The Robert M. Berne Cardiovascular Research Center Presents

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Understanding the Molecular and Genetic Basis of Fibromuscular Dysplasia

In this talk, Dr. Kovacic will cover the findings from this study, and what this has taught us of the molecular and genetic basis of this disease.

Fibromuscular dysplasia (FMD) is an understudied medical enigma that can cause arterial fibrosis, stenosis, dissection, tortuosity, aneurysm, dilation and occlusion, throughout the entire body. Mean age at diagnosis is 50-55 yrs and 94% are female. Importantly, FMD is not rare - its prevalence is as high as 5% in females. FMD commonly affects the renal arteries where it may cause hypertension, while cervical or coronary artery involvement may cause stroke or myocardial infarction, respectively. Death from FMD may arise from aneurysm rupture, stroke, myocardial infarction, or from other arterial beds like the mesenteric system causing fatal gut ischemia.

Our lack of pathobiologic knowledge of FMD is profound. Of the little we know, from the 1970’s-80’s when managed surgically (and arterial samples were available) we learned that FMD involves disarray of SMCs and myofibroblasts, with increased vascular collagen and matrix. However, due to a change in management from surgery to catheter-based therapy (e.g. angioplasty), FMD vascular samples are now rarely obtained, mandating a novel approach such as patient-specific fibroblasts as in our DEFINE-FMD study. Led by Dr. Jason Kovacic and initiated in 2012, the DEFINE-FMD study is aiming to use disease-relevant samples from FMD patients and matched healthy control subjects to DEFINE the molecular and cellular basis of FMD. In the DEFINE-FMD study, we are collecting DNA, plasma, serum, fibroblast cells (via skin punch biopsy) and culture supernatant from rigorously phenotyped FMD patients with multifocal disease and healthy controls. Healthy controls are also carefully screened, and are matched to FMD patients by gender, race/ethnicity, age, smoking status, body mass index and number of anti-hypertensive medications. As of July 2019, 380 subjects have been enrolled and recruitment is ongoing.

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11:00 AM-12:00 PM
MR5 3005

Hosted by: Clint Miller, PhD
Refreshments Served