**BACKGROUND**

- Down syndrome (DS) is known to have higher rates of autism spectrum disorder (ASD) than in the general population, with prevalence estimates around 16% vs. 1.7%.
- Individuals with DS + ASD receive their ASD diagnosis considerably later than ASD-only children: 14.4 years old vs. 5.04 years old.
- For those individuals with DS + ASD, research is ongoing about the distinct medical and behavioral profile, as differentiated from those with DS or ASD alone.
- The medical comorbidities of each separate population are well-characterized. However, we know little about whether there is also a distinct profile of co-occurring medical conditions in the DS + ASD population, or if their medical complexity affects the presentation and timing of their ASD diagnosis.

**OBJECTIVES**

1. Compare early medical risk factors among the DS-only, ASD-only, and DS + ASD populations.
2. Identify common presenting concerns prior to DS + ASD diagnosis.
3. Characterize the profile of co-occurring medical conditions in DS-only, ASD-only, and DS + ASD pediatric populations.

**METHODS**

- **Study Design:** Retrospective chart review of DS + ASD cases from the Sie Center for Down Syndrome at Children’s Hospital Colorado.
- **Inclusion Criteria:** Trisomy 21 patients who were diagnosed with ASD by the Sie Center between 2014 and 2020.
- **Exclusion Criteria:** Individuals over the age of 18 years at time of evaluation; individuals with translocation or mosaic DS.
- **Data Abstracted into REDCap:**
  - **Demographic information**
  - **Birth history**
  - **Medical comorbidities**
  - **ASD evaluation results**
  - **Initial behavioral concerns for autism**
  - **Age of child at ASD diagnosis**
  - **Time interval between first concerns and ASD diagnosis**

**RESULTS**

- **Table 1: Sample Characteristics**
  - **Group**
    - Down Syndrome (DS)
    - Autism Spectrum Disorder (ASD)
    - Down Syndrome + Autism Spectrum Disorder (DS+ASD)
  - **n**
    - 41
    - 41
    - 41
  - **Males (%)**
    - 68.3
    - 67.7
    - 65.9
  - **Hispanic (%)**
    - 22
    - 25.0
    - 24.4
  - **Medicaid (%)**
    - 35.8
    - 25.0
    - 22
  - **Family History of ASD (%)**
    - 51.2
    - 25.0
    - 43.9
  - **Family History of DS (%)**
    - 67.7 (SD 10.5)
  - **Tools (mean)**
    - Vineland AD/ASD Standard Score
      - 68.3 (SD 5.36)
      - 65.9 (SD 7.80)
      - 63.4 (SD 7.34)

- **Table 2: Presenting Concerns in ASD**
  - **Presenting Concerns**
    - Decrease in social-emotional reciprocity/social withdrawal
    - Deficits in non-verbal communicative behaviors
    - Deficits in developing, maintaining, and understanding routines
    - Stereotypy or repetitive motor movements, use of objects, or speech
    - Irritability or tantrums, inflexible adherence to routines or ritualized patterns of verbal or nonverbal behavior
    - Highly restricted, fixated interests that are abnormal in intensity or focus
    - Hyper or hypo-reactivity to sensory input or unusual interests in sensory aspects of the environment
  - **ASD Diagnosis (%)**
    - 51.2
    - 22
    - 49.2
  - **Group**
    - DS
    - ASD
    - DS+ASD
  - **p-value**
    - 0.006*
    - 0.137
    - 0.806

**CONCLUSIONS**

- Among the early medical risk factors studied, there was a significant association between diagnosis type and prematurity, with DS + ASD patients most likely to be born premature. Although there was an increased frequency of infantile spasms in the DS + ASD cohort, the association was not statistically significant.
- Children with ASD-only were significantly more likely to present with difficulties understanding relationships, repetitive behaviors, behavioral rigidity, and sensory differences than those with DS + ASD.

**NEXT STEPS**

- Complete the profile of co-occurring medical conditions: Perform regression analyses to assess for associations between comorbidities and diagnosis type.
- Determine if there is a reliable set of variables that predicts each diagnostic category.

**REFERENCES**