

Stickler Involved People and the University of Colorado School of Medicine are partners for groundbreaking research at our July 11-13 2014 Denver SIP conference.

International Study of Genetic Syndromes that Include Facial Abnormalities

The SIP Board of Directors agreed to participate in an international study to better characterize genetic syndromes that include facial abnormalities, including Stickler Syndrome. We hope you will take part in this study.

They 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 joins the study, the University of Colorado Team will then collect some information, they may collect some relevant medical records, and then they 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.

This project will be supervised by: Dr. Richard Spritz, Professor and Director, Human Medical Genetics and Genomics Program, University of Colorado School of Medicine

NOTE: Participants need documentation of diagnosis (molecular whenever possible; if you have had testing but don't have the report, then they will need info as to where it was done so they can ask for that item of medical records). The photo session will take ~5 minutes per person and you will be selected during Saturday's presentations.

Stickler
Involved People