

# Polygenic scores in ELSA

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# Introduction

- Polygenic scores (PGS) is a single value estimate of an individual's propensity to a given trait or health outcomes
- They are specific to each individual
- Many health outcomes and behaviours are polygenic in nature
- PGS have also been instrumental in highlighting that many diseases have overlapping, though to varying degree, molecular underpinnings

## Polygenic scores in ELSA

We have prepared polygenic scores for 55 human conditions, behaviours and traits. These can be downloaded either from the ELSA website or on request from Olesya Ajnakina (o.ajnakina@ucl.ac.uk)

Phenotype		
<b><i>Psychosocial</i></b>	<b><i>Childhood psychopathology</i></b>	Longevity
Educational Attainment-2	Aggressive behaviour in childhood	Sleep Duration
Educational Attainment-3	Aggression	Chronic pain
Social Deprivation	Pre-school internalising	Migraine
	Childhood trauma	Gait speed
<b><i>Personality types</i></b>	<b><i>Behavioural traits</i></b>	<b><i>Anthropomorphic traits</i></b>
Neuroticism	Smoking Initiation (ever/never) (2010)	Body Mass Index (2015)
Extraversion	Number of cigarettes per day (2010)	Body Mass Index (2018)
Agreeableness	Age of smoking initiation (2019)	Height
Openness	Smoking Cessation (2019)	Waist circumference
Conscientiousness	Smoking initiation (2019)	Waist-to-hip ratio
	Number of cigarettes per day (2019)	Hand grip
<b><i>Adult psychopathology</i></b>	Daily Alcohol Intake (2016)	<b><i>Biological outcomes</i></b>
Alzheimer's disease (2013)	Drinking alcohol per week (2019)	Plasma cortisol (morning)
Alzheimer's disease (2019)		C-reactive protein
Depressive symptoms	<b><i>Physical health &amp; Longevity</i></b>	
Major Depressive Disorder (2018)	Coronary Artery Disease	<b><i>Reproductive behaviour</i></b>
Anxiety Disorders (factor score)	Type II Diabetes	Age at Menarche
Anxiety Disorders (case-control)	General cognitive function (2015)	Age at Menopause
Insomnia Complaints	General cognitive function (2018)	Age at first birth – Female
Schizophrenia (2014)	IQ	Age at first birth – Male
Subjective Well-Being	Rheumatoid arthritis	Number of children – Female
Loneliness	Myocardial infarction	Number of children – Male
ASD		
ADHD		

## How PGSs were calculated

- These PGSs were built following methods as outlined by Health and Retirement Study (Ware et al. BiorXiv 2017: 1-13)
- PGSs are based on the directly genotyped data rather than imputed data
- PGSs for each phenotype are based on a single genome-wide association study (GWAS). These scores will be updated as sufficiently large GWAS are published for new phenotypes or as updated meta-analyses for existing phenotypes are released.
- If ELSA was included in any of GWASs, we request that consortia to repeat the analysis with ELSA removed.
- If specific polygenic score is not available in the list, we can calculate them upon request

## What else is available in ELSA?

- Several genetic data products derived from the ELSA sample are available. Some data are distributed through the European Genome-Phenome Archive and others are available through the ELSA website.
- The genome-wide genotypes (total 2,230,767) performed on 7,412 ELSA participants of European ancestry genotyped using the Illumina HumanOmni2.5 BeadChips (HumanOmni2.5-4v1, HumanOmni2.5-8v1.3)
- Quality controlled genotyped data for 1,372,240 (61.5% of 2,230,767) variants and 7,183 (96.9% of 7,412 original cohort)
- Imputed data using the Haplotype Reference Consortium (HRC.r1-1.GRCh37) as a reference panel - 7.2M SNPs
- Imputed data using the 1000Genomes Project as a reference panel – ~ 4.5M SNPs

## Other genetic work that we do:

Create polygenic scores using different tools in addition to PRSice, such as LDpred and PRS-CS

Run genome-wide association analyses and prepare summary statistics as part of consortiums using SNPTTEST, SAIGE

Estimate SNP heritability and partitioned heritability (LDSC regression)

Gene mapping and functional annotation

- If you have any questions, you would like to learn more about the genetic data in ELSA, please do not hesitate to contact me ([o.ajnakina@ucl.ac.uk](mailto:o.ajnakina@ucl.ac.uk)).
- Thank you for your attention and I am happy to take your questions.

The screenshot shows the ELSA website interface. At the top, there is a dark blue navigation bar with the ELSA logo and the text "English Longitudinal Study of Ageing". To the right of the logo are navigation links: "DATA & DOCUMENTATION", "COVID-19", "RESEARCH", "RESOURCES", and "More". A search bar is located at the top right of the navigation bar. Below the navigation bar, there is a section of text explaining that PGSs are publicly available and based on a single, replicated genome-wide association study (GWAS). It also mentions that scores will be updated as new GWAS are published. There are two links: "Download a detailed report describing the methods employed." and "Download a detailed list of the PGS available." Below this is a section titled "Genetic imputation" with a dropdown arrow. Underneath is a mailing list sign-up form with the text "Join our mailing list to be alerted with new data is added to UKDS". The form includes input fields for "Name" and "Email address", a "Subscribe Now" button, and a checkbox for "I consent to receiving email updates from ELSA about new data added to UKDS".

ELSA English Longitudinal Study of Ageing

DATA & DOCUMENTATION COVID-19 RESEARCH RESOURCES More

By making these PGSs publicly available, it is hoped that they will facilitate wide use among the ELSA data users. PGSs for each phenotype are based on a single, replicated genome-wide association study (GWAS).

These scores will be updated as sufficiently large GWAS are published for new phenotypes or as updated meta-analyses for existing phenotypes are released.

[Download a detailed report describing the methods employed.](#)

[Download a detailed list of the PGS available.](#)

**Genetic imputation** ▾

Join our mailing list to be alerted with new data is added to UKDS

Name  Email address

I consent to receiving email updates from ELSA about new data added to UKDS