Clinical characteristics and outcomes of cardiac disease in Barth Syndrome: the 10 year UK experience

Kang S-L., Forsey J., Walsh M., Dudley D., Tsai-Goodman B.
Bristol Royal Hospital for Children, Bristol, United Kingdom

Objective: Barth syndrome (BTHS) is an X-linked disorder characterized by cardiomyopathy, neutropenia, skeletal myopathy, and growth delay. This study aims to describe our clinical experience and outcome of cardiac disease in BTHS.

Methods: A retrospective study was performed to evaluate the cardiac status in all patients with BTHS in the UK over a 10 year period. Data collected included demographics, clinical course, echocardiographic and electrographic parameters. Serial echocardiographic data were analysed to assess trend in myocardial function and morphology. Strain analysis was performed during the most recent echocardiogram.

Results: 26 patients were included in the study. The median age of the 22 surviving patients at the time of evaluation was 12.6 years (range 2.0 to 23.8 years). Seven patients underwent cardiac transplantation at a median age of 3.6 years (range 0.33 to 11.9 years) and 6 recipients are alive at last follow up with good cardiac function. Four patients died at a median age of 2.13 years (range 0.67-4.22 years), and all deaths were related to cardiomyopathy. The 5 year survival rate was 85%, and no deaths were observed after 5 years of age. 92% (24/26) of patients presented with cardiomyopathy; whilst one had profound lactic acidosis and another had a positive family history of BTHS. Left ventricular diastolic dimension and systolic function measured by fractional shortening tended to normalise and stabilise after the first 3 years of life in the majority of patients. However, patients with BTHS (n=16) had statistically significant reduction in global longitudinal and circumferential strain compared to controls (n=18), p<0.001, despite normal fractional shortening and ejection fraction. There was also significantly reduced apical rotation or reversed apical rotation, p=0.001; and reduced LV torsion, p=0.046, in the patient group.

Conclusion: There is a broad spectrum of cardiac phenotype in BTHS, however longer term outcome in our cohort suggest good prognosis after the first 5 years of life, with the majority of patients remaining asymptomatic and recovering normal cardiac function by conventional echocardiographic measures independent of heart failure medication. However, strain imaging may provide valuable insight into the abnormal myocardial mechanics in Barth cardiomyopathy and potentially aid management.