

PPMD Care Project: Psychosocial Issues in Duchenne - Interim Report
January 28, 2022

PPMD is happy to provide an update on progress in the Psychosocial Issues in Duchenne Project, outlined below in four parts:

1. Comprehensive literature review
2. Creation and implementation of patient/caregiver and clinician surveys to better understand current prevalence, assessment and management practices
3. Development of a clinical neurobehavioral screening tool for Duchenne and Becker muscular dystrophy
4. Expert consensus meetings

Updates on Progress:

1. Comprehensive Literature Review

We are happy to share that the comprehensive literature view was completed by Dr. Molly Colvin, PhD, Massachusetts General Hospital. This review acted to inform the 2021 Duchenne and the Brain Expert Consensus Meeting (additional information below). In tandem, Dr. Jos Hendriksen, PhD, Leiden University, is working on a meta-analysis publication. In an effort to not duplicate efforts, PPMD opted to not publish the findings of the comprehensive literature review, but instead use them to inform discussions at the consensus meeting. Dr. Hendriksen's publication is pending.

2. Surveys to establish prevalence, assessment, and management

Parent/Caregiver survey: This survey was created with the goal of collecting data on management practices from the patient/caregiver perspective. Participants were recruited via The Duchenne Registry, and **320 respondents** completed the survey, which far exceeded recruitment goals. A majority of respondents reports *Nationwide Children's Hospital (Columbus, Ohio)*, *Cincinnati Children's Hospital Medical Center (Cincinnati, Ohio)*, *Duke Children's Hospital (Durham, North Carolina)*, and *UCLA (Los Angeles, California)* as their primary neuromuscular care site. Other sites from across the United States were represented.

Pertinent learnings to date include:

- Most families bring concerns of behavior, learning, and cognition to the primary care team/pediatrician, *not* the neuromuscular team.
- However, most primary care providers (68%) did not screen for the presence of neuropsychiatric or neurobehavioral problems.

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- Most patients (74%) reported no use of medications for the treatment of neuropsychiatric or neurobehavioral conditions.
- Barriers to obtaining care for neuropsychiatric or neurobehavioral issues most often included “treating other medical problems,” “no access to providers,” “PCP not receptive,” “not enough time,” “insurance coverage issues,” or “logistical problems.”
- Caregivers endorse higher use of therapy services than medication

Analysis of this data set is ongoing, with additional data points to be made available in the coming months. PPMD is pleased to be working with a data scientist to better understand the data elicited from this survey, and looks forward to sharing additional learnings in the future.

Provider Survey: This survey was created with the goal of understanding current prevalence, assessment, and management practices from the provider perspective. Participants were recruited via the Certified Duchenne Care Center network and the Decode Duchenne provider network, and **74 physicians completed the survey**. Top clinic locations to submit responses included *Children’s Hospital Colorado (Aurora, Colorado)*, *Helen DeVos Children’s Hospital (Grand Rapids, Michigan)*, and *University of Virginia Children’s Hospital (Charlottesville, Virginia)*.



This chart represents geographic spread of caregiver respondents (green) vs physician respondents (purple). In several locations, the icons overlap, representing families receiving care from centers who also responded to the survey.

Pertinent learnings to date include:

- Providers estimate that 25-75% of their patients with Duchenne meet criteria for the diagnosis of a neuropsychiatric or neurobehavioral condition, but that only 10-15% have received a formal diagnosis and appropriate treatment for their diagnosis
- A majority of neuromuscular providers do screen for these conditions in their clinic as part of routine care:
 - o 44% complete informal assessment

- 25% review the IEP/504 plan
- 14% complete a verbal formal assessment
- 16% complete a formal written (paper/electronic) assessment
- Most neuromuscular providers (55%) have a designated psychologist and/or neuropsychologist to whom they refer, but do not have access to that provider in real time
- However, a large majority (90%) of neuromuscular providers *do not* have access to a psychiatrist, **and** a majority (70%) do not feel comfortable prescribing medications for neuropsychiatric or emotional/behavioral concerns
- Of providers who **do** prescribe medications for these reasons, 27% are SSRIS or SNRIs for the treatment of depression, anxiety, and OCD; 14% are stimulant medications for the treatment of ADHD, and 12.5% are a-Adrenoceptor agonists for the treatment of ADHD. 10% are antipsychotics for the treatment of anger or emotional dysregulation, and another 10% are beta-blockers for the treatment of anxiety.

Analysis of this data set is ongoing, with additional data points to be made available in the coming months. PPMD is pleased to be working with a data scientist to better understand the data elicited from this survey, and looks forward to sharing additional learnings in the future. Additionally, PPMD is considering a second recruitment notification to elicit additional survey responses.

3. Development of a clinical neurobehavioral screening tool for Duchenne and Becker muscular dystrophy

To fill the gap as illustrated in survey findings, PPMD partnered with key psychologists and neuropsychologists in the development of a comprehensive screening tool, the “DBMD BELS Questionnaire.” This tool is currently being piloted under neuropsychologist Dr. Seth Sorensen at Arkansas Children’s Hospital in Little Rock, Arkansas.

The BELS (Behavior, Emotion, Learning, Social) tool was developed by Natalie Truba, PhD, and Molly Colvin, PhD to assess the full spectrum of DBMD symptoms with the goal of completing a broad assessment of symptoms across domains, with the goal of guiding referral decision making. The tool is not intended to gather sufficient data to make diagnosis, but instead acts as a general screener. As the tool crosses domains, it serves to minimize the number of assessments needed to screen for a variety of conditions, and minimizes the need to complete multiple assessments or refer for baseline screening.

The tool covers four domains: Behavior (ADHD, sensory, RRBI), Emotion (anxiety, depression, OCD), Learning (executive function, ID, LDs, Memory, Language), and Social (ASD, social pragmatics), and includes 45 items (yes/no/unsure) that parents/caregivers complete.

The tool is currently being used in ACH's multi-disciplinary dystrophinopathy clinic and has been in use since July 2021. Surveys are completed in clinic via an iPad. It takes about 10 minutes to complete.

- As of January 2022, 41 patients have participated, ranging in age from 4 – 19 years (M=12.68).
- 32 subjects carried a diagnosis of Duchenne, and 9 a diagnosis of Becker.
- 27 subjects (65%) were on steroids.
- 63% (N=26) of caregivers who completed the survey were the subject's mother, 22% (N=9) were the father, 10% (N=4) were the patient, and 5% (N=2) were "other guardian."
- 93% reported having previously seen a psychologist or psychiatrist
- 71% previously carried a psychological/psychiatric diagnosis prior to the time of the screening tool completion, with 51% (N=21) have at least two.
- Most frequently endorsed items within the tool include those associated with the behavior domain (5/12 items), with additional endorsement of learning (3/11 items).
- Social items are least endorsed (4/11 items).
- Total number of completed assessments was negatively impacted by COVID, including patients completed telemedicine visits or declining to attend in-person clinic appointments. Additionally, initial data analysis brought up concern for data skewing due to increased stress and anxiety due to the pandemic.

This pilot continues to collect data and data will be analyzed once the pilot is complete in July 2022. At that time, analysis will guide next steps including roll-out to additional clinic sites to aid in additional data collection and tool validation. A publication including pilot data and early learnings will be generated.

4. Expert Consensus Meeting

PPMD was happy to host the Dystrophinopathy and the Brain meeting from November 11-12, 2021 at the TWA Hotel at JFK Airport, New York City, New York. This meeting brought together a spectrum of experts from the field including neurologists, psychologists, neuropsychologists, therapists, behavior experts, and research from across the globe to discuss the issues families face due to lack of dystrophin the brain in Duchenne. This two-day meeting covered a variety of topics including work being done via the PPMD Psychosocial Issues in Duchenne project, and facilitated rich discussion and consensus around screening, diagnosis, and treatment. A meeting report is currently in process to be submitted for publication.

Themes that emerged from the meeting included enthusiasm for the BELS and eagerness to validate its use across the country and development of a psychiatric drug prescribing algorithm to support neuromuscular experts in their management of patients with Duchenne who require medical intervention but may not have access to appropriate professionals in attempt to standardize drugs prescribed and minimize polypharmacy.

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PPMD is grateful for the funding to complete this work, and is excited to share subsequent learnings in 2022 and beyond. Thank you for your generous support of this crucial work as we navigate how to better care for all people living with Duchenne.