SUPPLEMENTARY MATERIALS

**Supplementary Table 1. Scope of the literature review**

|  |  |
| --- | --- |
| **Population** | Individuals with LGMDR |
| **Exposures/Comparators** | Subgroups:   * Subtype/genotype   + Sarcoglycanopathies (LGMDR3-6)   + Dysferlinopathies (LGMDR2/MM)   + Calpainopathy (LGMDR1)   + FKRP-related dystroglycanopathy (LGMDR9)   + Anoctaminoapthy (LGMDR12) |
| **Outcomes** | Clinical measures:   * Serum creatinine kinase levels * MRI * ECG * Echocardiogram   Functional measures:   * Myometry measures * Muscle strength and functional testing * 6MWD * MMT * Brooke scale * AMS * Pulmonary function (e.g. FVC, FEV1) * ADL (e.g. Barthel Index) * Pain and fatigue (e.g. VAS) * General or LGMD-specific health-related quality of life (HRQoL) measures (e.g. Short Form-36, ACTIVLIM)   Longer-term outcomes, including but not limited to:   * LGMD disease progression by serial functional measures over time * Key clinical outcomes (e.g. need for wheelchair, need for ventilation, ambulation difficulties, heart failure, respiratory failure) * Mortality |
| **Study Design** | Epidemiologic (prospective or retrospective) studies  Randomized controlled trials  Case series (n>5)  Case reports |

**Abbreviations:** 6MWD, 6-minute walk distance; ADL, activities of daily living; AMS, average muscle score; ECG, electrocardiograph; FEV1, forced expiratory volume in 1 second; FKRP, fukutin-related protein; FVC, forced vital capacity; HRQoL, health-related quality of life; LGMDR, autosomal recessive limb girdle muscular dystrophy; MM, Miyoshi myopathy; MMT, manual muscle testing; MRI, magnetic resonance imaging; VAS, Visual Analog Scale.

**Supplementary Table 2. Characteristics of studies contributing patient data to the analysis**

| **Author, year** | **Study design** | **Country / Region** | **Subtypes** | **Number of patients** | **Study period / duration** |
| --- | --- | --- | --- | --- | --- |
| Aharoni et al, 2011 [1] | Case report | Israel | LGMDR2 | 1 | NR |
| Akarsu et al, 2018 [2] | Case report | Turkey | LGMDR12 | 1 | NR |
| Alavi et al, 2017 [3] | Retrospective | Iran | LGMDR3-6 | 25 | 2010-2015 |
| Algahtani et al, 2018 [4] | Case report | Saudi Arabia | LGMDR2 | 1 | NR |
| Alhamidi et al, 2017 [5] | Retrospective | Norway | LGMDR9 | 25 | NR |
| Al-Harbi et al, 2016 [6] | Case report | Saudi Arabia | LGMDR3 | 1 | NR |
| Angelini et al, 1998 [7] | Case report | Italy | LGMDR5 | 2 | NR |
| Angelini et al, 2010 [8] | Retrospective | Italy | LGMDR1, LGMDR2/MM | 43 | NR |
| Angelini et al, 2011 [9] | Case report | Italy | LGMDR2/MM | 2 | NR |
| Aoki et al, 2001 [10] | Retrospective | USA | MM | 24 | NR |
| Argov et al, 2000 [11] | Retrospective | Libya | LGMDR2 | 29 | 1997-98 |
| Arrigoni et al, 2018 [12] | Prospective | Italy | LGMDR1, LGMDR2 | 20 | NR |
| Bandyopadhyay et al, 2018 [13] | Case report | USA | LGMDR2 | 1 | NR |
| Barresi et al, 2000 [14] | Case report | Italy | LGMDR4 | 2 | NR |
| Baumeister et al, 2009 [15] | Case report | Germany | LGMDR5 | 1 | NR |
| Bhatt et al, 2019 [16] | Case report | India | LGMDR1 | 1 | NR |
| Bird et al, 2008 [17] | Case report | USA | LGMDR1 | 1 | NR |
| Blackburn et al, 2017 [18] | Case report | USA | LGMDR12 | 1 | NR |
| Boito et al, 2005 [19] | Retrospective | Italy | LGMDR9 | 20 | NR |
| Bonnemann et al, 1996 [20] | NR | Brazil | LGMDR4 | 7 | NR |
| Bouquet et al, 2012 [21] | Case report | France | LGMDR12 | 2 | NR |
| Burke et al, 2010 [22] | Case report | UK | LGMDR1 | 1 | NR |
| Cagliani et al, 2001 [23] | Case report | Italy | LGMDR4 | 1 | NR |
| Cagliani et al, 2003 [24] | NR | Italy | LGMDR2, MM | 7 | NR |
| Cagliani et al, 2005 [25] | Retrospective | Italy | LGMDR2/MM | 25 | NR |
| Cai et al, 2019 [26] | Retrospective | China | LGMDR12 | 5 | 2005-2018 |
| Calvo et al, 2000 [27] | NR | Spain | LGMDR5 | 10 | NR |
| Carman et al, 2018 [28] | Case report | Turkey | LGMDR2 | 1 | NR |
| Catteruccia et al, 2018 [29] | Case report | Italy | LGMDR3 | 2 | NR |
| Chourasia et al, 2017 [30] | Case report | USA | LGMDR9 | 1 | NR |
| Connolly et al, 1998 [31] | Case report | USA | LGMDR5 | 1 | NR |
| D'Amico et al, 2008 [32] | Case report | Italy | LGMDR9 | 1 | NR |
| Darin et al, 2007 [33] | Case report | Sweden | LGMDR9 | 2 | NR |
| de Albuquerque et al, 2015 [34] | Retrospective | Brazil | LGMDR1 | 6 | 3 years |
| de Paula et al, 2003 [35] | NR | Brazil | LGMDR9 | 16 | NR |
| Dicapua et al, 2014 [36] | Case report | USA | LGMDR5 | 2 | NR |
| Diers et al, 2007 [37] | Case report | Germany | LGMDR2 | 1 | NR |
| Dincer et al, 2000 [38] | Cross-sectional | Turkey | LGMDR1, LGMDR2, LGMDR3-6 | 25 | NR |
| Diniz et al, 2014 [39] | Case report | Turkey | LGMDR3 | 1 | NR |
| Diniz et al, 2015 [40] | Case report | Turkey | LGMDR4 | 1 | NR |
| Dirik et al, 2001 [41] | Case report | Turkey | LGMDR1 | 1 | NR |
| Driss et al, 2000 [42] | NR | Tunisia | LGMDR9 | 13 | NR |
| Fanin et al, 2003 [43] | Case series | Italy | LGMDR4 | 6 | NR |
| Fanin et al, 2004 [44] | Retrospective | Italy | LGMDR1 | 58 | NR |
| Fatehi et al, 2015 [45] | Prospective | Iran | LGMDR2 | 16 | NR |
| Fayssoil et al, 2018 [46] | Retrospective | France | LGMDR3-6 | 10 | NR |
| Feng et al, 2018 [47] | Cross-sectional | China | LGMDR1 | 32 | 2006-2016 |
| Figarella-Branger et al, 2002 [48] | Retrospective | France | LGMDR1, LGMDR3 | 5 | NR |
| Fischer et al, 2005 [49] | NR | Germany | LGMDR1, LGMDR2, LGMDR3, LGMDR9 | 17 | NR |
| Frosk et al, 2005 [50] | Case report | Canada | LGMDR9 | 3 | NR |
| Fu et al, 2016 [51] | Prospective | China | LGMDR9 | 12 | 2007-2016 |
| Fuschillo et al, 2010 [52] | Case report | Italy | LGMDR2/MM | 1 | NR |
| Gaul et al, 2006 [53] | Prospective | Germany | LGMDR9 | 9 | NR |
| Gemelli et al, 2017 [54] | Case report | Italy | LGMDR12 | 1 | NR |
| Ghafouri-Fard et al, 2017 [55] | Case report | Iran | LGMDR4 | 2 | NR |
| Ginjaar et al, 2000 [56] | Prospective | Netherlands | LGMDR3-5 | 8 | NR |
| Gomez-Andres et al, 2019 [57] | NR | Chile | LGMDR2/MM | 33 | 2011-2017 |
| Guglieri et al, 2008 [58] | Retrospective | Italy | LGMDR1, LGMDR2, LGMDR3-6, LGMDR9 | 181 | NR |
| Gulati et al, 2003 [59] | Case report | India | LGMDR5 | 1 | NR |
| Habig et al, 2017 [60] | Case report | Germany | LGMDR1 | 1 | NR |
| Hanisch et al, 2007 [61] | Retrospective | Germany | LGMDR1 | 9 | NR |
| Heikali et al, 2017 [62] | Case report | USA | LGMDR9 | 1 | NR |
| Hicks et al, 2011 [63] | Case series | UK, Germany | LGMDR12 | 20 | NR |
| Hollingsworth et al, 2013 [64] | Prospective | UK | LGMDR9 | 10 | NR |
| Hong et al, 2011 [65] | Case report | China | LGMDR9 | 2 | NR |
| Illa et al, 2007 [66] | Case report | Spain | LGMDR2 | 5 | NR |
| Izzedine et al, 2006 [67] | Case report | France | LGMDR2 | 1 | NR |
| Jenne et al, 2005 [68] | Case report | USA | LGMDR1 | 1 | 1995-2004 |
| Jethwa et al, 2013 [69] | Case report | UK | LGMDR2 | 1 | NR |
| Jomaa et al, 2009 [70] | Case report | Tunisia | LGMDR5 | 1 | NR |
| Kang et al, 2007 [71] | Prospective | USA | LGMDR9 | 7 | NR |
| Kapoor et al, 2005 [72] | Case report | India | LGMDR4 | 1 | NR |
| Kefi et al, 2008 [73] | Case report | Tunisia | LGMDR9 | 4 | NR |
| Khadilkar et al, 2008 [74] | Case report | India | LGMDR2/MM | 2 | 22 years |
| Khaiboullina et al, 2017 [75] | Case report | Russia | LGMDR2 | 3 | NR |
| Kida et al, 2015 [76] | Case report | Japan | LGMDR12 | 1 | 2004 |
| Klinge et al, 2008 [77] | Case report | UK | LGMDR2/MM | 1 | NR |
| Kocak Eker et al, 2019 [78] | Case report | Turkey | LGMDR6 | 1 | NR |
| Koken et al, 2018 [79] | Case report | Turkey | LGMDR9 | 1 | NR |
| Krishnaiah et al, 2016 [80] | Case report | USA | LGMDR3 | 1 | NR |
| Lanzillo et al, 2006 [81] | Case report | Italy | LGMDR1 | 1 | NR |
| Lerario et al, 2010 [82] | Case report | Italy | LGMDR2/MM | 2 | NR |
| Li et al, 2014 [83] | Case report | China | LGMDR2 | 1 | NR |
| Liewluck et al, 2012 [84] | Case report | USA | LGMDR1 | 1 | NR |
| Liewluck et al, 2013 [85] | Retrospective | USA | LGMDR12 | 7 | 1998-2012 |
| Lin et al, 2007 [86] | Case report | Tiawan | LGMDR9 | 1 | NR |
| Lindberg et al, 2011 [87] | Case report | Sweden | LGMDR9 | 5 | 2 years |
| Little et al, 2013 [88] | Case report | USA | LGMDR12 | 1 | NR |
| Lokken et al, 2016 [89] | Cross-sectional | Denmark | LGMDR9 | 11 | NR |
| Lomma et al, 2018 [90] | Case report | Australia | LGMDR1 | 1 | NR |
| Lu et al, 2019 [91] | Case report | China | LGMDR3 | 1 | NR |
| Margeta et al, 2009 [92] | Case report | USA | LGMDR9 | 2 | NR |
| Martinez-Thompson et al, 2018 [93] | Case report | USA | LGMDR1 | 1 | NR |
| Mashiah et al, 2016 [94] | Case report | Israel | LGMDR2 | 1 | NR |
| Matsubara et al, 2007 [95] | Case report | Japan | LGMDR1 | 2 | NR |
| McMillan et al, 2009 [96] | Case report | Canada | LGMDR5 | 1 | NR |
| McNally et al, 1996 [97] | Case report | Brazil | LGMDR5 | 11 | NR |
| Mendell et al, 2010 [98] | RCT | USA | LGMDR3 | 6 | 2 years |
| Mercuri et al, 2003 [99] | Prospective | UK | LGMDR9 | 22 | NR |
| Meznaric-Petrusa et al, 2009 [100] | Case report | Slovenia | LGMDR3 | 1 | NR |
| Miskew Nichols et al, 2018 [101] | Case report | Canada | LGMDR9 | 1 | NR |
| Mojbafan et al, 2016 [102] | Case report | Iran | LGMDR3 | 5 | NR |
| Moody et al, 2013 [103] | Case report | USA | LGMDR2 | 1 | NR |
| Muller et al, 2005 [104] | Case report | Germany | LGMDR9 | 3 | NR |
| Mustafa et al, 2014 [105] | Case report | Bangladesh | LGMDR4 | 1 | NR |
| Nagashima et al, 2004 [106] | Case report | Japan | LGMDR2, MM | 1 | NR |
| Negrao et al, 2012 [107] | Case report | Portugal | LGMDR12 | 3 | NR |
| Nemes et al, 2017 [108] | Case report | Hungary | LGMDR1 | 1 | NR |
| Neto et al, 2018 [109] | Case report | NR | LGMDR1 | 2 | NR |
| Nowak et al, 2000 [110] | Case report | Australia | LGMDR5 | 2 | NR |
| Oexle et al, 1996 [111] | Case report | Bulgaria | LGMDR5 | 2 | NR |
| Oflazer et al, 2009 [112] | Case report | Turkey | LGMDR1 | 1 | NR |
| Okahashi et al, 2008 [113] | Case report | Japan | LGMDR2 | 1 | NR |
| Okere et al, 2013 [114] | Case report | USA | LGMDR1 | 1 | NR |
| Oliveira Santos et al, 2018 [115] | Case report | Guinea-Bissau | LGMDR1 | 2 | NR |
| Paradas et al, 2010 [116] | Retrospective | Spain | LGMD2, MM | 26 | NR |
| Park et al, 2012 [117] | Retrospective | Korea | LGMDR2, MM | 28 | NR |
| Passos-Bueno et al, 1996 [118] | Retrospective | Brazil | LGMDR3 | 63 | NR |
| Patel et al, 2017 [119] | Case report | USA | LGMDR2/MM | 2 | NR |
| Peddareddygari et al, 2018 [120] | Case report | USA | LGMDR12 | 1 | NR |
| Pena et al, 2010 [121] | Case report | USA | LGMDR5 | 1 | NR |
| Penisson-Besnier et al, 1998 [122] | Case report | France | LGMDR1 | 2 | NR |
| Penttila et al, 2012 [123] | Retrospective | Finland | LGMDR12 | 25 | NR |
| Peric et al, 2019 [124] | NR | Serbia | LGMDR1 | 19 | 2008-2017 |
| Petersen et al, 2015 [125] | Retrospective | Switzerland | LGMDR2 | 13 | 1989-2015 |
| Pimentel et al, 2008 [126] | Case report | Brazil | LGMDR2 | 3 | NR |
| Pizzanelli et al, 2006 [127] | Case report | Italy | LGMDR1 | 1 | NR |
| Pollitt et al, 2001 [128] | Retrospective | UK | LGMDR1 | 13 | NR |
| Poppe et al, 2003 [129] | Retrospective | UK | LGMDR9 | 16 | NR |
| Prahm et al, 2017 [130] | Case report | Denmark | LGMDR1 | 1 | NR |
| Prelle et al, 1998 [131] | NR | Italy | LGMDR3-6 | 12 | NR |
| Prelle et al, 2003 [132] | Case series | Italy | LGMDR2/MM | 6 | NR |
| Quick et al, 2015 [133] | NR | Germany | LGMDR1 | 10 | January-September 2013 |
| Quinlivan et al, 1997 [134] | Case report | UK | LGMDR3 | 1 | NR |
| Rasmussen et al, 2014 [135] | Retrospective | Norway | LGMDR9 | 17 | 2004-2012 |
| Rekik et al, 2019 [136] | Case report | Tunisia | LGMDR1 | 1 | NR |
| Renard et al, 2012 [137] | Case report | France | LGMDR9 | 1 | NR |
| Ro et al, 2004 [138] | Prospective | Taiwan | MM | 4 | NR |
| Rosales et al, 2010 [139] | Retrospective | USA | LGMDR2 | 21 | NR |
| Rosales et al, 2011 [140] | Prospective | USA | LGMDR2, LGMDR9 | 16 | October 2006-January 2009 |
| Saenz et al, 2011 [141] | Case report | Spain | LGMDR1 | 2 | NR |
| Sahai et al, 2019 [142] | Case report | USA | LGMDR1 | 1 | NR |
| Sarkozy et al, 2012 [143] | NR | UK | LGMDR12 | 25 | NR |
| Sarkozy et al, 2013 [144] | Retrospective | UK | LGMDR12 | 44 | NR |
| Schessl et al, 2012 [145] | Case report | Germany | LGMDR12 | 4 | NR |
| Schneider et al, 2014 [146] | Case report | Germany | LGMDR12 | 1 | NR |
| Schutz et al, 2017 [147] | Case report | UK | LGMDR1 | 1 | NR |
| Sczesny-Kaiser et al, 2017 [148] | Case report | Germany | LGMDR1, LGMDR9 | 2 | NR |
| Selva-O'Callaghan et al, 2006 [149] | Case report | Spain | LGMDR2 | 1 | NR |
| Seong et al, 2016 [150] | Prospective | Korea | LGMDR1, LGMDR2 | 13 | NR |
| Sharma et al, 2004 [151] | Retrospective and prospective | India | LGMDR3-6 | 13 | 1998-2004 |
| Shouman et al, 2018 [152] | Case report | USA | LGMDR12 | 2 | NR |
| Strafella et al, 2019 [153] | Case report | Italy | LGMDR1 | 1 | NR |
| Svahn et al, 2015 [154] | Case report | France | LGMDR9 | 1 | NR |
| Szymanska et al, 2014 [155] | Case report | Poland | LGMDR2 | 1 | NR |
| Takahashi et al, 2003 [156] | Retrospective | Japan | MM | 25 | NR |
| Takahashi et al, 2006 [157] | Case report | Japan | LGMDR2 | 1 | NR |
| Takahashi et al, 2019 [158] | Case report | Japan | LGMDR2 | 1 | NR |
| Takano et al, 2000 [159] | Case report | Japan | LGMDR5 | 2 | 1991-1998 |
| Tarnopolsky et al, 2015 [160] | Case report | USA | LGMDR3 | 2 | NR |
| Tasca et al, 2014 [161] | Case report | Italy | LGMDR12 | 1 | NR |
| Tasca et al, 2018 [162] | Prospective | Europe | LGMDR3-6 | 69 | NR |
| Tetreault et al, 2011 [163] | Retrospective | Canada | LGMDR3 | 9 | NR |
| Todorova et al, 2007 [164] | Retrospective | Bulgaria | LGMDR1 | 22 | NR |
| Topaloglu et al, 1997 [165] | Prospective | Turkey | LGMDR1 | 10 | NR |
| Tsuburaya et al, 2011 [166] | Case report | Japan | LGMDR1 | 1 | NR |
| Ueyama et al, 2001 [167] | NR | Japan | LGMDR2, MM | 5 | NR |
| Umakhanova et al, 2017 [168] | Prospective | Russia | LGMDR2 | 12 | NR |
| Vainzof et al, 2000 [169] | NR | Brazil | LGMDR3 | 3 | NR |
| Van der Kooi et al, 1998 [170] | Case report | The Netherlands | LGMDR5 | 5 | NR |
| Van der Kooi et al, 2013 [171] | Retrospective | The Netherlands | LGMDR12 | 13 | >15 years |
| Viloria-Alebesque et al, 2019 [172] | Case report | Spain | LGMDR1 | 1 | NR |
| Vissing et al, 2014 [173] | Prospective | Denmark | LGMDR12 | 6 | NR |
| Von Der Hagen et al, 2006 [174] | Case report | Germany | LGMDR9 | 2 | NR |
| Von Guionneau et al, 2018 [175] | Case report | UK | LGMDR9 | 1 | NR |
| Vorgerd et al, 2001 [176] | Case report | Germany | LGMDR5 | 1 | NR |
| Wahbi et al, 2008 [177] | Prospective | France | LGMDR9 | 23 | NR |
| Walsh et al, 2011 [178] | Case report | Ireland | LGMDR2 | 1 | NR |
| Walter et al, 2003 [179] | Case report | Germany | LGMDR2/MM | 4 | NR |
| Walter et al, 2004 [180] | Case report | Germany | LGMDR3 | 2 | NR |
| Walter et al, 2013 [181] | RCT | Germany | LGMDR2, MM | 23 | 2003-2008 |
| Wang et al, 2015 [182] | Case report | Taiwan | LGMDR1 | 1 | NR |
| Wang et al, 2018 [183] | Case report | China | LGMDR9 | 2 | NR |
| Weiler et al, 1996 [184] | Retrospective | Canada | LGMDR2, MM | 9 | NR |
| Willis et al, 2014 [185] | Cross-sectional | Europe | LGMDR9 | 38 | NR |
| Wong-Kisiel et al, 2010 [186] | Case report | USA | LGMDR4 | 2 | NR |
| Woudt et al, 2016 [187] | Prospective | Chile | LGMDR2, MM | 31 | 2009-2012 |
| Xi et al, 2014 [188] | Prospective | China | LGMDR2/MM | 29 | 2005-2009 |
| Xie et al, 2018 [189] | Prospective | China | LGMDR9 | 10 | NR |
| Xie et al, 2019 [190] | Prospective | China | LGMDR3-6 | 25 | January 2013-August 2018 |
| Xu et al, 2018 [191] | Case report | China | LGMDR2 | 1 | NR |
| Yilmaz et al, 2009 [192] | Case report | Germany | LGMDR5 | 1 | NR |
| Ylikallio et al, 2016 [193] | Retrospective | Finland | LGMDR12 | 14 | NR |
| Yu et al, 2017 [194] | Prospective | China | LGMDR1, LGMDR2, LGMDR3, LGMDR9, LGMDR12 | 92 | January 2013-May 2015 |
| Yuksel et al, 2018 [195] | Case report | NR | LGMDR4 | 1 | NR |

**Abbreviations:** LGMD, limb girdle muscular dystrophy; MM, Miyoshi myopathy; NR, not reported; RCT, randomized controlled trial, UK, United Kingdom; USA, United States of America.

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