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Modeling clinical inherited cardiac diseases: Implications for personalized medicine

Hypertrophic Cardiomyopathy (HCM) was the first of the cardiomyopathies to be attributed a genetic etiology. It is also considered to be one of the commonest hereditary cardiac disorders with a prevalence of about 1:500 in young adults. Yet, after 25 years although much has been gained in unraveling the genetic basis for HCM; clinical management of these patients is still based on symptom control and the prophylactic implantation of internal cardio defibrillators (ICD) for the prevention of fatal cardiac arrhythmias. Risk stratifying those patients who are most likely to benefit from device therapy given some mutations have a higher propensity for sudden cardiac death than others has proved difficult. However, the potential for pharmacological therapy in such cases has often not been considered given a lack of understanding of the inherent biological systems perturbed as a result of these underlying genetic changes, we put forward a platform to better understand the mechanism of action of some of the HCM phenotypes using an *in silico* then *in vitro* model which attempts to define the structural and physical characteristics of mutations within the confines of clinical datasets to define prognostic implications of sequences. This was then integrated "omics" model and later verified experimentally, potential drugs were used which could be repurposed to target some of the main pathways shown to be perturbed in this process. This represents a possible methodology for which in cardiovascular and inherited cardiac disorders may aid the clinical potentials for personalized therapeutics.

Biography

Rameen Shakur is the Wellcome Trust Fellow in Regenerative Medicine and Cardiology for the University of Cambridge, UK. He is the author of a number of papers and 3 text books in medicine.

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