

Using Genomics to Understand an Individual's Risk for Common Diseases



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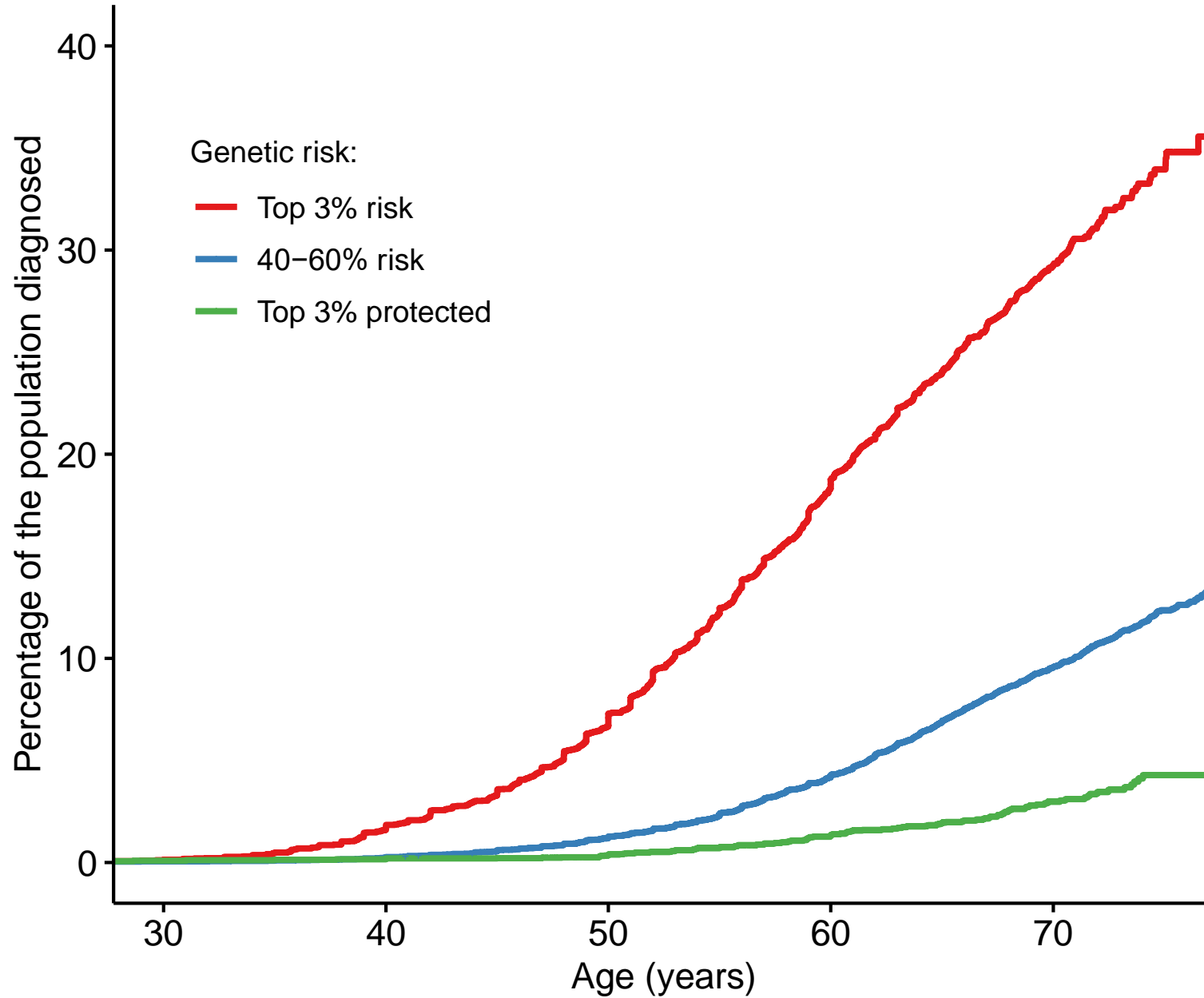
Polygenic Risk Scores: Individual Risk Prediction

For any common disease, we now know hundreds of common genetic variants across the genome which each individually have a small effect on risk.

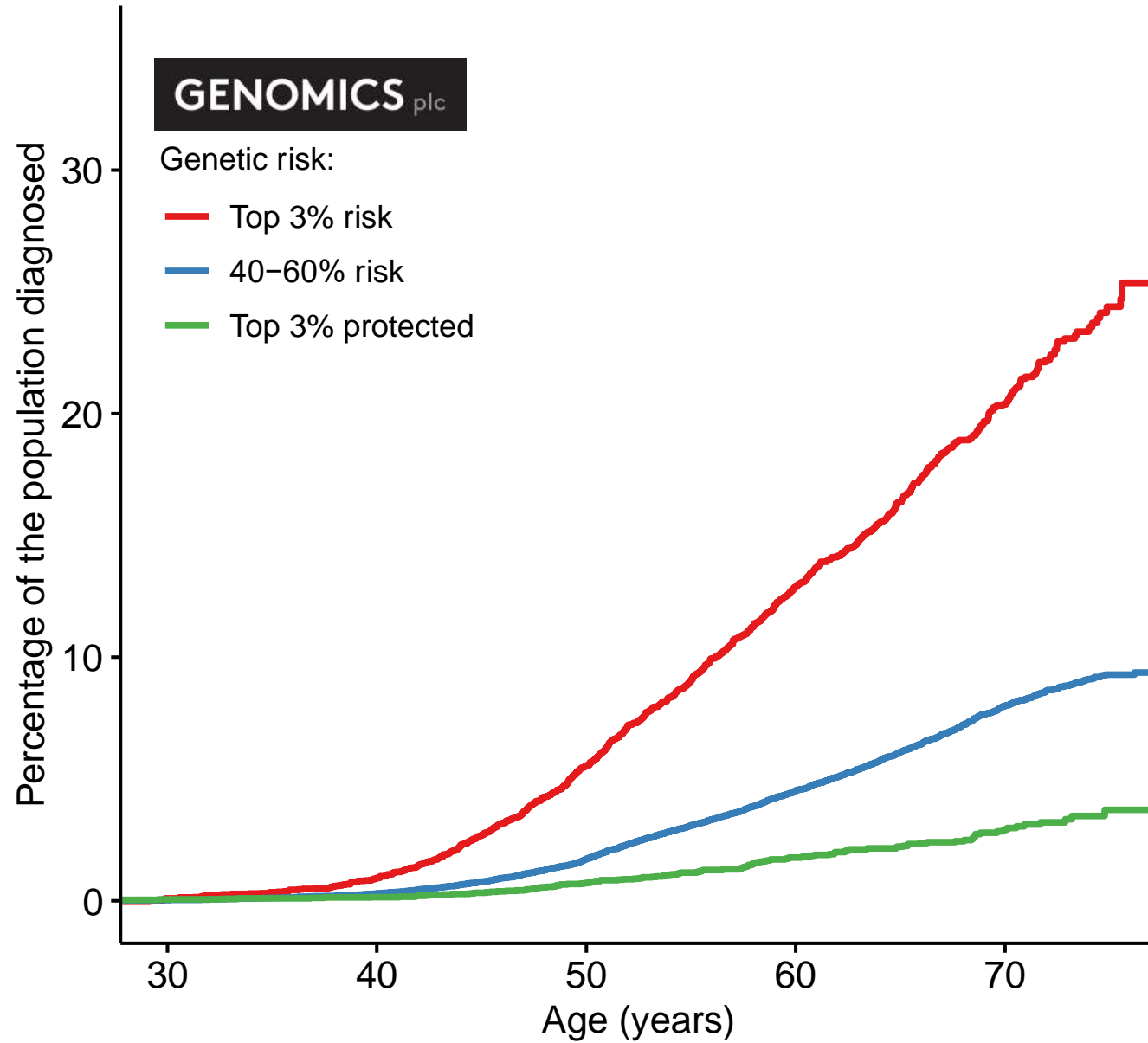
Information from these can be combined to give a “polygenic risk score” – a numerical summary of an individual’s genetic predisposition to that disease.

Across a population, there will be a distribution of these risk scores. Individuals with high values of the scores can be at significant (and clinically actionable) increased risk.

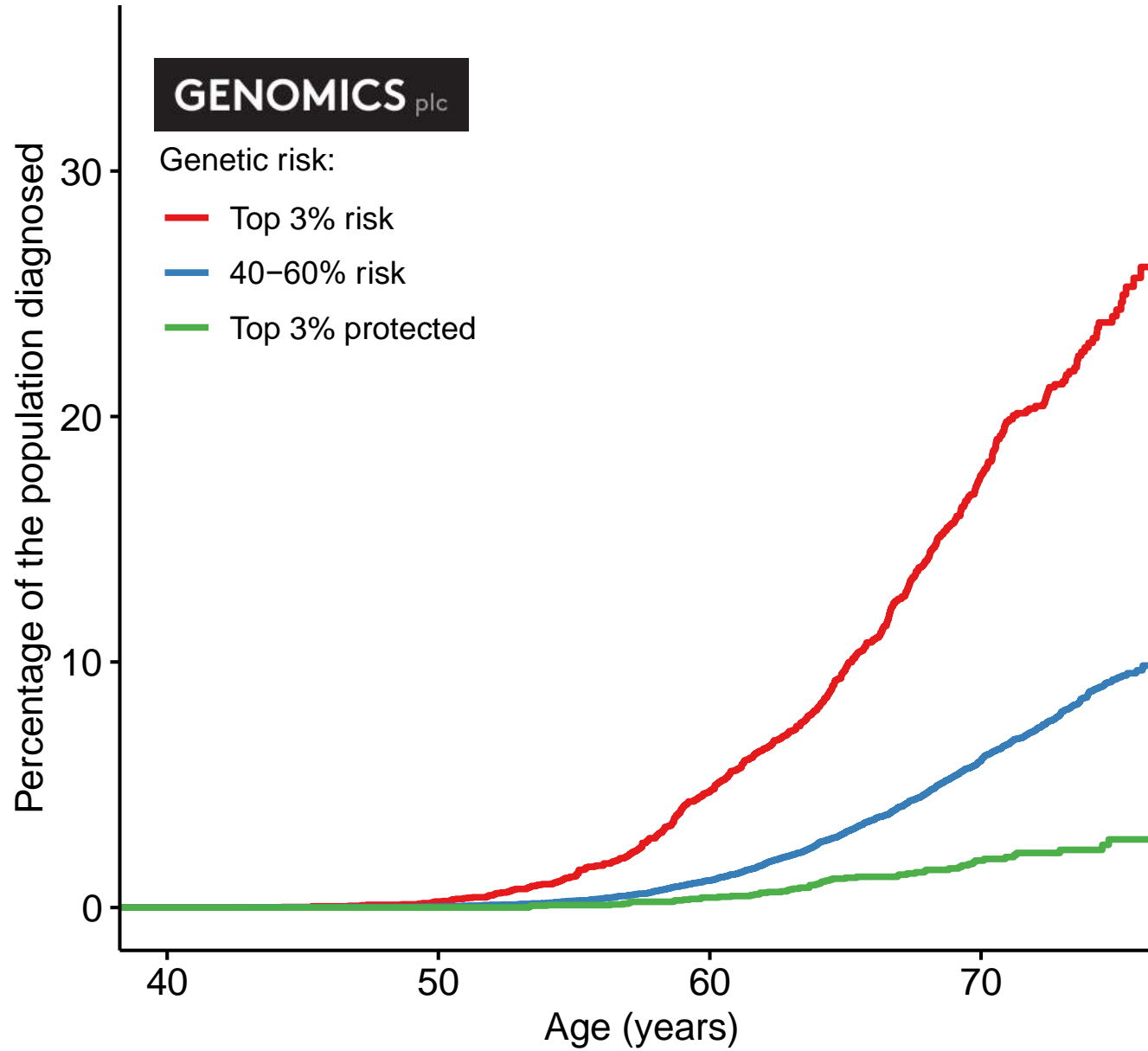
Coronary Artery Disease (men only)



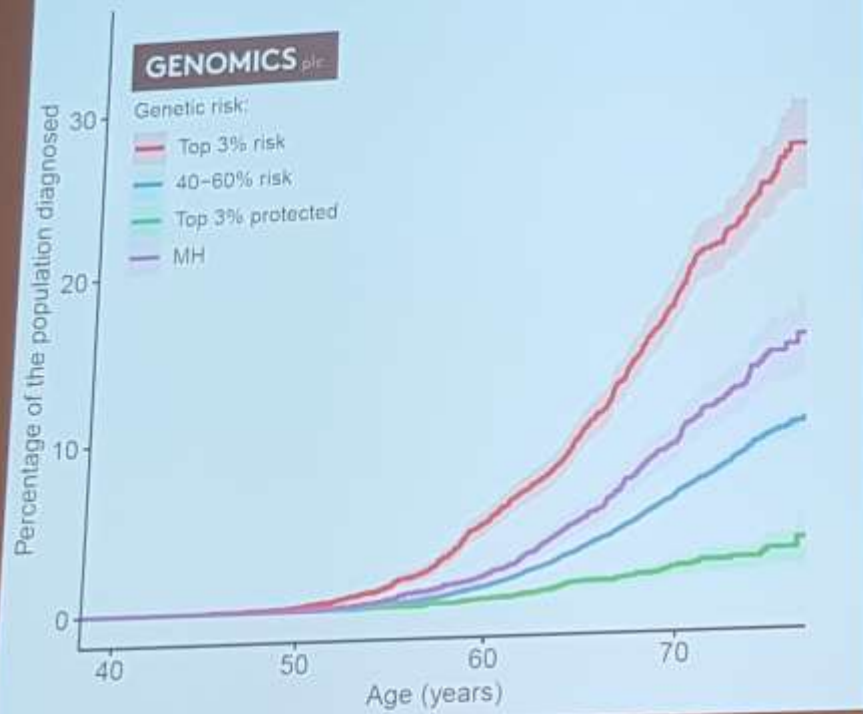
Breast Cancer (women only)



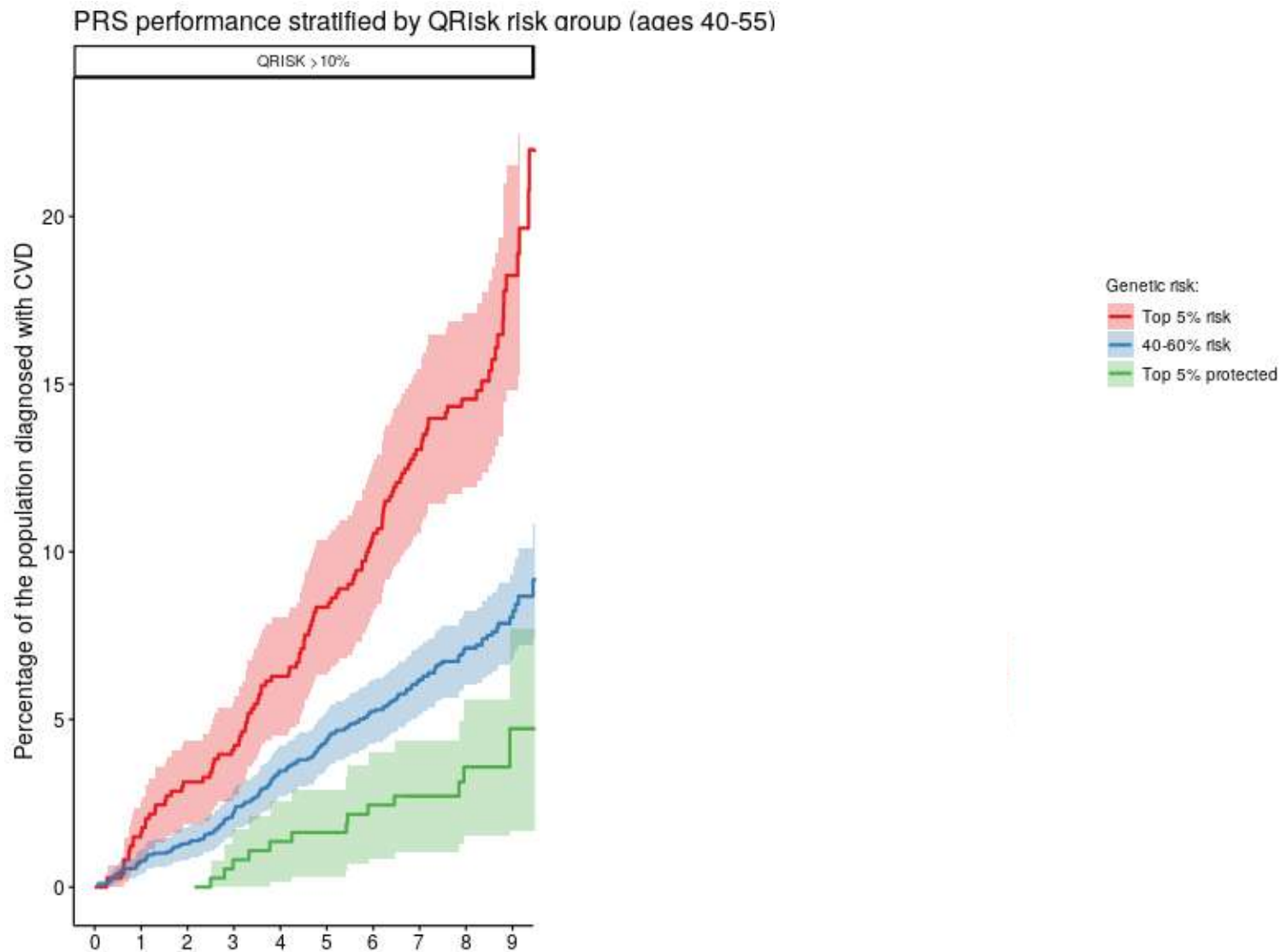
Prostate Cancer (men only)



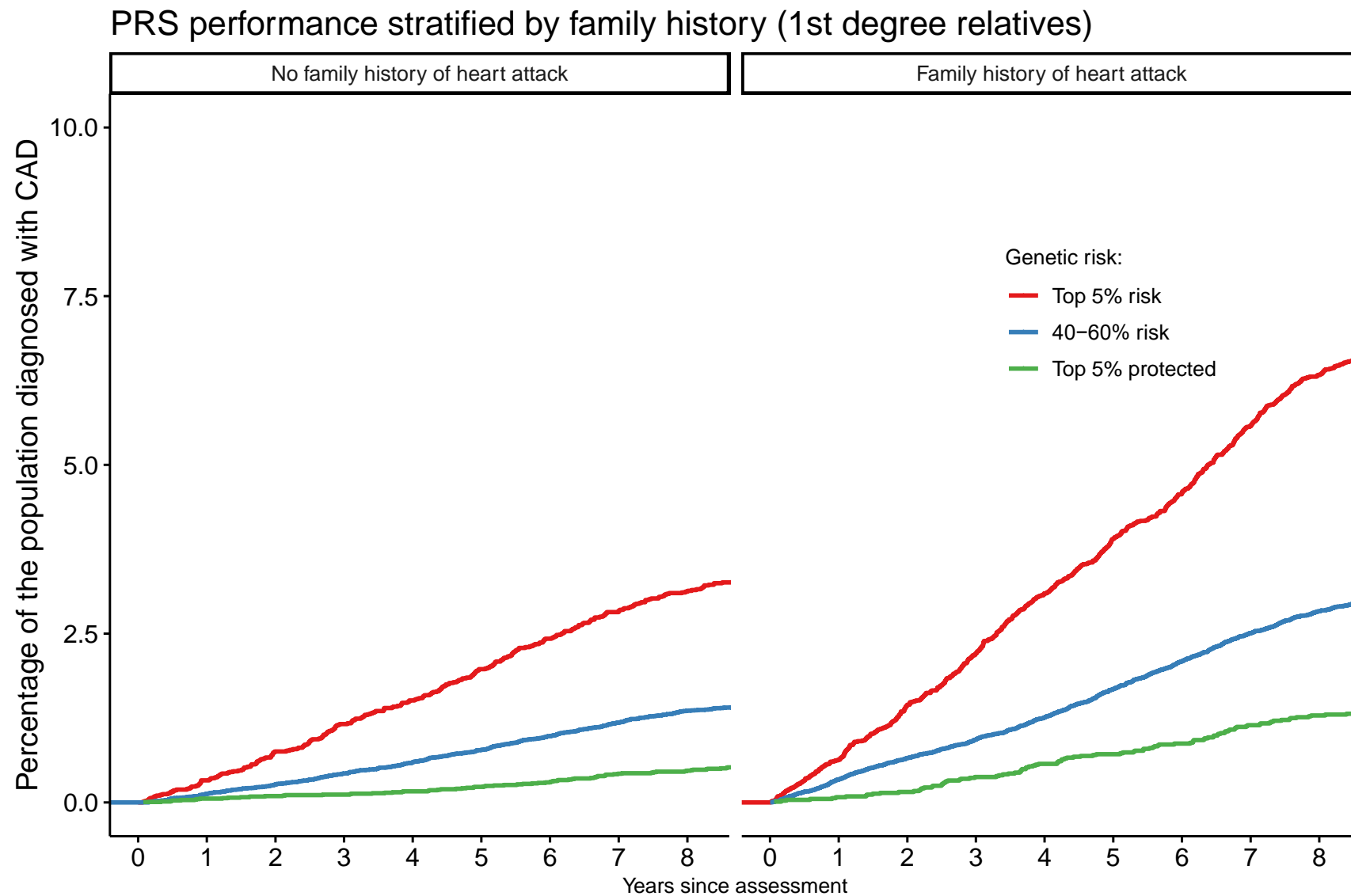
Prostate Cancer (men only)



PRS and Non-Genetic Risk Factors



PRS and family history



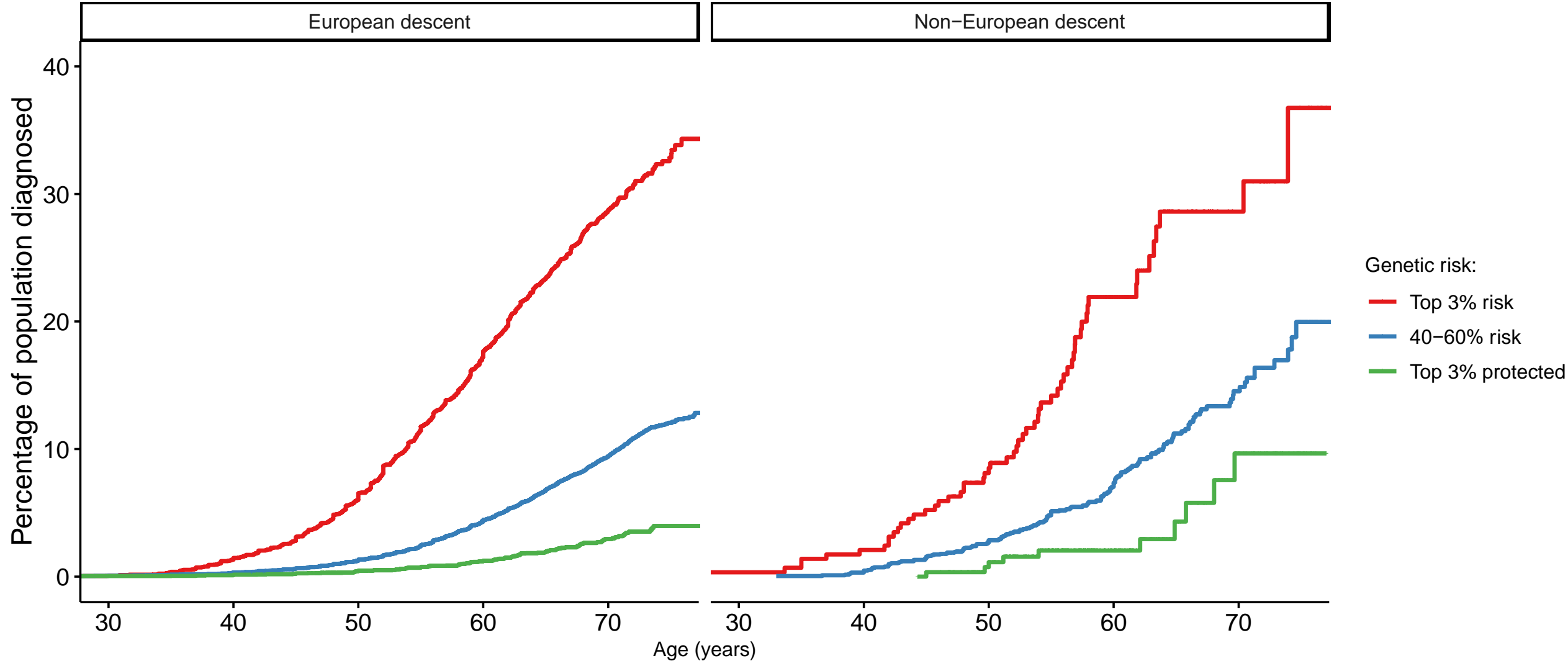
QRISK alone or with PRS

Consider 13M UK adults aged 40-55, using QRISK > 10%

	QRISK only		QRISK + PRS		CAD Diagnoses in group at 8yrs
Above Absolute risk for statins	1,040K		1,326K	798K	
				528K	14.7K
Below absolute risk for statins	11,960K		11,674K	242K	3.9K
				11,432K	

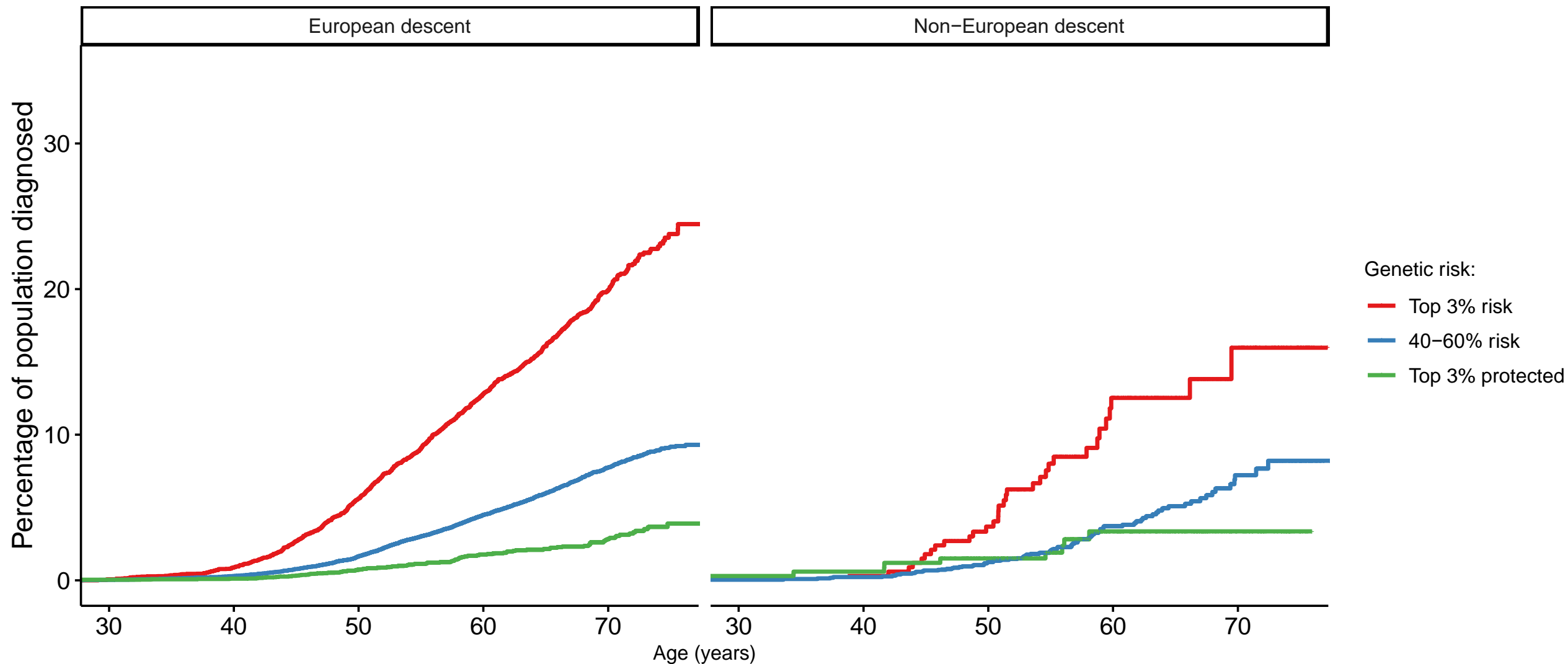
Non-European Ancestries are challenging

Coronary artery disease (men only)



Non-European Ancestries are challenging

Breast cancer (women only)



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