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CONCOMITANT BRUGADA-LIKE AND SHORT QT ELECTROCARDIOGRAM LINKED TO SCN5A MUTATION

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Objectives Mutations in the α-subunit of cardiac sodium channel gene SCN5A can lead to the overlapping phenotypes of both the Brugada and type 3 long QT syndromes. However, the combination of Brugada and a short QT phenotype resulting from mutation in SCN5A has not previously been described. A man with concomitant Brugada-like ST-T and short QT electrocardiogram was identified and the SCN5A gene was sequenced. Whole cell patch clamp analysis of HEK293 cells expressing a SCN5A channel with the patient's sequence was used to investigate the biophysical properties of the channel. The patient with the family history of sudden death showed Brugada-like and short QT interval electrocardiogram. Sequence analysis of the coding region of the SCN5A gene, identified a G to A missense mutation at nucleotide site 2066 that resulted in an amino acid substitution of arginine to histidine at amino acid site 689 (R689H). Patch clamp analysis showed that the R689H failed to generate current when heterologously expressed in HEK293 cells, indicating it was a loss-of-function mutation. Our finding firstly shows that a R689H in SCN5A results in a loss of protein function and the co-existents of the

Brugada-like and short QT interval electrocardiogram phenotypes.
Methods
Conclusions