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Heart Failure and Cardiomyopathies

THE HEART IN GAUCHER DISEASE: AN ECHOCARDIOGRAPHIC STUDY

Moderated Poster Contributions

Heart Failure and Cardiomyopathies Moderated Poster Theater, Poster Hall B1

Saturday, March 14, 2015, 4:15 p.m.-4:25 p.m.

Session Title: Novel Applications of Echo Imaging in Heart Failure

Abstract Category: 14. Heart Failure and Cardiomyopathies: Clinical

Presentation Number: 1166M-07

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Background: Gaucher disease (GD) is an autosomal recessive disorder of lysosomal storage resulting in accumulation of glucosylceramide in various organs. Cardiovascular involvement is well recognized, but the complete spectrum of cardiovascular phenotypes has never been documented. The aim of our study was to describe the echocardiographic findings in a series of well-characterized and genotyped GD patients.

Methods: We reviewed TTE performed on 33 patients (m=14, f=19, mean age 46 ± 17 years) with GD. We evaluated traditional (2-D, color, M-mode, tissue Doppler) and novel (speckle strain analysis) echocardiographic parameters.

Results: Concentric LV remodeling was noted in 10 of 33 patients. 5 had LVMI of >100 gm/m². Only 1 patient had PASP > 35 (mean 25 ± 6) mmHg. Premature valvular calcification of mitral and aortic (age <65) was seen in 45% (n=15). Of these, 8 had genotype N370S/N370S and 13 were on enzyme therapy. Average global longitudinal strain (GLS) and strain rate (SR) were 18.35 ± 2.4 and 0.84 ± 0.14 , respectively. In patients with GLS < 16 the LVMI was significantly higher (92 ± 40 vs. 71 ± 18 , $p = 0.05$).

Conclusion: This is the first comprehensive echocardiographic series demonstrating the spectrum of cardiac phenotypes in GD. Important observations include concentric remodeling, high LVMI, valvular calcification and abnormal GLS and SR. SR and GLS were lower in general and were negatively related to LVMI. These abnormalities will need to be studied in a larger cohort and compared to controls.

Genotype	N370S/N370S	N370S/L444P	N370S/84GG	other	all
PASP (mmHg)	25.6 ± 6	26.2 ± 6	27.9 ± 8	25.3 ± 6	26 ± 6
LVMI (gm/m ²)	79 ± 30	75 ± 27	78 ± 38	75 ± 25	75 ± 24
Valve abnormalities	7/14	1/5	5/5	5/9	18/33
Global longitudinal strain	17.7 ± 1.9	17.8 ± 2.2	17.1 ± 1.6	17.7 ± 1.9	18.4 ± 2.4
LV strain rate	0.9 ± 0.12	0.9 ± 0.14	0.9 ± 0.16	0.9 ± 0.12	0.8 ± 0.14
Left ventricular end diastolic volume Index (ml/m ²)	48 ± 11	47 ± 11	47 ± 7	48 ± 11	48 ± 10
Left ventricular end systolic volume Index (ml/m ²)	17 ± 5	16 ± 4	15 ± 4	17 ± 5	16 ± 4.5
LVEF (%)	66 ± 5	66 ± 5	66 ± 6	66 ± 5	66 ± 5
Aortic root dimension (cm)	3.0 ± 0.45	2.9 ± 0.35	3.0 ± 0.34	3.0 ± 0.4	3.0 ± 0.5