

Penn Medicine Awards 2016 MDBR Research Grant Awards for Snyder-Robinson Syndrome

The University of Pennsylvania Health System (Penn Medicine) announced the recipients of the 2016 Million Dollar Bike Ride (MDBR) research grants. Two of these grants were awarded for Snyder-Robinson research. Congratulations to Dr. Schwartz and Dr. Fierro!

Research Award #1

Charles Schwartz, PhD

Greenwood Genetic Center

Snyder-Robinson Syndrome pilot for \$53,000

Title: Role of Mitochondria and ROS in Etiology of SRS

Dr. Schwartz's research will explore the possibility that SRS patients may have a mitochondrial dysfunction. The following research objectives will take place, as a result of this study: 1) Determine the levels of calcium within the cell and the mitochondria of patients and controls, 2) Mitochondrial function will be determined by how well certain substrates are utilized by the mitochondria of patients and controls and measure the level of ROS, and 3) Examine if compounds affecting calcium balance, ROS accumulation and mitochondrial transport can reduce the metabolic phenotype observed in patient cells.

Research Award #2

Fernando Fierro, PhD

University of California, Davis

Snyder-Robinson Syndrome pilot for \$53,000

Title: Modelling Osteogenesis of Snyder-Robinson Patients by targeting Spermine Synthase using CRISPR/Cas9 in Human Bone Marrow Derived Mesenchymal Stem Cells

Dr. Fierro's research will explore why MSC with a dysfunctional SMS gene cannot become normal, mature osteoblasts. This study will study the cells harboring the SRS mutations in the context of bone formation, and compare them to MSC without mutations. The aim will be to identify with precision what alterations occur, in order to identify potential targets to treat the bone defects of SRS patients.

The full announcement from Penn Medicine, including research abstracts, can be found here:

<http://www.med.upenn.edu/orphandisease/odc-grants-awarded.html>