twin studies: what can they tell us about nature and nurture?

Twin studies used to be almost the only way to compare the influence of genes against the environment on personality and behavior. Recent advances in genetics, however, suggest that opposing "nature" to "nurture" is misleading. Genes combine with the environment to produce complex human traits.

Harold T. Shapiro and Bernard J. Shapiro are identical twins who both became heads of major universities, Princeton and McGill, respectively. Along the way, they both went to graduate school, specialized in statistics, had academic careers, and held positions as university provosts. Ann Landers and Abigail "Dear Abby" Van Buren were also identical twins. Each became a world-famous newspaper advice columnist, and the two of them dominated the advice-column business for more than 40 years, counseling more than 200 million people. Although their personal rift was widely publicized, the twin sisters agreed on almost everything they discussed in their columns.

Twin stories like these fascinate the general public. As identical twins, each pair shares essentially 100 percent of their genes. And having been raised together, these identical twins also shared the same family environment. So if both common genes and common environment contribute to twins' similarity, how much does each matter? How much do genes and environment shape our lives? These questions can be answered by twin studies, which generally show that both genes and environment influence human traits and behaviors.

traditional twin studies

Scholars have long studied twins to address the nature-nurture question. Identical twins separated at birth and brought up in separate environments provide the ideal test cases, but such cases are exceedingly difficult to find. Instead, most researchers study twins who have grown up together and carefully compare the experiences of identical twins to those of fraternal twins. Since identical twins are monozygotic, developed from a single sperm fertilizing a single egg, which then divides into two separate cell masses within the first two weeks of development, these twins are essentially clones. Fraternal twins, on the other hand, are dizygotic, developed when two eggs are each separately fertilized by different sperm. Identical twins like Dear Abby and Ann Landers have all their genes in common; fraternal twins have, on average, half their genes in common, as do any two full siblings. Comparing identical and fraternal twins enables researchers to separate genetic from environmental influences without measuring genes directly. If a physical or behavioral trait, like hair color or success at math, is shared more often by identical twins than by fraternal twins, researchers reason, genes must have played a role in developing the trait.

There is a big difference between stories about twins and studies of twins. The Shapiro brothers' story is fascinating and suggestive; it may be excellent material for television programs and magazine coverage. But by itself it is not particularly informative. Career similarity between twins could be a result of coincidence, genetic likeness, similar family influences, or a combination of any of these factors. A scholarly study may not be as sensational, but it must be more systematic; it must analyze a relatively large number of both identical and fraternal twin pairs. In particular, it must avoid emphasizing for dramatic effect only identical twins with similar traits and fraternal twins with dissimilar ones.

Twin and related studies provide most of the evidence that genes determine human traits and behaviors. For example, schizophrenia runs in families. Twin studies show that when one twin in an identical pair suffers from schizophrenia, the chance that the other twin will be schizophrenic is about one in two; it is only one in six for fraternal twins. This gap is evidence for the existence of a genetic tendency toward schizophrenia. Similar evidence suggests that genes influence such traits and behaviors as height, weight, manic-depressive psychosis, alcoholism, cognitive development, reading skills, parenting style, rate of accident occurrence in childhood, television-viewing habits, peer-group selection, timing of first sexual intercourse, marital disruption, and educational and economic attainment.

Scholars use the results of the identical versus fraternal twin comparisons (and similar studies of families) to estimate

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table 1:
proportion of variation in selected traits attributable to genes, shared environment, and unshared environment

	Heritability	Shared Environment	Unshared Environment
Personality	0.43	0.06	0.51
Hyperactivity	0.7	0	0.3
Cigarette Smoking	0.43	0	0.57
Cognitive Ability	0.5	0.35	0.15
Weight	0.74	0.06	0.2

Source: Robert Plomin et al., Behavioral Genetics in the Postgenomic Era

how much of the total variation among them in a trait, such as scores on cognitive skills tests, can be attributed to (1) variations in genes (this estimate is called "heritability"); (2) the family environments they share; and (3) everything else (including measurement errors), categorized as unshared environments. Table 1 shows estimates for selected traits for heritability, variations due to shared environment, and variations due to unshared, individually idiosyncratic environments. (The personality measure used in the table is based on what psychologists call the "Big Five" dimensions of personality: neuroticism, extraversion, openness, agreeableness, and conscientiousness.) Genetic factors appear to have a significant effect on all the selected traits in the table. Contrary to traditional views, environmental factors that twins or families share tend to play a much smaller role. Environmental factors unique to each individual also seem important. Lacking direct observations of genes, traditional twin studies rely on statistical analyses of differences in identical and fraternal twins to "decompose" the variations between them into these three sources. These studies cannot, however, tell us about the specific effects of particular genes or particular environments—only about the contributions of genes, shared environment, and individual environment.

problems with traditional twin studies

Traditional twin study designs rely on two assumptions, both of which have prompted criticism. First, the "equal environments" assumption states that the environments of identical twins are no more similar than the environments of fraternal twins. However, if the experiences of identical twins are more similar, genetic influences would be overestimated. Critics note that identical twins tend to be treated more alike than fraternal twins and suspect that this greater environmental similarity may explain the greater trait similarity of identical twins. For example, identical twins are probably more often dressed alike than fraternal twins. Ann Landers and Dear Abby were almost mirror images of one another as young children. If being treated identically, in dressing for instance, makes identical twins more similar than fraternal twins, we could mistakenly attribute effects to genes that are really due to differences in treatment.

More examples of similar treatment of identical twins in certain aspects of life, however, does not automatically discredit twin studies. What is crucial is whether the special way identical twins are treated affects the outcome of interest. If dressing does not, for instance, affect schizophrenia, the validity of the equal environments assumption is not relevant as far as the conclusions about genes and schizophrenia are concerned.

The second assumption is that there is little or no "assortative mating"—the tendency of people to marry people who are like them in intelligence, personality, looks, and so forth. Assortative mating could distort estimates of genetic influences in family studies. Children of similar parents would be more likely to receive the same genes for some traits than children of more dissimilar parents. For this reason, assortative mating would exaggerate genetic similarity for *fraternal* twins, but it would not affect genetic similarity for *identical* twins because they are 100 percent similar genetically, with or without parental assortative mating. In a twin study, assortative mating could underestimate the influence of genetics, which is calculated by measuring the differences in similarity for identical twin pairs against the same similarities for fraternal twins. Violations of the equal environments and the assortative mating assumptions thus have opposite effects and may, to some extent, cancel each other.

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Another type of skepticism about twin studies is more fundamental. Until recently, researchers could only observe genetically related siblings, not their actual genes. They had little understanding of the genetic mechanisms that might lead individuals to exhibit specific traits. The dependence of twin studies on genetically related family members, who also tend to share similar environments, makes many twin studies appear unconvincing and, at times, even suspicious. The tremendous advances in molecular genetics over the past two decades, however, are helping to improve the evidence. Twin studies and other related studies are no longer the only strategy for studying the genetic influence on human traits and behaviors. Molecular work adds further support to twin-based research.

molecular genetics

Before the early 1980s, human genes could only be identified through the biochemical defects and altered proteins of the gene involved. Even relatively simple genes proved quite difficult to analyze biochemically. The study of human genes changed markedly about two decades ago, with the discovery that the natural variations in human DNA sequences at a single chromosomal location can be used as markers to map the position of a gene along a chromosome. The development of human genetics since then has been dazzling. The discovery of superior classes of markers has contributed to mapping hundreds of chromosomal regions that contain genes affecting human diseases. These procedures have led to the identification of the specific genes that cause such disorders as Huntington's Disease, Duchenne muscular dystrophy, cystic fibrosis, hereditary non-polyposis colon cancer, and heritable breast cancers.

Most of the human disorders whose genes have been identified are genetically "simple," or Mendelian traits, in which there is a connection between some observable feature of people—for example, cystic fibrosis—and a single point on a chromosome. These simple Mendelian traits are rare. Almost all common disorders, such as heart disease, hypertension, diabetes, cancer, and infection, and almost all human traits interesting to social scientists, such as cognitive ability and personality, are non-Mendelian, or "complex."

Complex traits are usually influenced by multiple genes, multiple environmental factors, and interactions among these factors. In such cases, many people with the appropriate genes do not show the trait, while others with the genes do. In addition, different genes may stimulate similar traits. Major efforts are currently underway to map and identify the genes underlying some of these complex traits. Hirschprung's disease and Crohn's disease, for example, are two success stories. Both exemplify how molecular geneticists can successfully identify the genetic components of a complex human trait.

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twin studies in the post-genomic era

The extraordinary developments in molecular genetics have affected twin studies in several ways. First, advances in molecular genetics have substantiated hypotheses generated by the traditional twin research design. The importance of genes suggested by earlier twin studies has often been confirmed by later molecular genetic studies. For example, numerous twin studies have consistently supported a significant role for genetic factors in reading disability or dyslexia. In general, twin studies have attributed to genetic factors 40 to 70 percent of the variation among people in measures of reading. More recent molecular genetic studies point to a small number of chromosomal regions that may be related to susceptibility to dyslexia. These confirmations of the results of twin studies suggest that we need to move beyond endless debates about the assumptions of the traditional twin research design. Many molecular genetic studies have shown the usefulness of twin studies as an exploratory tool, whether or not the assumptions of equal environments and assortative mating are exactly met.

Molecular genetic studies have shown the usefulness of twin studies as an exploratory tool.

Second, twin studies of genetic influence can only yield preliminary results. The proof of the genetic basis of a human trait must await molecular genetic studies. Twin and other family studies are still useful as exploratory tools even when DNA-based studies are feasible. In the early stages of many large, DNA-based studies, including the well-publicized breast-cancer-gene study, researchers have used twin and family studies to explore the possibility of a genetic basis of a trait. These results are valuable indicators of whether further DNA-based research on a specific trait is worthwhile.

Third, even in DNA-based studies, researching identical and fraternal twins is still extremely valuable. Researchers can use identical twins to estimate the proportion of individuals with a certain gene that expresses a certain trait. While geneticists who study nonhumans can rely on experimental animal and plant studies, identical twins provide almost the only "quasi-experimental" possibility for human geneticists. If one person in an identical twin pair is affected by a trait, the probability of the other twin being affected supplies a ready estimate of what proportion of people with a gene in a given environment will exhibit the trait. Fraternal twins provide excellent data for genetic mapping. One advantage of fraternal twins is that they are exactly the same age. The effects of age sometimes confound genetic research. For instance, researchers studying reading disability found that efforts to correct reading deficits make it difficult to diagnose disabilities in older children. Twin studies allow comparisons of children of the same age and make it easier to explore genetic sources.

Cartoon courtesy of www.CartoonStock.com

Fourth, and perhaps most important for sociologists, despite major efforts and numerous technological breakthroughs, successful genetic mapping of "complex" disorders such as schizophrenia, manic-depressive (bipolar) disorder, autism, and type 1 and type 2 diabetes are rare. Social and behavioral scientists who study complex human traits and behaviors are likely to continue using twin and other traditional family studies as a major methodological strategy. Such studies will also remain useful for understanding how much a particular environment influences the expression of a genetically influenced trait.

using twins to study gene-by-environment interactions

Counterpoising genetics and environmental influences is somewhat misleading, despite the partisan claims of people on each side. Many human traits and behaviors result from both genetic and environmental factors. Moreover, genetic and environmental influences on a trait are not simply additive. Genes "interact" with the environment. That is, genes provide the potential for a trait, but environmental conditions determine whether that potential will be realized. The same genetic codes may be expressed at different levels in different environments.

Compare, for example, Asian immigrants in the United States to U.S.–born Asian Americans. U.S.–born Asians are twice as likely as immigrants to suffer from prostate cancer, and Asian-American adolescents born in the United States are more than twice as likely to be obese as Asian-American adolescents who recently immigrated to the United States. U.S.–born Asians and immigrant Asians are likely to have similar genetic predispositions for prostate cancer and obesity. The differences between the two groups in the prevalence of these disorders are, therefore, likely to be caused by environmental conditions such as lifestyle and diet.

To understand gene-by-environment interactions, we must evaluate the estimated heritability of traits in particular environments. The term *heritability* is often misunderstood. The traditional twin study design generally produces a single heritability estimate. Heritability, however, is not a fixed property of a trait; it could vary from one population to another. For example, under different social circumstances, the heritability for cognitive development may differ. We could make one estimate of heritability based on a U.S. middle-class population and another based on a low-caste population in India. We would expect the former to be significantly larger than the latter. In a modern liberal democracy, individuals enjoy more access to educational opportunities than in a traditional aristocratic society. As a result, the differences in cognitive achievement in a modern democracy should be due more to genetic differences than in a traditional society. An egalitarian, democratic society can be thought of as a "normal" environment for estimating the heritability of cognitive development, where the "genetic potential" for cognitive development can be realized. We can compare heritability in a traditional or more hierarchical society against this potential.

We know that even in a modern democracy access to educational opportunities varies considerably by ethnic or social group. In an impoverished urban neighborhood, the genes for cognitive development might not be as fully expressed as the

same genes in a suburban neighborhood. Using a sample of the statistics on more than 1,500 pairs of twins and siblings from the National Longitudinal Study of Adolescent Health, Elizabeth Stearns and I investigated children's differential opportunities to achieve their genetic potential for cognitive development. We found that African-American children and children whose parents are unemployed are less likely to realize their genetic potential for cognitive development than white children and children of employed parents, respectively. Thus, improving the social circumstances of disadvantaged children may help them reach their full cognitive potential.

In another study based on the same sample of adolescents, my colleagues and I found that friends' drinking behaviors seem to influence the expression of one's own genetic propensities for drinking. Regardless of a youth's genetic predisposition for alcoholism, having friends who drink heavily increases the chances that he or she will drink. Of course, this conclusion might have come from a standard sociological study of adolescents. So what then is the added value of a twin study? The study points to a probable environmental effect: friends' drinking behavior. Major DNA-based studies that aim at identifying the relevant genes are under way. When the genes are identified, studies may be carried out to examine whether friends' drinking behavior moderates the expression of each of the identified genes, as the twin research suggests. If so, appropriate strategies for treatment may be designed.

While disagreeing about the relative influence of the environment and genetics in the expression of certain traits, most social scientists probably agree that human traits and behaviors are influenced to some degree by both. However, until very recently, empirical work by social scientists has rarely recognized this, perhaps because we lacked DNA measures and perhaps because of the weaknesses of twin studies discussed earlier. The situation is beginning to change. As noted, molecular genetics has had remarkable success over the past twenty years in identifying the genes for simple, or Mendelian, traits. The human traits and behaviors that interest social scientists are almost always non-Mendelian and complex. For complex traits, twin studies remain important as an exploratory tool, and social science is indispensable to understanding environmental factors. For example, schizophrenia occurs in both identical twins 48 percent of the time, which means that about half the time only one of a pair of genetically identical twins develops schizophrenia. This is unmistakable evidence for the influence of non-genetic factors and the continuing relevance of social science to what may appear at first glance to be a genetically determined disorder.

Twin studies will continue to inform us about the relative importance of genes and environment on traits in ways that no molecular genetic breakthrough can completely elucidate. Moreover, twin studies may help shed light on specific environmental factors that moderate a genetic predisposition toward a trait or behavior. Although molecular genetics now occupies center stage, twin studies will continue to make noteworthy contributions to understanding how environmental factors and genetics combine to create human traits and behaviors, a fundamental scientific enterprise for the 21st century.

"Patriotism is supporting your country all the time and the government when it deserves it."
—Mark Twain

Photo by Helen Stummer

recommended resources

Guang Guo and Elizabeth Stearns. "The Social Influences on the Realization of Genetic Potential for Intellectual Development." *Social Forces* 80 (2002): 881–910. This article shows how, in the absence of DNA data, twins and siblings can be used to study the interactions between environment and heritability.

Online Mendelian Inheritance in Man. http://www.ncbi.nlm.nih.gov. This database is a catalog of human genes and genetic disorders authored and edited by Dr. Victor A. McKusick and his colleagues at Johns Hopkins and elsewhere. It was developed for the World Wide Web by NCBI, the National Center for Biotechnology Information.

Robert Plomin, John C. DeFries, Ian W. Craig, and Peter McGuffin, eds. *Behavioral Genetics in the Postgenomic Era* (American Psychological Association, 2002). Contains numerous articles by experts in molecular genetic studies of human traits and behaviors.

Robert Plomin, John C. DeFries, Gerald E. McClean, and Michael Rutter. *Behavioral Genetics*. 3rd ed. (W. H. Freeman, 1997). A good introduction to the field of genetic analysis of human traits and behaviors.

Neil J. Risch. "Searching for Genetic Determinants in the New Millennium." *Nature* 405 (2002): 847–50. Risch reviews achievements in molecular genetics over the past two decades and discusses some current issues from the perspective of a genetic epidemiologist.