Resource Summary Report

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Washington University Neuromuscular Disease Center

RRID:SCR_002059
Type: Tool

Proper Citation

Washington University Neuromuscular Disease Center (RRID:SCR_002059)

Resource Information

URL: http://neuromuscular.wustl.edu/

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Description: Organization portal for neuromuscular disease community and contains comprehensive listing of biological and clinical aspects of neuromuscular disorders.This knowledge base contains information on the physiology, structure of ion channels, neurotransmitters, neuroreceptors, and associated diseases. Major categories include DISORDERS & SYNDROMES, INDEXES, NEUROMUSCULAR EVALUATION, ANTIBODY TESTING and NEUROMUSCULAR DIVISION.

Abbreviations: WU Neuromuscular

Synonyms: Neuromuscular Disease Center

Resource Type: portal, data or information resource, organization portal

Keywords: neuromuscular disease, cerebellum, motor cortex, spinal cord

Availability: Free, Freely available

Resource Name: Washington University Neuromuscular Disease Center

Resource ID: SCR_002059

Alternate IDs: nif-0000-00158, SCR_007305, nif-0000-12503

Alternate URLs: http://neuromuscular.wustl.edu/over/overview.html

Old URLs: http://www.neuro.wustl.edu/neuromuscular/

Record Creation Time: 20220129T080211+0000

Record Last Update: 20260821T042535+0000

Ratings and Alerts

No rating or validation information has been found for Washington University Neuromuscular Disease Center.

No alerts have been found for Washington University Neuromuscular Disease Center.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 25 mentions in open access literature.

Listed below are recent publications. The full list is available at dkNET .

Ma X, et al. (2023) Screening for PRX mutations in a large Chinese Charcot-Marie-Tooth disease cohort and literature review. Frontiers in neurology, 14, 1148044.

Ando M, et al. (2022) Novel de novo POLR3B mutations responsible for demyelinating Charcot-Marie-Tooth disease in Japan. Annals of clinical and translational neurology, 9(5), 747.

Parissis D, et al. (2022) Genetic causes of acute encephalopathy in adults: beyond inherited metabolic and epileptic disorders. Neurological sciences : official journal of the Italian Neurological Society and of the Italian Society of Clinical Neurophysiology, 43(3), 1617.

Nan H, et al. (2022) Coexistence of Charcot-Marie-Tooth 1A and nondystrophic myotonia due to PMP22 duplication and SCN4A pathogenic variants: a case report. BMC neurology, 22(1), 17.

Ando M, et al. (2022) Novel heterozygous variants of SLC12A6 in Japanese families with Charcot-Marie-Tooth disease. Annals of clinical and translational neurology, 9(7), 902.

Bartikoski BJ, et al. (2022) Protocol to segment and characterize the morphometry of individual cross-sectioned striated muscle fibers from??adult murine models. STAR protocols, 3(4), 101855.

Yuan JH, et al. (2022) Multi-type RFC1 repeat expansions as the most common cause of hereditary sensory and autonomic neuropathy. Frontiers in neurology, 13, 986504.

Jacquier A, et al. (2022) Microrchidia CW-Type Zinc Finger 2, a Chromatin Modifier in a Spectrum of Peripheral Neuropathies. Frontiers in cellular neuroscience, 16, 896854.

Ando M, et al. (2022) Comprehensive Genetic Analyses of Inherited Peripheral Neuropathies in Japan: Making Early Diagnosis Possible. Biomedicines, 10(7).

Stavrou M, et al. (2021) Genetic mechanisms of peripheral nerve disease. Neuroscience letters, 742, 135357.

Vavouraki N, et al. (2021) Integrating protein networks and machine learning for disease stratification in the Hereditary Spastic Paraplegias. iScience, 24(5), 102484.

Dong HL, et al. (2021) Biallelic SORD pathogenic variants cause Chinese patients with distal hereditary motor neuropathy. NPJ genomic medicine, 6(1), 1.

Hentschel A, et al. (2021) Protein signature of human skin fibroblasts allows the study of the molecular etiology of rare neurological diseases. Orphanet journal of rare diseases, 16(1), 73.

Shi J, et al. (2021) Whole-exome sequencing identifies a heterozygous mutation in SLC12A6 associated with hereditary sensory and motor neuropathy. Neuromuscular disorders : NMD, 31(2), 149.

Kitani-Morii F, et al. (2020) Recent Advances in Drosophila Models of Charcot-Marie-Tooth Disease. International journal of molecular sciences, 21(19).

Chen Y, et al. (2020) TGM6 L517W is not a pathogenic variant for spinocerebellar ataxia type 35. Neurology. Genetics, 6(3), e424.

Xie Y, et al. (2020) Genetic and Clinical Features in 24 Chinese Distal Hereditary Motor Neuropathy Families. Frontiers in neurology, 11, 603003.

Lai LL, et al. (2020) Novel CAPN1 mutations extend the phenotypic heterogeneity in combined spastic paraplegia and ataxia. Annals of clinical and translational neurology, 7(10), 1862.

Mauri N, et al. (2017) A SINE Insertion in ATP1B2 in Belgian Shepherd Dogs Affected by Spongy Degeneration with Cerebellar Ataxia (SDCA2). G3 (Bethesda, Md.), 7(8), 2729.

Blaeser A, et al. (2016) Progressive Dystrophic Pathology in Diaphragm and Impairment of Cardiac Function in FKRP P448L Mutant Mice. PloS one, 11(10), e0164187.