Duchenne muscular dystrophy (DMD) is an X-chromosome-linked genetic disease that is caused by a mutation in the dystrophin gene and affects 1 in every 3500 boys. DMD patients suffer progressive muscle wasting and eventual cardiorespiratory failure that results in an early death in the second or third decade of life. Although extensive research effort has been invested, lack of a good mouse model that mimics the cardiac failure hinders research. We have developed a novel mouse model that exhibit all the symptoms found in DMD patients and our research is aimed at understanding the cardiac failure in DMD for future therapeutic interventions. Our mouse model fully recapitulates the DMD symptoms because we also took into account of the size of human protection DNA on chromosomal ends (telomere) compared to...
mouse. We would like to study the cause of cardiac failure in our mouse model by 1) determine if telomere shortening is specific to cardiomyocytes, 2) evaluate the level of cellular damage caused by oxidative stress and 3) identify the source of oxidative stress. These experiments will help us to better understand cardiac failure in DMD patients and allow testing of therapeutic interventions.

PROJECTS

- Role of Telomere Erosion in Lethal Cardiomyopathy in Duchenne Muscular Dystrophy - Stanford University
- Development of Biomarkers for Duchenne Dilated Cardiomyopathy

Publications

PUBLICATIONS

- Telomere shortening is a hallmark of genetic cardiomyopathies. Proceedings of the National Academy of Sciences of the United States of America

- Apelin and APJ orchestrate complex tissue-specific control of cardiomyocyte hypertrophy and contractility in the hypertrophy-heart failure transition. American journal of physiology. Heart and circulatory physiology

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- A Notch-dependent transcriptional hierarchy promotes mesenchymal transdifferentiation in the cardiac cushion. Developmental dynamics : an official publication of the American Association of Anatomists

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**PRESENTATIONS**

- Telomere and mitochondrial dysfunction in Duchenne Muscular Dystrophy. - 2014 EMBO conference on Telomeres, Telomerase and Disease (April 20, 2014)
- Telomere and mitochondrial dysfunction in Duchenne Muscular Dystrophy. - AHA - Basic Cardiovascular Sciences 2015 Scientific Sessions (7/13/2015)
- Telomere shortening as a hallmark of lethal dilated cardiomyopathy. - Weinstein Heart Conference 2016 (5/21/2016)
- Telomere shortening as a hallmark of lethal dilated cardiomyopathy - Stanford-Karolinska Cardiovascular Symposium (October 20, 2016)