American Heart Association awards $300,000 grant to Masonic Medical Research Institute

Maria Kontaridis, Ph.D., to study whether some genes could cause autism and congenital heart defects

For immediate release

Utica, December 28, 2020 - Is there a link between cardiac abnormalities and autism?

That's what Dr. Maria Kontaridis, Executive Director and Gordon K. Moe Professor of Biomedical Research and Translational Medicine at the Masonic Medical Research Institute (MMRI) in Utica, will be studying for the next three years, thanks to a $299,835.00 three-year Transformational Project Award from the American Heart Association. On Jan. 1, Dr. Kontaridis and her co-Investigator, Dr. Gulhan Ercan-Sencicek, an Instructor at the MMRI, will begin their project, titled "The role of *PTPN11* mutations in autism and heart pathogenesis," to understand how mutations in the same gene differentially affect the normal processes of heart and brain development.

"We think there are genes that have dual functionality in development, the abnormal regulation of which can cause neurocognitive and cardiac anomalies," Dr. Kontaridis said. "Genes regulate signaling relays within a cell that tell that cell what to do, whether it be to grow, divide, or die. Here, we are looking at the effects of a specific gene, which, when mutated, alters the relay process, thereby leading to the development of growth defects, autism, and cardiac abnormalities."

In particular, Drs. Kontaridis and Ercan-Sencicek will look at disease-causing changes in the *PTPN11* gene, a nodal gene involved in critical signaling processes that regulate normal growth and differentiation of cells in multiple tissues, including brain and heart.

"We have identified two novel mutations that we think link autism with heart abnormalities in human patients," Dr. Ercan-Sencicek said. "To understand the role of these mutations, we will reprogram somatic cells obtained from patients with these unique mutations and convert them into inducible pluripotent stem cells (iPSCs), cells that have the ability to differentiate into any tissue type of interest. For this project, we will differentiate these iPSCs into heart muscle cells and brain organoid cells to study the effects of *PTPN11* mutations on heart disease and brain development. These results will help us to identify potential novel therapeutics that can be used to treat patients."

"It's our honor to award this grant to Dr. Kontaridis," said Steve Gassner, chair of the Board of Directors of the American Heart Association in the Mohawk Valley. "We know that 1 in 100 children is born with a heart defect, and some of those children have been our Red Cap Ambassadors. Like heart disease, autism affects the whole family. We look forward to hearing from Drs. Kontaridis and Ercan-Sencicek in the coming years as progress continues on this exciting and ground-breaking work."

Additional Resources:

- www.heart.org

- www.mmr.edu

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About the American Heart Association
The American Heart Association is devoted to saving people from heart disease and stroke – the two leading causes of death in the world. We team with millions of volunteers to fund innovative research, fight for stronger public health policies, and provide lifesaving tools and information to prevent and treat these diseases. The Dallas-based association is the nation's oldest and largest voluntary organization dedicated to fighting heart disease and stroke. To learn more or to get involved, call 1-800-AHA-USA1, visit heart.org or call any of our offices around the country. Follow us on Facebook and Twitter.

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