Comparison of Hypertrophic Cardiomyopathy Candidate Gene Homologs in Human and Rhesus Macaque Sudden Cardiac Death Victims

Hypertrophic cardiomyopathy is a familial cardiomyopathy with autosomal dominant pattern of inheritance in humans. It is estimated to affect approximately 1 in 500 people and is a common cause of sudden cardiac death. Mutations have been identified in at least 10 genes. Over 1400 different gene variants have been identified, however presently it is not known which are causative of the disease and which are benign polymorphisms.

HCM has recently been described post-mortem in a colony of rhesus macaque primates who died suddenly at the California National Primate Research Center. Non-human primates are ideal animal models to study HCM because of their similarities to humans genetically, physiologically and anatomically. There are no comparative evolutionary medical studies involving human and non-human primate genomics data to date.

The availability of biological samples from rhesus macaques that died suddenly allows a unique opportunity to perform a comparative genomics study of this disease between these two species for the first time. Identification of shared gene variants between patients of both species may help to identify gene variants causative of HCM in humans. These can then be used to screen for the disease in sudden cardiac death survivors and relatives of patients with HCM.

Students who want to participate in the project will be expected to spend at least 6 hours in the lab. He/she will participate in the design and execution of the DNA analysis as well as sequence data analysis.

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