Background: Mutations in genes encoding the cardiac sodium channel and associated compounds have been linked with atrial fibrillation (AF). Nevertheless, current expert consensus does not support genetic testing in AF which is in part based on the fact that “there is no therapeutic impact derived from AF genetic test results”. However, there are no studies available supporting this recommendation. Consequently, this study analyzed the impact of mutations affecting the cardiac sodium channel on rhythm outcome of AF catheter ablation.

Methods and Results: In 137 consecutive patients with lone AF enrolled in the Leipzig Heart Center AF ablation registry, screening for mutations in SCN5A, SCN1B - 4B, CAV3, GPD1L, SNTA1 and MOG1 was performed. We identified 3 rare non-synonymous variants in SCN5A, 5 in SCN1B, 1 in SCN4B, 1 in CAV3, 6 in GPD1L, 3 in SNTA1 and 3 in MOG1 (16%). Variant carriers were otherwise comparable with non-variant carriers. Analysis of AF recurrence rates after radiofrequency AF catheter ablation by serial 7-day Holter-ECG monitoring between 3 and 12 months revealed no difference between groups, i.e. 37 % in variant carriers vs. 39 % in non-variant carriers (p=0.9).

Conclusions: Mutations in genes encoding the cardiac sodium channel and associated compounds are frequently found in lone AF but seem not to impact on outcome of AF catheter ablation. This finding supports current recommendations not to screen for mutations in AF and highlights the role of the pulmonary veins for AF initiation and maintenance even in the presence of mutations. Larger studies are, however, needed to further explore the possible association between genotype and response to AF therapies.