Echocardiographic Cardiac Size & Function in Adults with Down syndrome

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Introduction

Adults with Down syndrome (Ds) are living longer, revealing unique cardiac characteristics that go beyond congenital anomalies. Preliminary evidence points to smaller left-side heart chambers, hyperdynamic systolic function, altered diastolic function, and potential compensatory mechanisms to maintain cardiac output. However, baseline data in this population remain scarce. Without baseline descriptive reports of the cardiac size and function in the hearts of adults with Ds, the scientific and medical community are disadvantaged in understanding this population's cardiovascular function.

Methods

- **Design & Participants:** Cross-sectional, observational comparison of 18–35-year-old adults with Ds (diagnosed trisomy 21) and controls matched for age and physical activity.
- **Exclusion Criteria:** Unrepaired congenital heart defects, significant cardiometabolic disease, or medications affecting cardiovascular function.
- **Procedures:** All participants underwent a comprehensive echocardiogram following standard guidelines. Data were indexed to body size and analyzed with appropriate parametric/nonparametric tests. The effect size was calculated using Cohen's d.

Aims

Aim 1: Cardiac Size & Systolic Function
Compare left ventricular dimensions, volumes, and systolic indices (e.g., stroke volume, ejection fraction, fractional shortening) in adults with Ds versus age- and activity-matched controls.

Aim 2: Diastolic Function
Evaluate diastolic parameters (e.g., left atrial volume indexed to body surface area, Tissue Doppler Imaging myocardial velocity Em, and mitral inflow velocity eV) to determine whether diastolic filling differs between adults with and without Ds.

Aim 3: Ventricular-Arterial Coupling
Assess heart–arterial system interactions (e.g., pre-ejection period & ventricular-arterial coupling) to clarify whether unique coupling adaptations exist in Ds.

Adults with Down syndrome demonstrated a unique cardiac phenotype compared to adults without Down syndrome

Parameter	Ds (n)	Ds Mean ± SD	Non-Ds (n)	Non-Ds Mean ± SD	p-value	Cohen's D Effect size
Aim 1						
Left Ventricular Internal Dimension Diastole (mm)	36	40.9 ± 4.8	20	43.1 ± 4.2	0.038*	N/A
Left Ventricular Internal Dimension Systole (mm)	36	25.3 ± 4.4	20	29.1 ± 3.7	0.002*	N/A
Left Ventricular End-Systolic Volume Indexed to Body Surface Area (mL/m^2)	29	10.2 ± 3.2	19	13.6 ± 6.8	0.032*	N/A
Stroke Volume Indexed to Body Surface Area (mL/m^2)	35	27.5 ± 6.8	20	31.6 ± 4.8	0.024*	0.651
Left Ventricular Ejection Fraction	28	0.66 ± 0.05	18	0.60 ± 0.04	<0.001*	-1.27
Fractional Shortening	36	35.8 ± 5.9	20	31.2 ± 4.5	<0.004*	-0.848
Aim 2						
Left Atria Volume Indexed to Body Surface Area	30	5.40 ± 3.0	18	7.75 ± 4.5	0.049*	N/A
Tissue Doppler Image Lateral Myocardial Velocity Em (cm/s)	29	14.01 ± 3.0	18	17.08 ± 3.0	0.002*	1.002
Mitral Inflow Velocity eV (cm/s)	29	98.24 ± 18.4	18	84.27 ± 11.9	0.006*	-0.759
Aim 3						
Pre-Ejection Time (ms)	28	100.43 ± 13.4	18	115.33 ± 16.8	0.002*	1.01
Ventricular-Arterial Coupling	28	0.92 ± 0.1	18	1.06 ± 0.1	0.002*	0.969

Results

- Adults with Ds had smaller cardiac dimensions and volumes, even when indexed to body surface area
- Adults with Ds demonstrated systolic hyperfunction with higher ejection fraction and fractional shortening
- Adults with Ds demonstrated altered diastolic mechanics, with an increased mitral inflow velocity (eV) and lower myocardial velocity (Em)
- Adults with Ds showed a different ventricular-arterial coupling profile, with a shorter pre-ejection time and lower ventricular-arterial coupling ratio

Conclusion and Discussion

These findings suggest adults with Ds demonstrated physiological cardiac adaptations that supported adequate cardiac output at rest.

Adaptive but Limited Reserve:
Smaller ventricular volumes in Ds seem offset by hyperdynamic systolic function, potentially limiting cardiovascular reserve under stress.

Diastolic Function:
Smaller left atria volume, reduced diastolic relaxation, and increased mitral flow velocity suggest a mixed diastolic and left ventricular function profile.

Ventricular-Arterial Coupling:
Shorter, faster pre-ejection time and a lower ventricular-arterial coupling ratio suggest that adults with Ds hearts spend less time in systole, potentially increasing the workload of the heart.

Future Investigations & Interventions:
Longitudinal studies using 3D echocardiography, strain analysis, and exercise interventions are critical to identifying how these adaptations evolve, informing tailored strategies for improving cardiac health in individuals with Ds.

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