IDENTIFICATION OF DISEASE PROGRESSION STAGES IN PATIENTS WITH DUCHENNE MUSCULAR DYSTROPHY USING ADMINISTRATIVE CLAIMS DATA SUPPLEMENTED BY RELEVANT ELECTRONIC MEDICAL RECORDS (EMR) FIELDS

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INTRODUCTION

Duchenne muscular dystrophy (DMD) is a rare, X-linked, severely debilitating, and ultimately fatal neuromuscular disease characterised by progressive muscle weakness.[1] Loss of ambulation occurs at approximately 12 years.[2, 3] The median age of death with standard of care is 26–28 years,[4, 5] with the major causes of death being respiratory insufficiency and cardiomyopathy[1, 4]

The rarity of DMD has limited real-world study of disease progression in these patients.

OBJECTIVE

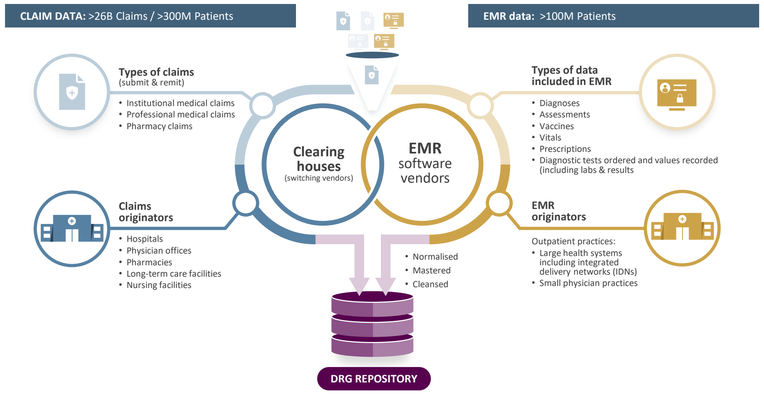
The objective of this study was to classify DMD patients by stage of progression using administrative claims data fields supported by relevant electronic medical records (EMR) fields.

METHODS

DATA

Data were drawn from an administrative claims and EMR dataset from Decision Resources Group, covering more than 300 million U.S. patients during 2011-2020. DMD patients were identified using EMR codes specific to DMD (SNOMED-CT). Clinical diagnosis and procedure markers reflecting disease progression and patient age were used as classification criteria.

Figure 1. DRG data repository



STAGE IDENTIFICATION

DMD progression was classified into 4 stages based on indicators of ambulatory and pulmonary ability as well as patient age, with expert clinical input: early ambulatory, late ambulatory, early non-ambulatory, and late non-ambulatory. Stage was assessed monthly for each patient based on observed stage classification markers and evaluated beginning at 12 months of data availability to ensure accurate classification. Identified stage was carried forward until markers of a later stage were observed. Progression logic was applied to ensure that patients could only progress to worse disease stages and not revert back to a better stage.

Figure 2. Identification of stage of progression based on age and observed signal markers in claims and EMR

STAGE 1 Early ambulatory	STAGE 2 Late ambulatory	STAGE 3 Early non-ambulatory	STAGE 4 Late non-ambulatory
1. Genetic test 2. Genetic counselling 3. Ankle-foot orthoses 4. Psychosocial management 5. Rehabilitation management	1. Ankle-foot orthoses 2. Manual wheelchair 3. Power assist wheelchair 4. Scooter 5. Cough assist device 6. Orthotic or prosthetic therapy 7. Steroid 8. Cardiac medication	1. Motorized wheelchair 2. Scoliosis 3. Orthopedic management 4. Rehabilitation management 5. Hospital bed 6. Pulmonary management 7. Cardiac medication 8. Bisphosphonate therapy	1. Tracheostomy 2. G-tube 3. Gastrointestinal management 4. Hospice and home health care 5. Pulmonary management 6. Assisted ventilation
Level 1: #1 or #2 or (#3 and age 0-8) **Level 2:** (#4 in conjunction with #5 and age 0-8) or (#5 and age 0-8) Early ambulatory is identified: • primarily by patients age and the absence of the markers characteristic of later stages • By observation of the markers listed above	**Level 1:** #1 or #2 or #3 or #4 **Level 2:** #5 or (#6 and age 9-13) **Pharma:** Combined use of #7 and #8	**Level 1:** (#1 + ≥2 #7) or #1 for >6 months or #2 **Level 2:** #3 or #4 or #5 if appears individually **Pharma:** (≥2 of #7) or (#7 and #8)	**Level 1:** #1 or #2 or #3 or #4 **Level 2:** (≥2 of #5) or #6

RESULTS

HEALTH STAGE DISTRIBUTION

94% of EMR-identified DMD patients had claims and markers allowing for stage classification.

Table 1. Health stage distribution 6th and 12th month from first observation among SNOMED patients

	DISTRIBUTION N (%)		AGE (MEAN)	
HEALTH STAGE	6th month	12th month	6th month	12th month
1	**383** (40.84%)	**362** (38.59%)	**7.17**	**7.44**
2	**269** (28.68%)	**243** (25.91%)	**13.15**	**13.10**
3	**206** (21.96%)	**214** (22.81%)	**18.57**	**18.05**
4	**80** (8.53%)	**119** (12.69%)	**22.73**	**23.06**
N (valid stage)	**938**	**938**	**938**	**938**
N (SNOMED)	**993**	**993**	**993**	**993**

SNOMED patients with at least one medical/pharma claim are included. The distribution is based on a "(k-1)" imputation assumption: if a patient's first observed valid health stage is k in month j, then he is assumed to be at stage (k-1) before month j (i.e., from the beginning of observation to month (j-1)).

Compared with a previously published age-based classification,[6] this method puts more patients in earlier stages. This may be reflective of improvements in standard of care treatment. It is also possible that because this method relies on positive observations of certain procedure and diagnosis codes to place patients at a certain stage, missing data could lead to a stage identification at a later age.

MAIN DRIVING IDENTIFICATION MARKERS BY STAGE

Driving Events by Stage

	STAGE 1 (N=83)	STAGE 2 (N=328)	STAGE 3 (N=575)	STAGE 4 (N=406)
Marker 1 (N, %)	Genetic testing **(27, 33%)**	Manual wheelchair **(165, 50%)**	Scoliosis **(193, 34%)**	Assisted ventilation **(225, 55%)**
Marker 2 (N, %)	Rehabilitation management **(26, 31%)**	Steroid and cardiac medication[2] **(78, 24%)**	Multiple cardiac medication[2] **(121, 21%)**	Pulmonary management **(104, 26%)**
Marker 3 (N, %)	Use of ankle-foot orthoses[1] **(20, 24%)**	Orthotic or prosthetic therapy[1] **(66, 20%)**	Motorized wheelchair[3] **(75, 13%)**	Tracheostomy **(99, 24%)**
Marker 4 (N, %)	Psychosocial management **(10, 12%)**	Cough-assist device **(22, 7%)**	Orthopedic management **(69, 12%)**	Gastrointestinal management **(58, 14%)**
Marker 5 (N, %)	Genetic counselling **(5, 6%)**	Use of ankle-foot orthoses[1] **(17, 7%)**	Rehabilitation management **(57, 10%)**	Gastric tube **(21, 5%)**
Marker 6 (N, %)		Scooter **(3, 1%)**	Hospital bed **(31, 5%)**	Hospice and home health care **(20, 5%)**
Marker 7 (N, %)		Power-assist wheelchair **(2, 1%)**	Pulmonary management **(28, 5%)**	
Marker 8 (N, %)			Cardiac medication and bisphosphonate therapy[2] **(24, 4%)**	
Marker 9 (N, %)			Cardiac medication and motorized wheelchair[2] **(12, 2%)**	

1. The criterion is joint with an age requirement; **2.** The criterion includes pharmaceutical codes; **3.** The criterion determines stage a set time after first instance

CONCLUSION

Classification of DMD disease progression within claims data provides insight into the real-world distribution of patients by stage and can enable future use of claims data to study additional topics such as disease burden by stage and rates of disease progression

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