

**INFLUENCE OF GENETIC FACTORS ON LEFT VENTRICULAR  
STRUCTURE AND FUNCTION IN FRIEDREICH'S ATAXIA**

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 **CARDIAC FUNCTION AND HEART FAILURE**

**INFLUENCE OF GENETIC FACTORS ON LEFT VENTRICULAR STRUCTURE AND FUNCTION IN FRIEDREICH'S ATAXIA**

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**Background:** In Friedreich ataxia (FA), the proportion of patients that develop cardiomyopathy (EF<40%) and the factors underlying its severity are unknown. Understanding these issues is complicated by the small size of most cohorts, and the variability of measures among different labs. This study examines the relationship between clinical and genetic features and echocardiographic parameters in FA.

**Methods:** Original echocardiograms from 142 patients with confirmed FA were analyzed at a single institution for ejection fraction (EF), percent change in LV internal diameter (%LVD), relative wall thickness in diastole (RWTd), and the tissue Doppler imaging E/A velocity ratio in the lateral wall (TDI E/A). Factors altering these parameters were assessed using summary statistics, linear and logistic regression analysis.

**Results:** In cross sectional analysis, mean EF was  $56.0 \pm 6.7\%$ , mean %LVD was  $32.9 \pm 8.4\%$ , mean RWTd was  $0.53 \pm 0.15$ , and mean TDI E/A was  $2.05 \pm 0.68$ . The mean RWTd and TDI E/A values are outside the normal ranges for the lab. 1.6% of patients had cardiomyopathy (EF<40%) and 70.9% of patients had left ventricular hypertrophy (LVH) (RWTd>0.45).

Disease-related factors correlated with specific echo parameters. By linear regression, age of onset ( $p=0.04$ ) and age ( $p=0.001$ ) were significant predictors of %LVD after adjusting for sex. RWTd correlated with sex ( $p=0.01$ ) and genetic severity of FA (assessed by shorter GAA repeat length) ( $p=0.06$ ) in a model adjusting for age. TDI E/A correlated inversely with GAA repeat length ( $p=0.03$ ) and age ( $p=0.01$ ) after adjusting for sex. Using logistic regression, an abnormal EF (<50%) was predicted by age ( $p=0.02$ ), but not by GAA repeat length or sex.

**Conclusions:** Echocardiography demonstrates the frequency of LVH in FA and variability of LV systolic and diastolic dysfunction with a minority of patients in this cohort having cardiomyopathy. Markers of systolic and diastolic dysfunction were frequently associated with longer GAA repeat lengths and older age. Longitudinal analysis and association of echocardiographic parameters with specific outcomes such as mortality can be used in this large cohort to define LV disease progression in FA.

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