

Autism Spectrum Disorder in Children with Down Syndrome

Perspectives and Experiences of Healthcare Practitioners and Caregivers in Canada

Julija Atanasova^{1,2}, Rudaina Banihani^{3,4}, Olaf Kraus de Camargo^{5,6,7}, Kerry Boyd⁸, Edyta Marcon¹, Joseph Ferenbok¹, Oksana Hlyva⁷, Jennifer Crowson^{9,10}

¹Translational Research Program, University of Toronto, Toronto, Canada ²The Hospital for Sick Children, Toronto, Canada ³Department of Newborn and Developmental Pediatrics Sunnybrook Hospital, Toronto, Canada ⁴Department of Pediatrics, University of Toronto, Toronto, Canada ⁵Ron Joyce Children's Health Centre, Hamilton, Canada ⁶Department of Pediatrics, McMaster University, Hamilton, Canada ⁷CanChild Centre for Childhood Disability Research, Hamilton, Canada ⁸Department of Psychiatry & Behavioural Neurosciences, McMaster University, Hamilton, Canada ⁹School of Social Work, McMaster University, Hamilton, Canada ¹⁰Canadian Down Syndrome Society

1 INTRODUCTION

While Autism Spectrum Disorder (ASD) in the general population has received extensive attention, its co-occurrence in children with Down Syndrome (DS) remains significantly underexplored. Studies suggest a startling prevalence rate—up to 39%¹ of individuals with Down Syndrome may also have Autism Spectrum Disorder, compared to about 2% in the general population. Despite this high likelihood of co-occurrence, specific challenges related to ASD in children with DS are not sufficiently addressed. This lack of information leads to significant delays in diagnosis, often up to five years after the onset of symptoms² compounded by overlapping symptoms, inadequate screening tools, and limited professional awareness³.

RESEARCH QUESTIONS:

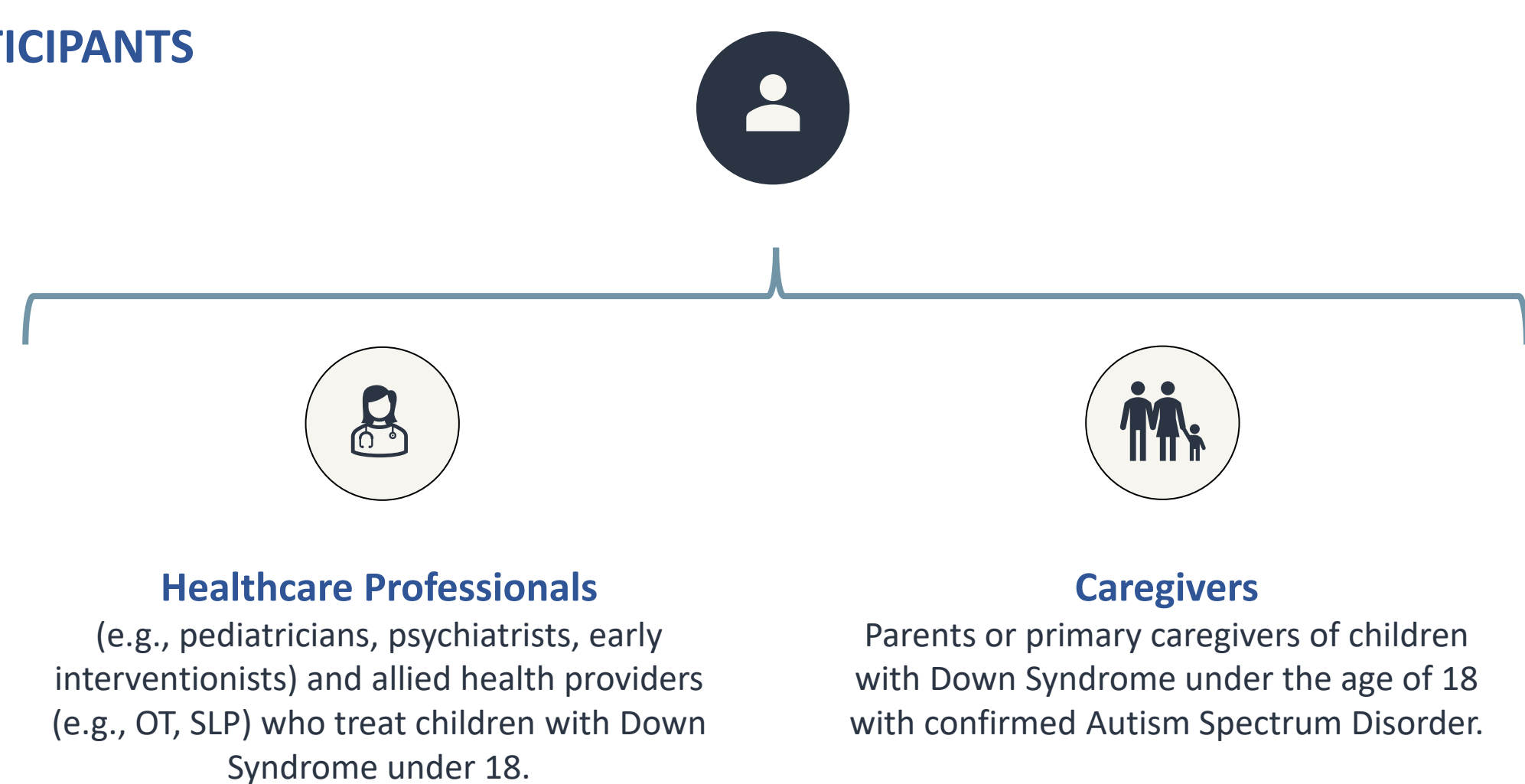
- What are the perspectives of healthcare professionals in Canada regarding the co-occurrence of ASD in children with DS and what barriers they face in identifying ASD?
- What are the diagnostic journeys and challenges experienced by families?

2 METHODS

STUDY DESIGN

This research study employed a **mixed methods design** with a **convergent parallel** approach, where both **quantitative and qualitative data were simultaneously collected and analyzed separately**. Presented data is from February to May 2024.

PARTICIPANTS



DATA COLLECTION METHODS

- Electronic anonymous **surveys** to collect demographic data, awareness, and clinical practices from health professionals; experiences and challenges from caregivers.
- One-on-one **semi-structured interviews** with caregivers only.
- **Procedure:** Participants identified through health associations and Down Syndrome associations across Canada. Surveys distributed electronically through REDCap. Interviews conducted over Zoom.
- **Data analysis:** Quantitative data from surveys were statistically analyzed for trends and correlations. The interviews were transcribed verbatim, coded in NVivo, and analyzed using thematic analysis to identify key themes and insights.

3 RESULTS



Figure A: Awareness Levels of DS-ASD

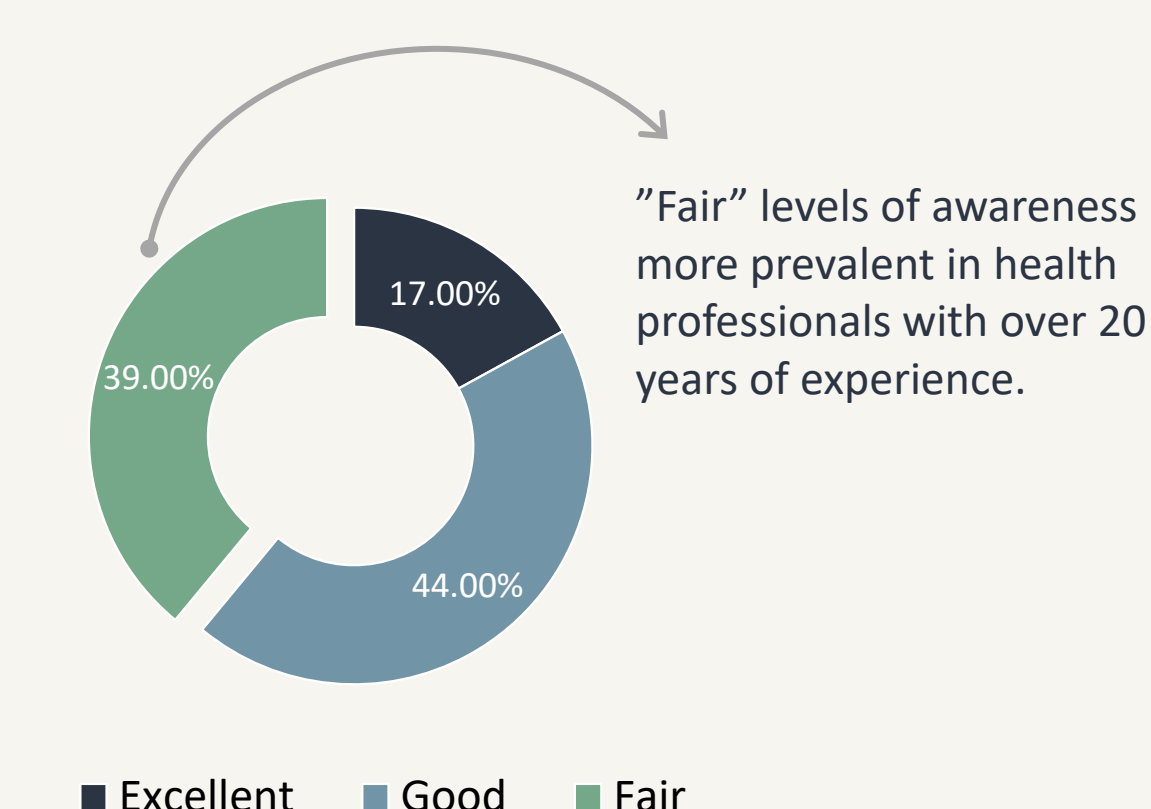
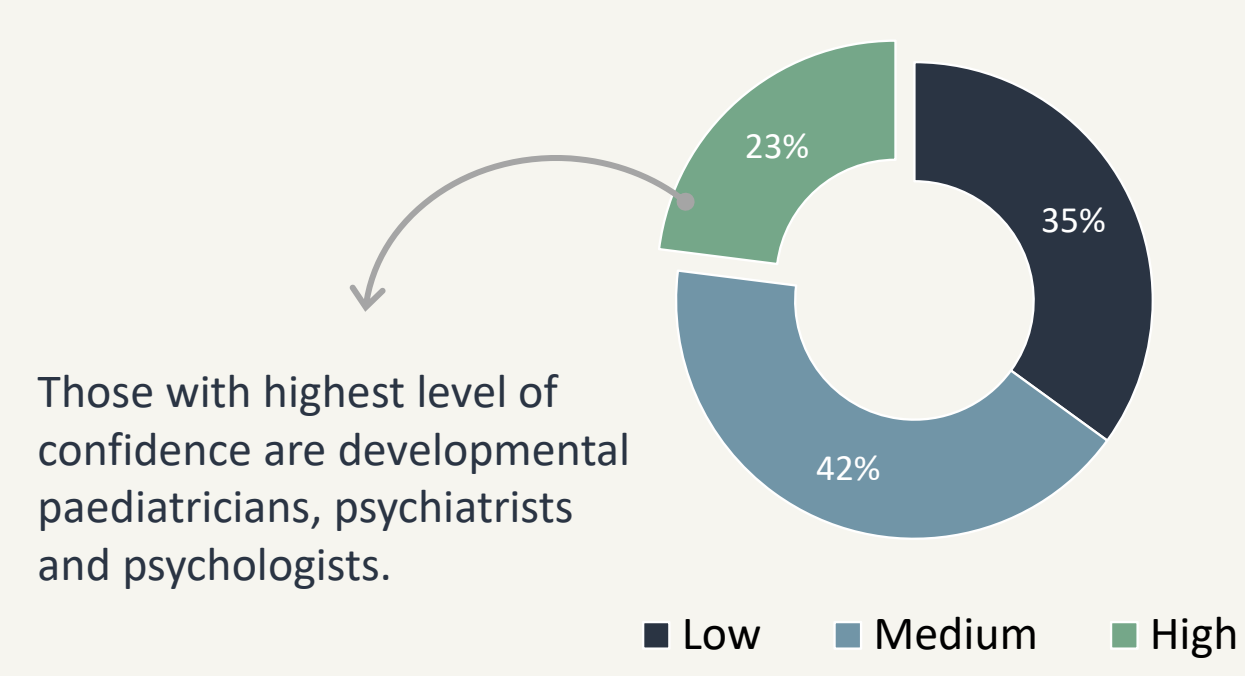


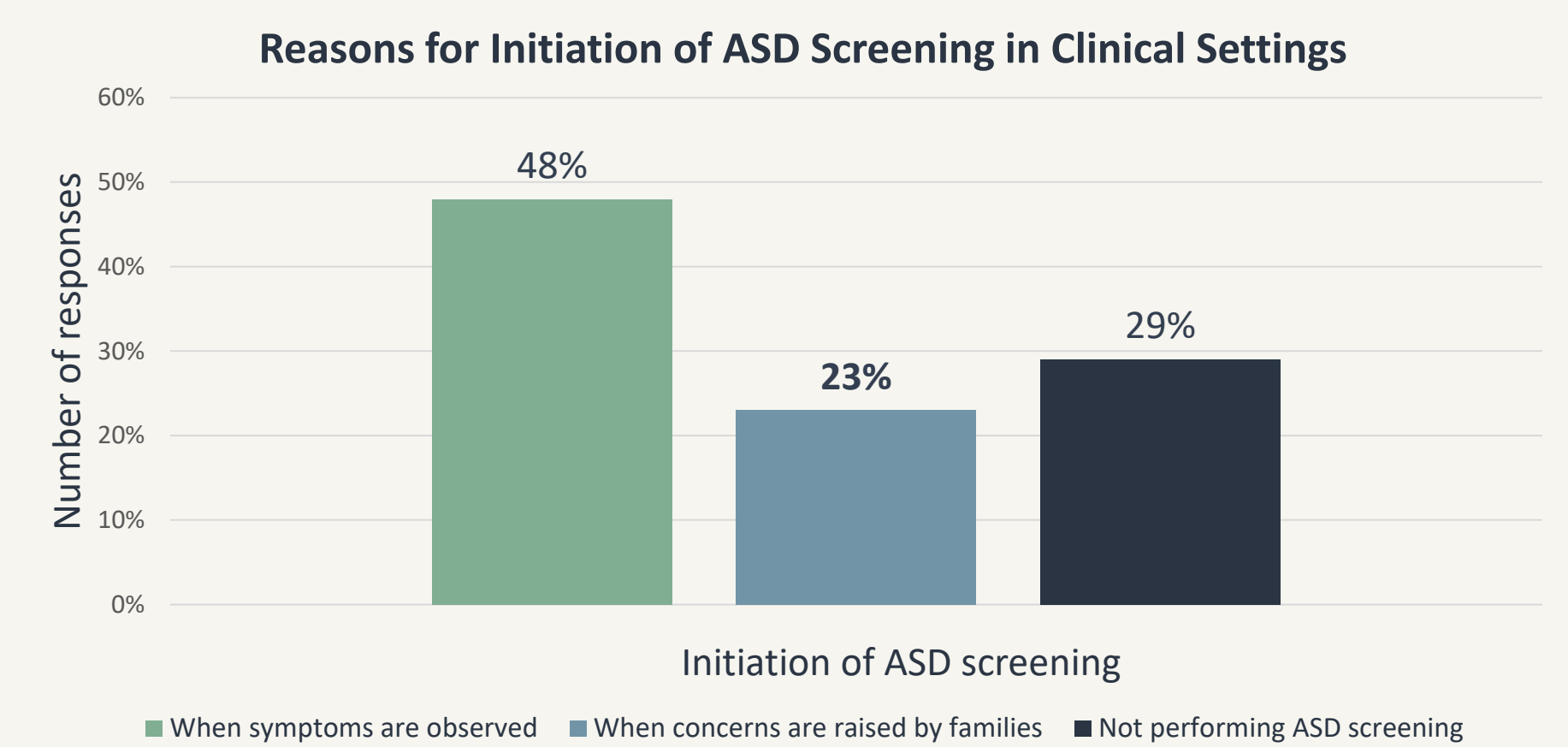
Figure B: Level of confidence to identify ASD in children with DS



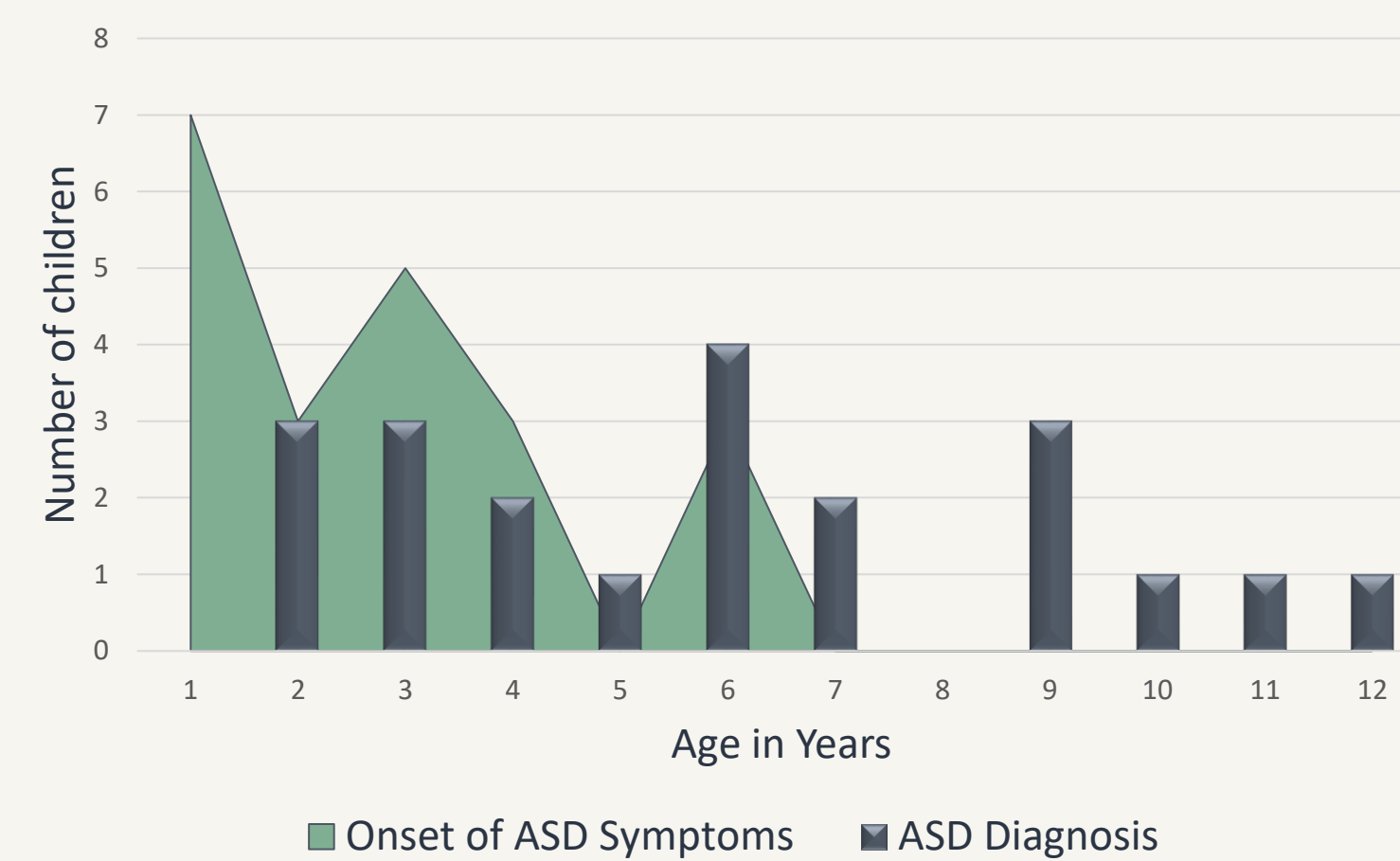
Data is based on 64 survey responses from healthcare professionals across 8 provinces in Canada.

Challenges in early identification of ASD symptoms in children with Down syndrome:

- Overlapping symptoms
- Lack of training
- Limited screening tools



Onset of ASD Symptoms and Age of Diagnosis in Children with Down Syndrome



Data based on 21 survey responses from parents of children with confirmed DS-ASD.

ASD Symptom Onset

- Mean: 2.76 years
- Standard Deviation: 1.73
- Median: 3 years

ASD Diagnosis

- Mean age at diagnosis: 6 years
- Standard Deviation: 3.08
- Median: 6 years

INTERVIEWS – 10 parents of children with dual diagnosis DS-ASD.

To identify common challenges and delays in diagnosis, as well as the impact on families.

QUALITATIVE ANALYSIS – IDENTIFIED THEMES

- 1 **Awareness Gaps** – Notable lack of awareness among caregivers about the possibility of co-occurring ASD in children with DS
- 2 **Diagnostic Challenges** – Caregivers frequently encountered delays or misdiagnoses, often due to unrecognized ASD signs.
- 3 **Healthcare Navigation Barriers** – Difficulties in navigating the health system, getting accurate information and accessing appropriate care.

Early observed ASD signs in children with DS:

- Repetitive behavior (21, 100%)
- Speech delay/regression (19, 90.5%)
- Unusual reactions to sounds, textures, or visual stimuli (13, 62%)

Prevalence of neurodevelopmental conditions in family histories:

38%	• ADHD
33%	• ASD
29%	• Learning Disabilities

4 DISCUSSION

COMPARISON WITH PRIOR STUDIES

This study confirms the challenges of delayed diagnosis and limited professional awareness seen in other research on co-occurring ASD in children with Down Syndrome. It distinctly reveals persistent gaps in Canada's healthcare approach, emphasizing the need for more proactive screening protocols compared to other countries.

STUDY LIMITATIONS

Include a small sample size and potential selection bias, favoring more vocal participants. The reliance on retrospective accounts may also introduce recall bias.

FUTURE DIRECTIONS

This study highlights the need for tailored ASD screening tools for children with DS, educational initiatives for health professionals in Canada. Future studies should aim for larger, diverse samples and use prospective methodologies to address these issues. Data collection until 10/2024: <https://redcap.utoronto.ca/surveys/?s=8FNAXMTTH3TP4LEX>

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CONTACT DETAILS

Julija Atanasova
Email: julija.Atanasova@mail.utoronto.ca

