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The Genetic Roots of Language

What SLI and Williams Syndrome Can Teach Us About the Roles Played by Genes in the Development of Language and Intelligence

by Andrea McColl

I'm sitting here in the student lounge, listening to my friends discuss their physics homework, tonight's dinner, and whether the "big ass party" someone put up fliers about is going to be a very big party, or a party for large-bottomed people. While the physics itself is proving difficult, something else they're doing is effortless—the sharing of ideas, abstract concepts, and common experiences through the production and interpretation of a complex pattern of sound waves. Surrounded by people with whom we can communicate in this manner, we don't question our own language abilities, yet we base judgments about others' intelligence on their language ability. We (consciously or not) rank those who speak in complicated, fully grammatical sentences as more intelligent than those who use only short sentences or make frequent grammatical errors. This apparent connection can be deceptive, and looking at where it breaks down reveals especially important pieces in the jigsaw puzzle that is language.

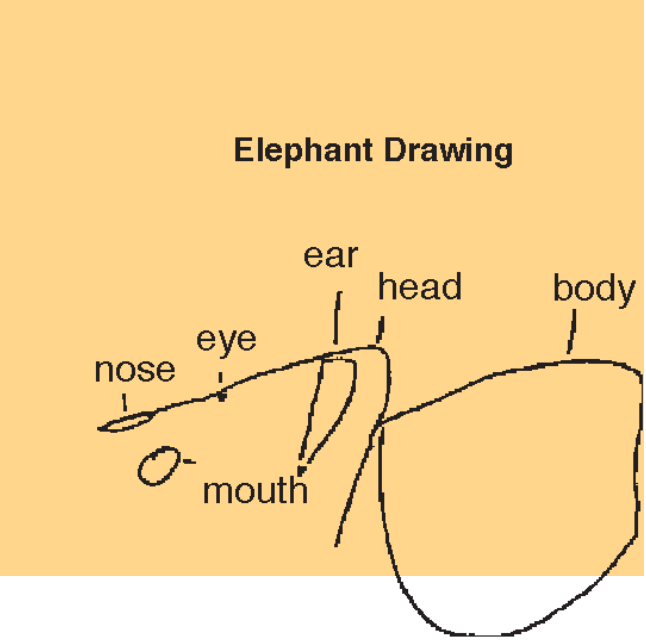
Six-year-old Hannah Gadlage (she's now eight) has the characteristic elfin features of Williams Syndrome. (Photo courtesy of the Gadlage family.)

Goo-Goo Genes?

Although born not knowing language, most of us are born with the ability to acquire it. Babies come prepared to learn language, and they will easily acquire the intricate structures of a fully developed language even when their environment is deficient. For example, there are many recorded cases of deaf children whose hearing parents learned sign language. Being second-language learners, these parents didn't achieve fluency—and yet their children, learning only from them, became completely fluent, with a grammar more complex and accurate than that used by their parents. This suggests a genetic predisposition to acquiring language.

Some argue that humans are born with a universal grammar, a framework into which the details of one's native language are placed. Since all languages contain, for example, nouns and verbs in some form, it can be argued that nouns and verbs are part of the structure of this universal grammar. However, this still is just a theory; the definitive proof depends on genetic evidence. More specifically, were it to be shown that identifiable genetic changes lead to identifiable linguistic problems (ideally, for example, that a mutation in chromosome 13 affects only verb conjugating abilities and nothing else), that would be enough proof for most researchers.

There are two disorders that suggest particularly compelling links between genetics and language: Williams Syndrome and Specific Language Impairment (SLI). In brief, with Williams Syndrome, individuals have incredibly low intelligence and unexpectedly fluent speech; with SLI, individuals have otherwise normal intelligence and significant language difficulties. While this in itself is a striking contrast, it is even more significant that individuals with Williams Syndrome are missing roughly 20 consecutive genes on one copy of their chromosome 7. (Humans have 23 pairs of chromosomes and at least 20 to 30 thousand genes.) SLI patients show strong patterns of inheritance as well. Many researchers herald these



This illustration of an elephant, drawn by an individual with Williams Syndrome, is indicative of the visuospatial deficits common to this disorder. (© Dr. Ursula Bellugi, The Salk Institute)

disorders as proof of the independence of language and intelligence. Some suggest that these disorders show a link between genes and language. Is the evidence sufficient?

Elephants and Elves: Williams Syndrome

When a 15-year-old with Williams Syndrome was asked to draw an elephant, she produced the drawing shown above, unidentifiable without the verbal description she produced while drawing (as described by Ursula Bellugi). "And what it has, it has long grey ears, fan ears, ears that can blow in the wind. It has a long trunk that can pick up grass, or pick up hay . . . If they're in a bad mood it can be terrible." Her sentences are nearly as complex as her drawing is simple, and are far more complex than one would expect from an individual with an IQ of 49.

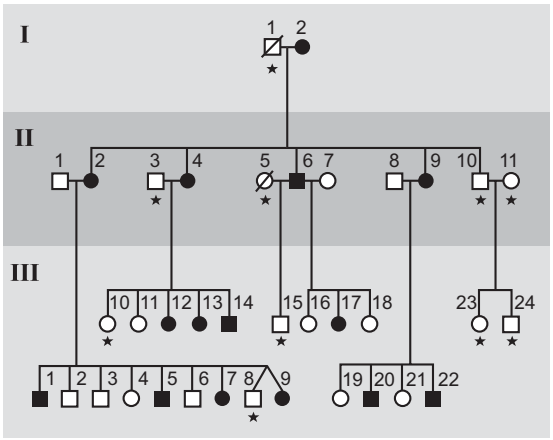
In addition to this extreme disparity between linguistic and visuospatial abilities, people with Williams Syndrome show other distinctive characteristics, both intellectual and biological. They generally have a strong affinity for music and show remarkable musical abilities, being able to write and sing songs, and to play songs by ear after hearing them only once. Perfect pitch also appears to occur more frequently in the Williams Syndrome population than in the general population. Physically, they have very distinctive "elfin" facial features, heart problems (such as aortic narrowing), high blood-calcium levels, and difficulty producing elastin, a protein that normally works with collagen to regulate the ability of joints and tendons to stretch. It was these last two biologic characteristics that enabled researchers to isolate the exact genetic problem that leads to the disorder. In 1993, the gene for elastin was identified as one of those in the region of chromosome 7 that is missing in Williams Syndrome individuals. With this discovery came the question: What role does this missing information play in producing the symptoms of the syndrome? While it is clear that losing one copy of the gene for elastin could affect its production, the other missing genes have not yet been mapped to the other characteristics of Williams Syndrome.

With recent sequencing of the human genome, it is becoming far simpler to identify the genetic source of various syndromes. Scientists are using this knowledge to figure out which genes affect which proteins. Moving from this microscopic evidence to a particular physical trait is a much more difficult leap: Would a missing gene have produced a protein essential to the developing brain, therefore making the effects of its absence irreversible? Would the missing gene have produced a protein that leads to lower levels of, for example, sodium in the blood, leading to an effect that can be easily treated by adding sodium to an affected child's diet? Is there still some other, more complex connection that has yet to be determined?

While the discovery of the genetic causes of Williams Syndrome is significant, it does not tell the whole story about the disorder. What we have learned about Williams syndrome strongly supports the idea that intelligence and language are not irrevocably interconnected, as had previously been thought, and this has further reinforced the theory that our ability to use language is based in our biology, not in our training.

Missing the Miscellaneous: SLI

Williams Syndrome is a very specific diagnosis that includes particular physiological and biochemical symptoms, as well as the more visible ones, and occurs in roughly one out of every 30 thousand births. In contrast SLI, Specific Language Impairment, is a much less well-categorized disorder. SLI is a title used to describe a family of related language disorders, occurring in approximately 7 percent of the population. An SLI



The pedigree of the KE family shows a dominant inheritance pattern, as roughly half of the children of affected individuals also have the disorder. Circles are females; squares are males. A shaded box represents an individual with language problems. In level II, numbers 2, 4, 6, 9, and 10 are siblings.

diagnosis requires that a patient have normal (or higher) IQ (as measured in a series of nonverbal tests), and score significantly low on several language tests that verify lower-than-normal language ability. There can be no external factor that potentially contributes to the language deficit: Hearing must be normal, speech must be physically and developmentally possible (SLI cannot be diagnosed in pre-language infants), and there must be no significant neurological damage.

Some individuals with SLI appear to be merely linguistically delayed, starting out a few years behind their peers with respect to language acquisition but eventually catching up and developing normal speech. Others never achieve full normalcy, suffering with their particular linguistic problems throughout their lives. Among the patterns found in SLI patients (although no single patient had every symptom) are:

- frequently pluralizing nouns improperly;
- frequently omitting the verb to be, as in "That man in a dark room."
- never using the past-tense marker "-ed";
- comprehending metaphors with difficulty (even when other comprehension of speech is normal); and
- using pronouns incorrectly, as in "Her eat. And her get clothes on."

These and other problems have been seen in both spontaneous and prompted speech.

Most English-speaking SLI individuals have a unique weakness in their grammatical morphology. They are unable to add necessary bits to words in order to indicate that they are plural (cat – cat**s**), have been done by someone (climb – climb**s**), happened in the past (walk – walk**ed**), and so on. In English, the words without these fragments added on are still legal words; however, this is not always the case in other languages. For example, in Italian, most words never appear in their stem form, but always have something added to provide more information. It is acceptable to say *parlo* (I speak), *parlate* (y'all speak), and *parlano* (they speak), but no Italian speaker would ever say the bare stem, *parl*. Although Italian-speaking SLI children have problems with morphology, they never eliminate modifiers. Instead, they substitute another word form, following a relatively standard pattern, such as using the third-person singular form of the verb instead of the third person plural, as in *vende* (he or she sells) instead of *vendono* (they sell). One theory is that, instead of changing the words to their proper form, they have memorized one or two forms of the words, and produce those whenever any form of the word is needed.

This may also be a learning deficit due to difficulties in processing fast sounds; and if so, in hearing the small sounds that make up these grammatical morphemes. Some studies have shown that individuals with SLI are worse at sequencing fast sounds, but this theory is not widely accepted. What *is* known is that there is still a great deal to learn about SLI, and this theory suggests a way in which genetic problems could impact language acquisition—through the hearing pathway. Without being able to properly receive or distinguish the sounds heard, making it to the next steps in language processing becomes far more difficult.

Several trends suggest the influence of genetics on SLI: Males are more likely to have SLI, and children with SLI are more likely to have at least one parent with a language problem. Many of this latter group of children have nonimpaired siblings. Additionally, studies of identical and fraternal twins have revealed that, especially among the fraternal twins, a statistically significant number of pairs consist of one impaired and one non-impaired twin. This in particular indicates that "nurture," or the role of environment in development, is not the sole cause of

language problems, as there have been many studies showing that twin children receive essentially the same linguistic input.

One large family, the KE family (whose pedigree is shown in diagram on the opposite page), in which many members have the same speech disorder, provides striking evidence of the role of genetics. This family is especially significant because the disorder exhibits the very clear inheritance patterns of a dominant gene. For example, individual II 10 in the pedigree is the only child of I 1 and I 2 not to have the disorder, and did not produce any children (III) with this disorder. However, half of his siblings and nephews were impaired. This inheritance does not appear linked to gender. It is also significant to note that impaired individuals produced both normal and impaired children; thus the environment in which these children were raised was the same, and likely had little influence on whether or not they developed the disorder.

The genes responsible for this particular family's disorders have been mapped to a particular area on chromosome 7, and genetic analysis of an unrelated patient who exhibited similar language problems revealed a mutation in this same region. While further studies are necessary, it may be that this mutation leads to a lack of a particular protein that affects the development of neural structures important for speech and language, the way similar proteins have been shown to influence neuronal development in other organisms. The overall genetic pattern exhibited by the KE family, especially with the discovery of the gene deletion, strongly suggests that a gene, or a small set of genes, has a major impact on language development.

Intelligence doesn't require language. There are thousands of recorded cases of acquired aphasia, where a subject has lost language ability due to some physical or physiological cause, such as a stroke or other nonintellectual brain damage.

Current studies are attempting to divide SLI patients into distinct subgroups, based on the details of their impairment, so that testing can determine more precisely the causes of the impairment. Perhaps one day, as with Williams Syndrome and the KE family, other types of SLI can be linked to specific chromosomes and genes and tell us more about the language acquisition pathway.

Broken Brains

Intelligence doesn't require language. There are thousands of recorded cases of acquired aphasia, where a subject has lost language ability due to some physical or physiological cause, such as a stroke or other nonintellectual brain damage. Well over a century ago, studies of brain-damaged individuals concluded that language ability resides almost entirely in the left hemisphere of the brain, which is also the case for most normal individuals. A few have right-hemisphere language; these people are commonly also left-handed and show other signs of hemisphere role-reversal. Among the most famous of these studies were those done by Pierre Paul Broca in 1861. One of his patients was so severely aphasic that he could utter only one word, "tan." An autopsy revealed that neurosyphilis, a degenerative disease, had damaged a very specific zone in this patient's brain (subsequently called Broca's area), a region later determined to be very important for control of speech production. Damage to Broca's area leads to great difficulties in speaking, but does not affect understanding of speech. Although it takes great effort for a Broca's aphasic to articulate words, when they do manage to name items, they do so correctly.

Damage to another region of the brain produces essentially the opposite effect. Patients with damage to Wernicke's area retain fluent and grammatically correct speech—but cannot understand what they are saying, or what anyone is saying to them. (Noam Chomsky, a famous linguist, once illustrated the separation between meaning and grammar in this completely grammatical yet meaningless sentence: "Colorless green ideas sleep furiously.") The speech of a Wernicke's aphasic is filled with nonsense words and incoherent trains of thought. While Broca's aphasics have very slow, stilted speech (when they can speak at all), Wernicke's aphasics talk a great deal, but when asked to name words, they either use a completely incorrect name, or select a related but incorrect word (such as knee for elbow).

Modern studies, especially those using magnetic resonance imaging, or MRI (a fast and non-intrusive technique that uses powerful magnetic

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fields to image the inside of a living body, which can be used to observe a brain in action and take pictures of the brain regions activated during various tasks) have verified the diagnosed function of these regions, as both Wernicke's and Broca's areas show distinct activity as impaired and nonimpaired patients perform a variety of linguistic tasks.

The distinct localization of these two areas of language in very specific regions of the brain is further evidence of a biological basis for language. That these target regions serve the same purpose in everyone shows that these areas must form during brain development; thus, genetic mechanisms must shape their creation.

Is the Evidence Enough?

Contrasting the SLI and Williams Syndrome studies neither proves nor disproves a genetic basis for language acquisition. However, it does show that language and intelligence are in fact independent. Since there exist people with normal intelligence who cannot speak properly, and people with dramatically low intelligence who speak normally, it is illogical to use language alone as a sign of intelligence. Rather, the origins of each should be explored separately.

SLI research is an especially promising source for information about language-specific genetic disorders. By identifying a group of unrelated SLI patients with the same particular language problems, researchers may be able to isolate a genetic marker for their particular variant of the syndrome. Imaging studies are being used to pinpoint the brain locations used in performing different linguistic tasks, and these studies may help divide SLI patients into different categories. A brain that does not activate along normal patterns could lead to some of the problems observed in SLI, and thus it might be possible to categorize some varieties of SLI based on patterns of brain activity. This would make it possible to examine each category, looking for common traits not found in either normal individuals or in other SLI categories. Such imaging studies may reveal whether SLI is more a hearing or linguistic deficit, or even perhaps two different disorders, one with each cause, that coincidentally appear similar to linguistic researchers.

Research continues on the linguistic front as well. Linguists are looking at the details of particular language deficits and are trying to characterize them. Then, using these deficits to characterize the essentials of language, they are seeking universal, separable properties of language that are coded in the DNA. If, for example, the ability to pluralize words can be "broken" genetically (by deleting a gene) without impacting the individual's other language, it would be plausible that this ability is an inherent structure, somehow separable from the rest of language. Cross-linguistic studies (such as the Italian one mentioned previously) have added more depth to this research; finding speakers who exhibit the same pattern of broken structures, regardless of native language, will add support to the independence of that structure.

The basic set of assumptions are that language is more than just a learned skill, that humans are either biologically predisposed towards language acquisition or have a built-in universal grammar framework upon which to build an acquired language, and that language is not tied to intelligence. These assumptions seem simple, but that is only because researchers have uncovered evidence such as the striking contrast between SLI and Williams Syndrome. These findings have led to far greater understanding of language and intelligence, as discussed here, and also of genetics, the structure of the brain, and the unexpected roles of various proteins in development.

So the next time you're talking with someone, take a moment to appreciate the complicated set of factors that enable you to communicate. And take a moment to thank your parents, as well, because it was their genetic contribution that made it possible for you to get your friends to help you finish your physics homework in time to go to that very large party. □

Andrea McColl

PICTURE CREDIT: Doug Cummings

Andrea McColl majored in biology and is now a first-year grad student in linguistics at the University of Southern California, where she will be working on a project on semantic deficits in Alzheimer's patients. As a Caltech undergraduate, she was a member of the fencing team and choreographed sword fights for several Theater Arts productions. Her faculty mentor on the Core 1 paper was Fiona Cowie, associate professor of philosophy, and the editor was Dian De Sha.