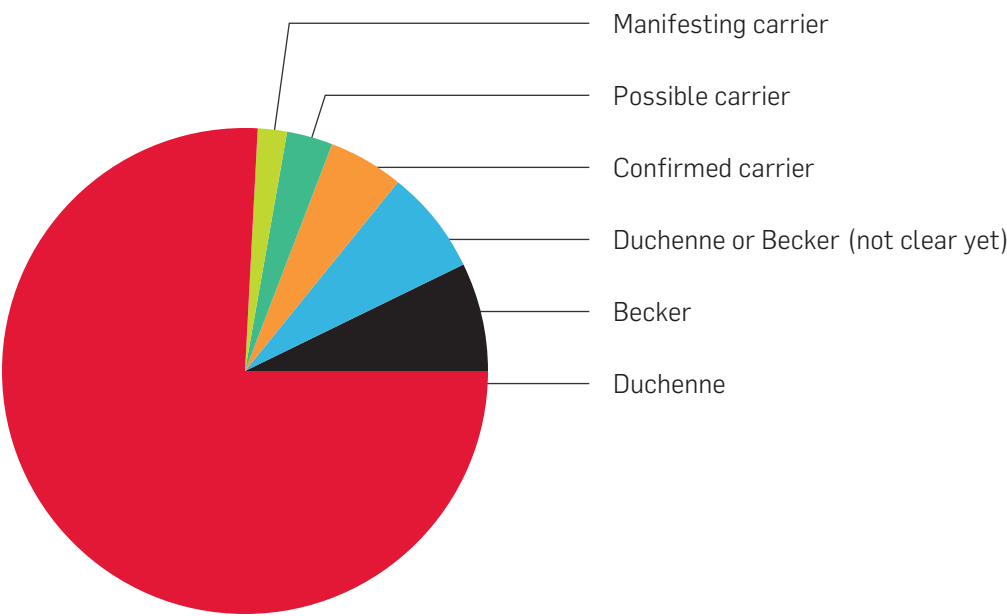


“We are very thankful for the Registry. You will never know how much your work means to us. It is a lifeline to hope.”



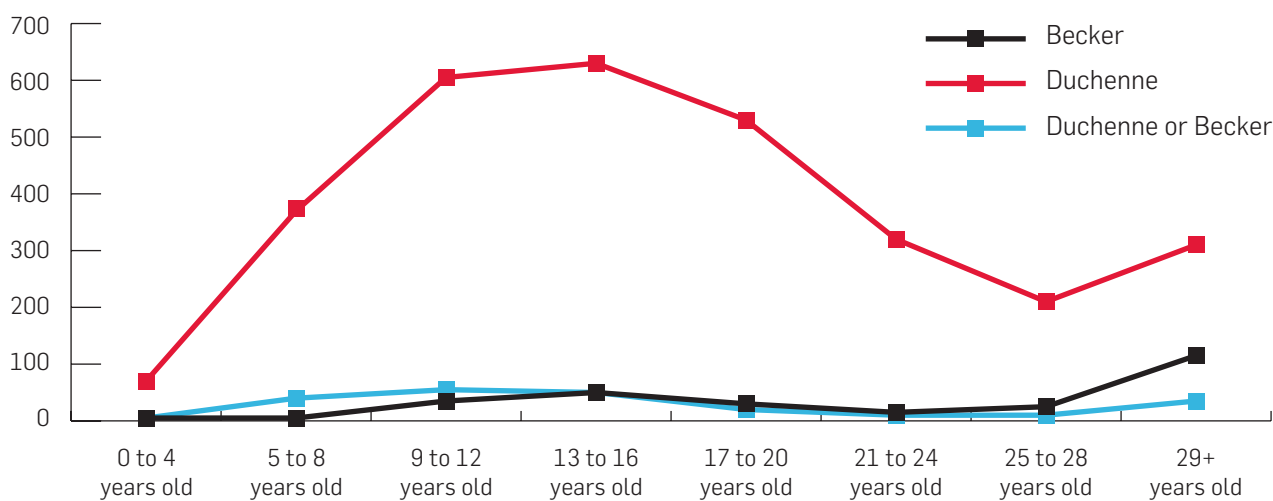
**For United States registrants,
the average age for a Duchenne
diagnosis was 4 years old, and
for Becker muscular dystrophy
was 10 years old.**

Diagnosis



Registry participants were asked about their diagnosis. Most registrants (3054 people, or 76% of registrants) self-reported that they are individuals with Duchenne muscular dystrophy, while 306 (7% of registrants) reported they are individuals with Becker muscular dystrophy. Carriers were also invited to register, with 155 women (5% of registrants) identifying as confirmed carriers without symptoms, 87 women (2%) identifying as manifesting carriers with symptoms, and 103 women (3%) identifying themselves as possible carriers.

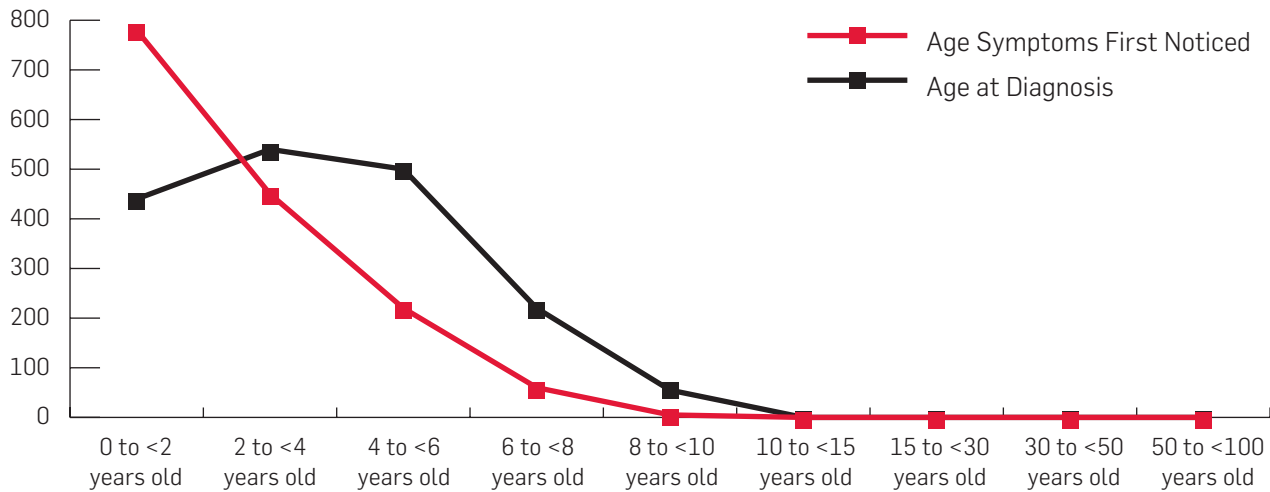
Age By Diagnosis



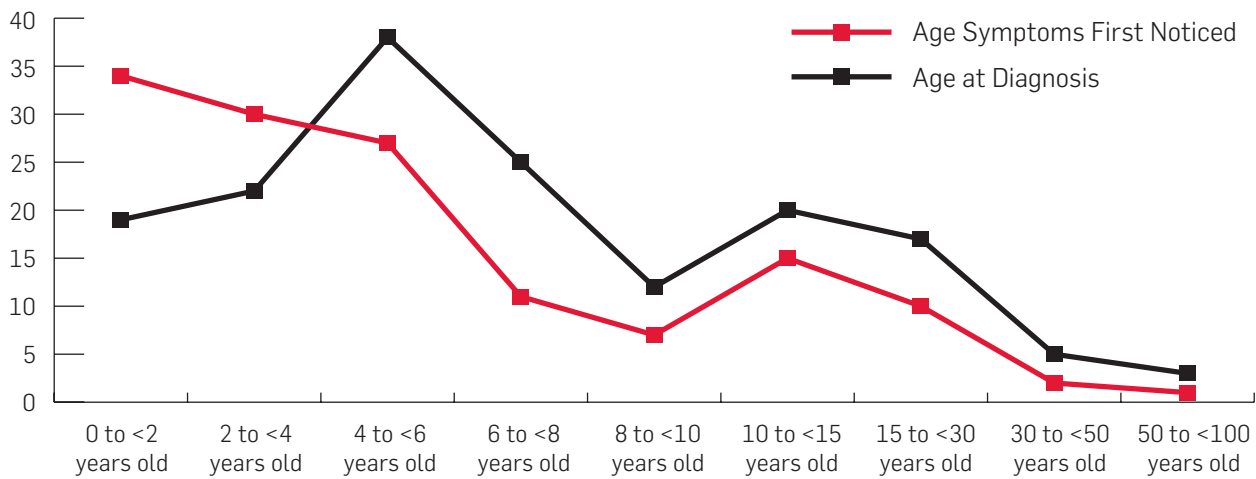
The above graph shows the current age ranges for registrants with Duchenne, Becker, and those who are unclear of their diagnosis. We asked registrants about the age in which they/their child first received their diagnosis. For United States registrants, the average age for a Duchenne diagnosis was 4 years old, and for Becker muscular dystrophy was 10 years old. This includes registrants both with and without a family history of muscular dystrophy.

We also asked registrants the age in which symptoms were first noticed. These graphs show the average age that registrants in the United States first noticed symptoms and the average age at diagnosis for Duchenne and Becker.

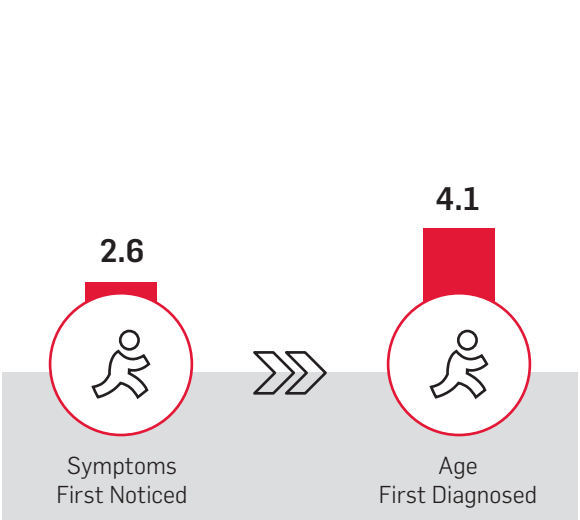
Age Symptoms First Noticed and Age at Diagnosis in U.S. — Duchenne



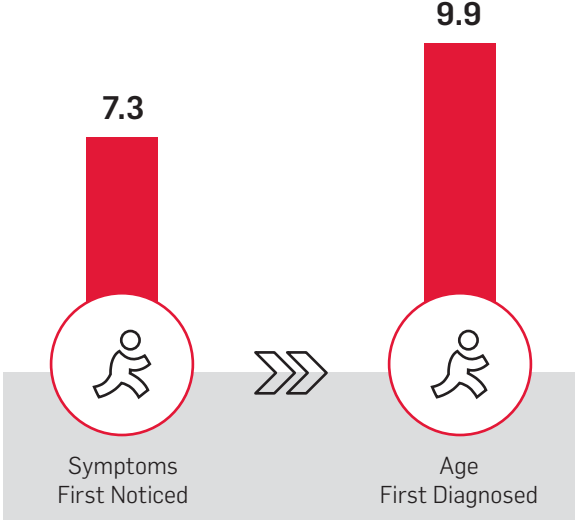
Age Symptoms First Noticed and Age at Diagnosis in U.S. — Becker



Average Age First Diagnosed in U.S. — Duchenne



Average Age First Diagnosed in U.S. — Becker



Genetic Information

Participants in The Duchenne Registry are requested to send in a copy of their genetic test report. The genetic test report is required in order to have a verified account according to TREAT-NMD guidelines. The Registry team reviews each of these reports and carefully enters the genetic mutation for each registrant. This provides vital information that verifies the diagnosis that is self-reported by the registrant and helps answer important research questions.

The table on the next page describes the categories of mutations found and their frequency after reviewing all the genetic test reports submitted to the Registry (n=1344, or 40% of Duchenne and Becker registrants). The Registry team found that deletions are the most common mutation type (65%), followed by nonsense mutations (13%) and duplications (10%).



Genetic Categories of Registrants with Verified Results

| | Count | Percent |
|------------------------------|-------|---------|
| Deletion | 868 | 64.6% |
| Nonsense | 168 | 12.5% |
| Duplication | 129 | 9.6% |
| Deletion — Small mutation | 73 | 5.4% |
| Splice site | 53 | 3.9% |
| Duplication — Small mutation | 29 | 2.2% |
| Insertion | 11 | 0.8% |
| Missense | 7 | 0.5% |
| Other | 4 | 0.3% |
| In / del | 2 | 0.1% |

Definitions

Deletion: One or more exons are missing from gene, and depending on the exons involved, the deletion can be “in-frame” or “out-of-frame”. Exons are small coding segments within the gene and the Duchenne gene has 79 exons.

Nonsense: A small mutation, or change in the DNA, that creates a premature stop codon so the resulting protein is much shorter than normal and usually nonfunctional.

Duplication: One or more exons are repeated in gene.

Deletion — Small mutation: A small deletion, or missing piece of DNA, that does not include an entire exon.

Splice site: A small mutation, or change in the DNA, within an intron, but near the exon borders, so splicing (removing) of the introns does not occur properly. Introns are small non-coding segments within the gene.

Duplication — Small mutation: A small duplication, or extra copies of a piece of DNA, that does not involve an entire exon.

Insertion: A small piece of DNA (smaller than an exon) is inserted into gene.

Missense: A small mutation that results in a change in the message of the DNA so the resulting protein may not function correctly.

Other: Any other rare mutation not otherwise described.

In / del: A rare mutation that involves a combination of an insertion and a deletion.

Since 2013, PPMD's genetic counselors have directed a FREE genetic testing program called **Decode Duchenne**. To date the Decode Duchenne program has received over 1000 applications, and over 900 individuals have completed genetic testing. We believe that financial barriers should not limit access to genetic testing in the Duchenne community. The application and testing process is fast and easy, and includes interpretation of results and counseling. The Decode Duchenne program is generously sponsored by Sarepta Therapeutics and PTC Therapeutics. Learn more at ParentProjectMD.org/Decode.



Review of the genetic reports provided key information regarding potential treatment options. The Registry team identified 325 people with Duchenne who have verified results demonstrating they may potentially benefit from treatments that cause **exon skipping** of exons 51, 45, and 53. In the Registry, 10% of registrants are amenable to skipping exon 51; 8% of registrants are amenable to skipping exon 45; and 6% of registrants are amenable to skipping exon 53.



A team of researchers at UCLA, led by Richard Wang, PhD in Dr. Stan Nelson's Laboratory and Florian Barthelemy, PhD in Dr. Carrie Miceli's Laboratory, analyzed Registry data for a study that was published in the journal *Human Mutation* in 2018 [*Human Mutation* 2018 Sep;39(9):1193-1202]. They found a correlation between certain deletions and the age at loss of ambulation. In particular, they found that boys with exon 45 deletions (exon 44 skippable) and boys with exon 3-7 deletions (exon 8 skippable) had a later age at loss of ambulation (they walked longer) than other boys in the Registry. Cultured muscle cells from Duchenne boys with deletions of exons 3-7 or exon 45 showed higher naturally occurring skipping than other mutations, providing a potential biological rationale for the prolonged ambulation.

These results highlight the power of aggregating large data sets in rare diseases such as Duchenne. The Registry allows researchers to have access to thousands of patients with Duchenne without ever having to see a patient in person, and at the fraction of the cost of a traditional natural history study.

Muscle Function

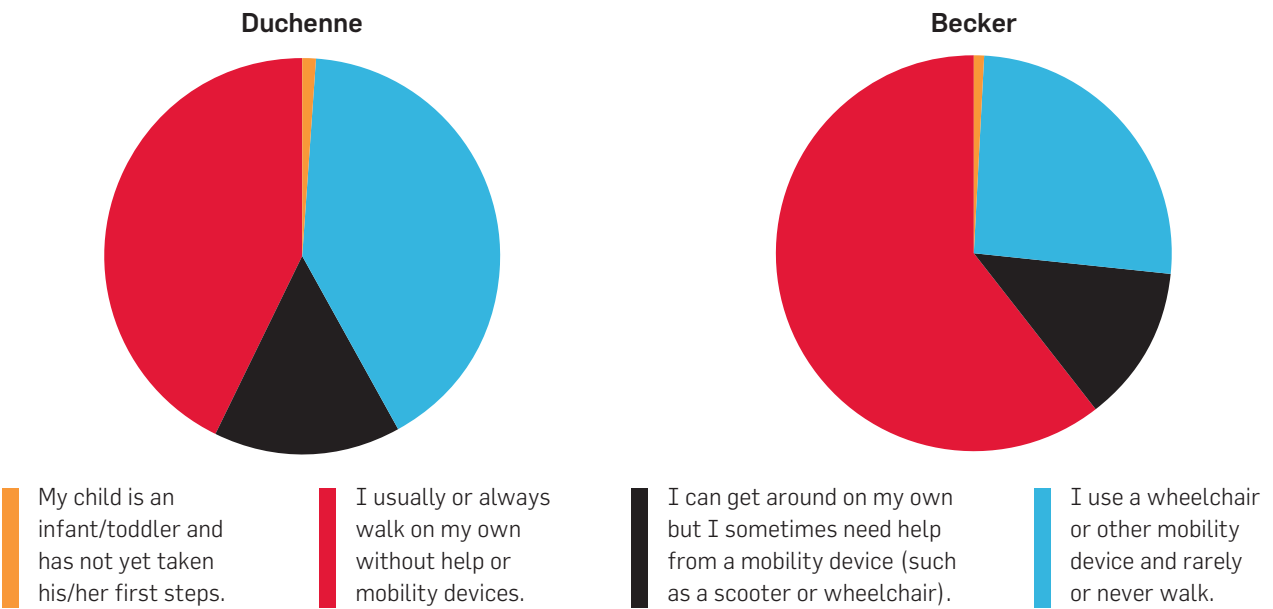
The Duchenne Registry has several Medical History surveys (modules) that together form the core of the data we collect. We ask registrants to complete all relevant surveys at the time of registration, and we request annual updates.

The better the data we collect, the better the Registry. The strength of the Registry really does depend on you, the registrants!

In the first survey, we asked registrants about their/their child's mobility. 44% of respondents reported that they usually or always walk on their own without help or mobility devices. 40% reported that they use a wheelchair or other mobility device and rarely or never walk. 15% of respondents reported that they can get around on their own but sometimes need mobility device assistance.

Mobility by Diagnosis

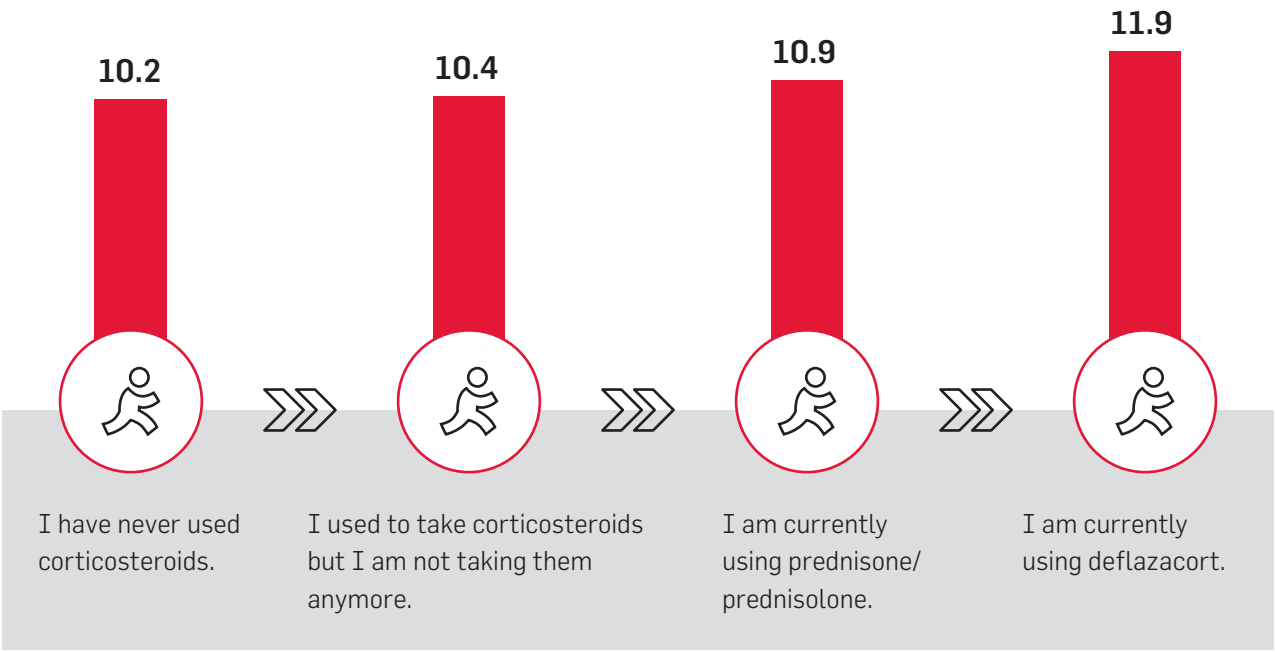
| | Duchenne | Becker | All |
|--|----------|--------|------|
| I usually or always walk on my own without help or mobility devices. | 1250 | 174 | 1424 |
| I can get around on my own but I sometimes need help from a mobility device (such as a scooter or wheelchair). | 444 | 37 | 481 |
| I use a wheelchair or other mobility device and rarely or never walk. | 1198 | 74 | 1272 |
| My child is an infant/toddler and has not yet taken his/her first steps. | 39 | 3 | 42 |
| All | 2931 | 288 | 3219 |



We used The Duchenne Registry data to look at the relationship between taking corticosteroids and time to loss of the ability to walk. We see the expected steroid benefit in the Registry participants: those who used steroids, or used to take steroids, are older when they need to start using a full-time mobility device like a wheelchair. On average, individuals with Duchenne who never used steroids lost their mobility at the age of 10. Those using prednisone/prednisolone had mobility until 11 years old, on average. Those using deflazacort had mobility until 12 years old, on average. The ages in the graphic below are based on averages of 10 years of Registry data. Average calculations include everyone, even those registrants who are outliers, meaning they either stop walking much earlier than expected or they walk much longer than expected.

It is important to note that other studies, including studies using the Registry data, have shown different ages for loss of ambulation. More complex statistical analyses (e.g. Kaplan-Meier) take into consideration multiple confounding factors and are more difficult to generate than an average. The more complex analyses have shown different ages for loss of ambulation, ranging from 11–13 for those individuals on prednisone and 12–14 for those individuals on deflazacort. For example, Richard Wang, PhD, Stan Nelson, MD and their team at UCLA published a study using the Duchenne Registry data in *PLoS Currents* in 2014 and found that the age at loss of ambulation for boys on prednisone was 13 and for boys on deflazacort was 14 [*PLoS Currents Muscular Dystrophy* 2014 October 17;6].

Average Age of Loss of Ambulation by Steroid Use for Duchenne Registrants



For people with Duchenne and Becker in the Registry, activities requiring upper body and arm function started to decline in the early teen years. Such activities include using a fork or spoon or writing with a pencil.



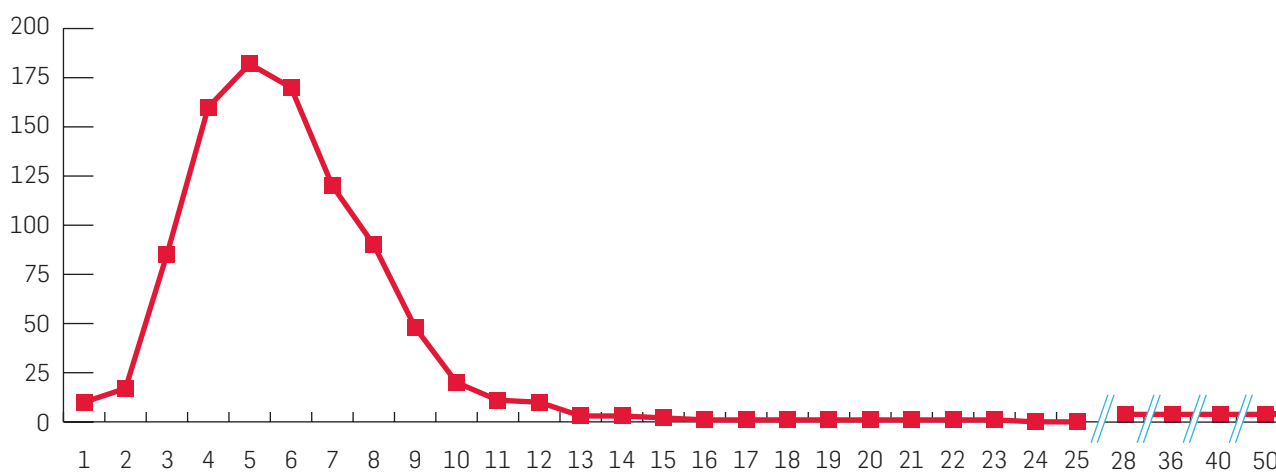
Corticosteroids

Corticosteroid Use for Registrants Diagnosed with Duchenne or Becker

| | Duchenne | Becker | All |
|---|-------------|------------|-------------|
| I am currently using deflazacort. | 808 | 13 | 821 |
| I am currently using prednisone/ prednisolone. | 643 | 23 | 666 |
| I used to take corticosteroids but I am not taking them anymore. | 279 | 22 | 301 |
| I have never used corticosteroids. | 806 | 184 | 990 |
| I don't know. | 61 | 9 | 70 |
| All | 2597 | 251 | 2848 |

We asked registrants about their/their child's current or previous use of corticosteroids. 29% of registrants reported that they currently were using deflazacort. 23% reported that they/their child was currently using prednisone or prednisolone. 11% of registrants reported that they used to take corticosteroids but they/their child were no longer taking them. 35% of registrants reported that they have never used corticosteroids.

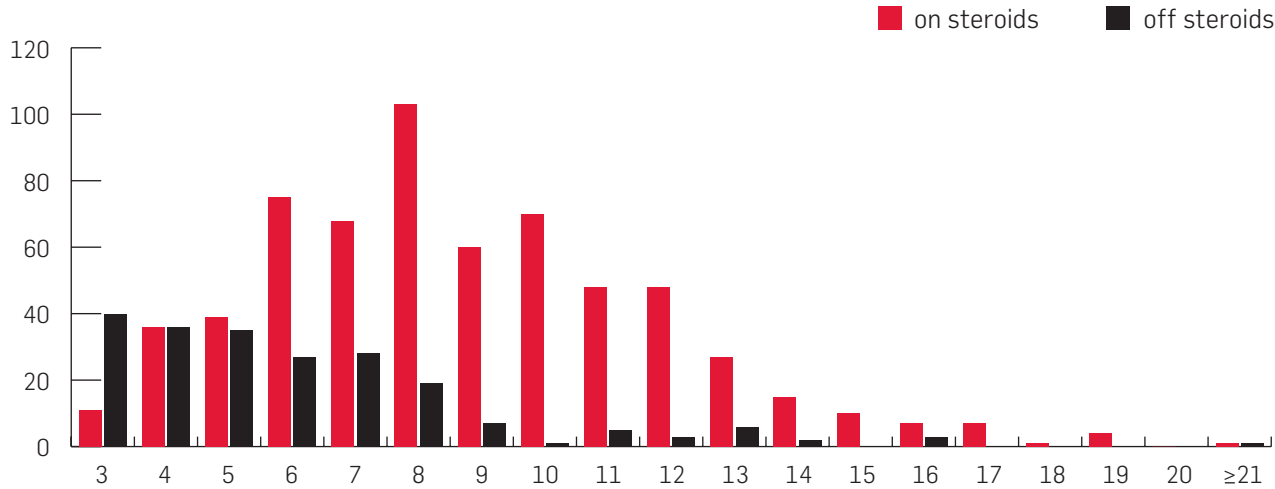
Age at First Corticosteroid Use — Duchenne



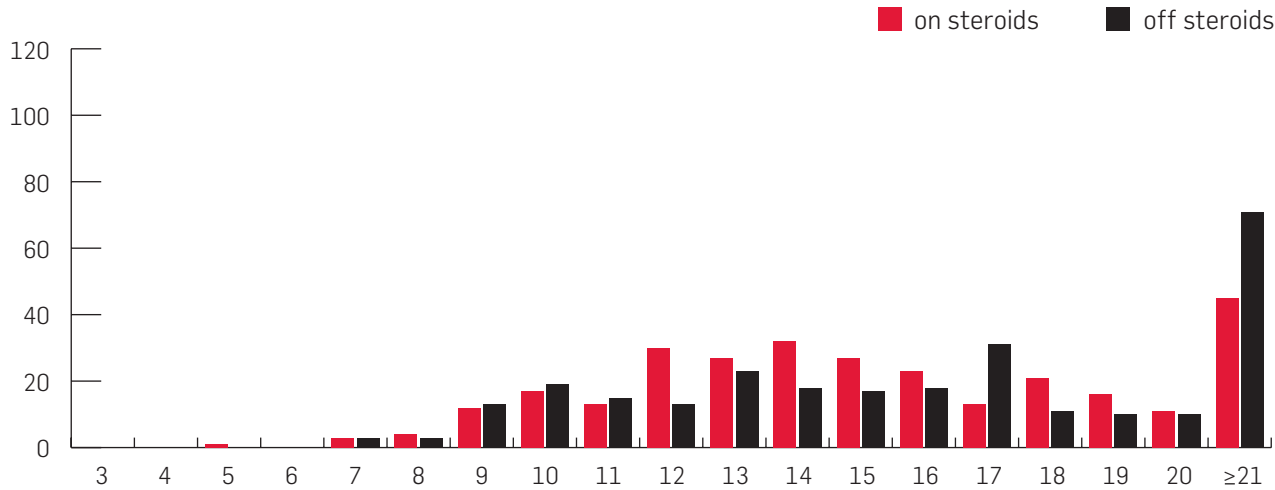
The average age of Duchenne registrants starting their first dose of corticosteroids was 5 years old. The average age of Becker registrants starting their first dose of corticosteroids was 11 years old.

Catabasis Pharmaceuticals performed an in-depth analysis of the corticosteroid data in The Duchenne Registry in 2017 (manuscript in progress). Corticosteroid use by age for ambulatory and nonambulatory Duchenne registrants in the United States is shown below. The majority of ambulatory Duchenne registrants under the age of 10 (63%) and over the age of 10 (92%) were using corticosteroid therapy. In contrast among nonambulatory registrants less than and greater than 10 years of age, 37% and 49%, respectively were using corticosteroid therapy.

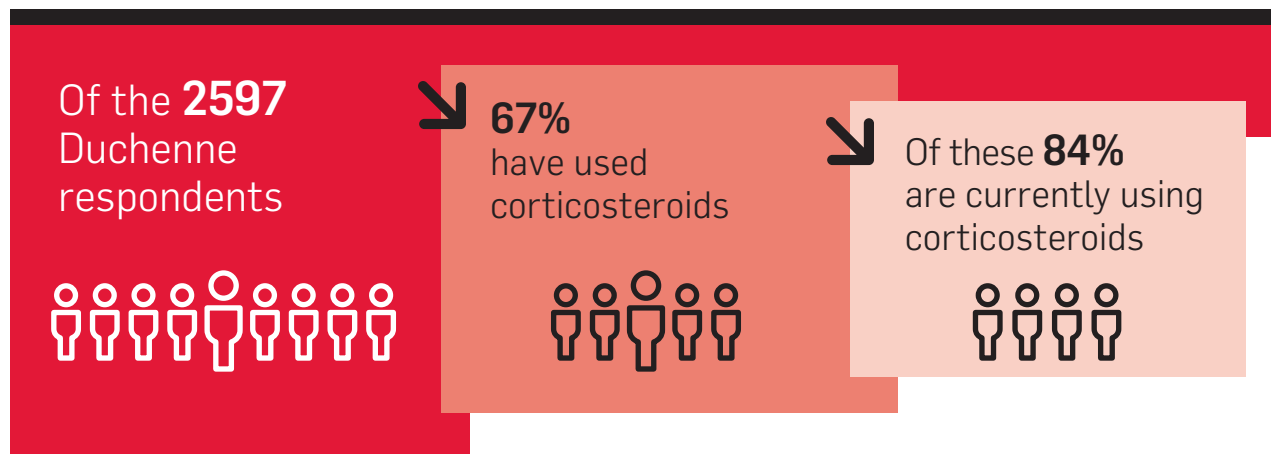
Ambulatory Duchenne Registrants in the United States



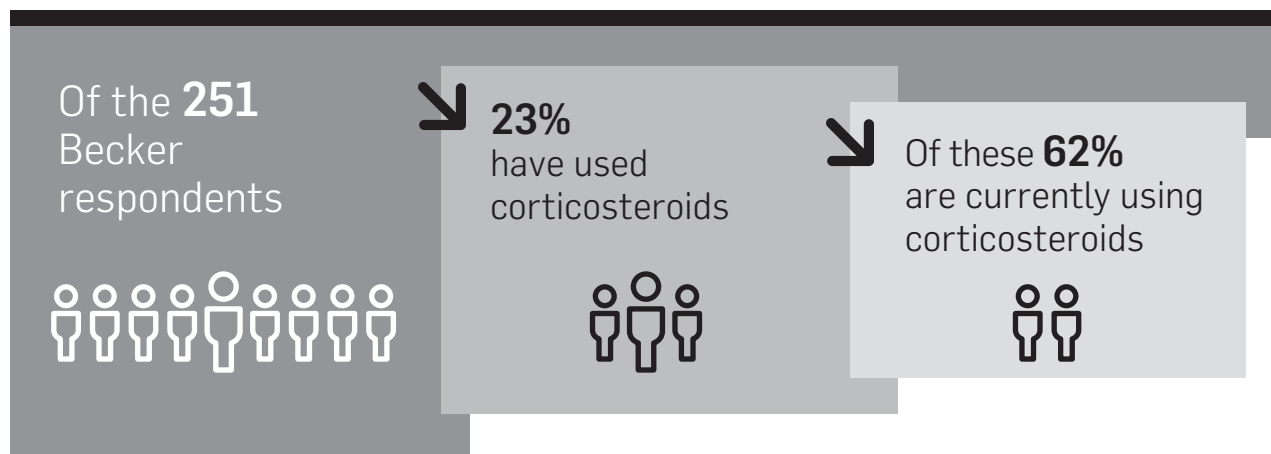
Nonambulatory Duchenne Registrants in the United States



Corticosteroids (Duchenne)

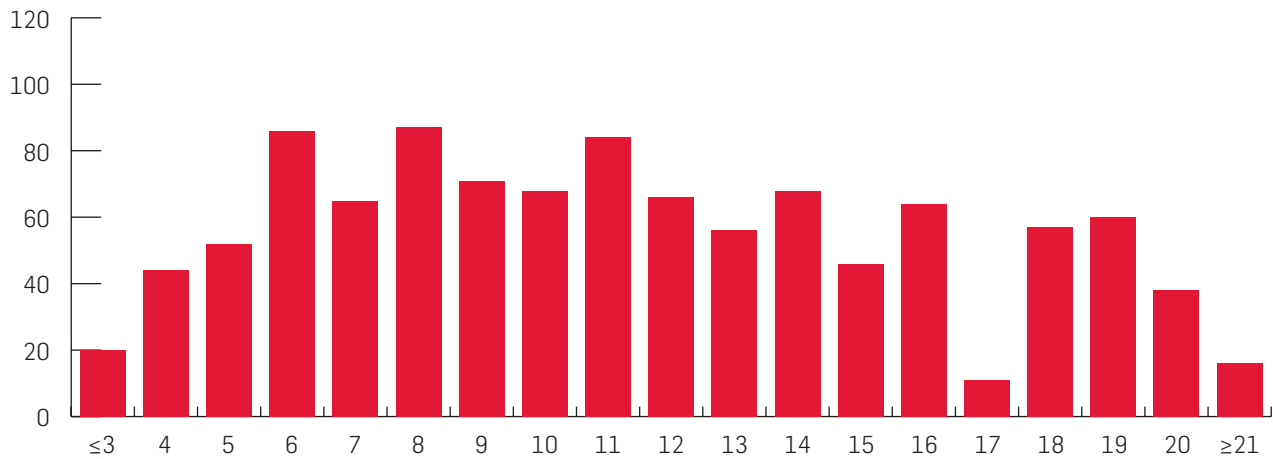


Corticosteroids (Becker)

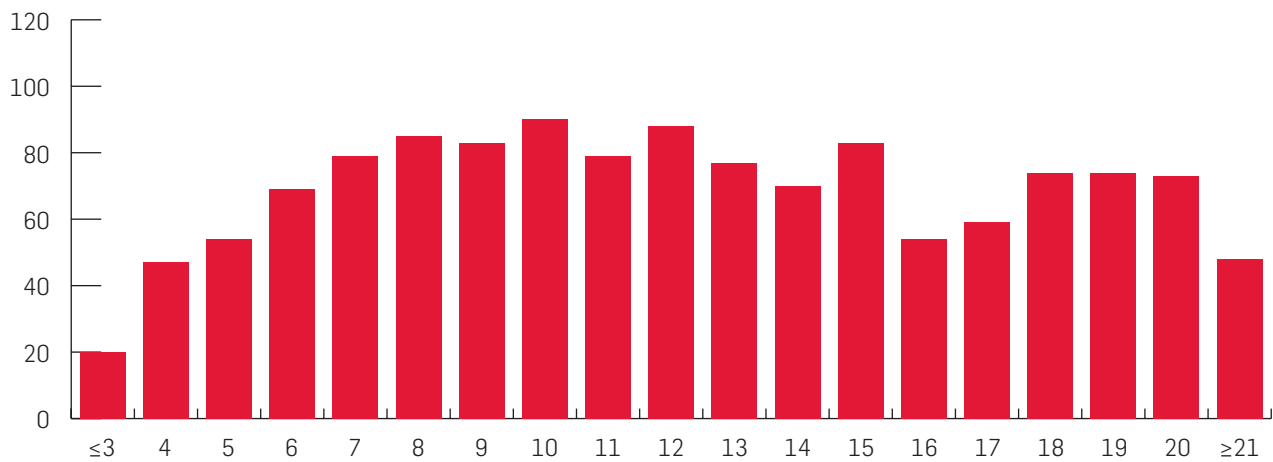


Catabasis also analyzed steroid use in five-year increments (2007–2011 and 2012–2016) by registrant age (below). The use of corticosteroids in registrants age 4–9 remained largely unchanged: 70% and 72% of registrants age 4–9 used corticosteroids from 2007–2011 and 2012–2016, respectively. Corticosteroid use in registrants age 10–25 was variable from 2007–2011 (between 0% and 84% by age), and tended to be more consistent from 2012–2016 (38% to 90% use by age). More consistent use of corticosteroids in the older registrants from 2012–2016 may be a reflection of better adherence to the standard of care guidelines.

Corticosteroid Use from 2007–2011



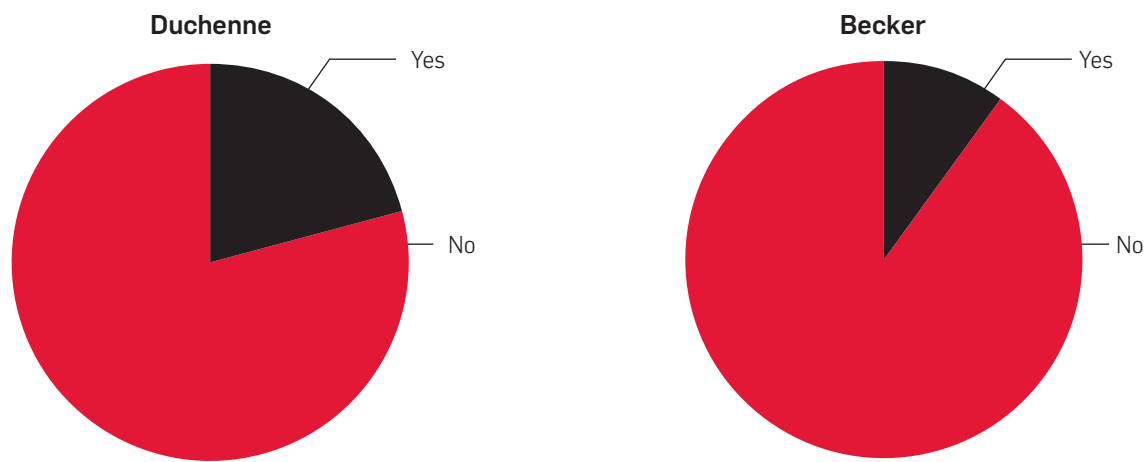
Corticosteroid Use from 2012–2016



Respiratory

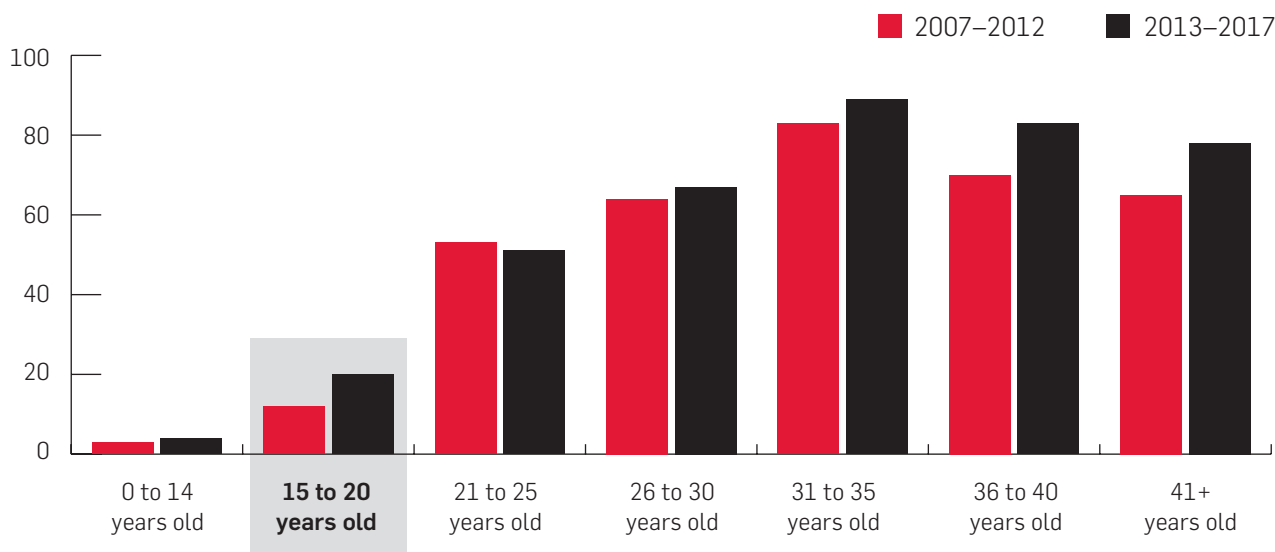
We asked registrants about their use of breathing devices, such as cough assist, BiPAP, CPAP, and ventilator. 21% of people with Duchenne in the Registry use a breathing device, compared to 10% of people with Becker.

Do you or your child use a breathing device?



For Duchenne registrants across all countries, the majority of ages showed an increase in the use of breathing devices in the years 2013–2017 compared to 2007–2012. The increase was **statistically significant** for the 15–20 year old age bracket (p-value = 0.019).

Percentage of Duchenne Respondents Using a Breathing Device By Age



Statistical significance refers to the claim that a result from data generated by testing is not likely to occur randomly or by chance, but is instead likely to be attributable to a specific cause. A p-value < 0.05 is usually considered statistically significant and a p-value < 0.001 is considered highly statistically significant.

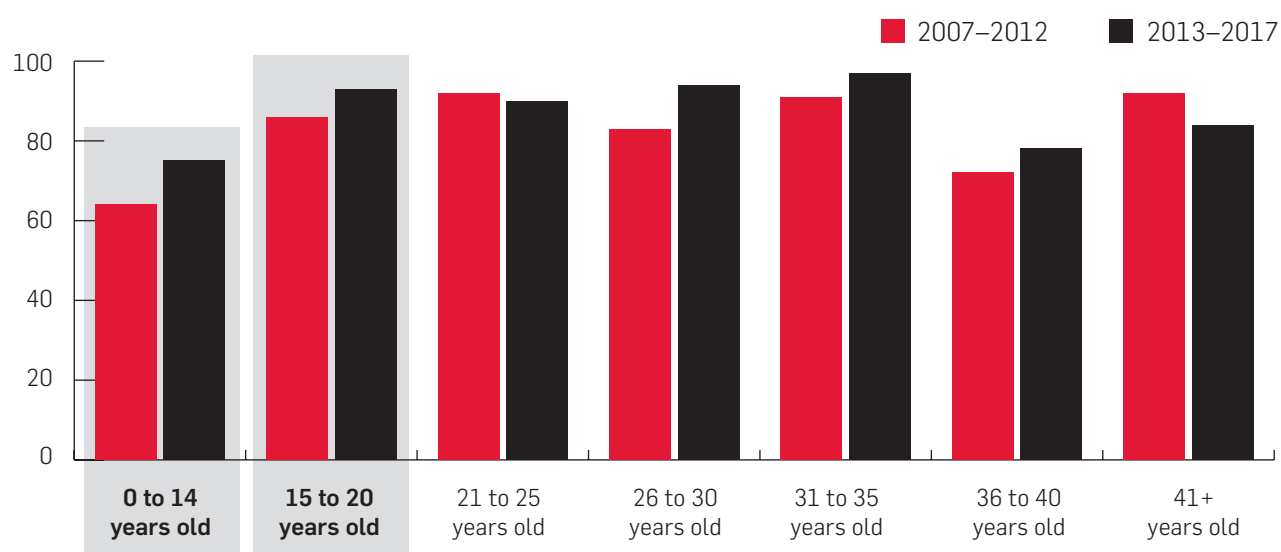
Cardiac

Have you had an echocardiogram, heart ultrasound, or cardiac MRI?

| Diagnosis | Yes | No |
|------------------------------------|------|-----|
| Duchenne | 1957 | 370 |
| Becker | 195 | 40 |
| Duchenne or Becker (not clear yet) | 105 | 58 |
| Manifesting carrier | 46 | 12 |
| Confirmed carrier | 77 | 51 |
| All | 2380 | 531 |

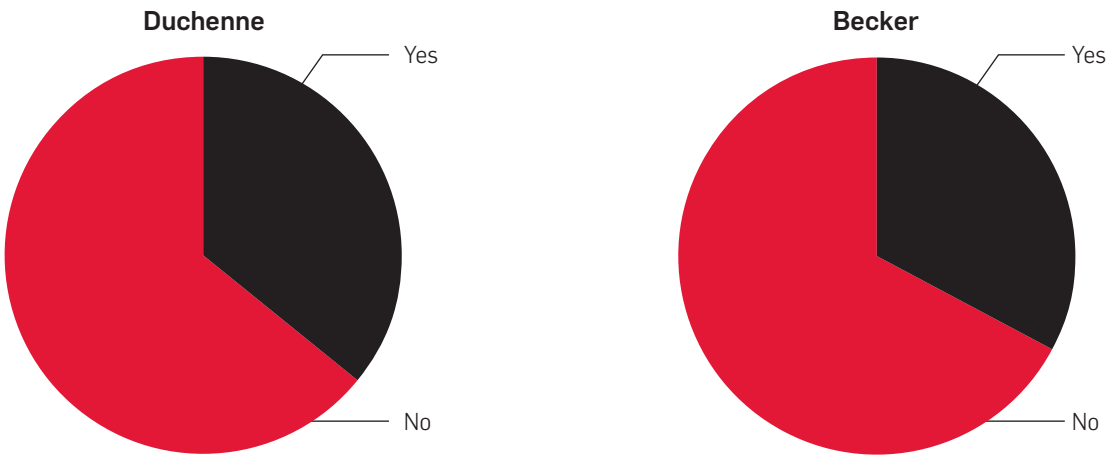
Most people with Duchenne (81%) and Becker (80%) have had an echocardiogram, heart ultrasound, or cardiac MRI. Many confirmed carriers (60%) also reported having one of these heart tests. For Duchenne registrants across all countries, the majority of ages showed an increase in echocardiogram in the years 2013–2017 compared to 2007–2012. The increase was **statistically significant** for the 0–14 (p-value: 0.002) and 15–20 (p-value: 0.002) year old age brackets.

Percentage of Duchenne Respondents Who Received an Echocardiogram By Age



36% of respondents with Duchenne and 33% of respondents with Becker have taken heart medication.

Have you ever taken any heart medications?



Heart Medications

| | Duchenne | | | Becker | | |
|---|-----------------|------------|-----------------|-----------------|------------|-----------------|
| | Currently Using | Never Used | Previously Used | Currently Using | Never Used | Previously Used |
| Beta blockers^a | | | | | | |
| Count | 254 | 104 | 9 | 27 | 5 | 3 |
| Percent (%) | 68.6 | 28.1 | 2.4 | 75.0 | 13.9 | 8.3 |
| ACE inhibitors^b | | | | | | |
| Count | 545 | 35 | 37 | 35 | 4 | 4 |
| Percent (%) | 87.9 | 5.6 | 6.0 | 81.4 | 9.3 | 9.3 |
| Angiotensin^c | | | | | | |
| Count | 112 | 149 | 10 | 11 | 10 | 1 |
| Percent (%) | 40.3 | 53.6 | 3.6 | 45.8 | 41.7 | 4.2 |
| Diuretics^d | | | | | | |
| Count | 113 | 130 | 11 | 10 | 10 | 2 |
| Percent (%) | 43.8 | 50.4 | 4.3 | 45.5 | 45.5 | 9.1 |
| Heart failure medications^e | | | | | | |
| Count | 50 | 159 | 13 | 6 | 11 | 2 |
| Percent (%) | 21.9 | 69.7 | 5.7 | 30.0 | 55.0 | 10.0 |
| Mineralcorticoid receptor antagonist^f | | | | | | |
| Count | 23 | 113 | 2 | — | 7 | — |
| Percent (%) | 16.2 | 79.6 | 1.4 | — | 87.5 | — |

^aexamples include: Tenormin (atenolol), Coreg (carvedilol), Toprol and Lopressor (metoprolol), Inderal (propranolol).

^bexamples include: Aceon (perindopril), Capoten (captopril), Vasotec (enalapril), Prinivil and Zestril (lisinopril), Altace (ramipril).

^cexamples include: Cozaar (losartan).

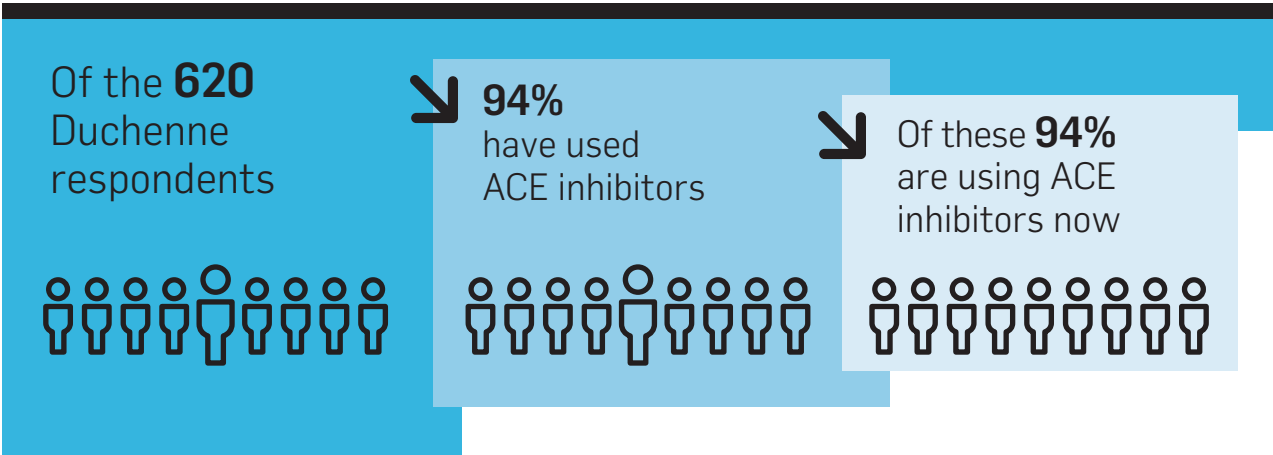
^dexamples include: Diuril (chlorothiazide), Lasix (furosemide), Esidrix (hydrochlorothiazide), Aldactone (spironolactone).

^eexamples include: Lanoxin (digoxin) and Primacor (milrinone).

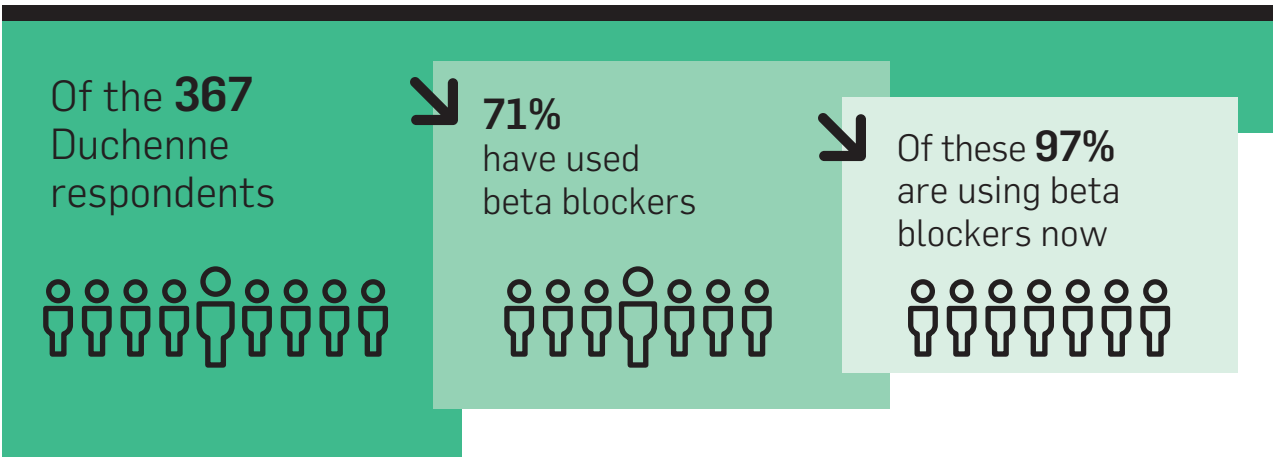
^fexamples include: Inspra (eplerenone).

We asked registrants about their use of specific heart medicines. Most people with Duchenne who completed the Heart Module have taken ACE inhibitors (94%), and 94% of those are currently using ACE inhibitors. Similarly, most people with Duchenne who completed the Heart Module have taken beta blockers (71%), and of those 97% are currently using beta blockers.

ACE inhibitors



Beta blockers

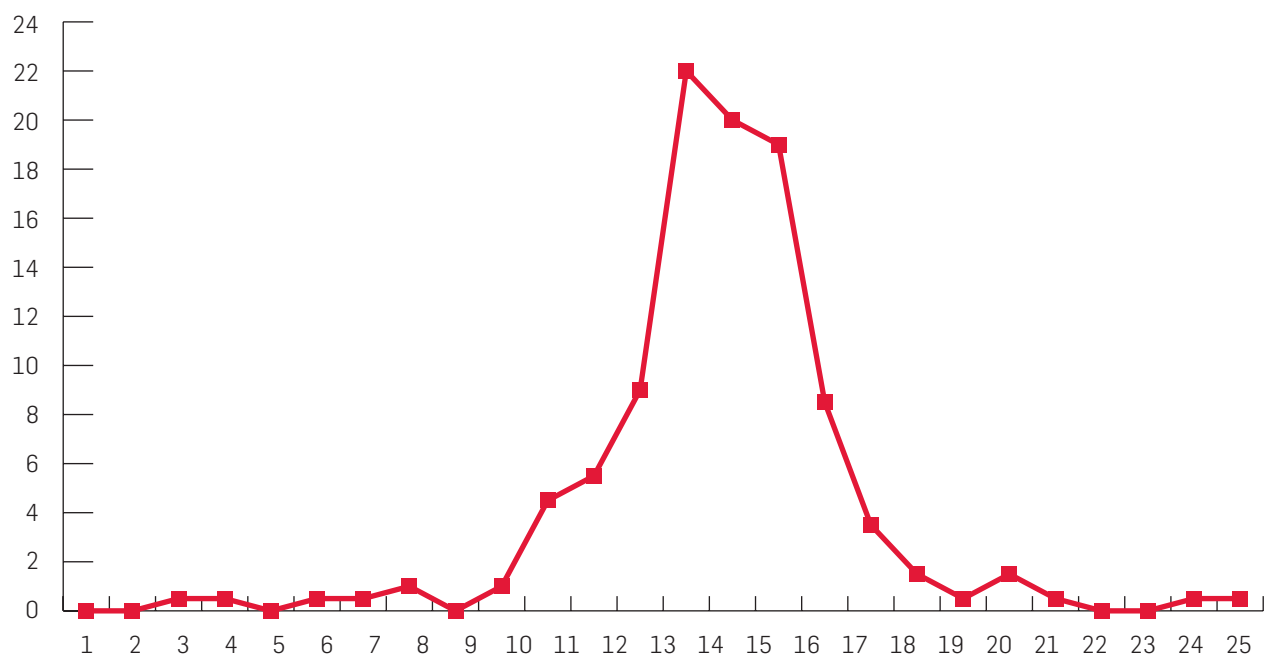


Bone

We asked registrants whether they/their child ever had an x-ray to evaluate scoliosis or curvature of the spine. More than half of respondents (60%) reported that a spine x-ray was never offered or that their doctor said that they did not need an x-ray. 31% of all registrants reported that they/their child did have an X-ray of the spine.

For registrants who had scoliosis surgery, the average age for scoliosis surgery was 14 years old.

Occurrence of Scoliosis Surgery By Age



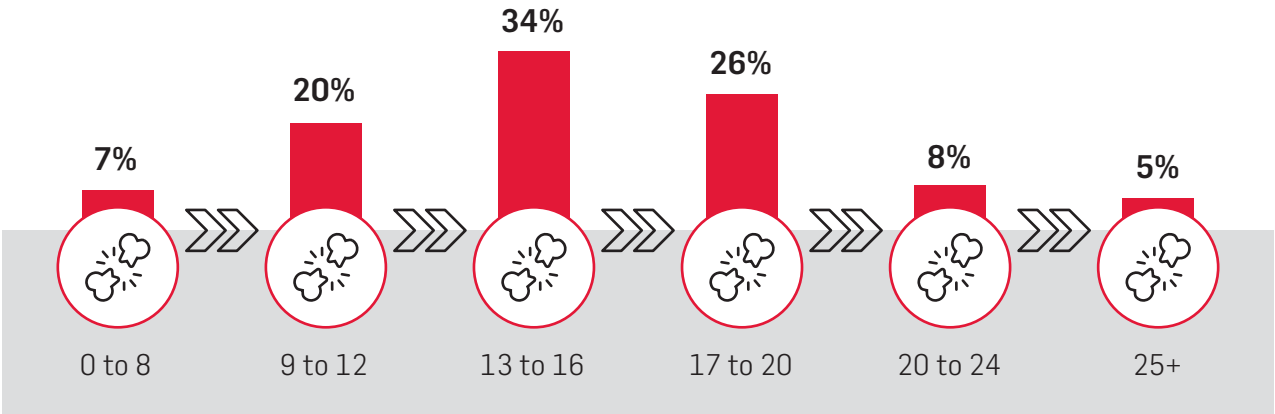
We asked registrants about the occurrence of broken bones after an incident of either minor trauma or no trauma at all. This was true for only a minority of registry participants (21%) and was more common for those with Duchenne than those with Becker.

Broken Bone(s) By Age and Diagnosis

| | | Yes | No |
|----------|-----------|-----|------|
| Age | Diagnosis | | |
| 0 to 4 | Duchenne | 8 | 221 |
| | Becker | 0 | 10 |
| 5 to 8 | Duchenne | 47 | 588 |
| | Becker | 4 | 30 |
| 9 to 12 | Duchenne | 87 | 515 |
| | Becker | 1 | 35 |
| 13 to 16 | Duchenne | 138 | 283 |
| | Becker | 6 | 20 |
| 17 to 20 | Duchenne | 107 | 125 |
| | Becker | 4 | 15 |
| 21 to 24 | Duchenne | 55 | 81 |
| | Becker | 4 | 21 |
| 25 to 28 | Duchenne | 36 | 40 |
| | Becker | 2 | 10 |
| 29+ | Duchenne | 46 | 71 |
| | Becker | 23 | 57 |
| All | | 568 | 2122 |

236 respondents diagnosed with Duchenne answered all the questions regarding broken bones. Of the total number of Duchenne respondents, 22.4% reported fracture after minor or no trauma, 8.7% reported vertebral fractures, and 25.9% reported having either type of fracture. For Duchenne registrants with long bone fractures, 60% of them happened in the teen years (ages 13–20).

Age When Fractures Occur



We asked registrants whether they/their child had ever been diagnosed with a fracture of the spine. The majority (93%) of respondents had not had a fracture of the spine.



“We registered our son in the Duchenne Registry because we felt the effort to track as many boys as possible with Duchenne is very important in moving the science forward.”

Spine Fracture By Age and Diagnosis

| | | Yes | No |
|----------|-----------|-----|------|
| Age | Diagnosis | | |
| 0 to 4 | Duchenne | 0 | 151 |
| | Becker | 0 | 5 |
| 5 to 8 | Duchenne | 6 | 394 |
| | Becker | 0 | 24 |
| 9 to 12 | Duchenne | 17 | 309 |
| | Becker | 0 | 16 |
| 13 to 16 | Duchenne | 30 | 172 |
| | Becker | 0 | 15 |
| 17 to 20 | Duchenne | 23 | 87 |
| | Becker | 0 | 6 |
| 21 to 24 | Duchenne | 7 | 54 |
| | Becker | 1 | 13 |
| 25 to 28 | Duchenne | 4 | 34 |
| | Becker | 0 | 4 |
| 29+ | Duchenne | 0 | 52 |
| | Becker | 1 | 35 |
| All | | 89 | 1371 |

Registry data show that individuals with Duchenne were more likely to experience broken bones or spine fractures than individuals with Becker. Broken bones and spinal fractures are more likely to occur as the individual with Duchenne or Becker gets into the teenage and adult years.

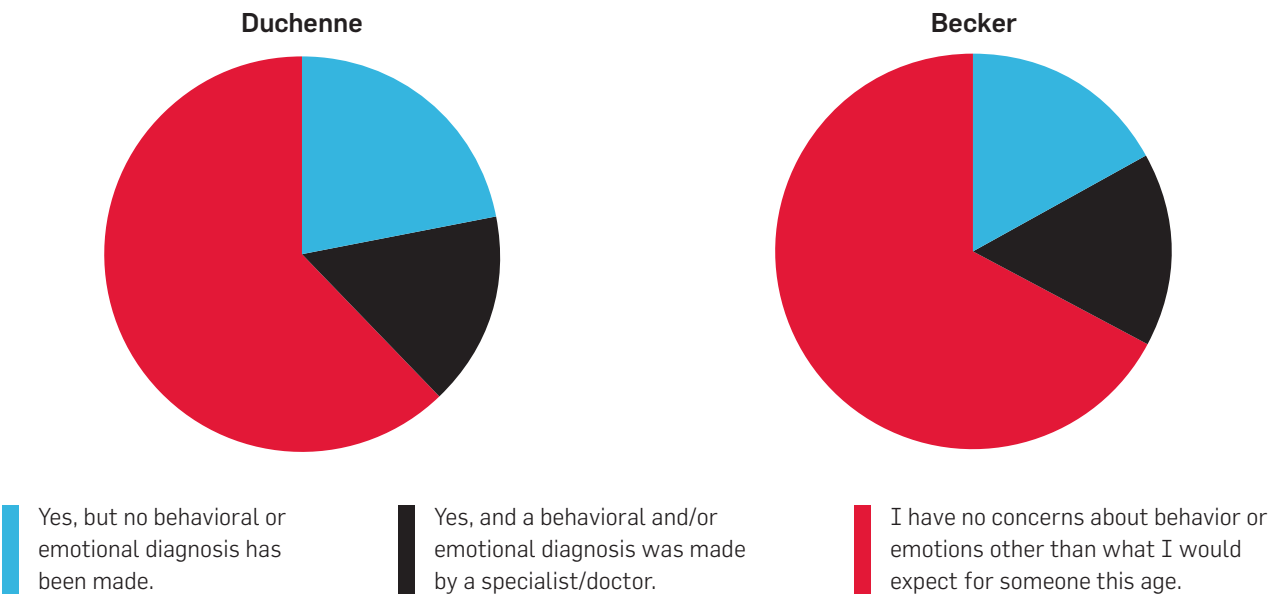
We also asked registrants about use of bisphosphonates for bone health. Only a small number of registrants (11%) reported taking such medications.

Behavior and Learning

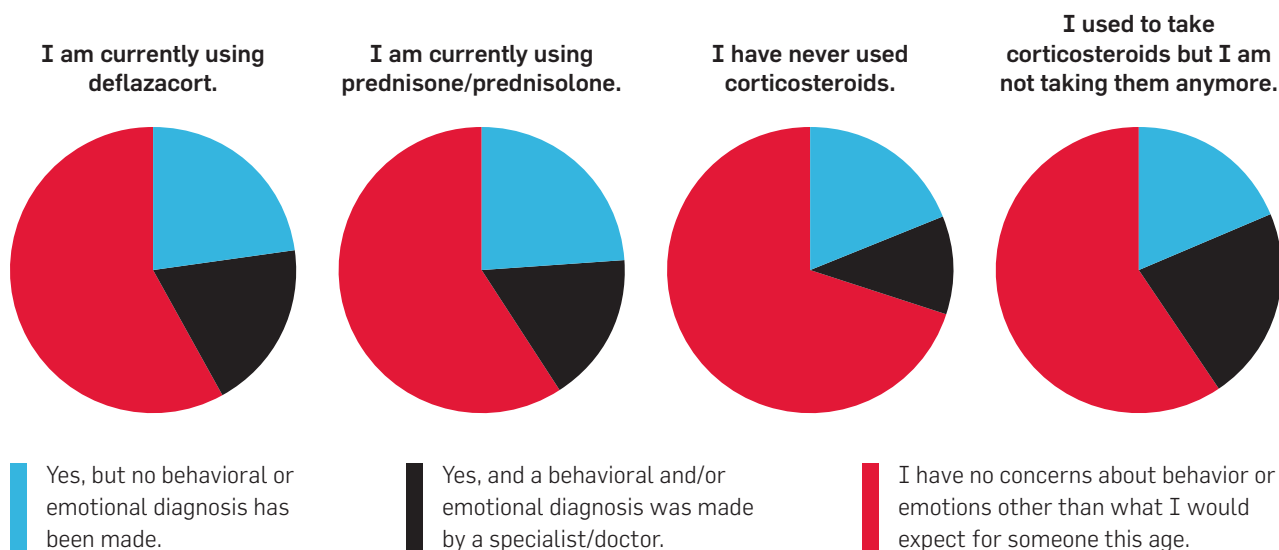
An increased chance for developmental, cognitive, learning, and behavioral difficulties has been documented in people with Duchenne and Becker. Many researchers think this occurs because the dystrophin protein is usually present in the brain (not just in the muscles), so when dystrophin is missing in Duchenne, the brain cells may not function as efficiently as they should. However, not every child with Duchenne or Becker is going to have behavioral or learning problems.

We asked registrants about the existence of any behavioral or emotional disorders. Only 16% of people with Duchenne were diagnosed with behavioral or emotional disorders, while 22% of people with Duchenne or their parents expressed concerns but were not yet diagnosed. 62% of people with Duchenne or their parents had no concerns about behavior or emotions. For Becker, 16% were diagnosed with behavioral or emotional disorders, while 17% expressed concerns but were undiagnosed. 67% of people with Becker had no concerns about behavior or emotions.

Behavioral Concerns By Diagnosis



Behavioral and Emotional Concerns and Corticosteroid Use



Of 739 individuals using deflazacort, 19% had a behavioral or emotional diagnosis (black). 58% reported having no concerns about behavior or emotions (red), while 23% have concerns but have no diagnosis (blue).

Of 579 people with Duchenne or Becker taking prednisone/prednisolone, we see very similar results. 17% of registrants taking prednisone/prednisolone have been diagnosed with emotional or behavioral disorders (black). 59% do not have any concerns about behaviors or emotions (red), whereas 24% are concerned about behavioral or emotional disorders but a diagnosis has not been made (blue).

Of the 831 registrants with Duchenne or Becker who have never used corticosteroids, 11% have a behavioral or emotional diagnosis (black). 70% do not have concerns about behavior or emotional disorders (red). 19% said they were concerned but a diagnosis was not yet made (blue).

There are 272 registrants who have taken corticosteroids in the past, but no longer take them. 22% have been diagnosed with a behavioral or emotional disorder (black). 60% of these individuals do not have concerns about behavioral or emotional disorders (red). 19% indicate they are concerned but have no diagnosis (blue).

In summary, the use of corticosteroids by registrants did appear to increase the risk for a behavioral diagnosis compared to those individuals who never used corticosteroids. However, even the registrants who never used corticosteroids had behavioral disorders, so we know that multiple factors (not just corticosteroid use) contribute to the development of behavioral issues.

Behavioral / Emotional Diagnosis: Duchenne and Becker



Behavioral Concerns: Duchenne and Becker

| | Yes | No |
|--|-----------|------------|
| Behavioral / Emotional Diagnosis | 792 (33%) | 1629 (67%) |
| Anxiety | 134 (6%) | 2287 (94%) |
| Attention Deficit Hyperactivity Disorder | 102 (4%) | 2319 (96%) |
| Attention Deficit Disorder | 84 (3%) | 2337 (97%) |
| Depression | 72 (3%) | 2349 (97%) |
| Obsessive Compulsive Disorder | 68 (3%) | 2353 (97%) |
| Autism | 59 (2%) | 2362 (98%) |
| Asperger Syndrome | 26 (1%) | 2394 (99%) |
| Oppositional Defiant Disorder | 26 (1%) | 2395 (99%) |
| Pervasive Developmental Disorder | 21 (1%) | 2400 (99%) |
| Other Behavioral / Emotional Diagnoses | 19 (1%) | 2402 (99%) |

This table shows the counts and percent of registrants who marked a concern about different types of behavioral problems. Registrants could choose more than one. One third (33%) of registrants marked at least one behavioral problem.

Learning Concerns: Duchenne and Becker

| | Yes | No |
|--|------------|------------|
| Writing Disability (dysgraphia) | 2298 (95%) | 123 (5%) |
| Learning Disability | 1301 (54%) | 1120 (46%) |
| Learning Disability (unsure what type) | 176 (7%) | 2245 (93%) |
| Learning Disability in Reading | 137 (6%) | 2284 (94%) |
| Short Term Memory Problems | 91 (4%) | 2330 (96%) |
| Receptive Language Delay | 90 (4%) | 2331 (96%) |
| Global Developmental Delay | 84 (4%) | 2337 (96%) |
| Verbal Apraxia / Dyspraxia | 72 (3%) | 2349 (97%) |
| Auditory Processing Deficits | 61 (3%) | 2360 (97%) |
| Mental Retardation | 45 (2%) | 2376 (98%) |
| Dyslexia | 43 (2%) | 2378 (98%) |
| Visual Processing Deficits | 24 (1%) | 2397 (99%) |
| Sensory Processing Disorder | 14 (1%) | 2407 (99%) |

This table shows the counts and percent of registrants who marked a concern about different types of learning problems. Registrants could choose more than one. More than half of registrants marked at least one learning problem.

A writing disability (dysgraphia) was reported by 95% of registrants. Writing is perhaps the most difficult academic task to master, as it requires the successful combination of a number of skills. Problems with muscle strength and fine motor dexterity can make the physical act of writing difficult, and problems with language skills can make spelling and grammar difficult. In addition, weaknesses in executive functioning can result in difficulty starting, planning, and organizing longer written projects.



**19% of registrants
participated in a clinical trial.**

**21% of registrants
participated in a research study.**

Clinical Trial and Research Experience

Are you currently participating in a clinical trial?

| | Duchenne | Becker | All |
|---|----------|--------|------|
| I am in a clinical trial right now. | 169 | 3 | 172 |
| I was in a clinical trial that is now over. | 130 | 8 | 138 |
| No, I have never been in a clinical trial. | 1236 | 120 | 1356 |
| All | 1535 | 131 | 1666 |

We asked registrants about their participation in a clinical trial—that is, participation in a study that is designed to see if an experimental therapy or treatment works. Most registrants (81% of all registry participants) reported that they had never participated in clinical trial. This represented 1236 with Duchenne (80%) and 120 individuals with Becker (91%).

Registry participants who did have experience with clinical trials (19% of all registry participants) were either in a trial at the time they were responding to this item (172 or 10% of all registrants) or had previously participated in a clinical trial (138 or 8% of all registrants). Trial participation was slightly higher by individuals with Duchenne as compared to those with Becker. 11% of those with Duchenne were currently participating in a trial compared to 2% of those with Becker. 8% of individuals with Duchenne reported being a trial that has ended compared to 6% of those with Becker.



“I joined the Duchenne Registry to keep up-to-date with trials —I love getting the emails when my son appears to match criteria for a trial. We also joined to provide data so research can progress faster.”

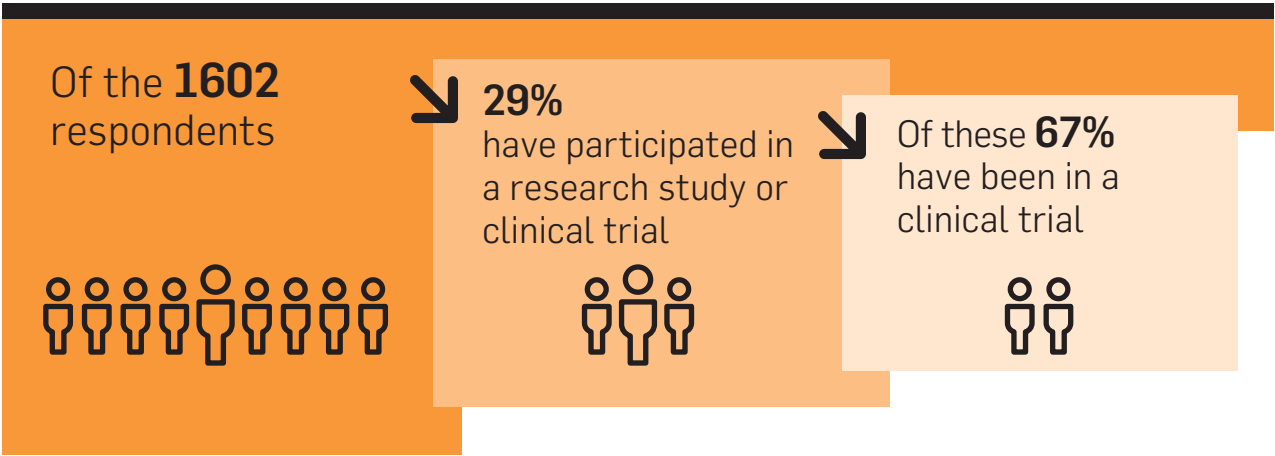
Are you currently participating in a research study other than a clinical trial?

| | Duchenne | Becker | All |
|---|----------|--------|------|
| I am in a research study right now. | 165 | 10 | 175 |
| I was in a research study that is now over. | 148 | 9 | 157 |
| No, I have never been in a research study. | 1170 | 108 | 1278 |
| All | 1483 | 127 | 1610 |

We also asked registrants about their participation in Duchenne or Becker-related research activities other than clinical trials. This would include participation in activities such as natural history and observational studies, survey studies and interview studies. Similar to participation in clinical trials, the majority reported that they had never been in a research study (79% of all registrants). This represented 1170 (79%) of individuals with Duchenne and 108 (85%) individuals with Becker.

21% of all registrants did report they were either currently participating in or previously had participated in a research study. Participation was again higher in those with Duchenne compared to individuals with Becker muscular dystrophy. 11% of those with Duchenne were currently participating in a research study compared to 8% of individuals with Becker. Similarly, 10% of those with Duchenne participated in a research study that has since ended compared to 7% of registrants with Becker.

Clinical Trials



Conclusion and Acknowledgements

In summary, we would like to thank all the families in our community who have joined The Duchenne Registry and contributed data over the past 10 years. We are grateful for every person who registered and updated their profile. We truly appreciate your time and dedication!

In addition to The Duchenne Registry team, we would like to thank the team at RTI International Center for Newborn Screening, Ethics, and Disability Studies for their meticulous work on the Ten Year Registry Report. All statistical analyses and graphics were generated by RTI International. A special thanks to the Powers family for sharing their beautiful family photos.

In order to expand on this important work, we strongly encourage your continued participation in the next phase of the Registry. We will be launching a new registry platform in 2019 that will make your user experience much easier and more engaging. Help advance research and speed the development of new therapies by participating in The Duchenne Registry.

**You have the power
to make a difference!**

Parent Project Muscular Dystrophy

JOIN THE FIGHT.
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