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Use of Artificial Intelligence to Identify Predictors of Functional Outcomes in Patients with Facioscapulohumeral Muscular Dystrophy

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Use of Artificial Intelligence to Identify Predictors of Functional Outcomes in Patients with Facioscapulohumeral Muscular Dystrophy

Submitting/Presenting Author (must be a trainee): Natalie Katz, MD, PhD
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☐ Medical Student
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Other authors/contributors involved in project: John Hogan, PhD, Colin Cernik, Ryan Delbango

IRB Number: n/a; project submitted for IRB review at outside institution (University of Rochester where database is held) and was deemed exempt

Describe role of Submitting/Presenting Trainee in this project (limit 150 words):
This project involved investigators in multiple locations: Jeffery Statland, MD from KU Medical Center served as the Principal Investigator for this project. Natalie Katz, MD, PhD was involved as a junior investigator on the clinical team to assist with study design, data preparation, analysis and interpretation. The team also included Colin Cernik, a junior statistician from KU. John Hogan, PhD is the principal engineer and founder of AIbytes, who, along with Ryan Delbango, completed the AI (machine learning) analysis.

Background, Objectives/Goal, Methods/Design, Results, Conclusions limited to 500 words

Background: Facioscapulohumeral muscular dystrophy (FSHD) is one of the most common forms of muscular dystrophy characterized by slowly progressive muscle weakness. Symptoms initially involve the facial and shoulder girdle muscles, with later involvement of the distal lower extremities, pelvic girdle and proximal lower extremities. FSHD type 1 (FHSD1) is the most common form (~95% of individuals) and is caused by deletion of microsatellite repeats in the D4Z4 region on chromosome 4 (4q35). Normal individuals have >10 repeats whereas individuals with FSHD1 have 1-10 repeats. There is significant variability in regards to disease severity, rates of progression and functional outcomes, but few studies have followed patients long enough to understand what drives functional outcomes, creating gaps in clinical care.

Objectives/Goal: The goal of this project was to analyze data from participants enrolled in the US National Registry for FSHD to identify factors that might be predictive of functional outcomes.
Methods/Design: We performed a retrospective cohort study using de-identified, longitudinally collected data from 578 participants with FSHD1 and an average of 9 years of follow up (range 1-18 years). Data were analyzed using traditional epidemiological methods and machine learning (artificial intelligence; AI) techniques to assess interactions between characteristics including: age, gender, genetics (# of D4Z4 repeats), age of symptom onset and diagnosis, wheelchair use, job loss due to FSHD, medication use and medical comorbidities. These data were also used to develop AI random forest algorithms to identify risk factors that were predictive of wheelchair use.

Results: More than half of patients (55%) reported symptom onset prior to age 18, however, there was an average diagnostic delay of 13 years. Consistent with the literature, we found that small allele size (1-3 D4Z4 repeat units) was associated with earlier diagnosis (median 14 years, 95% CI 11, 17), facial weakness as the initial symptom (53.7%), and higher risk of wheelchair use. Across all repeat lengths, we showed that women were more likely to use a wheelchair. Our AI final model identified disease duration, number of medications, age at diagnosis or symptom onset, and medical comorbidity (e.g. breathing difficulties, pneumonia, or arthritis) as the most significant predictors of wheelchair use; genetics provided only a small contribution to wheelchair use in the AI model.

Conclusions: Analysis of Registry data provided a unique opportunity to study a rare, slowly progressive disease using years of longitudinally collected data to identify predictors of functional outcome. Further, AI techniques allowed us to incorporate data that is commonly collected but rarely analyzed due to the complexity of analysis when incorporating multiple variables. Early AI modeling identified several features associated with wheelchair use, including disease duration, number of medications and medical comorbidities, and showed that genetics had surprisingly little influence on wheelchair use. This raises the possibility that better medical management may impact functional outcomes in patients with FSHD, independent of genetics, and may be important for clinical trial design and outcomes measurements.