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Rapid Detection and Prevalence of the AGT Deletion/Insertion Mutation underlying Autosomal Recessive Renal Tubular Dysgenesis in Taiwan

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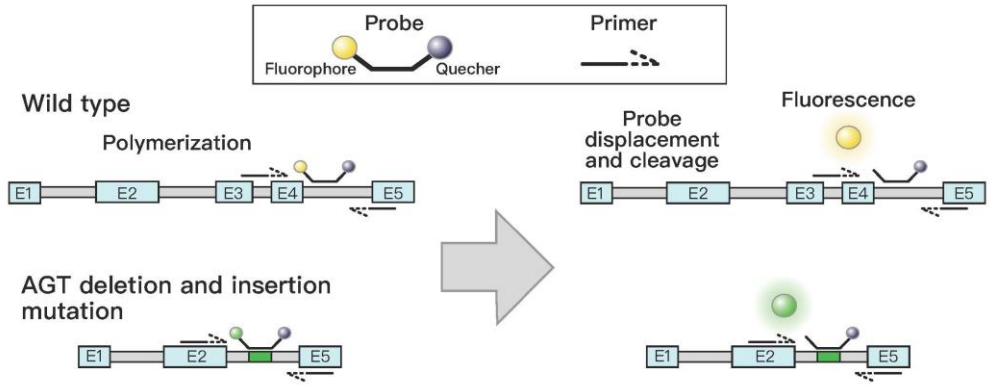
Objectives : A homozygous deletion/insertion (del/ins) mutation (E3_E4:2870bp del ins9bp) in the AGT gene responsible for autosomal recessive renal tubular dysgenesis (ARRTD) is frequently reported in Taiwan. Rapid and accurate detection of this peculiar mutation is crucial for genetic counseling and knowledge of the allele frequency in the population may help to better characterize the a priori risk for ARRTD in Taiwan.
Methods : Using a TaqMan probe-based real-time polymerase chain reaction, we first designed a mutation detection plate for the E3_E4:2870bp del ins9bp mutation of the AGT gene. The allelic frequency of heterozygous AGT mutation was determined in 5000 healthy subjects. The demographic data and serum AGT, angiotensin I (AgI), and AgII concentration were also collected in 2 affected patients, 20 carriers, and 9 normal subjects.
Results : The designed detection plate thoroughly validated by direct sequencing showed almost perfect sensitivity and specificity in genetically-diagnosed patients, carriers, and healthy subjects. The overall allelic frequency of positive AGT heterozygosity was 1% (50/5000). There was a significantly higher serum AGT concentration in heterozygous AGT carriers than wild-type subjects despite no difference in blood pressure.
Conclusions : This del/ins mutation in AGT can be rapidly and accurately identified by this allele-specific mutation plate. Due to its high prevalence in the Taiwanese population, it is likely that ARRTD may be that rare in Taiwan.

KSN 1.jpg



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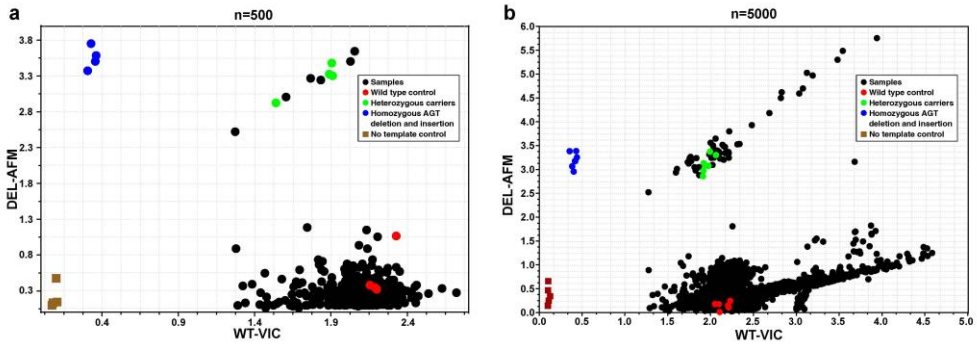
a
n=500
DEL-AFM
WT-VIC
Samples
Wild type control
Heterozygous carriers
Homozygous AGT deletion and insertion
No template control
b
n=5000
DEL-AFM
WT-VIC
Samples
Wild type control
Heterozygous carriers
Homozygous AGT deletion and insertion
No template control