PRIMED Consortium

Polygenic Risk Methods Development

PRIMED at ASHG

Motivation

Due to the historical oversampling of populations with European ancestries in genetic research, polygenic risk scores (PRS) currently perform less well in other, understudied populations, leading to concerns that clinical use in their current forms could widen health care disparities.

The PRIMED Consortium is developing and evaluating methods to improve PRS performance and overall risk prediction in diverse ancestry populations.

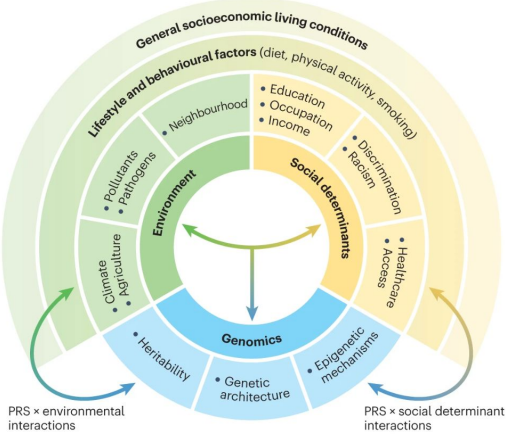

Figure 3a from Kachuri et al. (2023), showing the complex interrelationship among different genomic, environmental, and social risk factors. PMID:37620596

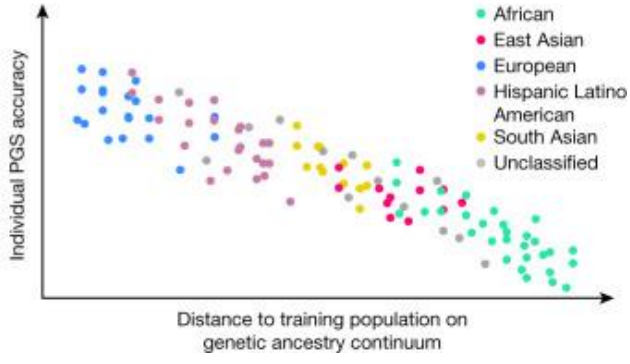

Figure 1d from Ding et al. (2023), illustrating how individual PRS accuracy decreases with increased genetic distance from the training population. PMID:37198491

Connect with PRIMED online

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PRIMED Overview

Consortium Goals

- Gather diverse datasets
- Develop new PRS methods
- Foster global collaboration

Study Sites and Centers

- CAPE
- CARDINAL
- D-PRISM
- FFAIRR-PRS
- Coordinating Center
- PREVENT
- EPIC-PRS
- PRIMED-Cancer

Committees

- Executive
- Steering
- Publications
- Agenda Planning

Working Groups

- Data Analysis
- Data Sharing
- Methods
- Population Descriptors, Social and Ethical Implications (POPSEI)
- Social Determinants of Health (SDoH)

Funders and Advisors

- NIH NHGRI & NCI
- External Scientific Panel

Partners and Affiliates

- Affiliate Members
- AnVIL Analysis & Data Infrastructure
- Partner Programs & Consortia

Consortium Products

- Population descriptors recommendations
- PRIMED common data model
- Analysis workflows
- Methods innovation

PRIMED consortium

NHGRI & NCI funding

U01HG011697
U01HG011717
U01CA261339
U01HG011719
U01HG011720
U01HG011723
U01HG011715
U01HG011710

Research Highlights

Perspectives on the field of PRS methods and development

PRS methods and software to improve prediction across diverse and admixed populations

PRS models for key traits to reduce health inequities

PRS models to improve biomarker accuracy

Environmental and social determinants to refine risk estimation

Innovative approaches to representing and using diversity in PRS

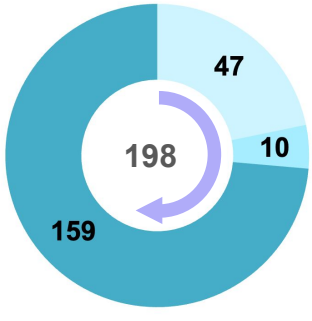
Social and ethical considerations to responsibly develop and implement PRS

Cloud-based approach to collaboratively share and analyze data on the AnVIL platform

Research Highlights with select publications

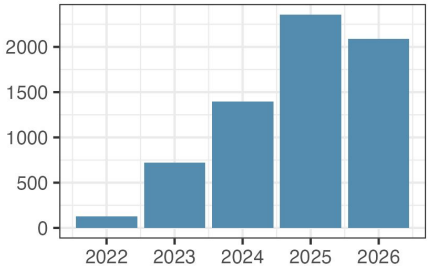
Publications

- Proposals in Progress
- Pre-prints
- Publications



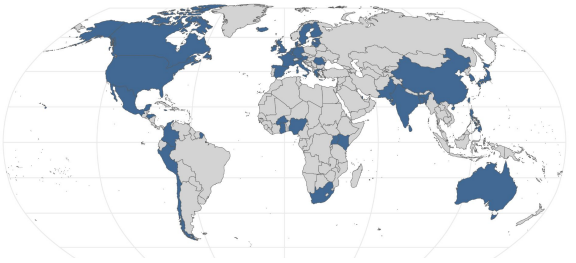
PRIMED Publications in PGS Catalog

Citations by year cited
(6,700 total)
Median RCR: 2.14



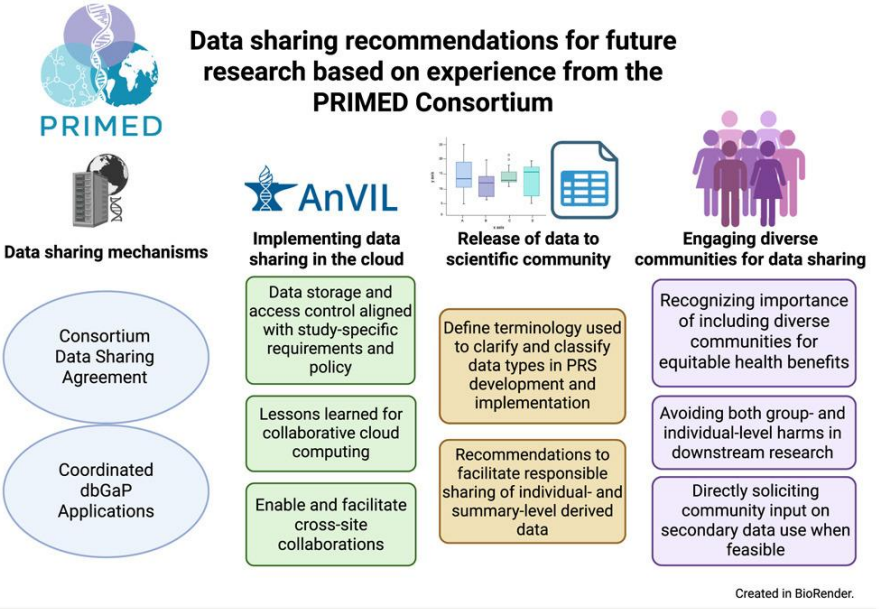
Icons from Flaticon

Data Overview



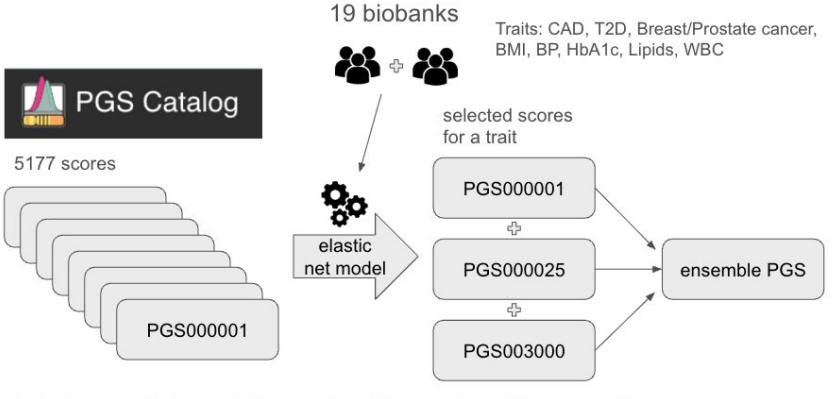
Over **40 countries** are represented among study participants whose data are used by the PRIMED Consortium to improve PRS development and use in diverse genetic ancestry populations.

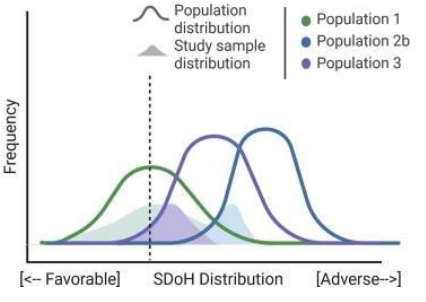
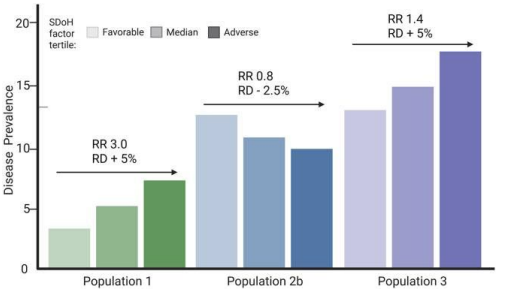
PRIMED developed multiple **data sharing mechanisms** to accommodate different data sources, types, and use restrictions. Data is shared and analyzed on the **NHGRI AnVIL cloud platform**. PRIMED has outlined **policy recommendations** to facilitate responsible sharing and future re-use of data products to improve polygenic risk prediction. (Smith et al. 2025; PMID 40628271)



Methods

PRIMED researchers are **developing and evaluating methods** that improve PRS performance across diverse ancestry populations and social contexts (two examples below). Analysis tools and workflows are made available in the **PRIMED Dockstore Organization**.



The PRIMED Legacy Project is developing new PRS to improve predictions of complex traits by combining existing PRS from the PGS Catalog to leverage cross-trait prediction.

Social determinants of health (SDoH) impact PRS performance. Incorporating SDoH frameworks of relationships between risk and disease is critical for PRS accuracy (Cromer 2026, PMID: 41785862).