

# Utility of Polygenic Risk Score (PGS) for Prediction of Dilated Cardiomyopathy (DCM) and Modulation of Severity of LV Dysfunction Among Cases

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## Background

### Clinical characteristics of DCM:

- ↑ LV volumes, ↓ LVEF;

### High burden:

- Heart transplantations;
- Heart failure;

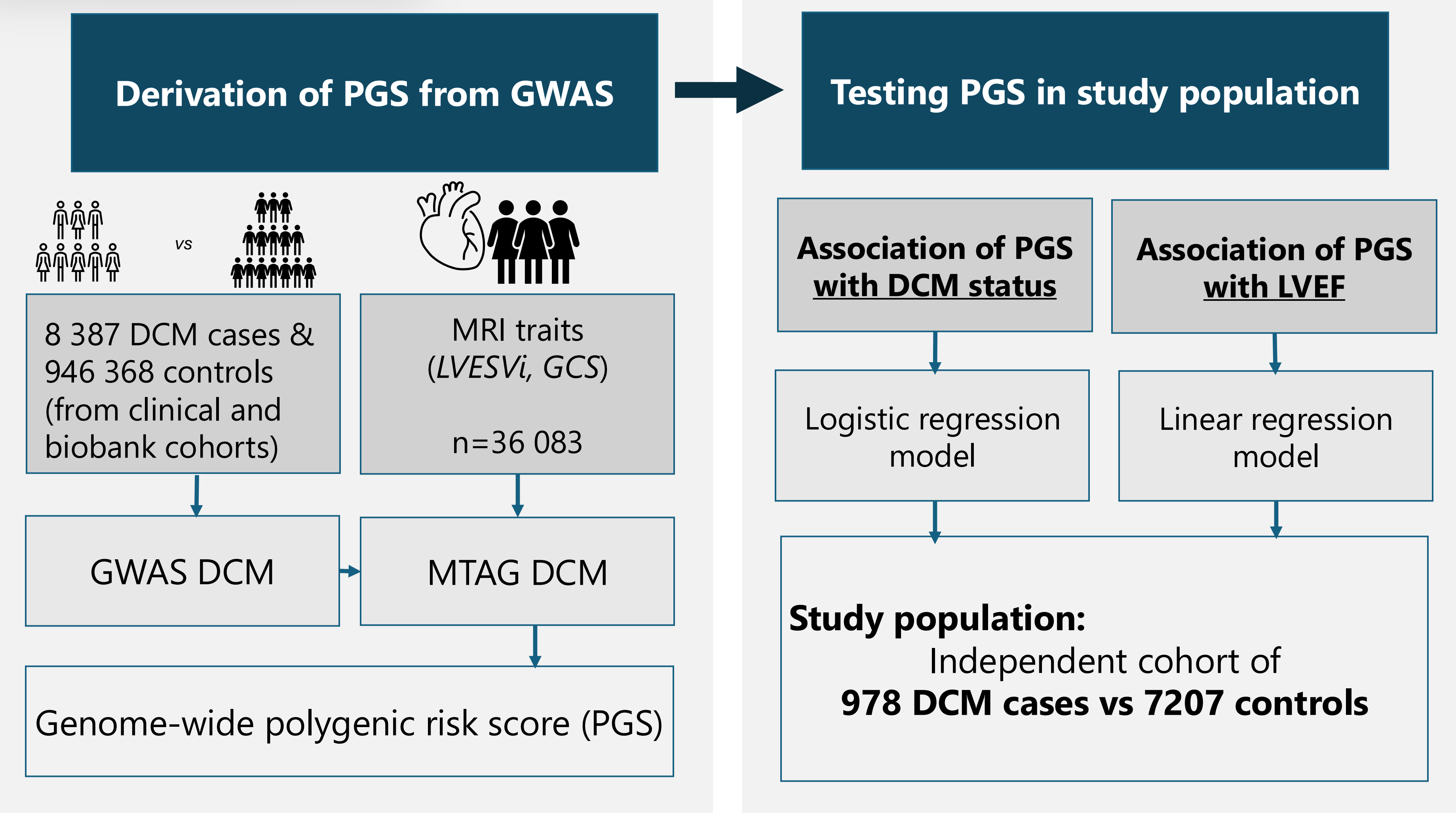
### Aetiology:

- Not fully elucidated;
- Rare genetic variants in 10-50%:
  - Incomplete penetrance;
  - Variable severity;
- Contribution of common genetic variation;

## Purpose

- Evaluate the utility of **PGS** for prediction of DCM.
- Evaluate the **association of PGS with left ventricular ejection fraction (LVEF)**.

## Methods

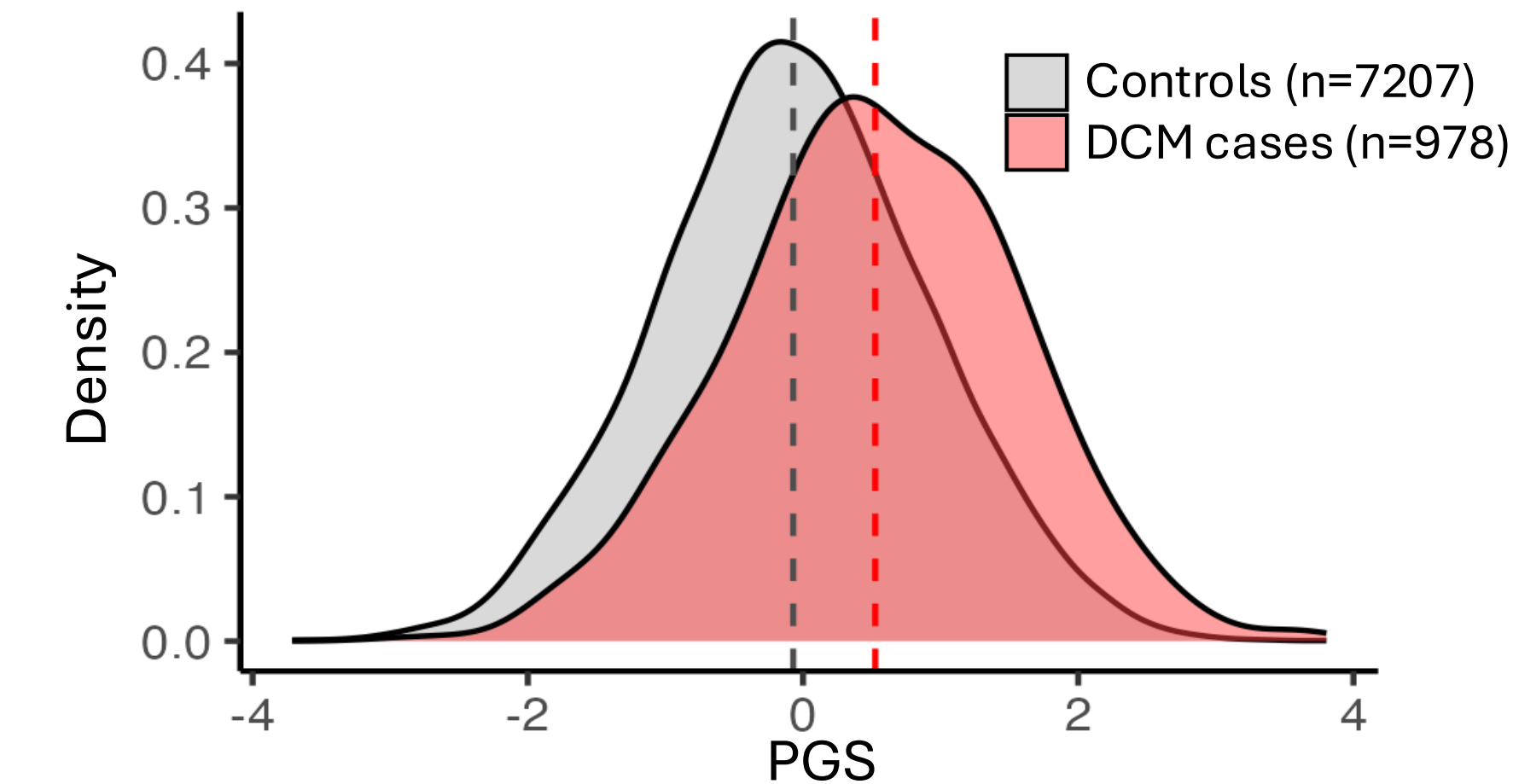


## Results

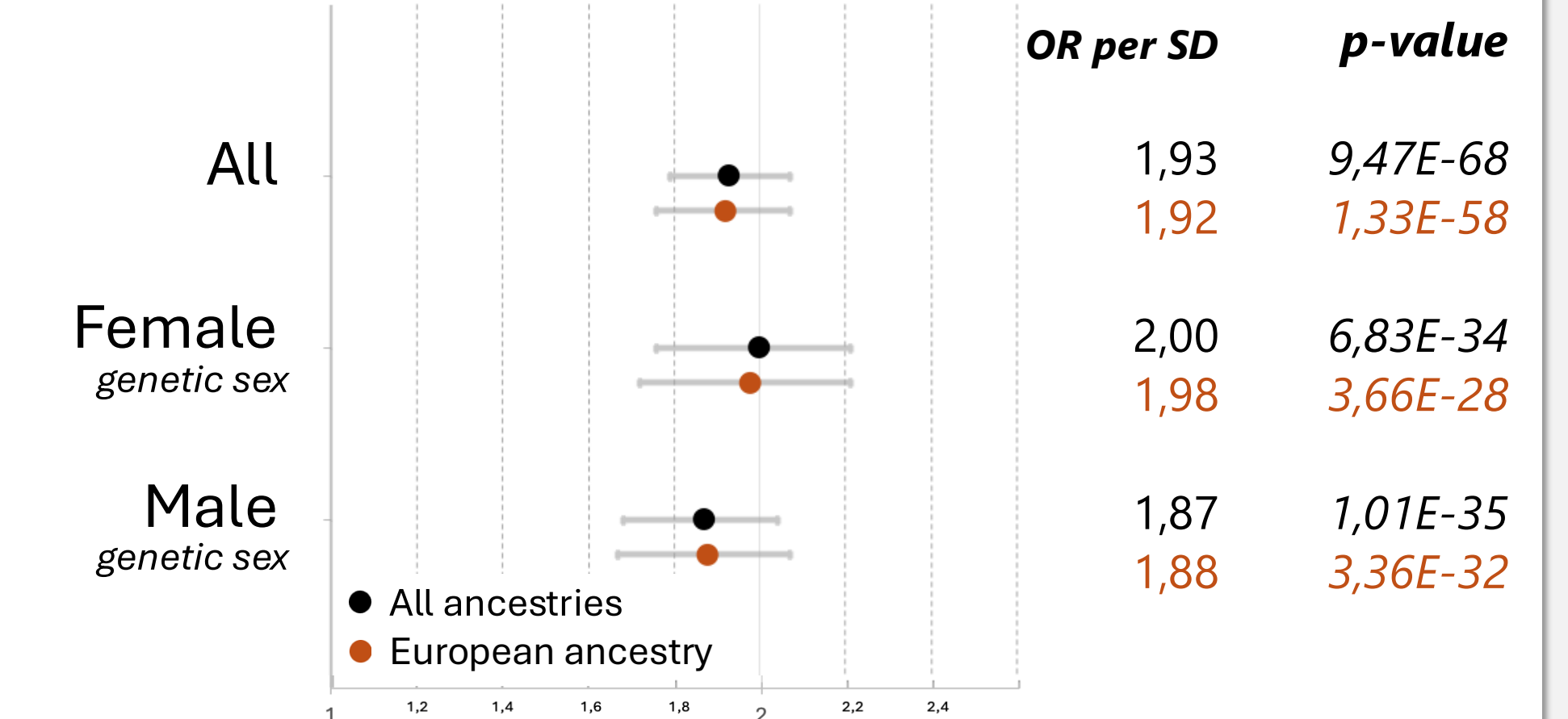
### PGS in DCM

- all
- males
- females

Distribution of PGS among DCM patients and controls

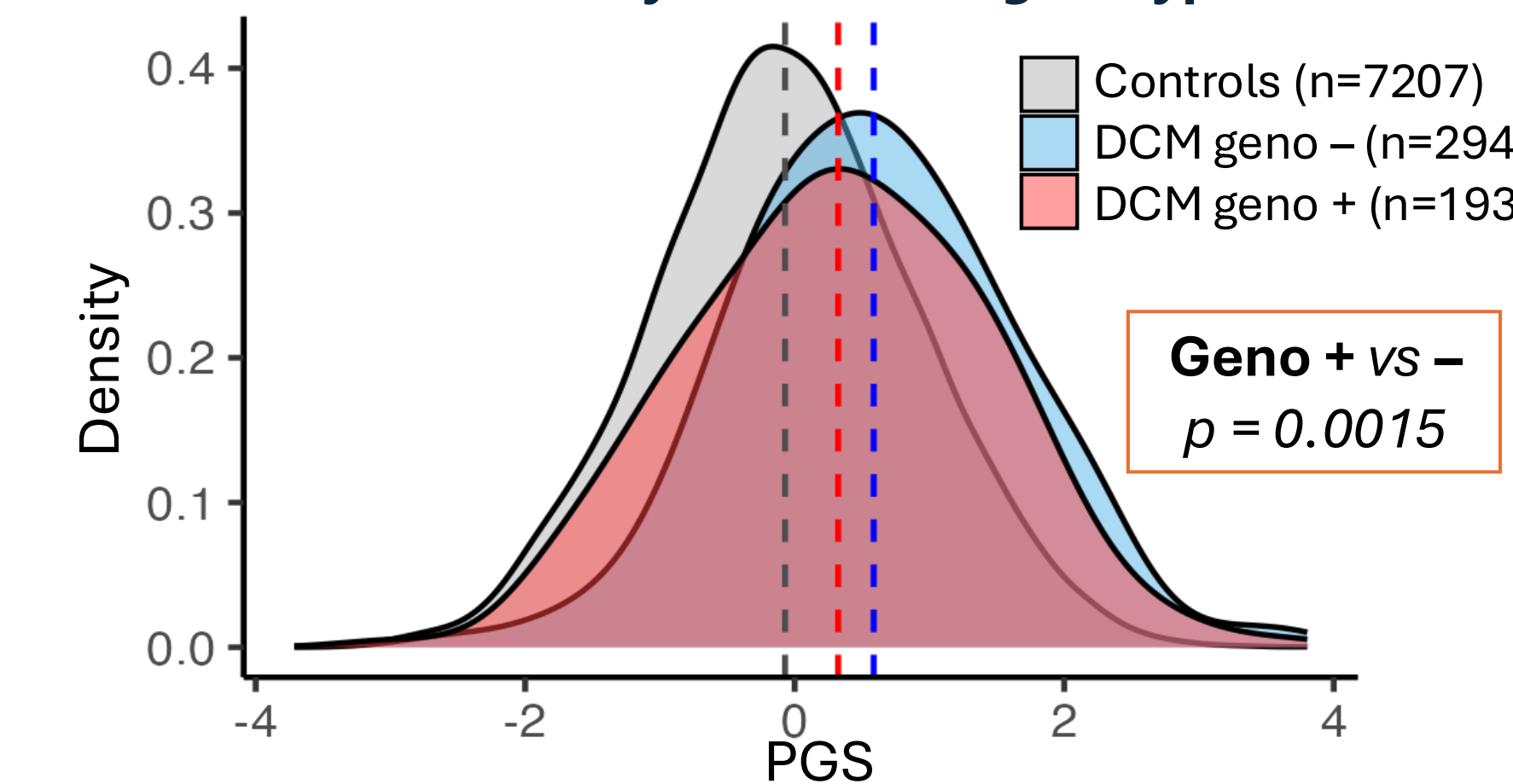


Forest plot with effect sizes for the PGS

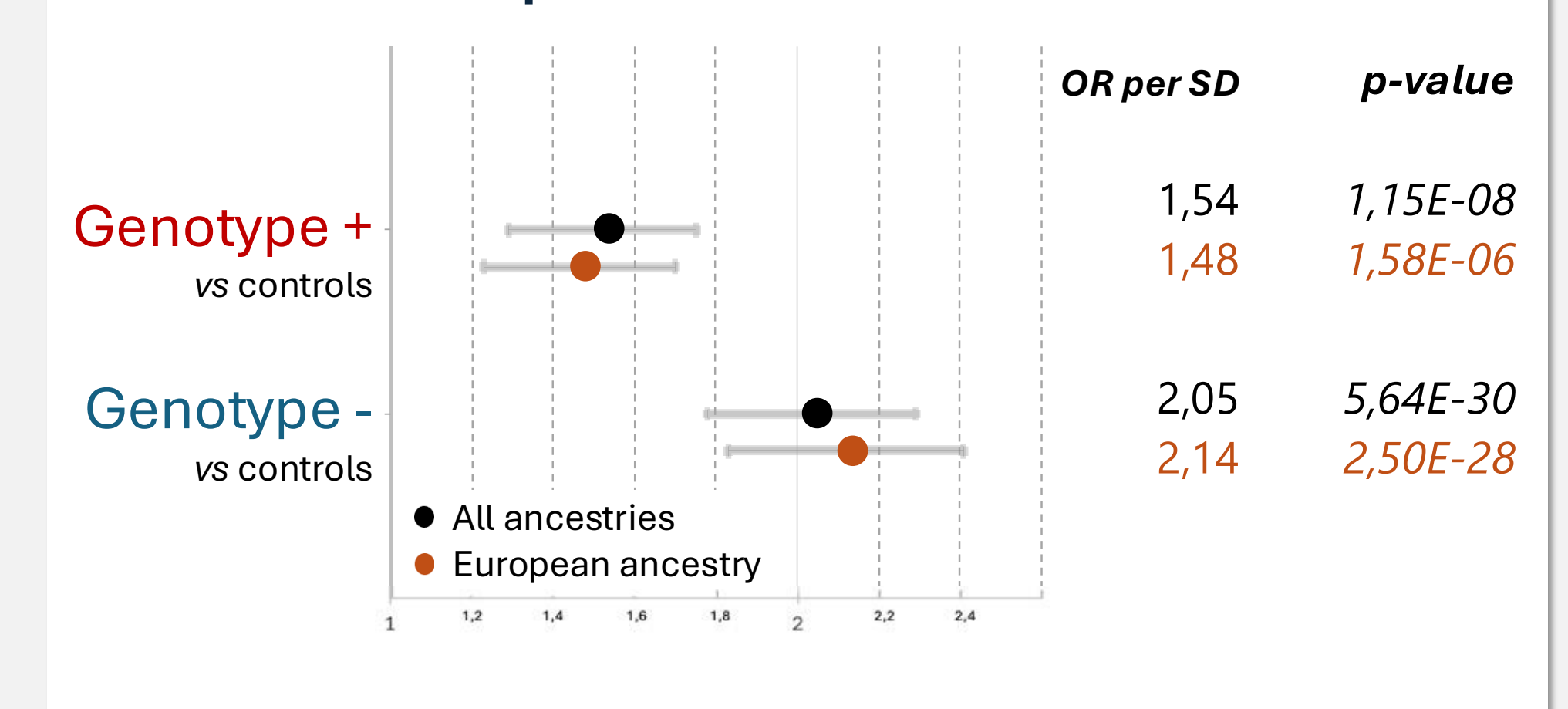


### Genotype + VS Genotype -

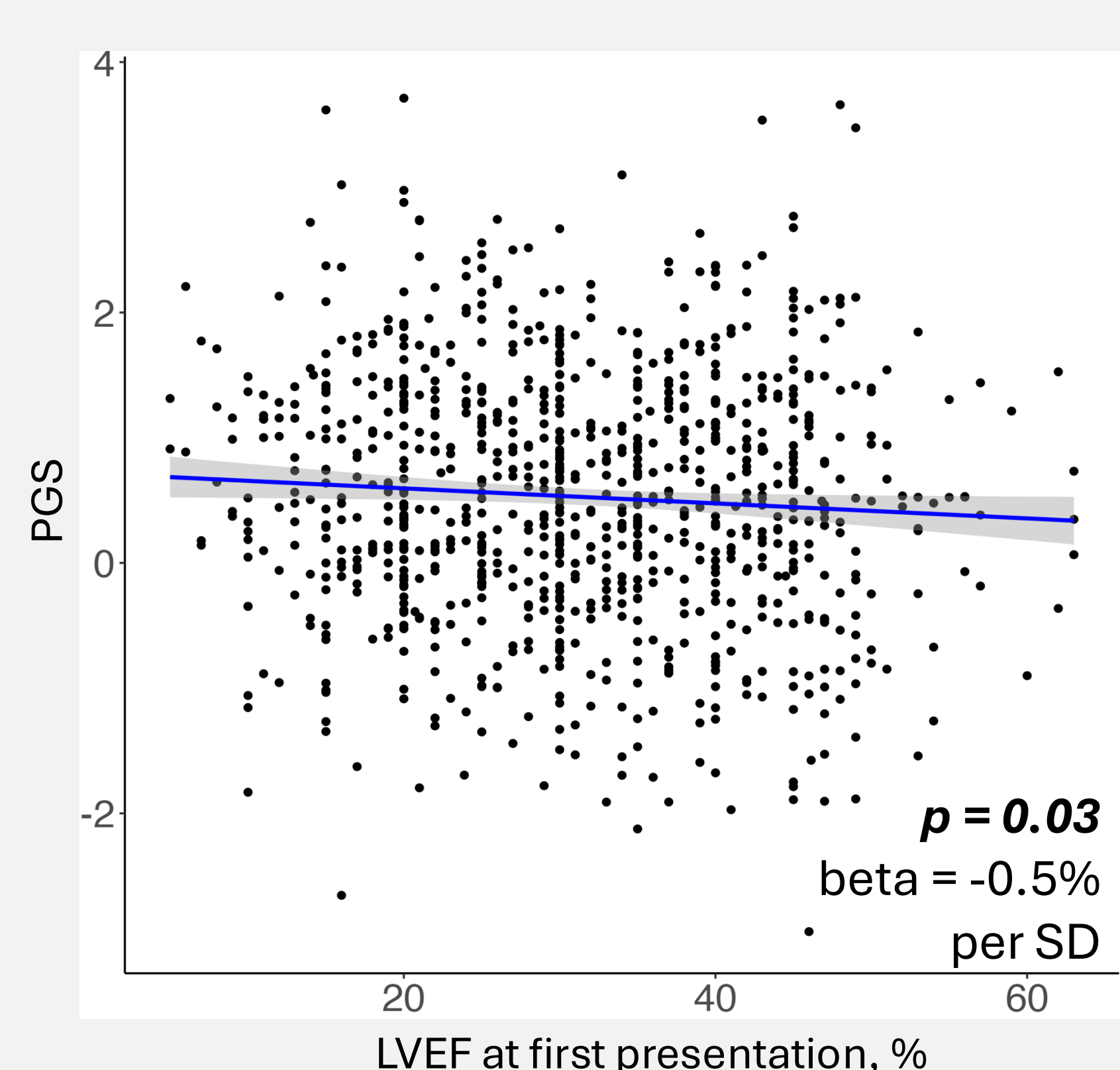
Distribution of PGS by rare variant genotype status



Forest plot with effect sizes for the PGS



### Association of PGS with LVEF at the time of the first presentation



## Conclusions

- PGS is strongly associated with risk of DCM
- PGS may modulate the severity of LV dysfunction among DCM cases.
- Effect present in both genotype + & genotype -

**Potential clinical applicability for PGS in DCM risk stratification.**