H

Heritability

Kate E. Lynch
Department of Philosophy, University of Sydney,
Sydney, Australia

Synonyms

Genetic inheritance; Inheritance

Definition

Heritability is the statistical metric that estimates the relative influence of genetic variation (measured as genetic variance) on phenotypic variation (phenotypic variance), relative to the influence of environmental differences (environmental variance).

Introduction

Heritability is a statistical parameter that is often used in discussions of genetic causation and the nature versus nurture debate. The heritability of a trait, represented by either H^2 (for broad heritability) or h^2 (for narrow heritability) is between 0 and 1. Traits with a high heritability are generally seen as genetically influenced, and those with a low heritability as environmentally caused (Lynch and Bourrat 2017).

The heritability concept most often invoked in psychological science is broad sense heritability. Broad sense heritability estimates the proportion of phenotypic variance (V_P) that can be accounted for by genetic variance (V_G), relative to the influence of environmental variance (V_E):

$$H^2 = \frac{V_G}{V_P} \qquad (1)$$

$$V_P = V_G + V_E \qquad (2)$$

Narrow sense heritability (h^2) is an alternative heritability measure which is primarily used in breeding experiments by quantitative geneticists using animal models. It is concerned only with additive genetic effects (V_A), as these are reliably transmitted between generations. This is opposed to other kinds of genetic effects like dominance and epistasis (V_{D+I}), which encompass gene–gene interactions.

$$h^2 = \frac{V_A}{V_P} \qquad (3)$$

$$V_P = V_A + V_{D+I} + V_E \qquad (4)$$

Knowledge of h^2 allows one to forecast the adaptive potential of a given trait within a population, using the breeder's equation (Lush 1937). Usually these kinds of estimates are confined to traits in agricultural animals such as body weight,

milk yield, life span, fecundity, and litter size (Falconer and McKay 1996).

Human research is less concerned about the adaptive potential of traits, and more interested in the sources of trait variation. Because of this, broad sense heritability is used, as it encompasses the total genetic contribution to the trait of interest. In order to calculate heritability, phenotypic data is collected from individuals who share a common family environment, and are genetically related to varying degrees. As environments are assumed not to vary between individuals in a family environment, the phenotypic similarities observed between parents and their children, or between siblings (especially twins), are assessed in light of their degree of genetic relatedness. See ▶ "Calculating Heritability" and ▶ "Twin Studies" for details.

Populations and Individuals

Heritability is a population-level parameter, and cannot be used to make causal inferences about individuals. For example, the heritability of height is ≈0.8 in most human populations. This means that if you lined up a population in accordance to their height, and wanted to know what accounted for differences in stature – you would find that environmental differences between individuals were largely unrelated to height differences, and that genetic differences were associated with differences in height.

A heritability of 0.8 does *not* mean that only 80% of people have their height genetically influenced. It also does not mean that a given individual has 80% of their height genetically influenced, such that an individual who is 165 cm tall can attribute 132 cm to their genes.

At an individual level, both genes and environment are causally necessary for the production of phenotypes, and assigning values of relative causal importance to each is incoherent. Keller (2010) illustrates this with an analogy. Imagine two individuals, Billy and Suzy, who want to fill a bucket of water. Suzy holds a hose to the bucket, while Billy turns on the tap. Once the bucket is full, one could ask: "How much of the water is due to Billy's contributions, and how much to Suzy's?" The example is meant to illustrate that this question does not make sense, as partitioning causes in this case is not possible. Both the hose-holding and the turning of the tap are required to fill the bucket, and it is not possible to assess "how much" each caused the bucket to fill, relative to the other.

The Utility of Heritability Estimates

In addition to the pure explanatory utility of heritability estimates, broad-sense estimates can be used as a tool to investigate the causes of phenotypic variation in detail. For example, they may be used as justification for the search for genetic markers which are the first step of genetic mapping, and can lead to the isolation of candidate genes via methods such as genome-wide association studies.

Broad heritability estimates can also be used to estimate and predict the phenotypes of families and offspring, including complex diseases and behaviors. This information can be utilized for preventative procedures and genetic counselling strategies, as well as to provide extra intervention and support for those who may be genetically susceptible to undesirable phenotypic variants.

The heritability of a trait has also been invoked to discuss broader philosophical issues such as free will and moral responsibility. For instance, Kaebnick (2006) raises the question: If no environmental interventions (that have yet been studied) seem able to alter the V_P of a trait, how responsible are individuals possessing a certain variant? Kaebnick believes that heritability analyzes help to inform, or at least introduce, some of these broader philosophical questions and should be considered by ethicists. Others such as Parens et al. (2006) have similarly theorized that knowledge about the heritability of a trait can impact on individual feelings of blame, responsibility, and human identity.

Heritability of Psychological Traits

One of Eric Turkheimer's (2000) three laws of behavior genetics is that all behavioral traits are heritable. This is also true in the study of personality, which indicates that all personality traits are heritable to some degree (Bratko et al. 2017). Studies of personality using the five-factor model (extraversion, agreeableness, conscientiousness, neuroticism, and openness for experience) have yielded significant heritabilities (Power and Pluess 2015), as well as other models of personality such as Eysenck's PEN (psychoticism, extraversion, and neuroticism), and Tellegen's (positive affect, negative affect, and constraint) (Bratko et al. 2017). Overall, personality differences have a heritability estimate around 0.40, meaning that 40% of the variance is due to genetic differences, and 60% due to environmental influences (Bratko et al. 2017).

Intelligence is another trait of interest to psychologists. It was one of the first traits to be analyzed using heritability and is to date the most studied phenotype in behavior genetics. The heritability of intelligence is estimated to be 0.5–0.8 (Plomin and Spinath 2004). However, these estimates have been questioned due to phenomena such as the Flynn effect (Flynn 1984) which documents the rise in IQ test performance over generational time scales. While the IQ scores of a population reliably trend towards a normal distribution, the median raw scores of later generations are higher than those of earlier ones, resulting in a continual re-adjustment raw scores, averaging a 3 point per decade adjustment (Flynn 1984). Another piece of evidence that indicates a significant environment influence on intelligence is observed IQ gains through adoption into higher socioeconomic (SES) homes, as well as through migration into higher SES countries (Sauce and Matzel 2018).

Mental illnesses and disorders have also gained a lot of attention in heritability studies. For example, schizophrenia has a $H^2 = 0.79$ (Hilker et al. 2018), unipolar depression $H^2 = 0.37$ (Sullivan et al. 2000), bipolar disorder $0.71 \leq H^2 \leq 0.77$ (Edvardsen et al. 2008), and Autism spectrum disorder $0.64 \leq H^2 \leq 0.91$ (Tick

et al. 2016). Some of these results have been criticized on the grounds of nonadditive effects biasing heritability estimates. These are discussed below.

Nonadditivity

Standard models of heritability assume additivity of genetic and environmental variance. This means that phenotypic variance can be calculated as the sum of genetic and environmental variances as two independent variables. However, empirical evidence suggests that genes and environments interact nonadditively for generating trait differences. This can happen in two ways. The first is gene-environment interaction (GxE), which occurs when the effects of genotypes are differentially expressed depending upon their environment. Likewise, environmental influences act differentially depending upon the genotype of an individual (Tabery 2014). For example, there is a genetic interaction with cannabis usage for development of schizophreniform psychosis. Individuals with a certain variant of the *COMT* gene are more likely to develop psychosis when exposed to cannabis, compared to those with a different gene variant (Caspi et al. 2005).

If GxE effects are not detected, then the resulting heritability estimate will be incorrect, as it is using an erroneous model of variance contributions. When they are detected, they can be accounted for by updating the heritability model to include an interaction effect:

$$V_P = V_G + V_E + V_{GxE} + r_{GE} \qquad (5)$$

Even if no GxEs are detected, or if they are detected and adjusted for in a nonadditive model (Eq. 5), they still pose a problem for the extrapolation-worthiness of heritability estimates. GxE occurs when genetic differences make a difference to phenotypes in different ways depending upon the environments experienced by the population under study. This makes it difficult to make general claims about the genetic causation of traits across populations. This problem was first identified by Lewontin (1974), who

argued that GxEs are problematic for heritability estimates even in cases where none have been identified. Although genes and environments may behave additively in one population, resulting in a particular H^2, that does not rule out that there could be interactions in different, unstudied environments in unobserved populations. This problem has led some to reject the utility of heritability estimates all together.

The second form of nonadditivity is gene environment correlation (rGE). This occurs when certain genotypes and certain environments are more likely to be found together, and so at a population level genetic variance and environmental variance become associated. There are three forms of rGE (Plomin et al. 1977). The first is passive rGE, which occurs when parents pass on associated genetic and environmental influences to their children. For instance, children of avid readers might inherit genes which provide good comprehension skills, as well as environments filled with many books. Thus, both genetic and environmental influences on reading ability are correlated within the population. Passive rGEs can be isolated using adoption studies, or by controlling for potential correlated environments in study designs. Active rGEs occur when certain genes predispose an individual to seek out certain developmental environments. In this case, the environmental exposure is an indirect effect of genotype, and so genes and environments are correlated at the population level. For example, bright children with a genetic advantage for academic study may, thanks in part to their genes, seek out more stimulating environments, further advancing their academic performance. A third form of rGE is reactive cases. These occur when individuals experience certain environments because of the way society reacts to some aspect of a genetically influenced genotype. This is thought to occur in problematic society-based reactions to different racial groups. Individuals possess genes which express themselves in physiological traits such as skin color, and because of these traits, those individuals are subject to harassment, or deprived of environmental resources, compared to counterparts with a different genetic background.

Active and reactive rGEs are difficult to detect in the study of heritability, and there is debate as to whether or not active cases should be included as part of the additive effects of V_G (Lynch 2017). Evidence for active and reactive rGEs comes from the heritability of environmental variables, such as socioeconomic status, television viewing, and quality of social support (Plomin and Bergeman 1991). Rutter, Moffitt and Caspi (2006) and Rutter and Silberg (2002) have proposed that rGE contribute significantly to the heritability of psychiatric disorders. For instance, aggressively prone offspring are likely to promote harsher treatment from others, compounding their development along a psychiatric trajectory. This work has been extended by Jaffee and Price (2007) who have identified particular allelic variations and their associations with environments, such as parental rejection. These associations are then predictive of later psychiatric illness, indicating that genetic variation may account rGE with parental engagement. Meek et al. (2013) have suggested that heritability estimates for autism may also be largely accounted for by active and reactive rGEs. Children who begin as mildly autistic are likely to select environments lacking in social stimuli (active rGE), and others around them can react to them in a way which compounds both their autistic phenotype (reactive rGE) and their subsequent environmental selection (further compounding the active rGE pathway).

Conclusion

Heritability is a statistical parameter that estimates the influence of genetic variation on phenotypic variation, relative to environmental variation. Broad heritability (H^2) is the most widely used statistic in the human sciences, and is used to explain the causes of trait differences at a population level. Many psychiatric disorders, personality differences, human abilities, and behaviors have significant heritabilities. These results can be used to understand the causes of trait differences in a population; however, causal claims about relative influences of genes and the environment cannot be made about individuals using heritability

results as heritability is strictly a population-level parameter. The population relativity of H^2 has led to some criticisms of the utility of the estimates. In particular, the existence of gene-environment interactions – both within actual and hypothetical populations – is problematic for extrapolating heritability results. Additionally, gene-environment correlations undermine the additive model used for heritability estimates, and are difficult to study and control for. There is growing evidence that gene-environment correlations account for some of the estimated heritability for psychiatric traits.

Cross-References

▸ Calculating Heritability
▸ Genetic Determinism
▸ Genetic Influences
▸ Problem of Genetic Inheritance, The
▸ Twin Studies

References

Bratko, D., Butković, A., & Vukasović Hlupić, T. (2017). Heritability of personality. *Psychological Topics, 26*(1), 1–24.

Caspi, A., Moffitt, T. E., Cannon, M., McClay, J., Murray, R., Harrington, H., . . ., & Poulton, R. (2005). Moderation of the effect of adolescent-onset cannabis use on adult psychosis by a functional polymorphism in the catechol-O-methyltransferase gene: longitudinal evidence of a gene X environment interaction. *Biological Psychiatry, 57*(10), 1117–1127.

Edvardsen, J., Torgersen, S., Røysamb, E., Lygren, S., Skre, I., Onstad, S., & Øien, P. A. (2008). Heritability of bipolar spectrum disorders. Unity or heterogeneity? *Journal of Affective Disorders, 106*(3), 229–240.

Falconer, D. S., & McKay, R. (1996). *Introduction to quantitative genetics* (3rd ed.). Burnt Mill: Longman.

Flynn, J. R. (1984). The mean IQ of Americans: Massive gains 1932 to 1978. *Psychological Bulletin, 95*(1), 29.

Hilker, R., Helenius, D., Fagerlund, B., Skytthe, A., Christensen, K., Werge, T. M., . . ., & Glenthøj, B. (2018). Heritability of schizophrenia and schizophrenia spectrum based on the nationwide Danish twin register. *Biological Psychiatry, 83*(6), 492–498.

Jaffee, S. R., & Price, T. S. (2007). Gene–environment correlations: A review of the evidence and implications for prevention of mental illness. *Molecular Psychiatry, 12*(5), 432.

Kaebnick, G. E. (2006). Behavioral genetics and moral responsibility. In E. Parens, A. R. Chapman, & N. Press (Eds.), *Wrestling with behavioral genetics: Science, ethics, and public conversation*. Baltimore: The Johns Hopkins University Press.

Keller, E. F. (2010). *The mirage of a space between nature and nurture*. Durham and London: Duke University Press.

Lewontin, R. C. (1974). Annotation: The analysis of variance and the analysis of causes. *American Journal of Human Genetics, 26*(3), 400.

Lush, J. (1937). *Animal breeding plans*. Iowa: Iowa State College Press.

Lynch, K. E. (2017). Heritability and causal reasoning. *Biology and Philosophy, 32*(1), 25–49.

Lynch, K. E., & Bourrat, P. (2017). Interpreting heritability causally. *Philosophy of Science, 84*(1), 14–34.

Meek, S. E., Lemery-Chalfant, K., Jahromi, L. B., & Valiente, C. (2013). A review of gene–environment correlations and their implications for autism: A conceptual model. *Psychological Review, 120*(3), 497.

Parens, E., Chapman, A. R., & Press, N. (Eds.). (2006). *Wrestling with behavioral genetics: Science, ethics, and public conversation*. Baltimore, Maryland: JHU Press.

Plomin, R., & Bergeman, C. S. (1991). The nature of nurture: Genetic influence on "environmental" measures. *Behavioral and Brain Sciences, 14*(3), 373–386.

Plomin, R., & Spinath, F. M. (2004). Intelligence: Genetics, genes, and genomics. *Journal of Personality and Social Psychology, 86*(1), 112.

Plomin, R., DeFries, J. C., & Loehlin, J. C. (1977). Genotype-environment interaction and correlation in the analysis of human behavior. *Psychological Bulletin, 84*(2), 309.

Power, R. A., & Pluess, M. (2015). Heritability estimates of the Big Five personality traits based on common genetic variants. *Translational Psychiatry, 5*(7), e604.

Rutter, M., & Silberg, J. (2002). Gene-environment interplay in relation to emotional and behavioral disturbance. *Annual Review of Psychology, 53*(1), 463–490.

Rutter, M., Moffitt, T. E., & Caspi, A. (2006). Gene–environment interplay and psychopathology: Multiple varieties but real effects. *Journal of Child Psychology and Psychiatry, 47*(3–4), 226–261.

Sauce, B., & Matzel, L. D. (2018). The paradox of intelligence: Heritability and malleability coexist in hidden gene-environment interplay. *Psychological Bulletin, 144*(1), 26.

Sullivan, P. F., Neale, M. C., & Kendler, K. S. (2000). Genetic epidemiology of major depression: Review and meta-analysis. *American Journal of Psychiatry, 157*(10), 1552–1562.

Tabery, J. (2014). *Beyond versus: The struggle to understand the interaction of nature and nurture*. cambridge massachusetts: MIT Press.

Tick, B., Bolton, P., Happé, F., Rutter, M., & Rijsdijk, F. (2016). Heritability of autism spectrum disorders: A meta-analysis of twin studies. *Journal of Child Psychology and Psychiatry, 57*(5), 585–595.

Turkheimer, E. (2000). Three laws of behavior genetics and what they mean. *Current Directions in Psychological Science, 9*(5), 160–164.