VANDERBILT KENNEDY CENTER

Measuring Receptive Communication in Angelman Syndrome

A Research Report for Families

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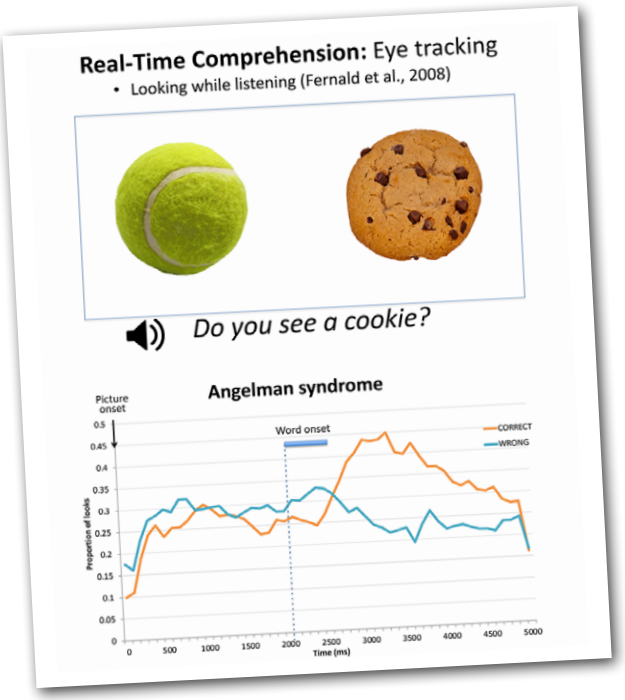
Introduction

Angelman syndrome is a rare genetic disorder that causes developmental delay, absent or limited speech, and difficulties with coordinated movements. As a result, the full range of abilities in individuals with Angelman syndrome may not be accurately captured by the conventional behavioral measures. This lack of sensitive measures poses a challenge for evaluating developmental changes and/or the effects of interventions.

To date, most available data about communicative and cognitive processes in Angelman syndrome has come from observations and reports by caregivers. In this study, our aim was to investigate receptive communication abilities of children and adults with Angelman syndrome using objective measures of brain activity and eye gaze recorded directly from the participants to obtain a more complete understanding of their speech processing.



What the Study Involved

This study focused on speech processing – the ability to pay attention, recognize the sound of familiar words, and understand their meaning. These questions have not yet been examined in great detail in Angelman syndrome, even though most daily activities involve processing of spoken language.

We used EEG (electroencephalography) to measure brain responses called auditory event-related potentials. An event-related potential (ERP) is the change in brain electrical activity in response to a specific stimulus event. Wearing a soft sensor net with many sponges, the participants listened to novel and familiar spoken words. The first set included made-up nonsense words, one of which was presented repeatedly, while the rest were played only once. Another set consisted of familiar words (such as dog, book, or cookie) mixed with nonsense words. We also used a computer camera to record eye movements while the participants looked at pictures of familiar objects and listened to the computer naming them (e.g., "Do you see a cookie?"). Our previous work established these tasks as effective measures of auditory attention and comprehension in children and adults with typical and atypical development.

The Study's Questions

The study addressed three main questions:

- ❑ Do children and adults with Angelman syndrome pay attention to speech sounds in their environment?
- ❑ Do their brain responses separate known words from nonsense sounds?
- ❑ What is the time course of spoken language comprehension in Angelman syndrome?

Study Design

You and your family member with Angelman syndrome were a part of a study sample of 43 families. All participants were between 4 and 45 years of age (mean age was 14 years). We recorded auditory brain responses and eye movements during a single research visit. Without the requirement of deliberate behavioral responses, these brief passive listening tests made the procedures suitable for all ages and ability levels. Our measure of communicative and social functioning came from the questionnaires about word knowledge and the daily living behaviors you had observed in your son or daughter over time. By relying on your extensive knowledge and experiences, we learned about what your family member could do not just at the time of our research visit, but on a regular basis at home and other settings.

Findings

All participants tolerated the sensors on their head for at least 6 minutes, which was long enough to record one set of brain responses. They were also able to pay attention to the computer screen for the 3-minute eye tracking task.

- ❑ The brain responses demonstrated the expected evidence of attention to spoken stimuli as reflected by greater difference between responses to repeated nonsense words compared to the novel stimuli heard only once. Better ability to detect word repetition was associated with parent reports of higher receptive communication and interpersonal adaptive functioning.
- ❑ Brain responses to the real vs. made-up spoken English words showed the pattern previously observed in young language learners with typical development. The expected larger brain responses to known words over the left hemisphere were present even in some

participants whose caregivers reported them not knowing some of the stimulus words, suggesting the possibility of a larger receptive vocabulary than previously thought.

- ❑ Eye tracking task demonstrated that children and adults with Angelman syndrome may take approximately 0.4 seconds longer than typical peers to process and respond to a spoken cue. The results also highlighted high variability in the ability to maintain visual attention while listening to brief spoken sentences. Faster gaze shifts to the named objects were observed in participants whose caregivers reported larger vocabularies, more adaptive communicative functioning, and fewer behavioral concerns with irritability and hyperactivity.

With your help, this study provided direct evidence that children and adults with Angelman syndrome are actively engaged with their language environment, rely on typical neural mechanisms to recognize known words, and may need additional time to react to what they hear. The objective nature of the brain- and gaze-based measures makes them useful for evaluating treatment effects in future clinical trials.

Additional Video Resource

Alexandra (Sasha) Key discussed this study and its findings in a Vanderbilt Kennedy Center Video Research Brief. You can view the 2 minute video at: vimeo.com/509901183

Heartfelt Thanks

We are grateful to the families who participated in this study. You are our partners in discovery. Without families taking part in research, we could not advance our understanding of the full range of functioning in individuals with and without developmental disabilities.

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