# Socio-demographic and genetic risk factors for drug adherence and persistence across 5 common medication classes

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### **Supplementary Methods**

### FinnGen

### Genotyping and quality control

FinnGen consists of prospectively recruited samples and a series of legacy cohorts with genotypes already available. Prospective samples were genotyped using the ThermoFisher Axiom custom array which tags a total of 655,973 variants. Genotype calling was performed using the Array Power Tools software. Legacy cohorts were genotyped using various Illumina arrays and genotype calling was performed using either GenCall or zCall algorithms. For both prospective and legacy cohorts the following quality control metrics were used.

Samples were removed if:

- Pihat was > 0.9 and the samples were not monozygotic or replicates
- There was a discrepancy between reported sex and genetically determined sex (F-value ≤ 0.3 for females and ≥ 0.8 for males)
- Missingness was  $\geq 5\%$
- Heterozygosity was ±4 standard deviations from the population average
- Pihat was > 0.1 with 14 or more samples
- Samples were ±4 standard deviations away from the population average according to the first two genetic principal components.
- Samples were tagged should there be evidence of a mendelian error or contain replicate samples with over 50,000 discrepancies.

Variants were removed if:

- The variant failed the Hardy-Weinberg Equilibrium test (p-value < 10-6)
- The variant had a call rate < 98%

#### Imputation

Pre-phasing was performed using Eagle 2.3.5 and samples were imputed using the SiSu v3 imputation reference panel. This reference panel is specific to the Finnish population, containing high-coverage (25-30x) whole-genome sequencing data from 3,775 Finns and 16,962,023 variants with minor allele count  $\geq$  3. After imputation, 16,387,711 variants were imputed with high quality (INFO > 0.6).

### Ancestry assignment

Firstly, the FinnGen samples were combined with the 1000 genomes phase 3 dataset. Genetic principal components were calculated using a subset of 49,451 pruned SNPs.

Aberrant was used to identify and remove samples that deviated from the main cluster. A probability of belonging to either a North-Western European or Finnish population was calculated by firstly performing PCA with individuals belonging to these ancestries from 1000 genomes data. FinnGen samples were then projected onto this PCA space and Mahalanobis distances calculated for each sample against each of the two ancestries. Samples were retained if there was  $\geq$  95% probability of belonging to the Finnish ancestry cluster.

### Estonian Biobank

### Genotyping and quality control

Genotyping of DNA samples from the Estonian Biobank was done at the Core Genotyping Lab of the Institute of Genomics, University of Tartu using the Illumina Global Screening Arrays (GSAv1.0, GSAv2.0, and GSAv2.0\_EST). Altogether 206,448 samples were genotyped and then PLINK format files were created using Illumina GenomeStudio v2.0.4. During the quality control all individuals with call-rate < 95% or mismatching sex that was defined based on the heterozygosity of X chromosome and sex in the phenotype data, were excluded from the analysis. Variants were filtered by call-rate < 95% and HWE p-value < 1e-4 (autosomal variants only). Variant positions were updated to Genome Reference Consortium Human Build 37 and all variants were changed to be from TOP strand using reference information provided by Dr. Will Rayner from the University of Oxford (https://www.well.ox.ac.uk/~wrayner/strand/). After QC the dataset contained 202,910 samples for imputation.

### Imputation

Before imputation variants with MAF<1% and Indels were removed. Prephasing was done using the Eagle v2.3 software <sup>1</sup> (number of conditioning haplotypes Eagle2 uses when phasing each sample was set to: --Kpbwt=20000) and imputation was carried out using Beagle v.18May20.d20 <sup>2,3</sup> with an effective population size ne=20,000. As a reference, Estonian population specific imputation reference of 2297 WGS samples was used <sup>4</sup>.

### Ancestry assignment

Further, EstBB samples were combined with the 1000 genomes phase 3 dataset for ancestry analysis. Genetic principal components were calculated using a subset of quality controlled and pruned genotyped SNPs. This was further used to identify and remove samples that deviated from the main cluster.

# Characterization of variants genome-wide significantly associated with adherence and persistence

Supplementary Data 13 reports the 4 variants associated with either adherence or persistence at  $P < 5 \times 10^{-8}$ . We further characterized of these variants based on evidences reported in Open Target Genetics <sup>5</sup>.

- *rs1339882991,* positively associated with both adherence and persistence to BP medications, is an intronic variant located in proximity of the *WNT2B* gene, showing a V2G assignment to the same gene based on evidence from brain tissue eQTLs. This variant was previously reported to be associated with increased risk of hypertension<sup>6</sup> and higher blood pressure<sup>7</sup>.
- *rs111349244,* associated with lower odds of persistence to BP medications, is an intronic variant located near the *LINC02227* gene and was associated with a lower number of antihypertensive medication purchases and decreased risk of hypertension in FinnGen.
- rs12149025, associated with lower persistence to DOAC, is an upstream gene variant located in proximity of the CBFA2T3 gene, with V2G assignment to CDH15 based on PCHi-C<sup>8</sup> evidences and evidences from muscle tissue eQTLs.
- *rs548379361,* associated with lower adherence to breast cancer medications, is an intronic variant near the *CFAP44* gene.

### Effect of polytherapy on adherence and persistence

We assessed the effect of polytherapy on drug adherence and persistence with respect to the five medications defined in the primary analysis. For each medication, we determined if any of the other four treatments were concurrent in the following manner: for adherence, we considered a medication regimen concurrent if the time between the first and last purchase recorded was overlapping at any time with the timespan used for adherence calculation; for persistence if the time between first and last purchase recorded of the potential concurrent treatment contained the purchase date used for persistence calculation. We fitted a linear model for persistence and adherence with a categorical variable with three levels (0 for no concurrent treatment, 1 for one concurrent treatment, and 2 for more than one concurrent treatment), adjusting for the baseline covariates used in the primary analysis (**Health and socio-demographic risk factors for persistence and adherence**). The percentages of change in adherence for polytherapy are reported and OR for persistence are reported in Supplementary Data 7,8.

We observed that the presence of at least once concurrent treatment was consistently associated with both increased adherence and higher odds of persistence. The percentage increase in adherence with one concurrent treatment ranged from 0.6% (blood pressure medications) to 3.6% (antiplatelets). The ORs of being persistent between one concurrent treatment and no concurrent treatments go from 1.04 (not statistically significant for breast cancer medications) to 2.79 (anticoagulants). Moreover, we find consistently larger effect sizes for two or more concurrent treatments compared with only one concurrent treatment, except for blood pressure medications.

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### **Supplementary Figure 1**



**Supplementary Figure 1 | Adherence as function of expected and observed consumption.** Hexagonal heatmap representing mean adherence levels at the varying of expected (number of days, x-axis) versus observed consumption (number of tablets, y-axis). Bins with less than 5 individuals are not displayed. The level lines represent the 2-dimensional density, each line including 10% of the individuals in the data.



**Supplementary Figure 2 | Genome-wide association results for persistence and adherence. a.** results of genome-wide association study of persistence to each medication. Number of cases and controls are reported next to each drug name. Red dotted line represents genome-wide significance at P-value = 1x10-8 (Bonferroni corrected for 5 medications). b. results of genome-wide association study of adherence to each medication. Total sample size is reported next to each drug name. Red dotted line represents genome-wide significance at P-value = 1x10-8 (Bonferroni corrected for 5 medications). B. Results for 5 variants with significance < 5x10-8 are reported in Supplementary Data 12.



**Supplementary Figure 3 | Associations between persistence and PGS for 33 clinically relevant traits.** Odds of persistence per 1-SD increase in trait PGS. ORs are from a logistic regression model adjusted for sex, age at initiation, first 10 genetic principal components. Error bars represent the 95% confidence interval for the estimates. Results are reported in Supplementary Data 16.







Supplementary Figure 5 | Associations between persistence (as defined for the sensitivity analysis) and PGS for 33 clinically relevant traits. Odds of persistence per 1-SD increase in trait PGS. ORs are from a logistic regression model adjusted for sex, age at initiation, first 10 genetic principal components. Error bars represent the 95% confidence interval for the estimates. Results are reported in Supplementary Data 21.

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