

# DISCORDANCE

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## DISCORDANCE

**Primary Disciplinary Field(s):** Behavioral Genetics, Psychology, Statistical Analysis

### 1. Core Definition

The term **discordance** fundamentally refers to a state of variance, disagreement, or lack of concurrence between two or more elements. In its most general application, it describes any situation where an observed condition or trait fails to align with an expected outcome or another corresponding measurement. This definition encompasses a broad range of scientific and philosophical contexts, suggesting a lack of harmony or consistency in data, behavior, or physical presentation. The concept is intrinsically relational, requiring at least two entities--such as observations, individuals, or symptoms--to assess whether they match or diverge. When divergence is observed, the state is defined as discordance, highlighting a significant deviation from uniformity or agreement.

However, the term holds a highly specialized and critical meaning within the fields of **behavioral genetics** and medical research, specifically in the context of twin studies. Here, discordance is quantified as the non-similarity observed between twins regarding a specific trait, disorder, or disease. If one twin exhibits the characteristic (e.g., a mental disorder or a physical ailment) while the other twin does not, they are considered discordant for that trait. This framework is essential for researchers attempting to disentangle the relative contributions of genetic endowment versus environmental influences, particularly when comparing monozygotic (identical) twins who share 100% of their segregating genes, and dizygotic (fraternal) twins who share, on average, 50% of their segregating genes.

The existence of discordance, particularly among monozygotic twins, provides compelling evidence that genetic predisposition alone is insufficient to guarantee the manifestation of a trait or disorder. Instead, it underscores the powerful role played by non-shared environmental factors--those experiences, exposures, or stochastic biological events that differ even between individuals raised in the same household. Analyzing the rate of discordance versus concordance allows researchers to statistically estimate parameters such as heritability (A), shared environment (C), and non-shared environment (E) in complex behavioral and medical phenotypes.

### 2. Etymology and Historical Development

The root of the word **discordance** derives from the Latin prefix "dis-" meaning "apart" or "away," and the noun "cordis" meaning "heart." Therefore, its literal meaning is a state of being "apart from the heart" or lacking agreement, which metaphorically translates to disharmony or strife. Historically, the term was primarily utilized in music and literature to denote a clash of sounds or

ideas. Its application in the scientific realm began to solidify alongside the development of systematic approaches to measuring variance and difference, particularly in statistics and early biometric studies during the late 19th and early 20th centuries.

The concept gained formal scientific rigor with the establishment of quantitative genetics, which sought to measure how much variation in a phenotypic trait could be attributed to genetic variation versus environmental variation. While scientists like Sir Francis Galton initiated the practice of comparing twins to understand inheritance, the formal quantification and rigorous distinction between concordance and discordance became paramount with the rise of modern epidemiological and genetic studies following World War II. Researchers needed a precise metric to describe the outcome when identical individuals did not share the same outcome, thereby enabling the sophisticated calculation of environmental effect sizes.

The historical utility of discordance lies in its capacity to serve as a negative proof in genetic determinism. Prior to the widespread acceptance of complex gene-environment interactions, there was a tendency to attribute high concordance rates in diseases like schizophrenia or heart disease primarily to genes. However, the observation of significant discordance rates, even among genetically identical individuals, forced a scientific paradigm shift, acknowledging the stochastic nature of biological development, epigenetic modifications, and the unique, personalized impact of environmental stressors throughout the lifespan. Thus, the history of **discordance** mirrors the history of refining the nature versus nurture debate into a more nuanced interactionist model.

### 3. Key Characteristics

Discordance is defined by several key characteristics that distinguish it from mere random variation. The first characteristic is **specificity to the trait**; a pair of twins may be highly concordant for height and eye color, yet remain discordant for a complex trait like major depressive disorder or the development of specific allergies. This specificity highlights that the underlying causes for non-shared outcomes are often highly localized, involving specific genetic loci interactions or environmental triggers unique to the individual.

Secondly, **discordance points to non-shared environment (E)**. In the classic ACE model (Additive genetic effects, Common environmental effects, and unique Environmental effects), discordance in MZ twins is the primary observable phenomenon used to calculate the 'E' component. Non-shared environments include prenatal differences (e.g., placental sharing variations), differing peer groups, accidents, unique pathogenic exposures, or even slight variations in parental treatment that are not perceived as shared. These unique experiences contribute to the outcome disparity observed in discordant pairs.

A third critical characteristic is the implication of **incomplete penetrance and variable expressivity**. Discordance for a genetically influenced disease often suggests that the underlying

genetic risk factor is necessary but not sufficient for the disease to manifest. Incomplete penetrance means that not everyone with the disease-causing genotype will develop the phenotype. Discordance is the phenotypic outcome of this incomplete penetrance, signaling that external or stochastic factors must cross a certain threshold alongside the genetic risk to trigger the observable trait. This makes discordance a measure of biological complexity rather than simply a measurement error.

#### 4. Significance in Behavioral Genetics

The analysis of **discordance rates** is the cornerstone of classical twin methodology in behavioral genetics. The entire mathematical framework of quantitative genetics relies on the comparative analysis of concordance and discordance between monozygotic (MZ) and dizygotic (DZ) twins to estimate the parameters of genetic and environmental influence. If a trait were entirely genetically determined, the concordance rate for MZ twins would theoretically be 100%, and any deviation from this rate is direct evidence of non-genetic influences.

When researchers study complex traits--such as personality dimensions, intelligence quotient (IQ), or susceptibility to psychological disorders like bipolar disorder--the degree of discordance among MZ twins is often substantial. For instance, studies on schizophrenia discordance consistently show that if one MZ twin is affected, the risk for the co-twin is significantly elevated compared to the general population, yet it rarely exceeds 50%. This 50% discordance rate is perhaps the most significant finding in the history of psychiatric genetics, proving conclusively that environmental factors are indispensable elements in the etiology of severe mental illnesses, even those with high heritability.

Furthermore, studying twins who are discordant for a specific disorder provides a unique and powerful research design known as the **discordant twin design**. In this model, the affected twin and the unaffected, genetically identical co-twin are compared across various biological or environmental metrics (e.g., epigenetic profiles, brain structure, early life stress exposures). Because the major confounding factor of genetic background is controlled for, any significant measurable differences between the two twins must be attributable to the non-shared environmental factors or stochastic events that led to the expression of the phenotype in one twin but not the other. This methodology offers unparalleled power for identifying critical risk and protective factors.

#### 5. Statistical Calculation and Interpretation

Statistically, discordance is quantified using specific formulas, primarily within the context of liability threshold models applied to categorical traits (e.g., presence or absence of a disease). In twin studies, the rate of discordance is typically calculated using pairwise or probandwise concordance

methods, focusing on the proportion of pairs where only one twin expresses the trait.

For a given population of twin pairs, researchers categorize them into four groups based on trait status: affected/affected (concordant), unaffected/unaffected (concordant), affected/unaffected (discordant), and unaffected/affected (discordant). The **discordance rate** is then the ratio of discordant pairs to the total number of pairs. High rates of discordance in MZ twins suggest low heritability and a dominant influence of unique environmental factors, whereas low discordance suggests high heritability and strong genetic control. Interpreting these rates allows for the statistical decomposition of variance using structural equation modeling, such as the aforementioned ACE model, which partitions total phenotypic variance ( $V_P$ ) into its constituent genetic and environmental components.

The statistical interpretation of discordance must always be viewed relative to concordance. If the concordance rate for MZ twins ( $C_{MZ}$ ) is significantly higher than the concordance rate for DZ twins ( $C_{DZ}$ ), this difference points strongly toward additive genetic influence. Conversely, if  $C_{MZ}$  is high, but the discordance rate is substantial (i.e.,  $C_{MZ} < 1.0$ ), it provides the precise magnitude of environmental input (E) that overrides the shared genetic blueprint. Therefore, **discordance** is not just a measure of difference, but a sophisticated statistical parameter reflecting the plasticity of the human phenotype in the face of identical genetic material.

## 6. Relationship to Concordance

Discordance and concordance are mutually exclusive and complementary concepts; they are two sides of the same coin when measuring trait similarity in twin pairs. **Concordance** signifies agreement or similarity--both twins share the trait or both lack the trait. It is typically associated with the influence of shared factors, including shared genes (A) and shared environment (C). The higher the concordance, particularly in MZ twins, the stronger the indication of genetic determination for that specific trait.

In direct contrast, **discordance** signifies dissimilarity or variance--one twin has the trait and the other does not. It serves as the metric for non-shared, unique influences (E). While concordance maximizes the perceived influence of shared factors, discordance isolates and highlights the power of individuality arising from non-genetic sources. The relationship between the two terms is crucial for the calculation of heritability estimates. For example, Falconer's formula, which estimates heritability based on the difference between MZ and DZ correlation coefficients ( $h^2 \approx 2(r_{MZ} - r_{DZ})$ ), implicitly utilizes the relationship between their respective concordance and discordance rates to derive the proportion of variance attributable to genes.

Understanding the dynamic interplay between these two states is essential for clinical and public health interventions. If a disorder shows very high concordance (low discordance), prevention strategies should focus heavily on genetic screening and early life factors that affect all carriers

equally. Conversely, if a disorder shows high discordance among MZ twins, interventions must shift focus toward identifying specific, idiosyncratic environmental triggers, unique lifestyle choices, or epigenetic modifiers that differentiate the affected twin from the unaffected co-twin. Thus, the balance between concordance and discordance dictates the focus of both basic research and applied prevention efforts.

## 7. Applications Beyond Genetics

While its primary and most rigorous application is found in behavioral genetics, the principle of **discordance** is broadly applied across multiple scientific and psychological disciplines to describe non-agreement or lack of functional correspondence.

In **clinical psychology and medicine**, discordance can refer to the mismatch between a patient's subjective experience (symptoms) and objective clinical findings (diagnosis or physiological markers). For example, patient-physician discordance occurs when the patient's self-reported pain level is inconsistent with objective physical findings, posing challenges for treatment planning. Similarly, symptom-diagnosis discordance may exist when a person exhibits symptoms that do not neatly fit into a single diagnostic category, highlighting the heterogeneity of psychopathology.

In **organizational psychology and management theory**, discordance often describes a lack of alignment between organizational goals and employee actions, or a variance between stated corporate values and actual organizational culture. This operational discordance can lead to inefficiency, low morale, and high turnover. Managers use surveys and performance metrics to identify areas where employee behavior is discordant with strategic objectives, necessitating structural or cultural intervention.

Finally, in the realm of **psychological measurement and statistics**, discordance is a measure of inter-rater reliability or agreement. When two independent observers assess a behavior or score a test, their observations are measured for concordance. High discordance between raters suggests that the measurement instrument or the criteria used for scoring are ambiguous, unreliable, or subjectively applied, requiring refinement of the operational definitions to ensure consistent and valid data collection across researchers.

## Further Reading

[Twin study](#) (Wikipedia)

[Nature Genetics: Twin Studies Collection](#) (Academic Source)

[Heritability](#) (Wikipedia)