Shape-based Quantification of 3D Face Data for Craniofacial Research

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General Exam

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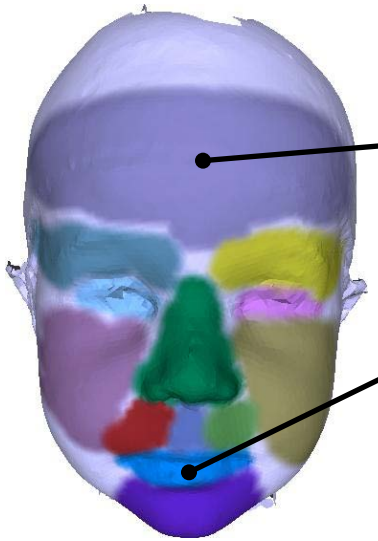
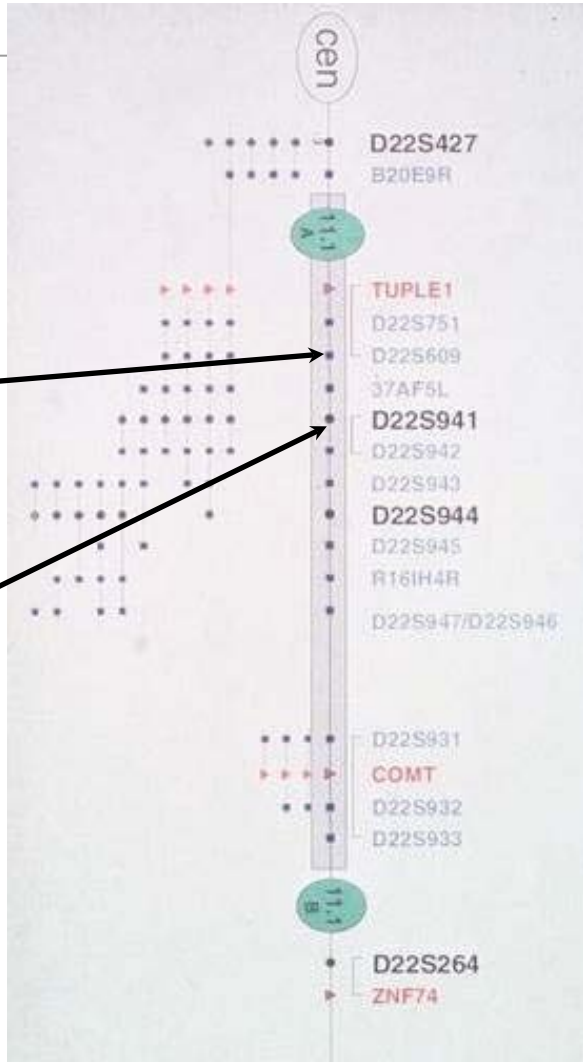
22q11.2 Deletion Syndrome

Motivation



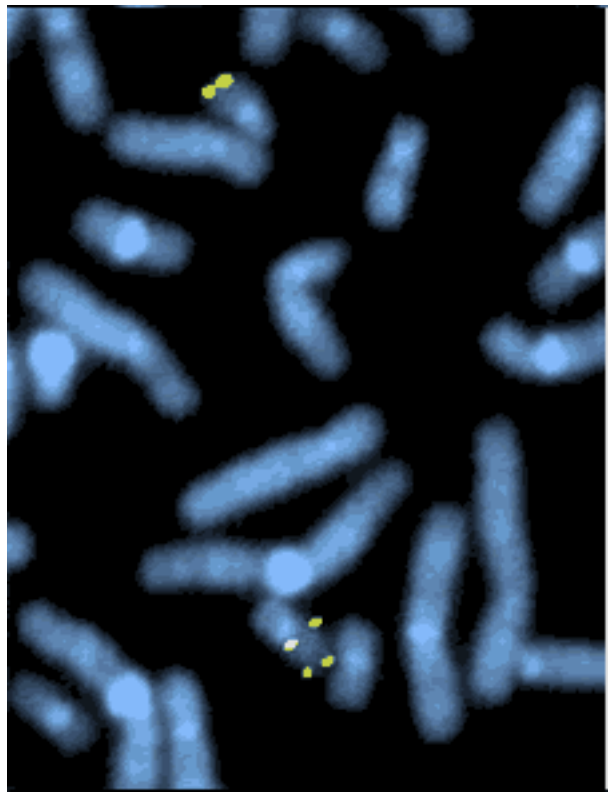


- Help detect disease more accurately

- Describe useful features

22q11.2 Deletion Syndrome (22q11.2DS)

- aka Velo-cardio-facial syndrome (VCFS)
- affects approximately 1 in 4000 individuals in the US
- early detection is important
  - cardiac anomalies
  - mild to moderate immune deficiencies
  - learning disabilities



genetic test for 22q11.2DS

22q11.2 Deletion Syndrome has Subtle Facial Features

VELO-CARDIO-FACIAL SYNDROME
Specialist Fact Sheet

Velo-cardio-facial syndrome (VCFS), also known as ***Shprintzen syndrome***, ***DiGeorge sequence or syndrome***, and ***22q11 deletion syndrome***, is caused by a deletion of a small segment of the long arm of chromosome 22. It is one of the most common genetic disorders in humans. The following list shows the anomalies that have bee findings have a 100% frequency, but all occur with sufficient frequency to warrant assessme check the web site of **The Velo-Cardio-Facial Syndrome Educational Foundation, Inc.** a

Craniofacial/Oral Findings

1. Overt, submucous or occult submucous cleft palate
2. Retrognathia (retruded lower jaw)
3. Platybasia (flat skull base)
4. Asymmetric crying facies in infancy
5. Structurally asymmetric face
6. Functionally asymmetric face
7. Vertical maxillary excess (long face)
8. Straight facial profile
9. Congenitally missing teeth
10. Small teeth (in primary dentition)
11. Enamel hypoplasia
12. Hypotonic, flaccid facies
13. Downturned oral commissures
14. Cleft lip (uncommon)
15. Microcephaly
16. Small posterior cranial fossa

Eye Findings

17. Tortuous retinal vessels
18. Suborbital congestion ("allergic
19. Strabismus
20. Narrow palpebral fissures
21. Posterior embryotoxon
22. Small optic disk
23. Prominent corneal nerves
24. Cataract
25. Iris nodules
26. Iris coloboma (uncommon)
27. Retinal coloboma (uncommon)
28. Small eyes
29. Mild orbital hypertelorism
30. Mild vertical orbital dystopia
31. Puffy or hooded upper eyelids

Ear/Hearing Findings

32. Overfolded helix
33. Attached lobules
34. Protuberant, cup-shaped ears
35. Small ears
36. Mildly asymmetric ears
37. Frequent otitis media

50. Tetralogy of Fallot
51. Right sided aorta
52. Truncus arteriosus
53. PDA (patent ductus
54. Interrupted aortic arc
55. Coarctation of the ac
56. Aortic valve anomal
57. Aberrant subclavian
58. Vascular ring
59. Anomalous origin of carotid artery
60. Transposition of the great vessels

74. Small cerebellar vermis
75. Cerebellar hypoplasia/dysgenesis
76. White matter hyperintensities
77. Generalized hypotonia
78. Cerebellar ataxia
79. Seizures
80. Strokes
81. Spina bifida/meningomyelocele
82. Mild developmental delay
83. Enlarged Sylvian fissure
84. Cavum septum pellucidum
85. Variations in size of various brain segments.

Abdominal/Kidney

99. Hypoplastic/aplastic kidney
100. Cystic kidneys
101. Inguinal hernias
102. Umbilical Hernias

148. Impulsiveness
149. Flat affect
150. Dysthymia, Cyclothymia
151. Social Immaturity
152. Obsessive compulsive disorder

Immunologic

157. Frequent upper respiratory infections
158. Frequent lower airway disease (pneumonia, bronchitis)
159. Reduced T cell populations
160. Reduced thymic hormone

Problems in Infancy

to-thrive
ER/GERD)

163. Hypothyroidism
164. Auto-immune thyroiditis
165. Mild growth deficiency, relative small stature (childhood)
166. Absent, hypoplastic thymus
167. Small pituitary gland (rare)

Problems in Infancy

115. Feeding difficulty, Failure-to-thrive
116. Gastroesophageal reflux (GER/GERD)
117. Nasal regurgitation
118. Irritability
119. Chronic constipation

Skeletal/Muscle/Orthopedic

168. Scoliosis
169. Osteopenia
170. Sprengel's anomaly, scapular deformation
171. Talipes equinovarus
172. Small skeletal muscles
173. Joint dislocations
174. Chronic leg pains

Psychiatric/Psychological

141. Bipolar affective disorder
142. Manic depressive illness and psychosis
143. Schizophrenia
144. Rapid or ultrarapid cycling of mood disorder
145. Mood disorder, depression
146. Autism spectrum disorder
147. Schizoaffective disorder

Nasal Findings

42. Prominent nasal bridge
43. Bulbous nasal tip
44. Mildly separated nasal domes (tip appears bifid)
45. Pinched alar base, narrow nostrils
46. Narrow nasal passages

ogical
ve disorder
ive illness and psychosis

apid cycling of mood disorder
, depression
m disorder
e disorder

Most common syndrome of cleft palate
Most common microdeletion syndrome in humans
Most common syndrome expressing conotruncal heart anomalies

Experts Looking at Photos

Becker et al. 2004

- 14 affected, 10 control

- one photo at infancy & one beyond 2 years old

Profession	#	Sensitivity	Specificity
Geneticist	9	0.72	0.51
Speech Pathologist	13	0.72	0.52
Surgeon	10	0.64	0.50

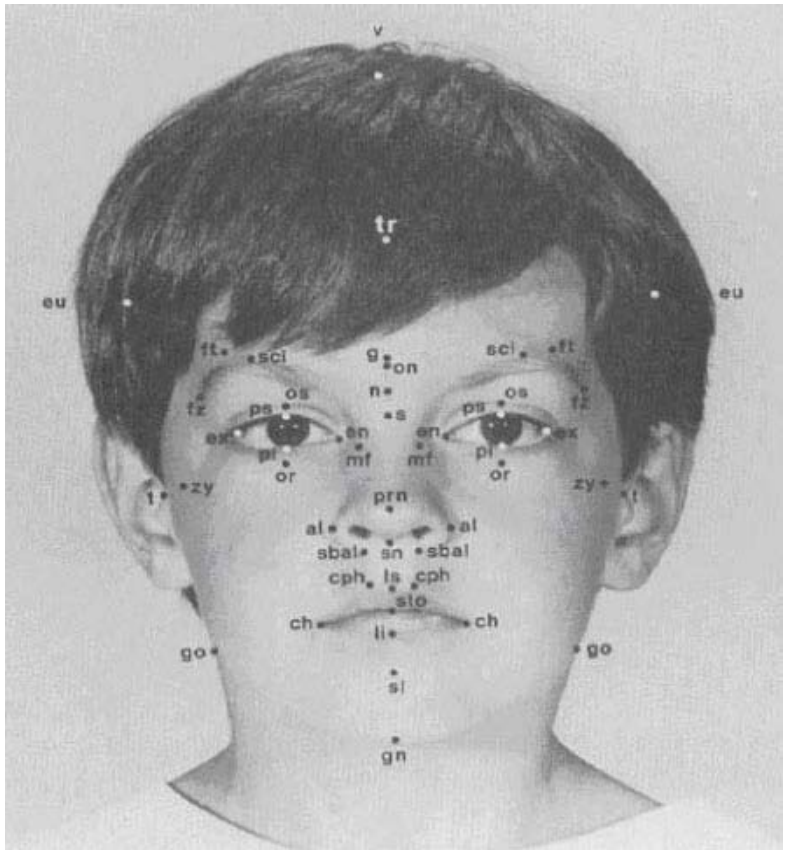
- Improve accuracy of genetic testing referrals

Research Objective

- Develop a successful methodology to
  - classify 22q11.2 deletion syndrome affected individuals
  - quantify the degree of dysmorphology in facial features
- Design consideration
  - Minimal human involvement

Related Literature

- Medical Craniofacial Assessment
  - calipers, manual landmarks
  - CT, MRI, Ultra Sound, Stereoscopic imaging
- Time consuming human involvement



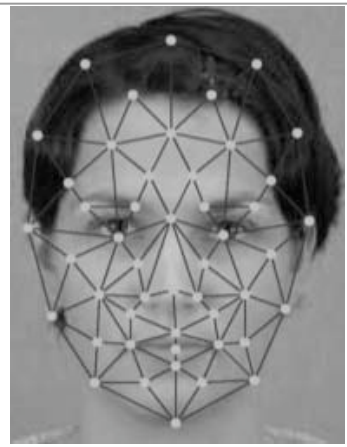
Related Literature

- Computer Vision Craniofacial Analysis
  - 1D waveforms
  - 2D images, landmarks
  - 3D morphable models, new representations, landmarks
  - hybrid 2D/3D systems
- Focus: biometric authentication and recognition

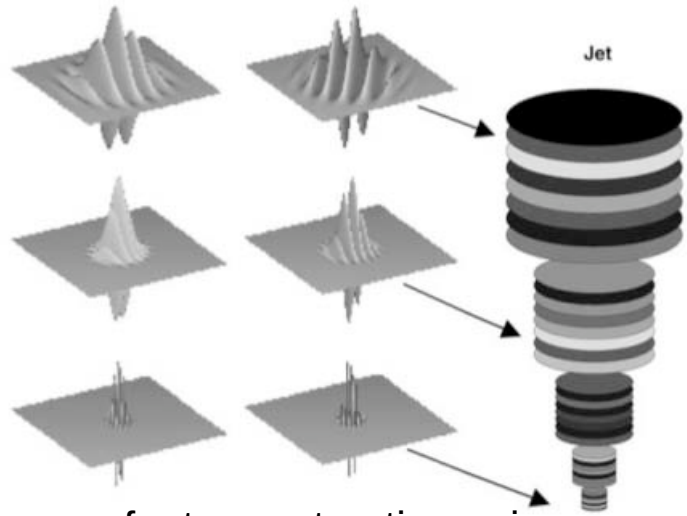
Recognition of Dysmorphic Faces

Boehringer et al. 2006

input



point labeling



feature extraction using wavelets

classify

PCA

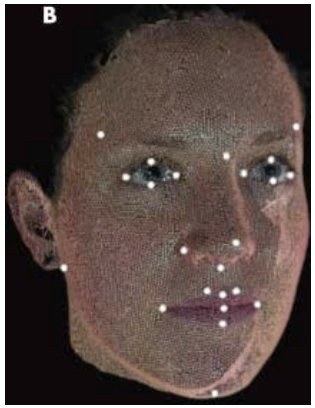
Recognition of Dysmorphic Faces Results

- Classifiers: LDA, SVM, kNN, JV
- Simultaneous classification
  - Manual 76%
  - Auto 52% => face localization
- Pairwise classification
  - 89% - 100%

Condition	*Age range*	*n*
Microdeletion 22q11.2	1–12	23
Cri-du-chat syndrome	1–17	16
Cornelia de Lange syndrome	5–33	12
Fragile X Syndrome	4–14	12
Mukopolysaccharidosis III	5–25	10
Noonan syndrome	1–40	18
Prader–Willi syndrome	4–21	12
Smith–Lemli–Opitz syndrome	1–18	13
Sotos syndrome	1–20	18
Williams–Beuren syndrome	1–29	13

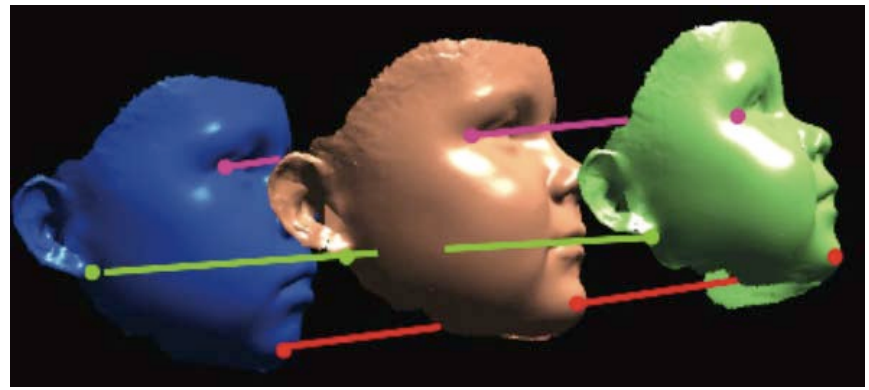
Dense Surface Models

Hutton 2004
Hammond et al. 2005
Hammond 2007



input

calculate mean landmarks



warp to mean landmarks

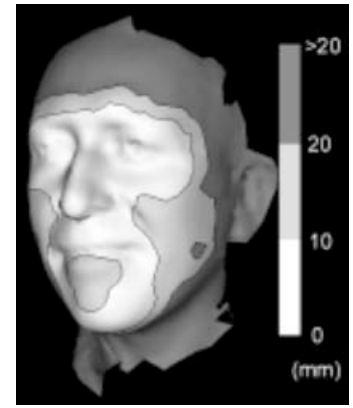
create dense correspondence to base mesh

average face

unwarp

PCA

classify



trim surfaces

Dense Surface Models: 22q11.2DS Results

Data characteristics	Classifiers	Best 22q11.2 results
60 VCFS 130 control	SVM, closest mean, Logistic regression, Neural networks, decision trees	Sensitivity 0.83 Specificity 0.92
115 VCFS 185 control	SVM, closest mean, linear discriminant analysis	Accuracy Face 0.94 Eyes 0.83 Nose 0.87 Mouth 0.85

Comparison to Previous Work

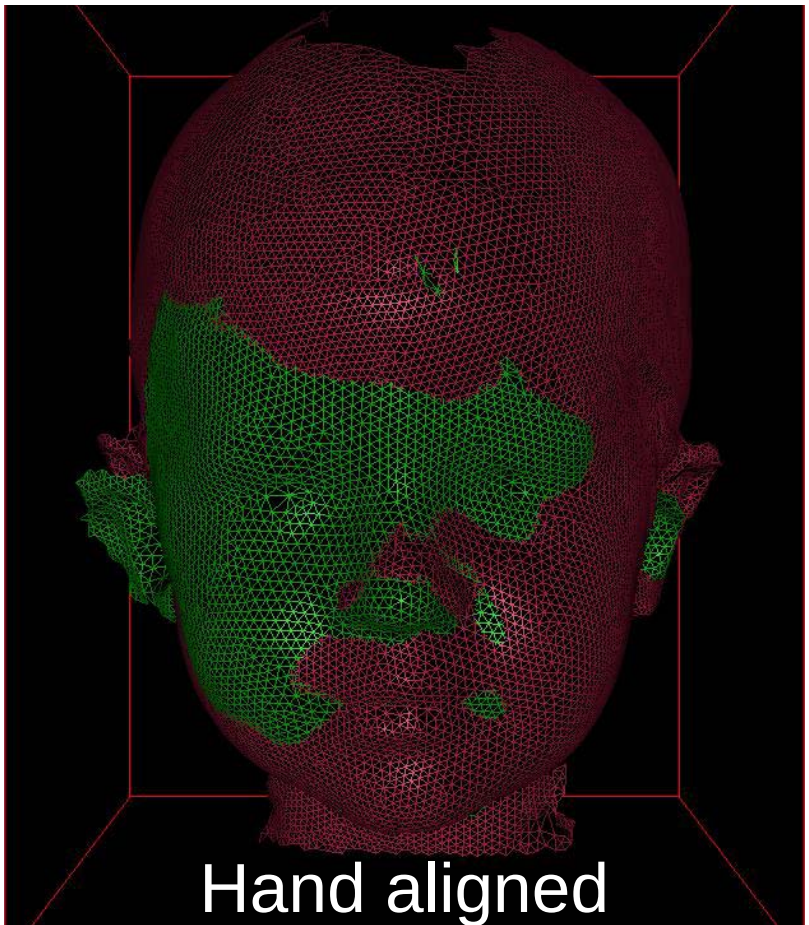
	Boehringer et al	Hutton et al	My work
Data representation	2D photographs	3D meshes	2D photographs 2.5D depth images curved lines
Control data	no	yes	yes
Data labeling	manual	manual	none [automatic]
Clean up	manual	empirically determined threshold	manual
Final goal	separate diseases	distance from average	facial features

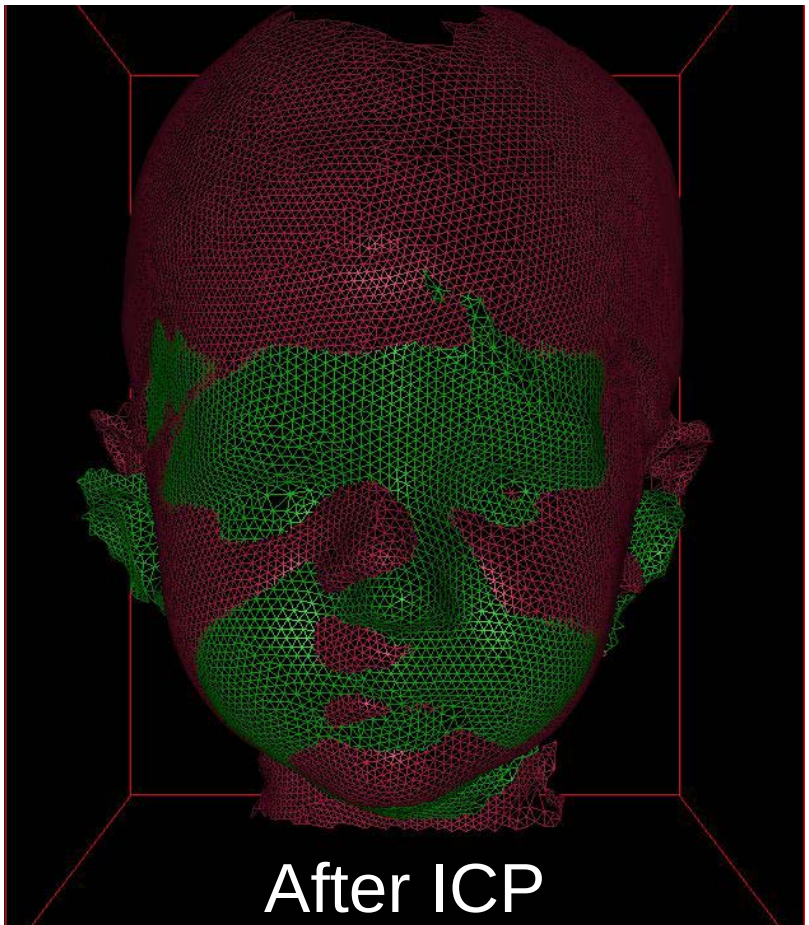
Data Preprocessing

Data Preprocessing: Pose Alignment 1st Attempt

- Goal: Align each head to same orientation

- Solution: Hand align with Iterative Closest Point (ICP) assistance

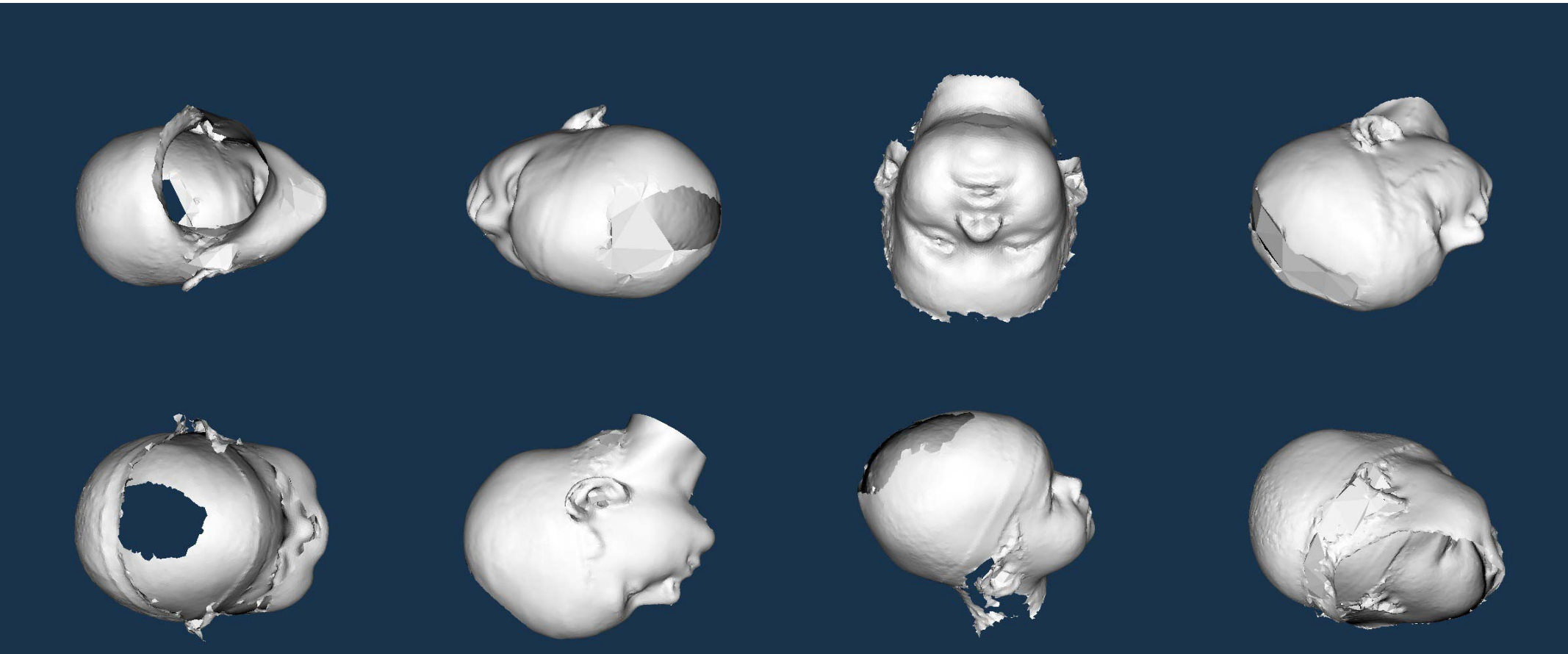




Data Preprocessing: Pose Alignment 2nd Attempt

- Goal: Align each head to same orientation

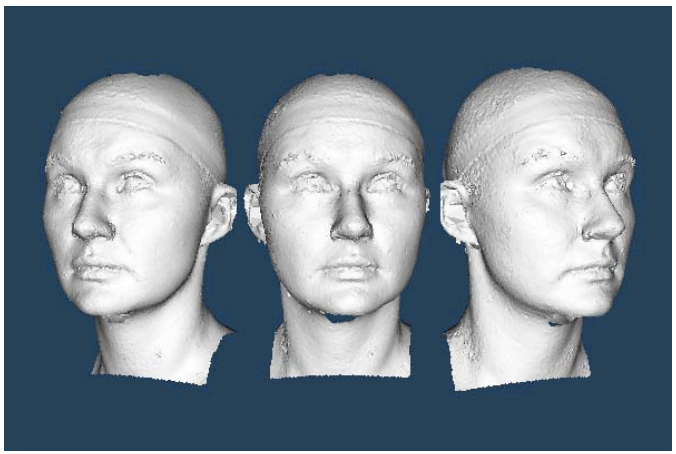
- Solution: Align using 1st principle component from PCA



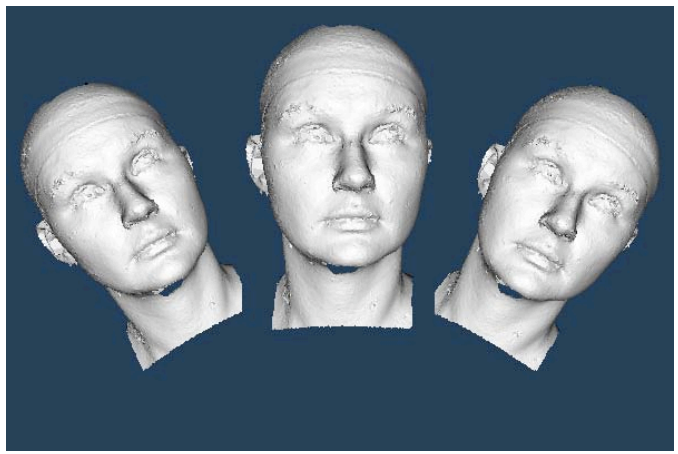
Data Preprocessing: Pose Alignment Final Solution

- Goal: Align each head to same orientation

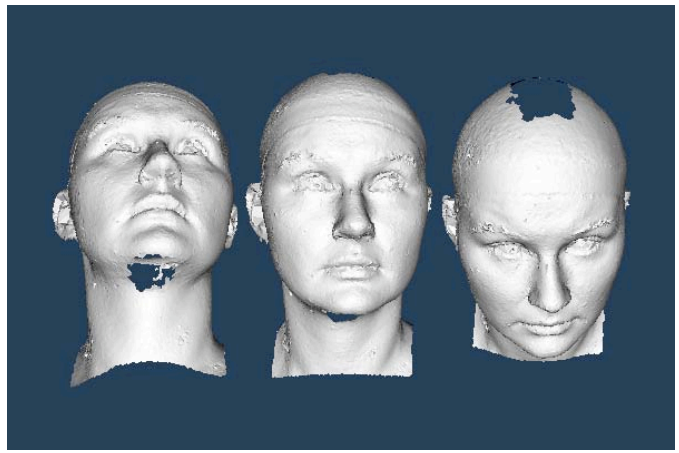
- Solution: Automatically calculate 3 rotation angles necessary to achieve goal



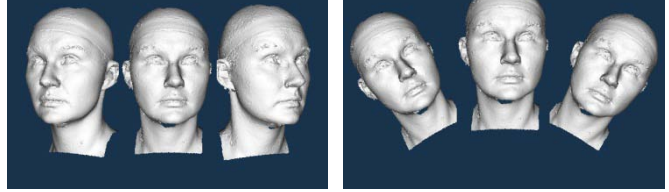
Yaw



Roll

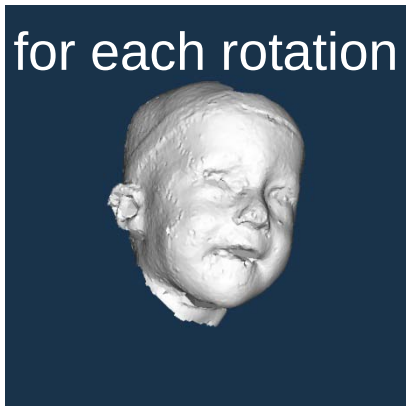


Pitch



Data Preprocessing: Yaw and Roll Alignment

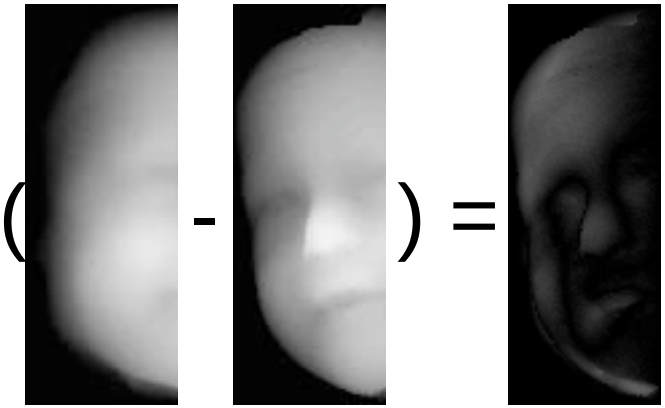
- Use symmetry to align



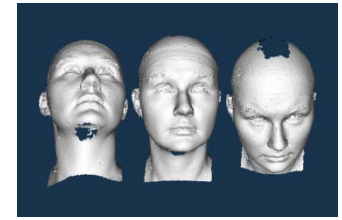
rasterize

choose rotation angle that minimizes difference

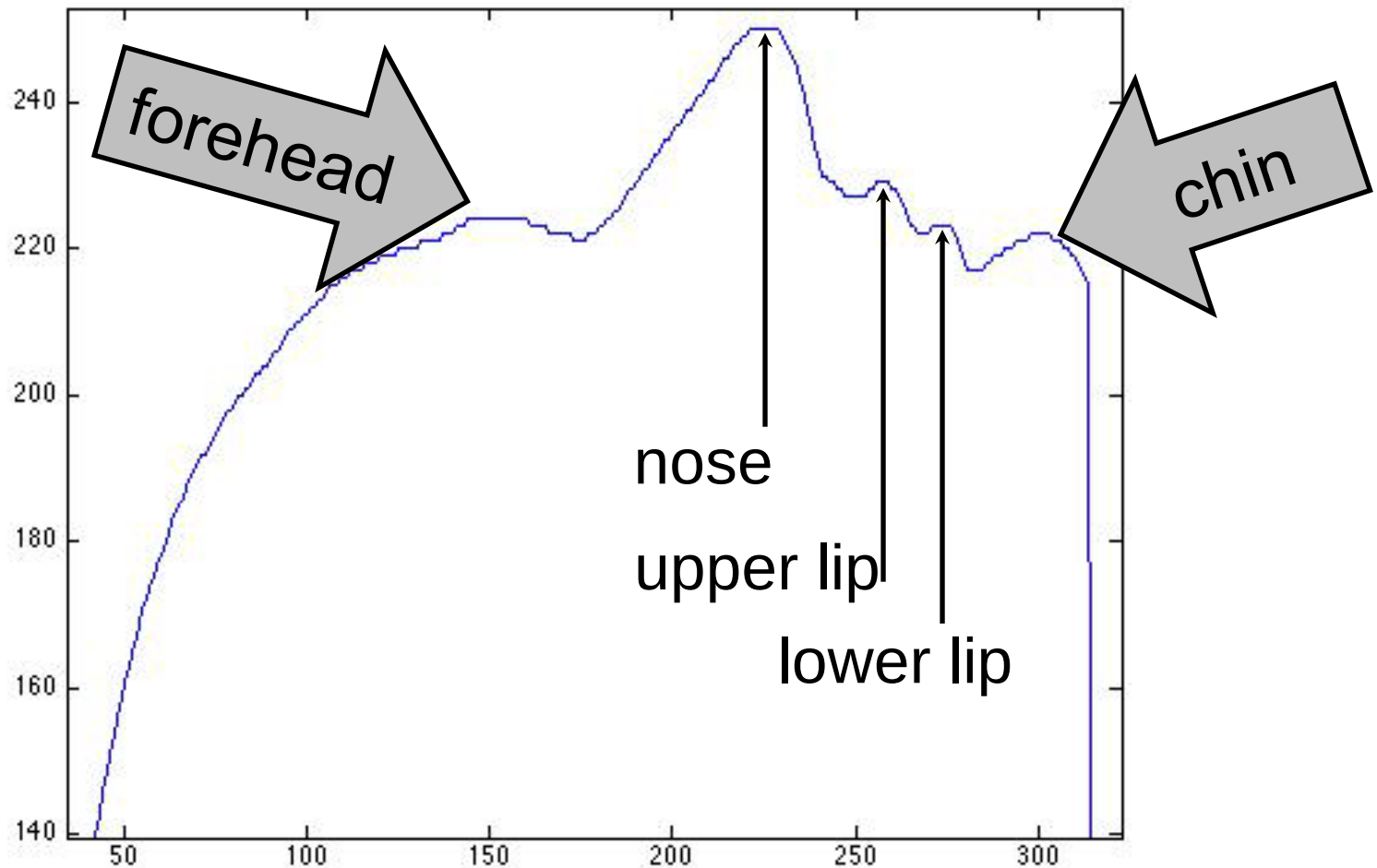
absolute value (-) =



Data Preprocessing: Pitch Alignment

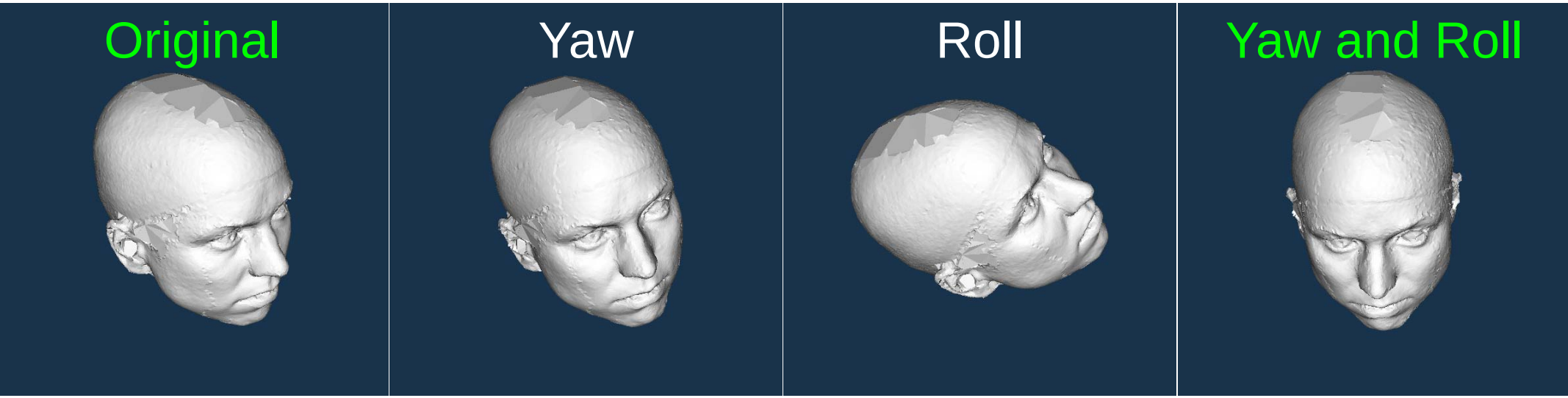


- Minimize difference between chin height and forehead height

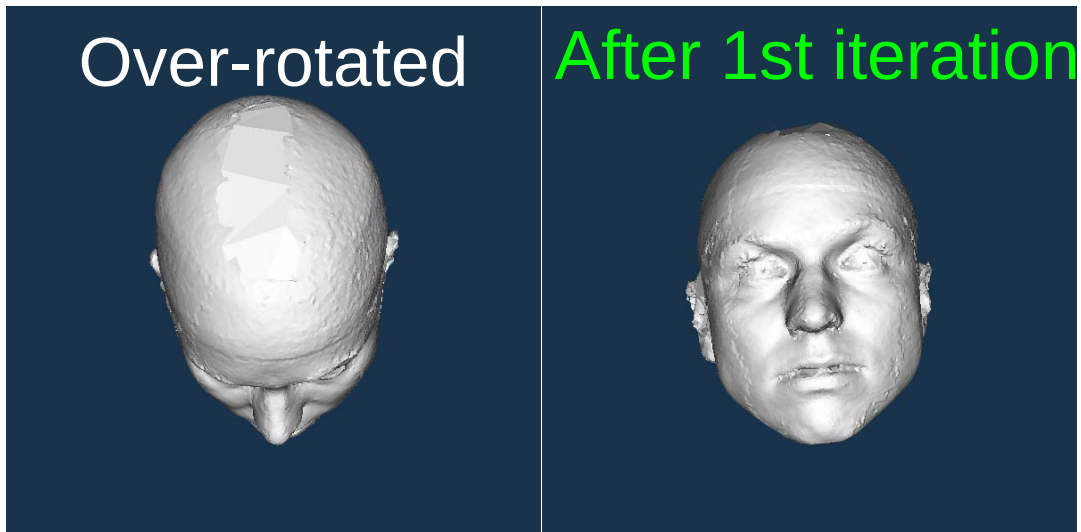


Data Preprocessing: Alignment Results

- Better results if both Yaw and Roll are aligned together



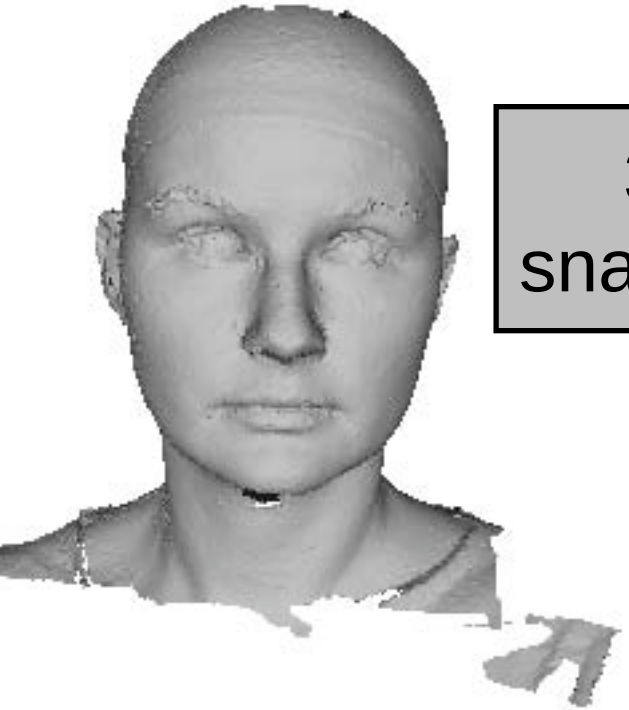
- Pitch rotation can fall into local minimum due to top of the head



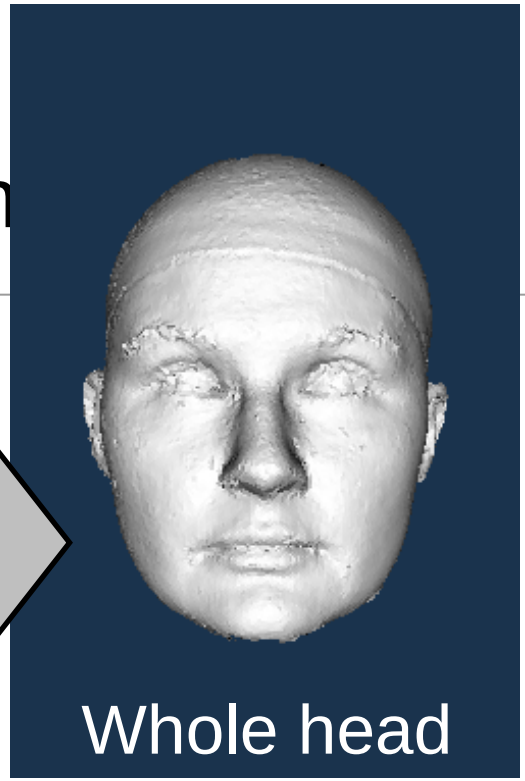
Data Representation

- 3D snapshot
- 2.5D depth image
- Curved lines

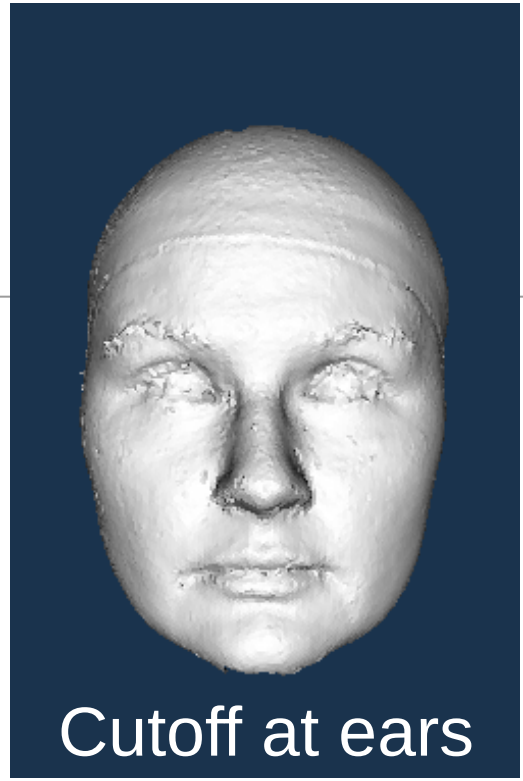
Data Representation



3D snapshot

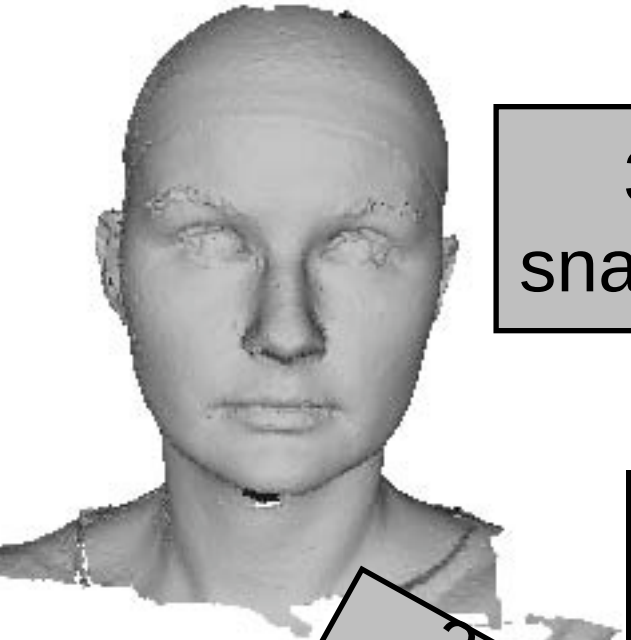


Whole head

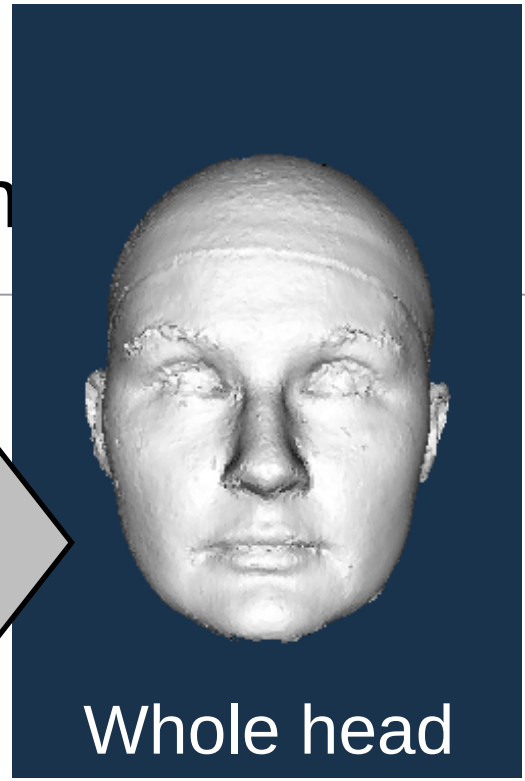


Cutoff at ears

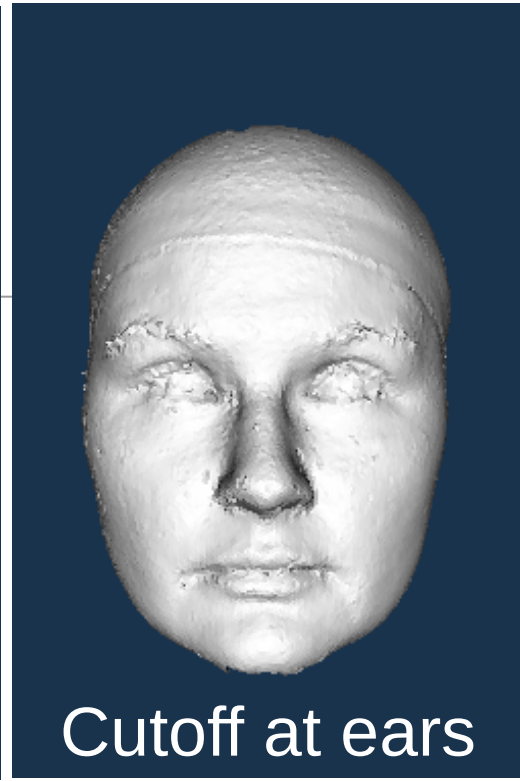
Data Representation



3D snapshot

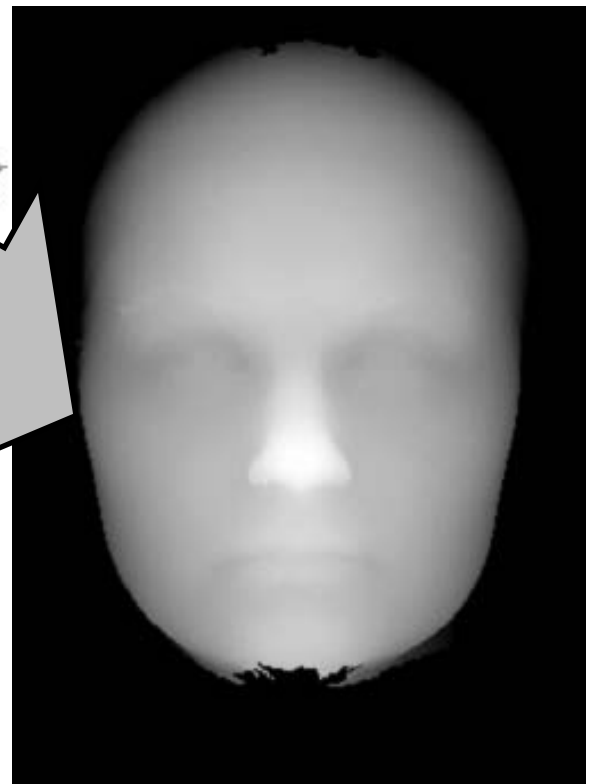


Whole head

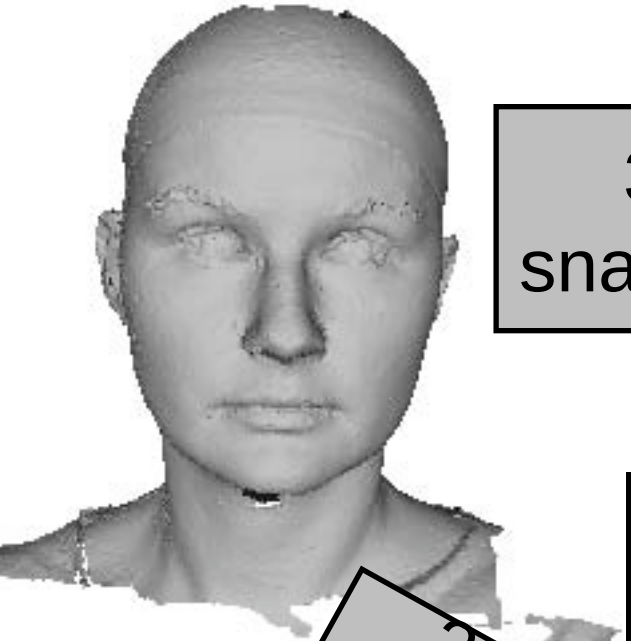


Cutoff at ears

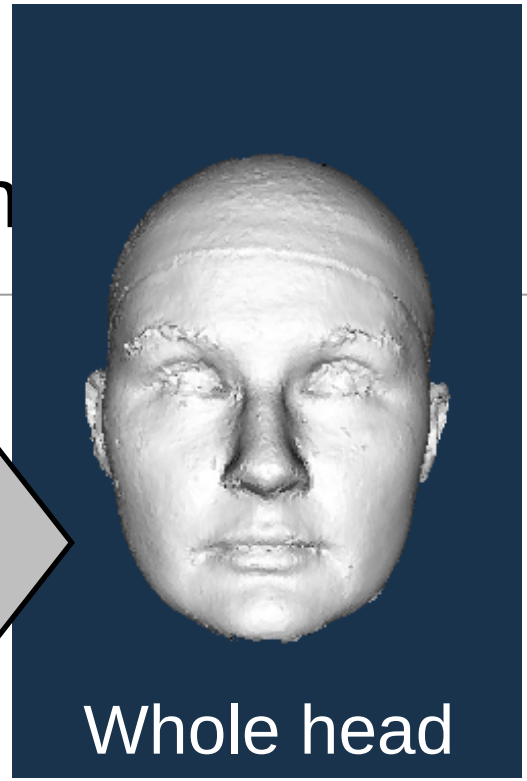
2.5D depth map



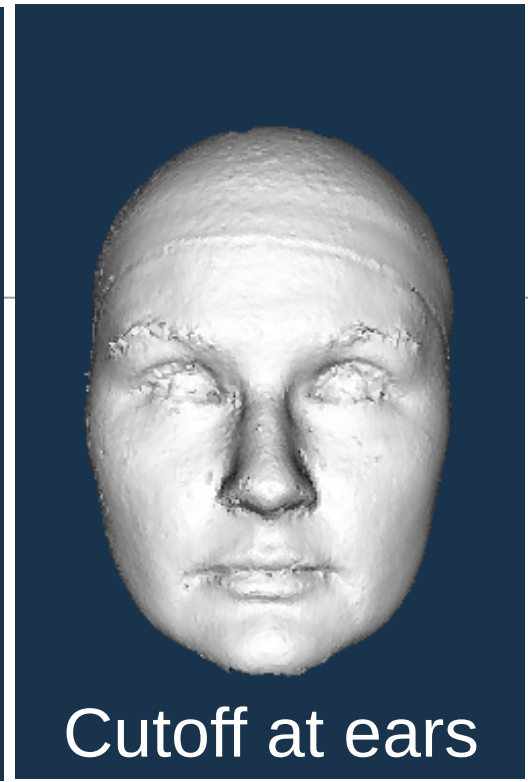
Data Representation



3D snapshot

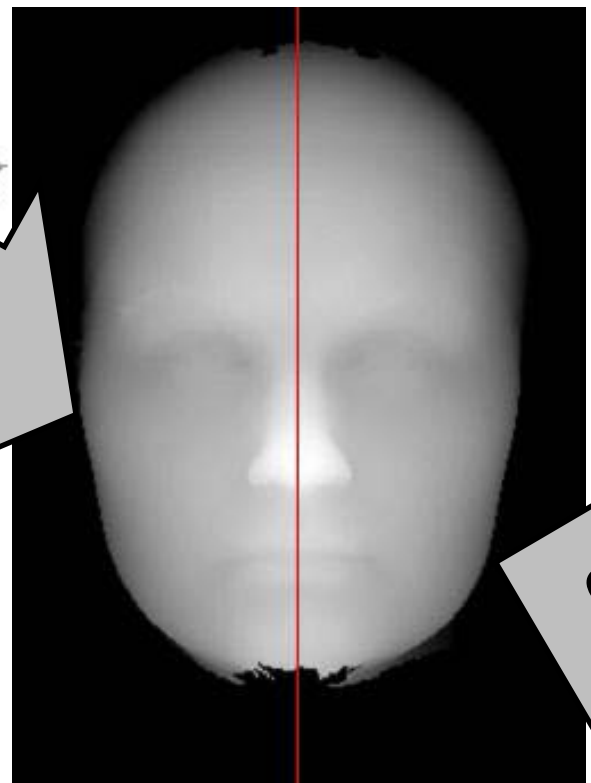


Whole head

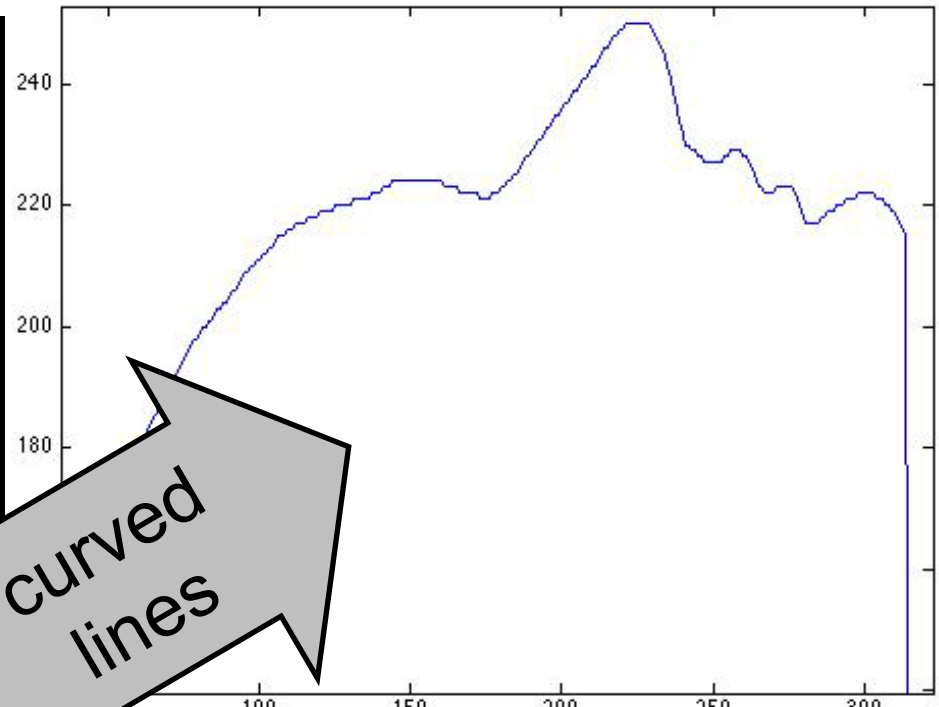


Cutoff at ears

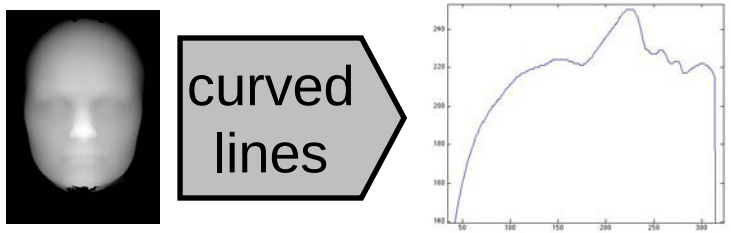
2.5D depth map

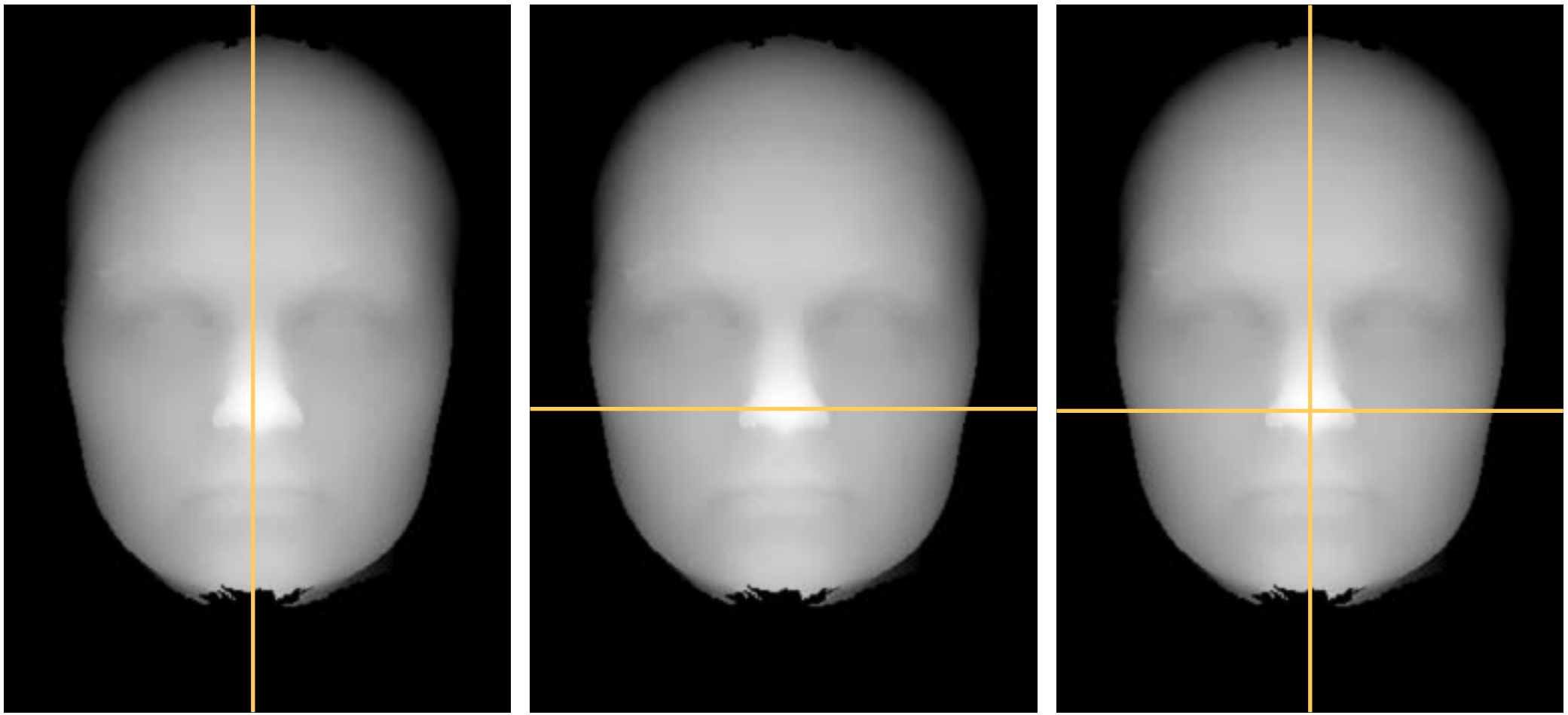


curved lines



Curved Lines Detail





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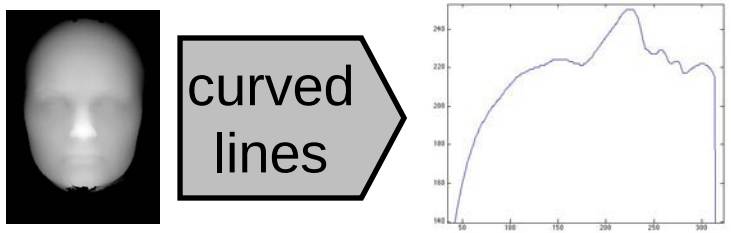
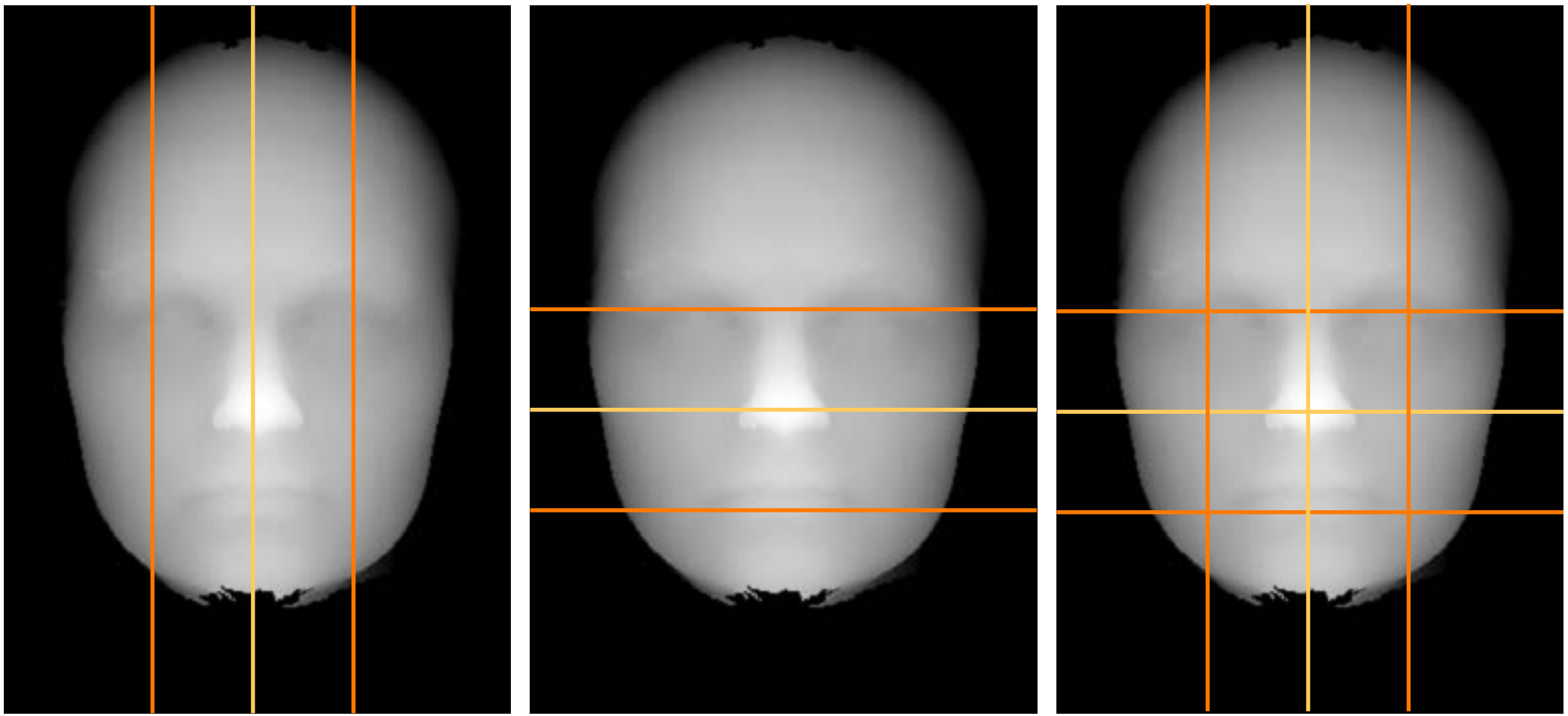
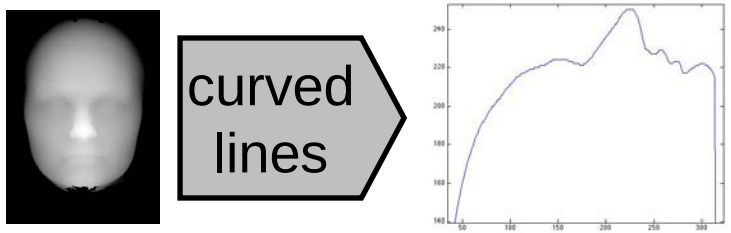
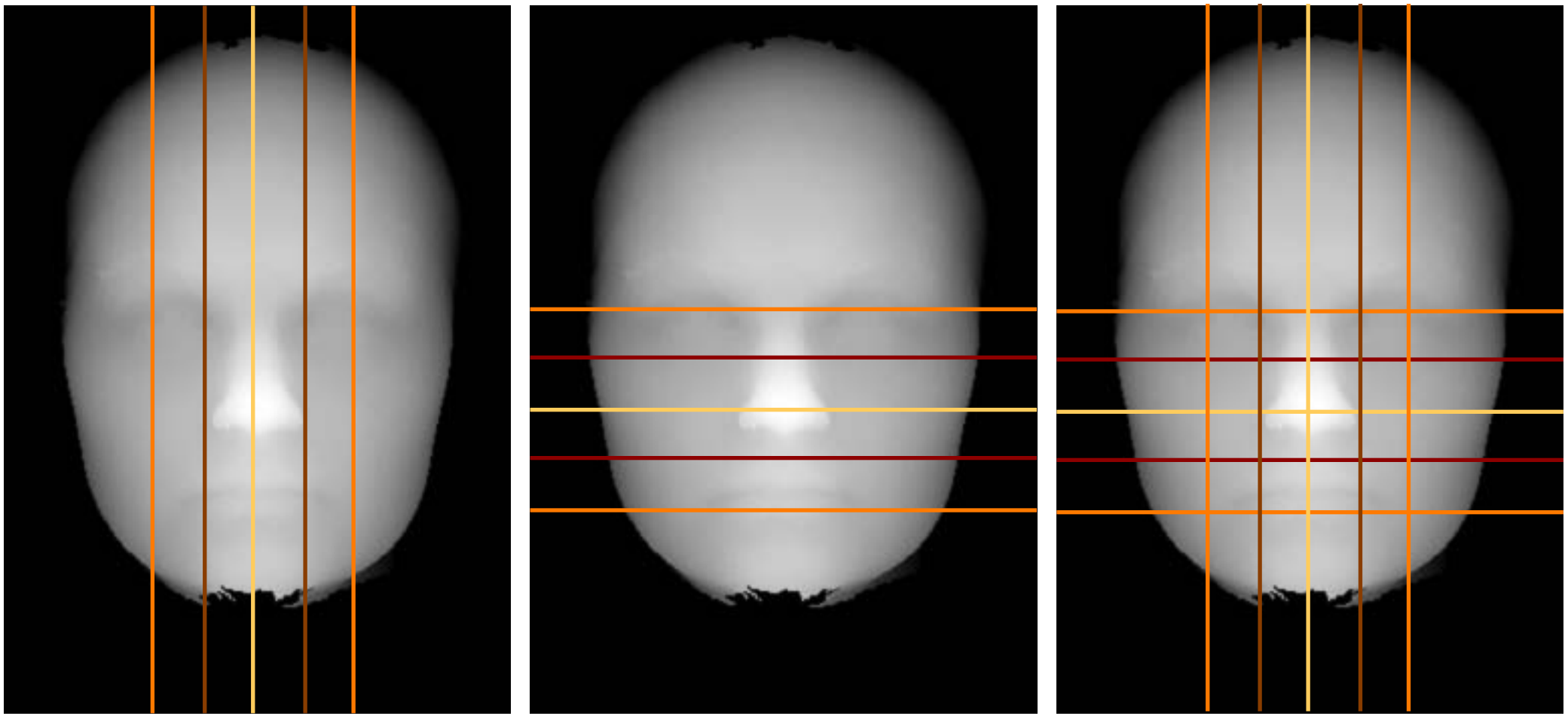


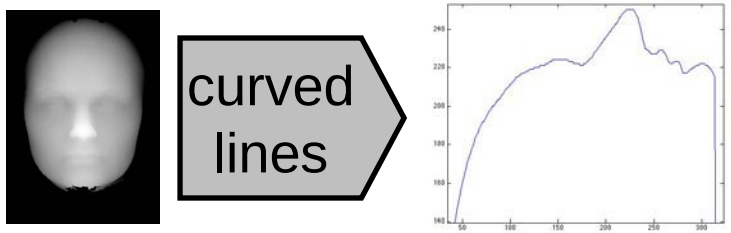


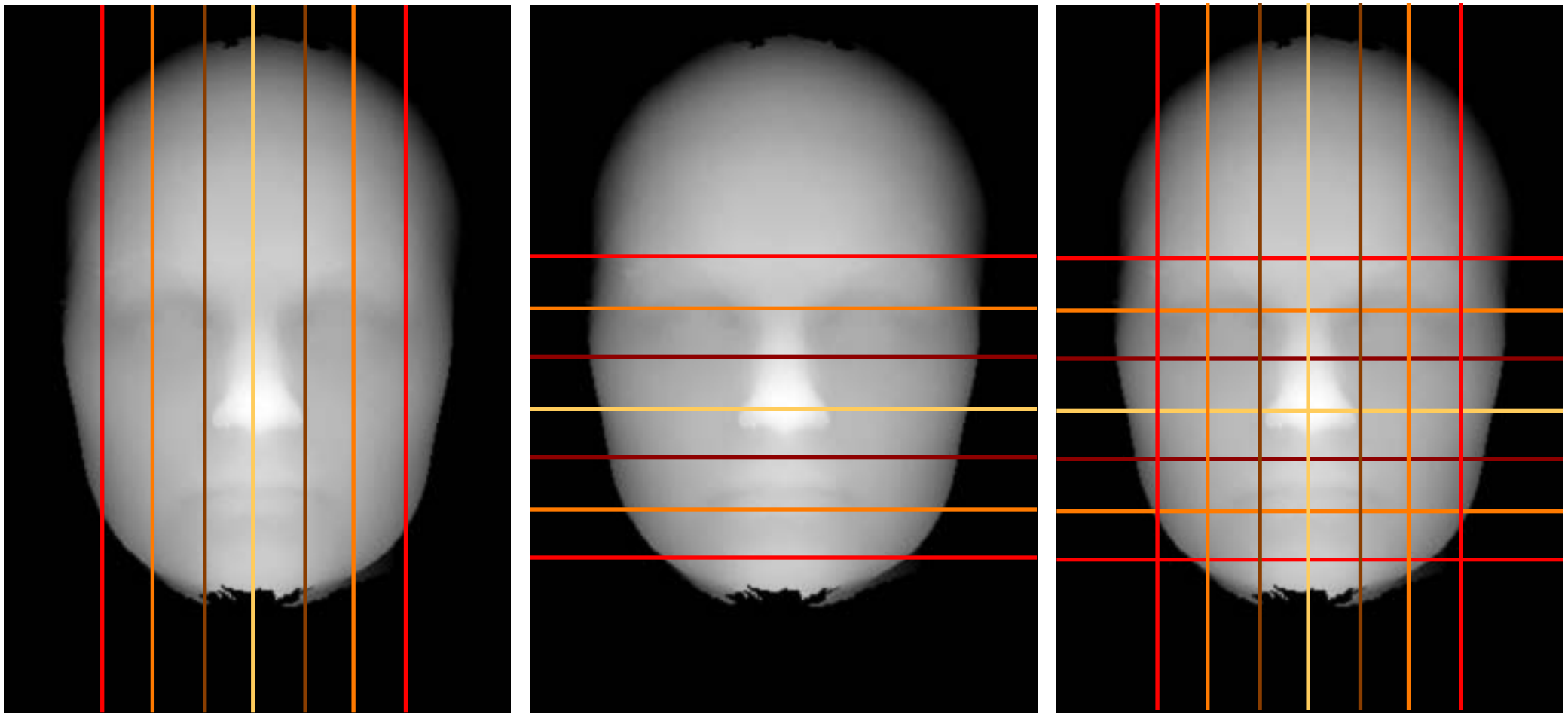
Curved Lines Detail





Curved Lines Detail

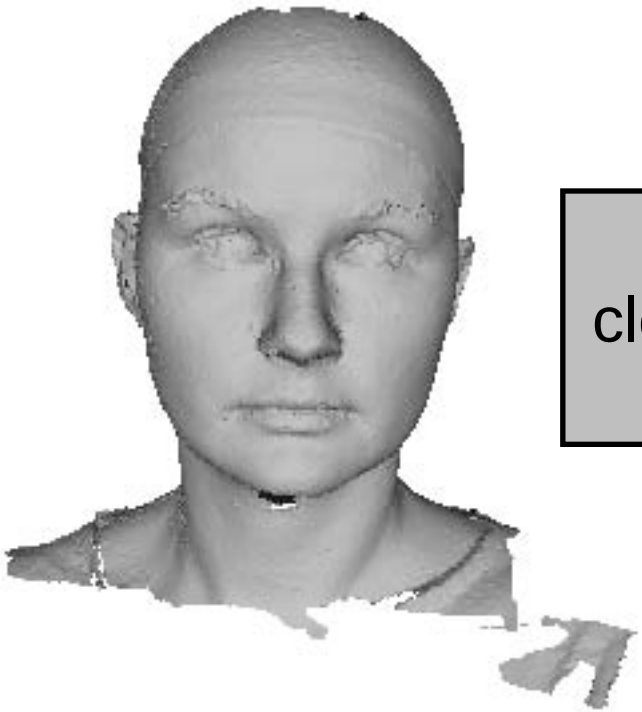




Experiment Setup

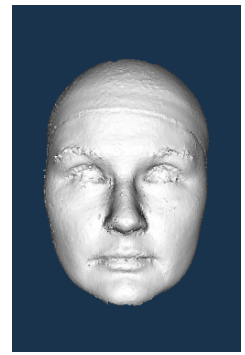
- 53 affected, 136 control individuals
- Age range 10 months to 39 years
- Data labeled status, gender & age
- Goal: classify each individual as affected or control

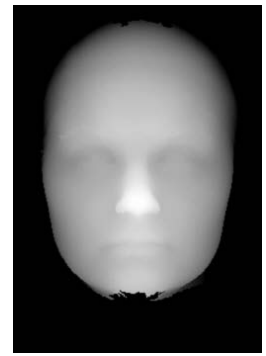
System Diagram

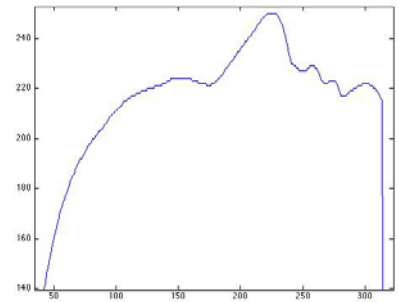


clean up

automatic alignment







facial feature selection and quantification

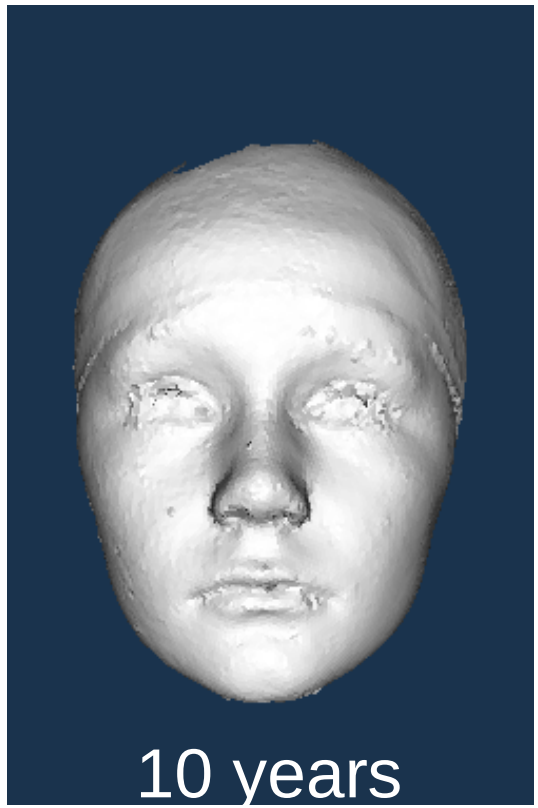
classify

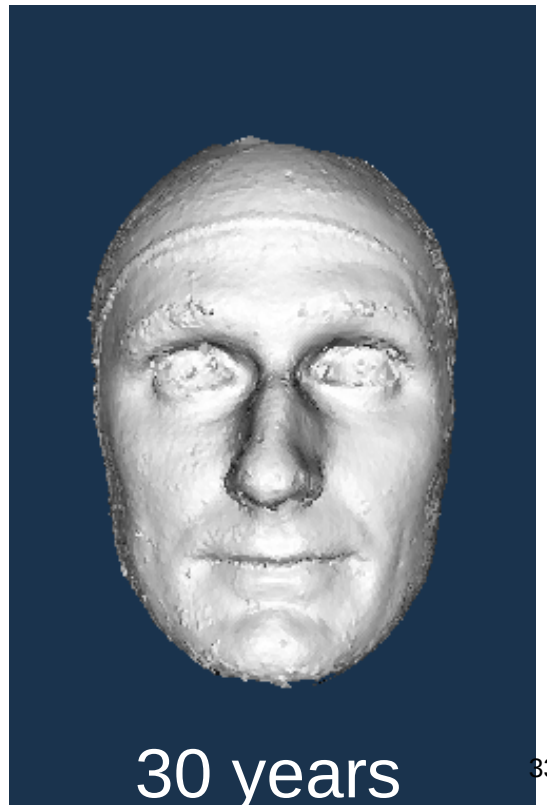
Naive Bayes

PCA

Experiments: Component Selection

Data divided by	Number of selected attributes	top 5 principal components	next 5 principal components
sex	64	d1, d7, d8, d9, d10	d11,d12,d14,d15,d16
age	47	d2, d3, d5, d6, d9	d13, d18, d20, d22, d23
affected	11	d1, d5, d8, d15, d25	d63, d66, d73, d75, d81 (d85)





Statistical measures

$$Accuracy = \frac{TP + TN}{TP + FP + TN + FN}$$

$$Recall = Sn = \frac{TP}{TP + FN}$$

$$Precision = \frac{TP}{TP + FP}$$

$$Sp = \frac{TN}{TN + FP}$$

$$F1 = \frac{2 * Precision * Recall}{Precision + Recall}$$

		Disease	
		Positive	Negative
Test	Positive	TP	FP
	Negative	FN	TN

Classifiers used

Table 1: F-measure with statistical significance t-test 0.05, two tailed

Data Set	ALL 3Dsnp	W86 3Dsnp	ALL 2.5D	W86 2.5D
ZeroR	0.00±0.00	0.45±0.30 ∘	0.00±0.00	0.45±0.30∘
NNge	0.32±0.23	0.66±0.19 ∘	0.29±0.25	0.70±0.17∘
JRip	0.38±0.21	0.59±0.21 ∘	0.42±0.19	0.66±0.18∘
J48	0.47±0.20	0.67±0.19 ∘	0.44±0.18	0.66±0.19
IB1	0.28±0.23	0.45±0.25	0.17±0.18	0.43±0.23
IBk=3	0.40±0.21	0.62±0.22 ∘	0.13±0.18●	0.37±0.24
NN:9,3	0.37±0.20	0.59±0.22 ∘	0.32±0.21	0.51±0.20
SVM default	0.35±0.20	0.59±0.23 ∘	0.30±0.21	0.49±0.21
SVM c=2	0.35±0.20	0.59±0.23 ∘	0.30±0.21	0.49±0.21
SVM c=3	0.35±0.20	0.59±0.23 ∘	0.30±0.21	0.49±0.21
SVM c=4	0.35±0.20	0.59±0.23 ∘	0.30±0.21	0.49±0.21
SVM e=2	0.35±0.20	0.60±0.22 ∘	0.29±0.21	0.57±0.18∘
SVM e=3	0.35±0.21	0.59±0.22 ∘	0.29±0.21	0.60±0.20∘
SVM e=4	0.34±0.21	0.59±0.22 ∘	0.27±0.21	0.61±0.20∘
SVM RBF	0.00±0.00	0.49±0.30 ∘	0.00±0.00	0.34±0.30∘
NaiveBayes	0.53±0.19	0.68±0.20	0.61±0.15	0.72±0.20∘

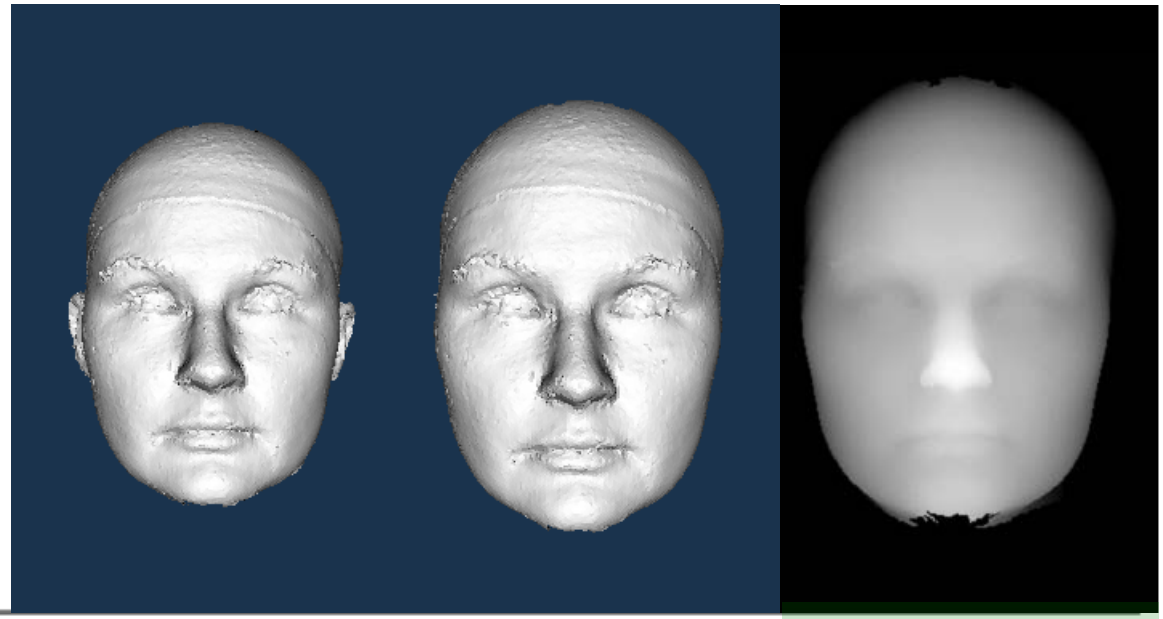
∘, ● statistically significant improvement or degradation

Results: Balancing Data Sets

Name	#total (#affected)	data set description
ALL	189 (53)	data collected from Children's Hospital
AS106	106 (53)	each affected matched by gender, then closest age
W86	86 (43)	only affected labeled white matched by gender, then closest age

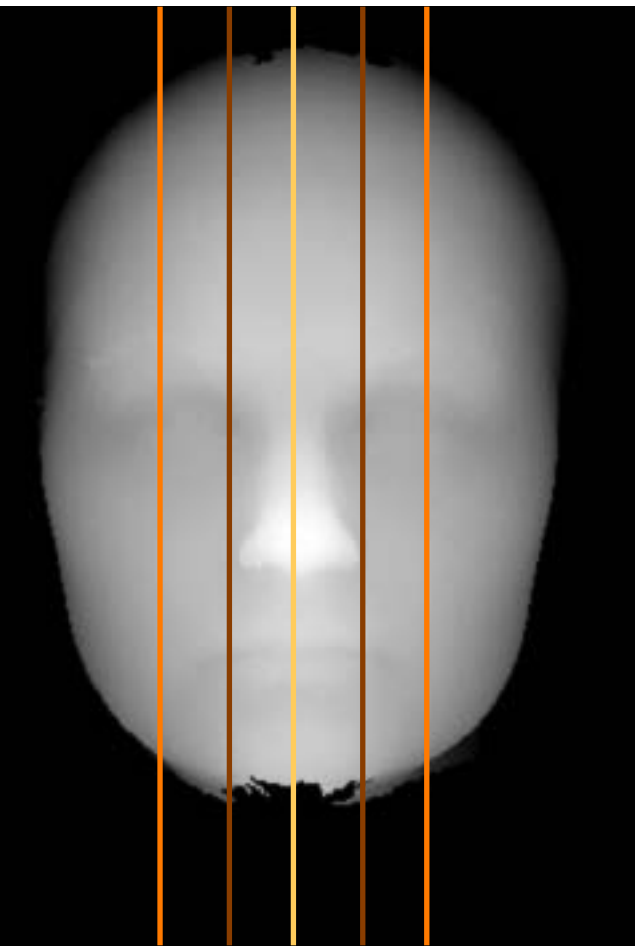
Data Set	ALL	AS106	W86	2 Experts	All Experts
F-measure	0.53±0.19	0.66±0.19	0.68±0.20	0.68±0.09	0.75±0.14
Precision	0.56±0.22	0.78±0.21	0.82±0.20	0.59±0.18	0.67±0.18
Recall	0.52±0.21	0.61±0.22	0.62±0.24	0.83±0.11	0.88±0.11
Accuracy	0.75±0.10	0.71±0.15	0.74±0.13	0.73±0.02	0.78±0.10

Results: 3D snapshot vs. 2.5D



Data Set	3Dsnp	3Dsnp cut	2.5D
F-measure	0.71±0.18	0.68±0.20	0.72±0.20
Precision	0.88±0.18	0.82±0.20	0.80±0.20
Recall	0.63±0.22	0.62±0.24	0.69±0.22
Accuracy	0.76±0.14	0.74±0.13	0.75±0.16

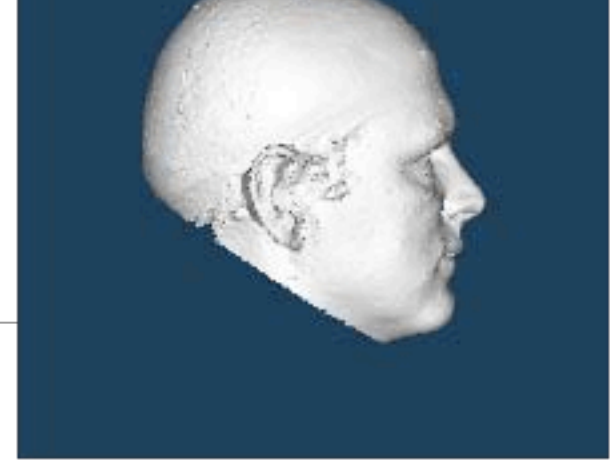
Results: Curved Lines



Data Set		Vertical Lines			
	2.5D	1	3	5	7
F-measure	0.72	0.71	0.76	0.78	0.67
Precision	0.80	0.81	0.88	0.88	0.79
Recall	0.69	0.68	0.70	0.73	0.62
Accuracy	0.75	0.75	0.79	0.82	0.72

Expert Survey

- 3 experts
- quantify features
- new insights



1

	Definitely YES	Probably YES	Probably NO	Definitely NO
Does this individual have 22q11?	○	○	○	○
Do you know this individual?	○	○	○	○

	Opposite of 22q11	Not 22q11	Moderate 22q11	Severe 22q11	Not enough data
Overall face					
22q Facial Phenotype	○	○	○	○	○
Asymmetric	○	○	○	○	○
Square/Rectangular	○	○	○	○	○
Hypotonic appearance	○	○	○	○	○
Eyes					
Hooded appearance	○	○	○	○	○
Nose					
Prominent nasal root	○	○	○	○	○
Tubular appearance	○	○	○	○	○
Bulbous nasal tip	○	○	○	○	○
Small nasal alae	○	○	○	○	○
Ears					
Small	○	○	○	○	○
Protuberant	○	○	○	○	○
Midface					
Relatively flat	○	○	○	○	○
Forehead					
Square	○	○	○	○	○
Prominent on profile	○	○	○	○	○
Mouth					
Small	○	○	○	○	○
Open	○	○	○	○	○

Comparison to experts

Data Set	ALL	AS106	W86	All Experts	2 Experts
F-measure	0.53±0.19	0.66±0.19	0.68±0.20	0.75±0.14	0.68±0.09
Precision	0.56±0.22	0.78±0.21	0.82±0.20	0.67±0.18	0.59±0.18
Recall	0.52±0.21	0.61±0.22	0.62±0.24	0.88±0.11	0.83±0.11
Accuracy	0.75±0.10	0.71±0.15	0.74±0.13	0.78±0.10	0.73±0.02

	W86	Experts	ExpA	ExpC	ExpM
F-measure	0.68±0.20	0.75±0.14	0.62	0.89	0.74
Precision	0.82±0.20	0.67±0.18	0.47	0.81	0.72
Recall	0.62±0.24	0.88±0.11	0.91	0.97	0.76
Accuracy	0.74±0.13	0.78±0.10	0.71	0.90	0.74

Proposal for Continued Work

- Distance from Average
- Approximation using Ellipsoids
- Creating new texture information
- Assessing facial asymmetry

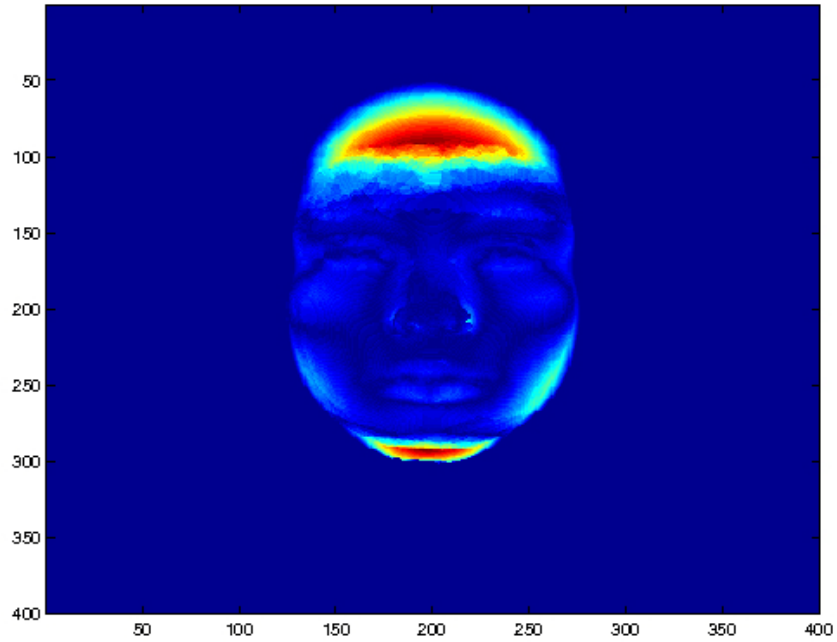
Global features

- Automatic Landmarks
- 3D Local features

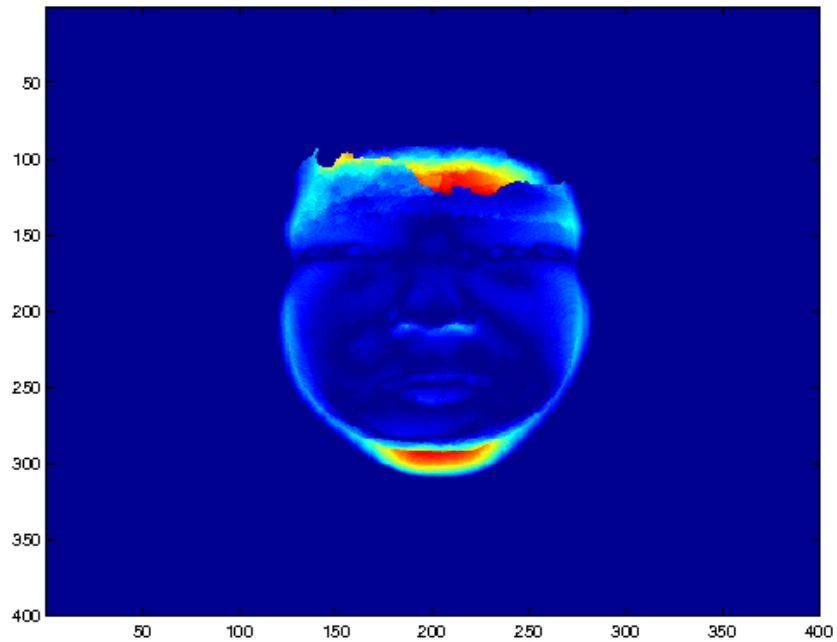
Local features

Global Feature
Distance From Average

$$A_{subset} = \frac{\sum_{subset} F}{|subset|}$$

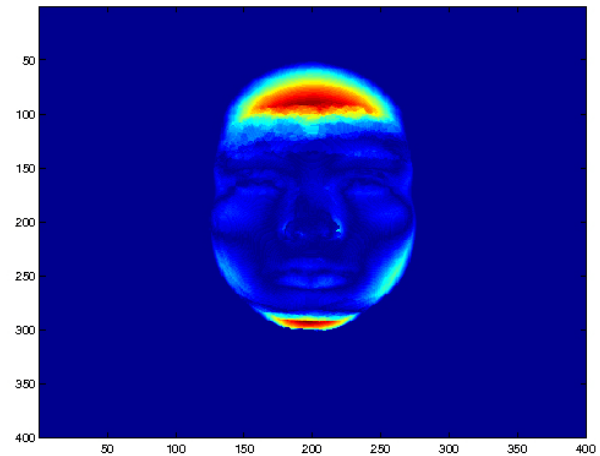


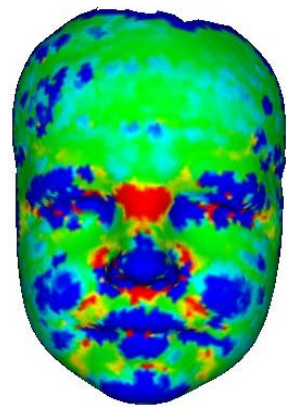
- Data sets: curved lines, 2.5D
- Separate by status, sex, age
- Similar approach as Hutton, but no landmarks necessary



Global Feature
Creating New Texture Information

- Average face
- Gaussian curvature
- Azimuth and elevation of normals
- Geodesic information





Contributions

- Fully automatic system
- Facial pose alignment method
- Different data representations for classification
- Classification of 22q11.2DS affected individuals rivals experts