

Gene	Disease	PI/Coordinator Institution Email	Website IT-base	Access for scientist/ Patient self registration	Publicly available	Scope and Objectives	Population scope	Items (number)	Patients registered
CONGENITAL MUSCULAR DYSTROPHIES and MYOPATHIES									
COL6A1 COL6A2 COL6A3	Ullrich congenital myopathy Bethlem myopathy	Valerie Allmand INSERM U582, Institute de Myology, Paris, France; v.allmand@institut-myologie.org	Planned website UMD	Free Internet access to published mutations. Unpublished data accessed only via Curators. Professional registration		Mutation database, epidemiology, natural history, genotype-phenotype correlation, pt recruitment for clinical trials.	Global	list of items available (short list of mandatory and highly encouraged)	
COL6A1 COL6A2 COL6A3	Ullrich congenital myopathy Bethlem myopathy	Anne Lampe South East of Scotland Clinical Genetic Service, Western General Hospital; Edinburgh, UK; anneklampe@doctors.org.uk Johan t. den Dunnen Leiden University Medical Centre, LUMC, Leiden, the Netherlands; ddunnen@HumGen.nl	http://www.dmd.nl/COL6A1 http://www.dmd.nl/COL6A2 http://www.dmd.nl/COL6A3 LSDB, LOVD	Different levels. Restricted IP address access. Professional registration	YES	Mutation database. Description of DNA variants, LSDB standardisation.	Global	34 patient, genetic and clinical data	37 (COL6A1) 32 (COL6A2) 21 (COL6A3) public entries
LAMA2	MDC1A	Christophe Beroud Montpellier University, Laboratoire de Genetique Moleculaire, Montpellier, France; Christophe.Beroud@igh.cnrs.fr	planned website UMD	Access for scientists. Professional registration		Mutation database, natural history, genotype-phenotype correlation, pt recruitment for clinical trials.	Global	8 topics	
LAMA2	MDC1A	Johan t. den Dunnen Leiden University Medical Centre, LUMC, Leiden, the Netherlands; ddunnen@HumGen.nl	http://www.dmd.nl/LAMA2 LSDB, LOVD	Different levels. Restricted IP address access. Professional registration	YES	Mutation database. Description of DNA variants, LSDB standardisation	Global	34 patient, genetic and clinical data	294 public entries
MTM1	Myotubular Myopathy	Valerie Biancalana Laboratoire de Diagnostic Genetique, Faculte' de Medecine-CHRU, Strasbourg, France; valerie.biancalana@chru-strasbourg.fr Jocelyn Laporte IGBMC, Illkirch, France; jocelyn@igbmc.u-strasbg.fr	Single-site, lab-based Mutation database Future plans: Website UMD	Access only for the curator, data can be obtained upon request Future plans: Free access to published mutations to scientists (anonymized data). Unpublished data accessed only by curator and participants. Professional registration	NO	Clinical and molecular database, genotype- phenotype correlation. Improve diagnosis and genotype-phenotype correlation, prognosis, future clinical trials. Involvement of patient organizations	Europe + some other countries Future plans : Global		431 families
MTM1	Myotubular Myopathy	Jorge Oliveira Instituto de Genética Médica J. Magalhães, Unidade de Genética Molecular, Porto, Portugal; jorge.oliveira@igm.min-saude.pt Johan t. den Dunnen Leiden University Medical Centre, LUMC, Leiden, the Netherlands; ddunnen@HumGen.nl	http://www.dmd.nl/MTM1 under construction LSDB, LOVD	Different levels. Restricted IP address access. Professional registration	YES	Mutation database. Description of DNA variants, LSDB standardisation	Global	34 patient, genetic and clinical data	21 public entries
BIN1	Centronuclear myopathy	Valerie Biancalana Laboratoire de Diagnostic Genetique, Faculte' de Medecine-CHRU, Strasbourg, France; valerie.biancalana@chru-strasbourg.fr Jocelyn Laporte IGBMC, Illkirch, France; jocelyn@igbmc.u-strasbg.fr	Future plans: Website UMD	Future plans: Free access to published mutations to scientists (anonymized data). Unpublished data accessed only by curator and participants. Professional registration	NO	Clinical and molecular database, genotype- phenotype correlation. Improve diagnosis and genotype-phenotype correlation, prognosis, future clinical trials	Global		7 patients
Dynamin 2	Centronuclear myopathy	Marc Bitoun INSERM U582, Institute de Myology, Paris, France; m.bitoun@institut-myologie.org	Planned website UMD	Access for scientists. Professional registration	NO	Clinical and molecular database, genotype- phenotype correlation, natural history	Global		
NEB	Nemaline	Carina Wallgren-Pettersson	Single-site, mainly clinical	Professional registration	NO	ENMC International	Global	170 items	370 pt

ACTA1	Myopathy	University of Helsinki and Folkhälsan, Dept. of Medical Genetics, Helsinki, Finland; Carina.Wallgren@helsinki.fi	database Mutational data on subset. Planned website			Consortium. Anonymous clinical data. Genotype-phenotype correlations.		clinical, familial, histological, genetic and other data	
ACTA1	Nemaline Myopathy	Nigel Laing Centre for Medical Research, University of Western Australia and Western Australian Institute for Medical Research, Nedlands, Western Australia, Australia; nlaing@cylene.uwa.edu.au	Website under development LSDB	Access for scientists. Professional registration		Mutation database	Global		
POMT1 POMT2 POMGnT1 LARGE FKRP FCMD	WWS MEB CMDs LGMDs	Francesco Muntoni Dubowitz Neuromuscular Centre, Institute of Child Health & Great Ormond street Hospital, London, UK; f.muntoni@imperial.ac.uk Judith Pagan, Steve Abbs Guy's & Thomas NHS Foundation Trust, London, UK; Judith.pagan@gstt.nhs.uk ; Stephen.Abbs@gstt.nhs.uk	Single-site, lab-based Mutation database Set up LOVD for all genes	Access for scientists. Professional registration	Variants publicly available in the Leiden database	Mutation and clinical database, natural history, treatment, ethnicity, consanguinity, patients' data, genotype-phenotype correlation, pt recruitment for clinical trials. ENMC International Consortium.	UK	Start with basic information that can be added to if required. LOVD: Clinical and patient data	
POMT1 POMT2 POMGnT1 FKTN (ex FCMD) FKRP LARGE	LGMDs WWS CMDs	Rosario dos Santos Instituto de Genética Médica Dr. Jacinto Magalhães, Unidade de Genética Molecular, Porto, Portugal; rosario.santos@igm.min-saude.pt Johan t. den Dunnen Leiden University Medical Centre, LUMC, Leiden, the Netherlands; cdunnen@HumGen.nl	http://www.dmd.nl/POMT1 http://www.dmd.nl/POMT2 http://www.dmd.nl/POMGnT1 http://www.dmd.nl/FCMD http://www.dmd.nl/FKRP http://www.dmd.nl/LARGE LSDB, LOVD	Different levels. Restricted IP address access. Professional registration	YES	Mutation database. Description of DNA variants, LSDB standardisation	Global	34 patient, genetic and clinical data	97 (POMGnT1) 149 (POMT1) 71 (POMT2) 91 (FKTN) 361 (FKRP) 24 (LARGE) public entries
RYR1	CCD MmD (CNM)	Heinz Jungbluth, Steve Abbs, Sue Lillis Guy's & Thomas NHS Foundation Trust, London, UK; King's College, London, UK; Heinz.Jungbluth@gstt.nhs.uk	Test data base Website planned	Access for scientists. Professional registration	NO	Mutation and clinical database, epidemiology, genotype-phenotype correlation, longitudinal and natural history, pt recruitment for clinical trials. Entry point: RYR1 mutation and congenital myopathy phenotype	UK Europe	40 items (genetic, clinical, pathological)	80 pt
SEPN1	Minicore RSM1	Ana Ferreira Institut National de la Sante et de la Recherche Médicale U582, Institut de Myologie, Paris, France; a.ferreiro@myologie.chups.jussieu.fr	Planned website UMD	Access for scientists. Professional registration		Mutation database, epidemiology, natural history, genotype-phenotype correlation, pt recruitment for clinical trials. ENMC International Consortium	Global		
GAA	Pompe Disease	Ria Broekgaarden Vereniging Sperziekten Nederland, Baam, the Nederland; Ria.broekgaarden@vsn.nl	The Pompe Registry http://www.lsdregistry.net/pomperegistry/ IPA/Erasmus Survey	Data collected by clinicians, and patient self-reported data	NO	Ongoing, observational database that tracks natural history and outcomes of patients with Pompe disease. Ongoing patient self-reported data	Global		>300 pt 110 Dutch
GAA	Pompe Disease	Ria Broekgaarden Vereniging Sperziekten Nederland, Baam, the Nederland; Ria.broekgaarden@vsn.nl	Vereniging Sperziekten Nederland http://www.vsn.nl/	VSN member registration (10.000) Data collected by the patient organisation.	NO	Databank of patients with confirmed diagnosis, date of birth, date of diagnosis, age and sex. On going member registration.	The Netherlands		
GAA	Pompe Disease	Pascal Laforet NMD reference center, Myology Institute Paris, France; pascal.laforet@psl.aphp.fr	The French Pompe registry https://eventa.kikamedical.com/maltase/ ; moving to Montpellier (C Beroud)	Access for scientists. Professional registration	NO	Epidemiology, genotype-phenotype correlations, natural history, evaluation of treatments	French		60
PYGM	McArdle Disease	Pascal Laforet NMD reference center, Myology Institute, Paris, France; pascal.laforet@psl.aphp.fr	Registry in progress Website planned: Montpellier (C Beroud)	Access for scientists. Professional registration		Epidemiology, genotype-phenotype correlations natural history, clinical trials	Several countries		

		<p>Andrea Martinuzzi Medea Scientific Institute, Conegliano Research Centre, Conegliano, Italy; andrea.martinuzzi@cn.Inf.it</p> <p>Ros Quinlivan The Wolfson Centre for Inherited Neuromuscular Diseases (CIND), Robert Jones and Agnes Hunt, NHS Trust, Oswestry, UK; Rcmq37@aol.com</p> <p>John Vissing Copenhagen Muscle Research Center, National University Hospital, Denmark; vissing@rh.dk</p>								
AGL	Glycogen storage disease type III	<p>Philippe Labrune Antoine Beclere Hospital, Université Paris Sud, UFR Kremlin Bicêtre, Clamart Cedex, France philippe.labrune@abc.aphp.fr (for children)</p> <p>Pascal Laforet NMD reference center, Myology Institute, Paris, France pascal.laforet@psl.aphp.fr (for adults)</p>	Registry in progress Website planned: Montpellier (C Beroud)	Access for scientists. Professional registration		Epidemiology, genotype-phenotype correlations, natural history	Several countries			
LGMDs and MFMs										
DYSF	LGMD2B Miyoshi Myopathy	<p>Laura Rufibach, Plavi Mittal Jain Foundation Inc. Redmond, WA, USA Lrufibach@jain-foundation.org plavimittal@jain-foundation.org</p>	The Jain Foundation https://jain-foundation.org/patients.php	Patient self registration and clinician/researcher referral	NO	Patient registry. Help for diagnosis (information and financial), epidemiology, and patient recruitment for clinical trials.	Global	Registration form asking basic information re diagnosis (6 items), family background (4), symptoms (20)	>208 pt	
CAPN3 DYSF	LGMD2A LGMD2B Miyoshi Myopathy	<p>Nicolas Levy, Martin Krahn INSERM U910 and APHM, Laboratoire de Genetique Moleculaire, Marseille, France; Nicolas.Levy@medecine.univ-mrs.fr martin.krahn@medecine.univ-mrs.fr</p>	Website from 2008 UMD	Access for scientists. Professional registration		mutation database: genotype-phenotype correlation, help for diagnosis, trial recruitment	Global		>200 >100	
CAPN3	LGMD2A	<p>Jacques Beckmann Department of Medical Genetics, University of Lausanne, Lausanne, Switzerland; Jacques.Beckmann@chuv.ch</p> <p>Johan t. den Dunnen Leiden University Medical Centre, LUMC, Leiden, the Netherlands; ddunnen@HumGen.nl</p>	http://www.dmd.nl/CAPN3 LSDB, LOVD	Different levels. Restricted IP address access. Professional registration	YES	Mutation database. Description of DNA variants, LSDB standardisation	Global	34 patient, genetic and clinical data	1665 public entries	
CAV3	LGMD1C Rippling muscle disease	<p>Johan t. den Dunnen Leiden University Medical Centre, LUMC, Leiden, the Netherlands; ddunnen@HumGen.nl</p>	http://www.dmd.nl/CAV3 LSDB, LOVD	Different levels. Restricted IP address access. Professional registration	YES	Mutation database. Description of DNA variants, LSDB standardisation	Global	34 patient, genetic and clinical data	130 public entries	
DYSF	LGMD2B Miyoshi Myopathy	<p>Johan t. den Dunnen Leiden University Medical Centre, LUMC, Leiden, the Netherlands; ddunnen@HumGen.nl</p>	http://www.dmd.nl/DYSF LSDB, LOVD	Different levels. Restricted IP address access. Professional registration	YES	Mutation database. Description of DNA variants, LSDB standardisation	Global	34 patient, genetic and clinical data	764 public entries	
DES MYOT ZASP CRYAB FLNC	MFm	<p>Montse' Olive IDIBELL Hospital de Bellvitge, Institute of Neuropathology, Barcelona, Spain; 25169mop@comb.es</p> <p>Lev Goldfarb The National Institute of Neurological Disorders and Stroke, National Institutes of Health, Bethesda, MD USA; goldfarbl@ninds.nih.gov</p>	Single-site, clinical data-base	Access for scientists	free access to published patients/ mutations	Epidemiology, define clinical phenotype, subclassification of patients, recruitment patients with unknown genetic defect. ENMC International Consortium	Global	99 items clinical, muscle imaging, histological, genetic and other data	35	
MYOT	MFm LGMD1A	<p>Johan t. den Dunnen Leiden University Medical Centre, LUMC, Leiden, the Netherlands; ddunnen@HumGen.nl</p>	http://www.dmd.nl/MYOT LSDB, LOVD	Different levels. Restricted IP address access. Professional registration	YES	Mutation database. Description of DNA variants, LSDB standardisation	Global	34 patient, genetic and clinical data	24 public entries	
LMNA	EDMD2	Gisèle Bonne	UMD central:	Free Internet access to published	free access	Mutation and clinical	Global	315 items	Total: 1434 LMNA	

	EDMD3 LGMD1B CMD1A	Institute National de la Sante' et de la Recherche Medicale, INSERM UR582, Institute de Myology, Paris, France; g.bonne@institut-myologie.org	http://www.umd.be : UMD-LMNA: http://www.umd.be:2000/ Euro-Laminopathies http://www.projects.mfpl.ac.a/euro-laminopathies/php/index.php?id=14	mutations. Unpublished data (291 LMNA mutated subjects) accessed only via Curators Professional registration	to published mutations	database. Natural History, understand pathogenetic mechanisms, epidemiology.		(Mycluster form); 120 (mini-myocuster form)	mutated subjects: 334 EDMD pt, 125 LGMD1B pt, 336 CMD1A pt. 142 published references, 291 unpublished subjects (non available via Internet)
LMNA	EDMD2 EDMD3 LGMD1B CMD1A	Johan t. den Dunnen Leiden University Medical Centre, LUMC, Leiden, the Netherlands; ddunnen@HumGen.nl	http://www.dmd.nl/LMA LSDB, LOVD	Different levels. Restricted IP address access Professional registration	YES	Mutation database Description of DNA variants, LSDB standardisation	Global	34 patient, genetic and clinical data	360 public entries
EMD	EDMD1	Cisèle Bonne Institute National de la Sante' et de la Recherche Medicale, INSERM UR582, Institute de Myology, Paris, France; g.bonne@institut-myologie.org	UMD central: http://www.umd.be : -UMD- EMD: http://www.umd.be:2010/ ; Euro-Laminopathies: http://www.projects.mfpl.ac.a/euro-laminopathies/php/index.php?id=14	Free Internet access to published mutations. Unpublished data (291 LMNA mutated subjects) accessed only via Curators. Professional registration	free access to published mutations	Mutation and clinical database. Natural History, understand pathogenetic mechanisms, epidemiology.	Global	315 items (Mycluster form); 120 (mini-myocuster form)	Total: 333 EMD mutated subjects: 45 published references, 112 unpublished subjects (non available via Internet)
EMD	EDMD1	Johan t. den Dunnen Leiden University Medical Centre, LUMC, Leiden, the Netherlands; ddunnen@HumGen.nl	http://www.dmd.nl/EMD LSDB, LOVD	Different levels. Restricted IP address access. Professional registration	YES	Mutation database. Description of DNA variants, LSDB standardisation	Global	34 patient, genetic and clinical data	140 public entries
SGCA SGCB SGCG SGCD	LGMD2 C-F	Gabriele Dekomien Humangenetik, Ruhr Universitat Bochum, Germany; gabriele.dekomienn@rub.de France Leturcq LBGM, Laboratoire Cassini, PARIS, France leturcq@cochin.insem.fr	Lab-based mutation database Planned	Patients self registration Access for scientists as well	NO	Mutation database	Germany, UK, Turkey, Arabia, Gipsy French and a few Mediterranean pt		203 pt + 90 family members investigated
SGCA SGCB SGCG SGCD SGCE	LGMD2 C-F Myoclonus-dystonia syndrome	Johan t. den Dunnen Leiden University Medical Centre, LUMC, Leiden, the Netherlands; ddunnen@HumGen.nl	http://www.dmd.nl/SGCA http://www.dmd.nl/SGCB http://www.dmd.nl/SGCG http://www.dmd.nl/SGCD http://www.dmd.nl/SGCE LSDB, LOVD	Different levels. Restricted IP address access. Professional registration	YES	Mutation database. Description of DNA variants, LSDB standardisation	Global	34 patient, genetic and clinical data	277 (SGCA) 167 (SGCB) 190 (SGCG) 40 (SGCD) 78 (SGCE) Public entries
TTN	TMD LGMD2J	Johan t. den Dunnen Leiden University Medical Centre, LUMC, Leiden, the Netherlands ddunnen@HumGen.nl	http://www.dmd.nl/ LSDB, LOVD	Different levels. Restricted IP address access. Professional registration	YES	Mutation database. Description of DNA variants, LSDB standardisation	Global	34 patient, genetic and clinical data	20 public entries
TCAP	LGMD2G	Johan t. den Dunnen Leiden University Medical Centre, LUMC, Leiden, the Netherlands ddunnen@HumGen.nl	http://www.dmd.nl/TCAP LSDB, LOVD	Different levels. Restricted IP address access. Professional registration	YES	Mutation database. Description of DNA variants, LSDB standardisation	Global	34 patient, genetic and clinical data	6 public entries
TRIM32	LGMD2H STM	Johan t. den Dunnen Leiden University Medical Centre, LUMC, Leiden, the Netherlands ddunnen@HumGen.nl	http://www.dmd.nl/TRIM32 LSDB, LOVD	Different levels. Restricted IP address access. Professional registration	YES	Mutation database. Description of DNA variants, LSDB standardisation	Global	34 patient, genetic and clinical data	13 public entries
DM1/DM2/FSHD/OPMD									

DM1/2 FSHD	Myotonic Dystrophy, FSHD	Richard Moxley, Charles Thornton, Rabi Tawil [co-investigator (FSHD)] Department of Neurology, University of Rochester Medical Center, Rochester, New York, USA dystrophy_registry@umc.rochester.edu	National Registry of Myotonic Dystrophy and Faciocapulohumeral Muscular Dystrophy Patients and Family Members http://www.umc.rochester.edu/nihregistry/	Patients self registration. Scientists can access anonymized data or apply to Registry if they need access to patients.	NO	Designed to promote research by facilitating contact between researchers and affected individuals, educational tool	USA Global		1400
DM1/2	Myotonic Dystrophy	Mark Rogers University Hospital of Wales, Cardiff, UK Kmark.rogers@cardiffandvale.wales.nhs.uk	Local clinical database	Access for scientists Professional registration	NO	Clinical database for systemic disease follow-up	Wales, UK		337 pt
FSHD	FSHD	Silvere van der Maarel Leiden University Medical Centre (LUMC), Leiden, the Netherlands maarel@lumc.nl	Nijmegen clinical registry Leiden Genetic registry LSDB, LOVD		NO	Genetic and clinical database. Natural history, epidemiology, genotype-phenotype correlation, pt. recruitment for clinical trials	Dutch More global	Some clinical details are available Genetic details	50% Dutch pop. 1600 pt
FSHD	FSHD	Rabi Tawil, Director of the Fields Center Charles Thornton Co-Principal Investigator of the Fields Center Silvere van der Maarel Co-Director of the Fields Center Rabi_Tawil@umc.rochester.edu charles_thornton@umc.rochester.edu maarel@lumc.nl	Fields Center for FSHD and Neuromuscular Research: http://www.umc.rochester.edu/FieldsCenter/index.cfm	Access for scientists	Genotyping data to be added to Leiden public LOVD database.	Genetic and clinical database, biobank, eventually clinical history and pt. recruitment for clinical trials	USA and NL		
PAPBN1	OPMD	Tobias Muller, Marcus Deschauer Dept. of Neurology, University of Halle, Halle, Germany; Tobias.muller@medizin.uni-halle.de Marcus.Deschauer@medizin.uni-halle.de	Local mutation database	Professional registration	NO	Genotype/phenotype correlation, genetic heterogeneity, epidemiology	Germany		219 pt

GENERAL DATABASES

Database	PI/Coordinator Institution Email	Website IT-base	Access for scientists/ Patient self registration	Publicly Available	Scope and Objectives	Population scope	Items (numbers)	Patients registered
CRAMP	Baziel Van Engelen Neuromuscular Centre Nijmegen, Department of Neurology, Nijmegen, the Netherlands; b.vanengelen@neuro.umcn.nl	The Dutch neuromuscular database CRAMP (Computer Registry of All Myopathies and Polyneuropathies) http://www.isno.nl/registratie.php	Access for neurologists, child neurologists, professional registration	NO	Computer Registry Common database for NMD in NL, epidemiology, pt. Recruitment, natural history	Dutch. (Belgium possible in the future)	Treelike structure 8 steps for making the diagnosis	10313 pt.