

FOUNDATION FUNDS RESEARCH ON HCM

HCM (Hypertrophic Cardiomyopathy) is a complex genetic disease for which there are currently upwards of a dozen different genes whose many associated mutations (more than 1,000) are involved in heart muscle function. Despite this large number of identified genetic mutations, a significant fraction of individuals with HCM (perhaps half of all patients) do not have any of these and are presumed to have as-yet-unidentified mutations.

A perhaps more intriguing aspect of HCM is the wide range of physical health status exhibited by individuals with identifiable mutations. It is estimated that up to 1 in 500 Americans may have a genetic mutation associated with HCM, and yet the number of individuals at risk for sudden cardiac death is much smaller. One of the most important areas for research is to understand the disease mechanism – how do genetic mutations lead to defects in the heart? What factors – genetic, environmental, and other – cause the mutations to sometimes be benign and other times result in disarray in heart muscle tissue, thickening of the ventricular wall, and serious risk of SCD?

The JTB Foundation is happy to announce the launching of our first research grant to Tufts Medical Center under the principal leadership of Dr Gordon Huggins to study genetic modifiers for HCM disease expression.

Dr Gordon Huggins in the Molecular Cardiology Research Institute at Tufts Medical Center has partnered with Dr Martin Maron, director of the Tufts Medical Center Hypertrophic Cardiomyopathy Clinic, for several years to research genetic factors that contribute to HCM. The broad goal of this research collaboration is to better define the many genetic factors that cause HCM as well to identify genes that alter the HCM disease presentation.

In the summer of 2011 the JTB Foundation funded a grant to Dr Huggins to study, in particular, the FHOD3 gene and its association with HCM disease expression. Early findings by Dr Huggins on FHOD3 were presented to the scientific community at the American Heart Association this year and, with the support of JTB, Dr Huggins and his team plan to publish a more complete analysis of this gene. Specific activities to be supported by the JTB grant funding include:

- The collection and purification of DNA from patients treated in the Tufts Medical Center HCM clinic to substantially increase the “bank” of DNA samples
- Further detailed scientific studies on FHOD3 to determine its role in disease expression and severity in HCM
- The support and development of a student trainee who intends to pursue a career in medicine or biomedical research and will work in Dr Huggins lab for a year on the grant

We are excited for this next step in our Foundation activities and look forward to sharing news and results from this project over the coming year.