The Duchenne Registry: Your Data Visualized

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1. Parent Project Muscular Dystrophy

Introduction

The Duchenne Registry (formerly DuchenneConnect) began in 2007, when 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. What they envisioned was a central hub that would bring together those living with Duchenne or Becker, along with their families and caregivers, to connect them with medical research, clinical care, clinical trials, and each other. At the same time, it would also be a resource for researchers and industries with an interest in Duchenne, allowing access to aggregate, de-identified data provided by patients and their families — information that could prove vital to advances in care and treatment. Today, the result of this endeavor is The Duchenne Registry, the largest, most comprehensive registry for Duchenne and Becker muscular dystrophy and women who are carriers. The Duchenne Registry (Registry) is a patient-report registry.

Benefits of participating in The Registry

- Receive clinical trial information specific to you/your family member
- Contribute to research from home!
- Gain a better understanding of EVERYONE with dystrophinopathies

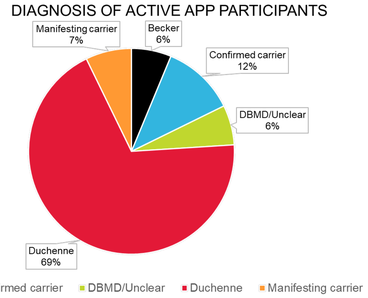
Methods

Participants in the Registry complete surveys about various health and daily experiences living with Duchenne/Becker or being a carrier. They can complete a total of 12 surveys, and we ask that they complete most surveys at least annually. We also ask participants to share a copy of their genetic test report. Participants can provide as much or as little data as they want. However, the more data we have, the more we can share with researchers and the better we can tailor information to a specific participant.

The Registry contacts participants, usually at least once a month, through app notifications and emails. Contact may be to complete a survey, share an educational newsletter, or notify registrants about new research studies and clinical trials or upcoming activities such as webinars and conferences.

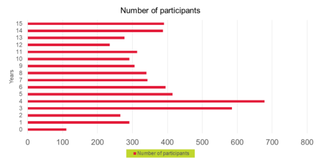
Demographic Information

DIAGNOSIS OF ACTIVE APP PARTICIPANTS



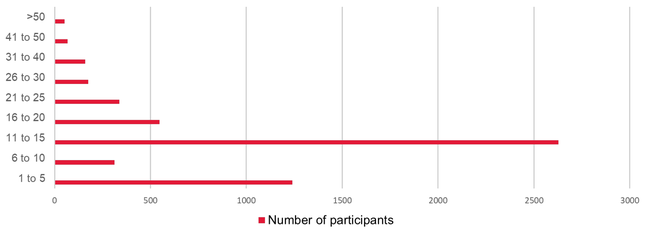
5632 individuals with dystrophinopathy have provided data to the Registry, and 1361 participants are currently active on the app.

YEARS PARTICIPATING IN THE REGISTRY



Top 5 Countries in the Registry	Count	Percentage
USA	3780	62%
India	336	6%
Australia	208	3%
Canada	188	3%
United Kingdom	108	2%

SURVEYS COMPLETED

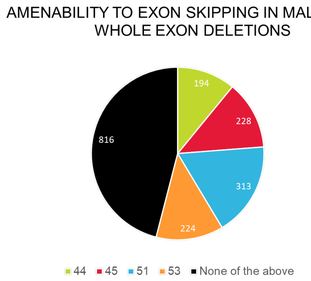


Genetic Variants

The Registry has one of the largest databases of genetic variants in dystrophinopathy. Genetic counselors review the genetic test reports and curate the variants so that the data can be searched and utilized by researchers.

Genetic Variant Type	Count	Percentage
Deletion of one or more exons	2185	67%
Duplication of one or more exons	373	11%
Nonsense	340	10%
Missense	22	1%
Splice site	108	3%
Other small sequence variants	209	6%

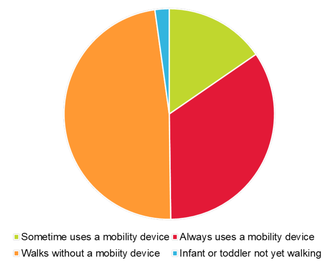
AMENABILITY TO EXON SKIPPING IN MALES WITH WHOLE EXON DELETIONS



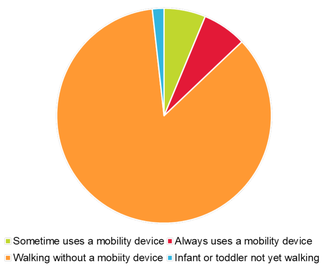
Muscle Function

A key survey in the Registry asks about muscle function, including ambulation and function of the upper and lower limbs. This survey is very important for determining clinical trial eligibility and is often used in research.

AMBULATION IN DUCHENNE AND BECKER



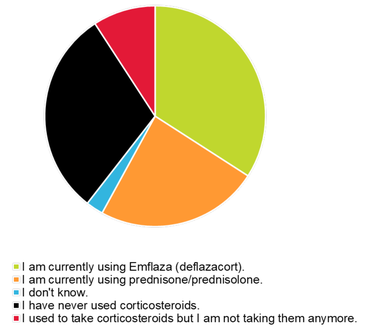
AMBULATION IN FEMALE CARRIERS



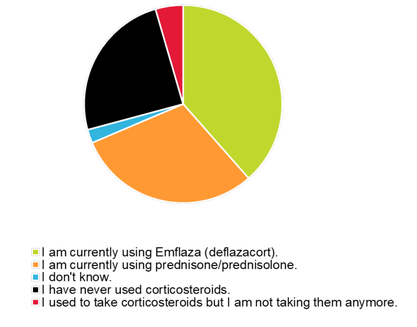
Corticosteroids

As a primary treatment for Duchenne, understanding use of corticosteroids is very important.

CORTICOSTEROID USE IN DUCHENNE (ALL AGES)

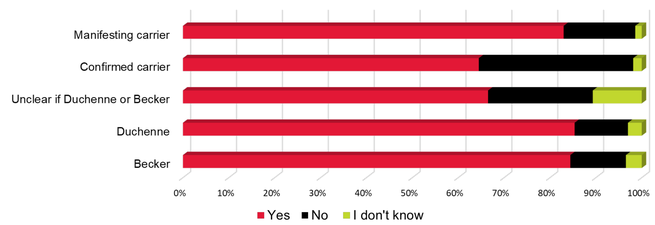


CORTICOSTEROID USE IN DUCHENNE (AGES 6-12)

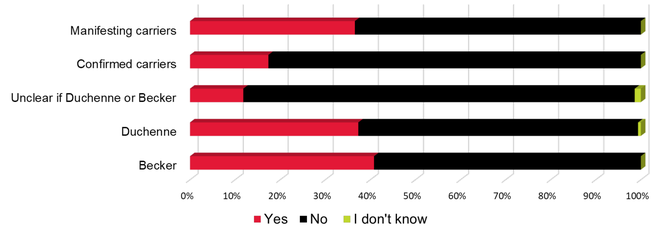


Heart Care and Medications

PERCENTAGE OF INDIVIDUALS OVER AGE 5 WHO HAVE HAD AN ECHOCARDIOGRAM



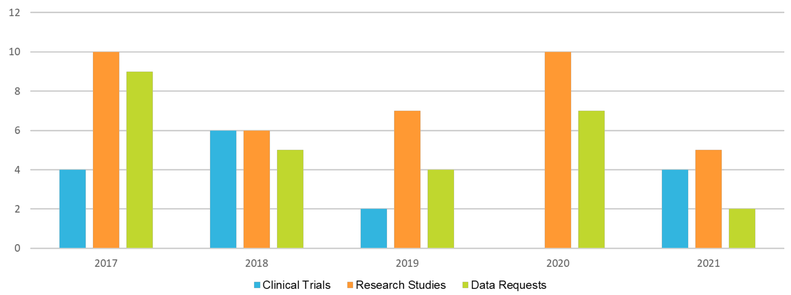
PERCENTAGE OF INDIVIDUALS TAKING CARDIAC MEDICATIONS



The Registry at Work

The Registry is helpful to both families and researchers. The graph below shows how the Registry has been used over the last five years, including the number of clinical trials recruited, the number of research studies recruited, and the number of researchers who have requested data.

REGISTRY AT WORK!



Acknowledgements

Your time and efforts are greatly appreciated and have advanced understanding of and research in Duchenne, Becker and carriers!