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Phospholamban p.Arg14del cardiomyopathy

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Phospholamban p.Arg14del cardiomyopathy

Clinical and morphological aspects supporting the concept of arrhythmogenic cardiomyopathy

- 1 Phospholamban p.Arg14del cardiomyopathy is characterized by large perinuclear and circumnuclear PLN protein aggregates, aggresomes and autophagic degradation. *(this thesis)*
- 2 Phospholamban immunostaining is a highly sensitive and specific method for diagnosing phospholamban p.Arg14del cardiomyopathy in complete heart- and myocardial LV assist device samples, but phospholamban aggregates are rarely detected in RV endomyocardial biopsy samples. *(this thesis)*
- 3 Phospholamban p.Arg14del cardiomyopathy is a distinct biventricular disease entity within the ACM spectrum. *(this thesis)*
- 4 Myocardial fibrosis occurs as an early phenomenon in PLN p.Arg14del mutation carriers, is most abundant in the LV inferolateral wall in line with electrocardiographical findings, and is independently associated with the occurrence of ventricular arrhythmia. *(this thesis)*
- 5 In an important subgroup of presymptomatic PLN p.Arg14del mutation carriers, subtle but significant structural remodeling as well as loss of LV diastolic function and RV systolic function can be observed using echocardiography. *(this thesis)*
- 6 The presence of late gadolinium enhancement is likely to be an important independent risk factor for malignant ventricular arrhythmia in PLN p.Arg14del mutation carriers, but to prove this a longer follow-up period is required. *(this thesis)*
- 7 The participation rate of presymptomatic mutations carriers in an intervention trial for an incompletely penetrant disease is easily overestimated. *(this thesis)*
- 8 You cannot change the wind, but you can adjust the sails. *(E. Edwards)*
A lesson learnt from animal experimental studies.
- 9 Gutta cavat lapidem, non vi, sed saepe cadendo. *(Ovidius)*
- 10 La vraie générosité envers l'avenir consiste à tout donner au présent. *(A. Camus, L'Homme Revolte)*
- 11 Een gezond mens heeft duizend wensen, een zieke mens maar één. *(J. Mens)*