DMDNBS Cloud: Creating the Evidence Base for Duchenne Muscular Dystrophy Newborn Screening

Amy Brower, PhD
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Piloting DMD NBS is a Priority

• Interest in screening all babies for Duchenne Muscular Dystrophy (DMD) has increased because of new therapies and management strategies

• Newborn screening (NBS) for DMD has not been nominated to the Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC)

• We undertook a consortia approach to developing the data required for nomination to the ACHDNC Recommended Uniform Screening Panel (RUSP)
Consortia Approach

• Capitalizes on each member’s expertise

• Distributes effort across the many stakeholders in NBS

• Reflects the multi-faceted NBS system in the United States

• Leverages the increased focus on drug development for rare disease by the pharmaceutical industry

• Pilots emerging technological and clinical practices

Please visit Poster #106 – The Duchenne Newborn Screening Consortium: Accelerating the Path to Nationwide Screening: Lloyd-Puryear et al.
1. Newborn Screen
- NBS Program at Wadsworth Center, New York Department of Health
- Principal Investigator: Michele Caggana, ScD, FACMG
- Analytical Validation of Creatine Kinase (CK) MM Isoform Assay
- Analytical Validation of Next Generation Sequencing of Genes for DMD and Other Neuromuscular Diseases
- Hospital-Based Recruitment with Informed Consent at Northwell Health and New York Presbyterian Hospital

2. Clinical Care
- Positive NBS Results Communicated to Parents by Genetic Counselors from Northwell Health or New York Presbyterian Hospital
- Clinical Decision Support Tool Developed by ACMG to Inform Pediatrician
- Female Carrier Results Communicated to Parents by Genetic Counselors
- Multi-disciplinary Comprehensive Care Tracked for All NBS Positive Results

3. Longitudinal Health Outcomes
- NBSTRN Led Group of Expert Clinicians Develop Longitudinal Set of Questions and Answers
- Focus on Asymptomatic Newborns Who Have a Genetic Diagnosis for DMD or Other Neuromuscular Condition
- Secondary Focus on Parent Experience with NBS Pilot and/or DMD or Other Neuromuscular Condition Diagnosis
- Electronic Case Report Form in the Longitudinal Pediatric Data Resource (LPDR) Details Diagnosis and Treatment
- Leverage Existing Patient Registries

4. Pilot Results
- NBS Screen CK-MM Performance
- NBS NGS of Genes for DMD and Other Neuromuscular Conditions Performance
- Clinical Exam Findings
- Number of Babies Screened
- Number of Diagnosed Babies
- Number of Carrier Females
- Estimates of Incidence
- Treatment Selection

5. RUSP Nomination
- Background on Conditions Identified by Using CK-MM as a Newborn Screen
- Background on Conditions Diagnosed by Using NGS of Gene for DMD and Other Neuromuscular Conditions
- NBS Screen Details
- Diagnosis Details
- Health Outcomes of NBS Identified Cases
- Treatment Choice for NBS Identified Cases
RUSP Review Informs Pilot Design

- Expansion of RUSP Begins with Nomination
- Evidence Review
- Screening Technology
- Diagnosis
- Treatment
- Health Outcome in Conditions Identified Through NBS
- Population Based Pilot

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New NBSTRN Tool

• Supports the analytical and clinical validation of new technologies
• Tracks emerging findings from pilots
• Includes ACHDNC Portal to inform Evidence Review Group
• Hosts the DMDNBS Cloud
Special Acknowledgement to the DMD NBS Consortium

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