3D imaging of people with CHARGE and their family members

Sheri Riccardi sheri.riccardi@ucdenver.edu

Who is eligible: all individuals

- Definite CHARGE syndrome (is your information in CSCDP?)
- First degree relatives (parents, siblings) of individuals with CHARGE

How the research will be conducted at conference

- Answer some some questions related to CHARGE (is your info in CSCDP?)
- Have 3D photo taken Estimated time: 10-15 minutes



Human Medical Genetics and Genomics Program

SCHOOL OF MEDICINE

UNIVERSITY OF COLORADO ANSCHUTZ MEDICAL CAMPUS 12800 E. 19th Ave. Mail Stop 8300 RC1 North | Room 3107 Aurora, CO 80045 | USA O: (303)724-3101 F: (303)724-3100 www.ucdenver.edu/humanmedicalgenetics

International Study of Genetic Syndromes that Include Facial Abnormalities

We are carrying out an international study to better characterize genetic syndromes that include facial abnormalities, including CHARGE Syndrome. We hope you will take part in this study.

We are building a database of special three-dimensional (3D) facial photographs of people with different such syndromes that can be used to precisely measure and define facial abnormalities and understand the underlying biological problems. This will become part of the National Institutes of Health "FaceBase" database (https://www.facebase.org/), and will be available to assist other qualified scientists who are working to better understand these syndromes. Additionally, this syndrome database will be used to help develop a computerized system to aid physicians in diagnosing these syndromes in the clinic.

If you or your child join the study, we will then collect some information, we may collect some relevant medical records, and we will take a 3D photograph, which will provide computerized information about the shape of your/his/her face. Total participation time is about 10-15 minutes.

This study is funded by the National Institutes of Health and has been approved by the Colorado Multiple Institutional Review Board. We hope you will consider taking part.

Study Director:

Richard A. Spritz, M.D. Professor and Director, Human Medical Genetics & Genomics Program University of Colorado School of Medicine 12800 E. 19th Ave., Rm. 3100, MS8300 Aurora, CO 80045 USA

Tel. 303-724-3108 FAX 303-724-3100 Email richard.spritz@ucdenver.edu

Study Coordinator:

Sheri Riccardi, BS Study Coordinator, Human Medical Genetics& Genomics University of Colorado 12800 E. 19th Ave., MS8300 Aurora, CO 80045 USA

Tel. 720-839-2802 FAX 303-724-3100 Email sheri.riccardi@ucdenver.edu