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The Problem

Humans are wired to be social, communicative creatures, but sometimes this circuitry goes awry. Researchers are striving to pinpoint the links between genes, brain activity and behavior to better understand psychological behaviors and mental disorders, such as autism. Understanding the basis for sign language or the cause of anxiety, for example, can help provide fundamental insights into the genetic mechanisms and neural circuits responsible for human social behavior.

The Approach

Ursula Bellugi pioneered the study of the biological foundation of language. She is regarded as the founder of the neurobiology of American Sign Language (ASL), because her work was the first to show it as a true language as processed by the brain, revealing more about how the brain learns, interprets and forgets language. Constantly seeking new avenues for illuminating the ties between neural and cognitive functions, Bellugi uses her expertise in neurobiological, genetic and behavioral studies to better understand Williams syndrome—a puzzling genetic disorder that results in low IQ and strong desire for social interactions—and autism. While patients with autism usually shy away from social interactions and eye contact, Williams syndrome patients do exactly the opposite, seeking out interactions with people. Bellugi is using imaging technologies to visualize how related gene deletions alter brain activity, mapping the affected neural

circuits, and developing stem cell reprogramming techniques to unveil the underlying biological basis for these drastically different disorders. Together, her studies on Williams syndrome, autism and sign language help paint a picture of the biology we use to interact with those around us.

The Innovations and Discoveries

- Bellugi discovered that ASL is processed by the same areas of the brain that interpret the grammar and syntax of spoken language. This was one of the first pieces of neurobiological evidence that ASL is treated as a true language by the brain.
- She found that people with Williams syndrome have boosted levels of oxytocin, the so-called “trust hormone,” explaining why they seek social interaction despite having other learning and cognitive disabilities. The finding helps researchers understand the normal role of oxytocin in the brain as well as which genes regulate the hormone's production and release.
- She compared the language-processing abilities of patients with Williams syndrome and autism and found a difference in brain patterns. When Williams syndrome patients heard a sentence with an out-of-place word, they showed a spike of activity in one area of the brain; the activity peak was absent in those with autism.

For more information, please visit:
<http://www.salk.edu/faculty/bellugi.html>

Autism, Genetics, Neurological Disease,
Neurobiology, Stem Cells, Williams Syndrome