

BACKGROUND

There is a high prevalence of gastrointestinal (GI) problems in individuals with neurodevelopmental disorders (NDDs). However, type, frequency and impact of these problems has been variable and difficult to ascertain across individuals with rare genetic disorders and NDDs and those with idiopathic NDDs. While there are variabilities in symptom presentation between different forms of NDDs, there are also differences across different subtypes. In addition, it is unclear if individuals who are either non-speaking or cognitively disabled have the ability to express the source of their GI distress or are able access or tolerate appropriate diagnostic evaluations and treatments.

SURVEY & MEETING

The [Consortium for Autism, Neurodevelopmental Disorders and Digestive Diseases \(CANDID\)](#) was formed in 2022. The goal of the collaboration was to better understand and helping those with co-morbid GI problems and NDDs across the spectrum with different abilities and disabilities.

A quantitative survey including the PEDSQL and PROMIS for GI symptoms and Quality of Life (QOL) was administered to 717 caregivers with minor or adult children with either syndromic or idiopathic forms of ASD. This was done to determine prevalence and type of GI problems in children and the impact on families. Recruitment was conducted through patient advocacy groups and through autism organizations. Families were invited to give their feedback on the usefulness of the QOL measures. The Indiana University reviewed the IRB.

Results were presented at the CANDID virtual conference, made possible by NIH grant R13 TR003938-01 and the work of organizations like ASF, PMSF, CDLK5 and Indiana University Recordings of the presentations are publicly available at www.candidgi.com or by scanning the QR code.

CANDID CONFERENCE AGENDA

Impact of GI Disorders on Quality of Life: A Family Panel
Family stakeholders¹

The Big Picture (and Survey Results)
Dr. William Bennett²

GI and Other Comorbidities in Neurodevelopmental Disorders
Dr. Andres Jimenez Gomez³

Brain - Gut Issues in an Autism GI Clinic
Dr. Kent Williams⁴

The Brain-Gut-Microbiome Axis in Autism Spectrum Disorder
Dr. Kara Gross Margolis⁵

Growth and Nutritional Deficiencies: Lessons Learned from Rett Syndrome
Dr. Kathleen Motil⁶

Traditional Diagnostic Strategies in Pediatric Gastroenterology and the Challenges of their Application to NDDs
Dr. Joseph Croffie⁷

A Need to Improve Detection and Monitoring of GI Distress in NDDs - The Adult Perspective
Dr. Baharak Moshiree⁸

Adapting Survey Instruments to Better Capture Gastrointestinal Distress in NDD Populations
Dr. Calliope Hologue⁹

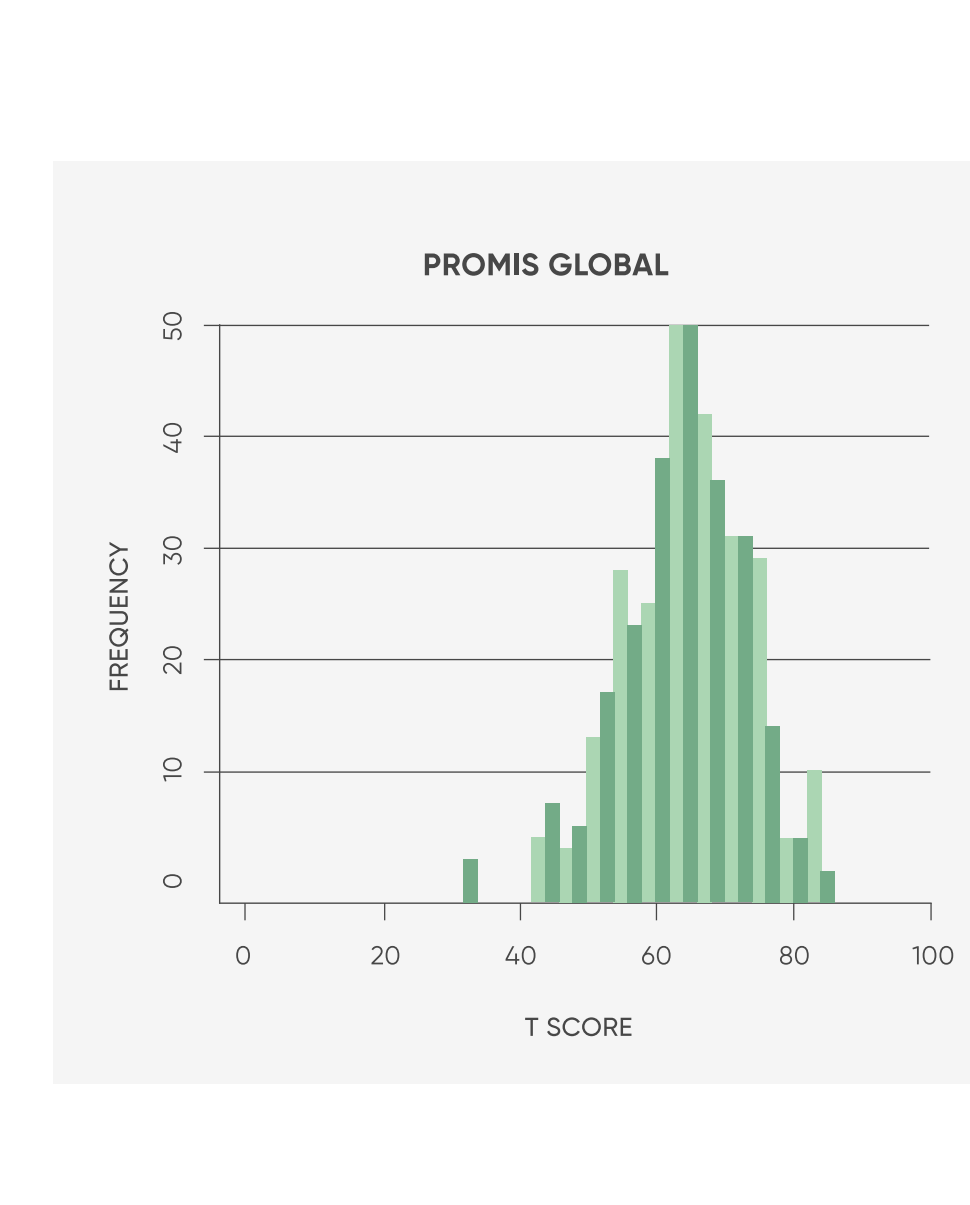
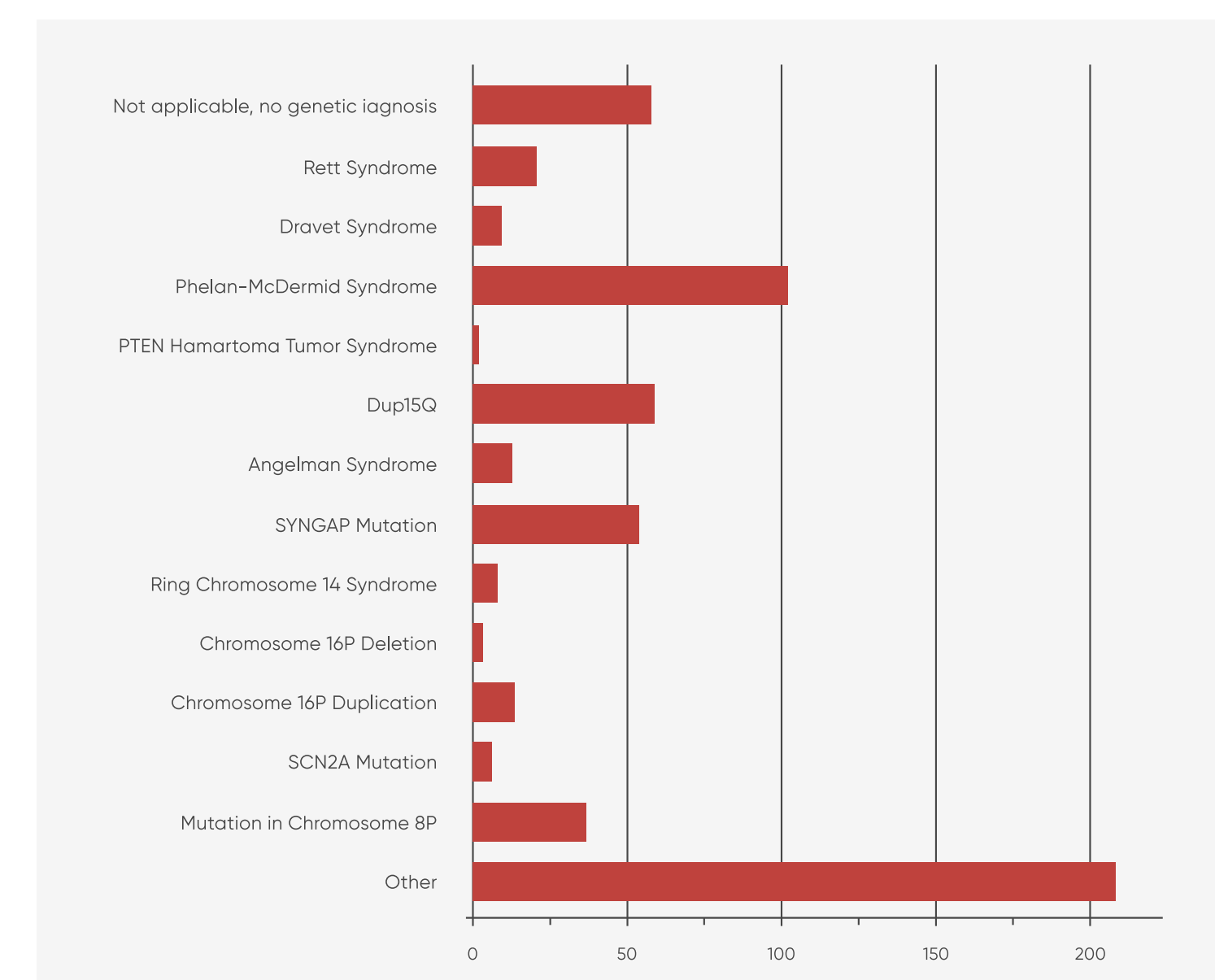
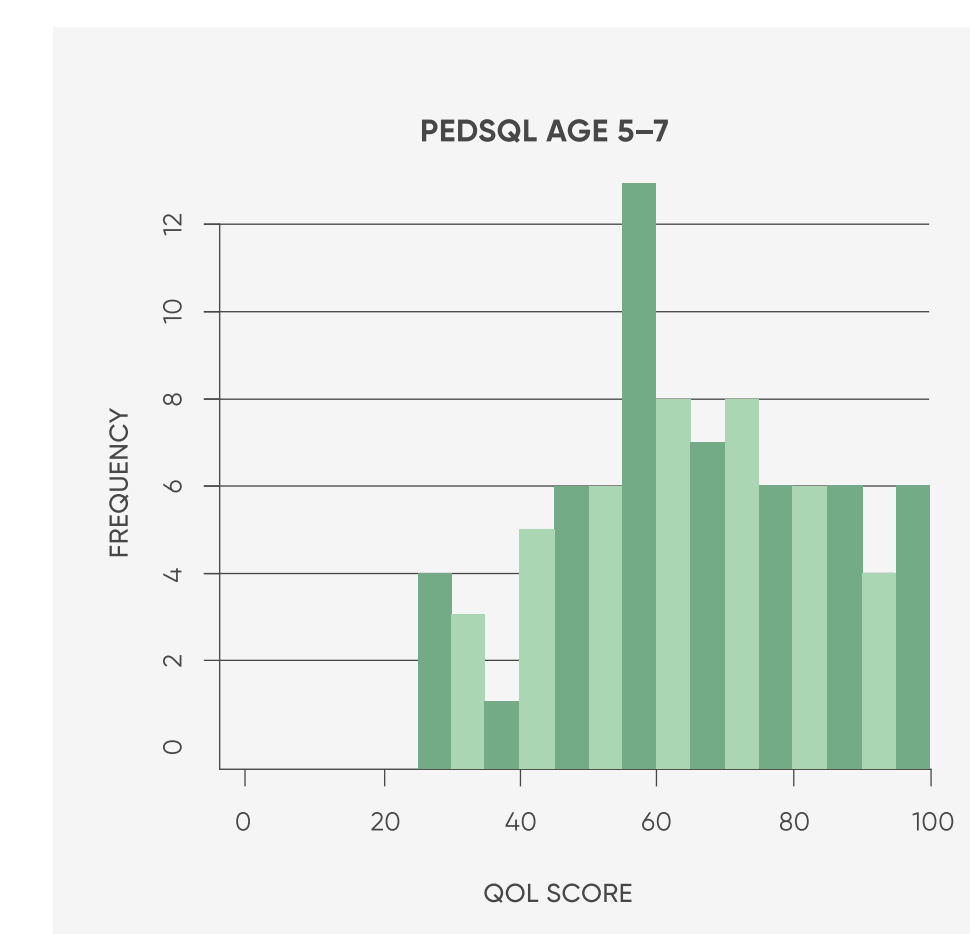
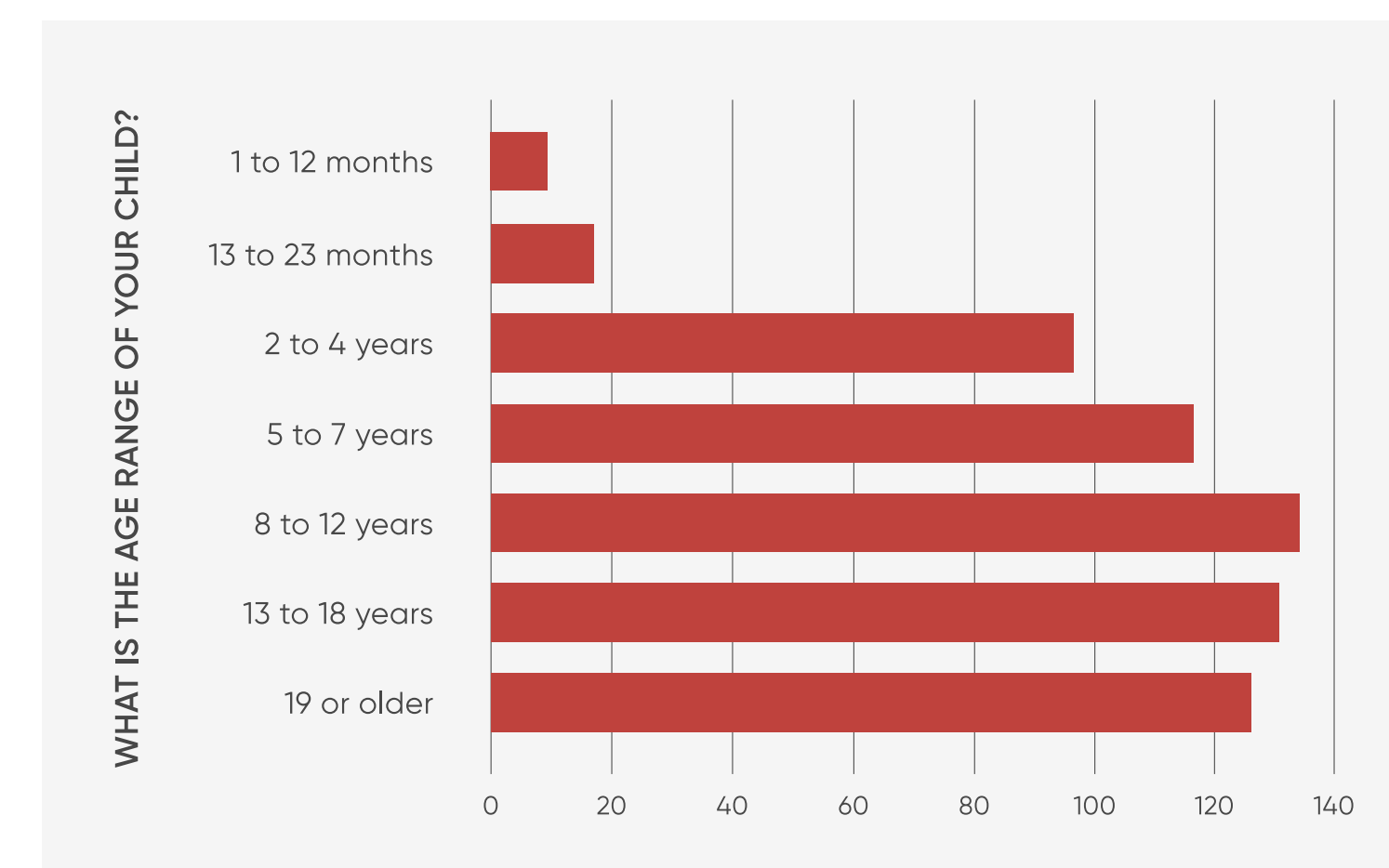
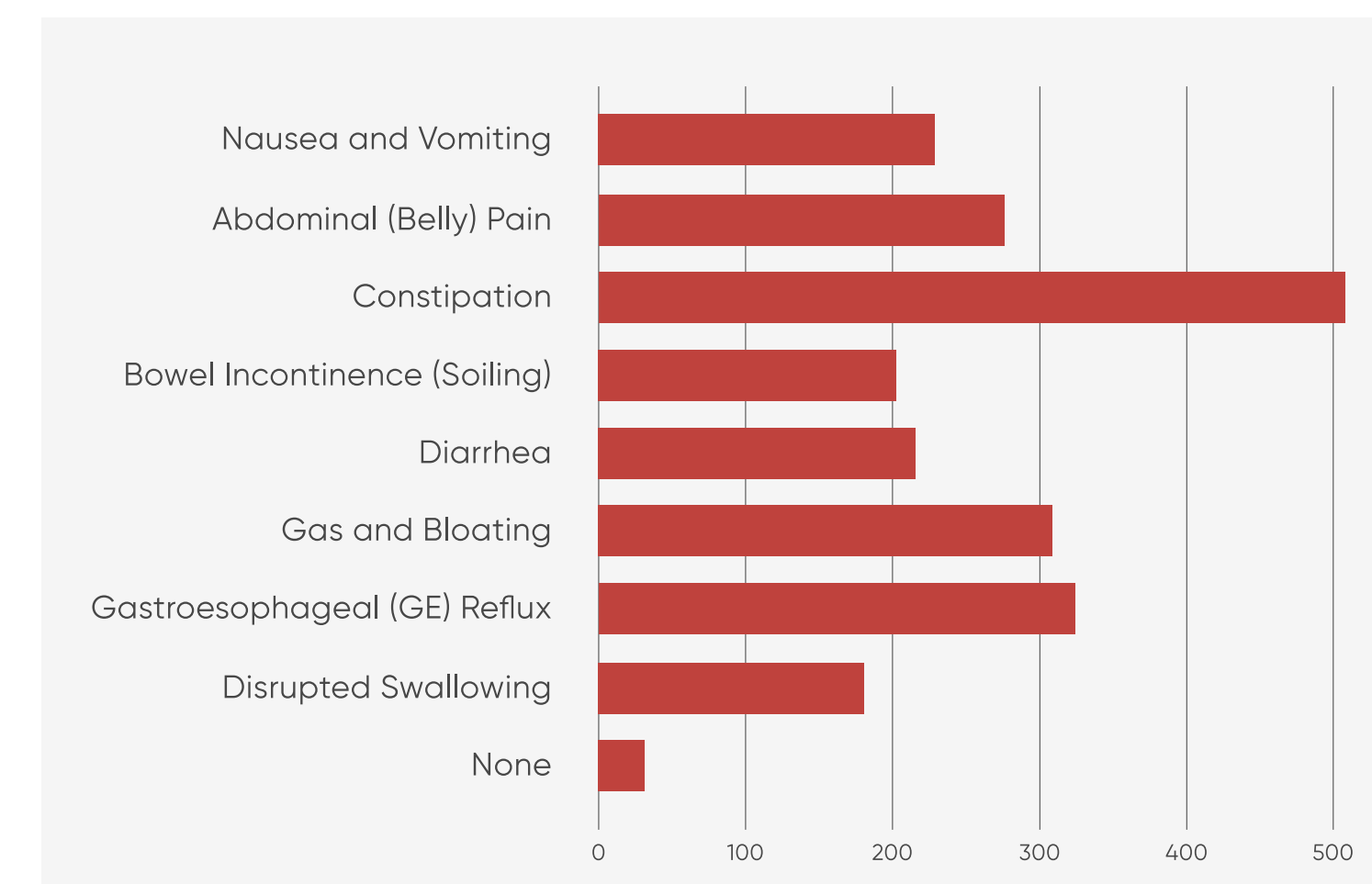
Harnessing the Power of Model Systems and Finding Non-Invasive Ways to Collect Human Data
Dr. Julia Dallman¹⁰

¹Lived experience with GI issues in family members with genetic diagnoses
²Pediatric gastroenterology, hepatology, and nutrition, Indiana University School of Medicine
³Pediatric neurology, Joe DiMaggio Children's Hospital
⁴Pediatric gastroenterology, Nationwide Children's Hospital
⁵Associate director for clinical and translational research and Director for gut-brain science for the NYU Pain Research Center

⁶Pediatric gastroenterology, hepatology, nutrition, Baylor College of Medicine
⁷Pediatric gastroenterology, Indiana University School of Medicine
⁸Gastroenterology, motility and digestive health, Atrium Health Carolinas
⁹Kennedy Krieger Institute
¹⁰Biology, University of Miami

RESULTS/QUANTITATIVE FINDINGS

Constipation was the most common ($p < 0.05$). A statistically significant ($p < 0.05$) decrease in quality of life associated with GI disorders was identified in the adult population compared with other age groups, suggesting that as children age, QoL decreases. Constipation and abdominal pain worsened QOL scores by at least 1 SD.



PARENT RESPONSE TO QOL MEASURES

- "The survey was **incredibly difficult** to complete given the wording of the questions. Our child ...cannot communicate the majority of her issues with us, we only see the results: vomiting, constipation, gas, crying when in pain from GI symptoms, etc. While your survey is inclusive of a variety of neurological disorders, it is not inclusive in its wording given the nonverbal and limited communication statuses of many of the participants."
- "The questions were SO out of reach for me. It makes me feel bad, because **most parents of neurotypical kids probably can answer these questions**. It feels like yet another way individuals with disabilities (and their parents) are **marginalized**. Our experiences are just so out of the norm, that what should be simple questions about GI symptoms doesn't work for us."
- "When talking about issues in the last 7 days, I answered no to almost every question. The issue is that the last 7 days would really be no indication of how prevalent and impactful my son's GI issues are, or the GI issues of any of our kids.."
- "While it has been great to have NDD disorders work together to understand QOL in GI that really impacts our families, our community felt it was **hard to answer the questions since most of our affected individuals are nonverbal** so we did not know the extent or stomach pain/discomfort or specifics to the bowel movements and level of difficulty, etc. Please keep this in mind in the results. I know N/A as an option does not help either but many of the caregivers had to guess..."

DISCUSSION & IMPLICATIONS

Constipation is the highest reported, but not only, GI problem experienced across those with NDDs. In order to better determine the source of GI distress, better subjective and objective measures need to be developed, especially for those who are non-speaking or have limited cognitive ability. Pediatric gastroenterologists need to be better trained to meet the needs of those with medically complex conditions.

- It is essential to involve families in each stage of measure development.
- Measures should be brief, easy, and relevant to prevent drop-out and fatigue, both in the clinic and in research. Any measure needs to account for an individual being non-verbal, or with limited communication.
- Modification of diagnostic criteria to be more inclusive of individuals with NDDs and ASD.
- Development of an extensive toolbox of validated measures for use in GI symptoms in NDDs (including rare diseases) and ASD.
- Enhance postgraduate medical education to improve clinicians' comfort caring for and awareness of GI disorders in NDDs and ASD.

