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European cardiomyopathies registry finds high use of defibrillators and genetic testing

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The most representative snapshot of real world practice in cardiomyopathies in Europe has shown a higher than expected use of defibrillators and genetic testing. The baseline results of the ESC's EORP Cardiomyopathy Registry Pilot are presented for the first time today at ESC Congress 2015.

"This is the first time that we have a snapshot of practice in cardiomyopathies across different European countries with diverse healthcare systems," said principal investigator Professor Perry Elliott, professor in inherited cardiovascular disease at The Heart Hospital and University College London, UK. "The literature is dominated by studies from a small number of large centres and none of the research has been so representative of Europe as a whole."

Cardiomyopathies are a common group of conditions that occur in one out of 200 to 300 people in Europe. They are genetic, which makes diagnosis important not only for patients but also for their families. All cardiomyopathies are associated with premature death, predominantly from sudden cardiac death and progressive heart failure but also from stroke. None of the conditions are curable but there are clear strategies for reducing or eliminating complications so diagnosis is crucial.

The EORP Cardiomyopathy Registry Pilot is a prospective, multicentre, observational study of patients presenting to 27 referral cardiomyopathy centres in 12 European countries. In 2012 and 2013 investigators examined the characteristics of 1 115 patients with hypertrophic (HCM),

dilated (DCM), arrhythmogenic right ventricular (ARVC) and restrictive (RCM) cardiomyopathy.

Participating centres were required to have dedicated cardiomyopathy clinics staffed by experienced medical and nursing teams and access to facilities for genetic testing with skills in interpreting the results. Each centre was asked to contribute up to 40 consecutive patients aged over 18 years during a 12 month period.

The commonest cardiomyopathy was HCM (61%) followed by DCM (31%), ARVC (5%) and RCM (3%). One of the most striking findings was that 316 patients (28.3%) had received an implantable cardioverter defibrillator (ICD). The proportion was highest in patients with ARVC (57.6%) and lowest in those with RCM (6.9%). The majority of ICDs were for primary prophylaxis. Some 8% of patients had a pacemaker alone and 5.2% had a cardiac resychronisation therapy (CRT) device.

"Nearly 60% of patients with ARVC had an ICD which is extremely high," said Professor Elliott. "We don't fully understand why that is yet. It may be because referral centres see the highest risk patients or therapy may be more aggressive in the real world than guidelines recommend, with a lower threshold for putting in a defibrillator than anticipated."

Also striking was the finding that 462 patients (41.4%) had genetic testing, with a causative mutation reported in 236 (51.1%). "This is the first time that data has been collected on the scale of genetic testing in patients with cardiomyopathies," said Professor Elliott. "The results show that the use of genetic testing is high and efficient, with more than half of the tests yielding a positive result."

Nearly 60% of HCM and ARVC patients and 70% of DCM patients were men. Slightly more than half of RCM patients were women. Professor Elliott said: "Most of the diseases were more common in men

which has been described before but is poorly understood. It could be that men have more severe disease or there may be systematic underdiagnosis in women."

The registry showed that one third of patients with a cardiomyopathy were diagnosed either incidentally or through family screening. "Cardiologists, GPs and other physicians need to be alert to the fact that these are quite common conditions that can appear through different routes," said Professor Elliott.

He concluded: "Patients with cardiomyopathies need intensive investigation using specialised diagnostic tests. Many of the findings reported today, including the very high use of ICDs in ARVC and the high yield from genetic testing, will be examined in substudies from the pilot and in the long-term registry which will also include non-referral centres, centres outside Europe, paediatric patients and, for the first time in a large international registry, myocarditis."

More information: Professor Elliott will give the lecture 'Presentation of the main results of the pilot phase of the Cardiomyopathy registry'

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