Novel Model for Decentralized Data and Biosample Repository for Duchenne and Becker Muscular Dystrophy

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Purpose

The purpose of this project is to create a decentralized, all-encompassing platform to connect researchers to coded data and biosamples from individuals with Duchenne and Becker muscular dystrophy (DMD/BMD), as well as carriers of these mutations.

Background

Duchenne muscular dystrophy and Becker muscular dystrophy are X-linked degenerative disorders of the dystrophin protein, which leads to progressive muscle weakness. DMD usually leads to death in early adulthood and is the most common neuromuscular disease of childhood[1]. Natural history[2] and observational studies[3] have been conducted in DMD and BMD worldwide. However, there is no single, comprehensive, widely accessible database in the US of DMD/BMD biological specimens with integrated clinical data.

CureDuchenne Link™ uses custom technology and world-class partnerships to deliver a seamless, patient-centric experience. Building on previously existing nation-wide community engagement efforts of our patient advocacy organization, CureDuchenne Link allows participation from anywhere in the US, no matter where participants reside or receive their care. Participants can choose from a menu of opportunities, all of which can be done in-home or near home.

Breaking the mold of siloed projects, CureDuchenne Link allows qualified researchers access to coded data, including whole genome sequencing data, and biosamples in one all-encompassing platform. This data is curated using industry-standard terminology, allowing harmonization with other research data sets.

Methods

Creating this platform required multiple different vendors to support infrastructure and operations. Firstly, electronic consent forms were required given the decentralized nature of this study. This was achieved using an online, 21 CFR part 11 compliant program. In the consent form, participants are asked if they are willing to provide data via surveys, medical records, and biosamples. These optional biosamples include blood, urine, saliva, skin, muscle, and any archived tissue samples.

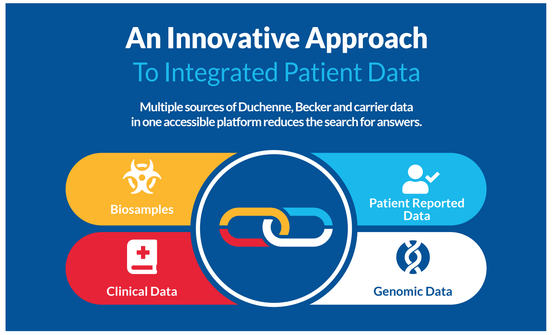

Figure 1: CureDuchenne Link Integration Diagram

To reduce burden of participation, custom software was created to facilitate participant registration and collection of participant reported data in one easy-to-use online portal. For participants with DMD/BMD that elect to provide medical records, data abstraction is performed by central project staff and is stored in a central data warehouse.

Data and biosamples provided by the participants are linked using a unique identifier and can then be shared with approved DMD researchers. CureDuchenne Link also connects researchers to participants, alerting participants when they qualify for additional opportunities or trials.

Results

When project planning commenced in early 2020, decentralized processes were not yet widely adopted and many vendors needed to make modifications to meet project requirements. Additionally, as a comprehensive solution was not readily available, cross-vendor platform integration was required. Vendor selection, protocol design, IRB review and approval, and platform testing took roughly 18 months.

Once the project was approved by WIRB-Copernicus Group (WCG), 18 participants from 14 households served as the pilot enrollment group. These participants further tested the functionality of the platform. 10 of these participants were surveyed regarding their experience using CureDuchenne Link. 100% of participants rated the overall experience good or excellent. The surveyed participants the website a 4.7/5 rating and 100% said that they were very likely to recommend participation.

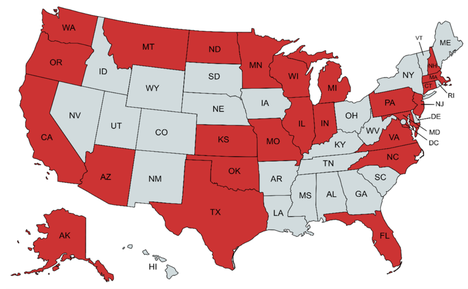

Figure 2: States Represented in CureDuchenne Link

In July 2021, CureDuchenne Link launched for nation-wide enrollment. After 5 days of open enrollment, there were 49 participants from 43 different households, representing 25 different states. Of the participants that have completed the consent process, 100% agreed to complete patient reported surveys, 97.1% of participants with DMD/BMD agreed to provide medical records, and 87.8% of participants agreed to provide at least one type of biosample.

Conclusions

CureDuchenne Link has created the first decentralized, all-encompassing platform of coded data and biosamples in the US for DMD/BMD. The field of decentralized research is still in its infancy, however CureDuchenne has developed a platform that is easily replicable in other disease areas. The rapid upscale from 18 to 49 participants in 5 days demonstrates the scalability of this platform.

Early feedback from pilot participants indicated that the online portal is easy to use, and the surveys are comprehensive, but not overwhelming. This is key to increasing participant retention. In addition to the user-friendly platform, CureDuchenne Link incentivizes participation by providing a menu of ways to participate, allowing for flexibility based on participant preference.

CureDuchenne Link has created an effective, comprehensive tool for decentralized research and biosample collection that patients are very willing to use. Beginning in 2022, CureDuchenne Link will expand to include archived muscle tissue, as well as facilitate distribution of coded data and samples to qualified researchers with the projects focusing on muscular dystrophy.

https://flowcode.com/p/erpgEla3y
Figure 3: QR code to references