Recessive cardiomyopathy genes – *BAG5*

BAG5

Co-chaperone of proteostasis regulation

Biallelic phenotype:



DCM

Year of first association: **2022**

Biallelic variant classes: **Truncating / LoF**

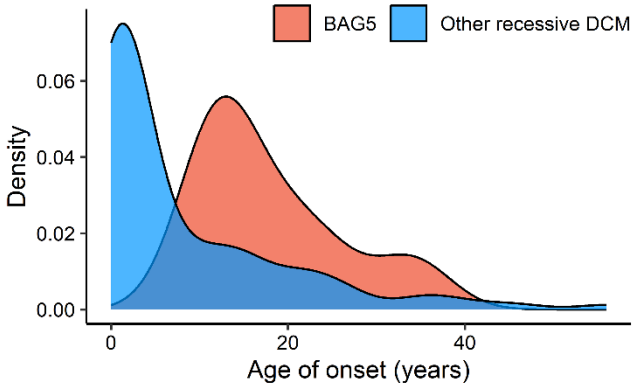
Mouse KO: **LV dilation and arrhythmogenicity**

Exome / Genome	5	2
Gene Panel	-	-

Number of biallelic families

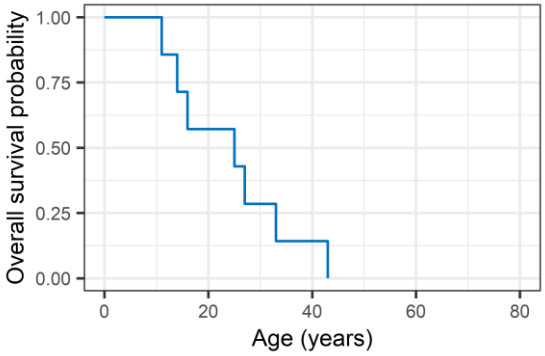
- Proven recessive (trio/family) or proband only
- Detected by genome-wide methods or panels

Clinical overview

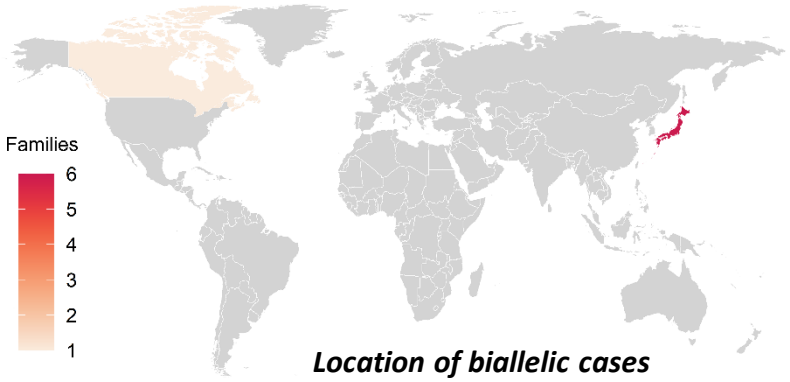


Outcomes

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%	0.00%	0.06%	0.06%

European	Finnish	Ashkenazi	American
0.09%	0.01%	0.00%	0.02%

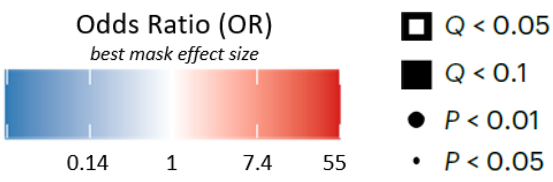
% of individuals with heterozygous LoF variants

Monoallelic associations

UK Biobank analysis

Effect sizes with p>0.05 displayed as white





Other evidence

Heterozygous *BAG5* truncating variants detected in 3/26 individuals with tachycardia-induced cardiomyopathy (PMID:35044787).

Biallelic reports (PMIDs): 35044787, 35288587, 37873655

#recessiveCMgenes

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

Nature Cardiovascular Research 2:1078–1094 (2023)