"New insights and advances in CHARGE syndrome: Diagnosis, etiologies, treatments, and research discoveries"
By Conny van Ravenswaaij-Arts and Donna M. Martin
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AUTHORS AND THEIR CONNECTION TO THE CHARGE SYNDROME FOUNDATION:

Conny van Ravenswaaij-Arts, MD, PhD, is a Clinical Geneticist at the University Medical Center Groningen, Netherlands, where she is head of a CHARGE Center which serves the entire European Union. She is a member of the team which identified *CHD7* as the major cause of CHARGE syndrome in 2004.

She and others from her team have been attending the biennial CSF conferences since 2005, where they have presented research findings, conducted research, collaborated with other researchers and met with families. Conny is on the CSF Scientific Advisory Board and is involved with many research projects on CHARGE and *CHD7*.

Donna Martin, MD, PhD, is Professor of Pediatrics and Human Genetics at the University of Michigan Medical School in Ann Arbor, Michigan. Her expertise includes evaluating and caring for individuals with CHARGE syndrome as well as research in mouse models of CHARGE and other developmental disorders of the nervous system.

Donna is a founding member of the Scientific Advisory Board of the CSF, which has awarded over a million dollars in research grants to date. She mentors graduate students and residents in medical genetics.

SUMMARY OF THE PAPER:

This paper starts with a historical perspective on CHARGE, a summary of the current state of research into *CHD7* and chromatin remodeling and an introduction to the 11 other manuscripts in the special issue. The authors note that with the availability of new clinical genetic testing (genome-wide screening), more and more people are being identified with *CHD7* variants. Whether or not they would all be considered to have "CHARGE syndrome" is up for debate. Is the best way to diagnose CHARGE by molecular testing, clinical findings, or some combination?

Animal models (mice, zebrafish, frogs, fruit flies) are helping us to understand the ways that variants in *Chd7* cause various features seen in people with CHARGE. Studies done at the cellular level may eventually help lead to treatment of some features of CS.

As an introduction to the other papers in this issue, the authors mention advancements in knowledge central nervous system, craniofacial, auditory and vestibular structures), neural crest, heart, immune system, endocrine system, gastrointestinal system and behavior. There is a summary of the features noted in 119 individuals with CHARGE in one paper and a summary of genetic diagnosis and counseling recommendations in another. All of these papers will be summarized in upcoming blasts.

The paper includes a table showing the frequency of features of CHARGE in individuals with and without known *CHD7* variants. There is also a table of the *Chd7* mouse model strains developed to date and an extensive discussion of the value of these models for learning more about CS. Finally, there is a discussion of what the future may hold for CHARGE research, including the roles of stem cells and CRISPR/Cas9 editing technology may play in eventually developing treatments.

WHAT DOES THIS MEAN TO FAMILY/PERSON WITH CHARGE?
Not a lot directly at this point in time. However, as the authors state, "Together, these successes paint an optimistic picture for the future of research and discovery in CHARGE syndrome. We hope that additional exciting breakthroughs will be made that ultimately translate to improvements for the lives of individuals with CHARGE syndrome and their families."

SHOULD I READ IT? SHOULD ONE OF MY DOCTORS READ IT?
If you are into the history of CHARGE and/or molecular genetics and animal models of genetic conditions, then yes, you will want to read the whole paper. Your doctors are more likely to be interested in individual papers more closely related to their specialties.

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