Background and aim

Personality traits are basic dimensions of behavioural variation, and twin, family, and adoption studies show that ~30% of the variation in personality traits is due to genetic influences. But how is genetic variation maintained in personality where there is selection for only the most advantageous genotypic trait values?

There are three main evolutionary mechanisms that could explain how genetic variation is maintained in traits:
- Selective neutrality
- Mutation-selection balance
- Balancing selection

Each of these mechanisms makes predictions (See Table) in terms of 1) The relative contribution of rare and common genetic variants to personality variation 2) The relative magnitude of non-additive genetic influences 3) Whether personality is affected by inbreeding

From genome-wide association and linkage studies, we already know that personality traits must be highly polygenic. Using genome-wide SNP data we tested further competing predictions of selective neutrality, mutation-selection balance, and balancing selection: A) The extent to which all common genetic variants contribute to variation in personality B) Whether or not inbreeding affects personality traits

We used genotypic and phenotypic data from four samples from Australia and Finland (total N>8,000) who were assessed on Cloninger’s personality traits Harm Avoidance, Novelty Seeking, Reward Dependence, and Persistence.

A) SNP-based heritability estimate

Method
To test A above, we used GCTA (Yang et al., 2010) to estimate the proportion of variation in personality traits that can be accounted for by ~270,000 SNPs taken together. This is achieved by determining to what extent genetic similarity (at the SNPs) between all individuals corresponds to their phenotypic similarity. This method captures the vast majority of the combined effect of common variants.

Results
Little variation in the Cloninger personality dimensions (7.2% on average) is due to the combined effect of common, additive genetic variants across the genome.

B) Inbreeding

Method
To test B above we examined the association between personality traits and the level of inbreeding in the ancestry of each individual. Using PLINK (Purcell et al 2007), we quantified individuals’ level of inbreeding by estimating the proportion of their genome that is in runs-of-homozyogosity (ROH; i.e. homologous stretches of DNA that can be observed in the offspring of even distant relatives), by summing the total length of all their autosomal ROHs divided by the total SNP-mappable autosomal genome length.

To test whether inbreeding influenced the personality traits we tested if the level of ROHs was correlated with the personality scale scores.

Results
Higher levels of inbreeding were significantly associated with less socially-desirable personality trait levels in three of the four personality dimensions.

Implications of these findings

A) The SNP-based heritability estimate shows that common additive genetic variants account for a small percentage (~20%) of the total genetic variation in all four personality traits. This is consistent with mutation-selection balance but not with selective neutrality or balancing selection models for highly polygenic traits.

The rest of the genetic variation is likely to comprise of rare variant effects and/or some combination of dominance and epistasis.

B) The finding that inbreeding affects personality traits in apparently deleterious directions is consistent with those traits being influenced by a load of mutations that tend to be rare, recessive, and deleterious, as predicted under mutation-selection balance.

These inbreeding effects are not consistent with selective neutrality or balancing selection models for highly polygenic traits, as these provide no reason to expect bias in the direction of dominance across many loci.

A and B are consistent with the conclusion that:

Genetic variation in personality traits has been maintained primarily by mutation-selection balance.

This study is published:

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