Limb Girdle Muscular Dystrophies

There are several major types and dozens of sub-types of muscular dystrophy.[1] The five most common types of muscular dystrophy are: Becker, Duchenne, Facioscapulohumeral (FSH), Myotonic, and Limb girdle.[2] Myotonic (G71.11), Becker and Duchenne (G71.01), and FSH (G71.02) muscular dystrophies all have specific ICD-10-CM codes. We propose adding codes for limb girdle muscular dystrophy (LGMD) and selected LGMD subtypes.

Limb girdle muscular dystrophies are a group of genetically inherited conditions that primarily affect proximal skeletal muscle leading to loss of muscle fibers and progressive, predominantly proximal muscle weakness.[3] To be considered an LGMD, the condition must be described in at least two unrelated families, individuals must demonstrate degenerative changes on muscle imaging over the course of the disease, and have dystrophic changes on muscle histology, ultimately leading to end-stage pathology for the most affected muscles. Most affected individuals achieve independent walking, and most individuals have an elevated serum creatine kinase activity.[4]

There are currently 34 identified subtypes of LGMD, each with a unique genetic cause.[5] While clinical presentations can be similar (thus explaining the initial grouping) these differing genetic causes result in varying presentations and have variation in pathophysiology. Rather than proposing ICD-10-CM codes for all 34 subtypes, we are proposing codes for a combination of the most prevalent LGMDs and those with advanced clinical therapeutic programs that could result in an FDA-approved treatment within five years. We propose subcodes for the six most prevalent LGMD subtypes (the autosomal recessive LGMDs caused by mutations in the genes that code for the proteins calpain-3, dysferlin, alpha-sarcoglycan, fukutin related protein, anoctamin5, and collagen-VI), as well as the autosomal dominant forms of calpain-3 and collagen-VI related LGMD to avoid coding confusion. We further propose subcodes for LGMD subtypes caused by beta-sarcoglycan and gamma-sarcoglycan dysfunctions due to advanced clinical therapeutic programs (most of the previously-mentioned most prevalent LGMDs also have ongoing clinical therapeutic programs).[6] In total, these ten new subcodes for individual LGMD subtypes will represent over half of the LGMD community.[7] We also propose an "other genetically confirmed limb girdle muscular dystrophy subtype" code for all individuals

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[1] Mercuri E, Muntoni F. Muscular Dystrophies. Lancet 2013; 381(9869):845-60. http://dx.doi.org/10.1016/S0140-6736(12)61897-2

[2] Theadom A, Rodrigues M, Roxburgh R, Balalla S, Higgins C, Bhattacharjee R, Jones K, Krishnamurthi R, Feigin V: Prevalence of Muscular Dystrophies: A Systematic Literature Review. Neuroepidemiology 2014;43:259-268. doi: 10.1159/000369343

[3] Limb-Girdle Muscular Dystrophy (LGMD). Muscular Dystrophy Association. Retrieved December 4, 2020 from https://www.mda.org/disease/limb-girdle-muscular-dystrophy

[4] Volker Straub, Alexander Murphy, Bjarne Udd, on behalf of the LGMD workshop study group. 229th ENMC international workshop: Limb girdle muscular dystrophies – Nomenclature and reformed classification Naarden, the Netherlands, 17–19 March 2017. Neuromuscular Disorders 28, issue 8, P702-710 (2018) https://doi.org/10.1016/j.nmd.2018.05.007

[5] Ibid.

[6] Building an industry-leading genetic medicine pipeline. Sarepta Therapeutics. Retrieved December 4, 2020 from https://www.sarepta.com/products-pipeline/pipeline

[7] Liu, W., Pajusalu, S., Lake, N.J. et al. Estimating prevalence for limb-girdle muscular dystrophy based on public sequencing databases. Genet Med 21, 2512–2520 (2019). https://doi.org/10.1038/s41436-019-0544-8

genetically diagnosed with an LGMD subtype that does not have a specific ICD-10-CM code, and a “limb girdle muscular dystrophy, unspecified” code for any individual diagnosed with an LGMD but the subtype has not been genetically confirmed (this diagnostic journey happens frequently for those with LGMD). The remaining currently recognized 24 LGMD subtypes can be considered for unique ICD-10-CM codes at a later date as therapeutic development advances.

Similar to the rationale used to create ICD-10-CM codes for Duchenne, Becker, and FSH muscular dystrophies, creating specific codes for the LGMDs will provide more accurate diagnoses; increase access to targeted care management and treatment; and inform patient decision making on clinical trials and resources for subtype-specific patient communities. Specific codes will facilitate the surveillance of these diseases; will allow more accurate estimates of their incidence, prevalence, survivorship, mortality and its causes, injuries, symptoms, and health visits; will help to identify factors that influence health status and secondary conditions, and will facilitate targeted therapeutic development and treatment at the LGMD subtype level. On a larger scale, ICD-10-CM codes can be used to compare health information across hospitals, regions, clinical settings, countries, and even across time in a given location and to facilitate the evaluation of clinical guidelines.

This proposal is submitted on behalf of a coalition of LGMD patient advocacy organizations and LGMD clinical experts, and reflects the input of clinicians, researchers, biopharmaceutical companies, physical therapists, coding experts, and other medical professionals familiar with LGMD.

TABULAR MODIFICATIONS

G71 Primary disorders of muscles

Excludes: arthrogryposis multiplex congenita (Q74.3)

metabolic disorders (E70-E88)

myositis (M60.-)

G71.0 Muscular dystrophy

G71.01 Duchenne or Becker muscular dystrophy

Autosomal recessive, childhood type, muscular dystrophy resembling

Benign [Becker] muscular dystrophy

Severe [Duchenne] muscular dystrophy

G71.02 Facioscapulohumeral muscular dystrophy

Scapulohumeral muscular dystrophy

New subcategory G71.03 Limb girdle muscular dystrophies

New sub-subcategory G71.030 Limb girdle muscular dystrophy due to calpain-3

		dysfunction
New code	G71.0300	Autosomal dominant limb girdle muscular dystrophy due to calpain-3 dysfunction
Add		Limb girdle muscular dystrophy type 1i
Add		LGMD D4 calpain-3-related
New code	G71.0301	Autosomal recessive limb girdle muscular dystrophy due to calpain-3 dysfunction
Add		Limb girdle muscular dystrophy type 2A
Add		LGMD R1 calpain-3-related
Add		Primary calpainopathy
New code	G71.031	Limb girdle muscular dystrophy due to dysferlin dysfunction
Add		Autosomal recessive limb girdle muscular dystrophy type 2B
Add		LGMD R2 dysferlin-related
Add		Dysferlinopathy
Add		Miyoshi Myopathy type 1
New code	G71.032	Limb girdle muscular dystrophy due to alpha-sarcoglycan dysfunction
Add		Autosomal recessive limb girdle muscular dystrophy type 2D
Add		LGMD R3 α-sarcoglycan-related
Add		Alpha-sarcoglycanopathy
New code	G71.033	Limb girdle muscular dystrophy due to beta-sarcoglycan dysfunction
Add		Autosomal recessive limb girdle muscular dystrophy type 2E
Add		LGMD R4 β-sarcoglycan-related
Add		Beta-sarcoglycanopathy
New code	G71.034	Limb girdle muscular dystrophy due to gamma-

			sarcoglycan dysfunction
Add			Autosomal recessive limb girdle muscular dystrophy type 2C
Add			LGMD R5 γ-sarcoglycan-related
Add			Gamma-sarcoglycanopathy
New code	G71.035		Limb girdle muscular dystrophy due to fukutin related protein dysfunction
Add			Autosomal recessive limb girdle muscular dystrophy type 2I
Add			LGMD R9 FKRP-related
New code	G71.036		Limb girdle muscular dystrophy due to anoctamin5 dysfunction
Add			Autosomal recessive limb girdle muscular dystrophy type 2L
Add			LGMD R12 anoctamin5-related
Add			Anoctaminopathy
Add			Miyoshi Myopathy type 3
New sub-subcategory	G71.037		Limb girdle muscular dystrophy due to collagen-VI dysfunction
New code		G71.0370	Autosomal dominant limb girdle muscular dystrophy due to collagen-VI dysfunction
Add			LGMD D5 collagen VI-related
Add			Bethlem myopathy dominant
New code		G71.0371	Autosomal recessive limb girdle muscular dystrophy due to collagen-VI dysfunction
Add			LGMD R22 collagen VI-related
Add			Bethlem myopathy recessive
New code	G71.038		Other genetically confirmed limb girdle muscular dystrophy subtype
Add			Other genetically confirmed LGMD subtype

New code	G71.039	Limb girdle muscular dystrophy, unspecified
Add		LGMD, unspecified
	G71.08	Other specified muscular dystrophies
		Benign scapuloperoneal muscular dystrophy with early contractures
		[Emery-Dreifuss]
		Congenital muscular dystrophy NOS
		Congenital muscular dystrophy with specific morphological abnormalities of the muscle fiber
		Distal muscular dystrophy
Delete		~~Limb-girdle muscular dystrophy~~
		Ocular muscular dystrophy
		Oculopharyngeal muscular dystrophy
		Scapuloperoneal muscular dystrophy
	G71.09	Muscular dystrophy, unspecified