

Table S1a

Timeline/Data

200,581 participants with whole exome sequencing data

Diagnostic code is present and first reported date precedes UKBB recruitment

Diagnostic code is present and first reported date after UKBB recruitment

Diagnostic code is present (any date, "first reported date" combined dataset)

DCM diagnostic code is present (any date) using ICD code (HES data only)

Non-ischaemic cardiomyopathy (any date)

21,129 with whole exome sequencing data and cardiac MRI imaging

Diagnostic code is present and first reported date precedes UKBB recruitment

Diagnostic code is present and first reported date after UKBB recruitment but precedes imaging

Diagnostic code is present at time of CMR ("first reported date" combined dataset)

Imaging criteria for DCM on CMR (increased LVEDV+ decreased LVEF without HCM or CAD), AND no CM repc

Diagnostic code present at time of CMR OR DCM criteria on CMR

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Table S1 a) Counts of CM and MACE phenotypes, and death, and estimates of CM prevalence, reported for the individuals of UKBB with exome sequencing data and a subset with imaging data available for analysis; b) Corresponding P-values of the burden analysis comparing heterozygotes to the rest of the population.

Prevalence of disease was estimated using diagnostic codes at 3-time points, enrollment, date of imaging, and at the most recent assessment. This was completed to mimic the participants that would be identified by secondary findings; known affecteds at recruitment, unrecognised affecteds identified at imaging, and heterozygotes that developed disease during follow up. At the time of imaging, we assessed prevalence using diagnostic codes & imaging definition. Incident cases were identified between these time points. The association between variant carrier status and diagnoses were tested using Chi-squared (normal coloured cell) or Fisher's exact (pink cell) tests. Non-significant associations were highlighted in red font. *, for each trait the total number subtraction of participants identified at previous incidence(s)

Description

Prevalence at ukbb recruitment

Incident cases post-recruitment (new cases) = value of surveillance

Cumulative prevalence

Latest prevalence (these data allow for the most specific phenotype code for DCM, which is only available for HE

An upper-bound estimate based on an inclusive definition of non-ischaemic cardiomyopathies

Prevalence at ukbb recruitment

Incident cases between recruitment & imaging

Prevalence of "known" DCM at time of imaging

Phenotype identified via MRI only

Total DCM by diagnostic code or imaging

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Total DCM by diagnostic code or imaging

						TTNtv	
Proportion with DCM or CM (%)	n	DCM (n)	any CM (n)	cardiac arrest (n)	MACE (n)		
1.4	877		12	1	80		
3.5	877*		30	5	141		
4.8	877		42	6	221		
4.3	877	38					
8.1	668	54					
1.1	88		1	0	10		
2.3	88*		2	0	10		
3.4	88		3	0	20		
2.4	85	2					
5.7	88	5					
						FLNCtv	
Proportion with DCM or CM (%)	n	DCM (n)	any CM (n)	cardiac arrest (n)	MACE (n)		
0	50		0	0	5		
0	50*		0	0	5		
0	50		0	0	10		
0	50	0					
0	37	0					
0	5		0	0	0		
0	5*		0	0	0		
0	5		0	0	0		
0	5	0					
0	5	0					

AF/arrhythmia (n)	stroke (n)	heart failure (n)	death (n)	Prevalence of DCM or CM (%)
59	13	24	0	0.09
112	6	75	60	0.25
171	19	99	60	0.33
				0.17
				0.73
8	1	2	0	0.02
9	0	3	0	0.08
17	1	5	0	0.10
				0.38
				0.48
AF/arrhythmia (n)	stroke (n)	heart failure (n)	death (n)	Prevalence of DCM or CM (%)
5	0	0	0	0.09
4	0	3	5	0.26
9	0	3	5	0.35
				0.19
				0.76
0	0	0	0	0.03
0	0	0	0	0.09
0	0	0	0	0.12
				0.39
				0.51

no TTNtv

n	DCM (n)	any CM (n)	cardiac arrest (n)	MACE (n)	AF/arrhythmia (n)	stroke (n)
199704		172	82	7503	4634	2527
199704*		491	894	14088	11498	953
199704		663	976	21591	16132	3480
199704	338					
158299	1149					
21041		5	3	572	406	148
21041*		17	13	606	448	121
21041		22	16	1178	854	269
21019	80					
21041	102					

no FLNCtv

n	DCM (n)	any CM (n)	cardiac arrest (n)	MACE (n)	AF/arrhythmia (n)	stroke (n)
200531		184	83	7578	4688	2540
200531*		521	899	14224	11606	959
200531		705	982	21802	16294	3499
200531	376					
158930	1203					
21124		6	3	582	414	149
21124*		19	13	616	457	121
21124		25	16	1198	871	270
21099	82					
21124	107					

heart failure (n)	death (n)
858	0
4776	11859
5634	11859
31	0
92	0
123	0

heart failure (n)	death (n)
882	0
4848	11914
5730	11914
33	0
95	0
128	0