Recessive Cardiomyopathy Gene Summaries

#recessiveCMgenes

TRIM63

E3 ubiquitin protein ligase (MuRF1)

Biallelic phenotype:



HCM

Year of first association: **2015**

Pathogenic variant classes:
Truncating & Missense (LoF)

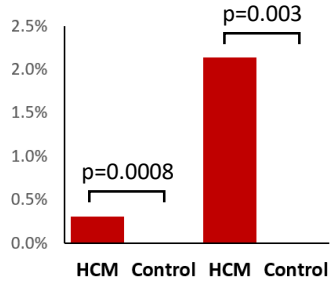
Mouse KO: **Severe hypertrophy**
(*with TAC model or TRIM63-TRIM55 double-KO*)

Exome / Genome	-	-
Gene Panel	7	21

Number of biallelic families

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

Case-Control

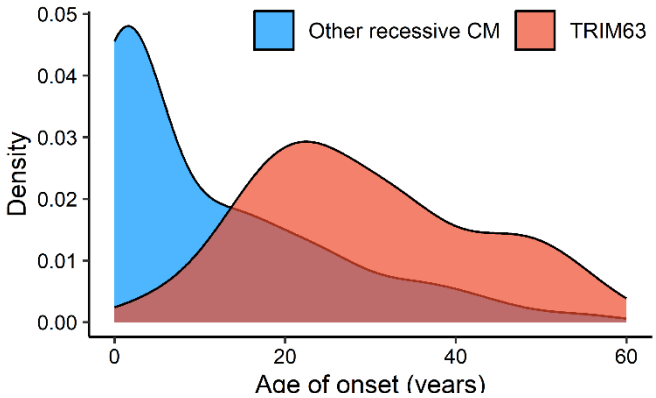






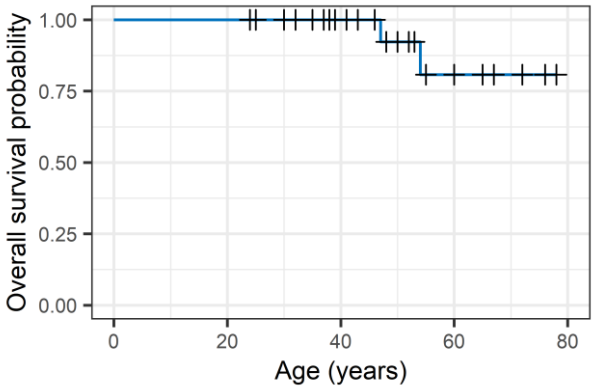
PMID: 32451364 37431535

Clinical overview

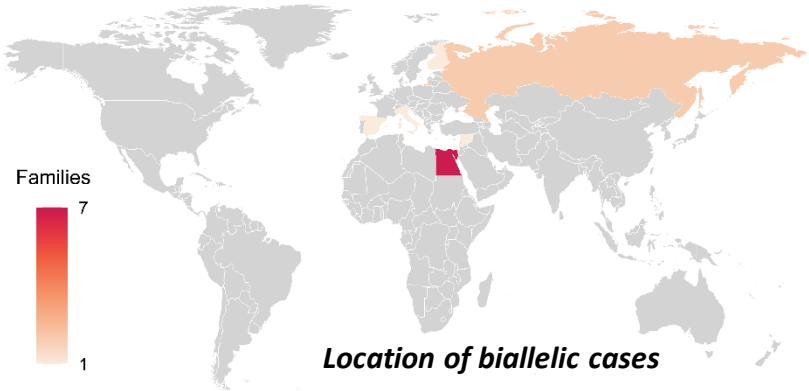


Outcomes

Freedom from death, heart transplantation or LVAD



Geographic and ancestral associations



Location of biallelic cases

Note: Most published cases were from a majority European ancestry cohort (PMID:32451364) but specific country of origin data was not available.

gnomAD v4 carrier frequencies

African	Middle East	South Asian	East Asian
0.07%	0.82%	0.05%	0.03%

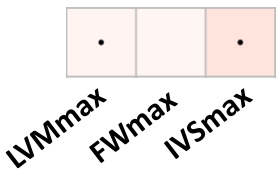
European	Finnish	Ashkenazi	American
0.19%	0.19%	1.48%	0.25%

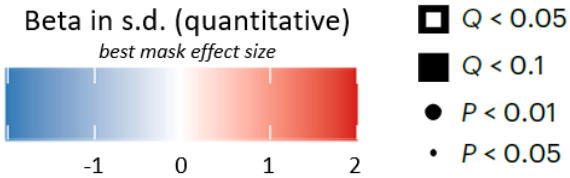
% of individuals with heterozygous LoF variants

Monoallelic associations

UK Biobank analysis

No significant association with cardiac disease traits, but nominal association observed for LV mass/hypertrophy traits





Biallelic reports (PMIDs): 25801283, 30372688, 32451364, 35273634, 36788754, 37431535

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Created on March 13th 2024

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

Nature Cardiovascular Research 2:1078–1094 (2023)