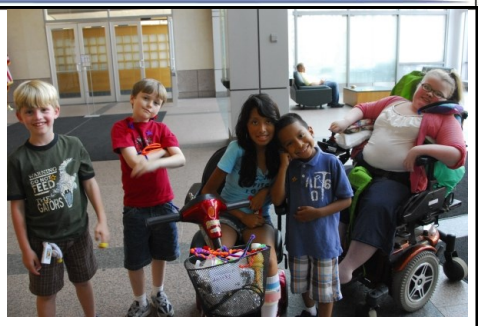
# CMDIR

## Congenital Muscle Disease International Registry

*Leading the Way to a Treatment and Cure*



The Congenital Muscle Disease International Registry (CMDIR) was created to identify the global congenital muscle disease population for the purpose of raising awareness, standards of care, clinical trials and in the future a treatment or cure.

Simply put, we will not be successful in finding a treatment or cure unless we know who the affected individuals are, what the diagnosis is and how the disease is affecting the individual.

The registry includes demographic, disease specific and diagnostic questions. The CMDIR has online help and genetic counselors you can access if you have questions. If you do not yet have genetic confirmation of disease, CMDIR counselors may be able to assist in finding a testing lab to help you determine your genetic mutation.

The registry includes people with Congenital Muscular Dystrophy, Congenital Myopathy, and Congenital Myasthenic Syndrome, and registers through the late onset spectrum for these disease groups. You do not have to have a genetic diagnosis to participate. Visit the CMDIR website to see the complete list of diagnoses registered.

## Register Today

**Registered affected individuals are required for:**

⇒ Clinical trials

⇒ Pharmaceutical investment

⇒ Government intervention and funding

⇒ Awareness and advocacy

**By registering you will also receive:**

⇒ CMDIR annual newsletter

⇒ Consensus Guidelines on Standards of Care

⇒ Notice of available clinical trials that apply to your registered profile

⇒ Notice of available therapies that apply to your registered profile when they become available

**To register, please visit**

**www.cmdir.org**


For more information please contact

Rachel Alvarez, Associate Director

| | |
|---|---|
| CMDIR | counselor@cmdir.org |
| 1712 Pelican Ave. | (323) 250-2399 |
| San Pedro, CA 90732 USA | www.cmdir.org |


CURE CMD

Congenital Muscular Dystrophy

Joshua Frase FOUNDATION

A Foundation BUILDING STRENGTH For Nemaline Myopathy

# CMDIR

## Congenital Muscle Disease International Registry

*Leading the Way to a Treatment and Cure*

**Congenital Muscular Dystrophy subtypes included in the CMDIR**

- Alpha 7/Alpha 9 Integrin Related Myopathy
- Collagen VI Related Myopathy (Ullrich through Bethlem CMD)
- Alpha-Dystroglycan Related Muscular Dystrophy (Dystroglycanopathy, WWS, MEB, Fukuyama, FKRP, LGMD2I, LGMD2K, LGMD2M, LGMD2N, LGMD2O)
- Choline Kinase B Receptor
- LAMA2 Related Muscular Dystrophy (Laminin Alpha 2 related dystrophy/MDC1A/Merosin deficient)
- LMNA Related Muscular Dystrophy (Laminopathy/LaminA/C, L-CMD, Emery Dreifuss muscular dystrophy)
- RYR1 Related Myopathy (with dystrophic presentation, including Malignant Hyperthermia, Exertional Myalgia with or without Rhabdomyolysis)
- SEPN1 Related Myopathy (Rigid Spine Muscular Dystrophy/RSMD1, Congenital Fiber Type Disproportion, Mallory Weiss Body Desmin, Multiminicore Myopathy)
- SYNE1 (Nesprin Related Muscular Dystrophy)
- Telethonin Related Muscular Dystrophy (TCAP/Titin-Cap)
- Congenital Muscular Dystrophy Not Otherwise Specified (including Merosin Positive)

**Congenital Myopathy subtypes included in CMDIR**

- Actin Aggregation Myopathy
- Cap Disease
- Central Core Disease (including Malignant Hyperthermia, Exertional Myalgia with or without Rhabdomyolysis)
- Centronuclear Myopathy (including Malignant Hyperthermia, Exertional Myalgia with or without Rhabdomyolysis)
- Congenital Fiber Type Disproportion (including Malignant Hyperthermia, Exertional Myalgia with or without Rhabdomyolysis)
- Core Rod Myopathy
- Hyaline Body Myopathy
- Multiminicore Myopathy
- Myotubular Myopathy
- Nemaline Myopathy
- Reducing Body Myopathy
- RYR1 Related Myopathy (including Malignant Hyperthermia, Exertional Myalgia with or without Rhabdomyolysis)
- Spheroid Body Myopathy
- Tubular Aggregate Myopathy
- Zebra Body Disease Myopathy
- Congenital Myopathy Not Otherwise Specified

**Congenital Myasthenic Syndrome (all subtypes included in CMDIR)**

**Myofibrillar Myopathy (all subtypes included in CMDIR)**