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# Congenital Heart Defects and Autism: Understanding the Breakdown of Associated Risk Factors In A Clinically Referred Sample

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## Background

- Children with Congenital Heart Defects (CHD) have **higher odds** of developing social difficulties and/or an Autism Spectrum Disorder (AuSD) than the general population (i.e., ~10% vs. ~1%).[1,2,3,4,5]
- CHD associated risk factors (e.g., genetic disorders, hypoxia/anoxia, neurological conditions) are associated with a higher incidence of AuSD, however, the drivers of these associations are not yet understood. [6,7,8]
- **Purpose:** The purpose of this study is to identify the rates of co-occurring cardiac, neurological, and genetic conditions to better understand associated risk factors in a patient sample from a medium-size children's hospital.

## Method

### Inclusion Criteria:

- Diagnosed with CHD < 18 years old
- High neurodevelopmental risk (AHA and AAP Position Statement, 2012)
- Cardiac Neurodevelopmental (CND) program neuropsychological evaluation (01/01/21 – 12/31/22)

### Participants:

- N = 356 (N=50 with autism)
- Gender: 57% male; 43% female
- Age: M = 5.29, SD = 4.30
- Race/Ethnicity: 77% White, 6% Black, 3% Asian, 4% Biracial, 2% Hispanic, 6% Other, 1% Missing

### Method:

- Patients attended regular CND follow-up clinic visits.
- Positive AuSD screenings further categorized by diagnosis, awaiting assessment, or evaluated with no AuSD dx (fig 1).
  - **35** patients were diagnosed with AuSD
  - **10** were awaiting assessment
  - **4** were evaluated and did not meet AuSD criteria
- Patients with AuSD or a pending evaluation were further characterized by the presence of comorbid conditions (Table 1; Figure 2).

## Results

**Figure 1: Percentage of Autism diagnosis in Total Sample**

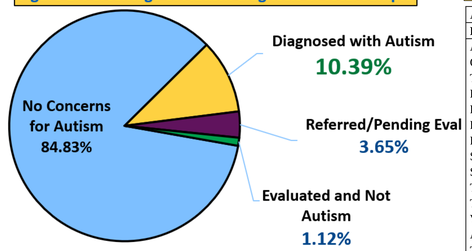
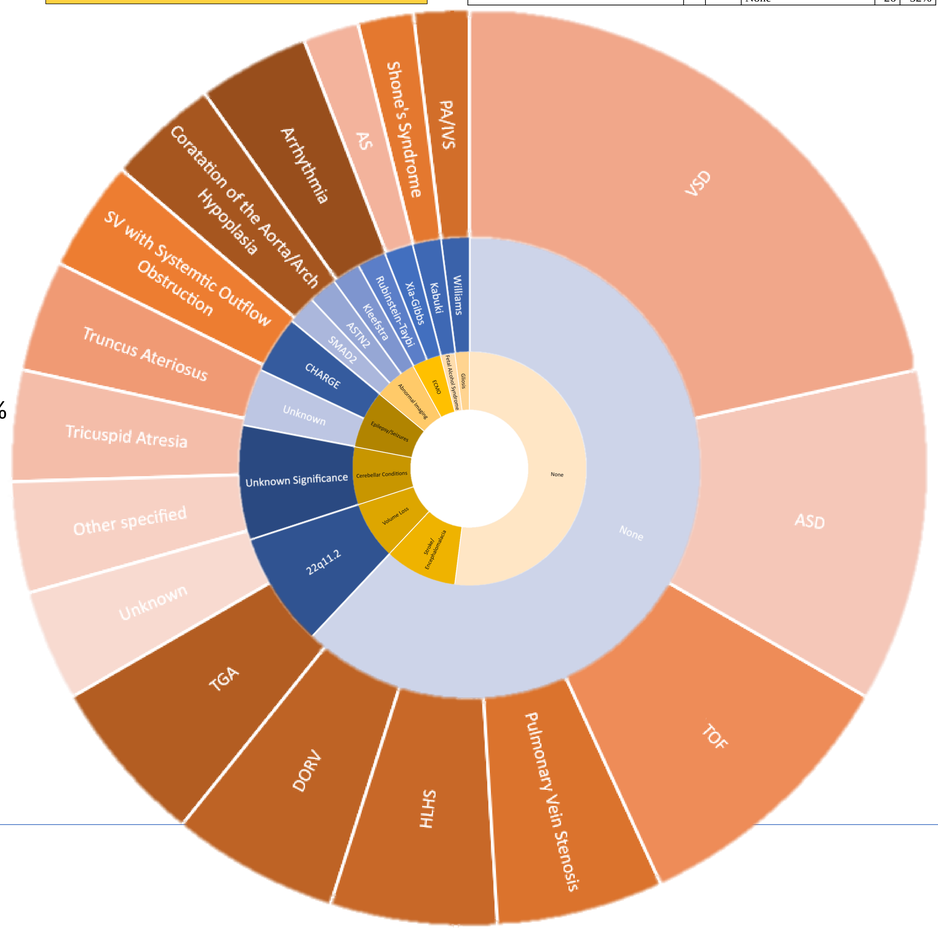



**Table 1: CHD type and comorbid genetic/neurological conditions among AuSD subsample**

| Autism Subsample (N=50) | # | % | | # | % |
|---|---|---|---|---|---|
| **Primary Cardiac Diagnosis** | | | **Genetic Condition** | | |
| Arrhythmia | 2 | 4% | Unknown Significance | 4 | 8% |
| Coarctation of the Aorta/Arch Hypoplasia | 2 | 4% | 22q11.2 | 4 | 8% |
| TGA | 3 | 6% | CHARGE | 2 | 4% |
| DORV | 3 | 6% | Williams | 1 | 2% |
| HLHS | 3 | 6% | Kabuki | 1 | 2% |
| PA/IVS | 1 | 2% | Xia-Gibbs | 1 | 2% |
| Pulmonary Vein Stenosis | 3 | 6% | Rubinstein-Taybi | 1 | 2% |
| Shone's Syndrome | 1 | 2% | Kleefstra | 1 | 2% |
| SV with Systemic Outflow Obstruction | 2 | 4% | ASTN2 | 1 | 2% |
| TOF | 5 | 10% | SMAD2 | 1 | 2% |
| Truncus Arteriosus | 2 | 4% | Unknown | 2 | 4% |
| VSD | 11 | 22% | None | 31 | 62% |
| AS | 1 | 2% | | | |
| Tricuspid Atresia | 2 | 4% | **Neurological Condition** | | |
| ASD | 6 | 12% | Epilepsy/Seizures | 4 | 8% |
| Other specified | 2 | 4% | Cerebellar Conditions | 4 | 8% |
| Unknown | 2 | 4% | Volume Loss | 4 | 8% |
| | | | Stroke/Encephalomalacia | 5 | 10% |
| **Oxygenation Status** | | | ECMO | 2 | 4% |
| Cyanotic | 20 | 40% | Abnormal Imaging | 3 | 6% |
| Acyanotic | 27 | 54% | Gliosis | 1 | 2% |
| Missing | 2 | 4% | Fetal Alcohol Syndrome | 1 | 2% |
| | | | None | 26 | 52% |

**Figure 2: CHD type (orange), genetic (blue), and neurological conditions (yellow) among AuSD subsample**




## Conclusion

- High prevalence of diagnosed AuSD in this sample compared to population data (i.e., ~10% vs. ~1%). [9]
- Diagnosis of AuSD was higher among those with acyanotic CHD.
- Children with CHD and AuSD had high prevalence of comorbid genetic and neurological conditions.

***Cardiac neurodevelopmental providers should include regular autism screening in clinics to ensure appropriate access to evaluation, treatment, and family support.***

- Among CHD diagnoses, VSD and autism were most often cooccurring (22%), followed by TOF (10%).
- Among genetic conditions, 22q11.2 (8%) and variants of unknown significance (8%) and autism were most often cooccurring.
- Among neurological conditions, Stroke/Encephalomalacia (10%) and autism were most commonly cooccurring, followed by epilepsy/seizures (8%), cerebellar conditions (8%), and volume loss (8%).
- Interestingly, most children with autism and CHD did NOT have a co-morbid neurological condition, which may suggest that neurological conditions/injuries do not drive the increased association between CHD and autism.
- Next Steps:
  - Further examine risk factors associated with an AuSD diagnosis in CHD population.
    - Examine patterns of associations between genetic and/or neurologic conditions in relation to AuSD within the CHD population.
  - It is imperative that providers include regular AuSD screening in clinics to ensure appropriate access to evaluation, treatment, and family support.

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