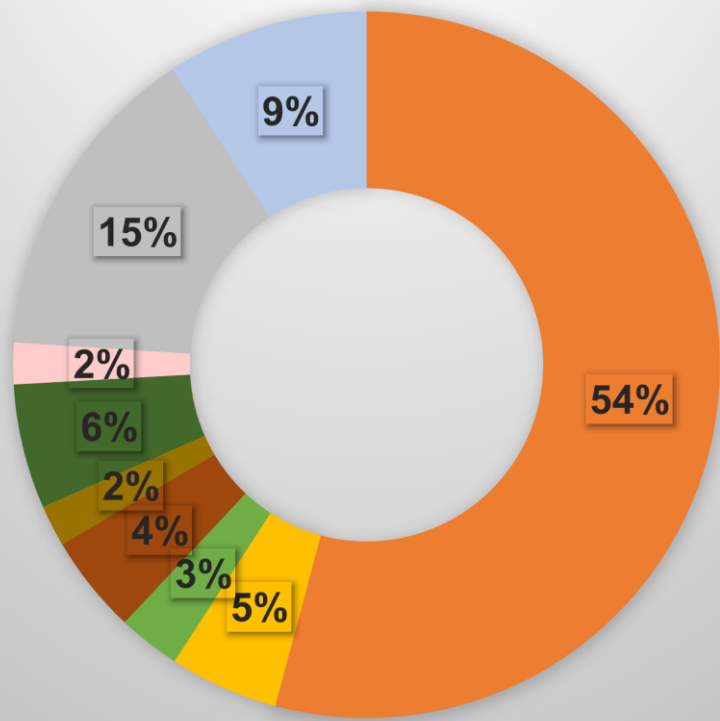


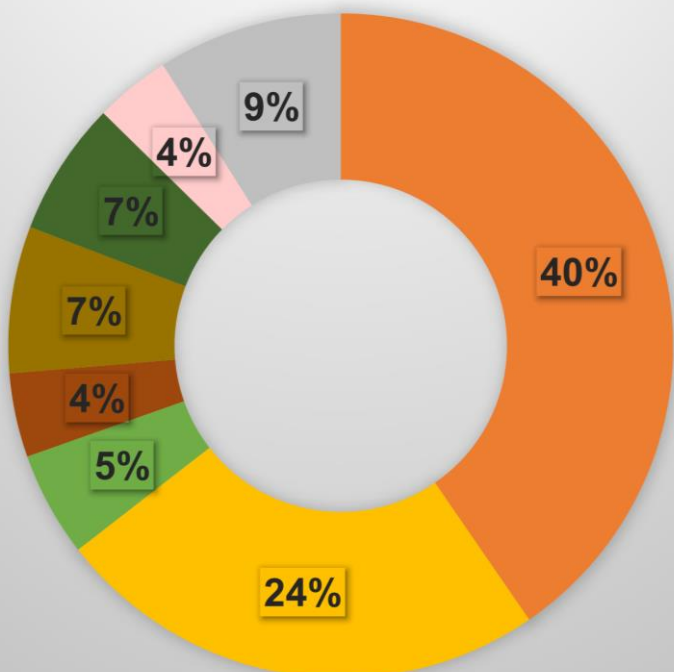
Figure 1. Aetiology of unexpected Sudden Cardiac Death

De Noronha N = 720 (all ages)
national referral autopsy results

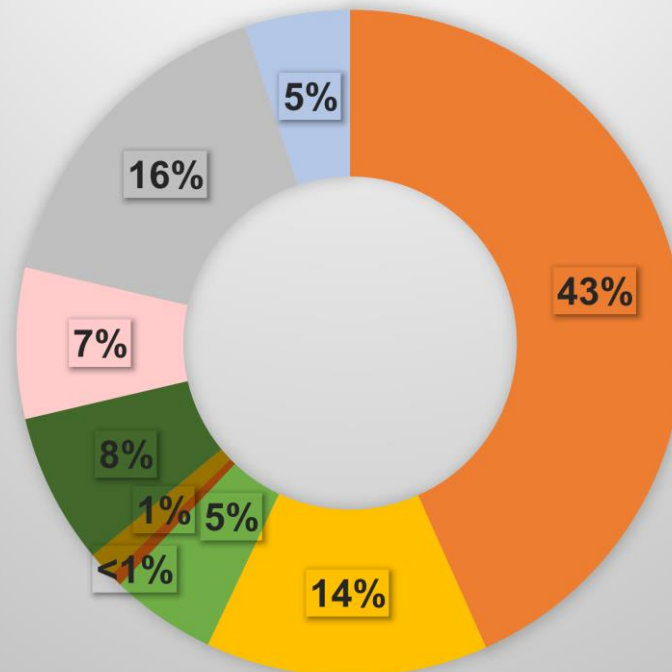


- Normal heart
- Coronary artery pathology /ischemic heart disease
- Arrhythmogenic right ventricular cardiomyopathy
- Hypertrophic cardiomyopathy
- Dilated cardiomyopathy
- Inflammation
- Aortic disease
- Other cardiac pathology/cardiomyopathy
- Idiopathic fibrosis/left ventricular hypertrophy

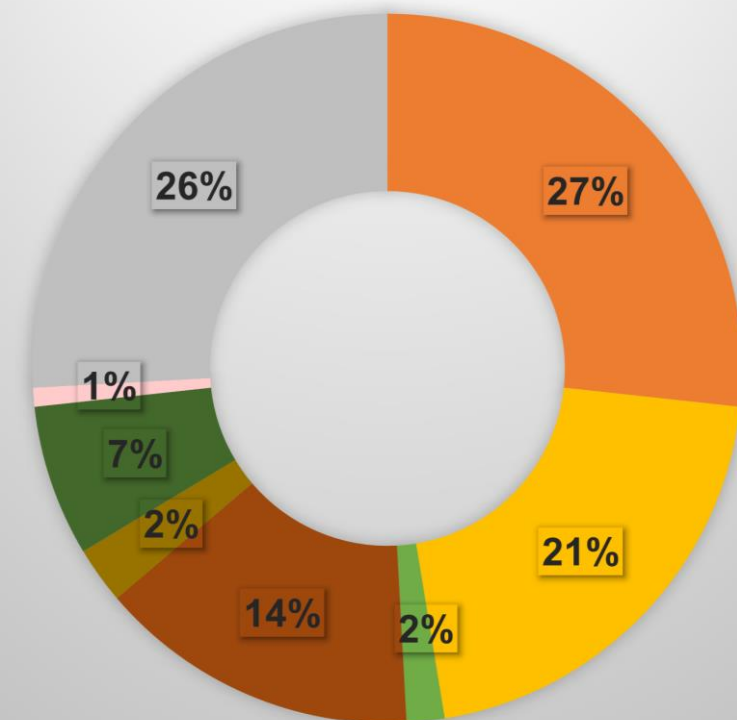
Bagnall N= 490 (1-35 y)
prospective international
autopsy survey



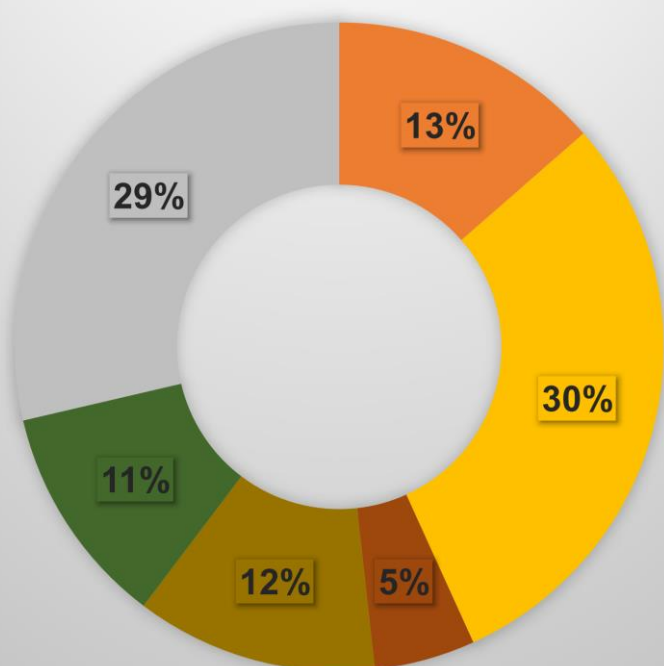
Winkel N= 314 (1-35 y)
national retrospective autopsy
results



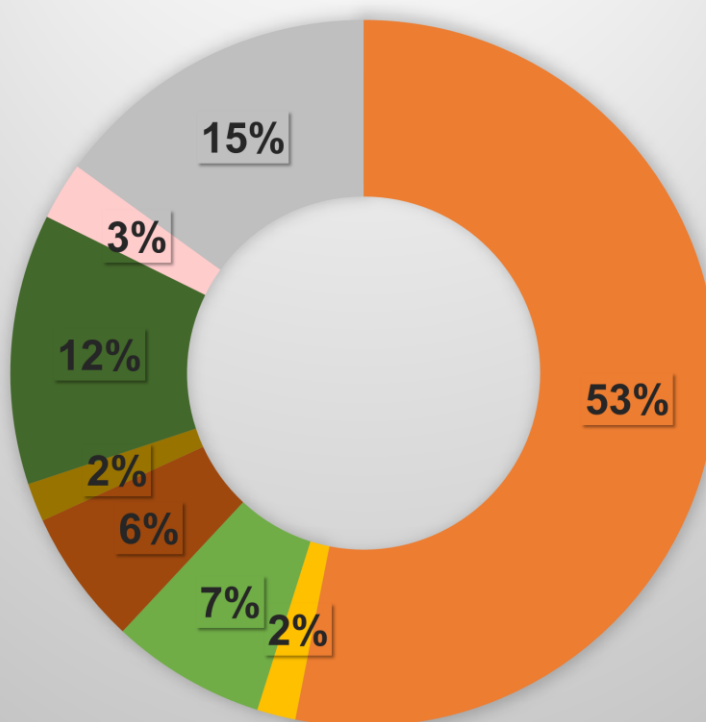
Margey N= 116 (15-34 y)
national SCD autopsy results



Papadakis N= 1677 (1-34 y)
national death certificates data



Pilmer N= 113 (1-19 y)
regional SCD autopsy results



Corrado N= 273 (1-35 y)
regional SCD autopsy results

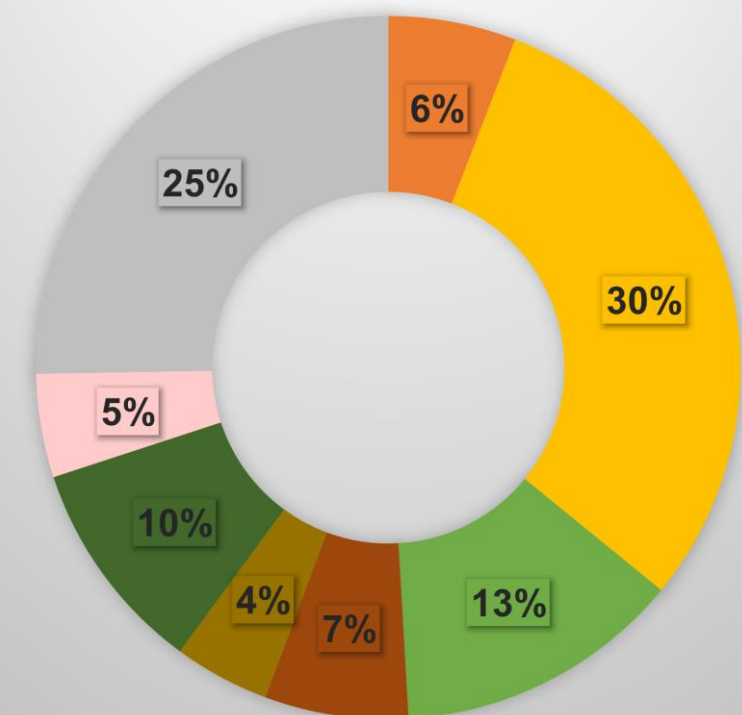


Figure 2. Aetiology of unexpected Sudden Cardiac Arrest

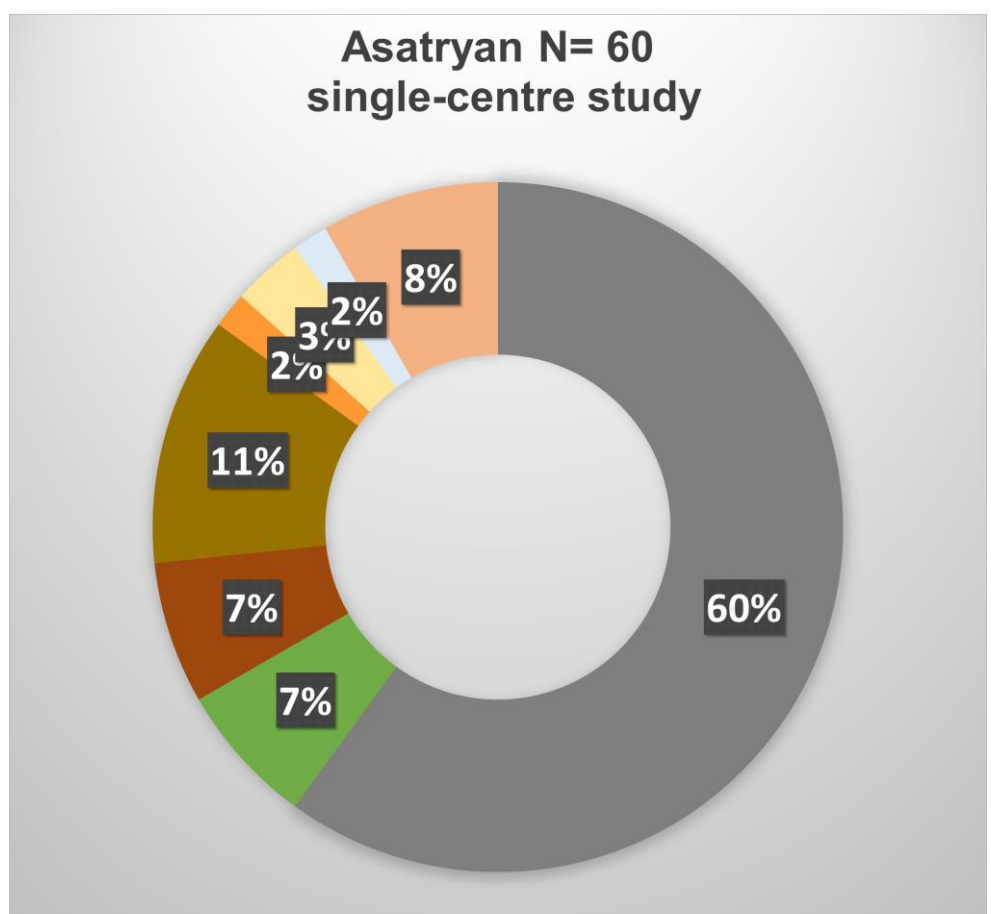
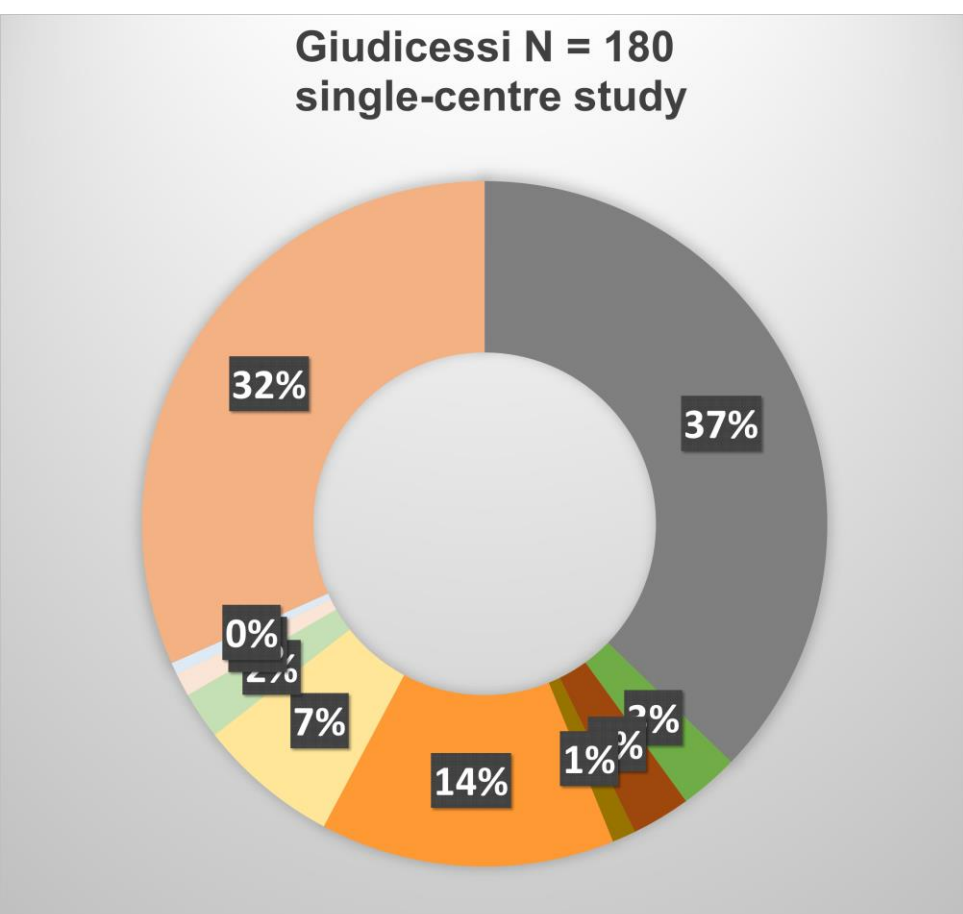
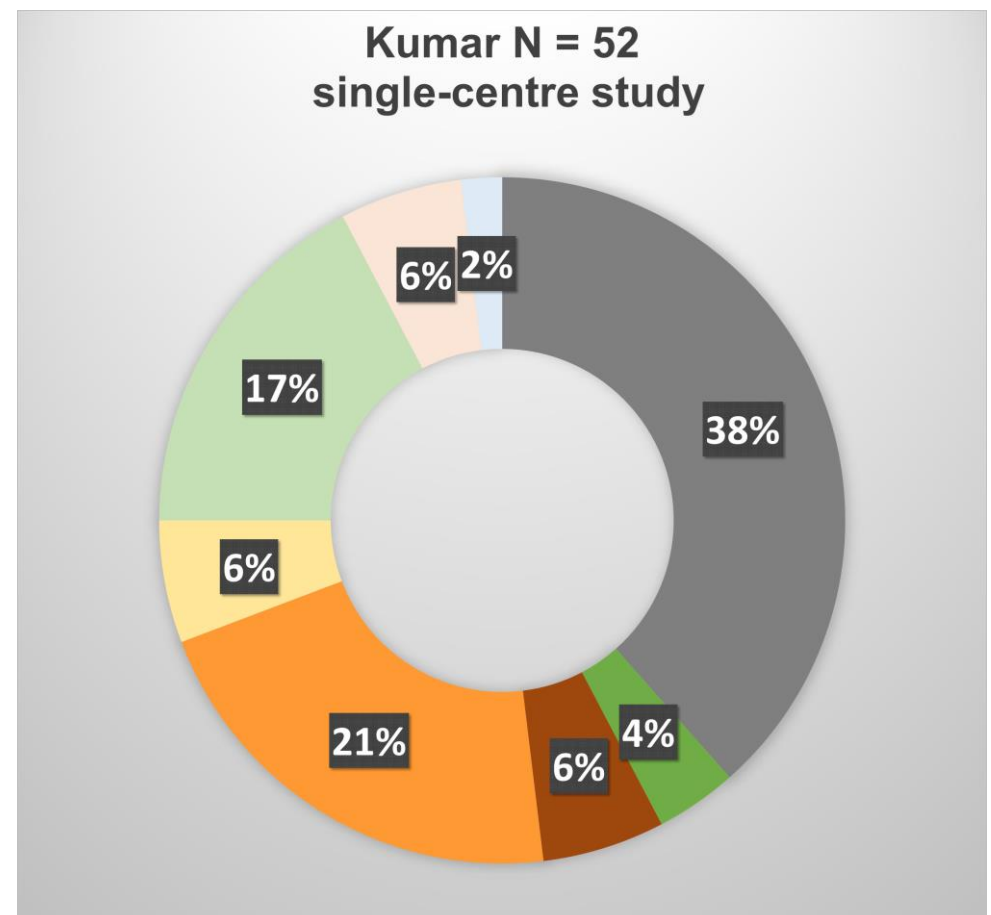
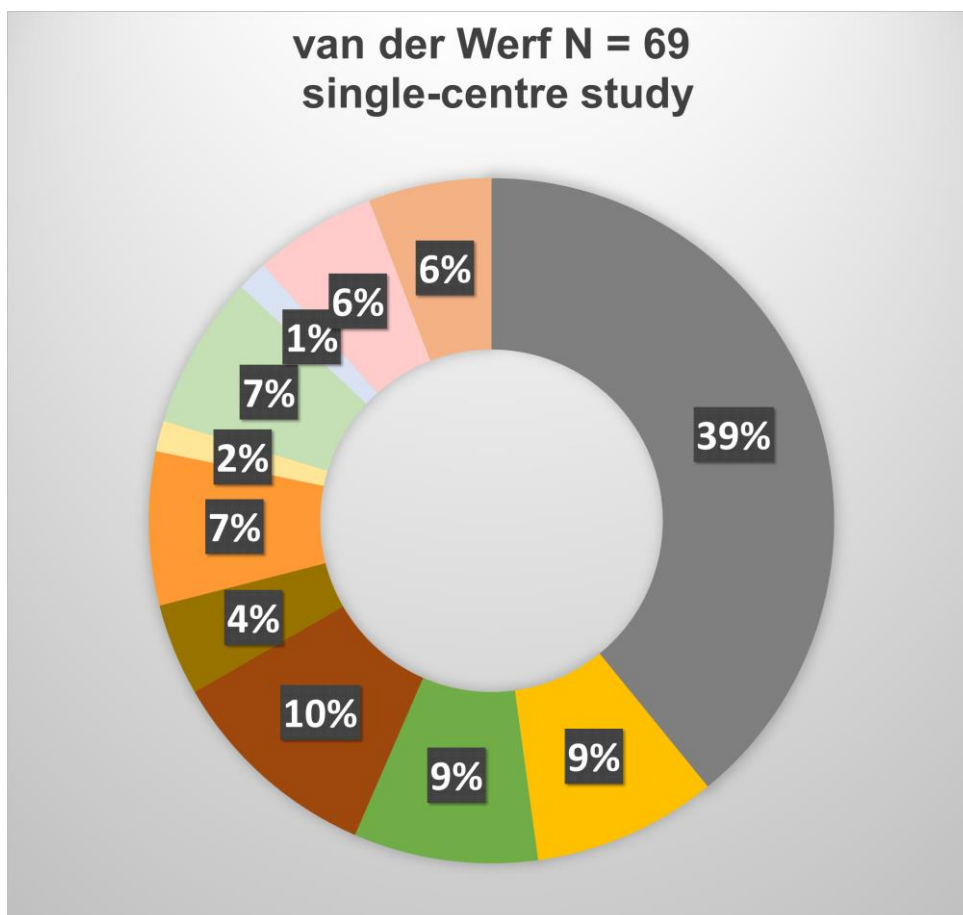
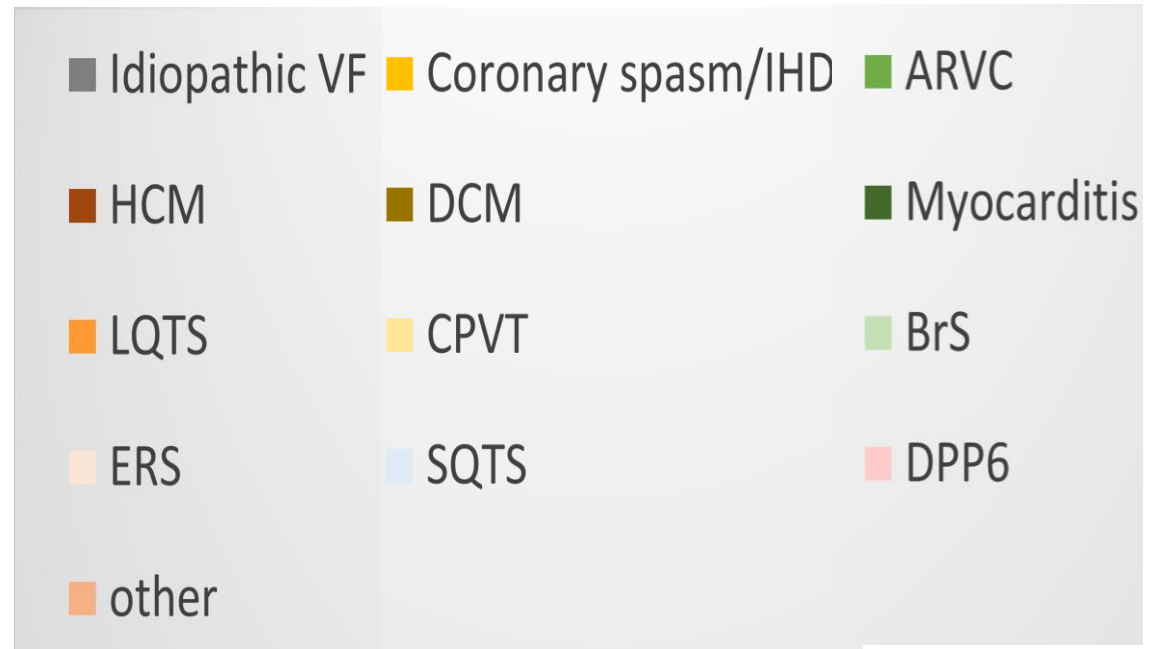
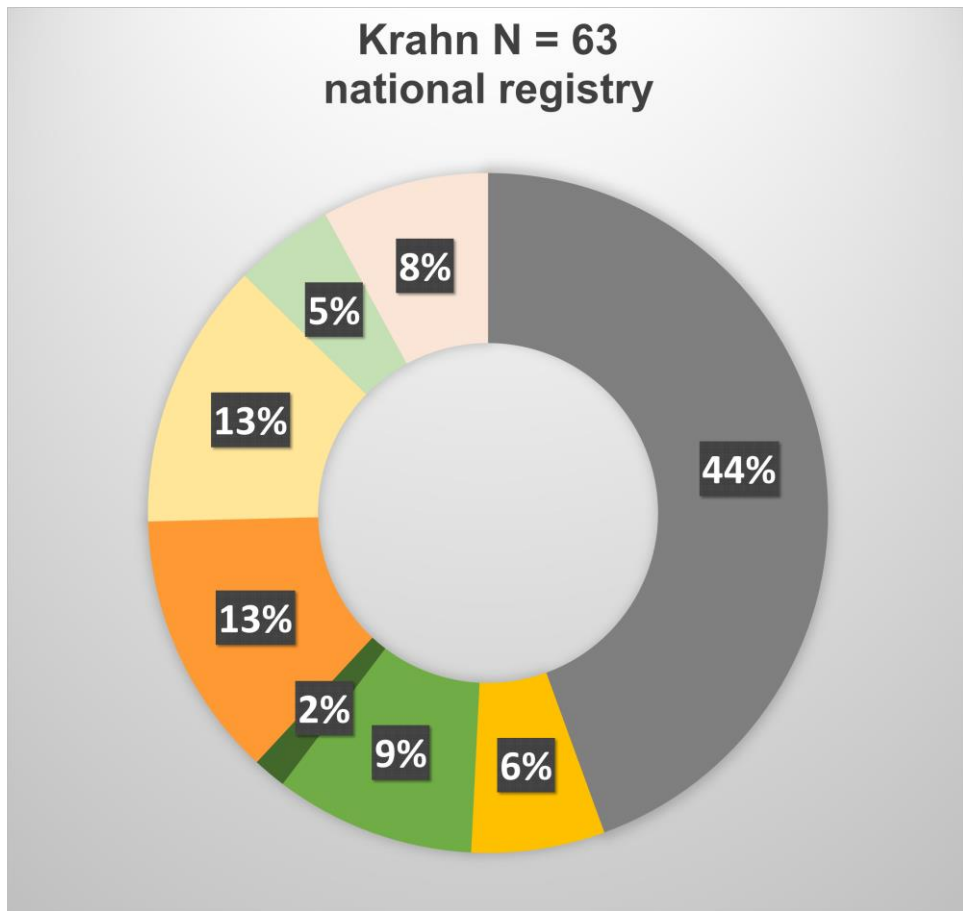
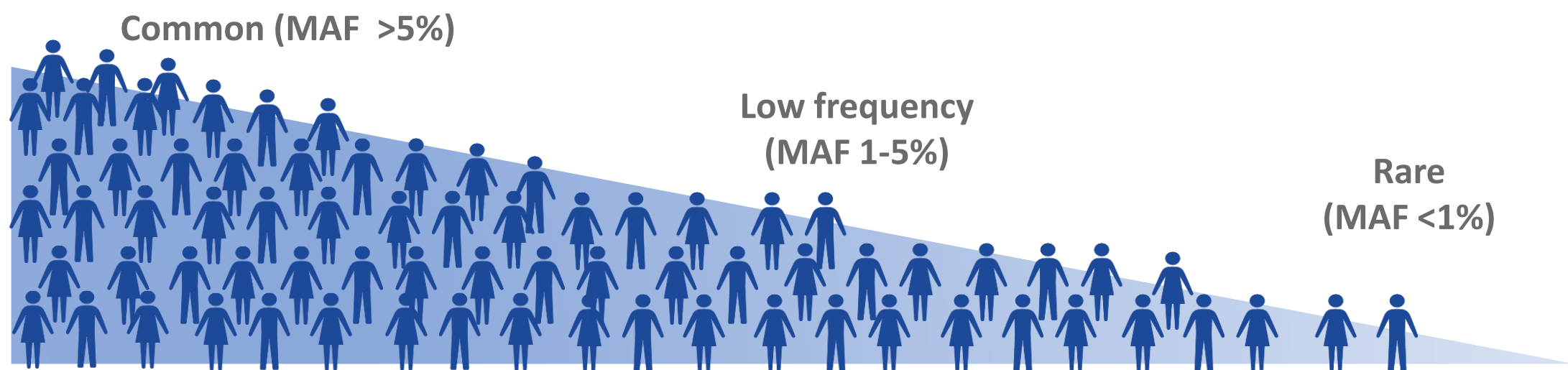
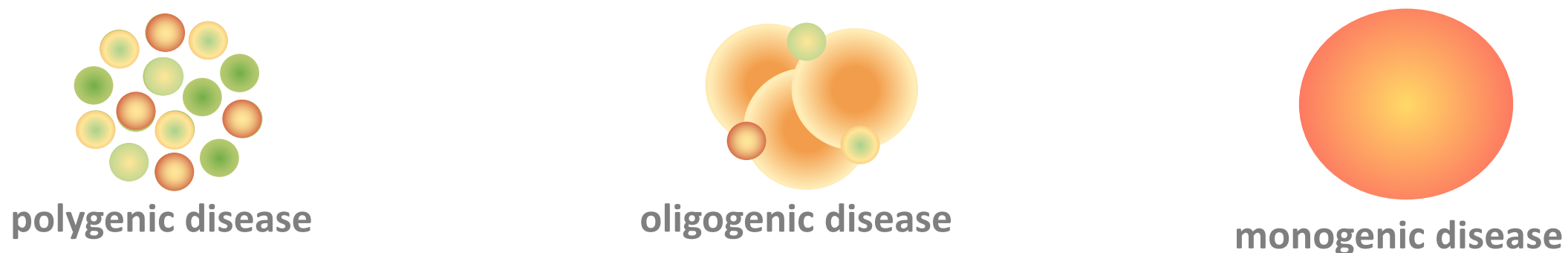


Figure 3 . Genetic variants frequency, effect size and classification

a) Variant frequency



b) Size of variant effect



c) Variant classification

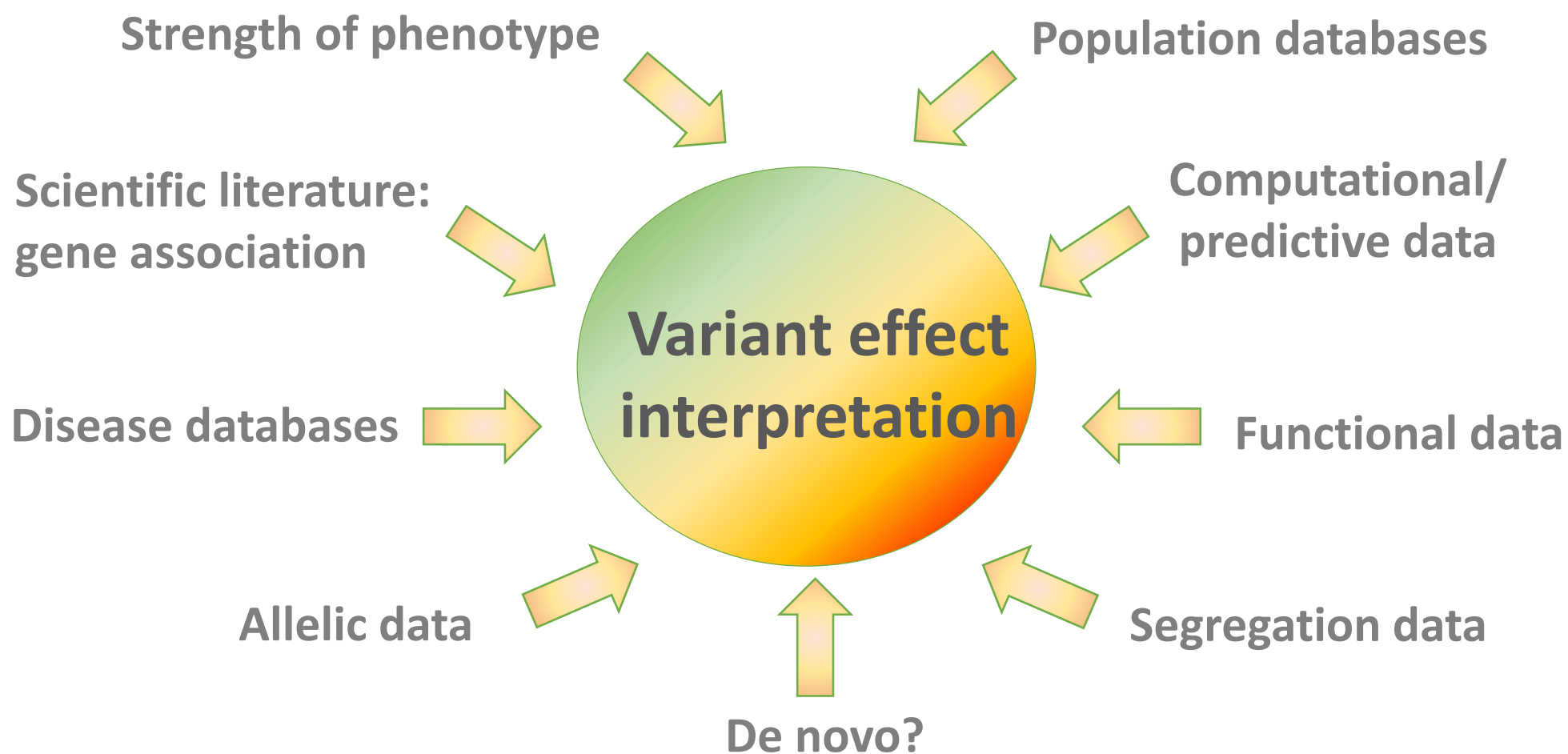
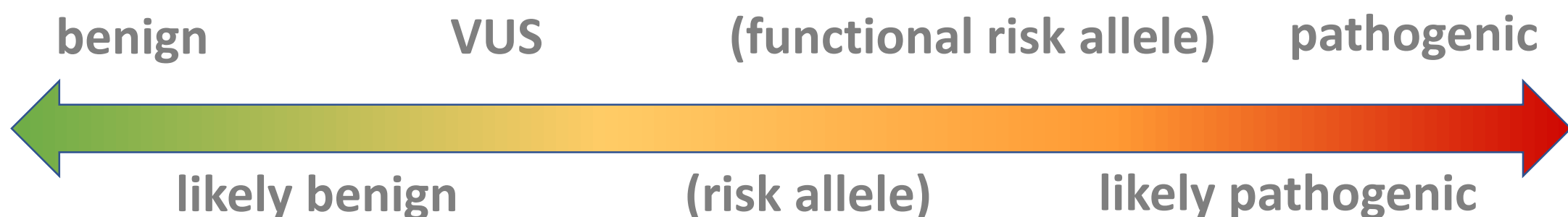
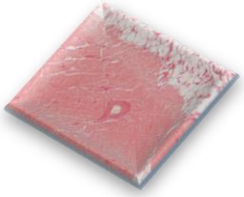


Figure 4 . Genetic approaches to rare and common diseases

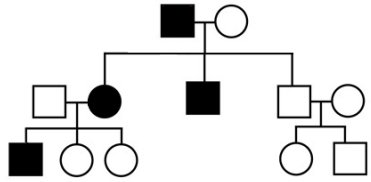
Genetic approaches to rare and common diseases

Rare or private variants – familial diseases

Molecular autopsy



Linkage analysis



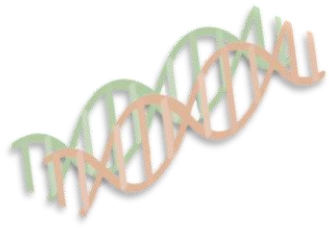
Variant classification (figure 3)

Genomic datasets



In vitro studies

Animal studies



Whole exome- or genome- sequencing



Variant calling

Common variants – population diseases

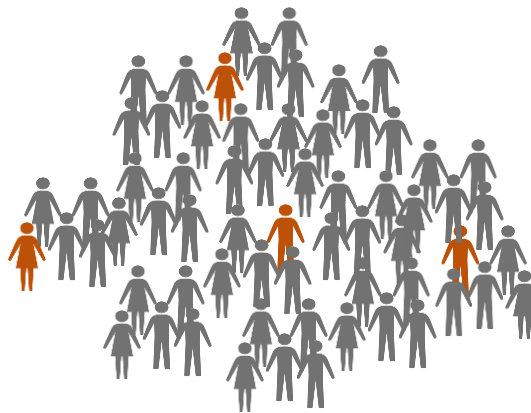
Cases



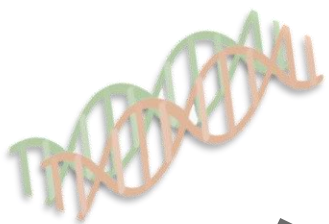
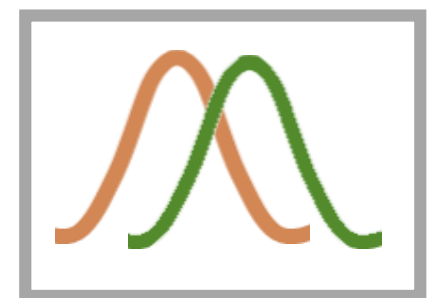
Controls



Risk prediction in general population



Polygenic risk scores



Genome-wide Association Studies

