Dear Editor,

I read with great interest the recent article by Saif et al.1 In a recent issue of your esteemed journal. The article is highly thought provoking. Interestingly, the past few years have seen the identification of a number of pathological conditions resulting because of mutations in the KCNQ1 gene.

For instance, increased disease severity is seen in patients with Long QT syndrome (LQTS) who demonstrate 3' UTR single nucleotide polymorphisms such as rs8234 in the KCNQ1 gene.2 In fact, there is an increased risk of mortality in individuals with type-1 LQTS who exhibit cytoplasmic loop mis-sense mutations of the KCNQ1 gene.3 Certain mutations of the KCNQ1 gene may also result in left ventricular non-compaction in patients with LQTS.4

Similarly, familial atrial fibrillation may occur because of the V141M mutation of the KCNQ1 gene.5 The L203P variant of the KCNQ1 gene may result in Torsades de pointes in patients, especially those with underlying Steinert syndrome, while the G643S variant predisposes patients with underlying myocardial ischaemia to developing Torsades de pointes.5,7 KCNQ1 gene polymorphisms may also play a pathological role in the development of Romano-Ward syndrome and Jervell and Lange-Nielsen syndromes.8 The rare Jervell and Lange-Nielsen syndrome occurs because of the p.S277del/c.921G>A mutation of the KCNQ1 gene.9

Similarly, an increased predisposition to develop “new onset diabetes” after tacrolimus-treated renal transplantation is seen in patients who demonstrate the rs2237895 single nucleotide polymorphisms of the KCNQ1 gene.10 Interestingly, the rs2074196 polymorphism of the KCNQ1 gene increases the risk of developing gestational diabetes mellitus.11 An increased risk of developing nephropathy is also seen in diabetic patients who exhibit the T allele of rs2237897 polymorphism.12

Clearly, mutations in the KCNQ1 gene are associated with a range of pathological conditions ranging from familial atrial fibrillation to Jervell and Lange-Nielsen syndrome. Further studies are needed to identify any other similar associations.

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REFERENCES