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Real-World Insights Into Limb-Girdle Muscular Dystrophy, Including Subtype 2I/R9: Treatment Patterns, Health Care Resource Utilization, and Costs

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OBJECTIVE

- This study describes the clinical and economic burden of individuals with limb-girdle muscular dystrophy (LGMD) identified by ICD-10-CM code G71.038 versus matched controls without muscular dystrophy

BACKGROUND

- Limb-girdle muscular dystrophy type 2I/R9 (LGMD2I/R9) is a rare, progressive, and irreversible neuromuscular disorder caused by autosomal recessive *FKRP* variants leading to impaired α-dystroglycan glycosylation[1,2]
- LGMD2I/R9 is associated with progressive functional decline and cardiopulmonary involvement, suggesting a substantial and growing healthcare burden[3]
- From October 2022 to October 2025, ICD-10-CM code G71.038 ("other LGMD") was used in administrative claims to identify individuals with LGMD2I/R9 and related autosomal recessive subtypes
- There is limited real-world evidence describing HCRU and costs associated with LGMD
- Real-world characterization of HCRU and costs is needed to inform clinical management and future evaluation of emerging therapies

METHODS

Study Design

Non-interventional, Retrospective Study

Study Period
January 1, 2016 to September 30, 2024

Patient Identification Period
October 1, 2022 to September 30, 2024

Index Date
Main cohort: Earliest record of ICD-10-CM code G71.038 during patient identification period
Matched cohort: Random record during patient identification period

Lookback Period	Follow-Up Period
Time period of 365 days before index date	Time period from the index date (inclusive) to the earliest day of (1) death, (2) latest day of claims, disenrollment, or end of EHR activity, or (3) end of study period

Data source
- Optum Market Clarity® integrated EHR and administrative claims database
- Nationwide, de-identified, multi-payer data with longitudinal follow-up

Patient population

Inclusion criteria
- Main cohort (other LGMD)
  - ≥2 EHR and/or claims records with ICD-10-CM G71.038 on separate days during the identification period
  - Linked EHR and claims data available
- Matched cohort (no MD)
  - ≥2 EHR and/or claims records for any reason during the identification period
  - Linked EHR and claims data available

Exclusion criteria
- Main cohort
  - Other LGMD subtype ICD-10-CM codes (eg, G71.031, G71.032, G71.033, G71.0340, G71.0341, G71.0342, G71.0349, G71.035, G71.09, G71.00)
  - Duchenne or Becker muscular dystrophy (G71.01)
  - Walker-Warburg syndrome (Q04.3)
  - Other congenital myopathy (G71.29)
  - G71.039 (other specified LGMD, unspecified subtype) after index date
- Matched cohort
  - Any muscular dystrophy diagnosis (G71.0X)
  - Walker-Warburg syndrome (Q04.3)
  - Other congenital myopathy (G71.29)

Outcomes of interest
- Demographic and clinical characteristics
- Treatment patterns
- HCRU (IP admissions, ED visits, OP visits, OP pharmacy claims)
- Medical costs

Statistical analysis
- Descriptive analyses
- 10:1 control cohort matched on age, sex, index month/year, geographic region, and payer type
- HCRU/costs as PPPM (95% CI)

RESULTS

- Of the 521 patients with other LGMD, 375 were eligible for HCRU/cost analyses (median follow-up, 10.6 months); the matched cohort included 5210 patients in the full cohort and 4093 in the HCRU cohort (patients with continuous medical and pharmacy enrollment in claims data for ≥1 day before and after the index date; median follow-up, 9.4 months)
- Mean age was 53.6 years; 68.5% were female **(Table 1)**
- Most patients were White (77.5%) and covered by Medicare (31.5%) or commercial insurance (39.0%) **(Table 1)**
- During follow-up, cardiac (29.0%) and pulmonary (39.2%) involvement exceeded that observed in controls (16.5% and 21.9%, respectively) **(Table 2)**

Table 1. Demographic characteristics of LGMD cohort

Characteristic	Patients with "other LGMD" (N=521)
Age at index[a], mean (SD)	53.6 (22.1)
Sex, n (%)	
Male	164 (31.5)
Female	357 (68.5)
Race, n (%)	
White	404 (77.5)
African American	34 (6.5)
Asian	13 (2.5)
Other/Unknown	70 (13.4)
Ethnicity, n (%)	
Hispanic	40 (7.7)
Non-Hispanic	408 (78.3)
Unknown	73 (14.0)
Geographic region	
Northeast	163 (31.3)
Midwest	230 (44.1)
South	49 (9.4)
West	56 (10.7)
Other/Unknown	23 (4.4)
Insurance payer, n (%)	
Commercial	203 (39.0)
Medicaid	55 (10.6)
Medicare	164 (31.5)
Multiple payers	9 (1.7)
Uninsured	<5[b]
Missing	89 (17.1)

[a]Some (<5) patients had an age of 0 years since "year of index date – birth year" was used to calculate age at index, and these patients have the same year for birth year and index year.
[b]<5: Counts of less than 5 were masked.

Table 2. Clinical characteristics of LGMD cohort

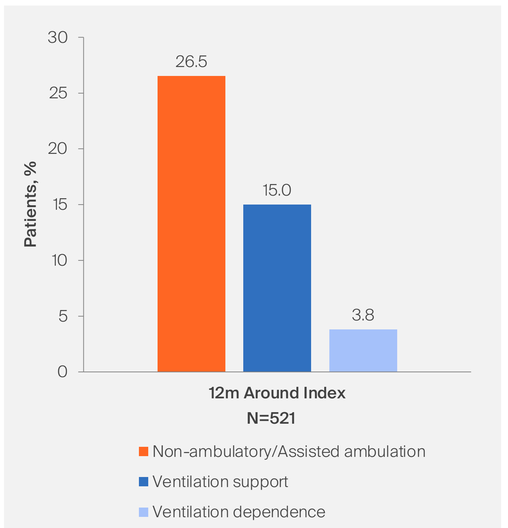
Characteristic, n (%)	LGMD cohort–follow-up (N=521)	Matched cohort–follow-up (N=5210)
Musculoskeletal abnormalities	19 (3.6)	–
Contractures	6 (1.2)	–
Exaggerated lordosis	<5[b]	–
Pectus excavatum	<5[b]	–
Scoliosis	11 (2.1)	–
Cardiac involvement	151 (29.0)	862 (16.5)
Arrhythmia	119 (22.8)	710 (13.6)
Cardiac fibrosis	0 (0.0)	0 (0.0)
Cardiac transplantation	<5[b]	0 (0.0)
Cardiomyopathy	25 (4.8)	80 (1.5)
Heart failure	58 (11.1)	303 (5.8)
Left ventricular hypertrophy	26 (5.0)	117 (2.2)
Myocardial fibrosis	0 (0.0)	0 (0.0)
Pacemaker	7 (1.3)	76 (1.5)
Pulmonary conditions[a]	204 (39.2)	1143 (21.9)
Dyspnea	129 (24.8)	491 (9.4)
Impaired regulation of breathing	<5[b]	16 (0.3)
Impaired regulation of breathing (hypoventilation)	13 (2.5)	45 (0.9)
Sleep disordered breathing	69 (13.2)	469 (9.0)
Sleep disordered breathing (hypoventilation)	<5[b]	19 (0.4)
Inefficient cough	70 (13.4)	395 (7.6)
Restrictive pulmonary function	21 (4.0)	64 (1.2)

[a]All pulumonary conditions listed are mutually exclusive.
[b]<5: Counts of less than 5 were masked.

RESULTS (cont'd)

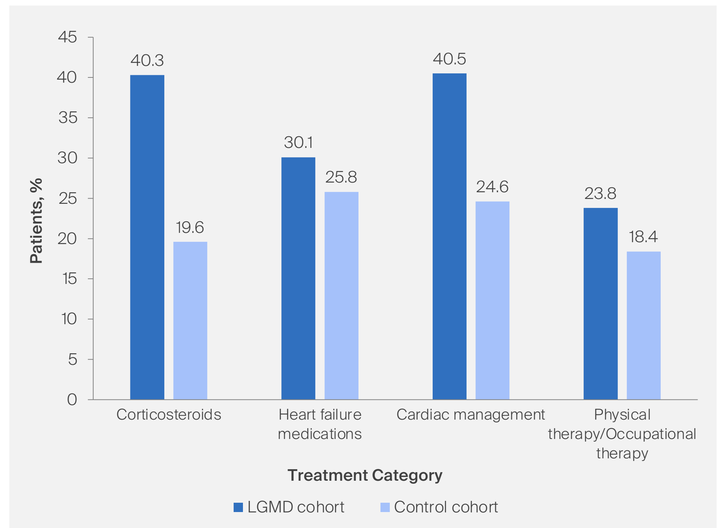
- In the 6 months before and after index, 26.5% of patients with LGMD were non-ambulatory/had assisted ambulation, 15.0% had ventilation support, and 3.8% had ventilation dependence **(Figure 1)**
- During follow-up, 40.3% of patients with LGMD received corticosteroids and 30.1% received heart failure medications, exceeding utilization among matched controls (19.6% and 25.8%, respectively) **(Figure 2)**

Figure 1. Functional and respiratory status during the 12 months around index[a]



[a]12 months around index defined as 6 months prior to index through 6 months following index.

Figure 2. Pharmacologic and supportive care utilization during follow-up



- In the HCRU cohort, 13.3% of patients with LGMD had ≥1 IP stay, 29.6% had ≥1 ED visit, and 98.4% had ≥1 OP visit, compared with 6.6%, 16.4%, and 95.7% of matched controls, respectively **(Figure 3)**
- In the LGMD cohort, IP and ED utilization was substantially higher in individuals who were non-ambulatory or required ambulation assistance (IP: 27.0%; ED: 48.0%) versus those who were ambulatory (IP: 8.4%; ED: 22.9%) **(Figure 4)**

Figure 3. All-cause HCRU during follow-up

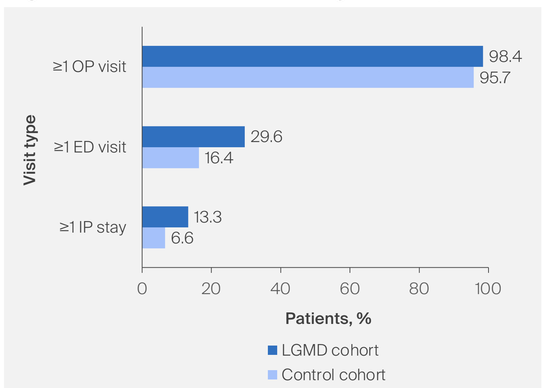
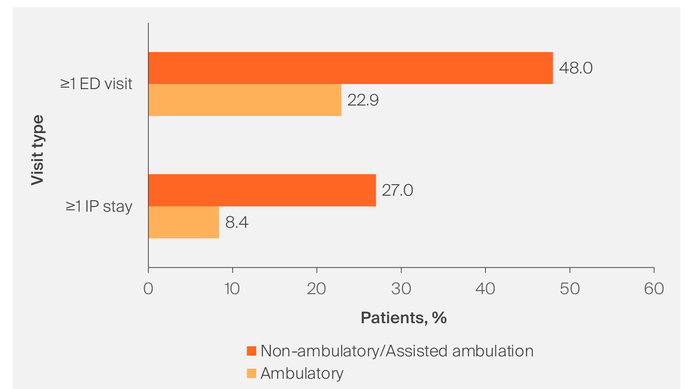
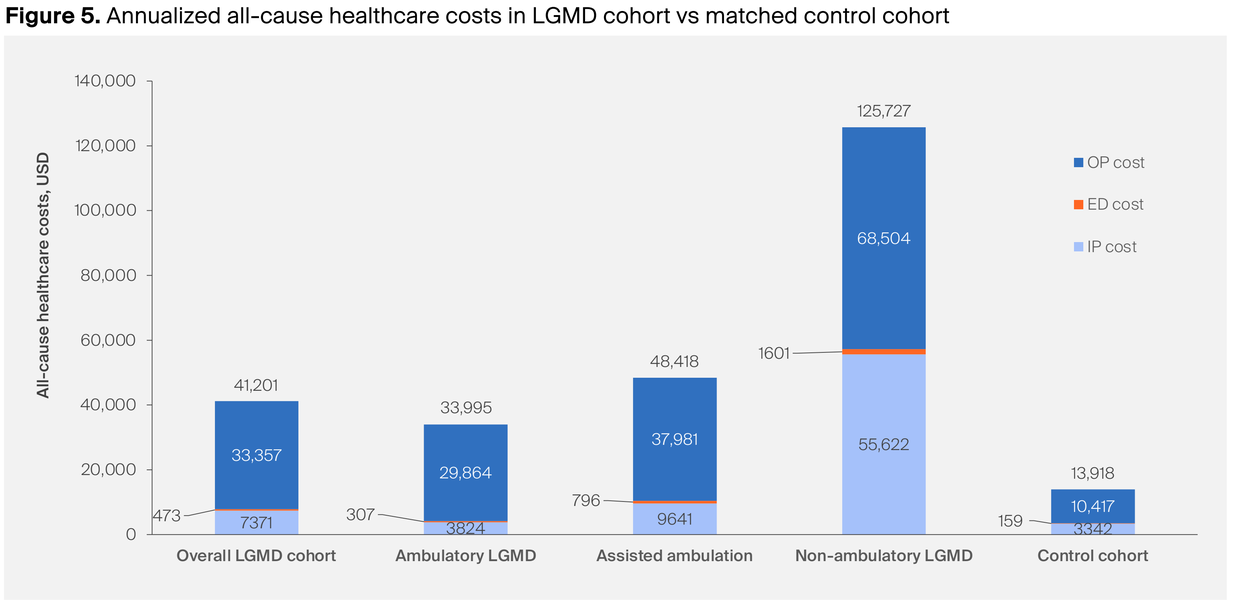


Figure 4. IP and ED utilization by ambulatory status, LGMD cohort



- Individuals in the LGMD cohort incurred approximately three-fold higher annualized all-cause healthcare costs than those in the control cohort ($41,201 vs $13,918), largely attributable to markedly high OP costs **(Figure 5)**
- Costs increased with functional impairment, with higher costs among assisted ambulation ($48,418) and non-ambulatory individuals ($125,727) versus those who were ambulatory ($33,995) **(Figure 5)**

Figure 5. Annualized all-cause healthcare costs in LGMD cohort vs matched control cohort



CONCLUSIONS

- Individuals living with LGMD2I/R9 and related subtypes experience considerable clinical burden and high HCRU and costs, with consistently higher comorbidity and medical costs than matched controls
- The newly released ICD-10-CM code (G71.036: limb girdle muscular dystrophy due to fukutin related protein dysfunction) for LGMD2I/R9 will enhance research and characterization of this population
- These findings underscore substantial unmet need and highlight the importance of advancing novel therapies and optimizing care strategies for individuals living with LGMD

LIMITATIONS

- Claims- and EHR-based analyses are subject to medical coding errors and incomplete capture of clinical details
- Use of the non-specific ICD-10-CM code for "other LGMD" may result in disease misclassification

STRENGTHS

- Use of a large, nationally representative linked EHR claims database with matched controls enables comprehensive real-world assessment of treatment, HCRU, and costs

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ABBREVIATIONS: CI, confidence interval; ED, emergency department; EHR, electronic health record; HCRU, healthcare resource utilization; ICD-10-CM, International Classification of Diseases, Tenth Revision, Clinical Modification; IP, inpatient; LGMD, limb-girdle muscular dystrophy; LGMD2I/R9, limb-girdle muscular dystrophy type 2I/R9; MD, muscular dystrophy; OP, outpatient; PPPM, per-patient-per-month; SD, standard deviation; USD, US dollars

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DISCLOSURES: K Bartley is a full-time employee and shareholder of BridgeBio Pharma, Inc., and a shareholder of Roche. **A Howarth, S Zhu, and F Tang** are full-time employees of Genesis Research Group. **A Talaga** is a full-time employee and shareholder of BridgeBio Pharma, Inc. **JM Statland** received grant funding from ALSA, CDC, Friends of FSH Research, FSHD Canada, FSHD Society, MDA, NCATS, and NINDS; consulted or served on scientific advisory boards for Alnylam, Armatus, Avidity, Dyne, Kate Therapeutics, Merck, Novartis, RegenXBio, Roche, Sarepta, Scholar Rock, and Vertex; and received stock options from Dyne.

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