What are the specific features of Fabry cardiomyopathy in comparison to amyloidosis cardiomyopathy ?

Les Hôpitaux Universitaires de STRASBOURG

S. El ghannudi [1,2,3] , A. Lefoulon [4] , E. Noel [5] , P. Germain [1], S. Doutreleau[4], Mi-Y. Jeung[1] , A. Gangi[1], C. Roy[1]

UNIVERSITÉ DE STRASBOURG

(1) Department of cardiovascular radiology, Nouvel Hôpital Civil (NHC) , Strasbourg, France, (2) Nuclear medicine department, NHC, Strasbourg, France, (3) UMR 7357, University of Strasbourg, (4) Cardiology department, NHC, Strasbourg, (5) Internal medicine department, NHC, Strasbourg

I. OBJECTIVES

The aim of the study was to determine specific clinical, ECG, echocardiography and cardiac magnetic resonance imaging (CMR) features of Fabry disease cardiomyopathy (FCM) in comparison to amyloidosis cardiomyopathy (ACM).

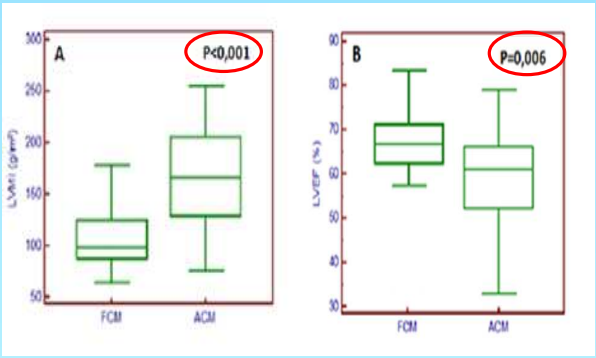
II. METHODS

We retrospectively included forty six patients, twenty-four patients with FCM, twenty-two with ACM between 2004 and 2013. Detailed demographic, clinical, ECG, echocardiography and CMR features were recorded.

III. RESULTATS

FCM were younger than ACM (41 [34.49-56] vs 76.5 [67-82.1] years, p<0.0001). Lower frequency of left ventricular (LV) heart failure in FCM (4.2% vs 86.4%, p<0.0001) and lower heart frequency in FCM vs ACM (66 [62-76.1] vs 80 [68.6-85.6] beats/min, p=0.04).

Using echocardiography, Lower left ventricular mass index (LVMI) in FCM vs ACM (117 [97.7-126.8], 165 [134-198.6], p=0.0006). Higher LV ejection fraction (LVEF) and cardiac index in FCM (68.5 [63.7-70]%, 61 [54.1-65.3], p=0.005, 3.5 [2.1-4.6], 1.65 [1.3-2.23] ml/min/m^2, p=0.02 respectively).



The border of the box plots are 25th and 75th percentiles of the median values. The line inside the box depicts the median. **(A)** LVMI in FCM and ACM **(B)** LVEF in FCM and ACM

Lower E/Ea in FCM (6 [6-8], 14.5 [11-19.2], p<0.0001). Higher S wave DTI at lateral mitral annulus and S wave DTI at lateral tricuspid annulus in FCM vs ACM (10.15 [8-11.7], 5.5 [4-8], p=0.0009), (13.6 [11.8-15.2], 10.25 [9-12.4], p=0.02) respectively.

	FCM (n=24)	ACM (n=22)	P Value
Echocardiography			
LVM increased, n (%)	16 (66.7)	20 (90.9)	0.02
LVMI (g/m²)	117 [97.7-127]	165 [134-198.6]	<0.001
IVS thickness > 10 mm, n (%)	15 (62.5)	22 (100)	<0.001
Thickness IVS (mm)	11 [10-12.2]	15.5 [14-19]	<0.001
PW thickness > 10 mm, n (%)	14 (58.3)	21 (95.4)	0.005
PW thickness (mm)	11 [10-12]	16 [14.4-18.6]	<0.001
LV dilated	0 (0)	0 (0)	1
LVED D (mm)	50 [47.3-51.6]	45 [38.5-47.8]	0.004
LVEF (%)	68.5 [63.7-70]	61 [54.1-65.3]	0.006
LVEF < 55%, n (%)	0 (0)	6 (27.3)	0.008
LV Kinetic disorder, n (%)	1 (4.2)	4 (18.2)	0.19
GLS ([N] %)	16 [15-20.5]	10 [6.2-12.8]	<0.001
D.F. abnormalities, n (%)	8 (33.3)	15 (68.2)	<0.001
E wave deceleration time (ms)	222 [211-230]	198 [174.2-246]	0.38
E/A	1.2 [1-1.45]	1.1 [0.7-4.5]	0.78
Ea Velocity (cm/s)	11.5 [5.6-22]	5.6 [2.5-10.5]	<0.001
E/Ea	6 [6-8]	14.5 [11-19.2]	<0.001
Sa wave velocity (cm/s)	11.15 [8-11.7]	5.5 [4-8]	<0.001
Mitral valvulopathy, n (%)	6 (25)	17 (77.3)	<0.001
Aortic valvulopathy, n (%)	2 (8.3)	5 (22.7)	0.23
Tricuspid valvulopathy, n (%)	1 (4.2)	5 (22.7)	0.03
RV abnormalities, n (%)	0 (0)	14 (63.6)	<0.001
RVFAC < 35%, n (%)	0 (0)	5 (22.7)	0.02
TAPSE ? 16 mm	0 (0)	9 (40.9)	0.001
S DTI (mm)	13.6 [11.8-15.2]	10.5 [9-12.4]	0.03
RVEDV indexed (mL/m²)	9.4 [7.9-10.7]	7.6 [6.6-9]	0.01
RVH	0 (0)	7 (31.8)	<0.001
SPAP (mmHg)	25 [21-28.2]	42.5 [38-45]	<0.001
LAV > 32 mL/m², n (%)	5 (20.8)	16 (72.7)	<0.001
LA surface (cm²)	20 [18-21]	26 [21-31.6]	0.002
CMR			
LVMI (g/m²)	83 [67.4-89.7]	109.5 [91-130]	0.002
IVS (mm)	11 [9-12]	17 [15-20]	<0.001
LVED V indexed (mL/m²)	69 [63.3-81.4]	61.5 [54.4-72.6]	0.16
LVEF (%)	69.5 [62.9-70]	63 [59.6-67.3]	0.047
LVLE (myocardial Fibrosis), n (%)	4 (26.7)	21 (95.5)	<0.001
Pericardial effusion, n (%)	0 (0)	6 (28.6)	0.007

No difference in right ventricular systolic function between the groups FCM (46 [38-56] vs ACM 39.5% [37.9-48.1], p=0.21) but decreased TAPSE (40.9% vs 0%, p=0.001) and higher pulmonary systolic pressure in ACM vs FCM (42.5 [38-45] vs 25 [21-28.2] mmHg, p<0.0001).

In MRI, lower myocardial fibrosis (26.7% vs 95.5 %, p<0.00001) and pericardial effusion (0% in FCM vs 28.6% in ACM, p=0.009).

IV.CONCLUSION

FCM appears to be less severe than ACM with a lower frequency of heart failure, regional and global left ventricular dysfunction and lower left ventricular myocardial fibrosis. Furthermore, there is a lower regional right ventricular abnormalities in FCM.