Data Supplement for Sullivan et al., Psychiatric Genomics: An Update and an Agenda. Am J Psychiatry (doi: 10.1176/appi.ajp.2017.17030283)

Supplement

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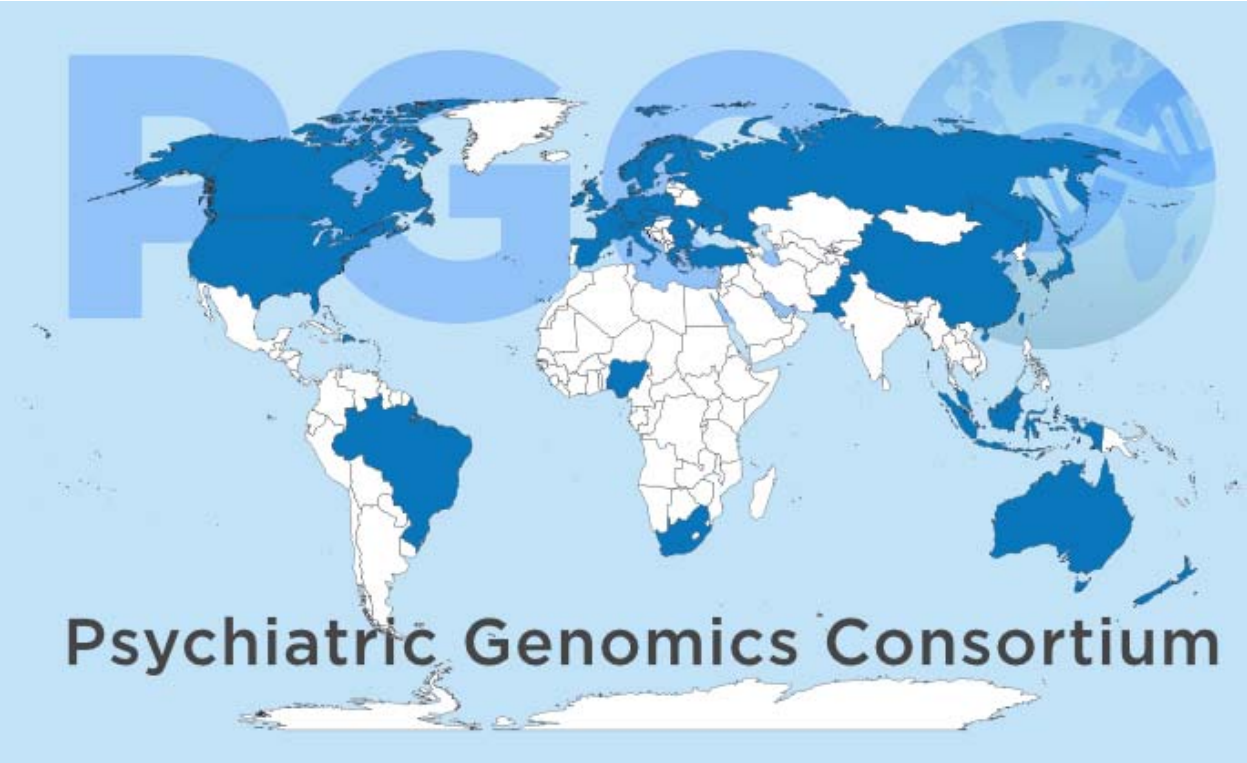
Figure S1. PGC world map



World map showing the numbers of cases contributed per country. There is an under-representation of subjects from countries in East Asia, central Asia, Africa, and south and central America. Countries with a historical preponderance of European-ancestry population are over-represented.

Table S1. Definition of terms

Term	Definition
Single-nucleotide polymorphism	SNP. Specific position where chromosomes carry different nucleic acids. Usually have two variants, which makes them more amenable to genotyping. Polymorphism means frequency is > 1% in a population. There are >10 million SNPs. As they are common, relatively easy to genotype, and widespread, these SNPs are targeted by GWAS arrays.
Single-nucleotide variant	SNV. Essentially the same definition as SNP but can have any frequency. In practice, can be common (>0.01), uncommon (0.001-0.01), rare (<0.001), or ultra-rare (i.e., reported just once in any study).
Twin or pedigree heritability	Twin-h^2, pedigree-h^2. Proportion of the variance in liability of a phenotype (disease, trait) that is due to genes, estimated from risks to twins or to other relatives.
SNP-heritability	SNP-h^2. Proportion of the variance in liability of a phenotype (disease, trait) that is due to directly assessed genetic variation, estimated from genome-wide SNP data.
Mendelian disease	Caused by a (usually rare) mutation in DNA sequence on one (dominant) or both (recessive) of an individual's pair of chromosomes.
Complex disease	Caused by an interaction of multiple genetic and/or environmental factors, does not exhibit classical Mendelian inheritance patterns.
Copy number variant	CNV. Chromosomal segment where DNA has been deleted or duplicated. Other structural variants include inversions and translocations.
Common-disease common-variant hypothesis	Genetic risk for common diseases is due to common SNPs.
Multiple rare variant hypothesis	Genetic risk for common disease is due to many different rare SNPs, especially in protein coding or gene regulatory regions.
Linkage disequilibrium	LD. Correlation between two SNPs that are close together (an allele of one SNP is usually inherited with a specific allele from a second SNP). LD makes GWAS possible: a carefully selected subset of SNPs contains information about many others.
Genome-wide association study	GWAS. A systematic search for common SNPs that influence a disease or trait, using a genome-wide SNP array for typing a cohort of individuals. Current arrays also provide information about large CNVs.
Genome-wide SNP array	A system for assaying hundreds of thousands or millions of SNPs for an individual subject. Can now be done inexpensively (<$US 50) and at high throughput (5,000 subjects per week is readily attainable).

Updated from Table 1 in reference (1).

Table S2. PGC papers

Main papers, primary reports and reviews (N=25)

1. Agrawal A, Edenberg HJ, Gelernter J. Meta-analyses of genome-wide association data hold new promise for addiction genetics. J Stud Alcohol Drugs. 2016;77:676-680.

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4. CNV Working Group of the Psychiatric Genomics Consortium, Schizophrenia Working Groups of the Psychiatric Genomics Consortium. Contribution of copy number variants to schizophrenia from a genome-wide study of 41,321 subjects. Nat Genet. 2016;49:27-35.

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Table S3. Case and control descriptions

Group	N_{case}	Brief description
ADHD (2)	39K	**Diagnosis**: DSM-IV/ICD-10 ADHD, structured interviews, national treatment/pharmacy registers, or validated web instruments (new samples: iPSYCH). Includes PGC study quality review. **Ascertainment:** clinical or national register. **Phenotypes**: severity, symptoms, age onset, comorbidity.
AN (3)	50K	**Diagnosis**: DSM-IV AN (excluding amenorrhea), structured interviews (new samples: ANGI & Charlotte's Helix). Includes PGC study quality review. **Ascertainment:** clinical or national register. **Phenotypes**: severity, symptoms, binging, purging, lowest/highest BMI, age onset, comorbidity.
AUT (4)	40K	**Diagnosis:** DSM-IV ASD, structured interviews (ADI-R and/or ADOS), national register samples (clinic-based, validated with ADI-R/ADOS). Includes PGC study quality review. **Ascertainment:** clinical or national register. **Phenotypes:** symptoms, severity, ADI-R/ADOS elements, IQ/comorbid ID.
BIP (5)	49K	**Diagnosis**: DSM-IV BIP1 or BIP2, structured interviews or validated national treatment or quality assurance registers. Includes PGC study quality review. **Ascertainment:** clinical or national register. **Phenotypes**: severity, symptoms, age onset, cognition, brain MRI.
MDD (6)	100K	**Diagnosis**: DSM-IV MDD, structured interviews, national treatment/pharmacy registers, or validated web instruments (new samples: iPSYCH & UK Biobank, Q1 2016). Includes PGC study quality review. **Ascertainment:** clinical or national register. **Phenotypes**: severity, symptoms, age onset, recurrence, comorbidity, sex abuse.
OCD/TS (7)	34K	**Diagnosis**: DSM-IV OCD or TS, structured interviews, national treatment registers, or validated web instruments (new samples: iPSYCH & funded collections. Includes PGC study quality review. **Ascertainment:** clinical or national register. **Phenotypes**: severity, symptoms, age onset, comorbidity.
PTSD (8)	75K	**Diagnosis**: DSM-IV PTSD, structured interviews, national treatment/pharmacy registers, or validated web instruments (new samples: DK Soldier). Includes PGC study quality review. **Ascertainment:** clinical, Army STARRS, or national register. **Controls** are trauma-exposed & separate from other PGC groups. **Phenotypes**: trauma details, severity, symptoms, age onset, comorbidity, trauma history.
SCZ (9)	100K	**Diagnosis**: DSM-IV SCZ or SAD, structured interviews or validated national treatment/pharmacy registers (new samples: CLOZUK-v3, China, EUGEI). Includes PGC study quality review. **Ascertainment:** clinical or national register. **Phenotypes**: pos/neg symptoms, age onset, cognition, environmental exposures, brain MRI.
SUD	60K	**Diagnosis**: DSM-IV cannabis, opioid, cocaine, nicotine and alcohol use disorders (new samples: "SmokeScreen", UK Biobank, other PGC). Includes PGC study quality review. **Ascertainment:** clinical or national register. **Phenotypes**: severity, quantity/frequency, symptoms, age onset, recurrence, craving, IV, poly-drug.
Controls	†	**Diagnosis**: For common disorders (K_P>0.01), screened for absence of case definition, and for some studies, exposure. For uncommon/rare disorders, most screened & a few unscreened; although the power implications are small (10) ,we test this carefully. Includes PGC study quality review. **Ascertainment:** most case collections have controls from population or national registers. **Phenotypes**: variable, but many have extensive psychiatric phenotyping.

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