Exploration of a Novel Biomarker for Thoracic Aortic Aneurysm and Dissection

LDS GRANT AWARDED TO DR. BART LOEYS GRANT RUNS FROM JULY 1, 2022 – JUNE 30, 2024

What is a biomarker?

A biomarker is a measurable substance in blood whose presence is indicative of a normal or abnormal process, or of a condition or disease.

During previous research of the expression profiles of genes in the aneurysmal aortic wall of three different Marfan and Loeys-Dietz syndrome mouse models, a gene was identified that was highly expressed in all three models. This protein is a novel player in the pathogenesis of thoracic aortic aneurysm and its highest expression is in the aortic wall.

Why is biomarker research important?

Imaging of the aorta (MRI, MRA, CT Scan) is the current method to determine rate of aortic root enlargement and aortic risk. At present, there are no biomarkers that could predict the existence of a thoracic aortic aneurysm or the occurrence of a thoracic aortic dissection.

THE IDENTIFICATION OF SUCH A BIOMARKER WOULD BE OF GREAT HELP TO:



Determine if new therapeutics are helping in the reduction of aortic enlargement.



Find methods to validate effectiveness of new drugs and make clinical trials less long and less expensive.



Develop an easy, efficient and effective blood test to monitor aortic health.

What will be done during the study?



INVESTIGATE if the serum levels of this protein in Marfan and Loeys-Dietz mouse models corresponds to the thoracic aortic aneurysm severity and progression.

GATHER EVIDENCE in serum samples of patients with thoracic aortic aneurysm and dissection to validate if this protein is also in humans and can possibly be used as a biomarker for thoracic aortic aneurysm and dissection.

IDENTIFY THE CELLULAR SOURCE of this protein in Marfan and Loeys-Dietz patients through a stem cell-derived model and understand the mechanisms that cause this protein to increase for future therapeutic targets.

Do we anticipate additional research to be continued after the grant cycle is completed?

If aim is successful, study would need a longer project with many additional samples to correlate blood protein level with thoracic aortic aneurysm progression and severity. If validated, this protein level could be used to test therapeutic compounds with the "simple" measurement of the blood levels and those therapeutics that diminish the protein level would be beneficial for the treatment of aortic aneurysm.

How will this help LDS patients?

Our hope is that this research can prove that the level of this protein in the blood can give an indication of aortic health and possibly be used as predictor of aortic dissection. This research could also be used to help determine if new therapeutics are helping in the reduction of aortic enlargement, which is greatly needed for drug companies to make clinical trials less long and less expensive.

Be a Part of Advancing Hope

Funding for this research came from the Loeys-Dietz syndrome community. Through individual donations of all sizes, we are working together to advance hope for Loeys-Dietz syndrome patients, families, and physicians. This study is supported by the hope and trust of our community. Thank you!

Donate Now





Contact Angela Crist at ACrist@marfan.org.