Establishing Common Terminology to Advance Efforts in Duchenne Muscular Dystrophy Newborn Screening

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BACKGROUND
The approval of new therapies for Duchenne Muscular Dystrophy (DMD) has accelerated the need for pre-symptomatic identification of individuals with DMD. Comprehensive pilots of newborn screening (NBS) for DMD have the potential to generate the evidence base that establishes the benefit of screening for DMD in the neonatal period. Parent Project Muscular Dystrophy (PPMD) created the Duchenne Newborn Screening Consortium (DMDNBSC) to conduct a state-based pilot funded by the biopharmaceutical and medical device industry, involving patient advocacy representatives, and leveraging national resources and tools. This novel consortia approach leverages the system of NBS in the United States which is a multi-faceted program of prenatal education, physiological and molecular screening shortly after birth, diagnosis of screen positive cases, and referral for clinical care and treatment. The involvement of stakeholders across each of these areas is critical to piloting DMD newborn screening.

The DMD NBS pilot will enroll and screen 100,000 newborns over a two-year period at birthing centers within the Northwell Health System and New York Presbyterian Hospitals. In addition to generating data about the analytical and clinical validity of the NBS method, the pilot will collect clinical and health information on screen positive cases and gather parent input through an online questionnaire and in person interview. To accomplish the DMD NBS pilot goals, the consortia will utilize tools, resources, and expertise from the Newborn Screening Translational Research Network (NBSTRN), a key component of the Eunice Kennedy Shriver National Institute of Child Health and Human Development Hunter Kelly Newborn Screening Research Program led by a team at the American College of Medical Genetics and Genomics (ACMG).

OBJECTIVE
To transform the DMDNBS Cloud CDEs into standardized terminology using the LOINC database, we exported and compared the CDEs to an existing data dictionary in LOINC. When possible, pre-existing LOINC codes were matched to questions from the DMD pilot and were identified as common data elements. Most questions were unique, thus the creation of new LOINC codes specific to DMD are required. These data elements and mappings will soon be uploaded to the NIH Common Data Elements Repository, which can be referenced for future research.

METHODS
NBSTRN developed the Longitudinal Pediatric Data Resource (LPDR) using REDCap in a secure environment to collect DMD pilot data using a series of question and answer sets, called common data elements (CDEs), established by clinical care experts. This tool, DMDNBS Cloud, will be launched in a cloud-based information technology environment. To facilitate the use of DMDNBS Cloud the NBSTRN partnered with the National Library of Medicine (NLM) to map ontology and electronic messaging standards to 339 CDEs. We report on an effort to map Logical Observation Identifiers Names and Codes (LOINC) to the CDEs. LOINC helps establish common terminology for laboratory and clinical observations and utilizes a different code for each test, measurement, or observation that has a clinically different meaning. The DMDNBS Cloud presents the CDEs organized into modules that match the clinical workflow from demographics and family history to genetic testing and longitudinal follow-up.

CONCLUSION
This partnership between the NBSTRN and the NLM was valuable for training medical students to become more familiarized with biomedical informatics and data management. Helping to build this set of LOINC codes for the DMD NBS pilot developed a greater appreciation for the process of approving candidate conditions for federally recommended newborn screening. Gaining insight on these steps in precision medicine helped to understand the importance of early diagnosis to minimize the impact of genetic diseases like DMD.