Identification of one novel genetic locus and gene associated with atrial fibrillation in the Taiwanese population

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Experience:

Market Needs:

The market need for an atrial fibrillation (AF) genetic test in health exams is driven by several factors:

1. **Early Risk Identification:** AF is a leading cause of stroke and other cardiovascular complications. Early identification through genetic testing of AF helps individuals at risk take preventive actions, especially in those with family history of arrhythmia
2. **Personalized Treatment:** Genetic testing can aid in tailoring treatments for AF, improving outcomes and reducing adverse reactions to medications, which aligns with the growing trend toward precision medicine.
3. **Growing AF Prevalence:** With the aging population and rising prevalence of cardiovascular diseases, routine AF genetic screening could help manage healthcare costs by identifying at-risk individuals earlier.
4. **Proactive Health Management:** Offering AF genetic tests as part of health exams appeals to consumers interesting in proactive health monitoring and personalized wellness plans, such as exercise and diet planning to prevent AF when the genetic test is positive.

Our Technology:

We performed genome-wide association study (GWAS) with whole genome genotypes (4,512,191 SNPs) in 516 young AF Patients and 5160 normal sinus rhythm controls. We found 3 loci associated with AF, which included one locus close to *PITX2* gene (chromosome 4q25, rs2723329, $P=1.53\times10^{-10}$), one locus close to *RAP1A* gene (also close to previous *KCND3* locus; chromosome 1p13.2, rs7525578, $P= 1.24\times10^{-26}$) and one novel locus close to *HNF4G* gene (chromosome 8q21.13, rs2980218, $P=2.19\times10^{-9}$). They were further validated in a replication population with 1002 independent AF patients and 2003 controls ($P=4.60\times10^{-9}$, 4.45×10^{-10} and 6.97×10^{-5} for *RAP1A, PITX2* and *HNF4G*, respectively). Transcriptome-wide association study (TWAS) revealed significant association of tissue *HNF4G* gene expression with AF ($P= 3.30\times10^{-7}$).

Strength:

We found one novel AF locus (*HNF4G*) which had never been reported before. *HNF4G* has also never been reported to be related to any cardiovascular disease.

Competing Products:

Our patent of using *KCNIP1* copy number variation (CNV) as an AF genetic test in Taiwan

Intellectual Properties:

Use of rs2980218 single nucleotide polymorphism (SNP) in chromosome 8q21.13 which is close to the *HNF4G* gene as a genetic test for AF

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