

John Taylor Babbitt Foundation – Stanford University

2019 Research Update

The JTB foundation is supporting work directed by Dr. Victoria Parikh, a cardiologist at Stanford University specializing in the care of patients with inherited cardiovascular diseases. Dr. Parikh's research investigates the multiple causes of cardiomyopathy in the laboratory with a particular clinical and scientific interest in inherited arrhythmogenic cardiomyopathies.

Supported by funding from the JTB foundation, Dr. Parikh is developing a deeper understanding of the gene *RBM20*. Her previous work established that *RBM20* mutations cause heart failure and sudden cardiac death; current work is investigating variant-specific pathogenicity in *RBM20*. Funding from JTB has allowed Dr. Parikh to expand her laboratory team to add Anna Kirillova, a recent graduate of UC Davis. The Parikh lab has established the armament of gene-editing reagents necessary for insertion of mutated copies of *RBM20* into genetically altered induced pluripotent stem cells. They are simultaneously establishing a library of all possible variants in *RBM20* and validating their test of pathogenicity (via a stably expressed splicing reporter). In addition, Dr. Parikh has leveraged the support of the JTB Foundation to expand these studies to other genes that cause arrhythmogenic cardiomyopathy.

Dr. Parikh expects to have preliminary results on a cadre of *RBM20* variants by November of this year, which she will present, along with her clinical characterization of *RBM20* cardiomyopathy, at the annual American Heart Association national meeting. Recent results from Dr. Parikh and colleagues have been published in two papers on heart failure and cardiac hypertrophy (in *Nature Communications*) and restrictive cardiomyopathy (in *Circulation*). Dr Parikh will also be presenting research results at the University of Alabama Birmingham's Cardiovascular Institute and Beth Israel Deaconess Medical Center in Boston this September. Lastly, she has recently been promoted to Assistant Professor of Medicine at Stanford School of Medicine, beginning in September 2019.