Assessment of Disease Progression and Costs Associated with Duchenne Muscular Dystrophy (DMD) Using the Optum Market Clarity Database

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Background and Objective

DMD is a severe hereditary neuromuscular disease caused by mutations in the dystrophin gene, primarily affecting males. These mutations lead to progressive muscle fiber degeneration and weakness, initially causing difficulty in ambulation, which may progress to wheelchair use. Understanding DMD through real-world data is essential to improve patient outcomes. This study aimed to assess disease progression and costs in DMD patients over 5 years.

Methodology

- A retrospective study was conducted using the Optum de-identified Market Clarity claims and EHR database. DMD patients were identified with ICD-10 code G71.01 from July 2017 through June 2019.
- The index date was the date of the first DMD diagnosis.
- Patients with ≥2 outpatient claims (≥30 days apart) or with 1 inpatient claim with continuous enrollment/eligibility (including both medical and pharmacy coverage) for 6 months pre and 5 years post-index date were included. From these, patients having elevated creatine kinase (≥10,000 U/L) were included if they had a positive mention of at least one DMD term in clinical notes.
- Patients diagnosed with DMD during the baseline period, those with a positive mention of Becker muscular dystrophy (BMD) in clinical notes, and those older than 7 years at the index date were excluded.
- Disease progression was assessed by tracking the emergence of disease signs, symptoms, and complications across age categories (1 to 3 years, 4 to 5 years, and 6 years) over a five-year follow-up period using clinical notes and the Market Clarity database.
- Cost analysis (medical and pharmacy) was also performed during the follow-up.

Results

- A total of 25 DMD patients were identified: five aged 1-3 years, thirteen aged 4-5 years, and seven aged 6 years. In the first-year post-diagnosis, common symptoms included reduced mobility, Gower's sign, feeding difficulties, and respiratory infections. By the second year, arrhythmia and cardiomegaly appeared. In the third year, severity increased, with scoliosis, encephalopathy, and lordosis emerging. During years 4 and 5, symptoms worsened, including lordosis, kyphosis, contractures, and wheelchair dependence (**Figure 1**).
- The average per member per year all-cause cost (Medical + Pharmacy) for all age groups was determined. A statistically significant upward trend ($p < 0.01$) was observed in relation to the severity of complications (**Figure 2**).

Figure 1. Progression of Disease Symptoms and Complications Over Follow-up Years

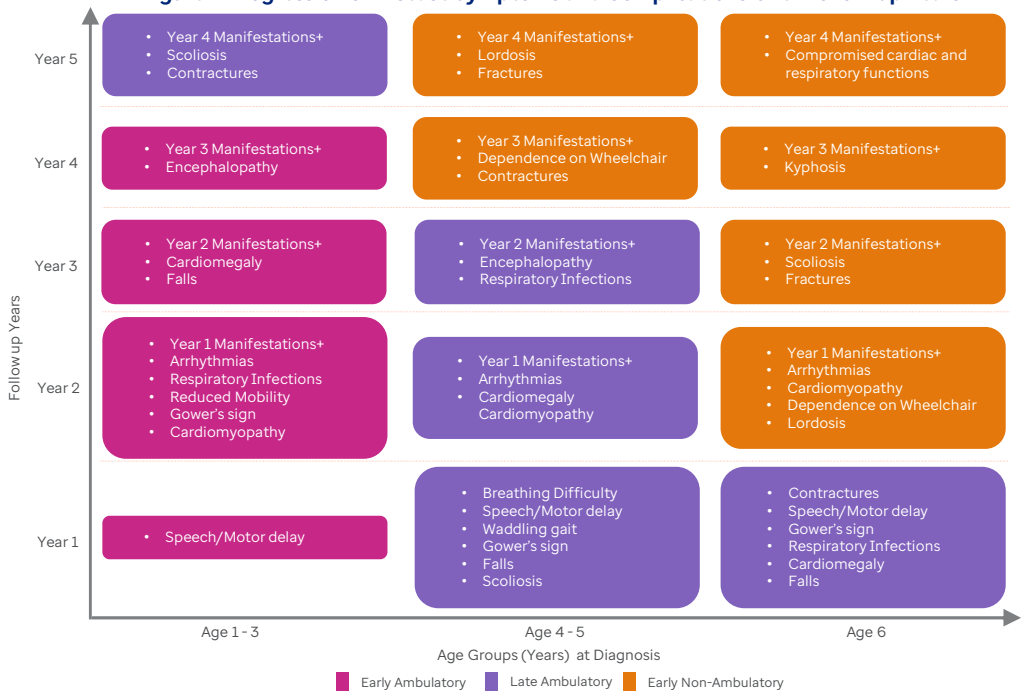
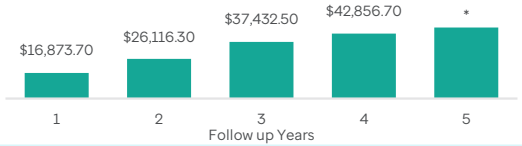


Figure 2. Annual Average All Cause Cost



* N too low to draw any inference

Conclusions

The findings of this study showed elevated resource utilization and complication patterns in DMD patients. The progression of the disease is slower at younger ages but accelerates with age, leading to severe complications such as loss of ambulation, scoliosis, lordosis, and kyphosis. These insights can help improve patient care and support the development of treatments for the progressive nature of DMD.

References: 1. Duan D, et al. Duchenne muscular dystrophy. Nat Rev Dis Primers. 2021 Feb 18;7(1):13.
2. Glanzman AM, Kusler A, Stuberg WA. Muscular Dystrophies and Spinal Muscular Atrophy. Campbell's Physical Therapy for Children Expert Consult-E-Book.2016 Dec 20:242. 2. Emery AE. Population frequencies of inherited neuromuscular diseases—a world survey. Neuromuscular disorders. 1991 Jan 1;1(1):19-29.
3. Shaw, M. L. (2023, October 25). Exon-skipping therapy for DMD linked to high care costs, health care resource utilization. The American Journal of Managed Care. Retrieved from https://www.ajmc.com/view/exon-skipping-therapy-for-dmd-linked-to-high-care-costs-health-care-resource-utilization

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