Real-World Demographic and Treatment Characteristics of Patients with Congenital Myasthenic Syndrome in the United States

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Introduction

- Congenital Myasthenic Syndromes (CMS) are a group of rare, inherited neuromuscular disorders caused by mutations in genes essential for neuromuscular junction structure and function.[1]
- CMS is estimated to affect between 1 in 45,000 and 1 in 555,000 people globally.[2]
- CMS are characterized by fatigability or persistent muscle weakness affecting ocular, bulbar, axial, respiratory, and/or limb muscles, with substantial phenotypic heterogeneity which can lead to potentially life-threatening respiratory insufficiency.[2,4]
- CMS generally appears during infancy or childhood, though the age of onset, intensity of symptoms, and progression of the condition can differ significantly depending on the patient and genetic subtype; despite early symptom onset (often <3 years), diagnosis is frequently delayed by several years or until adulthood.[3]
- CMS imposes a substantial lifelong burden driven by functional impairment, dependence on supportive care, caregiver impact, and sustained healthcare utilization.[5,6]
- Despite increasing recognition of CMS, real-world evidence describing patient characteristics and treatment utilization remains limited, particularly in the United States (U.S.).
- Given the rarity and heterogeneity of CMS, large-scale real-world claims data offer an important opportunity to characterize patient demographics, treatment utilization, and disease burden, informing care pathways, future research and therapeutic development.

Objective

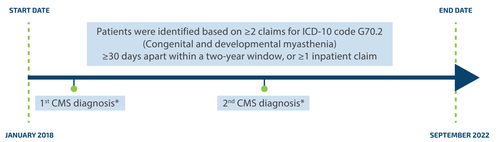
- The objective of the current study was to provide a real-world assessment of patient characteristics among individuals with CMS in the U.S, including demographic characteristics, insurance coverage and treatment utilization.

Method

Study design

- A retrospective analysis was conducted using Komodo Health's Prism platform, which records longitudinal patient journeys across the U.S. **(Figure 1).**
  - The platform integrates both open and closed medical and pharmacy claims data to enable real-world assessment of rare disease populations.
- Patients were identified based on ≥2 claims for ICD-10 code G70.2 (Congenital and developmental myasthenia) ≥30 days apart within a two-year window, or ≥1 inpatient claim
  - The patient identification period was defined as January 1, 2018 to September 30, 2022, to ensure all patients have at least 2 years of lookforward and lookback.
  - Earliest claim satisfying this during the selection period i.e 2018 -2022 was chosen as index date.

Figure 1: Study design



*Patients must have ≥2 recorded CMS diagnosis during 2018-2022
CMS, Congenital Myasthenic Syndromes

Inclusion criteria

- Patients with ≥2 outpatient or professional claims with the ICD-10 diagnosis code G70.2 recorded ≥30 days apart but no more than 2 years apart, to reduce the likelihood of rule-out or miscoded diagnoses

Or

- Patients with ≥1 inpatient hospitalization claim with a G70.2 diagnosis, reflecting a higher likelihood of confirmed disease.

Exclusion criteria

- Patients with a prior CMS diagnosis before 2018 (i.e., evidence of an ICD-10 G70.2 claim before the index date) were excluded to identify newly diagnosed patients in claims data and to characterize treatment utilization from disease onset.
- Patient with ≥2 claims for differential diagnoses (myasthenia gravis [MG], Lambert–Eaton myasthenic syndrome [LEMS], muscular atrophy, muscle wasting and atrophy, congenital myopathies, congenital myotonic dystrophy, Fetal Akinesia Deformation Sequence [FADS], muscular dystrophy, hereditary neuropathies, Congenital Disorders of Glycosylation [CDG], moebius syndrome and mitochondrial disorders) were excluded to increase confidence in the CMS cohort and reduce potential misdiagnosis.

Outcomes

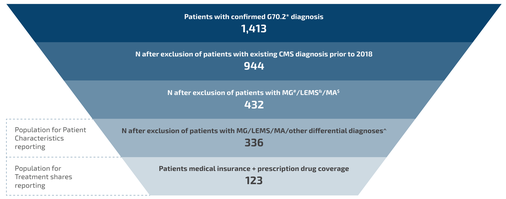
- Descriptive analyses were performed to characterize the study population, including assessments of age distribution, gender, insurance payer type, and patterns of treatment and supportive care utilization.

Results

Patient disposition

- The detailed patient disposition is shown in **Figure 2.**
- A total of 1413 patients with a confirmed ICD-10 diagnosis of G70.2 (congenital and developmental myasthenia) were initially identified.
- After excluding patients with prior evidence of CMS diagnosis (G70.2), 944 patients remained for further evaluation.
- After removing patients with autoimmune disorders of the neuromuscular junction (MG, LEMS), genetic motor neuron disorders (spinal muscular atrophy) resulted in a reduced cohort of 432 patients.
- Finally, 336 patients were included in the study, after removing patients with and other related neuromuscular conditions (differential diagnosis) and of which 123 patients had medical and pharmacy capture available.

Figure 2: Patient identification funnel



*G70.2: Congenital and developmental Myasthenia
#Myasthenia gravis/transient neonatal myasthenia gravis
&lambert eaton myasthenic syndrome
$Spinal muscular atrophy/"Muscle wasting and atrophy, not elsewhere classified"
^other differential diagnoses: congenital myopathies, congenital myotonic dystrophy, FADS, muscular dystrophy, hereditary neuropathies, CDG, moebius syndrome and mitochondrial disorders

Patient characteristics

Cohort size

- Among 336 patients included in the study, 212 were pediatric (63%) and 124 were adults (37%) patients.

Age

- Pediatric cohort consisted mainly patients aged 0–12 years (79%), followed by aged 13–17 years (21%).
- Adults were mainly between 18–44 (41%) years, 37% were of age ≥65 years and 22% aged 45–64 years.

Gender

- Gender was almost equally distributed in overall population (~50% male / 50% female).
- Among pediatric patients, 53% were male, while among adult patients, 43% were male.

Race

- In overall cohort, White patients were the most common (36% overall), followed by Hispanic/Latino and Black patients.
  - White patients represented over one-third of the overall cohort (48% adults, 28% pediatrics).
  - Hispanic/Latino representation was 11% overall cohort.
  - Black and Asian patients each accounted for a small proportion of the cohort.
  - A substantial proportion of patients had unavailable data for their race/ethnicity (43% overall).

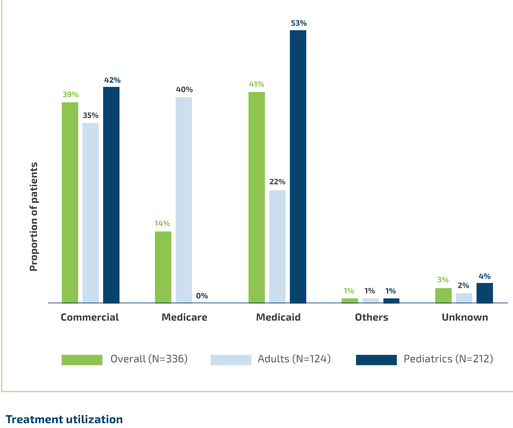
Table 1: Patient characteristics

Characteristic	Overall (N =336) N (%)	Adults (N=124) N (%)	Pediatrics (N=212) N (%)
Age Distribution			
0-12	168 (50%)	0	168 (79%)
13-17	44 (13%)	0	44 (21%)
18-44	51 (15%)	51 (41%)	0
45-64	28 (8%)	28 (22%)	0
65+	45 (14%)	45 (37%)	0
Gender			
Male	167 (49.7%)	54 (44%)	113 (53%)
Female	166 (49.5%)	68 (55%)	98 (46%)
Unknown	<11	<11	<11
Race			
White	120 (36%)	60 (48%)	60 (28%)
Hispanic or Latino	37 (11%)	14 (11%)	23 (11%)
Black	19 (6%)	<11	11 (5%)
Asian	<11	<11	<11
Other	<11	<11	<11
Unknown	143 (43%)	36 (29%)	107 (50%)

Insurance coverage

- Medicaid was the most common payer (41%), followed by commercial (39%) and Medicare (14%) **(Figure 3)**.
- Insurance coverage differed by age group.
  - Among pediatric patients, Medicaid was the most common payer (53%), followed by commercial insurance (42%). No Medicare insurance was reported.
  - Among adults, coverage was distributed across Medicare (40%), commercial insurance (35%), and Medicaid (22%).

Figure 3: Insurance coverage among patients with CMS



Treatment utilization

- Treatment and service utilization patterns differed between adults and pediatric patients over time **(Table 2)**.
- Use of cholinergic agonists including pyridostigmine was low but comparable at baseline between adults and pediatric patients but increased post index to 25% among pediatric patients at 1st year and 2nd year.
- Use of salbutamol (albuterol) was higher in pediatric patients than adults across all periods.
  - Albuterol was the most used medication, used by 37% of pediatrics and 27% of adult patients within a year of diagnosis.
- Utilization of occupational or physical therapy services was substantially higher among pediatric patients at all time points, increasing from 57% pre index to 71% within a year of diagnosis while only among adult patients use of occupational or physical therapy increased from 12% pre-index to 22% within a year of diagnosis
- Wheelchair/walker use remained low overall but was more common in pediatric patients than adults across periods (<15%).

Table 2: Treatment utilization among patients with CMS

Characteristic	Pre index 1 year		Post Index 1 year		Post Index 2nd year (365th-730th day)	
	Adults (N = 58) N (%)	Pediatric (N = 65) N (%)	Adults (N = 58) N (%)	Pediatric (N = 65) N (%)	Adults (N = 58) N (%)	Pediatric (N = 65) N (%)
Cholinergic agonists	<11	11	≤11	16 (25%)	<11	16 (25%)
Pyridostigmine	<11	<11	≤11	16 (25%)	<11	16 (25%)
Amifampridine/ 3,4-Diaminopyridine (3,4-DAP)	0	0	0	0	<11	0
Neostigmine	0	0	0	0	0	0
Adrenergic Agonists	≤11	21 (32%)	16 (27%)	24 (37%)	13 (22%)	21 (32%)
Salbutamol (albuterol)	≤11	21 (32%)	16 (27%)	24 (37%)	12 (21%)	21 (32%)
Ephedrine	0	0	0	0	<11	0
Open Channel Blockers	<11	<11	<11	<11	<11	<11
Quinidine	<11	0	<11	0	0	0
Fluoxetine	<11	<11	<11	<11	<11	<11
Occupational or physical therapy Services*	<11	37 (57%)	13 (22%)	46 (71%)	12 (21%)	47 (72%)
Wheelchair/walker use*	<11	<11	<11	<11	<11	<11

*Identification was based on procedure codes, with durable medical equipment (DME) captures defined using Healthcare Common Procedure Coding System (HCPCS) codes

Limitations

- CMS identification relied on ICD-10 coding (G70.2), which may lead to misclassification or under-ascertainment, particularly given diagnostic heterogeneity and frequent misdiagnosis of CMS as other neuromuscular conditions.
- Genotype, clinical severity, functional status, and confirmatory diagnostic information (e.g., genetic testing or electrophysiology) were not available in claims data, limiting clinical granularity and subtype-specific interpretation.
- Treatment utilization and supportive care may be underestimated or incompletely captured, particularly for services paid out-of-pocket or delivered outside reimbursed settings.
- The descriptive design and relatively small identified population precluded causal inference or detailed subgroup analyses.

Conclusions

- This real-world claims analysis provides one of the early U.S. characterizations of patients with CMS, confirming that CMS is an ultra rare condition predominantly affecting pediatric patients.
- The cohort demonstrated heterogeneity in age, insurance coverage, and treatment utilization, with clear differences between pediatric and adult patients.
- High utilization of occupational and physical therapy services particularly among pediatric patients suggests a substantial functional burden and ongoing supportive care needs among patients with CMS.
- Treatment patterns were broadly consistent with current clinical practice, with frequent use of adrenergic agonists such as albuterol and lower use of cholinergic agents and open channel blockers.
- Overall, these findings emphasize the complexity of CMS management and the value of real-world data in understanding care patterns in a population where prospective evidence remains limited.
- **Future insights:** Further research using complementary data sources and longitudinal designs is needed to better characterize the patient journey, clinical outcomes, and overall disease burden, and to improve case ascertainment in real-world datasets.

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Disclosures

Syed Raza, Rebecca Shilling and deMauri Mackie are employees of argenx. Amit Goyal, Charlotte E. Ward and Namith Dharani are employees of ZS Associates and serve as paid consultants for argenx.

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