



Identification of a strong genetic background for progressive cardiac conduction defect by epidemiological approach

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Résumé en anglais

Introduction Progressive cardiac conduction defect (PCCD) is a frequent disease attributed to degeneration and fibrosis of the His bundle. Over the past years, gene defects have been identified demonstrating that PCCD could be a genetic disease. The aim of this study was to show a familial aggregation for PCCD using a genetic epidemiological approach to improve in fine genetic knowledge of the transmission of the disease. Methods and results Using the French social security number, the authors have been able to determine the city of birth of the 6667 patients implanted with a pacemaker (PM) for PCCD between 1995 and 2005 in the western part of France. The authors then mapped the frequency of PM implantations for PCCD. A large heterogeneity of the frequency of the disease has been observed, with a frequency of 0.21% in the major city (Nantes) ranging up to 2.28% in specific parishes. Familial studies performed in the parishes with the highest frequency of the disease allowed the authors to identify five large families with PCCD. Clinical investigations demonstrated phenotype heterogeneity between families. Three patterns have been differentiated. Conclusions This study demonstrates a disparate geographical repartition of the frequency of PM implantation in the area of the authors at least in part related to a hereditary factor. The identification of five large families affected by PCCD using epidemiological approach underlines the existence of a major genetic background in PCCD.

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