



CHARGE SYNDROME GENETICS RESEARCH

Purpose of the study

Not all cases of CHARGE Syndrome are caused by mutations in the *CHD7* gene. We are interested in finding new genetic causes of CHARGE.

Participation Requirements

1. At least one individual in the family must have clinical features of CHARGE Syndrome.
2. Children of any developmental age can participate (we have cheek swabs for children who may have difficulty producing enough saliva).

Nature of participation

We are collecting saliva samples from individuals with CHARGE and their parents and siblings. We are asking families to participate in the CHARGE Syndrome Clinical Database Project (CSCDP) so that we can obtain clinical information on each participating family. DNA will be collected from the saliva and sequenced at The University of Michigan.

Time involved

Each family member will need to spend 10-15 minutes donating saliva in a cup, and another 15-20 minutes completing a clinical history/questionnaire, for a total of up to 30 minutes.

Contact information

Dr. Donna Martin (734) 645-3564/donnamm@umich.edu, Dr. Stephanie Bielas (858) 525-1757/sbielas@umich.edu or Ms. Jennifer Skidmore (734) 678-0463/camelot@umich.edu can provide information on how to participate in this research.