P6455

Differences in cardiac phenotype and natural history of laminopathies with and without neuromuscular presentation

R. Ditaranto[1], C. Rapezzi[1], G. Boriani[2], F. Pasquale[1], M. Graziosi[1], G. Vitale[1], A. Berardini[1], G. Lanati[1], A. Corsini[1], G. Caponetti[1], G. Lattanzi[3], L. Potena[1], M. Ziacchi[1], O. Leone[1], E. Biagini[1]

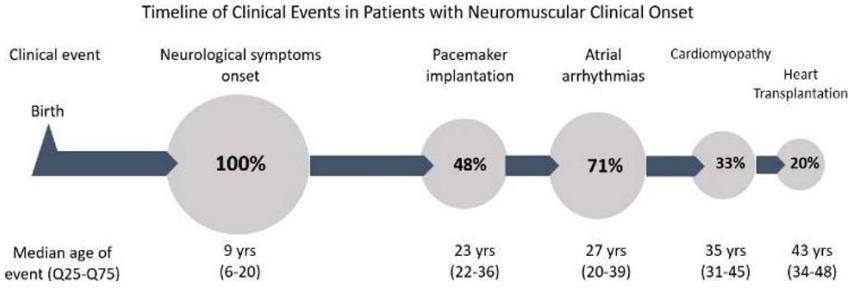
[1]University Hospital Policlinic S. Orsola-Malpighi, Bologna, Italy; [2]University of Modena & Reggio Emilia, Department of Biomedical, Metabolic and Neural Sciences, University of Modena and Reggio Emilia, Modena, Italy; [3]University of Bologna, Italian National Research Council | CNR • Institute of Molecular Genetics IGM Bologna, Bologna, Italy

Aim: To look for differences in cardiac phenotype and natural history of patients affected by laminopathy, according to the presence or less of neuromuscular involvement at clinical presentation.

Methods: We prospectively analyzed 47 consecutive pts with a genetic diagnosis of laminopathy followed at a single centre between 1994 and 2017. Additionally, reports of clinical and instrumental evaluations before referral at our centre were retrospectively evaluated.

Results: Neuromuscular presentation, mainly as Emery-Dreifuss muscular dystrophy (EDMD), was present in 21 (46%) cases (14 LMNA and 7 EMD gene mutations). These pts had symptoms earlier (9 vs 39 years, $p<0.001$) in life compared to pts without neuromuscular onset (26 LMNA gene mutations), and clinical manifestations anticipated the first evidence of cardiac disease by a mean time of 15±8 years (maximum time gap of 38 years). Despite a similar prevalence of atrial fibrillation/flutter (AF) (71% vs 65%, p=0.758) and atrio-ventricular blocks (48% vs 65%, p=0.250), pts with neuromuscular onset experienced AF and pace-maker implantation at a significantly younger age (27 vs 41 yrs, p=0.015 and 23 vs 44 yrs, p=0.027 respectively). Differently a higher prevalence of sinus node dysfunction (33% vs 4%; p=0.015) and atrial paralysis (14% vs 4%; p=0.311) was reported in pts with neuromuscular onset. Prevalence of cardiomyopathy (CMP) (73% vs 33%, p=0.008) and sustained ventricular tachyarrhythmias were higher among pts with cardiac onset (23% vs 4%, p=0.111) whereas the prevalence of heart transplantations and median age of recipients were similar in the two groups (24% vs 20%, p=1.000 and 46 vs 43, p=0.592 years respectively). All pts with neuromuscular onset who received a diagnosis of CMP had a previous history of rhythm disturbance except 2 cases, where a concomitant diagnosis of the 2 disorders was formulated. On the contrary a strict temporal progression from rhythm disturbances to CMP (or viceversa) was not appreciable in the other group: AF and AVBs could precede the diagnosis of CMP be diagnosed at the same time or later.

Conclusions: In pts affected by laminopathy neuromuscular involvement, when present, was most often the first clinical manifestation and preceded cardiological involvement, with a long time frame in some cases. Except for sinus node dysfunction, much more frequent in patients with EDMD, a similar prevalence of rhythm disturbances was reported, although pts with neuromuscular clinical onset were younger at diagnosis of AF and at PM implantation. Pts without neuromuscular presentation had a higher prevalence of CMP and ventricular arrhythmias, albeit a similar rate of heart transplantation. In pts with neuromuscular onset, cardiac involvement was characterized by a stepwise progression from rhythm disturbances to CMP, where a strict temporal progression from rhythm disturbances to CMP was not observed in the group of pts without neuromuscular clinical onset.



Downloaded from https://academic.oup.com/eurheartj/article/40/Supplement_1/ehz746.1048/5597399 by guest on 24 April 2024