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4. AN AUTOMATED SYSTEM FOR EARLY DIAGNOSIS, SEVERITY AND PROGRESSION IDENTIFICATION IN DUCHENNE MUSCULAR DYSTROPHY: A MACHINE LEARNING AND DEEP LEARNING APPROACH

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Background: Duchenne Muscular Dystrophy (DMD) is a fatal X-linked congenital genetic disorder affecting 1/5000 males that is caused by a lack of dystrophin, a protein critical for function of muscle cells. The disorder leads to progressive debilitating muscle weakness and loss of ambulation around the age of 12 with early death due primarily to cardiac and respiratory complications in the late teens or early 20s. Weakness is detectable from infancy, but symptoms are typically recognized during toddler or early childhood years. There is presently no cure for the disorder, but novel gene repair interventions and other preventive therapies are initiated as early as possible to slow progress of the disease and prevent secondary conditions. Tools are needed to 1) facilitate early diagnosis; 2) identify early indicators of clinical severity, and 3) quantify and track progression of muscle weakness across the ambulatory phase of the disease.

Methods: Here, we present an Artificial Intelligence (AI)-based detection of gait characteristics in toddlers and children with DMD and typically-developing peers. Our system collects data from mobile device acceleration sensors remotely and in real time using our novel Walk4Me smart phone application. Our web application extracts temporal/spatial gait characteristics and raw data signal characteristics, and then uses traditional machine learning and deep learning techniques to identify patterns that can 1) identify children with gait disturbances associated with DMD, 2) describe the degree of mobility limitation, and 3) identify characteristics that change over time with disease progression. Results: We have identified several machine learning techniques that differentiate between DMD and typically-developing children with >99% accuracy across the age range studied and have identified corresponding temporal/spatial gait characteristics associated with each group.

Conclusion: Our work manifests how the latest advances in mobile device and machine learning technology can be adapted to measure clinical outcomes regardless of point of care and that may be used to inform early clinical diagnosis, treatment decision making and to monitor disease progression.