Genetic counselling and testing of adult patients with cardiomyopathies: insight from the EORP cardiomyopathy and myocarditis registry of the European Society of Cardiology

Authors:
T Helio¹, P Elliott², JW Koskenvuo³, JG Gimeno⁴, L Tavazzi⁵, M Tendera⁶, P Kaski⁷, A Maggioni⁸, C Laroche⁹, ALP Caforio¹⁰, P Charron¹¹, ¹University of Helsinki and Helsinki University Hospital - Helsinki - Finland, ²University College London and St. Bartholomew’s Hospital - London - United Kingdom of Great Britain & Northern Ireland, ³Blueprint Genetics - Helsinki - Finland, ⁴Hospital Universitario Virgen Arrixaca - Murcia - Spain, ⁵Maria Cecilia Hospital - Cotignola - Italy, ⁶School of Medicine in Katowice, Medical University of Silesia - Katowice - Poland, ⁷Great Ormond Street Hospital for Children - London - United Kingdom of Great Britain & Northern Ireland, ⁸ANMCO Research Center - Florence - Italy, ⁹European Society of Cardiology, EUObservational Research Programme - Sophia-Antipolis - France, ¹⁰University of Padova, Cardiology, Dept of Cardiological, Thoracic and Vascular Sciences and Public Health - Padova - Italy, ¹¹Sorbonne University - Paris - France.

On behalf: EORP Cardiomyopathy Registry Investigators Group

Topic(s):
Myocardial Disease – Treatment

Introduction

Cardiomyopathies comprise a heterogeneous group of diseases, often of genetic origin.

Purpose

We assessed the current practice of genetic counselling and testing of adult cardiomyopathy patients in the prospective ESC EORP cardiomyopathy registry.

Methods

3 208 adult patients from sixty-nine centres in 18 countries were enrolled. Clinical data on genetic counselling and testing and on the presentation of cardiomyopathies were gathered.

Results

Genetic counselling was performed in 60.8% of all patients (75.4% in hypertrophic (HCM), 39.2% in dilated (DCM), 70.8% in arrhythmogenic right ventricular (ARVC) and 49.2% in restrictive cardiomyopathy (RCM), p<0.001). Comparing European geographical areas, genetic counselling was performed from 42.4% to 83.3% (p<0.001). It was provided by a cardiologist (85.3%), geneticist (15.1%), genetic counsellor (11.3%), or a nurse (7.5%), (p=0.001). Genetic testing was performed in 37.3% of all patients (48.8% in HCM, 18.6% in DCM, 55.6% in ARVC and 43.6% in RCM, p<0.001). Index patients with genetic testing were younger at diagnosis, had more familial disease, family history of sudden cardiac death or implanted cardioverter defibrillators but less comorbidities than those not tested (p<0.001 for each comparison). At least 1 disease causing variant was found in 41.7% of index patients with genetic testing (43.3% in HCM, 33.3% in DCM, 51.4% in ARVC and 42.9% in RCM, p=0.13).

Conclusion

We report on the practice of genetic counselling and testing in cardiomyopathies in Europe. Genetic counselling and testing were performed in a substantial proportion of patients but less often than recommended by European
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1 University of Helsinki and Helsinki University Hospital - Helsinki - Finland, 2 University College London and St. Bartholomew's Hospital - London - United Kingdom of Great Britain & Northern Ireland, 3 Blueprint Genetics - Helsinki - Finland, 4 Hospital Universitario Virgen Arrixaca - Murcia - Spain, 5 Maria Cecilia Hospital - Cotignola - Italy, 6 School of Medicine in Katowice, Medical University of Silesia - Katowice - Poland, 7 Great Ormond Street Hospital for Children - London - United Kingdom of Great Britain, 8 ANMCO Research Center - Florence - Italy, 9 European Society of Cardiology, EURObservational Research Programme - Sophia-Antipolis - France, 10 University of Padova, Cardiology, Dept of Cardiological, Thoracic and Vascular Sciences and Public Health - Padova - Italy, 11 Sorbonne University - Paris - France

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Genetic counselling was performed in 60.8% of all patients (75.4% in hypertrophic (HCM), 39.2% in dilated (DCM), 70.8% in arrhythmogenic right ventricular (ARVC) and 49.2% in restrictive cardiomyopathy (RCM), p<0.001). Comparing European geographical areas, genetic counselling was performed from 42.4% to 83.3% (p<0.001). It was provided by a cardiologist (85.3%), geneticist (15.1%), genetic counsellor (11.3%), or a nurse (7.5%), (p<0.001). Genetic testing was performed in 37.3% of all patients (48.8% in HCM, 18.6% in DCM, 55.6% in ARVC and 43.6% in RCM, p<0.001). Index patients with genetic testing were younger at diagnosis, had more familial disease, family history of sudden cardiac death or implanted cardioverter defibrillators but less comorbidities than those not tested (p<0.001 for each comparison). At least 1 disease causing variant was found in 41.7% of index patients with genetic testing (43.3% in HCM, 33.3% in DCM, 51.4% in ARVC and 42.9% in RCM, p=0.13).

Conclusion
We report on the practice of genetic counselling and testing in cardiomyopathies in Europe. Genetic counselling and testing were performed in a substantial proportion of patients but less often than recommended by European guidelines, and much less in DCM than in HCM and ARVC, despite evidence for genetic background.