

**Name of Journal:** *World Journal of Clinical Cases*

**Manuscript NO:** 62212

**Manuscript Type:** CASE REPORT

**A homozygous deletion, c. 1114-1116del, in exon 8 of the CRPPA gene causes congenital muscular dystrophy in Chinese family: A case report**

Mi Y, *et al.* Novel CRPPA variant in CMD.

Mi Yang, Ru-Xin Xing

## Match Overview

1	<b>Crossref</b> 24 words Liu, Xiaomin, Gaoting Ma, Zhangning Zhao, Fei Mao, Jiyou Tang, Xiuhua Li, and Meijia Zhu. "Novel mutation of SLC20A1 ...	1%
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## Genetic cause of heterogeneous inherited myopathies in a ...

<https://www.sciencedirect.com/science/article/pii/S2214426920301282>

Dec 01, 2020 · 1. Introduction. Inherited muscle diseases are a heterogeneous group of disorders, that includes **congenital**, metabolic and mitochondrial myopathies, **muscular** dystrophies and myotonias [1,2]. Each of these categories of genetic muscle disorders is in turn broad, with **muscular** dystrophies subdivided to Duchenne/Becker, myotonic, facioscapulohumeral, distal and limb-girdle **muscular** ...

Author: Ioannis Zaganas, Vasilios Mastorode...

Publish Year: 2020

## OMIM Entry - # 608807 - MUSCULAR DYSTROPHY, LIMB ...

<https://www.omim.org/entry/608807> ▾

A number sign (#) is used with this entry because of evidence that autosomal recessive limb-girdle **muscular dystrophy**-10 (LGMDR10) is caused by **homozygous** or compound heterozygous mutation in the titin **gene** (TTN; 188840) on chromosome 2q31. Heterozygous mutation in the titin **gene** causes tardive tibial **muscular dystrophy** (TMD; 600334). For a general description and a discussion of ...

## (PDF) ISPD gene mutations are a common cause of ...

<https://www.researchgate.net/publication/234047744...>

In this article, we report the involvement of the ISPD **gene** in milder dystroglycanopathy phenotypes ranging from **congenital muscular dystrophy** to limb-girdle **muscular dystrophy** and identified ...

## Muscular Dystrophy, Congenital, Lmna-Related disease ...

[https://www.malacards.org/card/muscular\\_dystrophy\\_congenital](https://www.malacards.org/card/muscular_dystrophy_congenital) ▾

Genetics Home Reference: 25 LMNA-related **congenital muscular dystrophy** (L-CMD) is a condition that primarily affects muscles used for movement (skeletal muscles). It is part of a group of genetic conditions called **congenital muscular dystrophies**, which cause weak muscle tone (hypotonia) and muscle wasting (atrophy) beginning very early in life.

## Limb-Girdle Muscular Dystrophies

<https://neuromuscular.wustl.edu/musdist/lg.html> ▾

Northern India: p.Asp780His, c.2099-1G>T, c.2051-1G>T, c.2338G>C Large (31,012-bp) **deletion** (Exons 2-8) (c.309+4469\_c.1116-1204del) Occurs in different ethnic backgrounds; Clinical correlations Severe phenotype **Homozygous** null mutations: Often but not always NS1 domain mutations; S86F Least severe phenotype: **Homozygous** missense mutations



Homozygous deletion, c. 1114-1116del, in exon 8 of the CRPPA ge



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### [\(PDF\) ISPD gene mutations are a common cause of ...](#)

<https://www.researchgate.net/publication/234047744...>

Dystroglycanopathies are a clinically and genetically diverse group of recessively inherited conditions ranging from the most severe of the **congenital muscular** dystrophies, Walker-Warburg syndrome ...

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<https://www.researchgate.net/publication/23481951...>

**congenital muscular** dystrophy (FCMD), caused by a mutation in the fukutin **gene**, is a severe form of dystro- glycanopath y. A retrotransposon insertion in fukutin is seen in almost all cases of ...

### [\(PDF\) ISPD loss-of-function mutations disrupt dystroglycan ...](#)

<https://www.researchgate.net/publication/224810956...>

(c) Schematic (not to scale) of the ISPD **exon**-intron **gene** structure. Human ISPD cDNA (5,524 bp; NM\_001101426) contains 10 coding exons spread across 333,796 bp of genomic DNA.

### [\(PDF\) Low Pass-Genome Sequencing: Validation and ...](#)

<https://www.researchgate.net/publication/340919264...>

Duchenne **muscular** dystrophy (DMD; **exon** 55) **deletion** was detected in a male with Duchenne **muscular dystroph** y, not previously detected by CMA analysis (because of lack

### [\(PDF\) The ties that bind: functional clusters in limb ...](#)

<https://www.researchgate.net/publication/343291910...>

with severe forms of **congenital muscular** dystrophy, a re- ... Duchenne **muscular** dystrophy **gene**. ... **causes muscular** dystrophy and arrhythmia by affecting protein trafficking. J Clin Invest. 2016; ...



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Cited by: 1

Author: Ioannis Zaganas, Vasilios Mastorodemos,...

Publish Year: 2020

## Abstracts from the 53rd European Society of Human Genetics ...

<https://www.nature.com/articles/s41431-020-00741-5>

Dec 01, 2020 · A recurrent **exon 4\_10 deletion** in the IKBKG **gene** accounts for 60 to 80% of IP-causing mutations, but other point mutations along the **gene** as well as **exon** ...

## (PDF) ISPD mutations account for a small proportion of ...

<https://www.researchgate.net/publication/282277834...>

**CRPPA gene** pathogenic variants have been reported to cause a spectrum of phenotypes ranging from **congenital muscular dystrophy** with cerebral and ocular involvement (Walker-Warburg syndrome ...

## Fukuyama Congenital Muscular Dystrophy - Abstract - Europe PMC

<https://europepmc.org/abstract/MED/20301385> ▾

Jan 26, 2006 · The CMDIR is a patient self-**report** registry with the goal to register the global **congenital** muscle disease population including persons with **congenital** myopathy, **congenital muscular dystrophy**, and **congenital** myasthenic syndrome. The CMDIR registers affected individuals of all ages with symptoms from birth through late onset (limb-girdle).

Cited by: 16

Author: Kayoko Saito

Publish Year: 2019

## Autosomal Recessive Limb-Girdle Muscular Dystrophy disease