Congenital Muscular Dystrophy

Last updated: May 08, 2013
Years published: 2007, 2013

Acknowledgment

NORD gratefully acknowledges Susan E. Sparks, MD, PhD, Clinical Genetics/Department of Pediatrics, Levine Children's Hospital at Carolinas Medical Center, for assistance in the preparation of this report.

Disease Overview

Summary

Congenital muscular dystrophy (CMD) is a general term for a group of genetic muscle diseases that occur at birth (congenital) or early during infancy. CMDs are generally characterized by diminished muscle tone (hypotonia), which is sometimes referred to as "floppy baby"; progressive muscle weakness and degeneration (atrophy); abnormally fixed joints that occur when thickening and shortening of tissue such as muscle fibers cause deformity and restrict the movement of an affected area (contractures); spinal rigidity, and delays in reaching motor milestones such as sitting or standing unassisted. Feeding difficulties and breathing (respiratory) complications can develop in some cases. Muscle weakness may improve, remain stable or worsen. Some forms of CMD may be associated with structural brain defects and, potentially, intellectual disability. The severity, specific symptoms, and progression of these disorders vary greatly. Most forms of CMD are inherited as autosomal recessive traits. Collage type VI-related disorders can be inherited as either autosomal dominant or autosomal recessive conditions. LMNA-related CMD is inherited in an autosomal dominant manner, with all

muscles and have different ages of onset, severity and inheritance patterns. As researchers have

learned more about the CMDs, such as identifying many of the specific genes involved, a broader picture of these diseases has emerged. The subtypes of CMD have considerable overlap with other disease classifications including the congenital myopathies, disorders of glycosylation, and the limb-girdle muscular dystrophies. CMDs are a rapidly growing disease family and information about these disorders is constantly changing.

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Programs & Resources

RareCare® Assistance Programs

Accepting Applications

Phone: 866-218-8172 | Email: DMD@rarediseases.org | Fax: 203-263-9597

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Phone: 866-218-8172 | Email: DMD@rarediseases.org | Fax: 203-263-9597

PTC 124 GD-016-DMD

Referral Required

Phone: 866-647-9325 | Email: PTC-016-DMD@rarediseases.org | Fax: 203-674-9937

Additional Assistance Programs

MedicAlert Assistance Program

NORD and MedicAlert Foundation have teamed up on a new program to provide protection to rare disease patients in emergency situations.
https://rarediseases.org/patient-assistance-programs/medicalert-assistance-program/

Rare Caregiver Respite Program

This first-of-its-kind assistance program is designed for caregivers of a child or adult diagnosed with a rare disorder.
https://rarediseases.org/patient-assistance-programs/caregiver-respite/

Patient Organizations

Child Neurology Foundation

NORD Member

Email: info@childneurologyfoundation.org

https://rarediseases.org/organizations/child-neurology-foundation/

Muscular Dystrophy Association

NORD Member

Phone: 800-572-1717 | Email: resourcecenter@mdausa.org

https://rarediseases.org/organizations/muscular-dystrophy-association/

Cure CMD (Congenital Muscular Dystrophy)

Email: info@curecmd.org

https://rarediseases.org/non-member-patient/cure-cmd-congenital-muscular-dystrophy/

European Alliance of Neuromuscular Disorders Associations

Email: eamda@hotmail.com

https://rarediseases.org/non-member-patient/european-alliance-of-neuromuscular-disorders-associations/

Global FKRP Registry

Email: coordinator@fkrp-registry.org

NIH/National Institute of Arthritis and Musculoskeletal and Skin Diseases

Phone: 301-495-4484 | Email: NIAMSinfo@mail.nih.gov | Fax: 301-718-6366

https://rarediseases.org/non-member-patient/nih-national-institute-of-arthritis-and-musculoskeletal-and-skin-diseases/

NIH/National Institute of Neurological Disorders and Stroke

Phone: 301-496-5751 | Fax: 301-402-2186

https://rarediseases.org/non-member-patient/nih-national-institute-of-neurological-disorders-and-stroke/

Society for Muscular Dystrophy Information International

Email: smdi@auracom.com

https://rarediseases.org/non-member-patient/society-for-muscular-dystrophy-information-international/
