A case report of juvenile onset familial atrial fibrillation due to genetic defect

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Introduction: Atrial fibrillation (AF) is a common arrhythmia and morbidity increases with age. Genetic factors are considered as the key in the development of AF, and familial forms of AF were reported more than 80 years ago. KCNQ1-V241F was reported to be a gene that causes both bradycardia and AF. However, it is rare for three relatives in family with a history of AF have same genetic defect.

Methods: A 24-year-old man suffered from bradycardia since birth and was diagnosed with AF at the age of 18 years. We performed cardiac examination of cardiomyopathy and coronary artery disease to differentiate juvenile onset familial AF. In family history, 2 relatives of him had AF, 1 had SSS with AF (sick sinus syndrome type 3; pacemaker was implanted), and we examined their genetic defects.

Result: There were no special findings in cardiac examination. But we identified same genetic defect of KCNQ1-V241F in his family tree, his sister, his mother and his grandmother.

Conclusion: This case report has highlighted of gene defect in KCNQ1-V241F. This gene defect causes both bradycardia and AF. Therefore, in order to prevent the onset and complications of juvenile onset familial AF, it is important to conduct genetic tests on close relatives, even in the case of a genetic defect where family history is rarely reported.