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.
WHAT WERE RESEARCHERS TRYING TO LEARN FROM THIS STUDY?

Researchers were interested in three primary areas. They wanted to:

1. Learn how individuals with NDDs typically communicate, from their caregiver's perspective.
2. See how well the measure captures key aspects of their children's communication ability.
3. 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 HNRNPH2-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

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– Caregiver of a child with Hao-Fountain syndrome
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
- HNRNPH2-related disorders
- Bosch-Boonstra-Schaaf optic atrophy syndrome (NR2F1)
- GRIN2B-related neurodevelopmental disorder
- Hao-Fountain syndrome

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** (SRF)
- **STXBP1 Foundation**
- **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**

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