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Speaking of twins

It has been said that all parents are environmentalists until they have their second child. The insight may come even sooner to those who have twins, and studies of identical and non-identical twins have provided geneticists with some of the best data for dissecting out the relative contributions of genes and environment to the emergence of human cognitive traits. The paper on page 324 from Robert Plomin and colleagues is one such example; by analyzing a large cohort of two-year-old twins, they have shown that delayed acquisition of language has a strong genetic component, and that whereas for most children the effect of genes is small relative to that of their environment, the children who are slowest to acquire language are delayed in large part because of their genetic makeup.

Twins provide an almost ideal natural experiment for studies of human heritability. Monozygotic twins are genetically identical, whereas dizygotic twins share on average 50% of their autosomal genes. Of course, parents and offspring, and non-twin siblings, are equally related, but twins provide a built-in control for many important sources of environmental variation. By comparing monozygotic and dizogotic twins with each other and with the population as a whole, it is possible to measure the extent to which a given trait is affected by genetic versus environmental influences. Plomin and colleagues took advantage of the UK Office for National Statistics to contact parents of almost every pair of twins born in England and Wales in 1994. The parents were sent a questionnaire shortly before the children's second birthday, in which they were asked to identify their twins as identical or non-identical (this is known to be about 96% accurate), and to provide information about their vocabulary as well as various non-verbal abilities. Vocabulary was estimated by identifying which words each child knew, from a test set of 100 words that had been selected as a good predictor of total vocabulary. The response rate was an impressive 44% (parents of twins are typically very supportive of genetic studies) and after discarding from the sample those children with deafness or other complications, the authors were left with more than 3000 pairs. This may be the largest cohort of twins ever examined, and is likely to be a reasonable representation of the British population as a whole.

By the age of two, children have typically passed the 'babbling' stage and are acquiring new words rapidly, with vocabularies ranging from a few to many hundreds of words. Two-year old children typically hear thousands of words per day, mainly from their parents, and it seems plausible that differences in linguistic environment may account for at least part of the variation in vocabulary. But as Chomsky has emphasized, children show an extraordinary ability to acquire language even from an impoverished stimulus, suggesting the existence of a 'language instinct' that is buffered against variations in the linguistic environment. Might this ability be affected by genetic differences between individuals?

Previous studies had already suggested that there would be some genetic effect, although they were based on smaller sample sizes and had not looked as early as two years, when language is first starting to emerge. The surprising result of this new study is that the relative contributions of genes and environment differ for different parts of the population distribution. Specifically, while the heritability of vocabulary for the population as a whole is fairly low (25%) with a correspondingly large environmental contribution, a much greater heritability (73%) is seen for the bottom 5% of the distribution, that is for those children who are slowest to acquire vocabulary and who would be typically classified as having specific language impairment (SLI).

These results suggests a number of conclusions. First, SLI is not simply the tail of the normal distribution, but rather a distinct genetic condition (or conditions; the data do not address the question of whether SLI is genetically or functionally heterogeneous). The genetic risk arises not from a single mutation but from the combined action of a number of alleles whose effects are individually weak and largely additive (rather than depending on specific combinations, as seems to be the case for certain other conditions such as autism). Importantly, the effects of most of these alleles appear to be specific for language; although some of the children in the bottom 5% for vocabulary also performed poorly on nonverbal tests, the majority did not, and the heritability estimate was hardly altered when the low nonverbal scorers were omitted from the analysis. So far, there is no evidence that the genetic factors predisposing children to SLI produce any other systematic effects.

The results therefore set the stage for identifying what might crudely be called 'genes for language'. How close is this prospect? The technological advances in molecular genetics over the last few years have been enormous, and continue to accelerate; on some recent estimates, the human genome may be completely sequenced within the next three years. Moreover, it is no longer necessary to perform laborious linkage studies to identify genes associated with particular quantitative traits; instead, once a trait is known to be highly heritable,

the entire genome can be scanned with a closely spaced array of polymorphic markers to determine which ones are associated with affected individuals. Identification of the nearby genes of interest still requires significant further searching, but this will be greatly facilitated by the emerging technologies of DNA chips and single nucleotide polymorphisms, as well as by the availability of the complete genome sequence.

What can we expect to learn from the identification of such genes? A plausible guess is that at least some of them will be involved in the development of brain regions that are specifically involved in the use of language. While these genes will probably not explain most of the variation in normal human linguistic ability (since the variant alleles affect mainly the bottom 5% of two-year-olds), their identification can hardly fail to provide new insights into the neural basis of language acquisition. A particularly powerful approach may be to combine genotyping with functional brain imaging and cognitive or psychophysical studies, to determine whether specific alleles have measurable effects on brain function. Moreover, since language is such a uniquely human attribute, it will be fascinating to determine whether such genes have homologs in other primates, and whether they can shed new light on human brain evolution.

Studies of human heritability, particularly of cognitive traits, have often been met with skepticism from those who fear their implications, particularly the suggestion that traits with high heritability are in some sense genetically predetermined. But such fears are based on a fallacy; heritability in a given regime of environmental variation says nothing about the possibility that different environments might lead to different effects. Plomin himself is no genetic determinist; indeed, he emphasizes that one goal of this work is to identify children who are at risk for SLI, so that they can be given preventative treatments that may help them avoid the sequelae of their condition (for instance dyslexia). And even if some of the problems prove refractory to intervention, a genetic explanation is not obviously less palatable than an environmental one that attributes the child's difficulties to the failings of the parents. Nevertheless, as similar methods are applied to other cognitive traits, some difficult issues will inevitably arise; not least of these is the danger inherent in drawing an apparently objective and genetically-based distinction between 'normal' and 'abnormal' personalities.