



Poster Presentation

An Interactive Seminar in Genetics: All You Want To Know About the Genetics of CHARGE Syndrome

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Presenter Information

Christa de Geus is a medical doctor in training to become a clinical geneticist. Within clinical genetics she has a particular interest in neurology and neuroradiology. In 2014 she joined the research group of Prof. Conny van Ravenswaaij as a PhD student. Her PhD focuses on neurological symptoms in CHARGE syndrome.

Presentation Abstract

After a short introduction, all questions of the participants regarding genetics, the CHD7 gene, mutations, recurrence risk, et cetera, will be answered using instructive illustrations. A handout that explains the main issues for a lay audience will be provided. We will also present the results of our recent update of the CHD7.org website. CHD7.org aims to be a comprehensive source of clinical and molecular genetic information on known CHD7 mutations. Based on the experience of previous years, we will offer parents the opportunity to discuss the specific genetic test results of their child with us in short sessions after the seminar.