

Cognition and language in Down syndrome: The role of congenital heart disease and the challenge of testing floor effects in the Pediatric Heart Network CHILD-DS study

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Background

- ~50% of children with Down syndrome (DS) have congenital heart disease (CHD), but its impact on neurodevelopmental (ND) outcomes remains unclear.
- In children without DS, CHD surgery has been associated with impairments in all ND areas.
- Prior findings on CHD's developmental impact in children with DS are inconclusive.
- In the NIH INCLUDE-funded **Congenital Heart Disease: Impact on Learning and Development in Down syndrome (CHILD-DS)** study, main results demonstrated no difference in cognitive scores by CHD status, but floor effects limited comparisons. Adaptive skills differences emerged, including in language.

Objective

To further investigate cognition and language outcomes in children with DS with complete atrioventricular septal defect (CAVSD, most common CHD in DS) versus no major CHD.

Study sample and methods

- Setting:** 13 PHN clinical sites in the U.S. and 1 in Canada, May 2022-Oct 2023
- Participants:** 259 children 5-12 years with DS (124 CAVSD repair and 135 non-CHD).
 - Study groups were similar in age and sex, with some differences in race and ethnicity.
 - The CAVSD group had lower median neighborhood SES (Table 1, $P=0.01$).
- Outcomes:**
 - Cognitive skills from the **Stanford Binet Intelligence Scales, 5th Ed. (SB-5)** were assessed using **deviation index scores** (Sansone SM et al. 2014), transformed into within-cohort z-scores to reduce floor effects and allow cross-measure comparison.
 - Receptive language on **Peabody Picture Vocabulary Test, 5th Edition (PPVT-5)** and Communication subdomain of **Vineland Adaptive Behavior Scales, 3rd Edition**
 - Expressive language on **Expressive Vocabulary Test, Third Edition (EVT-3)** and Communication subdomain of **Vineland**
 - Adaptive skills from **Vineland**
- Statistical analysis:** Group differences were evaluated using standard parametric and nonparametric tests for categorical and continuous variables. Multivariable regression models adjusted for *a priori* socio-demographic and medical history variables. Loess plots, by study group, show relationship of deviation index scores with age at testing.

Table 1. Sample Characteristics

Median (Q1, Q3) or n (%)	Overall (n=259)	CAVSD Repair Group (n=124)	Non-CHD Group (n=135)	P
Age at consent (years)	7.7 (6.5, 9.2)	7.6 (6.5, 10.3)	7.6 (6.5, 8.7)	0.38
Male	128 (49%)	63 (51%)	65 (48%)	0.67
Race				0.09
Asian	9 (3%)	3 (2%)	6 (4%)	
Black/African American	28 (11%)	19 (15%)	9 (7%)	
White/Caucasian	187 (72%)	87 (70%)	100 (74%)	
Other or multiracial	31 (12%)	12 (10%)	19 (14%)	
Ethnicity				0.05
Hispanic or Latino/Latina	37 (14%)	12 (10%)	25 (19%)	
Not Hispanic or Latino/Latina	217 (84%)	109 (88%)	108 (80%)	
Socioeconomic status Index*	0.1 (-0.5, 0.5)	-0.1 (-0.7, 0.4)	0.2 (-0.4, 0.6)	0.01

*Neighborhood SES was calculated using a modified Winkleby/Cubbin index.

Results

Cognition

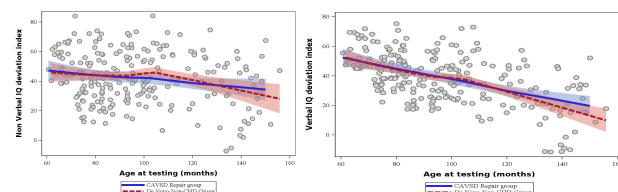
- Deviation index scores enabled cognitive IQ data on all but one child: 1/3 of children scored at SB-5 testing floor, equally distributed between groups.
- No group differences were seen for nonverbal or verbal deviation index scores (Table 2), or domain-specific deviation index scores.

Table 2. Stanford Binet-5 Cognitive Outcomes by Study Group

Median (Q1, Q3), Mean $\pm$ SD, or n (%)	Overall (n=236*)	CAVSD Repair Group (n=112)	Non-CHD Group (n=124)	P
Non-verbal IQ deviation score	42.3 $\pm$ 17.4	41.5 $\pm$ 17.8	43.0 $\pm$ 17.0	0.51
Verbal IQ deviation score	38.8 (25.8, 50.3)	39.1 (25.8, 51.0)	38.4 (26.2, 47.9)	0.87

*n provided is for children with data deemed valid by the neurodevelopmental evaluator.

- Deviation index scores by age at testing demonstrate widening gap between children with DS and age-based norms over time as cognitive development progresses more slowly among children with DS (see Figures).
- Nonverbal deviation index score shows less decline than verbal, suggesting stronger developmental progress in nonverbal abilities.



Figures. Nonverbal and Verbal Deviation Index Scores by Age at Testing

Receptive and Expressive Language, Adaptive Skills

- CAVSD group was ~half as likely to pass EVT-3 teaching items (i.e., "Verbally label 2 photos of common objects/items," Table 3), with no difference for PPVT-5.
- Children with CAVSD had lower scores in Adaptive Behavior Composite, communication, and expressive communication adjusting for socio-demographic and medical factors (Table 4).

Table 3. Receptive and Expressive Language in Direct Child Testing

Median (Q1, Q3), Mean $\pm$ SD, or n (%)	Overall (n=236*)	CAVSD Repair Group (n=112)	Non-CHD Group (n=124)	P
Peabody Picture Vocabulary Test, 5th Edition				
Did not pass teaching items	25 (11%)	16 (14%)	9 (7%)	0.10
Standard Score	51.0 (40.0, 61.0)	48.5 (40.0, 61.0)	52.0 (40.0, 60.0)	0.61
Expressive Vocabulary Test, 3rd Edition				
Did not pass teaching items	38 (17%)	24 (22%)	14 (12%)	0.03
Standard Score	63.0 (46.0, 71.0)	63.0 (40.0, 70.0)	62.0 (51.0, 72.0)	0.49

*n provided is for children with data deemed valid by the neurodevelopmental evaluator.

References

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Winkleby MA and Cubbin C. Influence of individual and neighbourhood socioeconomic status on mortality among black, Mexican-American, and white women and men in the United States. *J Epidemiol Community Health* 2003;57:444-52.

Receptive and Expressive Language, Adaptive Skills, Continued

- Higher expressive communication scores were also associated with higher SES index in multivariable analysis (0.9 ± 0.4 , $P=0.03$, data not shown).
- Study group and *a priori* sociodemographic and medical factors explained 16% of the variance in communication and 21% in the expressive subdomain.

Table 4. Adaptive Behavior and Communication in Children with Down syndrome in CAVSD Repair vs. Non-CHD Groups (Vineland, n=233*)

	β (SE)**	P	R ²
Adaptive Behavior Composite	-3.8 (1.7)	0.03	0.16
Communication Domain	-5.3 (2.4)	0.03	0.16
Receptive Subdomain	-1.0 (0.6)	0.08	0.14
Expressive Subdomain	-1.1 (0.5)	0.04	0.21

*n provided is for children with data deemed valid by the neurodevelopmental evaluator.

**Multivariable regression results, adjusting for *a priori* covariates. Group effect and the corresponding p-value are presented from each model, along with overall R-square.

Limitations/future directions

- Over 30% of participants scored at the SB-5 floor and over 20% of children with CAVSD did not pass EVT-3 teaching items, highlighting limitations of standard ND measures in children with DS.
- Both CAVSD group and lower SES were associated with lower expressive language scores, emphasizing the need for further research on social drivers of health in children with DS.
- Need to target interventions to support development of vulnerable adaptive skills, such as expressive communication, in children with DS and CAVSD.

Conclusions

- School-age children with DS and CAVSD had lower expressive language scores but similar cognitive scores compared with DS peers without major CHD after adjustment for floor effects.
- Traditional cognitive measures using standard scores demonstrated substantial floor effects, supporting the use of alternative assessment approaches (deviation scores) and greater emphasis on functional outcomes relevant to long-term quality of life in individuals with DS.
- Findings highlight the need for ND measures that better capture meaningful functional outcomes in children with DS, including communication, adaptive skills, and community participation.

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