

**RESEARCH SUMMARY**

MEASURING COMMUNICATION ABILITY FOR KIDS WITH NEURODEVELOPMENTAL DISORDERS

In the first two years of a five-year project, researchers at Duke University interviewed family caregivers of children and adolescents with neurodevelopmental disorders. Caregivers were asked to describe how their child communicates, helping to improve the way we measure communication ability.

Aidan Carter, who is affected by Hunter syndrome, is held by his mother Jen as he waits for his weekly 3 hour enzyme treatment at Children's Hospital in Waltham, MA.

OVERVIEW

A person's ability to communicate effectively is extremely important to both their quality of life and that of those around them. Communicating is more challenging for people with neurodevelopmental disorders (NDDs). As new treatments to improve communication ability are tested, researchers need ways to learn if they work.

Having a reliable way to assess a child's communication ability before, during, and after a new treatment is key. Since family caregivers (such as parents) spend the most time with their child, they need to be included in determining if the treatment is making a difference.

Many existing tools designed to assess communication have limitations—particularly when a child uses limited verbal speech or an assistive device (like a tablet) to help them communicate.



[Scan for more info about the ORCA Measure](#)

By partnering with families, researchers at Duke aimed to better understand the important ways that children with NDDs communicate with their families, especially those who have limited verbal communication or are primarily non-verbal. They also explored the relevance of an existing measure, the Observer-Reported Communication Ability (ORCA) measure.

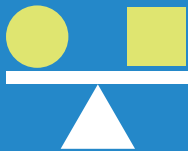
WHAT WERE RESEARCHERS TRYING TO LEARN FROM THIS STUDY?

Researchers were interested in three primary areas.

They wanted to:



Learn how individuals with NDDs typically communicate, from their caregiver's perspective



See how well the measure captures key aspects of their children's communication ability



Identify opportunities to refine the measure to make it more clear, and capture additional communication concepts that will expand the range of the measure

WHAT EVALUATION METHODS DID RESEARCHERS USE IN THE STUDY?

In the first part of the five-year study:

Researchers conducted 115 interviews with caregivers of children diagnosed with one of 12 different NDDs. These interviews helped the researchers understand

what aspects of their children's communication are most important and meaningful to families. They also interviewed clinicians (for example, speech language pathologists) to learn how different aspects of communication are different or similar across NDDs.



WHAT DID THE RESEARCHERS LEARN SO FAR?

Across NDDs, communication can take multiple forms, including: spoken words, spoken approximations of words (like 'baa' for 'bottle'), physical gestures and body movements, American Sign Language, eye gaze, and various communication systems (tablets, devices, pictures, boards, books). Considering these forms of communication, researchers asked caregivers to describe behaviors that they observe in their children.

Communication is complex. Caregivers spoke about many different reasons their child communicates with them, including to make requests, to seek attention, and to ask questions. They also spoke about how they know their child understands their communication, like following directions and responding to their name. They also talked about social communication, like greeting others and playing games.

WHAT DID CAREGIVERS TALK ABOUT?

"If she wants us to open a toy for her or get it to spin or light up and she can't do it, she'll take our hand and put it on the toy."

– Caregiver of child with SYNGAP1-related intellectual disability

"She most frequently will use the 'I want' button [on the AAC device] when she's going in or the people button...So, she can ask for people."

– Caregiver of child with STXBP1-related disorder

"Usually his hands, like to point, because if we don't understand him, he'll just say, 'Look! Look!' And he'll pull us in and show us what he's talking about."

– Caregiver of child with Hunter syndrome

"She touches something if she wants it. If she wants the TV on, she touches the fireplace because the TV sits above the fireplace."

– Caregiver of child with Phelan McDermid syndrome

"Like a typical two-year-old. You know, it's like fussing and kind of throwing her body around. Like I don't wanna do this. And diaper changes are a big one."

– Caregiver of a child with Malan syndrome

"So, most of her communication style is with her eyes – looking at things, smiling, kinda nodding – encouraging if it's something that she wants."

– Caregiver of a child with SCN2A-related disorder

"Sometimes, like if she wants to dance, she'll say like shake your butt, because she wants...her dad to put music on."

– Caregiver of child with SETBP1-HD

"If I say 'We're gonna go take a bath,' then sometimes he'll clap his hands."

– Caregiver of child with Schinzel-Giedion syndrome

"So, I'll say if we finished dinner, 'Put your cup in the sink, and throw your plate away'... she's very familiar with those, so she can follow those."

– Caregiver of child with HNRNP2-related disorder

"She'll either follow through with what you ask of her or she'll respond in an appropriate manner. So, if you tell her that you love her, she'll give you a hug, or she'll smile."

– Caregiver of child with BBSOAS

"If we give her options, like, 'Do you want bathtub or shower?' she'll say which one she wants to do."

– Caregiver of child with GRIN2B-related NDD

"She will sign, 'Please.' Or she'll sign, 'More.' and then, 'Food,' of course. But she'll sign the same thing for a drink. She has started pointing to things which is fairly new."

– Caregiver of a child with Hao-Fountain syndrome



THE ORCA MEASURE

The ORCA measure was designed originally for individuals with Angelman syndrome, who have similar communication challenges to the groups included in this study.

It was created specifically to evaluate a child's typical communication abilities over the previous 30 days.

It focuses on meaningful aspects of a person's communication ability from the caregiver's point of view.

In the first part of the five-year study, researchers found that, overall, the concepts that the caregivers and clinical experts talked about matched well with the concepts on the ORCA measure. Feedback from caregivers included changes to some of the current questions and the possibility of adding new questions.

THREE TYPES OF COMMUNICATION



1. EXPRESSIVE

- Use of symbols and words
- Seek attention
- Direct attention
- Refuse object
- Request object
- Request object out of view
- Request more
- Ask questions
- Understanding information
- Communicate with people



2. RECEPTIVE

- Turns in conversation
- Make choices
- Responding to name
- Respond to familiar directions
- Respond to new directions
- Understanding mood
- Understanding isolated words
- Respond to questions



3. PRAGMATIC

- Greeting
- Use names
- Playing games
- Comfort others

WHAT POPULATIONS WERE REPRESENTED IN THIS STUDY?

This study included children with rare and ultra rare NDDs including:

- SYNGAP1-related intellectual disability
- STXBP1-related disorders
- Hunter syndrome (MPS-II)
- Phelan-McDermid syndrome
- Malan syndrome
- SCN2A-related disorders
- SETBP1 haploinsufficiency disorder
- Schinzel-Giedion syndrome
- HNRNP2-related disorders
- Bosch-Boonstra-Schaaf optic atrophy syndrome (NR2F1)
- GRIN2B-related neurodevelopmental disorder
- Hao-Fountain syndrome



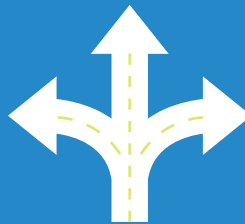
Elena Escoto, 6, who was born with Angelman syndrome, greets her mother at the bus stop after school.

The study population consisted of English-speaking families. Caregivers had to verify that their child's diagnosis was confirmed through genetic testing. The study group consisted of caregivers of children one-year-old and older.

Over 30% of caregivers in the study identified with a racial or ethnic group that is traditionally underrepresented in clinical research (e.g. African/American or Black, American Indian/Alaskan Native, Asian, Middle Eastern, mixed race, Hispanic-Latino). Most caregivers had at least some college or graduate education.

WHAT ARE THE NEXT STEPS?

Researchers will work to modify the ORCA measure and then get more feedback on the changes. They want to make sure all updates are easy to understand and relevant to caregivers. They will also conduct an electronic survey and statistical evaluation to explore if the measure is reliable and valid. The ultimate goal would be to use of the measure in future clinical trials.



DISCLOSURES

The study team members have developed the technology being discussed. If the technology is commercially successful in the future, the developers and Duke University may benefit financially.

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Through COMBINEDBrain, we also worked closely with representatives from these patient advocacy organizations:

SYNGAP RESEARCH FUND

Collaboration. Transparency. Urgency.

SynGAP Research Fund (SRF)



STXBP1 Foundation

PROJECT/ALIVE

Project Alive



CureSHANK



Phelan-McDermid Syndrome Foundation



Malan Syndrome Foundation



FamilieSCN2A Foundation



SETBP1 Society



Schinzel-Giedion Syndrome Foundation



Yellow Brick Road Project



NR2F1 Foundation



GRIN2B Foundation



Foundation for USP7-Related Diseases