“Williams syndrome is a perfect example of where a genetic predisposition interacts with the environment to sculpt the brain in unique ways. It provides a unique window for understanding how missing genes and the resulting changes in brain structure and function ultimately shape behavior.”

Autism spectrum disorders (ASD) and Williams syndrome (WS) are both neurodevelopmental disorders, but their manifestations couldn’t be more different. While autistic individuals live in a world where objects make much more sense than people do, people with WS are social butterflies who bask in other people’s attention. Despite myriad health problems, generally low IQs, and severe spatial problems, they are irresistibly drawn to strangers, look intently at people’s faces, remember names and faces with ease, and are colorful and skillful storytellers.

In recent studies, Ursula Bellugi and her team compared brain response patterns linked to face and language processing between individuals with ASD and WS to gain novel insights into their polar opposite social and communication profiles. To this end, they measured so-called event-related potentials, or ERPs for short, which are brainwaves directly reflecting electrical activity of the brain occurring in response to discrete events. They found that when viewing human faces, individuals with WS respond with a unique brain signature not found in ASD or any other group.

The observed differences may underlie gaze avoidance in ASD and reflect the increased interest in and attention to human faces in WS. Similarly, individuals with WS exhibited an abnormally large ERP response when a typical sentence finishes with an odd ending (“I take my coffee with sugar and shoes”), indicating that they are particularly sensitive and attuned to semantic aspects of language. In contrast, individuals with ASD did not show this negativity, suggesting that the inability to integrate lexical information into the ongoing context may underlie their communicative and language impairments.

To gain a better understanding of the neural and genetic correlates of social behavior in different social phenotypes, Bellugi’s team is now integrating these findings with the exquisitely mapped genetic profile of WS. They are hypothesizing that specific genes in the WS region may be involved in the dysregulation of specific neuropeptide and hormonal systems, which could explain the observed hypersocial behavior.

For more information, please visit salk.edu/faculty/bellugi.html