Evidence for Genetic Influences on Homosexuality

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It is hardly daring these days to hypothesise that a trait or characteristic has a genetic component. During the past decade studies in behavior genetics have shown substantial hereditary contributions in personality (Loehlin & Nichols, 1976), intelligence (Bouchard *et al.*, 1990), and psychopathology (Kendler *et al.*, 1987), as well as traits generally assumed to be socially determined, such as social attitudes (Martin *et al.*, 1986) and religiosity (Waller *et al.*, 1990). Findings of non-zero heritability have become so common that zero heritability is no longer an interesting null hypothesis. For most traits, it is almost certainly false.

There are, however, some exceptions. All other things being equal, a characteristic associated with decreased reproductive success should be less heritable than one which is uncorrelated with it. The decrease in expected heritability should be more pronounced, first, the greater the decrement in reproductive success, and second, the more common the characteristic. The reason for the former is that an allele for a characteristic with deleterious reproductive consequences should be eliminated in favour of alternative alleles with less negative consequences. The reason for the latter relationship is that the more common a characteristic, the more difficult it is to account for the existence of genes for it by the mutation rate.

Homosexual orientation is a trait that is both relatively common and has a strong negative association with reproductive success. The largest study of the question suggested that homosexual men and women have less than a quarter the number of offspring that heterosexuals have (Bell *et al.*, 1981). Although the frequently proffered frequency for homosexuality of 10% is almost certainly exaggerated, the more plausible lower estimates of 4% and 1.5% for male and female homosexuality, respectively (Gebhard, 1982), are still much higher than the highest known mutation rates. Thus, sexual orientation is a good candidate for diminished heritability. Given this argument, it is surprising that the bulk of the available evidence suggests moderate heritability for male sexual orientation. Female sexual orientation has been studied much less extensively, but recent studies are consistent with a genetic contribution for women as well.

Family Studies

The first step to establish a heritable component for a trait is often a study of familial aggregation. Heritable characteristics should run in families. Early studies (Henry, 1941) suggested a familial tendency for male and female homosexuality, but these studies were not systematic. There have been several recent family studies of male and female sexual orientation. In the first study of this type, Pillard and Weinrich (1986) recruited 51 homosexual and 50 heterosexual male probands using newspaper advertisements that did not mention the purpose of the study. Probands were asked to rate their siblings' sexual orientations. Furthermore, they were asked for permission to contact all siblings. Results suggested that probands were quite accurate in rating their siblings' sexual orientations. Furthermore, using either self-ratings or sibling-ratings, brothers of the homosexual probands were substantially more likely to be homosexual than were brothers of heterosexual probands – 20% vs. 4%, respectively. These results were consistent with another family study of male homosexuality, performed by Bailey and Pillard (1991). They obtained estimates of the number of brothers "known" to be homosexual from heterosexual male and female probands, as well as homosexual male probands, finding a 10% rate among the brothers of homosexual males compared to a 2% rate among the brothers of heterosexual males and females. Although brothers were not contacted to verify their sexual orientations, results of Pillard and Weinrich suggest that this is unnecessary. Thus, male sexual orientation appears to be familial.

There have now been two recent reports regarding female homosexuality, as well. Pillard (1990) found a 25% rate of homosexuality or bisexuality among 60 sisters of bisexual or homosexual female probands, compared to a rate of 11% for sisters of 53 heterosexual female probands. He also found a marginally significant elevation of homosexual brothers among their homosexual female probands, suggesting common influences in the development of male and female homosexuality. Bailey and Benishay (1993) found that 12% of the 99 sisters of the homosexual probands were also homosexual, compared to 2% of the 83 sisters of the heterosexual probands. Similar to Pillard's results, a slightly higher percentage of brothers of the homosexual probands were also homosexual (7% to 1%), although this difference was not significant. Both studies used the methodology of Pillard and Weinrich (1986), attempting to ascertain siblings' sexual

orientations directly, when possible. Both found proband reports to be highly predictive of sibling self-reports.

Thus, the available evidence supports the hypothesis that both male and female sexual orientation run in families. Some of these studies suggest co-familiality of male and female homosexuality, although this is less clear. Further studies using larger numbers of subjects are necessary to determine whether they are co-familial. The finding that a characteristic is familial supports the possibility of genetic factors. However, it is insufficient to establish genetic influences, as a trait may be familial for either genetic or environmental reasons. In order to distinguish these possibilities, more sophisticated methodologies are needed.

Genetic Studies

Kallmann (1952) performed the first, and until quite recently, the largest twin study of sexual orientation. He searched psychiatric and correctional institutions and the "homosexual underworld" for male homosexuals with twins, and found a 100% concordance rate for 37 male monozygotic (MZ) twin pairs, compared to a 15% rate for 26 male dizygotic (DZ) twin pairs. Kallmann's study has been severely criticized for its methodology, particularly its sampling biases and its anomalous findings. In contrast to Kallmann's results, several case studies and smaller twin series (Rosenthal, 1970) suggest that the true MZ concordance rate is substantially less than 100%, though it appears to be appreciable (Pillard, 1990). Although the criticisms of Kallmann's study are justified, his results have been rejected too swiftly and unconditionally. Despite all the problems of the study, it is difficult to imagine a plausible explanation for the findings that omits genetic influences.

A report of two pairs of male MZ pairs reared apart supports the likelihood of genetic factors (Eckert, 1986). Both members of one pair were unambiguously homosexual. In the other pair, one twin identified himself as homosexual and the other as heterosexual. However, the "heterosexual" twin had had a long term homosexual relationship, and admitted to sexual attraction to his twin. More likely he was at least bisexual, and thus both pairs were concordant for the presence of homosexual feelings. The same study reported on four separated female twin pairs with homosexuality. None was concordant, raising the possibility that genetic factors play less of a role for female than for male

homosexuality. However, the sample was much too small to be more than suggestive of sex differences in genetic transmission.

During the past two years there have been several larger reports of genetic studies of sexual orientation, primarily concerning males. The first, by Buhrich *et al.*, (1991), used male twins from the Australian Twin Registry, who completed mailed questionnaires. The response rate was respectable, with a 69% of individuals and 53% of pairs returning completed questionnaires. The final sample consisted of 95 MZ and 63 DZ pairs. The questionnaires contained items about sexual orientation, as well as the related traits of sexual identity (the subjective experience of oneself as male or female) and sex-dimorphic behaviours. All three traits were assessed for their present status and retrospectively, for childhood. Homosexual males have a relatively feminine pattern of sexual identity and sex-dimorphic behaviors, especially in childhood, compared to heterosexual males. The inclusion of these traits allowed the investigation of the reasons why the three traits are related, using multivariate statistical analyses.

Univariate analyses, which require relatively few assumptions, supported the likelihood of familial factors influencing all traits, with the exception of childhood sexual identity. That is, genetic and/or familial environmental (also called "shared environmental") factors were necessary to explain the pattern of twin correlations. However, the univariate analyses were generally unable to determine whether twin resemblance was due to genetic or shared environmental factors, or both. Subsequent multivariate analyses suggested that there was a genetic factor common to all the traits. Because the twin correlations were imperfect, the environment also appeared to be important. However, the effective environmental factors were comprised of aspects of the environment that even identical twins reared together do not share. This would seem to exclude, for example, an unconditionally "distant" father or an "overbearing" mother.

The study by Buhrich *et al.*, (1991) is the most systematically ascertained sample studied to date. Furthermore, the simultaneous consideration of related traits such as sexual identity also commends it. There are some limitations, however, that preclude it from being definitive. First, the study was relatively small, containing only 20 individuals who could plausibly be called bisexual, the rest of the individuals being heterosexual and hence providing little information. Second, despite the attempt to ascertain subjects systematically, there was a

higher rate of homosexuality among the MZ twins, suggesting a possible volunteer bias. The most plausible – but not the only – explanation is that MZ discordant pairs are more likely than discordant DZ pairs to participate. Another limitation is that the multivariate analyses, which were the most informative about the relative importance of genes and shared environment, relied on technical assumptions that were not well tested, due to the small sample. The approach of Buhrich *et al.*, is a sound one, and it is to be hoped that a replication will be attempted using a substantially larger sample.

An unselected population-based twin sample will have relatively few homosexual subjects, due to the low incidence of homosexuality in the general population. One alternative that has been employed in several studies is selection using the proband method. In this methodology, only gay men who have twins (or other relatives of interest) are recruited, ignoring, for instance, heterosexual men with twins and gay men without twins. The crucial data in such a study are the rates of homosexuality among the different types of relatives, such as MZ and DZ twins. There have been two recent reports using this method.

The first and larger study, by Bailey and Pillard (1991), recruited gay men with twins or adoptive brothers using advertisements in gay publications. (Adoptive brothers are biological unrelated males reared as siblings to the probands.) Probands telephoned the investigators and were interviewed, usually in person, about their sexual orientations and related traits. They were also asked about their brothers' sexual orientations and for permission to contact their cotwins or adoptive brothers. There was a concordance of 98% between sibling self-reports and proband reports. The rates of homosexuality (including bisexuality) among the relatives were: 52% (29/56) for MZ cotwins; 22% (12/54) for DZ cotwins; and 11% (6/57) for adoptive brothers. There was, however, one finding of the study difficult to reconcile with a simple genetic contribution. Non-twin siblings had a significantly lower rate (9%) than DZ twins, though the two kinds of relatives are equivalent in genetic similarity. Heritability estimates were computed varying two assumptions. The base rate of homosexuality was assumed to be either 4% or 10%. The effect of ascertainment bias was also examined, specifically the possibility that probands from concordant pairs would be more likely than probands from discordant pairs to volunteer. Heritabilities were computed assuming no ascertainment bias and two values of non-zero bias. Heritability estimates were all at least moderate, and most of them exceeded 0.50.

There is a serious methodological problem with studies such as this, however. Because probands are recruited as volunteers, they may be unrepresentative in important respects. Specifically, potential probands' willingness to volunteer may depend on the sexual orientation of their twins or adoptive brothers. Bailey and Pillard found that if the bias was similar for MZ and for DZ twins, heritability estimates were not lowered appreciably. However, if MZ twins from discordant pairs were substantially less likely than DZ twins from discordant pairs to volunteer, seriously misleading results could result. That kind of bias could even lead to a spurious finding of heritability when true heritability is zero. We doubt that this accounts for Bailey and Pillard's results, but that possibility cannot presently be ruled out. Unfortunately, the other available studies using the proband methodology suffer from the same flaw. The ideal study would ascertain probands systematically, to eliminate ascertainment bias. Studies of psychopathological disorders such as schizophrenia, for instance, ascertain probands as consecutive admissions into mental hospitals. Unfortunately, there is no equally efficient way of ascertaining homosexual twin probands.

Setting aside this limitation for the moment, and accepting Bailey and Pillard's conclusion of non-zero heritability, does this study have anything else to say about the origins of sexual orientation? Bailey and Pillard sought to elucidate the nature of the genetic contribution by seeing whether it was related to any measured phenotypic characteristics. If a characteristic is a marker for high genetic loading, then MZ probands with the characteristic should be particularly likely to have homosexual cotwins. For example, Bailey and Pillard investigated whether MZ probands' childhood gender nonconformity (CGN – analogous to Buhrich *et al.*'s "sex-dimorphic behaviours") predicted concordance (Green, 1987). However, MZ probands who recalled being effeminate in childhood were just as likely as more masculine probands to have homosexual cotwins. Bailey and Pillard were unable to find any characteristic that predicted distinguished probands from concordant and discordant pairs.

They also examined how similar MZ cotwins were for CGN. Cotwins in concordant MZ pairs were remarkably similar for CGN. However, discordant pairs were essentially uncorrelated for CGN. Thus, if an effeminate MZ proband had a homosexual cotwin, the cotwin also tended to be effeminate, and if a masculine MZ proband had a homosexual cotwin, the cotwin also tended to be masculine. (In contrast, if the cotwin was heterosexual, one could not make an

accurate prediction about his degree of CGN from the proband's score.) This is interesting because it suggests that genes and/or shared environmental factors may also affect the expression of salient differences among homosexual men, such as CGN.

Finally, Bailey and Pillard noted that among the cotwins of MZ probands, there was a bimodal distribution of sexual orientation, as measured by Kinsey scores. That is, cotwins tended either to be heterosexual or homosexual, and there were relatively few bisexuals. This suggests that on the phenotypic level, sexual orientation may be more of a categorical than a dimensional characteristic. This does not necessarily imply discontinuity on the etiological level, such as the presence or absence of a major gene. Indeed, bimodality among MZ cotwins is not relevant to the detection of a major gene.

A second genetic study, restricted to twins, was reported by King and McDonald (1992). They used a method of recruitment similar to that of Bailey and Pillard, and found a sample of 46 homosexuals with twins (38 male, 8 female). The reported concordances, 25% for MZ twins compared to 12% for DZ twins, are somewhat lower than those obtained by Bailey and Pillard. However, King and McDonald's sample was considerably smaller, so that the difference might largely be due to sampling error. Furthermore, the written report was brief, and some important information was omitted. For instance, it is unclear how zygosity was diagnosed in this study, cotwins were not contacted to verify orientations, nor were results reported separately for men and women. Finally, it is striking that 5 of 7 respondents considered their cotwins entirely heterosexual despite an apparently prolonged incestuous homosexual relationship. However, the differences between King and McDonald's results and those of Bailey and Pillard highlight the need for replication using more systematically ascertained samples.

Thus, the bulk of the available genetic evidence suggests that male sexual orientation is at least moderately heritable. However, the limitations of this literature must be recognized. The studies least subject to ascertainment bias, such as Eckert *et al.* (1986) and Buhrich *et al.* (1991) had too few homosexual twins to yield statistically powerful tests of hypotheses. Furthermore, there was some evidence for ascertainment bias in Buhrich *et al.*'s study. The studies with reasonably large numbers of homosexual subjects, including Kallmann's (1952), Bailey and Pillard's (1991), and King and McDonald's (1992), all were subject to

more serious forms of ascertainment bias, and yielded a wide range of concordances. Nevertheless, no study to date has produced results inconsistent with at least moderate heritability for male sexual orientation.

We have focused on male sexual orientation, since there is a paucity of genetic data for females. This is probably primarily due to the substantially lower base rate of female homosexuality, which appears to be less than half as common (Gebhard, 1982). A complete explanation of sexual orientation will only be possible, however, when females have been studied as well.

Future Directions

There are two main directions for future research. First, more scientifically rigorous behavioral genetic studies of sexual orientation are needed, in order to clearly confirm the promising results of available studies. Since at least two laboratories have begun molecular genetic studies of sexual orientation, this is particularly important, to ensure that these efforts are not wasted chasing chimerical genetic influences that are actually methodological artifacts. One method that should be pursued is that employed by Buhrich *et al.* (1991), in which members of a large twin registry are recruited for study. Another approach is the proband method, in which one would first locate a large sample of gay men, and then systematically study all those with twins and/or adoptive siblings. The key to both approaches is to minimize the likelihood that ascertainment will be associated with the sexual orientation of relevant subjects (e.g., cotwins of gay probands). As social tolerance of homosexuality increases, more methodologically rigorous studies will be possible.

Assuming that nontrivial heritability is proven beyond reasonable doubt, there remains the question of which genes are involved and what they are doing. Methodologies for the study of single genes, such as linkage analysis, are useful in theory for the identification of genes affecting a trait. However, to this date they have not been very useful in studying behavioral characteristics (Plomin, 1990).

Behavior genetics methods can also be used to study mechanisms indirectly. For example, one striking result of the more recent genetic studies is the high rate of discordance among the MZ twins. Except for Kallmann's study, concordances

have been well under 100%. This shows that environment must exert an influence on sexual orientation. Furthermore, the effective environment appears to comprise some aspect of experience (loosely defined) that differs between MZ twins of the same pair, reared together. It should be remembered that "environment" refers simply to all nongenetic influences, biological or social. There are no current theories of sexual orientation that would predict frequent discordance between MZ cotwins. Given theories of prenatal influences on sexual orientation (Dorner, 1990), it would be useful to know whether aspects of the prenatal environment such as hormonal exposure, can plausibly differ between MZ cotwins. Discordance in HIV infection in MZ co-twins born to HIV infected mothers certainly points to the possibility (Goedert *et al.*,1991) of differential prenatal biological influences. On the psychosocial side, the equivalent question is whether parental treatment of twins differs in ways likely to foster differences in sexual orientation.

One strategy to study genetic mechanisms indirectly is to determine whether probands of concordant MZ pairs differ from those in discordant MZ pairs in some identifiable way. Probands of concordant pairs presumably have relatively high genetic loadings compared to those of discordant pairs. As previously noted, Bailey and Pillard (1991) found that MZ probands' CGN was unrelated to concordance status. This finding should be replicated, and other candidates should be studied as well, such as history of heterosexual feelings and behavior.

Conclusions

Both male and female sexual orientation appear to be familial. Furthermore, the available evidence suggests that for males this familial tendency is, at least in part, due to genetic factors. No genetic study has avoided significant methodological problems. On the other hand, it is reassuring that all have found some evidence for genetic influences.

We conclude with a reconsideration of the paradox with which we began. How can genes that decrease their bearer's fertility resist elimination by natural selection? The general answer is simple: while such genes may detract from homosexuals' reproductive success, they facilitate the reproductive success of other (nonhomosexual) individuals who carry them. One possibility, for example,

is kin selection, whereby homosexuals reproduce less, but help their siblings reproduce more than they otherwise would have. This model suggests that homosexuals should invest more in their siblings than heterosexuals do, a prediction that has yet to be studied. Another possibility is that the relevant genes confer unknown biological advantage (e.g., disease resistance, avoidance of violent conflict) on their carriers. The model here is the allele sickle cell anemia, which present in heterozygous form, protects against malarial infection. If this model is correct, then it may be possible to detect biological differences between homosexuals and heterosexuals that are beneficial to the former. Empirical work on the fascinating question of how homosexual genes might persist has not begun. The evidence reviewed herein suggests that it is reasonable to ask the question.

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