



Professional Day

Friday, August 2, 2019

Updates on Research in CHARGE

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Presenter Information

Dr. Martin is Interim Chair and Professor of Pediatrics at The University of Michigan Medical School. She is also a Medical Geneticist and serves as Chair of the Scientific Advisory Board for the CHARGE Syndrome Foundation. She and her husband, Yehoash Raphael, Professor of Otolaryngology - Head and Neck Surgery at The University of Michigan Medical School, have a 24 year-old son, Noam, who has CHARGE. Their laboratories study mechanisms of CHD7, the gene involved in CHARGE, using human genetics and mouse models. They focus on understanding and treating symptoms related to the nervous system, including hearing loss and balance disorders.

Presentation Abstract

The CHARGE Syndrome Foundation is dedicated to promoting new knowledge and discoveries that positively impact individuals with CHARGE. Toward this end, the foundation provides funding to support a wide variety of basic science and clinical research, including Davenport fellowships, biennial symposia, human genetics, gene discovery, animal models (mice, fish, flies), and stem cells. In 2004, CHD7 was identified as the major gene involved in CHARGE, thus opening new opportunities to understand CHARGE mechanisms and develop therapies. In 2012, the Foundation established an annual pilot research award program to support individual researchers working on CHARGE-related studies. In 2014, the foundation established biennial symposia held in Ann Arbor at The University of Michigan to promote basic science discoveries in CHARGE. In this presentation, we will provide an overview of the past seven years of foundation supported pilot awards, and the three symposia. We will discuss the impact of these activities on our understanding of CHD7 and on the lives of individuals with CHARGE. We will also present an overview of new innovative technologies and progress toward development of therapies.

Learning Objectives

- Review research funding supported by the CHARGE Syndrome Foundation since 2012.
- Present current research supported by the NIH on CHD7 and CHARGE.
- Discuss active areas of investigation on understanding mechanisms and progress toward therapies related to CHD7 and CHARGE