Bibliographie sur les maladies neuromusculaires

Bibliography of neuromuscular disorders

n° 2020-06-1 du 9 au 21 juin 2020 (June 9 to 21, 2020)

Publiée tous les 15 jours par le service de documentation de l'AFM-Téléthon, la « Veille Neuromusculaire » contient les dernières références intégrées dans Pubmed. La liste des pathologies concernées par cette veille est issue des Fiches Techniques Savoir & Comprendre publiées par l'AFM-Téléthon intitulées « Principales maladies neuromusculaires » (Novembre 2017) et « Recherche Neuromusculaire : Etat des lieux, 6ème Edition » (Septembre 2018). Vous trouverez les veilles précédentes sur notre portail documentaire dédié aux maladies neuromusculaires Myobase.

Every two weeks, the AFM documentation service publishes the “Neuromuscular Bibliography” in which you will find latest references published in Pubmed. The list of diseases below comes from both resources: « Principales maladies neuromusculaires » (November 2017) and « Recherche Neuromusculaire : Etat des lieux, 6ème Edition » (September 2018) published by AFM-Téléthon in Fiches Techniques Savoir & Comprendre Serie. Previous reports are available on Myobase, the informations tool about neuromuscular diseases.

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1 Japan Society for the Promotion of Science (RPD), Tokyo, Japan. 2 Institute of Medical Genetics, Tokyo Women’s Medical University, Tokyo, Japan. 3 Tokyo Women’s Medical University Institute for Integrated Medical Sciences, Tokyo, Japan. 4 Department of Pediatrics, Hiroshima Prefectural Hospital, Hiroshima, Japan. 5 Department of Pediatrics, St. Marianna University School of Medicine, Kawasaki, Japan.
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KEYWORDS: LAMA2-CMD; dy2J/dy2J mouse model; laminin-211; muscular dystrophy; next-generation sequencing; satellite cells
PMID: 32523512 PMCID: PMC7261890 DOI: 10.3389/fnmol.2020.00069 Free PMC Article
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1Wolfson Centre for Inherited Neuromuscular Disease, RJAH Orthopaedic Hospital, Oswestry, SY10 7AG, UK; The School of Pharmacy and Bioengineering, Keele University, ST5 5BG, UK.

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**KEYWORDS:** Differentiation; Emerin; Emery-Dreifuss Muscular dystrophy; LINC complex; Lamin A/C; Nesprin

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**KEYWORDS:** Emery–Dreifuss muscular dystrophy; emerin; histone deacetylase; myogenic differentiation; nuclear envelope

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**KEYWORDS:** BAF; BANF1; DNA-damage response; EDMD1; chromatin; emerin; laminA/C; laminopathies; oxidative stress; prelamin A

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KEYWORDS: Cardiomyopathy; Duchenne Muscular Dystrophy; NHE-1; Pharmacokinetic; Rimeporide; Safety
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Comparison of Survival Analysis between Surgical and Non-surgical Treatments in Duchenne Muscular Dystrophy Scoliosis.
Yang JH1, Kim KS2, Lee GH3, Kim HS4.
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KEYWORDS: Duchenne muscular dystrophy; Forced vital capacity; Functional outcome; Mortality; Respiratory function; Scoliosis; Survival rate; Swinyard scale
PMID: 32535073 DOI:10.1016/j.spinee.2020.06.004

NATURAL HISTORY OF SERUM ENZYME LEVELS IN DUCHENNE MUSCULAR DYSTROPHY AND IMPLICATIONS FOR CLINICAL PRACTICE.
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**The Quest for the Prediction of Steroid Responsiveness in Duchenne Muscular Dystrophy.**

Suthar R1, Sahu JK2.

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**Effect of Aerobic Physical Exercise in an Animal Model of Duchenne Muscular Dystrophy.**

Hoepers A1,2, Freiberger V1, Ventura L1, Grigollo LR1, Andreu CS3, da Silva BB1, Martins DF1, Junior RJN4, Streck EL5, Comim CM1.

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KEYWORDS: Animal; Duchenne muscular dystrophy; Muscle; Oxidative stress; Physical exercise

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**Desmin prevents muscle wasting, exaggerated weakness and fragility, and fatigue in dystrophic mdx mouse.**

Ferry A1,2, Messéant J1, Parlakian A3, Lemaître M1, Roy P1, Delacroix C1, Lilienbaum A4, Hovhannisyan Y3, Furling D1, Klein A1, Li Z2, Akgulul O3.

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KEYWORDS: desmin; mdx mice; skeletal muscle function

PMID: 32515007 DOI:10.1113/JP279282

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**Pulmonary and upper limbs function in children with early stage Duchenne muscular dystrophy compared to their healthy peers.**

Bulut N1, Aydin G2, Alemdaroğlu-Gürbüz F, Karaduman A2, Yilmaz O2.

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KEYWORDS: Functional performance; Pulmonary function test; Rehabilitation; Upper limb

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KEYWORDS: Cognition; Duchenne muscular dystrophy; Dystrophin; Neuroimaging

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KEYWORDS: 4-stair climb; Duchenne muscular dystrophy; domagrozumab; myostatin inhibitor

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KEYWORDS: Duchenne muscular dystrophy (DMD); alarmins; cardiomyopathy; muscular dystrophy; pentraxin 3 (PTX3)

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3Department of Clinical Genetics, Rigshospitalet, University of Copenhagen, Copenhagen, Denmark.
KEYWORDS: calpain 3; calpainopathy; dominant inheritance; limb girdle muscular dystrophy
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KEYWORDS: ANALYSIS/QUANTITATION OF BONECLINICAL TRIALSDISEASES AND DISORDERS OF/RELATED TO BONEFIBRODYSPLASIA OSSICIFICANS PROGRESSIVARADIOLOGY
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KEYWORDS: ACVR1; ALK3-Fc; Activin A; Anti-ACVR1 antibody; Burn tenotomy; Fibrodysplasia ossificans progressiva; Heterotopic ossification; Inhba; Progenitor cells; Single cell RNA sequencing; Trauma
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KEYWORDS: Charcot-Marie-Tooth disease; aminocyl-IRNA synthetase; peripheral neuropathy; protein synthesis; tRNA
PMID: 32543048 DOI: 10.1111/febs.15449
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Madriz R1, Guariglia SR1, Haworth A1, Korosh W1, Gavin M1, Lyon GJ1.
1Jervis Clinic, NYS Institute for Basic Research in Developmental Disabilities (IBR), Staten Island, New York 10314, USA.
2Congenica Ltd, Biodata Innovation Centre, Wellcome Genome Campus, Hinxton, Cambridge CB10 1SA, United Kingdom.

KEYWORDS: progressive cerebellar ataxia; severe global developmental delay

PMID: 32532879 DOI: 10.1101/mcs.a005108 Free full text

Electrodiagnostic accuracy in polyneuropathies: supervised learning algorithms as a tool for practitioners.

Uncini A1, Aretusi G2,3, Manganelli F4, Sekiguchi Y5, Magy L6, Tozza S4, Tsuneyama A6, Lefour S6, Kuwabara S5, Santoro L4, Ippoliti L3.
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KEYWORDS: Diagnostic accuracy; Electrodiagnosis; Polyneuropathies; Supervised learning algorithms

PMID: 32518996 DOI: 10.1007/s10072-020-04499-y

A New Point Mutation in the PMP22 Gene in a Family Suffering From Atypical HNPP.

Benquey T1, Fockens E2, Kouton L2, Delmont E2, Martini N2,4, Attarian S2,3, Bonello-Palot N3,4.
1Service de Biochimie et Biologie moléculaire Grand Est, Unité Médicale Pathologies neurologiques et cardiologiques, Centre de Biologie et de Pathologie Est, Hospices Civils de Lyon, Bron, France.
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KEYWORDS: HNPP; PMP22 gene; phenotypic heterogeneity; point mutations

PMID: 32538861 DOI: 10.3233/JND-190460

Subcellular diversion of cholesterol by gain- and loss-of-function mutations in PMP22.

Zhou Y1, Borchelt D1, Bauson JC2, Fazio S2, Miles JP6, Tavori H4, Notterpek L1,3.
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KEYWORDS: Charcot-Marie-tooth disease Type 1A; Dejerine-Sottas syndrome; Schwann cell; cholesterol subcellular trafficking; fibroblast; peripheral myelin protein 22

PMID: 32511621 DOI: 10.1002/glia.23840

Peripheral myelin protein 22 preferentially partitions into ordered phase membrane domains.

Marinko JT1,2, Kenworthy AK3,4, Sanders CR2,2,6.

PMID: 202000508 DOI: 10.1073/pnas.2000508117

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KEYWORDS: membrane phase domain; ordered; peripheral myelin protein 22

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KEYWORDS: Charcot-Marie-Tooth disease; Patient-reported outcome measures; clinical trials; quality of life; translation and cultural adaptation

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KEYWORDS: PDXK; hereditary neuropathy; optic atrophy; pyridoxal kinase; pyridoxal phosphate; vitamin B6

PMID: 32522499 DOI: 10.1016/j.nmd.2020.04.004

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1Foot and Ankle Unit, Royal National Orthopaedic Hospital, Stanmore, United Kingdom; Department of Public Health, Trauma and Orthopaedics, University of Naples Federico II, Naples, Italy. Electronic address: alebernas@gmail.com.
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KEYWORDS: Cavovarus; Charcot-Marie-Tooth; Cone beam; High arch; Hindfoot; Weightbearing CT

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1Centre de Réadaptation Marie Enfant, Centre de Recherche du CHU Sainte Justine, Montréal, Canada; ToNIC Toulouse Neuroimaging Center, Université de Toulouse, Inserm, UPS, Toulouse, France. Electronic address: claire.cherriere@inserm.fr.
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1Grant MacEwan University, Massage Therapy Program, Canada. Electronic address: galitip@gmail.com.
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1Department of Basic Medical Sciences, Faculty of Medicine, University of La Laguna, 38071 Tenerife, Spain.
2Department of Pathology, Eurofins® Megalab-Hospiten Hospitals, 38100 Tenerife, Spain.
KEYWORDS: Meissner corpuscles; appendicular neurogenic hyperplasia; gallbladder neurogenic hyperplasia; nerves; peripheral nervous system tumours; telocytes
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1Department of Cardiology, The First Affiliated Hospital of Xi’an Jiao Tong University, Xi’an 710061, Shaanxi, China.
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5Harrington Discovery Institute at University Hospitals, 11407 Euclid Ave, Cleveland, Ohio 44106, United States.
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Gala-Bładzińska A1,2, Mazur K3, Debiec A4, Gargasz K5, Bartosik-Psiejek H6,4.
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KEYWORDS: autoimmune neurological diseases; efficacy; side effects; therapeutic plasma exchange
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1University Health Network, Neurology, Toronto, Ontario, Canada.
2University of Toronto, Neurology, Toronto, Ontario, Canada.
KEYWORDS: Myasthenia Gravis; patient acceptable symptom state; patient reported outcomes; single simple question; symptoms
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Sakano S1, Matsuyama H1, Ishikawa H1, Shindo A1, Li Y1, Matsuura K1, Mizutani M1, Kawada N1, Tomimoto H1.
1Department of Neurology, Mie University Graduate School of Medicine, 2-174 Edobashi, Tsu, Mie, 514-8507, Japan.
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4Department of Neurology, Matsusaka Central General Hospital, 102 Azakomon, Kawai-machi, Matsusaka, Mie, 515-0818, Japan.
KEYWORDS: Anti-muscle-specific tyrosine kinase antibodies; Bortezomib; Case report; Multiple myeloma; Myasthenia gravis
PMID: 32532281 PMCID: PMC7291755 DOI:10.1186/s12883-020-01813-1 Free PMC Article

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Lamb CJ1, Rubin DI1.
1Department of Neurology, Mayo Clinic, Jacksonville, Florida.
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Ding J1, Zhao S1, Ren K1, Dang D2, Li H1, Wu F3, Zhang M1, Li Z4, Guo J5.

1Department of Neurology, Tangdu Hospital, Fourth Military Medical University, Xi'an, 710038, Shaanxi Province, China.
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**KEYWORDS:** Generalization; Immunosuppressive therapy; Myasthenia gravis; Ocular; Predictive factor

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**Efficacy and safety of Rituximab in myasthenia gravis: A French multicentre real-life study.**

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7Department of Neurology, University Hospital of Rennes, Rennes, France.
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**KEYWORDS:** Myasthenia gravis; Rituximab; anti-ACh-R; anti-MusK

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**Adult Ocular Myasthenia Gravis Conversion: A Single-Center Retrospective Analysis in China.**

Feng X1, Huan X1, Yan C1, Song J1, Lu J1, Zhou L1, Wu H2, Qiao K3, Lu J4, Xi J5, Luo S1, Zhao C4.

1Department of Neurology, Huashan Hospital Fudan University, Shanghai, China.
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**KEYWORDS:** Adult myasthenia gravis; Anti-acetylcholine receptor antibody; Conversion; Ocular myasthenia gravis; Risk factors

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**The face of myasthenia gravis.**

Ruiter AM1, Naber WC2, Tannemaat MR2, Verschuuren JJGM2.

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**Myasthenia Gravis: From the Viewpoint of Pathogenicity Focusing on Acetylcholine Receptor Clustering, Trans-Synaptic Homeostasis and Synaptic Stability.**

Takamori M1.

1Neurological Center, Kanazawa-Nishi Hospital, Kanazawa, Japan.

**KEYWORDS:** Lrp4; MuSK; acetylcholine receptor; agrin; matrix proteins; myasthenia gravis; neuromuscular junction; wnts

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Fichtner ML, Jiang R, Bourke A, Nowak RJ, O'Connor KC.
1Department of Neurology, School of Medicine, Yale University, New Haven, CT, United States. 
2Department of Immunobiology, School of Medicine, Yale University, New Haven, CT, United States. 
3Trinity Hall, University of Cambridge, Cambridge, United Kingdom.
KEYWORDS: MuSK; AChR; B cells; B lymphocytes; autoantibodies; autoimmunity; immunopathology; myasthenia gravis
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1University of Kentucky, Department of Neurology, USA.
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1Department of Emergency Medicine, Shengjing Hospital of China Medical University, Shenyang, China. 
2Department of Obstetrics and Gynecology, Shengjing Hospital of China Medical University, Shenyang, China.
KEYWORDS: autoimmune; interleukin; microRNA; myasthenia gravis; neuromuscular disease
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8Department of Pathology, Istanbul Medical Faculty, Istanbul University, Istanbul, Turkey. 
9Department of Thoracic Surgery, Istanbul Medical Faculty, Istanbul University, Istanbul, Turkey.
KEYWORDS: CXCR5; ICOS; IL-17; IL-21; IL-4; PD-1; T follicular helper cells; myasthenia gravis
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1Neuroimmunology Laboratory, Mayo Clinic Rochester MN, USA. 
2Neuroimmunology Laboratory, Mayo Clinic Rochester MN, USA. Electronic address: lennon.vanda@mayo.edu.
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3INSERM U1258, Illkirch, France.
4CNRS UMR7104, Illkirch, France.
5Strasbourg University, Illkirch, France.
6Pharmalex, Mont-St-Guibert, Belgium.
7Hospital Armand Trousseau, Institute I-Motion, Institute of Myology, Paris, France.
8MDUK Neuromuscular Center, Department of Paediatrics, University of Oxford, Oxford, UK.
9Division of Child Neurology, Centre de Références des Maladies Neuromusculaires, Department of Pediatrics, University Hospital Liège & University of Liège, 4000 Liège, Belgium.
KEYWORDS: GDF8; MSTN; antisense oligonucleotides; biomarker; centronuclear myopathies; dynamin; myotubular myopathy; therapy
PMID: 32545974 DOI:10.33699/PIS.2020.99.5.226-231

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Lei Y1, Klionsky DJ1.
1Life Sciences Institute and Department of Molecular, Cellular and Developmental Biology, University of Michigan, Ann Arbor, MI, USA.
KEYWORDS: Autophagosome; RAB11; centronuclear myopathy; dynamin 2; recycling endosome; scission
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**Myopathies distales – Distal myopathies**

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**Knockdown of genes involved in axonal transport enhances the toxicity of human neuromuscular disease-related MATR3 mutations in Drosophila.**

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KEYWORDS: Drosophila; MATR3; amyotrophic lateral sclerosis (ALS); genetic screen; multisystem proteinopathy (MSP)

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**Myopathies liées à la cavéoline 3 – CAV3 related myopathies**

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**Immunofluorescence-Based Analysis of Caveolin-3 in the Diagnostic Management of Neuromuscular Diseases.**

Roos A1, Hathazi D2, Schara U3.

1Department of Neuropediatrics, Developmental Neurology and Social Pediatrics, Centre for Neuromuscular Disorders in Children, University Hospital Essen, University of Duisburg-Essen, Essen, Germany. andreas.roos@uk-essen.de. 2Department of Clinical Neurosciences, University of Cambridge, Cambridge, UK. 3Department of Neuropediatrics, Developmental Neurology and Social Pediatrics, Centre for Neuromuscular Disorders in Children, University Hospital Essen, University of Duisburg-Essen, Essen, Germany.

KEYWORDS: CAV3-related LGMD; Caveolin-3; Caveolinopathy; Immunofluorescence in muscular diseases; Protein analyses in muscular diseases; Rippling muscle disease

PMID: 32548831 DOI:10.1007/978-1-0716-0732-9_18

**Myopathies inflammatoires – Inflammatory myopathies**

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**Comparison of rapamycin and methylprednisolone for treating inflammatory muscle disease in a murine model of experimental autoimmune myositis.**

Kang J1, Feng D1, Yang F1, Tian X1, Han W1, Jia H1,2.

1Department of Neurology, Xijing Hospital, The Fourth Military Medical University, Xincheng, Xi'an, Shaanxi 710032, P.R. China. 2Department of Neurology, Shenzhen Hospital, Southern Medical University, Shenzhen, Guangdong 518034, P.R. China.

KEYWORDS: EAM; IL1Ms; TGF-β; Treg cells; rapamycin

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**Spontaneous symptomatic improvement in a pediatric patient with anti-3-hydroxy-3-methylglutaryl-coenzyme A reductase myopathy.**

Suárez B1, Jofré J2, Lozano-Arango A2, Ortega X3, Diaz J4, Calcagno G5, Bevilacqua JA6, Castiglioni C7.

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**KEYWORDS:** 3-hydroxy-3-methylglutaryl-coenzyme A reductase; HMGCR; Immune-mediated necrotizing-myopathy; LGMD; spontaneous improvement

PMID: 32518057 DOI:10.1016/j.nmd.2020.03.008

105. *Clin Rev Allergy Immunol.*, 2020 Jun 17. doi: 10.1007/s12016-020-08798-2. [Epub ahead of print]

**Type I Interferons in the Pathogenesis and Treatment of Autoimmune Diseases.**

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**KEYWORDS:** Autoimmune disease; Epigenetic modifications; Interferonopathies; Juvenile idiopathic arthritis; Sjogren's syndrome; Systemic lupus erythematosus; Type I interferon signaling pathway

PMID: 32557263 DOI:10.1007/s12016-020-08798-2

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**A Dramatic Presentation of Immune-Mediated Necrotizing Myopathy.**

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PMID: 32558677 DOI:10.1097/RHU.0000000000001443

107. *Ann Rheum Dis.*, 2020 Jun 16. pii: annrheumdis-2019-216599. [Epub ahead of print]

**Machine learning algorithms reveal unique gene expression profiles in muscle biopsies from patients with different types of myositis.**

Pinal-Fernandez I1,2,3, Casas-Dominguez M1,2, Derfoul A1, Pak K1, Miller FW7, Millisenda JC6, Grau-Junyent JM8, Selva-O’Callaghan A1, Carrion-Ribas C1, Pak JP1, Albayda JP1, Christopher-Stine L2,8, Lloyd TE2, Corse AM2, Mammen AL9,2,8.

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KEYWORDS: autoantibodies; autoimmune diseases; autoimmunity; dermatomyositis; polymyositis
PMID: 32546599 DOI: 10.1136/annrheumdis-2019-216599

108. J Rheumatol. 2020 Jun 15. pii: jrheum.200480. doi: 10.3899/jrheum.200480. [Epub ahead of print]
Proceedings of the 2019 Canadian Inflammatory Myopathy Study Symposium: Clinical Trial Readiness in Myositis.

Leclair V1, Landon-Cardinal O1, Aggarwal R1, Bansback N1, Campbell C1, Feldman BM1, Jarry M4, McNamara S1, White B1, Hudson M; CIMS Investigators.
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PMID: 32541080 DOI: 10.3899/jrheum.200480

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Multiple subcutaneous tuberculous abscesses in a dermatomyositis patient without pulmonary tuberculosis: a case report and literature review.

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KEYWORDS: Abscess; Dermatomyositis; Limb; Subcutaneous; Tuberculous
PMID: 32532200 PMCID:PMC7291664 DOI: 10.1186/s12879-020-05137-w Free PMC Article
110. *Autoimmun Rev.*, 2020 Jun 11:102586. doi: 10.1016/j.autrev.2020.102586. [Epub ahead of print]

**Immune checkpoint inhibitor-induced myositis, the earliest and most lethal complication among rheumatic and musculoskeletal toxicities.**

Allenbach Y¹, Anquetil C², Manouchehrí A³, Benveniste O⁴, Lebrun-Vignes B⁵, Spano JP⁶, Ederhy S⁵, Klatzmann D⁴, Rosenzweig M⁵, Fautrel B¹, Cadranel J⁵, Johnson DB², Moslehi J³, Salem JE¹⁰.

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**KEYWORDS:** Adverse drug reactions; Immune checkpoint inhibitors; Myocarditis; Myositis; Pharmacology; Rheumatology

PMID: 32535094 DOI: 10.1016/j.autrev.2020.102586

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**Rheumatic immune-related adverse events associated with cancer immunotherapy: A nationwide multi-center cohort.**

Roberts J¹, Ennis D², Hudson M³, Ye C⁴, Saltman A⁵, Rottapel R⁶, Pope J⁷, Hoa S⁷, Tisseverasinghe A⁸, Fifi-Mah A⁹, Maltez N¹⁰, Jamal S¹¹.

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**Renal disorders in rheumatologic diseases: the spectrum is changing (Part 1: connective tissue diseases).**

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**KEYWORDS:** Lupus nephritis; Malignant hypertension; Myoglobinuria; Polymyositis; Scleroderma; Sjögren syndrome

PMID: 32529559 DOI: 10.1007/s40620-020-00772-7
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*Evaluation of usefulness in surfactant protein D as a predictor of mortality in myositis-associated interstitial lung disease.*
Kaieda S¹, Gono T², Masui K³, Nishina N⁴, Sato S⁵, Kuvana M⁶; A Multicenter Retrospective Cohort of Japanese Patients with Myositis-associated ILD (JAMI) investigators.
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PMID: 32525903 PMCID: PMC7289364 DOI:10.1371/journal.pone.0234523 Free PMC Article

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*MicroRNA and mRNA profiling in the idiopathic inflammatory myopathies.*
Parkes JE¹,², Thoma A³, Lightfoot AP³, Dav P⁴, Chiny H⁵,⁶, Lamb JA¹.
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KEYWORDS: Dermatomyositis; Idiopathic inflammatory myopathies; Polymyositis; RNA sequencing; microRNA
PMID: 32529172 PMCID: PMC7285612 DOI:10.1186/s41927-020-00125-8 Free PMC Article

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*Anti-Ku antibody-positive systemic sclerosis-polymyositis overlap syndrome in an adolescent.*
Loo RJ¹, Nocton JJ², Harmelink MM³, Chiu Y⁴.
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KEYWORDS: anti-Ku antibody; myositis; overlap syndrome; polymyositis; scleroderma
PMID: 32519400 DOI:10.1111/pde.14243

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*[Juvenile dermatomyositis: A series of 22 cases].*
[Article in French]
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KEYWORDS: Dermatomyosite; Dermatomyosite juvénile; Dermatomyositis; Gottron's sign; Juvenile dermatomyositis; Myosite; Myositis; Signe de Gottron
PMID: 32532518 DOI:10.1016/j.annder.2020.04.016
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**Off-label studies on ruxolitinib in dermatology: a review.**
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**KEYWORDS:** Ruxolitinib; alopecia; off-label; psoriasis  
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**Macrophage activation syndrome as a complication of dermatomyositis: A case report.**
Zhu DX1, Qiao JJ1, Fang H2.

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**KEYWORDS:** Case report; Dermatomyositis; Hyperferritinemia; Inflammatory; Macrophage activation syndrome; Systemic juvenile idiopathic arthritis  
**PMID:** 32548165  
**PMCID:** PMC7281037  
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**Recommendations for the treatment of anti-melanoma differentiation-associated gene 5-positive dermatomyositis-associated rapidly progressive interstitial lung disease.**
Romero-Bueno F1, Díaz Del Campo E2, Trailler-Arguás E3, Ruiz-Rodríguez JC4, Castellvi J5, Rodríguez-Nieto MJ6, Martínez-Becerra MJ7, Sanchez-Pernaute O8, Pinal-Fernandez I9, Solanich X9, Gono T10, Gonzalez-Gay MA11, Plaona MN12, Selva-O’Callaghan A13, MEDRAS5 (Spanish MDA5 Register) group (listed contributors at the end of the article).

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**KEYWORDS:** Basiliximab; Cyclophosphamide; Cyclosporine; Dermatomyositis; Extracorporeal membrane oxygenation; Glucocorticoid; Intensive care; Intravenous immunoglobulins; Lung transplant; Mycophenolate; Plasmapheresis; Polymyxin B hemoperfusion; Rapidly progressive interstitial lung disease; Review; Rituximab; Tacrolimus; Tofacitinib; systematic  
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**Abatacept in the Treatment of Juvenile Dermatomyositis-Associated Calcifications in a 16-Year-Old Girl.**
Sukumaran S1, Vijayan V2.

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Maladie de Pompe – Pompe disease

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6Genetics Research Center, University of Social Welfare and Rehabilitation Sciences, Tehran, Iran.

KEYWORDS: Inherited metabolic disorders; Lipid Storage Myopathy; Metabolic Myopathy; Neuromuscular diseases; VLCAD Deficiency; Very Long-Chain Acyl-CoA Dehydrogenase

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1Department of Pediatrics, Prince Sultan Military Medical City, Riyadh, Kingdom of Saudi Arabia. E-mail. aalhashem@psmmc.med.sa.

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**Late-onset MELAS syndrome with mtDNA 14453G→A mutation masquerading as an acute encephalitis: a case report.**

Yokota Y1, Hara M2, Akimoto T1, Mizoguchi T1, Goto Y1,2,3,4, Nishino I5, Kamei S1,6, Nakaima H1.

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KEYWORDS: Encephalitis; Late-onset; MELAS; ND6 gene

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Wang S1, Song T, Wang S.

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KEYWORDS: MERRF; ataxia; dorsal root ganglia; ganglionopathy; myopathy

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1Department of Pharmacology, University of Minnesota, Minneapolis, MN 55455, USA.

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1Section on Developmental Neurobiology, National Institute of Neurological Disorders and Stroke, National Institutes of Health, Bethesda, MD, United States.

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1Division of Neuromuscular Medicine, Department of Neurology, Mayo Clinic, Rochester, Minnesota.
2Division of Speech Pathology, Department of Neurology, Mayo Clinic, Rochester, Minnesota.
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3Department of Neurology, Renji Hospital Shanghai Jiaotong University School of Medicine, Shanghai, China. Electronic address: yangtaiguan@sina.com.
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Sequential targeted exome sequencing of 1001 patients affected by unexplained limb-girdle weakness.
Töpf A1, Johnson K1,2, Bates A1, Phillips L1, Chao KR3,4, England EM3,4, Laricchia KM3,4, Mullen T3,4, Valkanas E3,4, Xu L3,4, Bertoli M1, Blain A1, Casasús AB1, Duff J1, Mroczek M1, Specht S1, Lek M3,4,6, Ensini M1,7, MacArthur DG3,4,8,9; MYO-SEQ consortium, Straub V10.
Collaborators (79)
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volker.straub@ncl.ac.uk.
KEYWORDS: genetic diagnosis; limb-girdle weakness; neuromuscular disease; next-generation sequencing; targeted exome analysis
PMID: 32528171 DOI:10.1038/s41436-020-0840-3

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Measures of nocturnal oxyhemoglobin desaturation in children with neuromuscular disease or Prader-Willi syndrome.
Kaditis AG1, Polytarchou A1, Moudaki A1, Panagiotopoulou-Gartagani P1, Kanaka-Gantenbein C1.
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KEYWORDS: central sleep apnea; desaturation; nocturnal hypoxemia; obstructive sleep apnea
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Exercise-Related Oxidative Stress as Mechanism to Fight Physical Dysfunction in Neuromuscular Disorders.
Siciliano G1, Chico L1, Lo Gerfo A1, Simoncini C1, Schirinzì E1, Ricci G1.
1Department of Clinical and Experimental Medicine, Neurological Clinic, University of Pisa, Pisa, Italy.
KEYWORDS: aerobic and anaerobic exercise; neuromuscular diseases; oxidative stress; physical exercise training; quality of life
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Morphological variability is greater at developing than mature mouse neuromuscular junctions.
Mech AM1, Brown AL1, Schiavo G2,3, Sleeth JN2,4.
1Department of Neuromuscular Diseases, UCL Queen Square Institute of Neurology, University College London, London, UK.
2UK Dementia Research Institute, University College London, London, UK.
3Discoveries Centre for Regenerative and Precision Medicine, University College London Campus, London, UK.
KEYWORDS: NMJ-morph; epitrochleoanconeus; fast twitch; flexor digitorum brevis; lumbricals; morphology; motor neuron; muscle fibre type; neuromuscular junction; slow twitch; synapse; transversus abdominis
PMID: 32533580 DOI:10.1111/joa.13228

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Profiling and Functional Analysis of Circular RNAs in Porcine Fast and Slow Muscles.
Li B1, Yin D2, Li P2, Zhang Z3, Zhang X1, Li H1,2, Li P2, Hou L1, Liu H2, Wu W1.
1College of Animal Science and Veterinary Medicine, Shenyang Agricultural University, Shenyang, China.
159. *Muscle Nerve*. 2020 Jun 12. doi: 10.1002/mus.27000. [Epub ahead of print]

**Quantitative electromyography: Normative data in paraspinal muscles.**

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3Dep. of Neurology Unit, AUSL-IRCCS di Reggio Emilia, Reggio Emilia, Italy.
4Dep. of Clinical Neurophysiology, Rigshospitalet, University of Copenhagen.

KEYWORDS: Anterior horn cell disease; EMG; Head drop; Myopathy; Neuromuscular disorder; Normative data; Paraspinal muscle; Spinal segment; quantitative electromyography

PMID: 32530492 DOI:10.1002/mus.27000

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**Spatially regularized parametric map reconstruction for fast magnetic resonance fingerprinting.**

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KEYWORDS: Convolutional neural network; Image reconstruction; Magnetic resonance fingerprinting; Quantitative magnetic resonance imaging

PMID: 32544842 DOI:10.1016/j.media.2020.101741 Free full text
Some of citations presented by diseases are sorted below by specialties.

### Anatomopathologie – Anatomical pathology

**Ann Rheum Dis.** 2020 Jun 16. pii: annrheumdis-2019-216599. [Epub ahead of print]

**Machine learning algorithms reveal unique gene expression profiles in muscle biopsies from patients with different types of myositis.**

Pinal-Fernandez I1,2,3, Casal-Dominguez M1,2, Derfoul A1, Pak K1, Miller FW5, Milisenda JC6, Grau-Junyent JM4, Selva-O'Calleghan A1, Carron-Ribas C1, Paik JJ2, Albadia J3, Christopher-Stine L2,3, Lloyd TE2, Corse AM2, Mammen AL2,3,8,9.

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**KEYWORDS:** autoantibodies; autoimmune diseases; autoimmunity; dermatomyositis; polymyositis

PMID: 32546599 DOI: 10.1136/annrheumdis-2019-216599

**J Neuropathol Exp Neurol.** 2020 Jul 1;79(7):719-733. doi: 10.1093/jnen/nlaa046.

**What Every Neuropathologist Needs to Know: The Muscle Biopsy.**

Nix JS1, Moore SA2.

1Department of Pathology, Johns Hopkins University School of Medicine, Baltimore, Maryland.

2Department of Pathology, Carver College of Medicine, The University of Iowa, Iowa City, Iowa.

**KEYWORDS:** Inflammatory myopathy; Muscle biopsy; Muscular dystrophy; Myopathy; Neurogenic atrophy

PMID: 32529201 DOI: 10.1093/jnen/nlaa046

### Cardiologie – Cardiology

**Heart Rhythm.** 2020 Jun 7. pii: S1547-5271(20)30546-4. [Epub ahead of print]

**Evaluation of mexiletine effect on conduction delay and bradyarrhythmic complications in patients with myotonic dystrophy type 1 over long-term follow-up.**

Vio R1, Zorzi A1, Bello L2, Boffozzoni V2, Botta A1, Rivezzoli F1, Leonori L1, Migliore F1, Bertaglia E1, Iliceto S1, Pegoraro E2, Corrado D1, Calore C1.

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2Department of Neuroscience, University of Padova, Padova, Italy.

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4Department of Cardiac, Thoracic and Vascular Sciences and Public Health, University of Padova, Padova, Italy.

Electronic address: domenico.corrado@unipd.it.

**KEYWORDS:** antiarrhythmic drugs; atrioventricular block; bundle branch block; myotonic dystrophy; neuromuscular disorder; pacemaker; sudden cardiac death

PMID: 32525073 DOI: 10.1016/j.hrthm.2020.05.043

**Mol Cell Proteomics.** 2020 Jun 15. pii: mcp.RA120.002071. [Epub ahead of print]

**Kir2.1 interactome mapping uncovers PKP4 as a modulator of the Kir2.1-regulated inward rectifier potassium currents.**

Park SS1, Ponce-Balbuena D2, Kuick R3, Guerrero-Serna G4, Yoon J5, Mellacheruvu D6, Conlon KP7, Basrur V8, Nesvizhskii AI9, Jalife J4, Rual JF10.

1University of Michigan, Dept. of Pathology, United States.

2University of Michigan, Dept. of Internal Med., United States.

3University of Michigan.
Rimeporide as a first-in-class NHE-1 inhibitor: Results of a phase Ib trial in young patients with Duchenne Muscular Dystrophy.

Previtalli SC1, Gidaro T2, Diaz-Manera J3, Zambon A1, Camesecchi S4, Roux-Lombard P5, Spitali P6, Signorelli M6, Szigwarto CA7, Johansson C8, Gray J9, Labolle D9, Thomé FP10, Pitchforth J11, Domingos J11, Muntoni F12.

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KEYWORDS: Cardiomyopathy; Duchenne Muscular Dystrophy; NHE-1; Pharmacokinetic; Rimeporide; Safety

PMID: 32535224 DOI:10.1016/j.phrs.2020.104999

PTX3 Predicts Myocardial Damage and Fibrosis in Duchenne Muscular Dystrophy.

Farini A1, Villa C1, Di Silvestre D2, Bella P1, Tripodi L1, Rossi R1, Sitzia C1, Gatti S1, Mauni P1, Torrente Y1.

1Stem Cell Laboratory, Department of Pathophysiology and Transplantation, Università degli Studi di Milano, Unit of Neurology, Fondazione IRCCS Ca’ Granda Ospedale Maggiore Policlinico, Centro Dino Ferrari, Milan, Italy. 2Institute of Technologies in Biomedicine, National Research Council (ITB-CNR), Milan, Italy. 3Residency Program in Clinical Pathology and Clinical Biochemistry, Università degli Studi di Milano, Milan, Italy. 4Center for Surgical Research, Fondazione IRCCS Ca’ Granda, Ospedale Maggiore Policlinico, Milan, Italy.

KEYWORDS: Duchenne muscular dystrophy (DMD); alarmins; cardiomyopathy; muscular dystrophy; pentraxin 3 (PTX3)

PMID: 32508664 PMCID: PMC7248204 DOI:10.3389/fphys.2020.00403 Free PMC Article

Multisystem Myotilinopathy, including Myopathy and Left Ventricular Noncompaction, due to the MYOT Variant c.179C>T.

Finsterer J1, Stöllberger C2, Hasun M2, Riedhammer K3,4, Wagner M5.

1Krankenanstalt Rudolfstiftung, Messerli Institute, Vienna, Austria. 22nd Medical Department with Cardiology and Intensive Care Medicine, Krankenanstalt Rudolfstiftung, Vienna, Austria. 3Institute of Human Genetics, Germany. 4Department of Nephrology, Klinikum Rechts der Isar, School of Medicine, Technical University of Munich, Munich, Germany.

PMID: 32509353 PMCID:PMC7244945 DOI:10.1155/2020/5128069 Free PMC Article
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*J Bodyw Mov Ther*. 2020 Apr;24(2):130-137. doi: 10.1016/j.jbmt.2019.10.014. Epub 2019 Oct 25.

**Massage therapy treatment and outcomes in a patient with Charcot-Marie-Tooth disease: A case report.**

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KEYWORDS: Charcot-Marie-Tooth; Massage therapy; Peripheral neuropathy

PMID: 32507138 DOI:10.1016/j.jbmt.2019.10.014

**Électromyographie – Electromyography**

*Respir Care*. 2020 Jun 16. pii: respcare.07426. doi: 10.4187/respcare.07426. [Epub ahead of print]

**Effects of Positioning on Cough Peak Flow and Muscular Electromyographic Activation in Duchenne Muscular Dystrophy.**

Marques L.¹,², de Freitas Fregonezi GA.¹,², Santos JP.¹,², Marcelino AA.¹,², Medeiros da Fonseca JD.¹,², Dourado-Júnior MET.³, Aliverti A.⁴, Sarmento A.¹,², Resqueti VR.¹,²

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KEYWORDS: Duchenne muscular dystrophy; cough; electromyography; plethysmography; respiratory muscles

PMID: 32546537 DOI:10.4187/respcare.07426

**Muscle Nerve**. 2020 Jun 12. doi: 10.1002/mus.27000. [Epub ahead of print]

**Quantitative electromyography: Normative data in paraspinal muscles.**

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³Dep. of Neurology Unit, AUSL-IRCCS di Reggio Emilia, Reggio Emilia, Italy.

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KEYWORDS: Anterior horn cell disease; EMG; Head drop; Myopathy; Neuromuscular disorder; Normative data; Paraspinal muscle; Spinal segment; quantitative electromyography

PMID: 32530492 DOI:10.1002/mus.27000

**J Electromyogr Kinesiol**. 2020 Jun 9;53:102437. doi: 10.1016/j.jelekin.2020.102437. [Epub ahead of print]

**Lambert-Eaton myasthenia syndrome: specified description of a response pattern to low-frequency repetitive nerve stimulation.**

Zhou X.¹, Wang Z.¹, Zhu Y.¹, Zhu D.¹, Xie C.¹, Guan Y.³

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KEYWORDS: Electrophysiology; Lambert-Eaton myasthenia syndrome; Low-frequency decrement; Myasthenia gravis; Neuromuscular disorder; Repetitive nerve stimulation

PMID: 32554206 DOI:10.1016/j.jelekin.2020.102437
**Gastroentérologie / Nutrition – Gastroenterology / Nutrition**

**Myopathies featuring early or prominent dysphagia.**

Triplett JD, Pinto MV, Hosfield EA, Milone M, Liewluck T.

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2Division of Speech Pathology, Department of Neurology, Mayo Clinic, Rochester, Minnesota.

KEYWORDS: dysphagia; inclusion body myositis; muscular dystrophy; myopathy; myositis; videofluoroscopy

PMID: 32510670 DOI: 10.1002/mus.26996

**Imagerie médicale – Medical imaging**

**Diagnostic Value of Magnetic Resonance Imaging in Fibrodysplasia Ossificans Progressiva.**

Botman E, Teunissen BP, Rajmakers P, de Graaf P, Yaqub M, Treurniet S, Van Ommen G, Bravenboer N, Micha D, Pals G, Bökenkamp A, Neteleinbos JC, Lammertsma AA, Eekhoff EM.

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KEYWORDS: ANALYSIS/QUANTITATION OF BONECLINICAL TRIALSDISEASES AND DISORDERS OF/RELATED TO BONEFIBRODYSPLASIA OSSIFICANS PROGRESSIVARADIOLOGY

PMID: 32537549 PMCID: PMC7285757 DOI: 10.1002/jbm4.10363 Free PMC Article

**Neurology.**

**Novel approaches to quantify CNS involvement in children with Pompe disease.**

Korlimarla A, Spiridigliozzi GA, Crisp K, Herbert M, Chen S, Malinzak M, Stefanescu M, Austin SL, Cope H, Zimmerman K, Jones H, Provenzale JM, Kishnani PS.

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PMID: 32518148 DOI: 10.1212/WNL.0000000000009979

**Med Image Anal.**

**Spatially regularized parameteric map reconstruction for fast magnetic resonance fingerprinting.**

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PMID: 32518148 DOI: 10.1016/j.media.2020.101741
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KEYWORDS: Convolutional neural network; Image reconstruction; Magnetic resonance fingerprinting; Quantitative magnetic resonance imaging
PMID: 32544842 DOI:10.1016/j.media.2020.101741 Free full text

BMJ Case Rep. 2020 Jun 11;13(6). pii: e236444. doi: 10.1136/bcr-2020-236444.

Use of muscle MRI in an atypical presentation of FSHD2.
Jesuthasan A1, Shah S2, Morrow JM3.
1University College Hospital, University College London Hospitals NHS Foundation Trust, London, UK.
2Lysholm Department of Neuroradiology, National Hospital for Neurology and Neurosurgery, London, UK.
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KEYWORDS: neurology; neuromuscular disease
PMID: 32532898 DOI:10.1016/j.bcr-2020-236444

Médecine physique et de réadaptation – Physical and rehabilitation medicine

J Bodyw Mov Ther. 2020 Apr;24(2):85-91. doi: 10.1016/j.jbmt.2019.09.009. Epub 2019 Oct 4.

An adapted dance program for children with Charcot-Marie-Tooth disease: An exploratory study.
Cherriere C1, Martel M2, Fortin S3, Raymond MJ4, Veilleux LN5, Lemay M6.
1Centre de Réadaptation Marie Enfant, Centre de Recherche du CHU Sainte Justine, Montréal, Canada; ToNiC Toulouse Neuroimaging Center, Université de Toulouse, Inserm, UPS, Toulouse, France. Electronic address: claire.cherriere@inserm.fr.
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PMID: 32507158 DOI:10.1016/j.jbmt.2019.09.009

J Bodyw Mov Ther. 2020 Apr;24(2):130-137. doi: 10.1016/j.jbmt.2019.10.014. Epub 2019 Oct 25.

Massage therapy treatment and outcomes in a patient with Charcot-Marie-Tooth disease: A case report.
Paz G1.
1Grant MacEwan University, Massage Therapy Program, Canada. Electronic address: galitip@gmail.com.

KEYWORDS: Charcot-Marie-Tooth; Massage therapy; Peripheral neuropathy
PMID: 32507138 DOI:10.1016/j.jbmt.2019.10.014

J Neurosci Nurs. 2020 Jun 5. doi: 10.1097/JNN.0000000000000519. [Epub ahead of print]

Assessing Motor Function in Congenital Muscular Dystrophy Patients Using Accelerometry.
Lawal TA1, Todd JJ, Elliott JS, Linton MM, Andres M, Witherspoon JW, Collins JP, Chrismer IC, Tounkara F, Waite MR, Nichols C, Bönnemann CG, Vuillerot C, Bendixen R, Jain MS, Meilleur KG.
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PMID: 32511172 DOI:10.1097/JNN.0000000000000519

Front Physiol. 2020 May 21;11:349. doi: 10.3389/fphys.2020.00349. eCollection 2020.

**Aerobic Exercise Training in Patients With mtDNA-Related Mitochondrial Myopathy.**

Jeppesen TD1.
1Copenhagen Neuromuscular Clinic, Department of Neurology, Rigshospitalet, University of Copenhagen, Copenhagen, Denmark.

KEYWORDS: mitochondrial DNA; mitochondrial myopathies; oxidative capacity; training; treatment
PMID: 32508662 PMCID: PMC7253634 DOI:10.3389/fphys.2020.00349 Free PMC Article

Front Physiol. 2020 May 20;11:451. doi: 10.3389/fphys.2020.00451. eCollection 2020.

**Exercise-Related Oxidative Stress as Mechanism to Fight Physical Dysfunction in Neuromuscular Disorders.**

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KEYWORDS: aerobic and anaerobic exercise; neuromuscular diseases; oxidative stress; physical exercise training; quality of life
PMID: 32508674 PMCID:PMC7251329 DOI:10.3389/fphys.2020.00451 Free PMC Article

Nephrologie – Nephrology

J Nephrol. 2020 Jun 11. doi: 10.1007/s40620-020-00772-7. [Epub ahead of print]

**Renal disorders in rheumatologic diseases: the spectrum is changing (Part 1: connective tissue diseases).**

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KEYWORDS: Lupus nephritis; Malignant hypertension; Myoglobinuria; Polymyositis; Scleroderma; Sjögren syndrome
PMID: 32529559 DOI:10.1007/s40620-020-00772-7

Optalmologie – Ophthalmology

BMC Neurol. 2020 Jun 11;20(1):238. doi: 10.1186/s12883-020-01805-1.

**Prediction of generalization of ocular myasthenia gravis under immunosuppressive therapy in Northwest China.**

Ding J1, Zhao S1, Ren K1, Dang D2, Li H1, Wu F3, Zhang M1, Li Z4, Guo J5.
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KEYWORDS: Generalization; Immunosuppressive therapy; Myasthenia gravis; Ocular; Predictive factor
PMID: 32527235 PMCID:PMC7288410 DOI:10.1186/s12883-020-01805-1 Free PMC Article

Eur Neurol. 2020 Jun 11;1:7. doi: 10.1159/000507853. [Epub ahead of print]

**Adult Ocular Myasthenia Gravis Conversion: A Single-Center Retrospective Analysis in China.**

Feng X1, Huan X1, Yan C1, Song J1, Lu J1, Zhou L1, Wu H2, Qiao K3, Lu J1, Xi J1, Luo S1, Zhao C1.
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KEYWORDS: Adult myasthenia gravis; Anti-acetylcholine receptor antibody; Conversion; Ocular myasthenia gravis; Risk factors
PMID: 32526733 DOI:10.1159/000507853

Pneumologie – Pulmonogy

Effects of Positioning on Cough Peak Flow and Muscular Electromyographic Activation in Duchenne Muscular Dystrophy.
Marques L1,2, de Freitas Fregonezi GA1,2, Santos IP1,2, Marcelino AA1,2, Medeiros da Fonsêca JD1,2, Dourado-Júnior MET3, Alverti A4, Sarmento A1,2, Resqueti VR4,2.
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KEYWORDS: Duchenne muscular dystrophy; cough; electromyography; plethysmography; respiratory muscles
PMID: 32546537 DOI:10.4187/respcare.07426

Evaluation of usefulness in surfactant protein D as a predictor of mortality in myositis-associated interstitial lung disease.
Kaieda S1, Gono T2, Masui K3, Nishina N4, Sato S5, Kuwana M2; A Multicenter Retrospective Cohort of Japanese Patients with Myositis-associated ILD (JAMI) investigators.
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PMID: 32525903 PMCID:PMC7289364 DOI:10.1371/journal.pone.0234523 Free PMC Article

Anti-Signal Recognition Particle Antibody-Associated Severe Interstitial Lung Disease Requiring Lung Transplantation.
Qureshi A1, Brown D2, Brent L3.
1Internal Medicine, Reading Hospital - Tower Health, West Reading, USA.
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3Rheumatology, Temple University Hospital, Philadelphia, USA.
KEYWORDS: anti-signal recognition particle; interstitial lung disease; lung transplantation; myopathy
PMID: 32523819 PMCID:PMC7273426 DOI:10.7759/cureus.7962 Free PMC Article

Respiratory function during enzyme replacement therapy in late-onset Pompe disease: longitudinal course, prognostic factors, and the impact of time from diagnosis to treatment start.
Stockton DW1, Kishnani P2, van der Ploeg A3, Llerena J Jr4, Boentert M5, Roberts M6, Byrne BJ7, Araujo R8, Maruti SS8, Thibaut N9, Verhulst K9, Berger K10.
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KEYWORDS: Alglucosidase alfa; Enzyme replacement therapy; Late-onset Pompe disease; Pompe disease; Registry; Respiratory function

PMID: 32524257 DOI: 10.1007/s00415-020-09936-8

Semin Arthritis Rheum. 2020 Jun 1;50(4):776-790. doi: 10.1016/j.semarthrit.2020.03.007. [Epub ahead of print]
Recommendations for the treatment of anti-melanoma differentiation-associated gene 5-positive dermatomyositis-associated rapidly progressive interstitial lung disease. Romero-Bueno F1, Diaz Del Campo D1, Trallero-Araguas E2, Ruiz-Rodriguez JC3, Castellvi P4, Rodriguez-Nieto MJ5, Martinez-Becerra MJ6, Sanchez-Fernaute O7, Pinal-Fernandez I8, Solanich X9, Sono T10, Gonzalez-Gay MA11, Pina M12, Selva-O’Callaghan A13. MEDRAS (Spanish MDA5 Register) group (listed contributors at the end of the article).
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13Systemic Autoimmune Diseases Unit. Medicine Dept. Vall d’Hebron University Hospital, GEAS group. Universitat Autònoma de Barcelona, Barcelona, Spain. Electronic address: aselva@vhebron.net.

KEYWORDS: Basiliximab; Cyclophosphamide; Cyclosporine; Dermatomyositis; Extracorporeal membrane oxygenation; Glucocorticoid; Intensive care; Intravenous immunoglobulins; Lung transplant; Mycophenolate; Plasmapheresis; Polymixin B hemoperfusion; Rapidly progressive interstitial lung disease; Review; Rituximab; Tacrolimus; Tofacitinib; systematic

PMID: 32534273 DOI: 10.1016/j.semarthrit.2020.03.007

Braz J Phys Ther. 2020 Jun 5. pii: S1413-3555(19)30364-8. [Epub ahead of print]
Pulmonary and upper limbs function in children with early stage Duchenne muscular dystrophy compared to their healthy peers. Bulut N6, Aydin O2, Alemdaroğlu-Gürbüz P1, Karaduman A2, Yilmaz O2.
1Faculty of Physical Therapy and Rehabilitation, Hacettepe University, Ankara, Turkey. Electronic address: nmn60_90@hotmail.com.
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KEYWORDS: Functional performance; Pulmonary function test; Rehabilitation; Upper limb

PMID: 32553415 DOI: 10.1016/j.bjpt.2020.05.012

Pediatr Pulmonol. 2020 Jun 11. doi: 10.1002/ppul.24899. [Epub ahead of print]
Measures of nocturnal oxyhemoglobin desaturation in children with neuromuscular disease or Prader-Willi syndrome.
Kaditis AG1, Polyarchou A1, Moudaki A1, Panagiotopoulou-Gartaganis P1, Kanaka-Gantenbein C1.

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1Division of Pediatric Pulmonology and Sleep Disorders Laboratory, First Department of Pediatrics, National and Kapodistrian University of Athens School of Medicine and Aghia Sophia Children's Hospital, Athens, Greece.
KEYWORDS: central sleep apnea; desaturation; nocturnal hypoxemia; obstructive sleep apnea
PMID: 32525614 DOI:10.1002/ppul.24899