Research Update – 2014 – John Taylor Babbitt Foundation

HCM, or Hypertrophic Cardiomyopathy, is the most common inherited cardiac disease and is estimated to affect up to 1 in 500 individuals. While most cases of HCM are benign, due to its high prevalence HCM is also the most common heart-related cause of sudden death in young athletes. The JTB Foundation's Research Initiative supports medical investigations aimed at uncovering the pathways by which diverse genetic HCM mutations, known and unknown, may lead to high risk of sudden cardiac death. The ultimate goal is to contribute to treatments that would disrupt these pathways and save lives. In 2014, our research initiate is funding work in the laboratory of Dr. M. Roselle Abraham at the Hypertrophic Cardiomyopathy Center of Excellence at Johns Hopkins.

Past research suggests that abnormal use of energy by the heart - burning too much energy for the same amount of work - is what results in the symptoms and serious health problems in some patients with HCM. Given the diversity of HCM-causing mutations and medical presentations, a major unanswered question is whether this energy abnormality occurs in all patients diagnosed with HCM and at all stages of the disease. The Abraham Laboratory at Johns Hopkins has focused on studying mitochondria in three different mouse models of HCM. Mitochondria are the so-called power houses of cells, responsible for synthesizing ATP, the cell's main source of chemical energy. The three mouse models are used to represent the spectrum of human HCM disease.

Through the generous funding support by the John Taylor Babbitt Foundation, Dr. Abraham's lab has discovered that mitochondrial function and calcium transport are affected to different degrees depending on the specific HCM mutation and the stage of the disease. For example, in one mutation that is known to cause high rates of sudden death in humans, mitochondrial number was reduced and function was impaired in 5-week-old mic, which correspond to teenage humans. The laboratory is now investigating why this happens in HCM with the goal being to develop novel therapies that prevent development of heart thickening, heart failure, arrhythmias and sudden cardiac death. The next major laboratory aim is to transform blood cells taken from the veins of HCM patients into beating heart cells. Success in this aim would allow various treatments to be tested on heart cells in a petri dish and the best treatment to be selected, resulting in targeted, personalized therapies.