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Psychological impact of predictive genetic testing in cardiomyopathies

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Background: Most Cardiomyopathies are inherited diseases with autosomal dominant inheritance and delayed cardiac expression. In families with a documented causal mutation, predictive genetic testing can be proposed to "healthy" relatives to appropriately manage medical follow-up. Psychological impact of predictive genetic testing is however poorly described in these diseases.

Aim and methods: We performed a prospective study about the psychological impact of predictive genetic testing in adults seen in our out-patient multidisciplinary consultation (cardiologist, genetic counsellor, and psychologist). Self-report questionnaires were proposed during the initial consultation (Q1) and 1 to 3 month after genetic result was given (Q2). Questionnaires used validated scales to evaluate anxiety, depression, hopelessness, self-esteem, impact of announcement, quality of life (QOL), and motivations.

Results: Sixty-six adults completed questionnaire Q1 and 38 completed questionnaire Q2 (15 mutation carriers, 23 non carriers). Analysis of psychometric tests revealed no significant modification before/after genetic results. However, genetic status influenced the level of general anxiety, depression score and QOL. The consultation process was evaluated as useful (97%) and reassuring (80%). The interview with the pyschologist was useful especially for the anticipation of the result (73%). Waiting period (minimal period fixed between consultation and blood sampling) was evaluated as reassuring (38%) but unnecessary (91%).

Conclusion: We report on the first French evaluation of the psychological impact of predictive genetic testing in cardiomyopathies. No deleterious effect of genetic results was observed. However the study underlines the usefulness of multidisciplinary management, especially to help the relatives to better anticipate the genetic result and consequences.

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Very long-term effects of pacing therapy in Hypertrophic Obstructive Cardiomyopathy (HOCM)

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The clinical value of DDD pacing as primary treatment of HOCM remains controversial. Very long-term data are lacking.

Aims: single-centre observational study aimed at describing the very long term effects on symptoms, clinical and echocardiographic outcomes

Patients: 54 patients (59±14 years) with symptomatic (NYHA Class >2) drug-refractory HOCM implanted with a DDD pacemaker with or without defibrillator between 1991 and 2007 and followed up to 20 years (mean 11.5; range 0,4-21,8).

Main results are summarised in table. No patient had myomectomy or septal ablation during follow-up (f/u). NYHA functional class and other symptoms were significantly improved at 1-2 years and at the end of f/u. Left ventricular outflow tract (LVOT) gradient decreased by a mean of 78% at 1-2 years and 89% at end f/u consistent with SAM resolution. LV ejection fraction decreased over time with a mean value of 56% at end f/u without evidence of cavity dilatation. The actuarial survival rate was 90% at 5-yrs and 65% at 10-yrs. 24 patients died, 19 from non cardiac cause and 5 cardiovascular. 2 patients had heart transplant after 8 and 13 yrs.

Conclusion: The clinical and echocardiographic outcome of HOCM patients treated by DDD pacing seems favourable, inviting to re-evaluate the exact value of the therapy in further controlled studies

Table – Main results

	Baseline	3 months	1-2 years	End f/u	P value
NYHA functional class, (%)					< 0,0001
Grade 1	0	31	35	36	
Grade 2	43	53	59	57	
Grade 3	52	16	6	7	
Grade 4	5	0	0	0	
Syncope/near-syncope (%)	76/48	2/2	2/2	2/2	<0,0001
Angina (%)	57	4	4	4	<0,0001
LVOT gradient (mmHg)	79±36	20±24	11±15	8±21	<0,0001
SAM (%)	96	38	30	16	<0,0001
LVEF (%)	63,5±7,5	61±7	59±7	56±9	0,05
LVEDD (%)	47±5	NA	NA	43±12	0,34

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Potential role of peptide natriuretic and troponin-T to predict cardiac echocardiographic findings in hereditary transthyretin amyloidosis patients

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Background: Transthyretin (TTR) familial amyloid polyneuropathy (FAP) is a fatal autosomal dominant neurodegenerative disease characterized by deposition of transthyretin targeting mainly the peripheral nervous system and the heart. Early noninvasive detection of cardiac impairment is of importance for the therapeutic management.

Aim: Assess if natriuretic peptide (NT-proBNP) or troponin T (cTnT) are reliable biomarkers to predict echocardiographic left ventricle (LV) impairment in a wide variety of TTR patients.

Methods: 36 asymptomatic carriers and patients with proven FAP genetic mutation had clinical, biological and echocardiography assessment of left ventricle (LV) systolic function (SD), filling pressure (FP) and hypertrophy (LVH) as marker of amyloid deposition

Results: In the all cohort, the median (IQR) age, NT-proBNP, LV ejection fraction were respectively 59 (41-74), 323pg/ml (58-1960) and 60% (51-66). 64% were men and 12 had an increased in cTnT. TTR gene mutations prevalence was 50% for Val30Met. 4 patients were asymptomatic, 6 had only neurologic clinical signs and 26 had echo-LV abnormalities with or without neurologic disorders. Their median NT-proBNP value were respectively: 33 (19-50), 54 (37-154) and 747 (253;2840). Using received-operator curve, NTproBNP identified significantly patients with echo-LV abnormalities (Area : 0.92;(0.83-0.99), p=0.001) with a threshold above 82pg/ml and a sensitivity of 92% and specificity of 90%. Elevated cTnT (superior to 0.01ng/ml) was only observed in patients combining impairment of LVH and SD or LVH, SD and FP.