Recessive Cardiomyopathy Gene Summaries

#recessiveCMgenes

NRAP

Nebulin-related anchoring protein (intercalated disc)

Biallelic phenotype:



DCM

Year of first association: **2017**

Pathogenic variant classes: **Truncating / LoF**

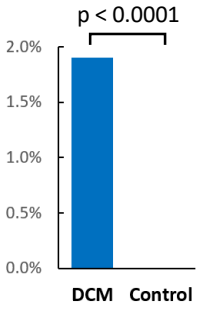
Mouse KO: **-**

Exome / Genome	7	12
Gene Panel	2	9

Number of biallelic families

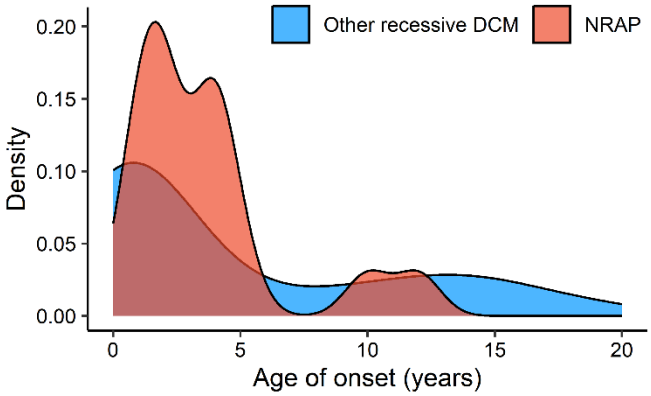
- Proven recessive (trio/family) or proband only
- Detected by genome-wide methods or panels

Case-Control



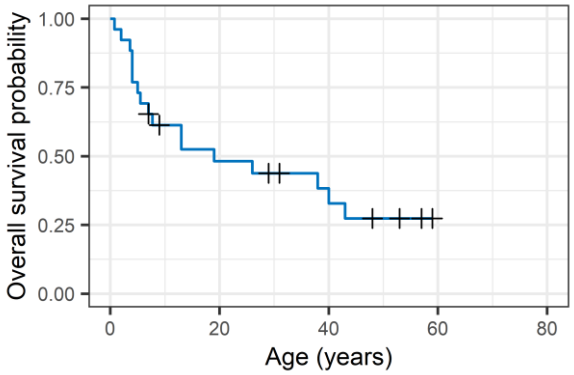
PMID:33534821

Clinical overview

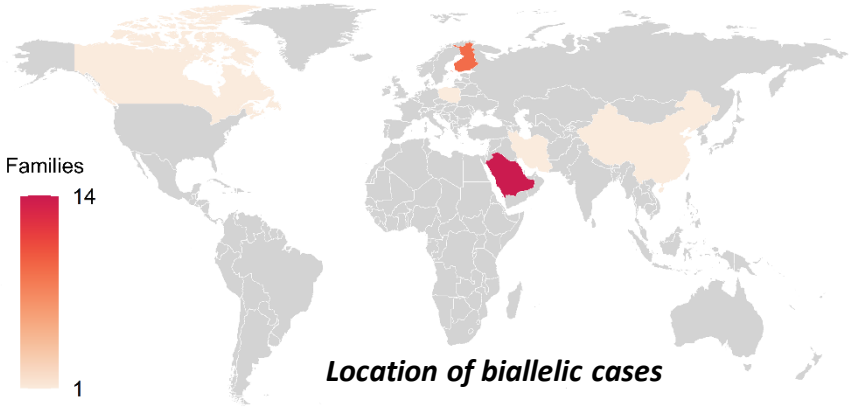


Outcomes

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
0.78%	0.46%	0.48%	0.20%

European	Finnish	Ashkenazi	American
0.24%	1.36%	1.03%	0.36%

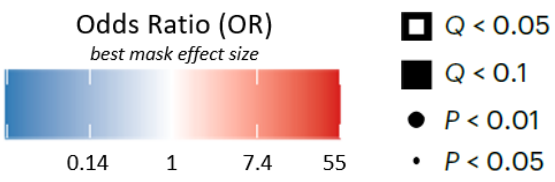
% of individuals with heterozygous LoF variants

Monoallelic associations

UK Biobank analysis

Effect sizes with p>0.05 displayed as white





Other evidence

Heterozygous *NRAP* truncating variants significantly enriched (p<0.0001) in DCM cases (1.9%, n=11/577) compared to controls (0.3%, n=75/25912), with an OR=6.7 (3.5–12.7), suggesting these variants may be risk factors of intermediate effect sizes in DCM patients (patient cohort likely enriched for individuals of Finnish ancestry) (PMID:33534821).

Biallelic reports (PMIDs): 28600779, 28611399, 30384889, 32870709, 33534821, 35288587, 36672924, 36815016

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Created on Feb 27th 2024

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

Nature Cardiovascular Research 2:1078–1094 (2023)