3D Analysis of Normal Facial Variation: Data Repository and Genetics

Principal Investigators: Weinberg, Marazita

Co-Investigators: Heike, Cunningham, Hecht

Grant Number: 1U01DE020078

FaceBase Annual Meeting: May 1-3, 2013

CCDG
CENTER FOR CRANIOFACIAL & DENTAL GENETICS

FaceBase
A Resource For Craniofacial Researchers

Restatement of Aims

Goal 1: To construct a web-based normative repository of 3D facial measurements and images

1a) Ascertain 3500 European Caucasian individuals – age 3 to 40 – from multiple sites: Pittsburgh, Seattle, and Houston (and Iowa!)

1b) Acquire 3D facial surface images, basic demographic descriptors and saliva samples from each participant

1c) Extract quantitative data from 3D surface images including landmarks and linear measurements

1d) Deposit clean data (including images and measures) into a web-based repository accessible through FaceBase.org (*3D Facial Norms*)

Goal 2: To investigate the genetic basis of variation in facial shape

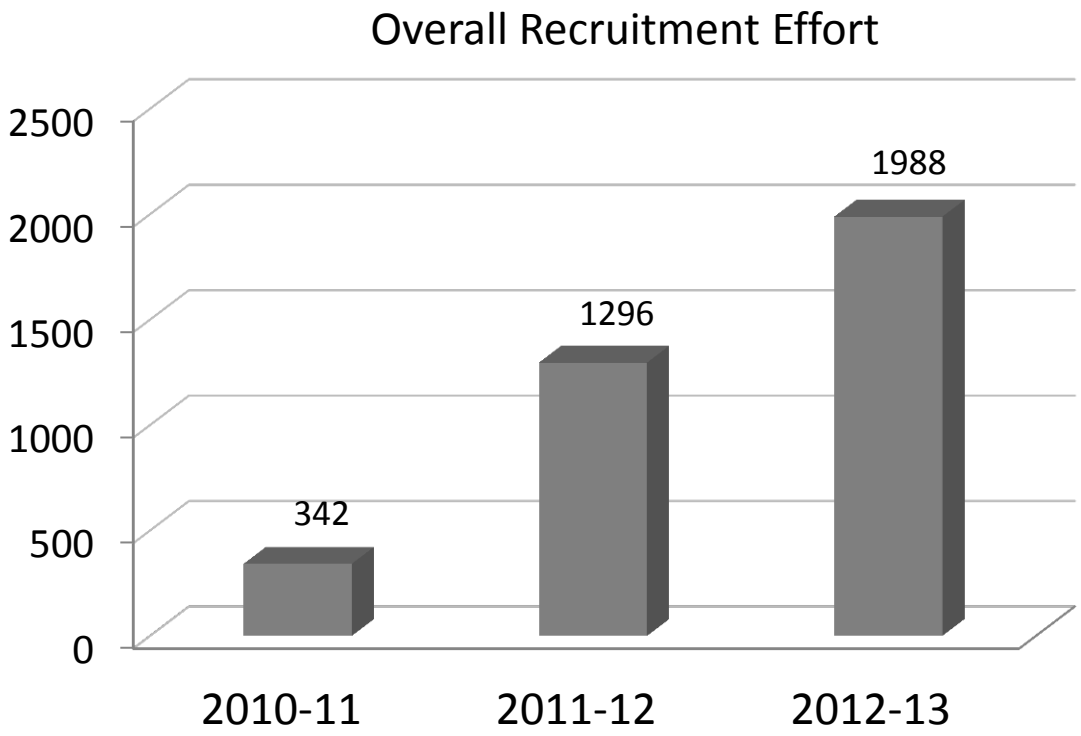
2a) Submit saliva samples for genotyping (proposed year 5)

2b) Perform morphometric analyses on 3D data set to derive shape variation descriptors

2c) Identify genetic variants associated with normal facial shape variation

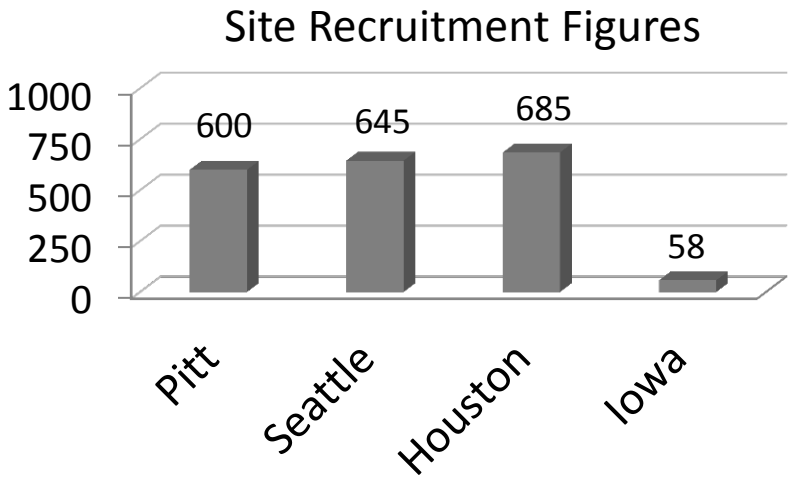
2d) Submit genotype data and results to FaceBase.org

Progress over the Past Year

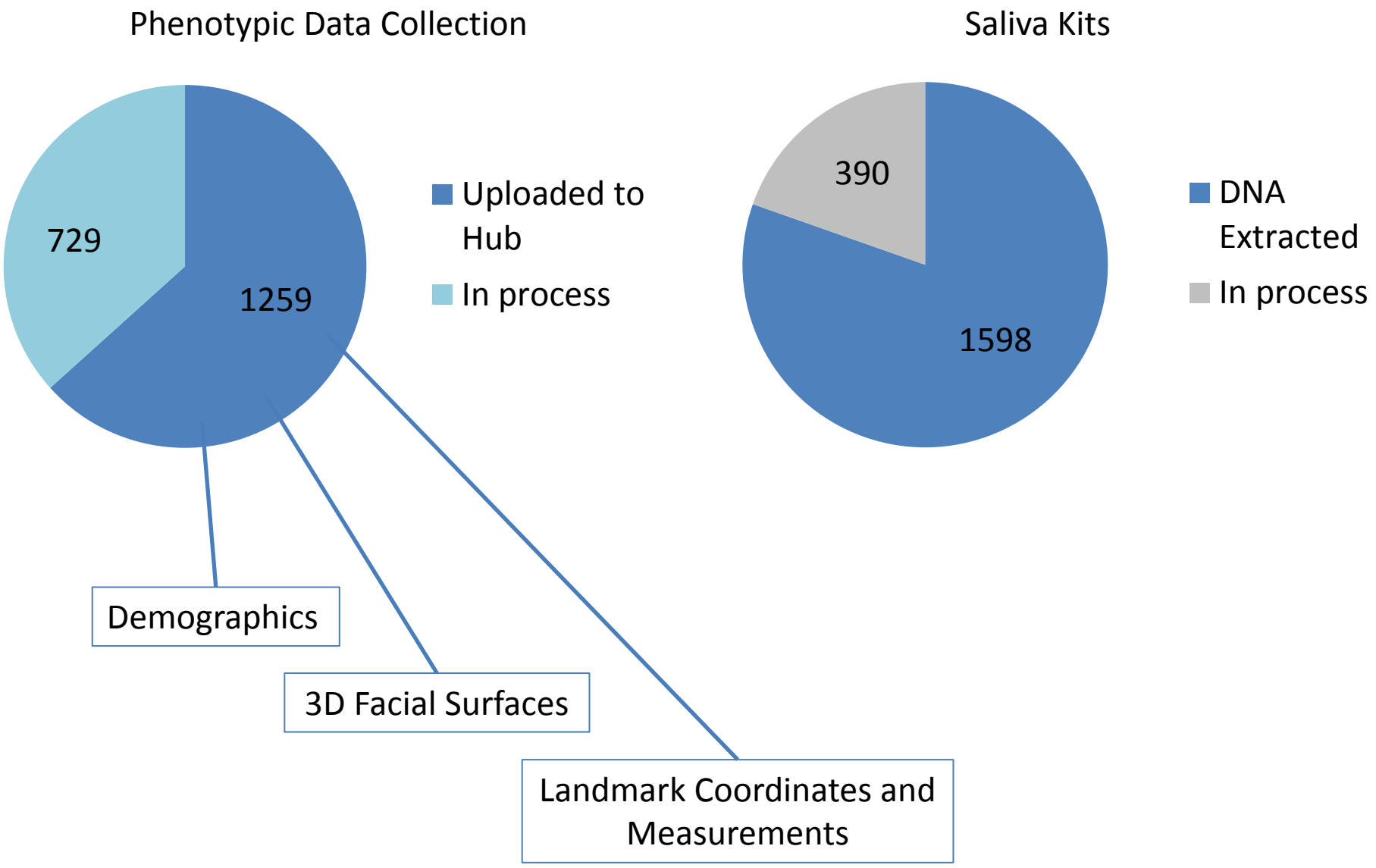


3D Facial Norms Repository

Recruitment Target: 3500 healthy unrelated individuals age 3-40



Progress over the Past Year



3D Facial Norms Web-Interface

FaceBase

USERNAME OR E-MAIL * PASSWORD *

LOG IN

Create new account Request new password

HOME | PROJECTS | DATA | GENOME BROWSER | FORUMS | RESOURCES | SEARCH | BIOREPOSITORY

YOU ARE HERE: Home » 3D Facial Norms Database

3D Facial Norms Database

Overview | 3D Database Search | Summary Statistics | Data Anaylsis | Tools | Technical Notes

3D DATABASE SEARCH

Explore the 3D Norms Database through a customizable search interface. This is the main portal for querying and downloading individual-level phenotype and genotype data based on user-defined parameters. Phenotypic data include 3D facial landmark coordinates, anthropometric facial measurements, demographic descriptors, and 3D facial surface models.

GO →

TOOLS

A collection of practical web applications for the clinical and research community.

GO →

SUMMARY STATISTICS

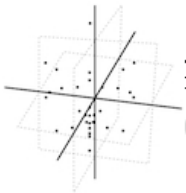
View normative age- and sex-based averages for selected craniofacial measurements and 3D facial surface models.

GO →

TECHNICAL NOTES

Detailed background information on all aspects of the 3D Facial Norms Project.

GO →



DATA ANALYSIS

A compendium of results from past analyses based on the 3D Facial Norms dataset.

GO →

ABSTRACT PAGE

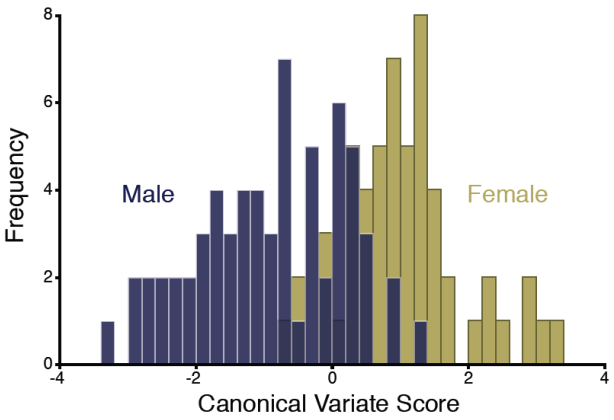
GO →

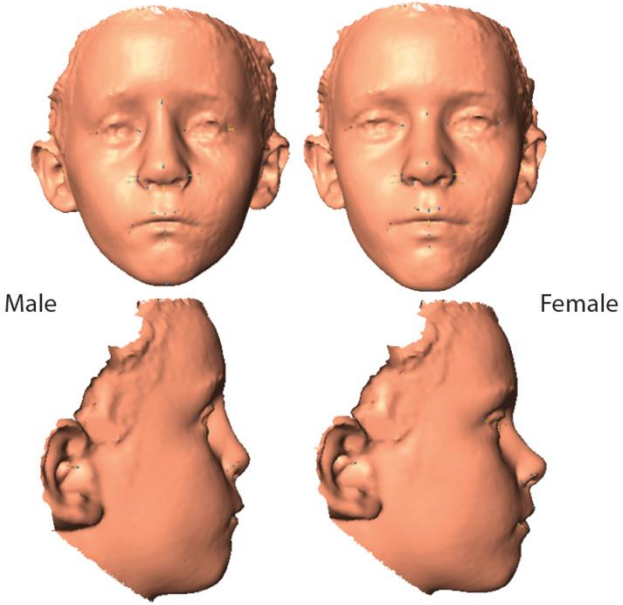
Using the 3D Norms Data

- Analyses of human craniofacial variation and growth patterns
- Normative control data for craniofacial comparisons
- Development of novel image analysis methods
- Genomic studies of craniofacial traits

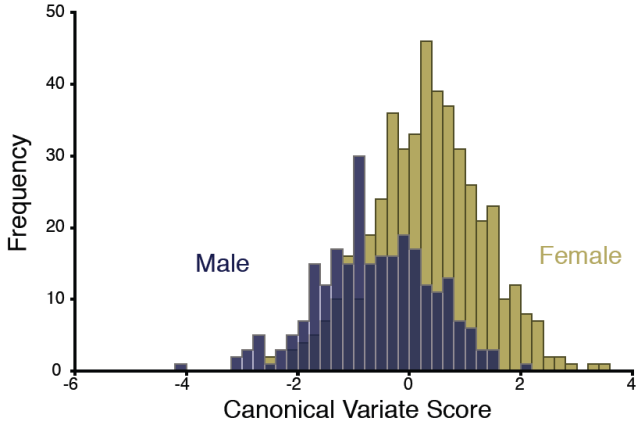
Sexual Dimorphism in facial shape across the lifespan

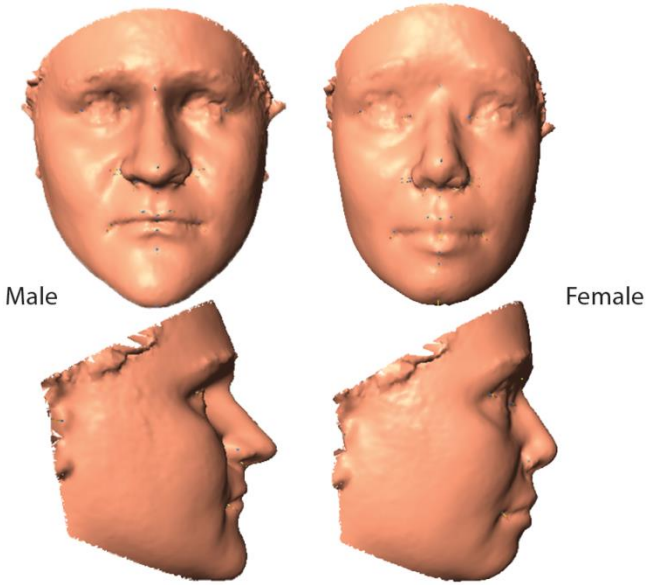
Juveniles 3-10 (n = 119)



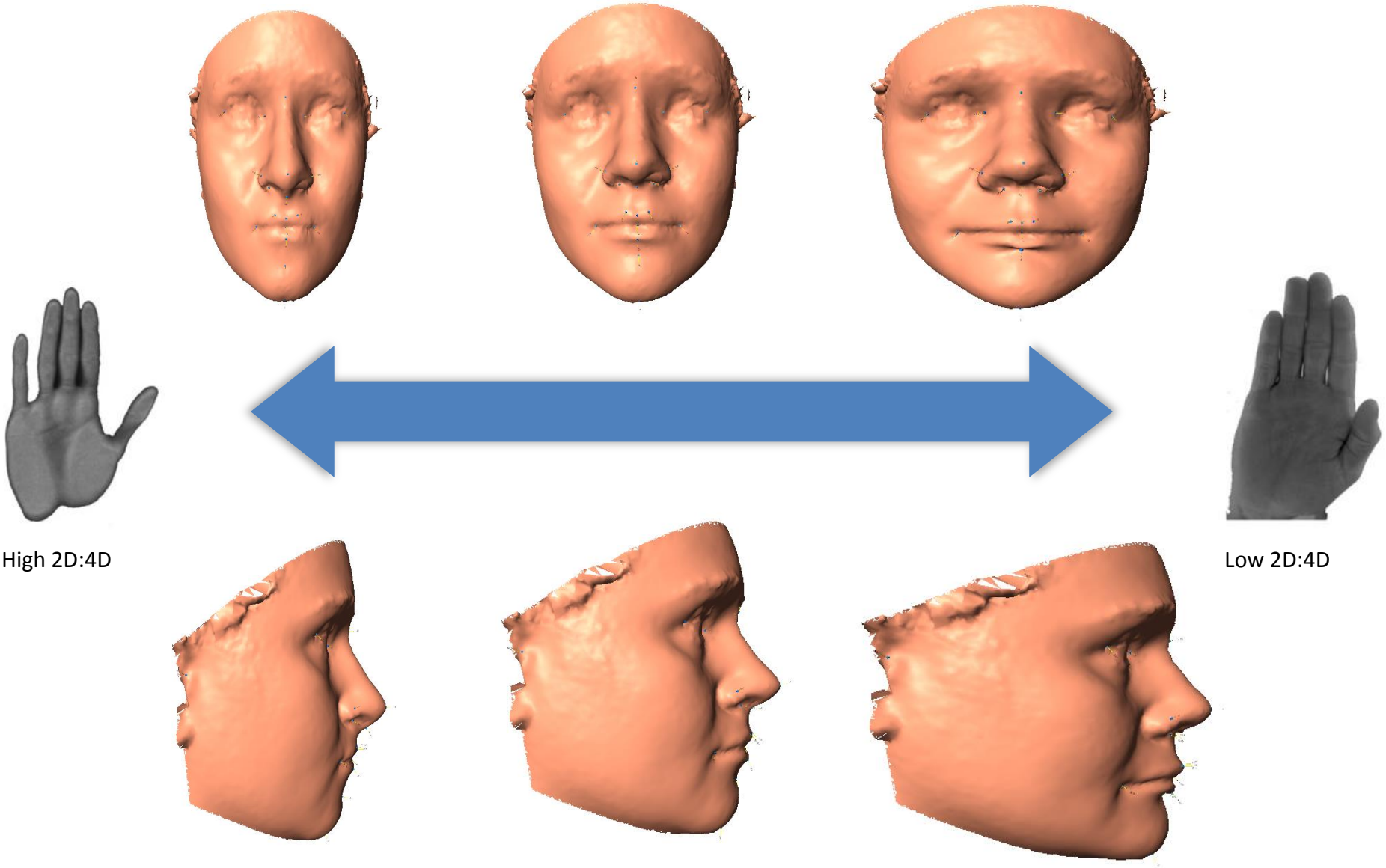


Adults 18-40 (n = 722)





Relationship between facial shape and digit ratio in males



High 2D:4D

Low 2D:4D

Using the 3D Norms Data

Analyses of human craniofacial variation and growth patterns

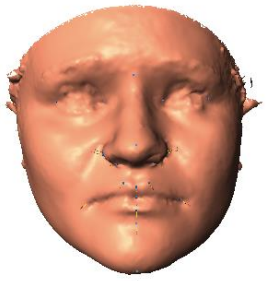
Normative control data for craniofacial comparisons

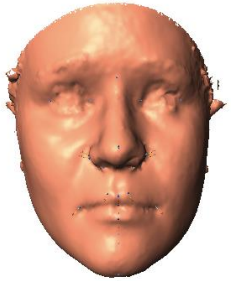
Development of novel image analysis methods

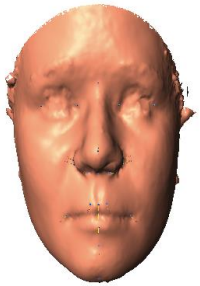
Genomic studies of craniofacial traits

Assessment of craniofacial dysmorphology in Cutis Laxa

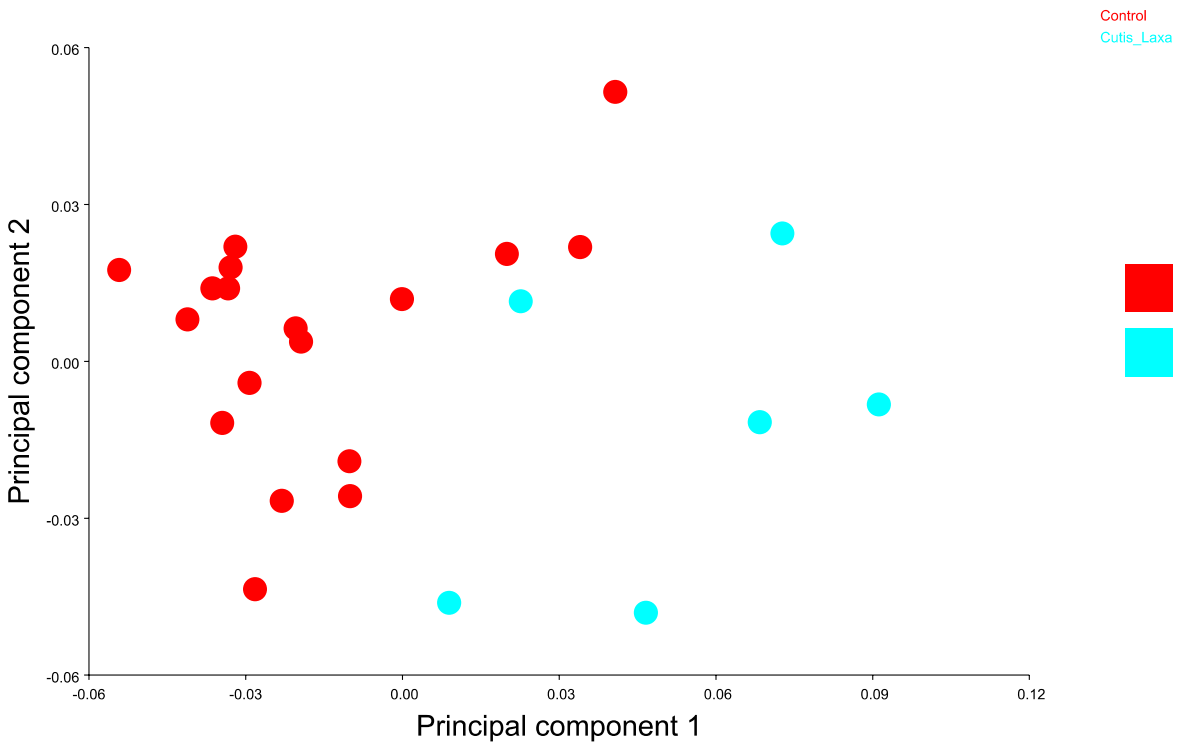
Control Morph

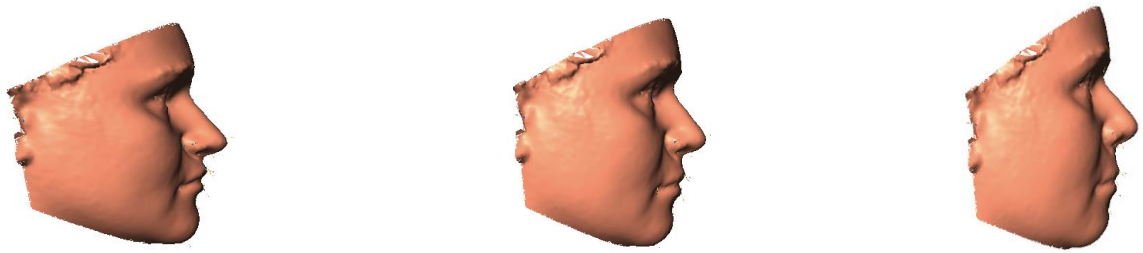




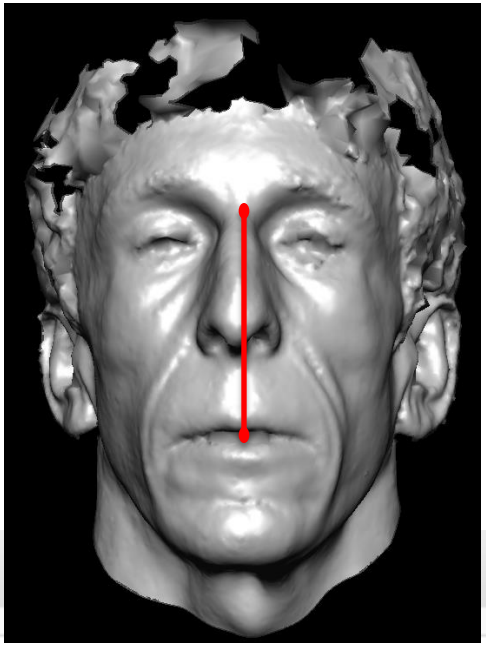


Cutis Laxa Morph





Using Z-Score Calculator Tool to compare upper facial height



MEASUREMENT	VALUE		Z-SCORE	
Lower Facial Depth Left		mm		σ
Morphological Facial Height		mm		σ
Upper Facial Height	92.10	mm	2.02	σ

Using the 3D Norms Data

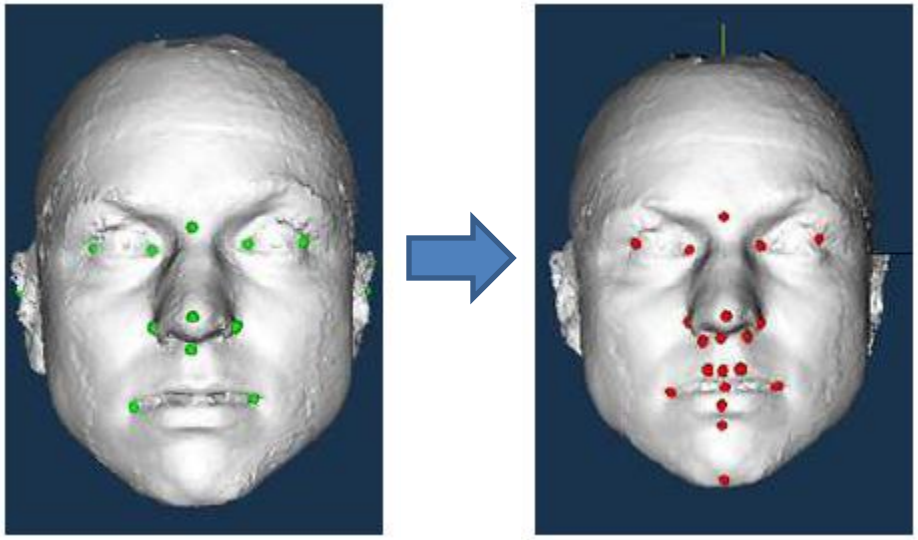
Analyses of human craniofacial variation and growth patterns

Normative control data for craniofacial comparisons

Development of novel image analysis methods

Genomic studies of craniofacial traits

Automated landmarking of facial surfaces



Step 1: Initial Identification of facial landmarks using geometric method

Step 2: Deformable registration – improves fit and adds additional landmarks

Compared to manual landmark placement

Point Name	Geometric Method Average Distance(mm)	Deformable Method Average Distance(mm)
Nasion	——	2.92±1.62
Pronasale	1.29±0.68	1.59±0.81
Subnasale	2.35±2.16	2.45±0.80
Alare(R)	3.24±2.61	1.78±1.15
Alare(L)	3.14±2.41	3.07±1.15
Labiale Superius	——	2.27±1.15
Stomion	——	1.49±0.90
Labiale Inferius	——	2.27±1.41
Sublabiale	——	3.17±1.87
Subalare(R)	——	2.36±1.06
Subalare(L)	——	1.59±0.93
Crista Philtri(R)	——	2.31±1.27
Crista Philtri(L)	——	1.99±1.03
Chelion(R)	3.14±2.41	3.08±2.14
Chelion(L)	2.80±2.38	3.08±1.64
Gnathion	——	5.31±3.54
Endocanthion(R)	4.78±1.45	2.39±1.09
Endocanthion(L)	4.58±1.70	2.78±1.50
Exocanthion(R)	3.15±2.21	3.34±1.63
Exocanthion(L)	2.72±1.86	3.68±1.91

Using the 3D Norms Data

Analyses of human craniofacial variation and growth patterns

Normative control data for craniofacial comparisons

Development of novel image analysis methods

Genomic studies of craniofacial traits

Genotyping Strategy

- CIDR application planned for summer
- Goal is to have 2200-2500 samples in hand at the time of application
- Key factor in thinking about genotyping: **what is most useful for the FaceBase user community**
  - A genome-wide panel probably makes most sense for FaceBase users
    - Illumina Core Panel (300k SNPs) + Exome, or…
    - OmniExpress Panel (750k SNPs) + Exome, or…
    - Omni 2.5 Panel (2.5mil SNPs) + Exome
  - Additional custom panel for testing relevant SNPs from recent analyses
  - Replication and extension dataset
  - Meta-analysis/mega-analysis dataset

Acknowledgements

Funding for this project provided by the following grant from the National Institute of Dental and Craniofacial Research: U01-DE20078

Pittsburgh Team
- Seth Weinberg
- Mary Marazita
- Pooja Gandhi
- Zach Raffensperger
- Trish Parsons
- Annette Kreg-Jensen
- Cristy Spino
- Wendy Carricato

Seattle Team
- Carrie Heike
- Michael Cunningham
- Laura Steuckle
- Linda Peters
- Erik Stuhaug
- Trylla Tuttle
- Eden Palmer

Houston Team
- Jacqueline Hecht
- Rosa Martinez
- Robert Plant
- Chung How Kau

Iowa Team
- Jeff Murray
- Nichole Nidey