# Recessive Cardiomyopathy Gene Summaries

#recessiveCMgenes

# *LMOD2*

*Thin filament protein (actin elongation)*

*Biallelic phenotype:*



**DCM**

Year of first association: **2019**

Pathogenic variant classes: **Truncating / LoF**

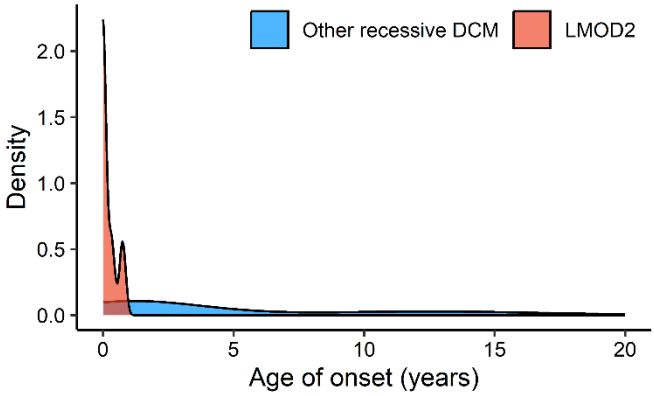
Mouse KO: **DCM-like phenotype, early death**

| | | |
|---|---|---|
| **Exome / Genome** | 4 | - |
| **Gene Panel** | 1 | - |

***Number of biallelic families***

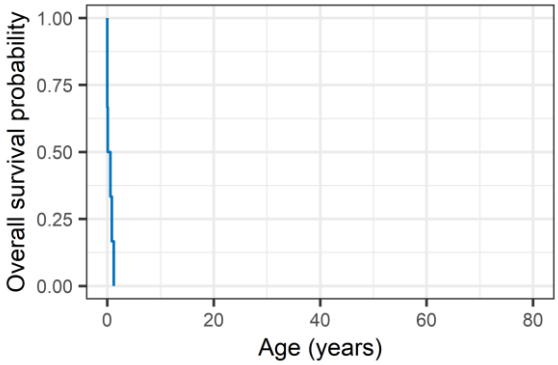
*- Proven recessive (trio/family) or proband only*
*- Detected by genome-wide methods or panels*

## *Clinical overview*

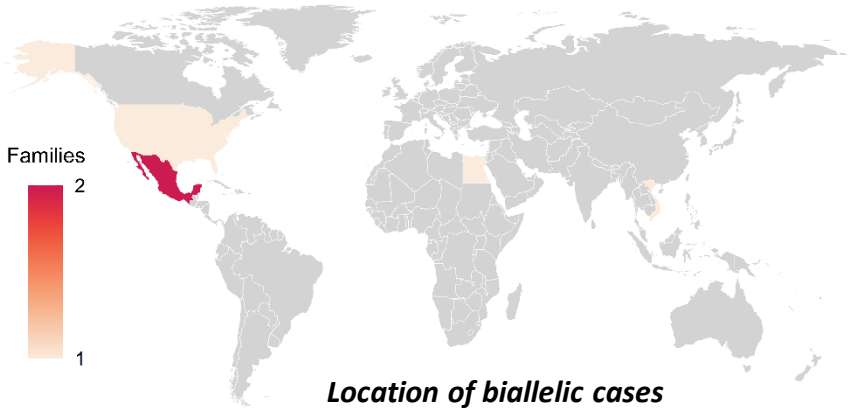



***Outcomes***

*Freedom from death, heart transplantation or LVAD*




## *Geographic and ancestral associations*




***Location of biallelic cases***

***gnomAD v4 carrier frequencies***

| African | Middle East | South Asian | East Asian |
|---|---|---|---|
| 0.03% | 0.03% | 0.02% | 0.02% |

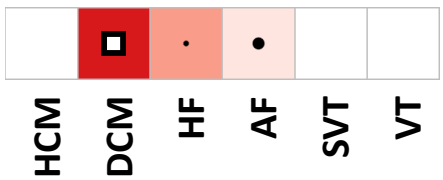
| European | Finnish | Ashkenazi | American |
|---|---|---|---|
| 0.04% | 0.00% | 0.01% | 0.02% |

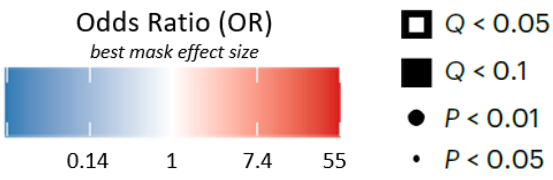
*% of individuals with heterozygous LoF variants*

## *Monoallelic associations*

***UK Biobank analysis***

*Effect sizes with p>0.05 displayed as white*




**DCM**: OR = 13.7 (5.4-35.2) for rare *LMOD2* heterozugous protein truncating variants in UK Biobank

*Biallelic reports (PMIDs): 31517052, 34888509, 35082396, 35188328, 37296576*

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*Created on Jan 31st 2024*

***Exploring the complex spectrum of dominance and recessiveness in genetic cardiomyopathies.***

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