台灣精準醫療計畫
Taiwan Precision Medicine Initiative
成果暨新聞發表會

中央研究院
郭沛恩 院士

台灣精準醫療計畫

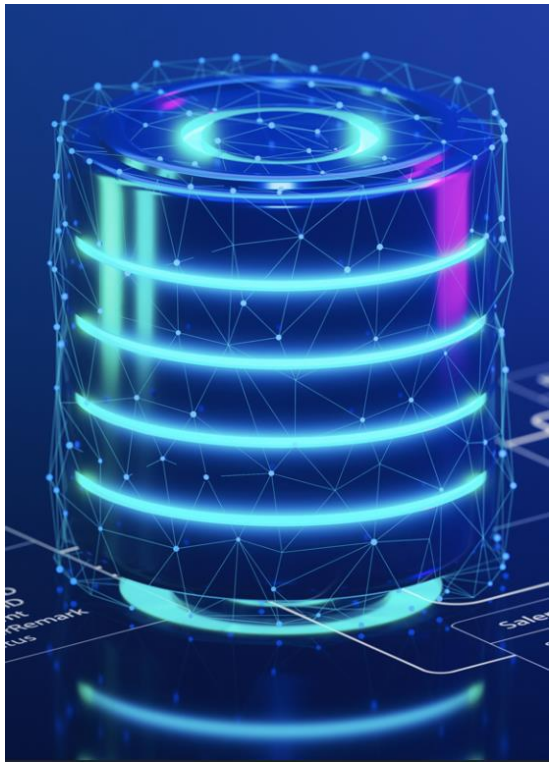
Taiwan Precision Medicine Initiative

中央研究院與16個醫療體系
共同執行的多中心研究計畫

精準醫療如何促進健康?

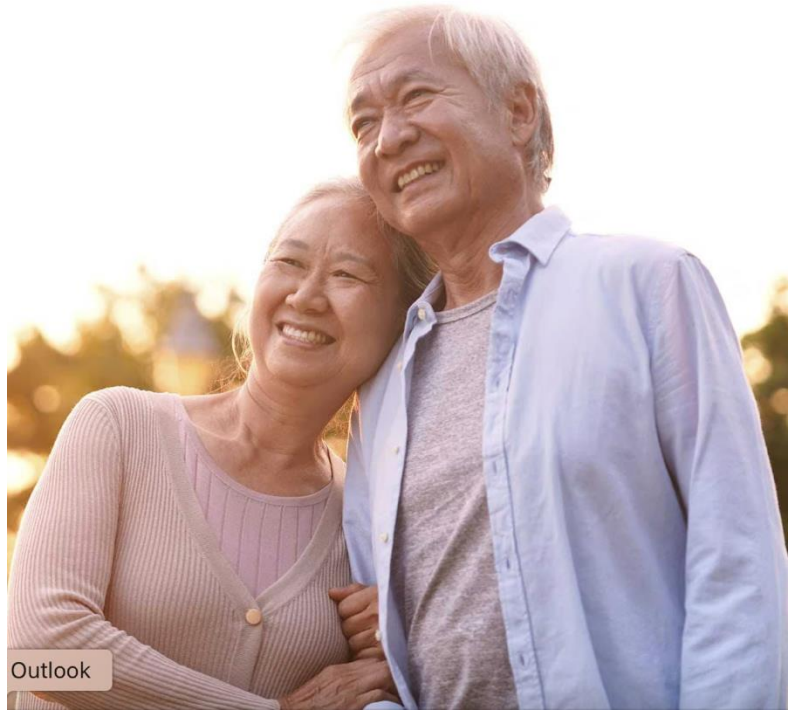
個人資訊

比對
族群資料



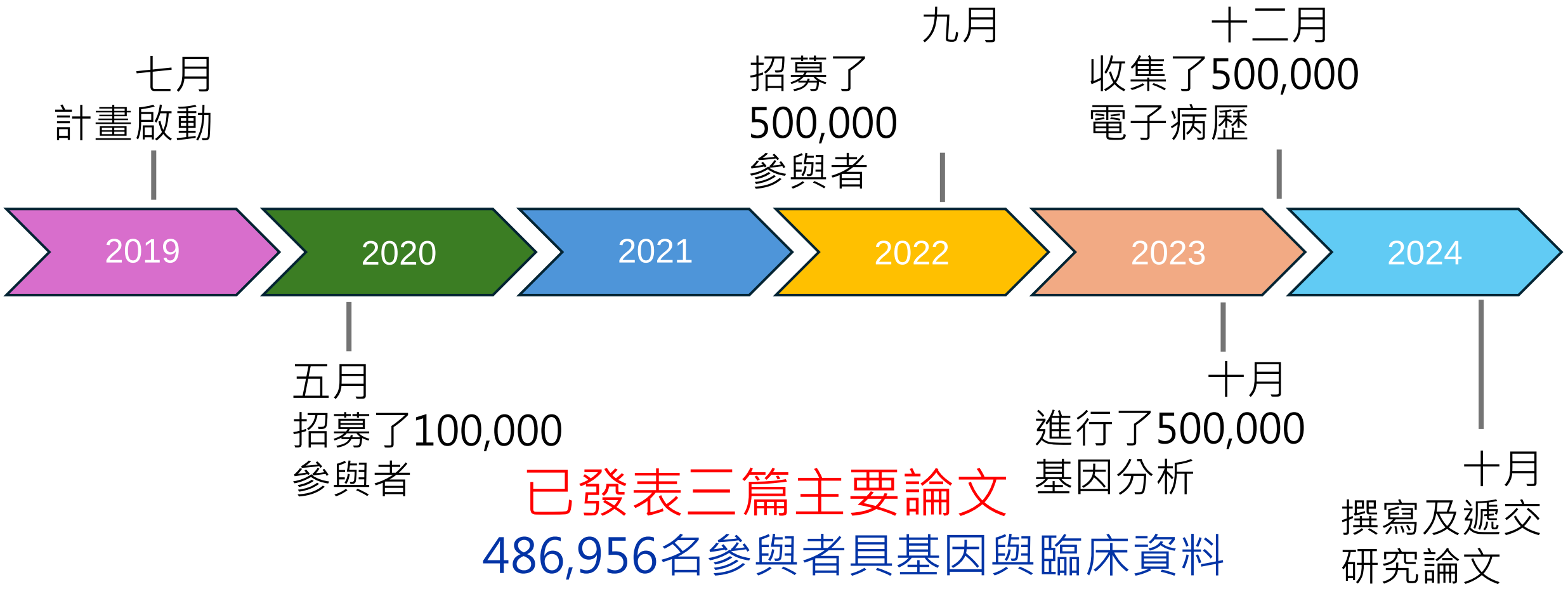
參考資料庫

最佳化
醫療照護



健康老

台灣精準醫療計劃時間表



已發表三篇主要論文

486,956名參與者具基因與臨床資料

每位參與者研究花費為新臺幣1800元

持續強化台灣在精準健康的國際競爭力與影響力

科學面向 (TPMI PHASE II)

- 進行再同意以完善 TPMI 參與者的臨床資料
- 針對所有國人常見疾病建立風險預測模型
- 驗證並優化疾病風險預測演算法
- 測試「風險導向健康管理」的有效性

公衛面向 (PRECISION HEALTH 2035)

- 儘速完成青壯年族群的基因型鑑定分析
- 推廣健康生活

整合式疾病風險計算器 (Assess 評估)
+ 健康管理策略 (Advise 建議)
+ 健康生活 (Act 行動)
= 健康老 (Healthy aging)

預期可降低常見疾病的醫療負擔愈社會成本負擔, 並改善占全球約20%人口的健康狀況

中央研究院 ACADEMIA SINICA
生物醫學科學研究所
INSTITUTE OF BIOMEDICAL SCIENCES

中央研究院
Academia Sinica

NSTC 國家科學及技術委員會
National Science and Technology Council

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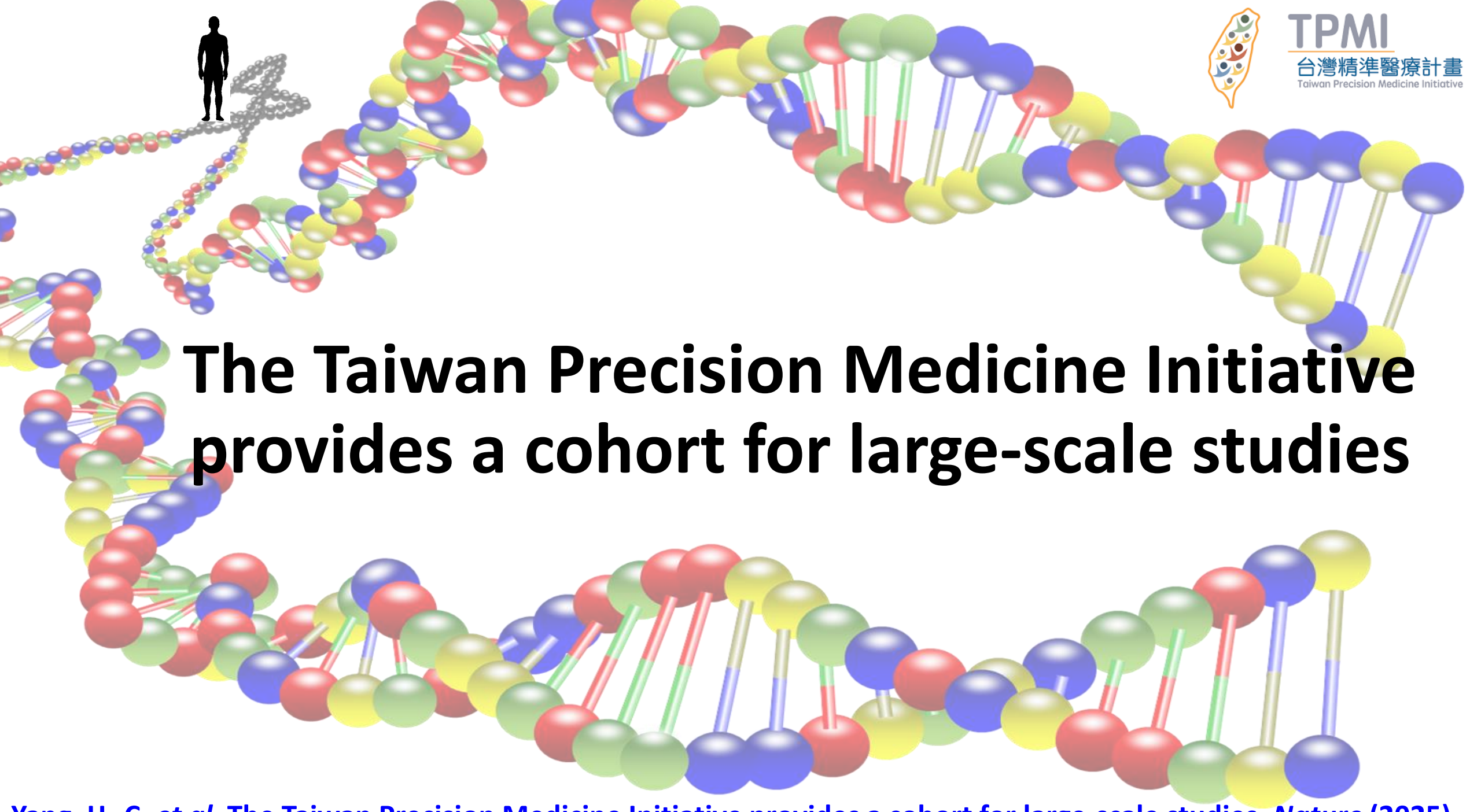
中央研究院

楊欣洲 研究員



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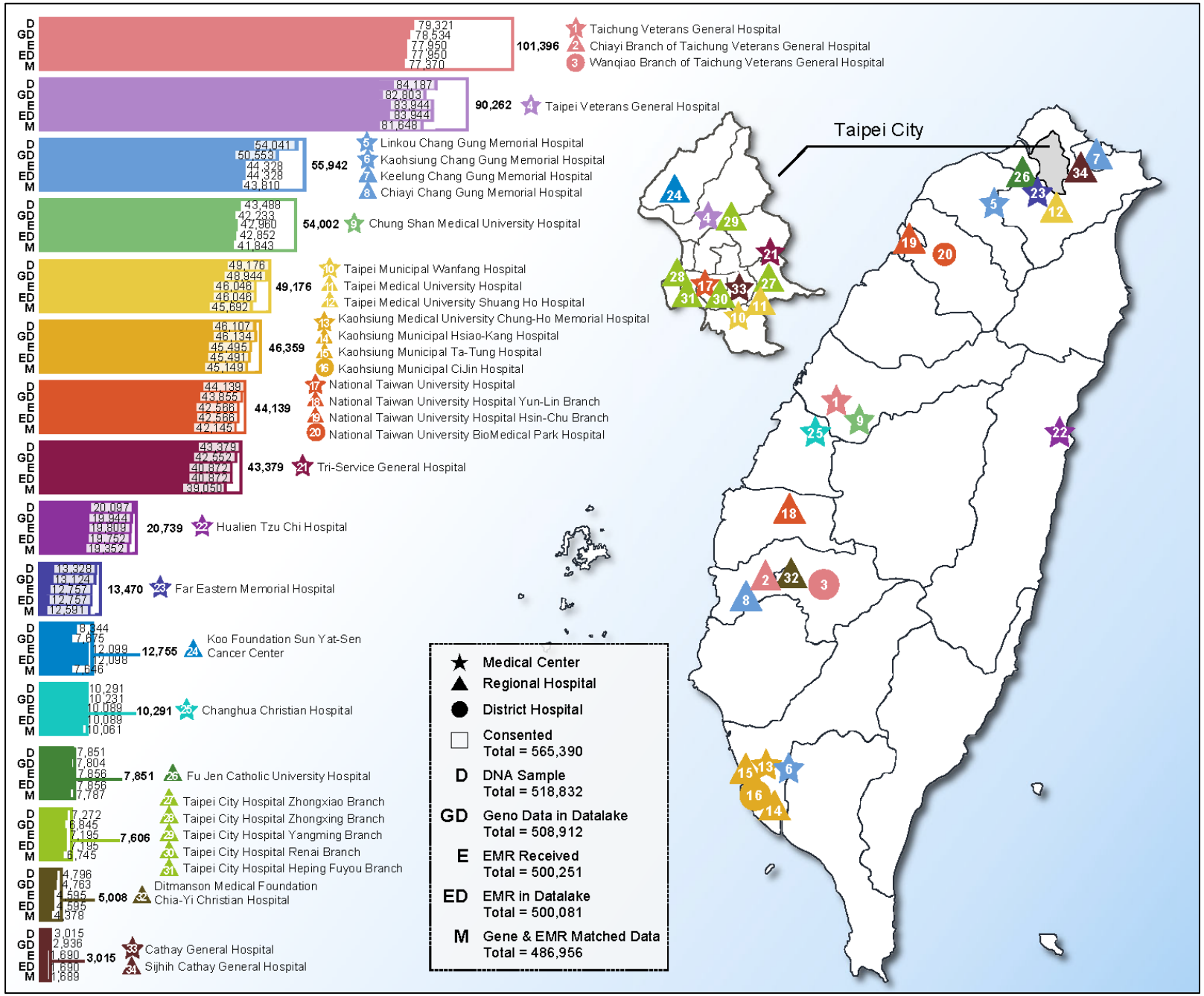
The Taiwan Precision Medicine Initiative provides a cohort for large-scale studies

Yang, H.-C. *et al.* The Taiwan Precision Medicine Initiative provides a cohort for large-scale studies. *Nature* (2025).



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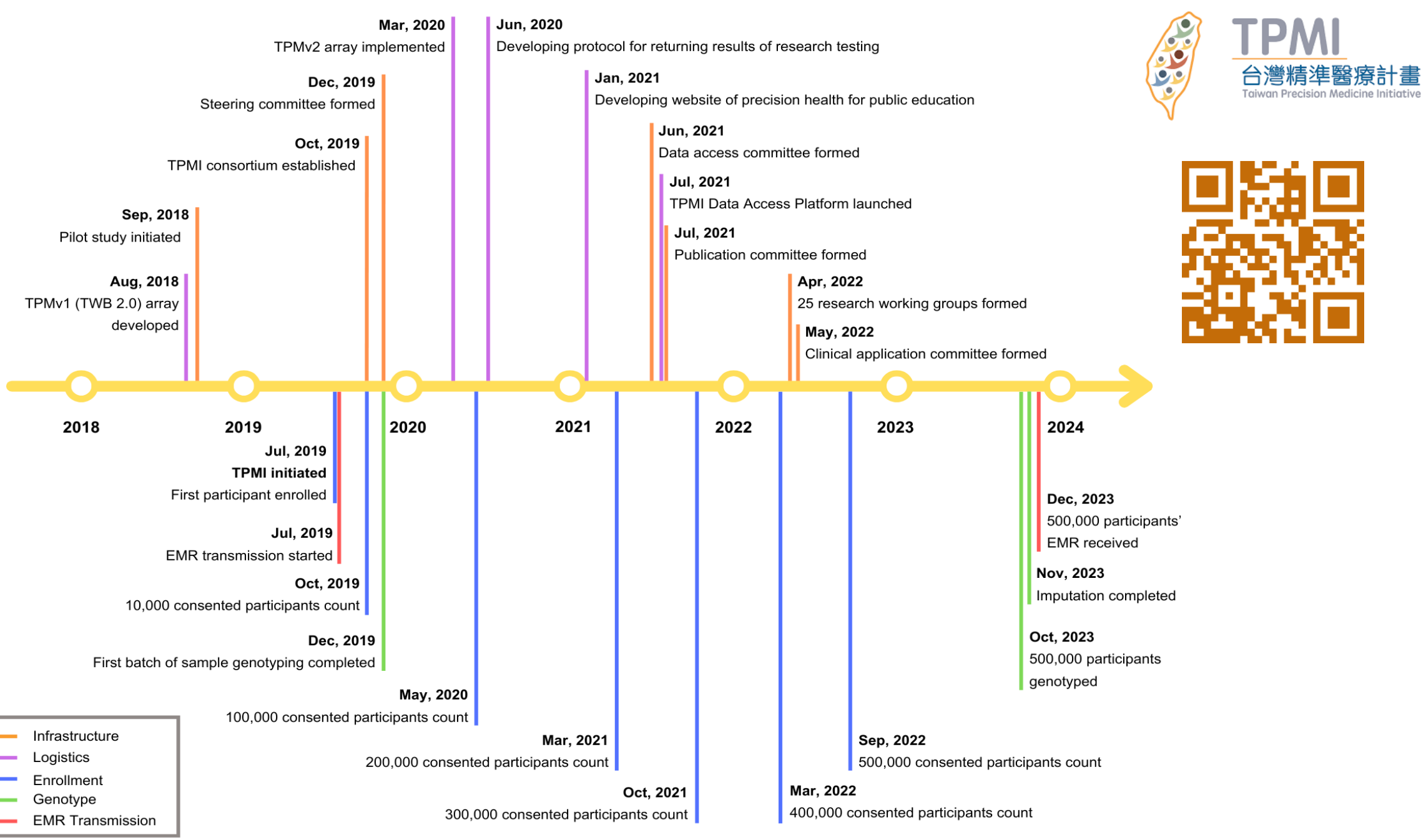

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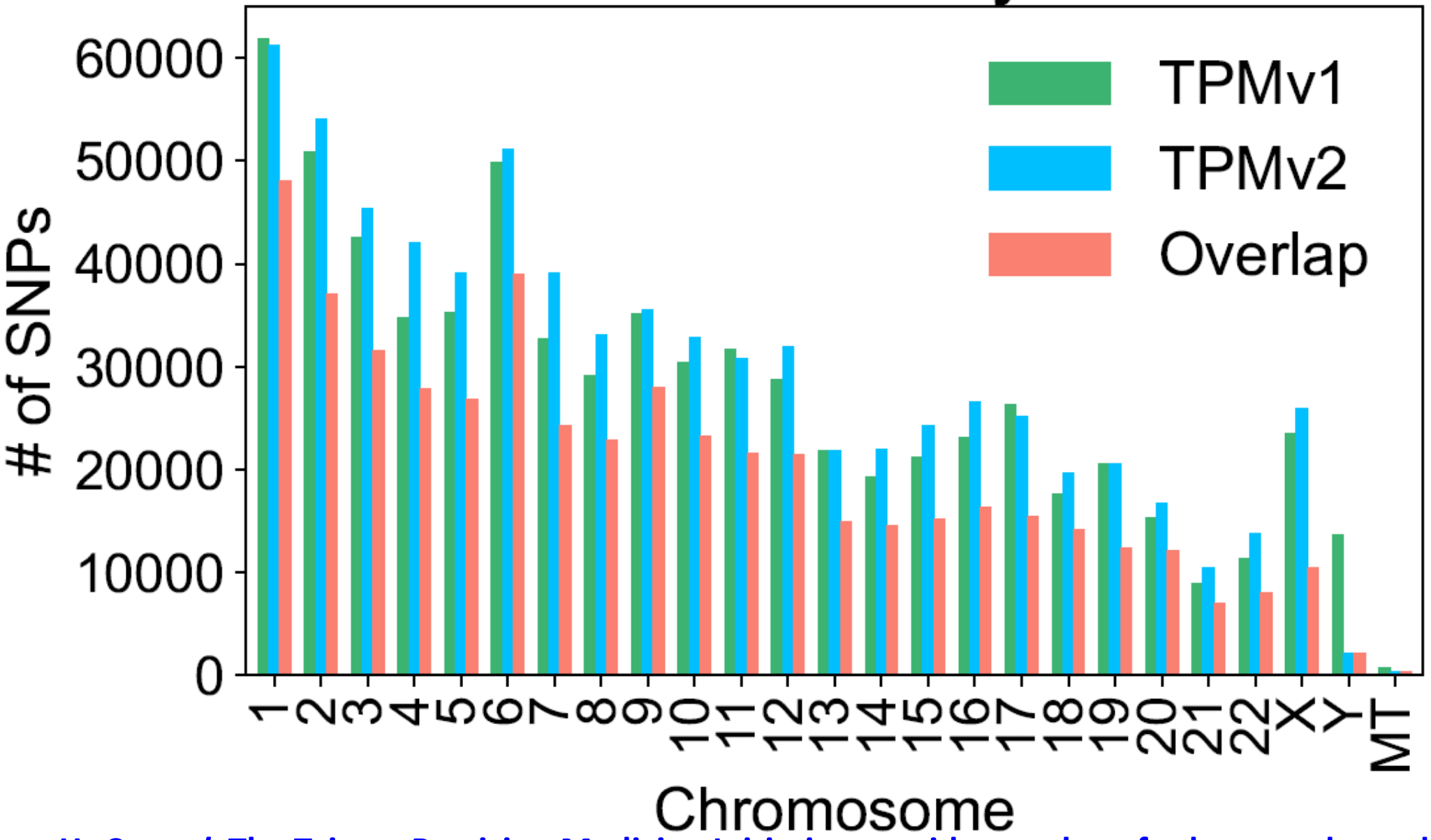
TPM array



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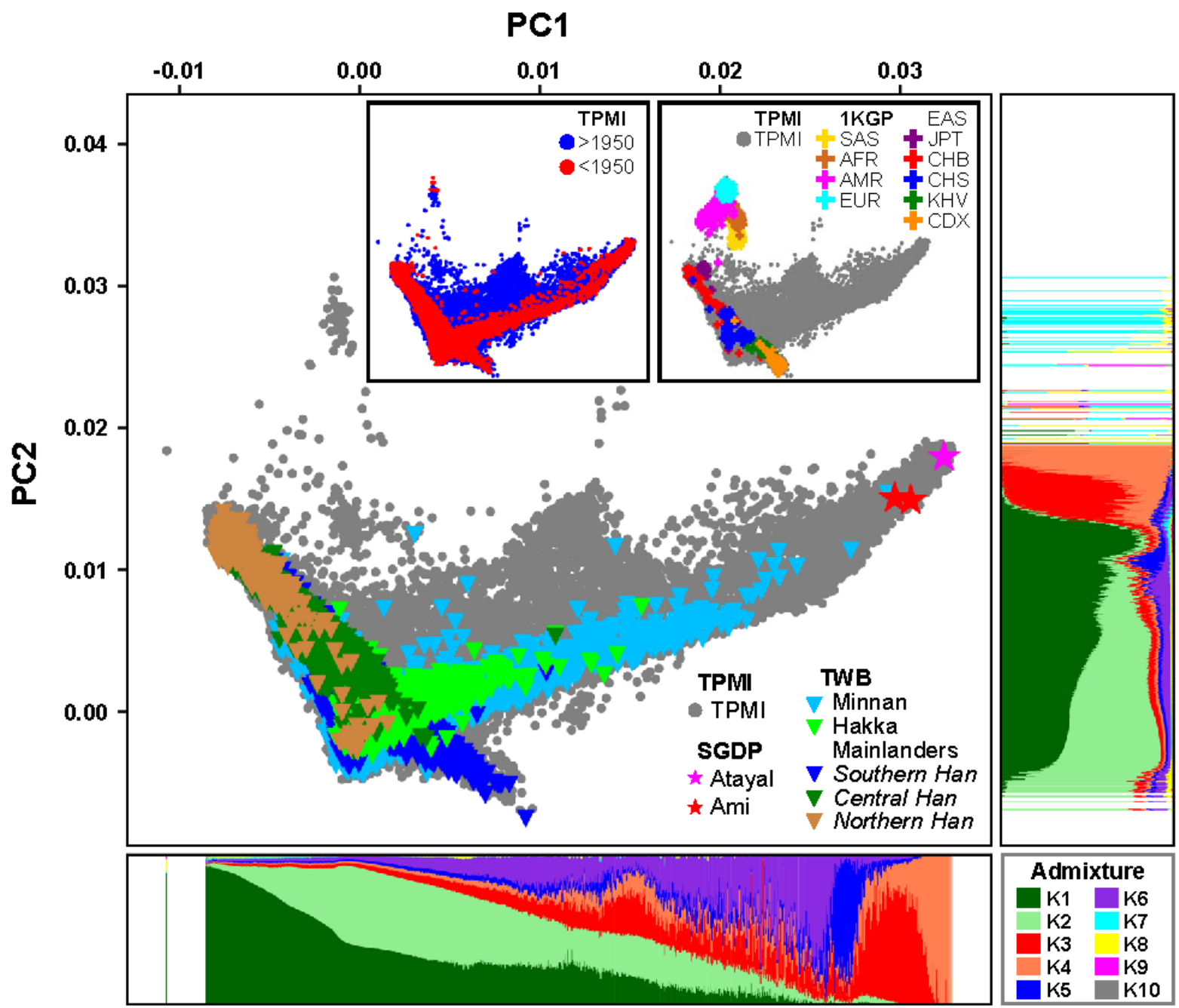

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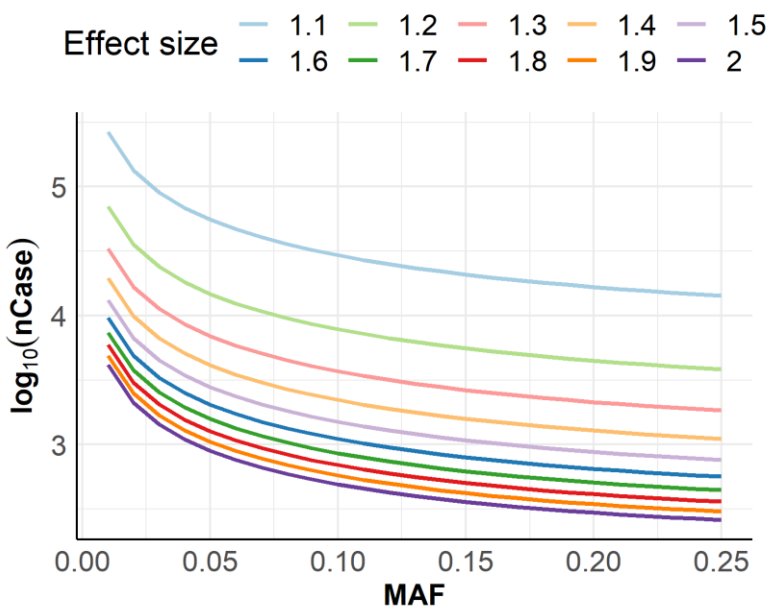

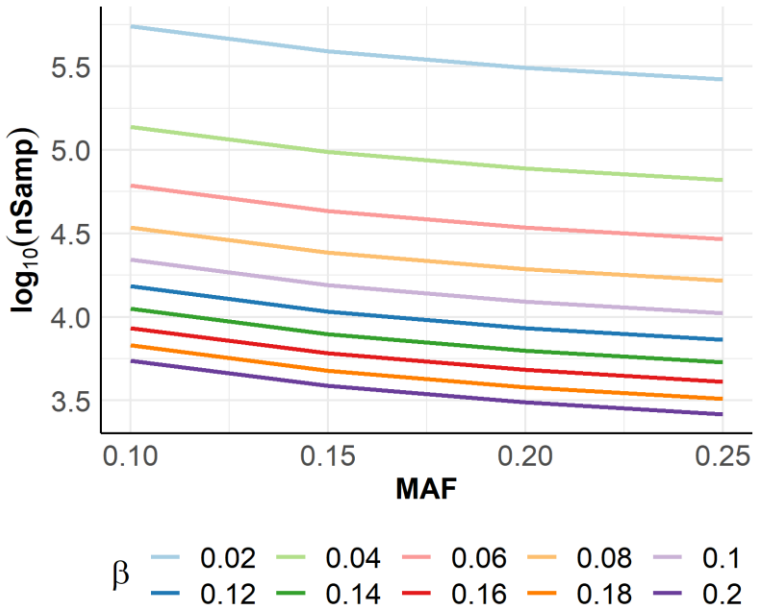

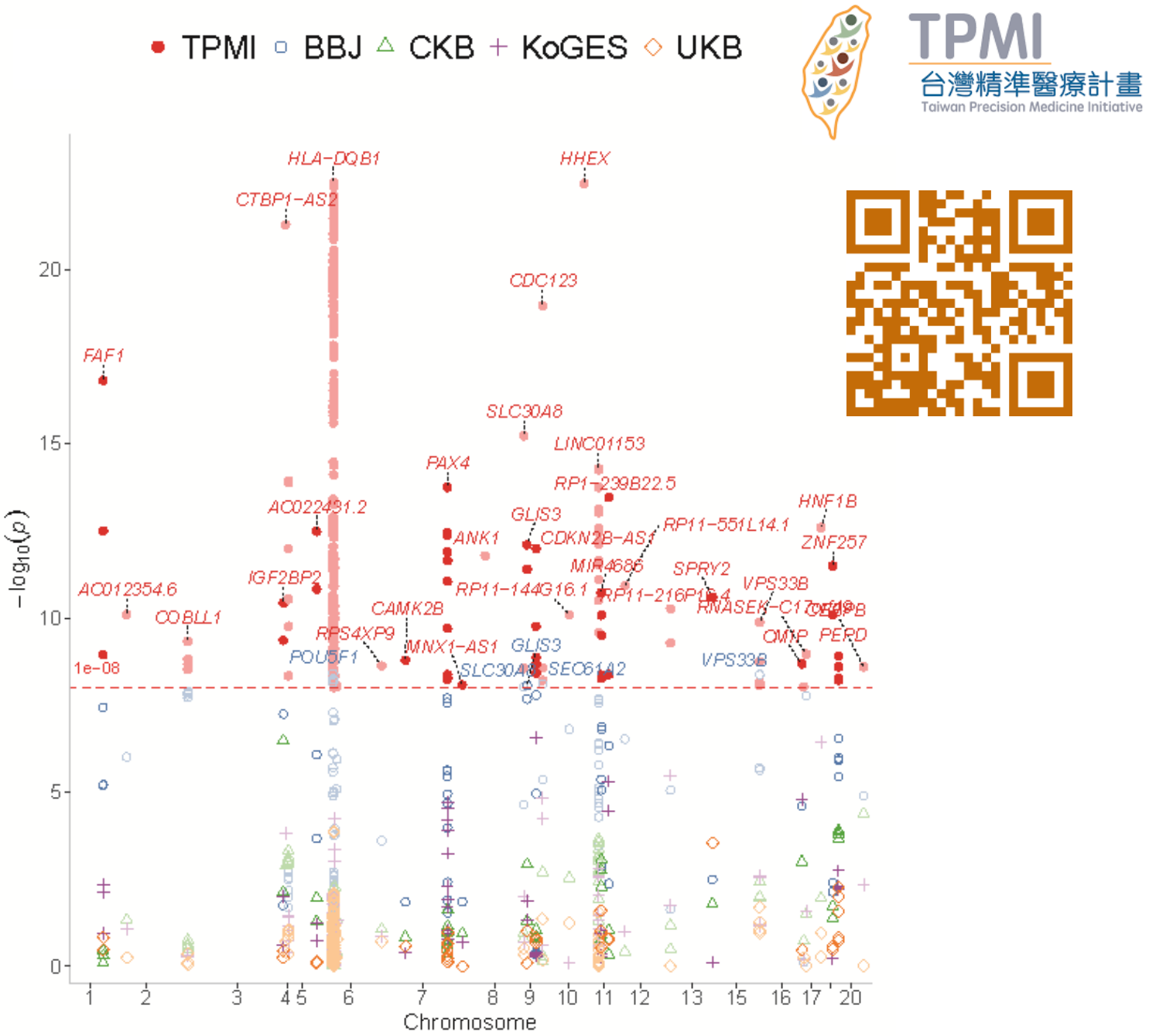

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謝謝聆聽

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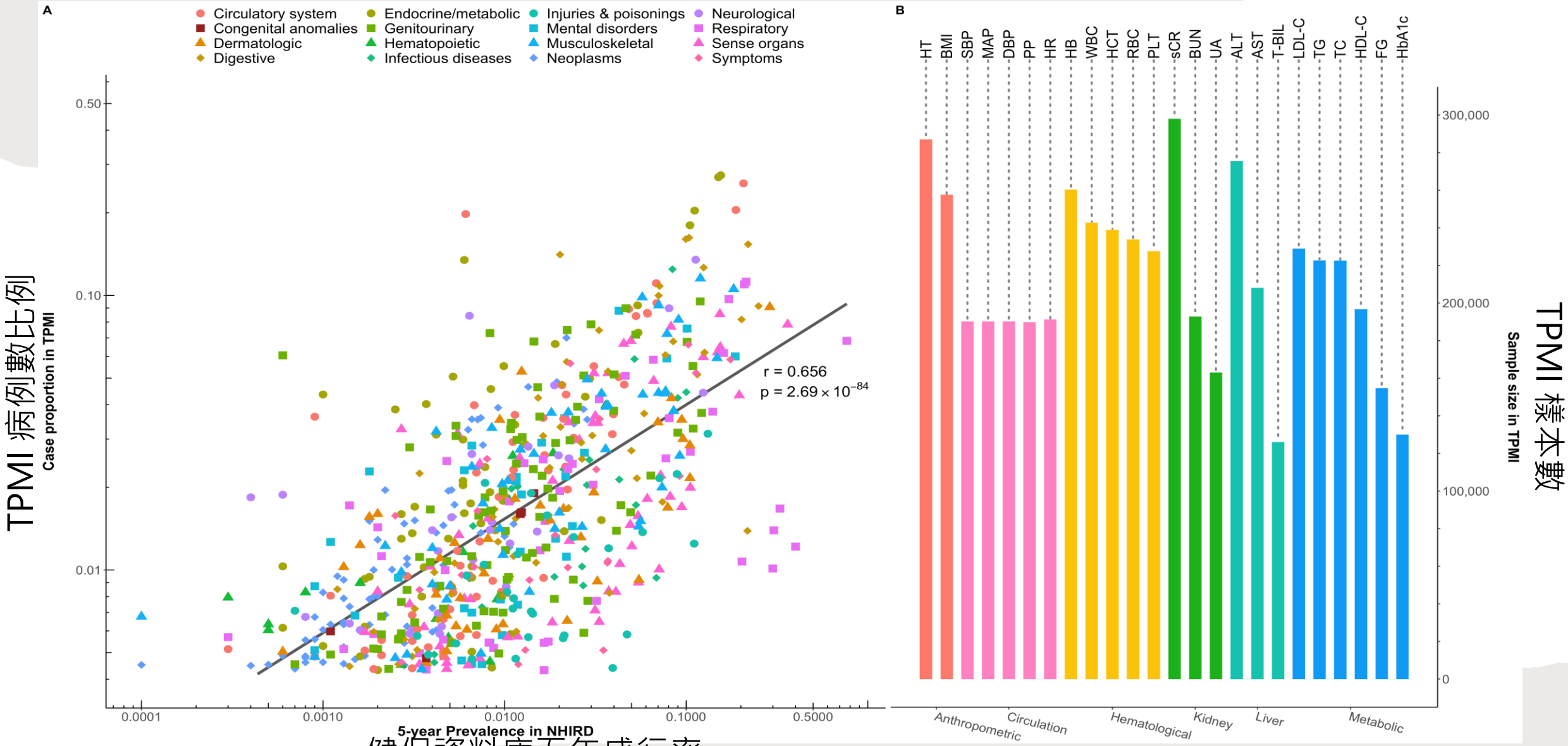
陳弘昕 助研究員

Population-specific polygenic risk scores for people of Han Chinese ancestry

Diseases and quantitative traits in TPMI

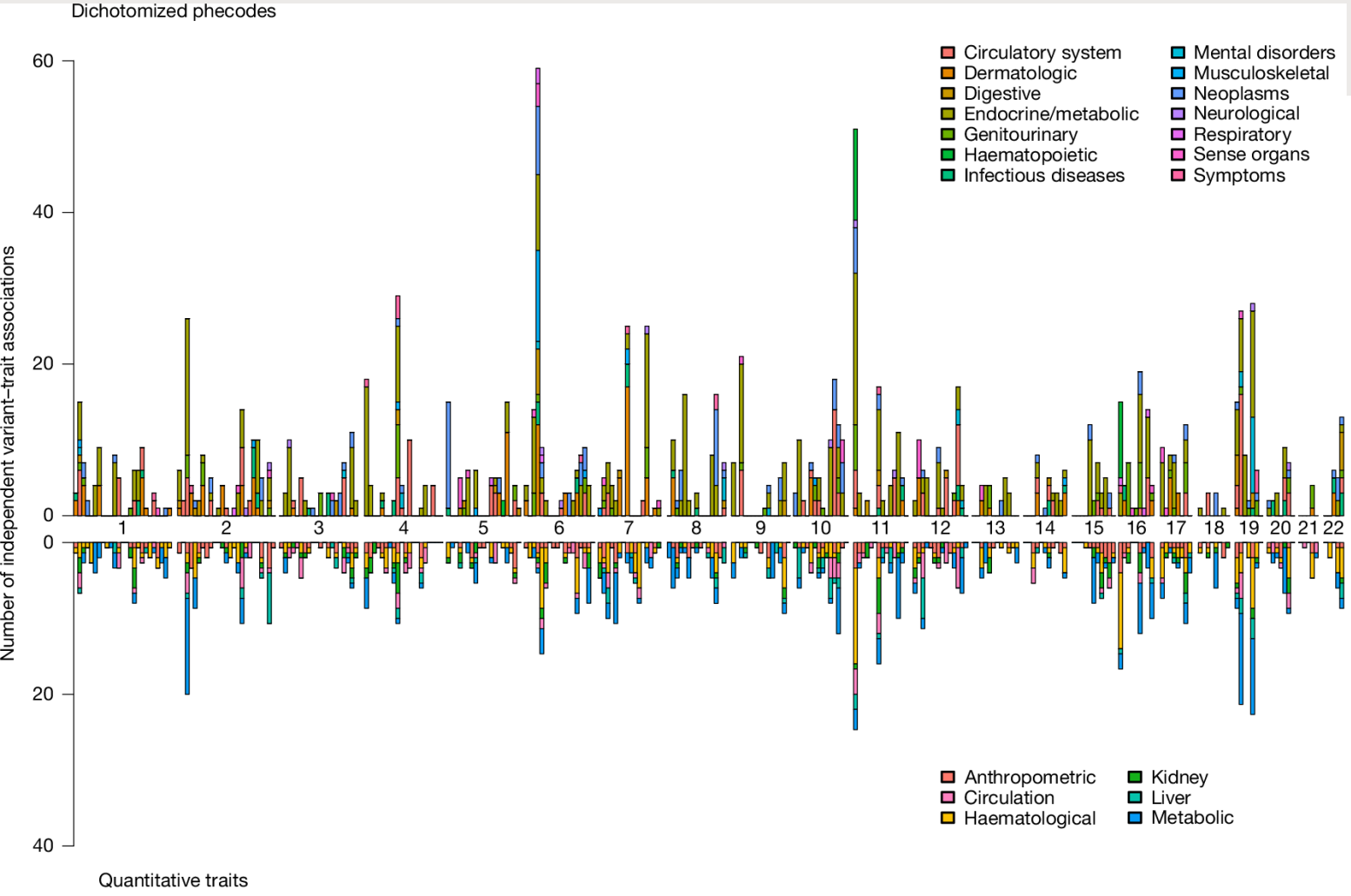


TPMI 病例數比例

健保資料庫五年盛行率

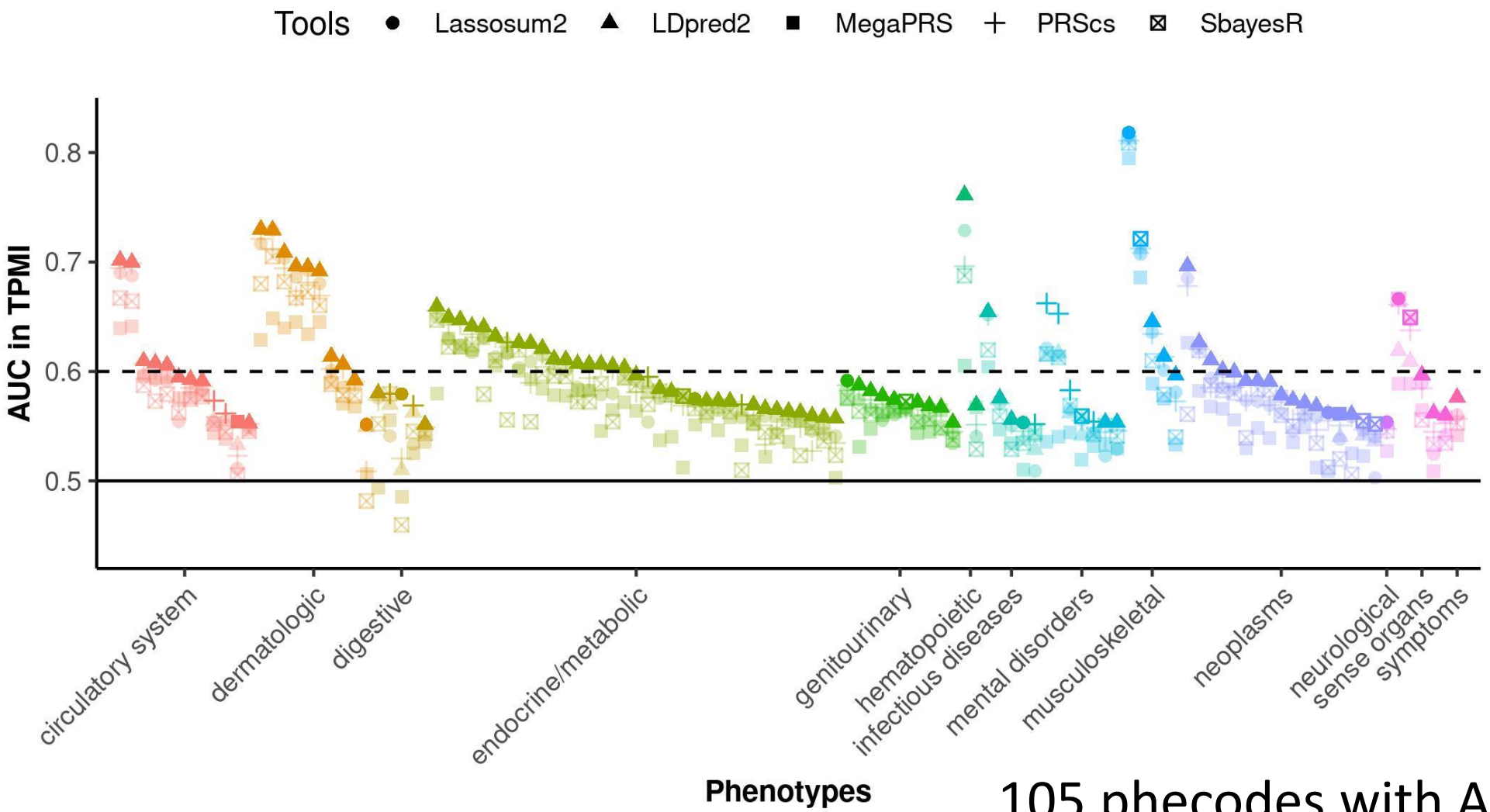
TPMI 樣本數

Phenome-wide Genome-wide association study

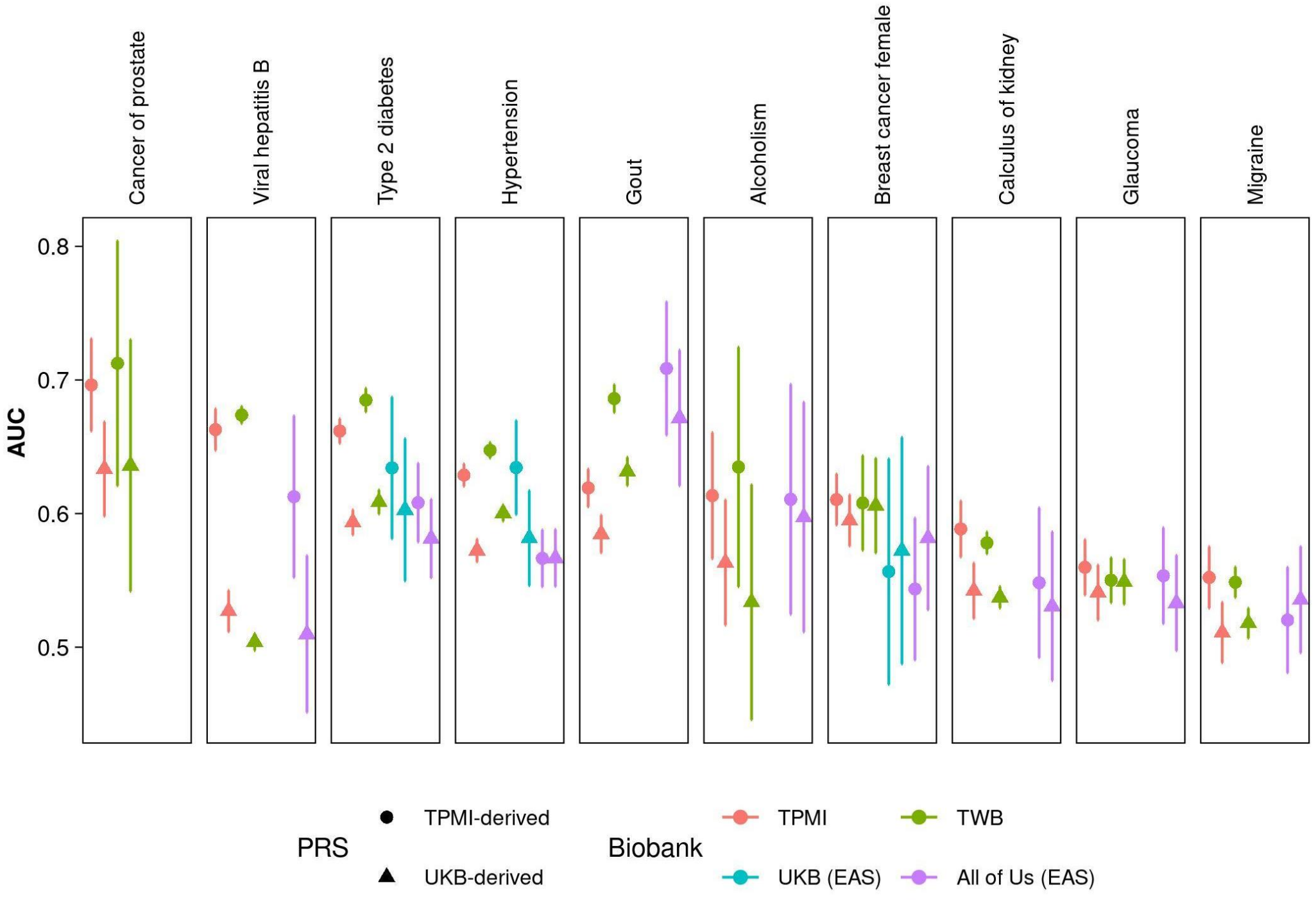


- 2,656 independent signals
  - 1,309 from disease GWAS
  - 1,347 from quantitative traits
- 95 new associations
  - 30 variants are rare in other populations
  - 25 exhibit a significant different effect size with UKB
  - 19 loci are from hepatitis B

PRS development



105 phecodes with AUC > 0.55


Cancer of prostate
Viral hepatitis B
Type 2 diabetes
Hypertension
Gout
Alcoholism
Breast cancer female
Calculus of kidney
Glaucoma
Migraine
AUC
0.8
0.7
0.6
0.5
PRS
TPMI-derived
UKB-derived
Biobank
TPMI
TWB
UKB (EAS)
All of Us (EAS)

Overall health

Table1. Proportion of disease burden explained by genetic risk

		top 5 % vs. bottom 5 %			
		Raw model		Adjusted model[1]	
Index	Genetic risk	R^2	p-value	R^2	p-value
Clinical visit	Cardiometabolic traits	0.93 %	2.30×10^{-4}	1.32 %	2.00×10^{-2}
	Autoimmune and infectious diseases	1.16 %	2.09×10^{-4}	1.11 %	4.96×10^{-2}
	Kidney-related traits	0.65%	3.48×10^{-3}	0.65 %	1.17×10^{-1}
	All predictable traits (131 traits)	7.26 %	1.79×10^{-26}	**8.47 %**	**2.69×10^{-14}**
Hospitalization	Cardiometabolic traits	2.64 %	4.83×10^{-9}	3.55 %	7.10×10^{-9}
	Autoimmune and infectious diseases	1.29 %	1.23×10^{-4}	1.68 %	6.38×10^{-4}
	Kidney-related traits	0.49 %	5.06×10^{-3}	0.64 %	2.21×10^{-2}
	All predictable traits (131 traits)	7.75 %	3.59×10^{-28}	**10.29 %**	**5.62×10^{-27}**

[1]Model adjusting for sex, age and enrollment hospital

- The count of clinical visits and duration of hospitalization were used to roughly describe individuals' overall health

Thank you for your attention