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. Approved by University of Michigan IRB MED. Study ID: HUM00032360 We do not have an end date for this study.

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 can provide cheek swabs for children who may have difficulty producing enough saliva).

Nature of participation: After consent is obtained, we will send you a kit to collect 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 tube, and another 15-20 minutes completing a clinical history/questionnaire, for a total of up to 30 minutes. Saliva collection kits will be mailed to participants.

Contact information: contact us for more information and to begin the consent process

Dr. Donna Martin (734) 645-3564/donnamm@umich.edu

Dr. Stephanie Bielas (858) 525-1757/sbielas@umich.edu

Ms. Jennifer Skidmore (734) 678-0463/camelot@umich.edu