UNIVERSITÀ DEGLI STUDI DI MILANO
DIPARTIMENTO DI
SCIENZE BIOMEDICHE PER LA SALUTE

Three-Dimensional Craniofacial Features of Glut1DS

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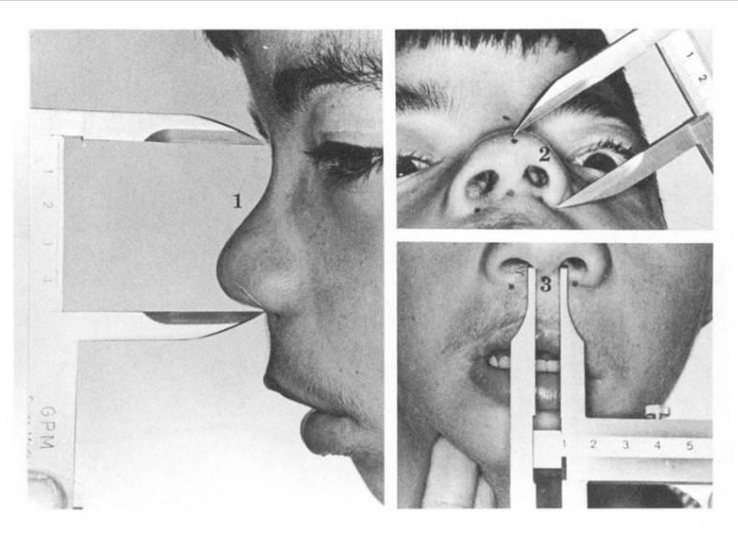
Measurements
RMS
Time [s]
CD133
CD49f
E12.5
E13.5
E14.5
E15.5
B
C
D

3D surface analysis

- Direct anthropometry
- 3D Cephalometry (CT/ CBCT)
- Ultrasounds
- Magnetic resonance
- Electromagnetic systems
- Electromechanical systems
- Optoelectronic systems
- Moiré/ fringe light projectors
- Laser scanning
- Stereophotogrammetry

- Simple, easy to use
- Danger free
- Fast
- Easy-to-manage data
- Treatment visualization and simulation
- Low cost

Conventional anthropometry --single measurements



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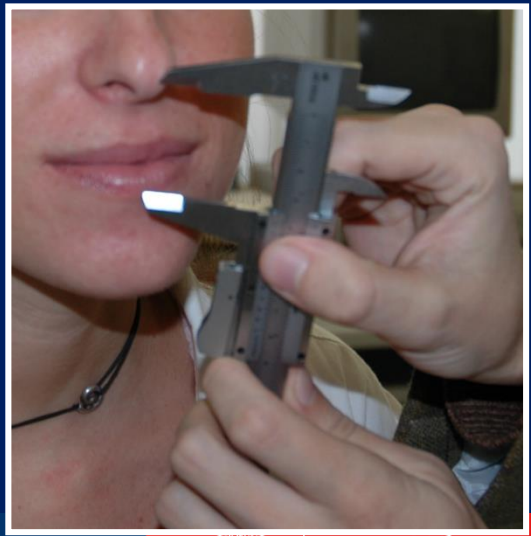

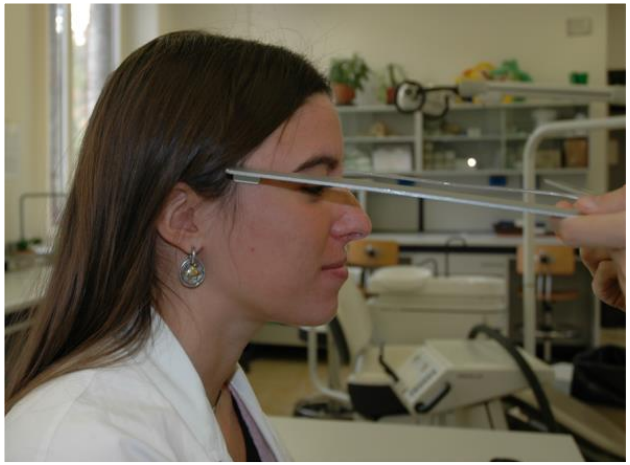

L.G. Farkas, Anthropometry of the head and face, 2nd Ed. New York: Raven Press, 1994.

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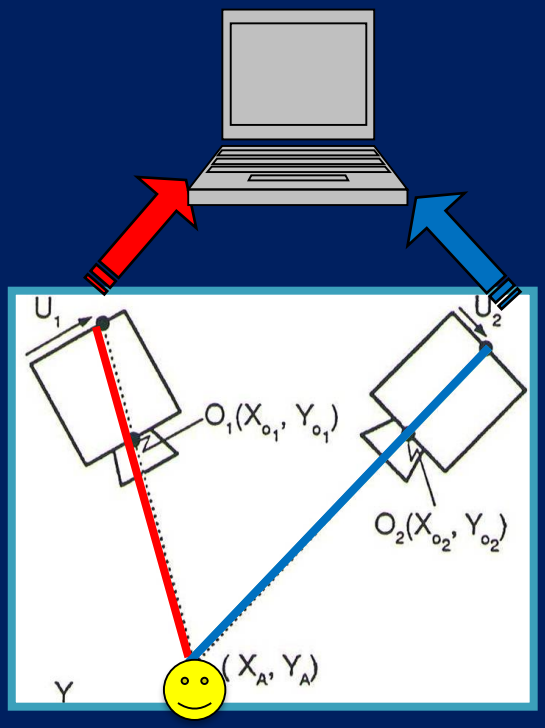
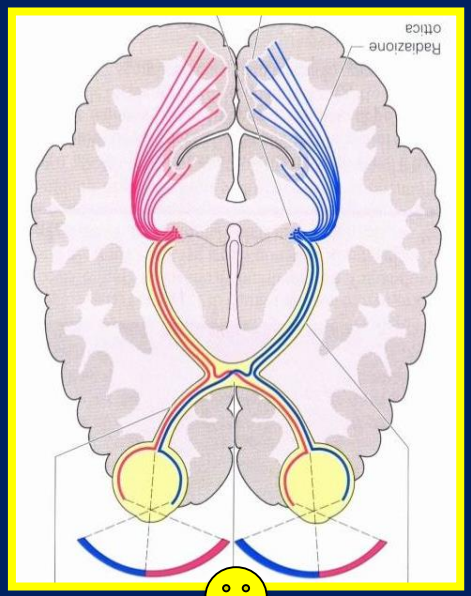
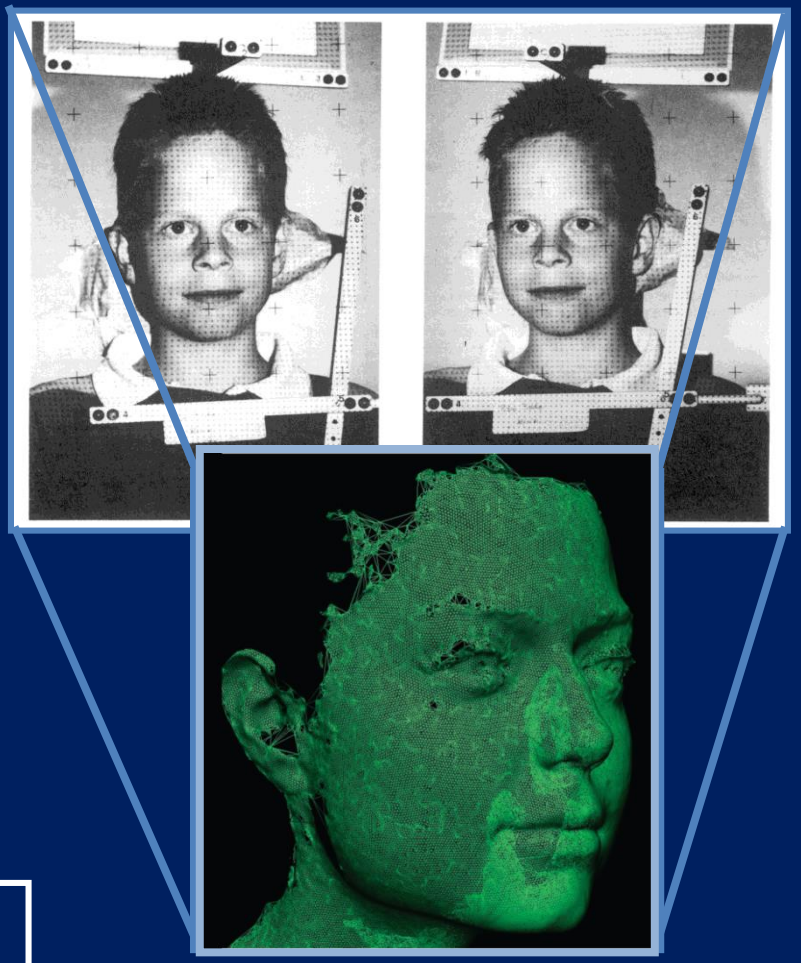
Optical methods

Stereophotogrammetry



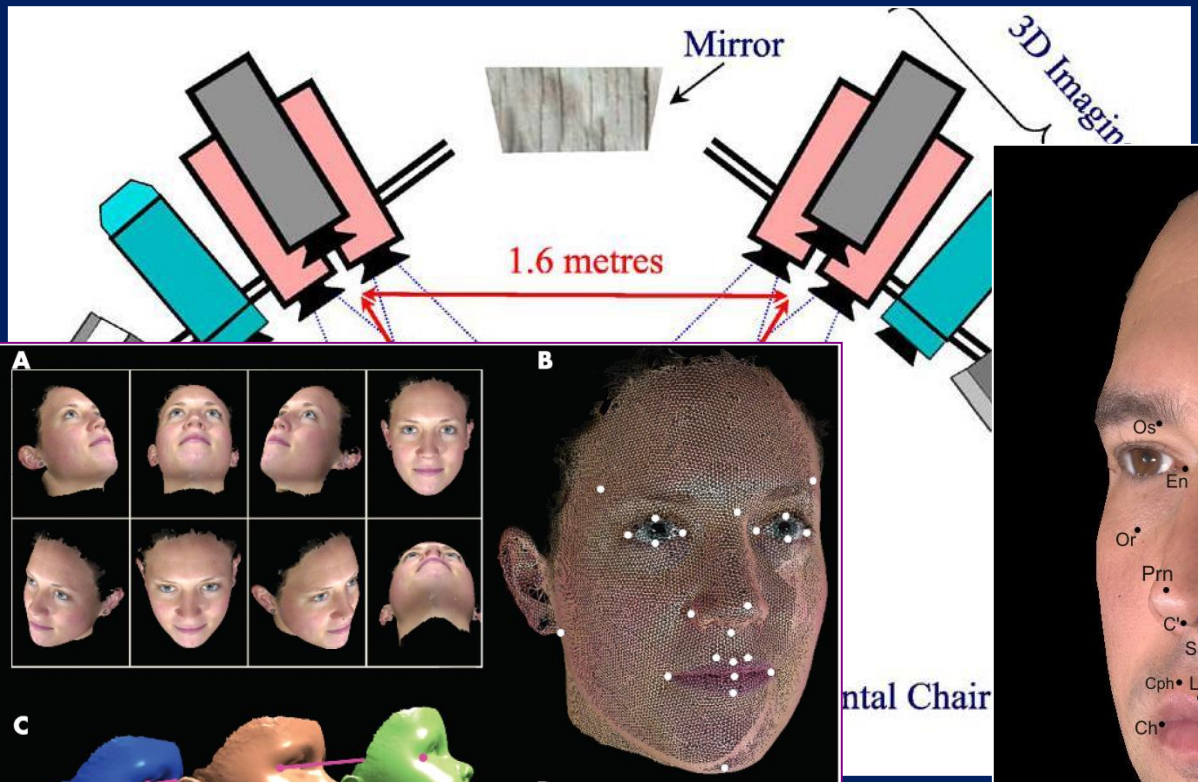


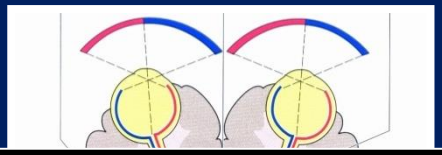


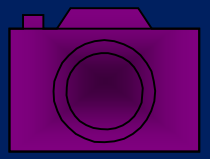
Each object is seen in each istant by 2 TVCs (x, y): stereoscopic vision (x, y, z)

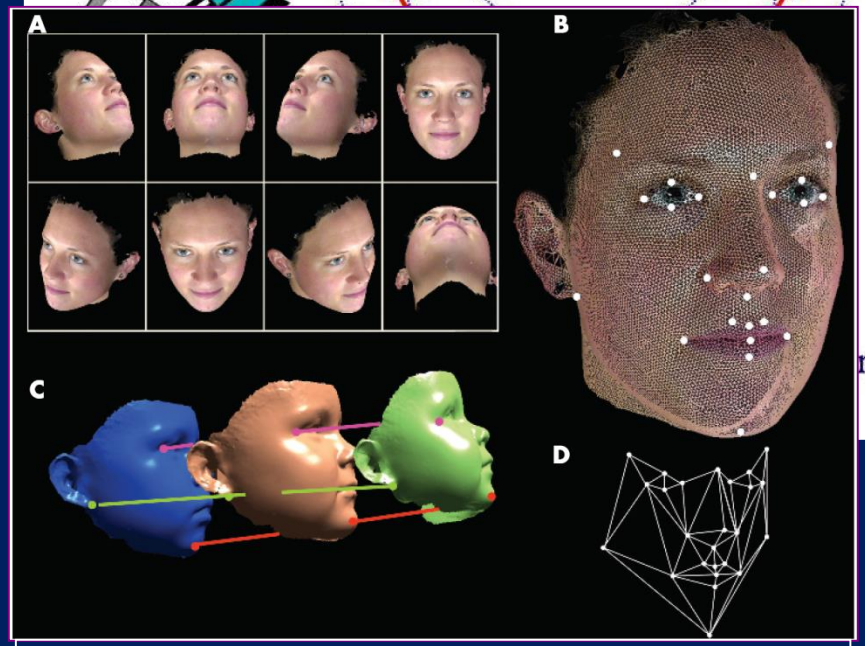
Ras et al. J Dent 1996;24:369-74
Burke & Hughes-Lawson, AJODO 1989;96:144-51

Stereophotogrammetry

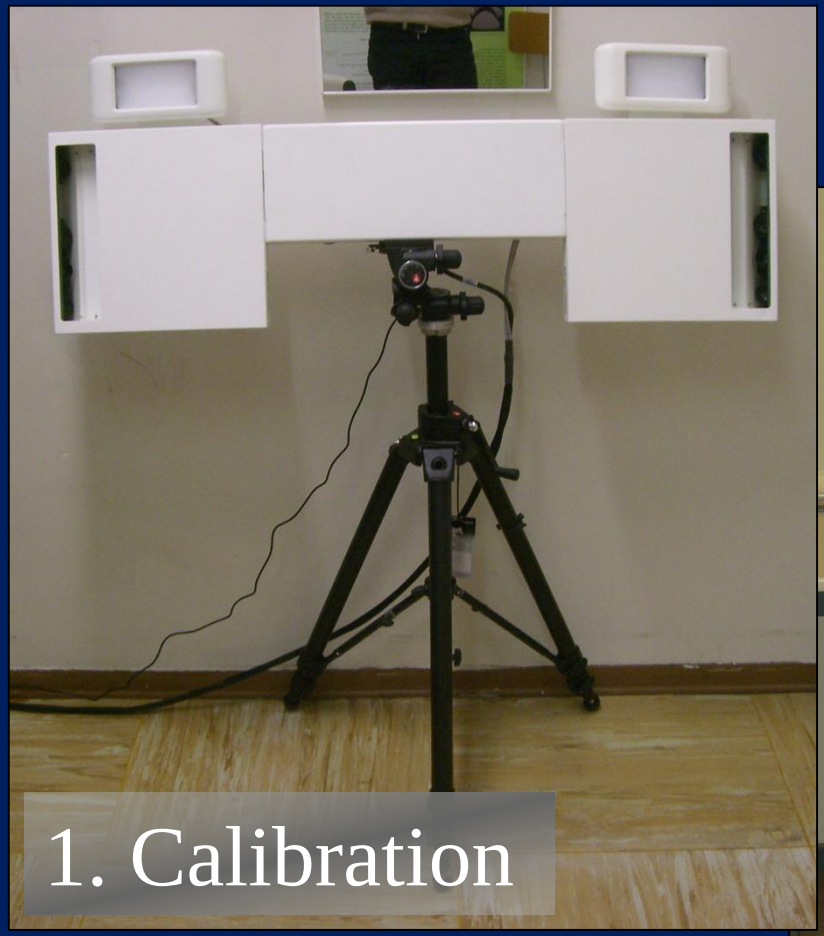








Hammond, Arch Dis Child 2007; 92:1120-6



1. Calibration

2. Acquisition

Stereophotogrammetry

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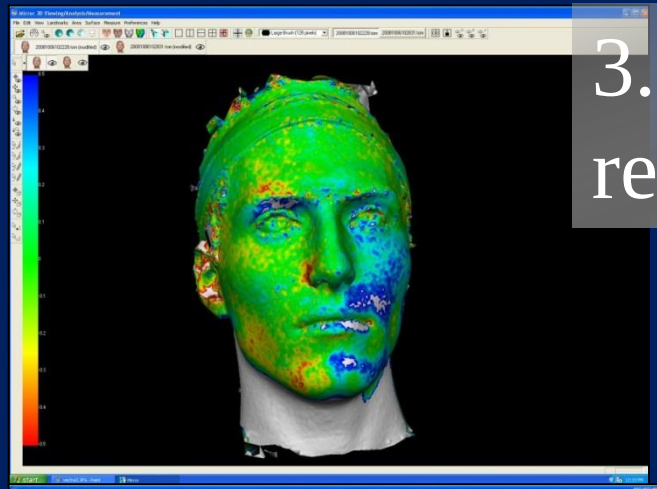
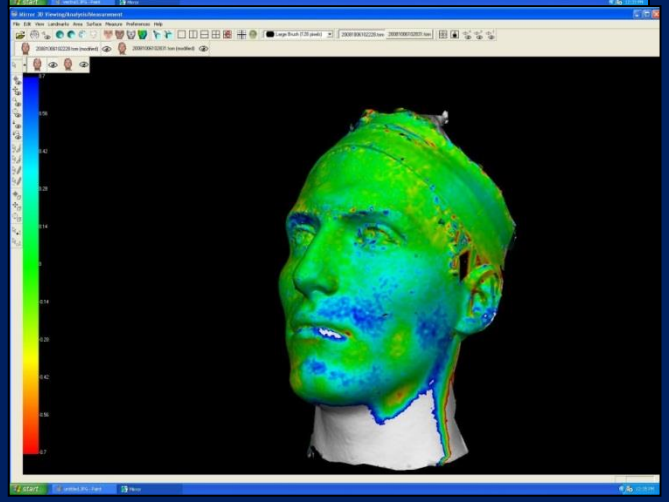
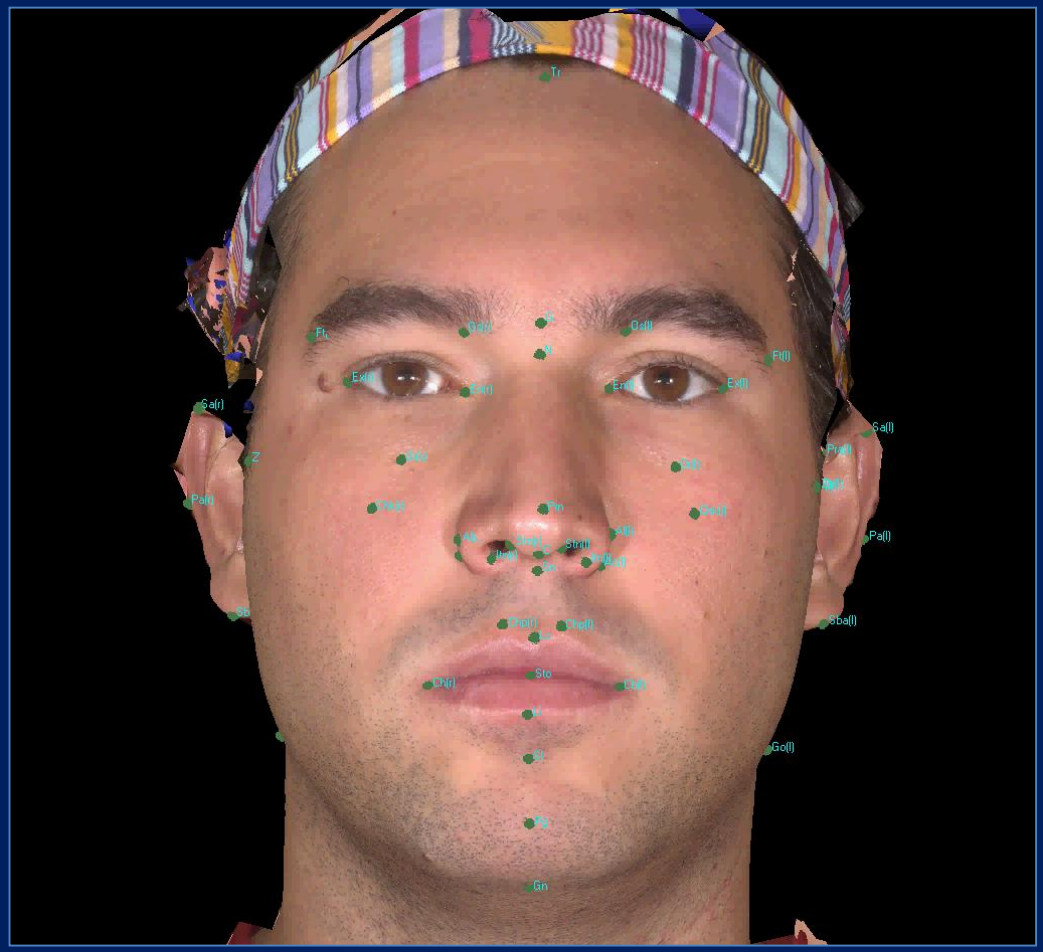




3. Digital reconstruction

4. Landmark digitization & calculations



Stereo-photogrammetry

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- Easy-to-manage data ??
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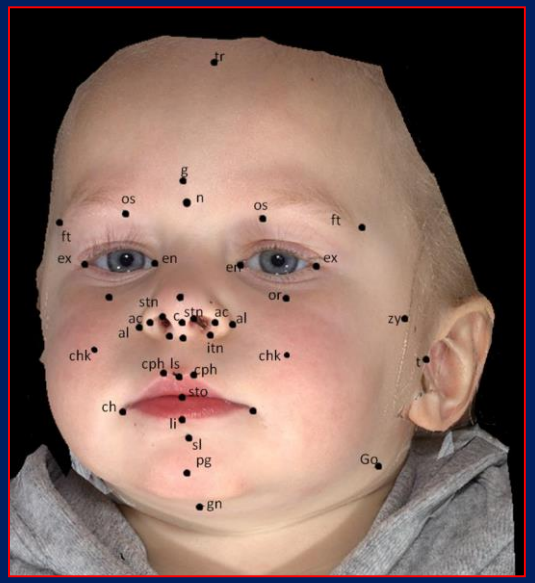
Optical methods

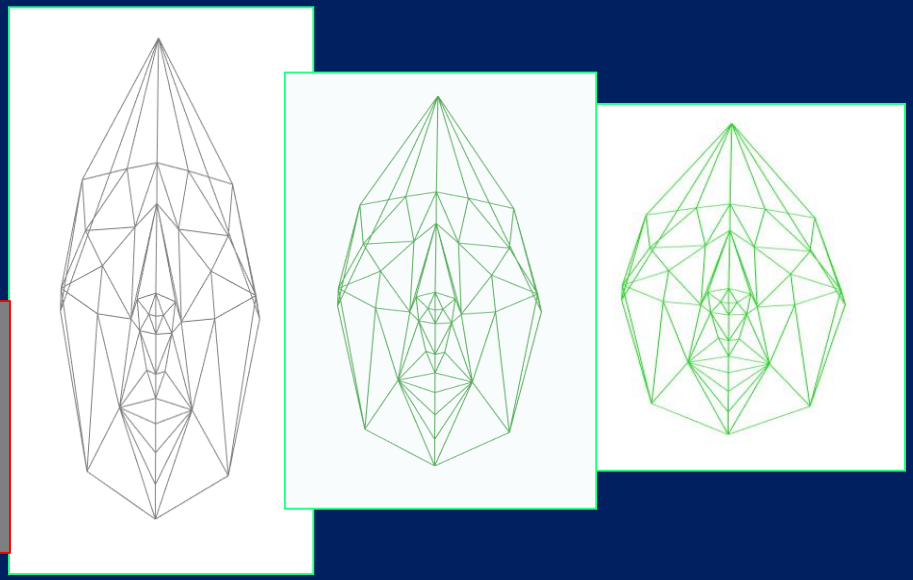
Università degli Studi di Milano, via Festa del Perdono 7 (MM2 Duomo)

Subjects & Methods

- 11 Glut-1 syndromic subjects
- Females, 3 – 32 years
- 205 control subjects
- 42 landmarks for each face (ears excluded)

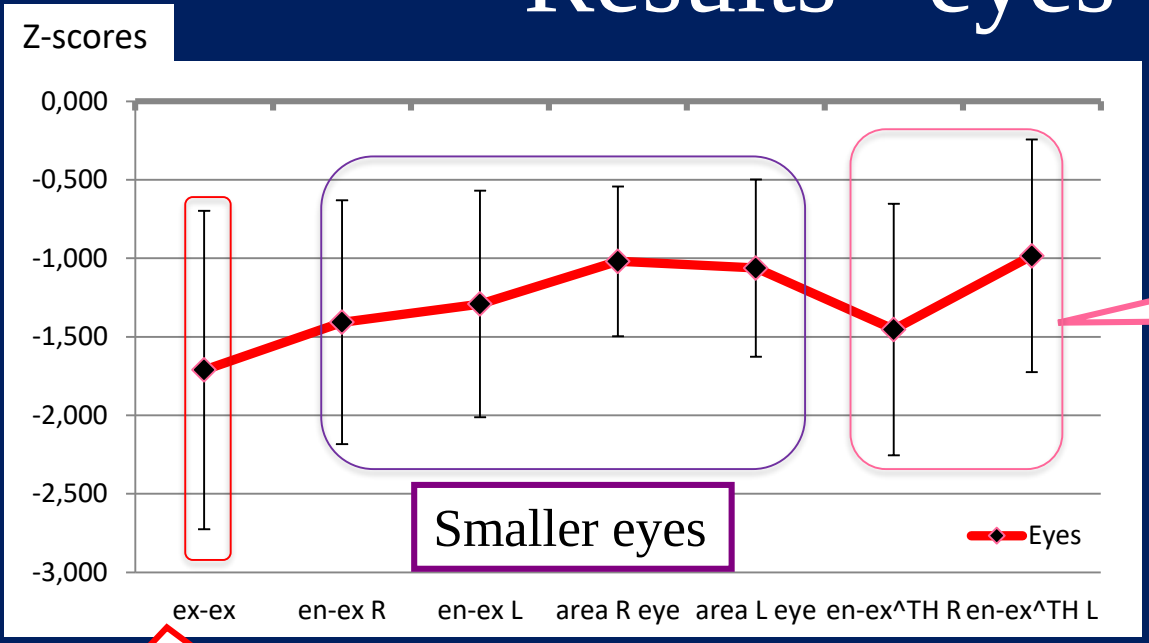


1. Landmark identification (skin)
2. Stereophoto
3. Landmark digitization (PC)
4. Calculations



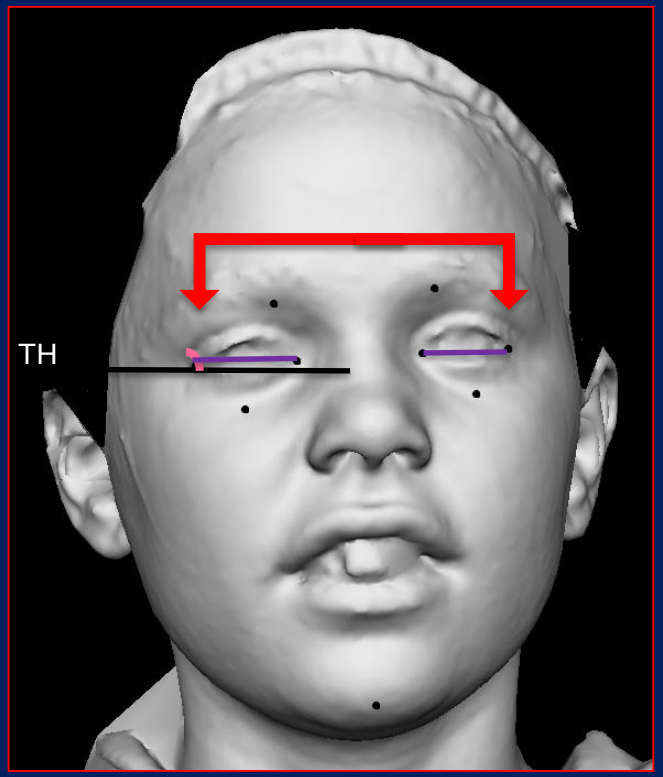
a. Landmark to landmark distances, ratios and angles (z-scores)
b. Principal Component Analysis

Results - eyes

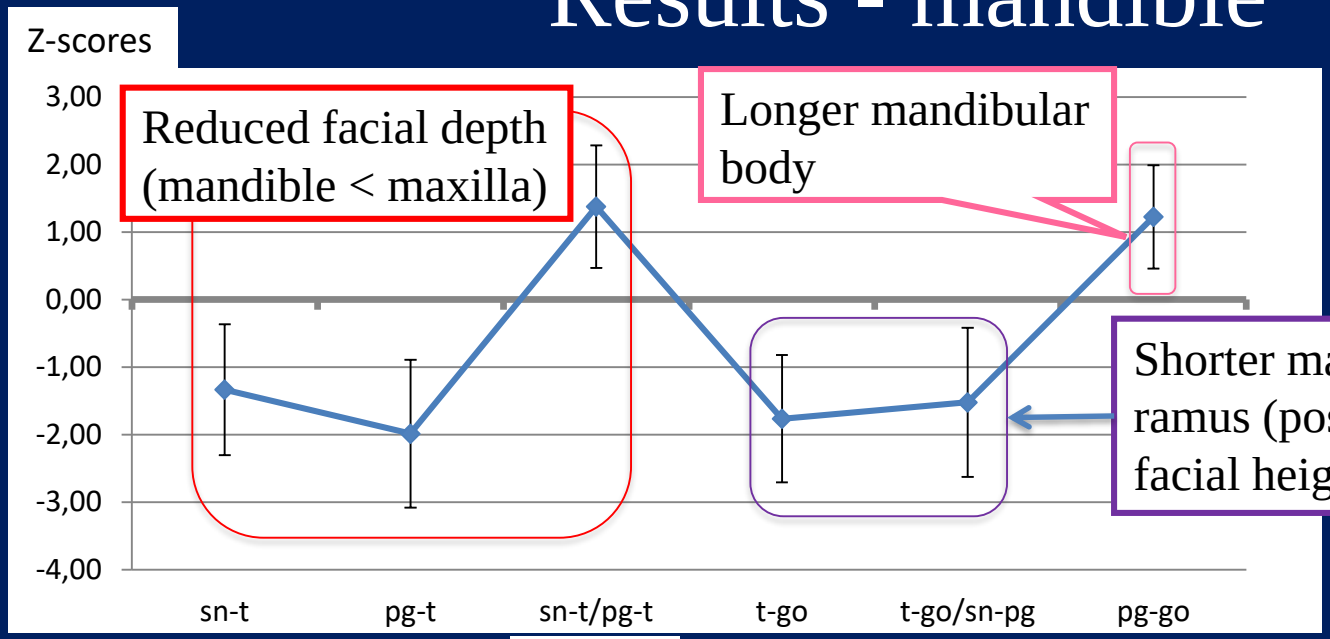


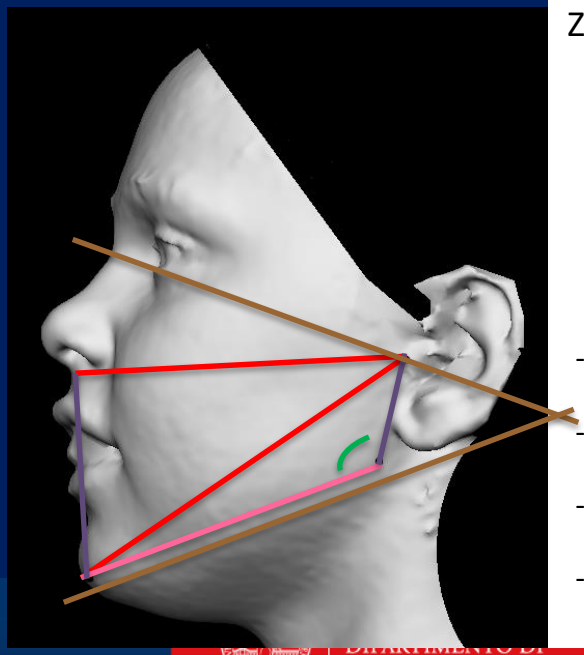
Down-slanted eyes

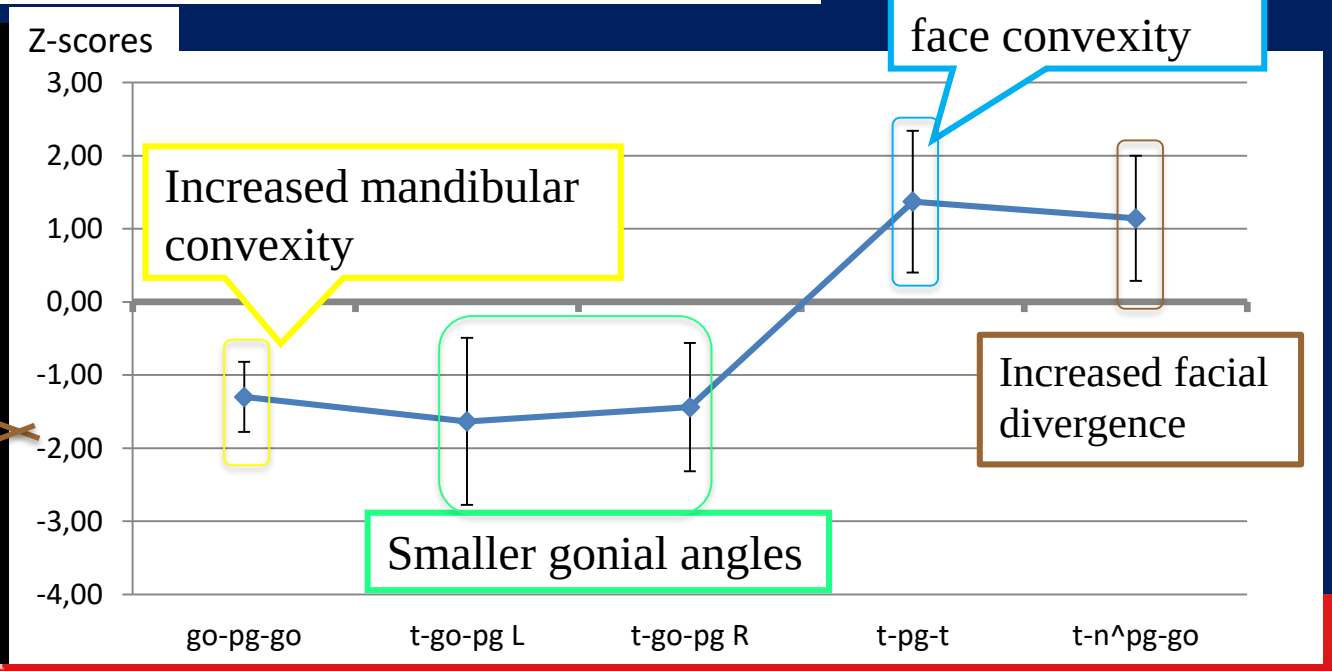
Reduced intercanthal distance



Results - mandible







Principal Component Shape Analysis

i) Procrustes registration
ii) Intra group analysis (controls, Glut1)
iii) Inter groups analysis & stepwise linear regression

205 control subjects

- 1st principal component: 43% of face variability
- 1st-70th components: 99% of face variability

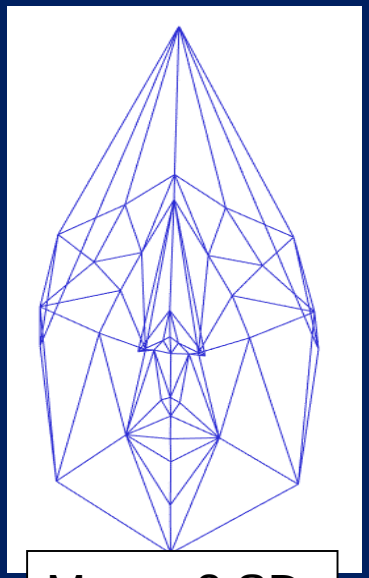
11 Glut-1 subjects

- 1st principal component: 24.9% of face variability
- 1st-9th components: 97% of face variability

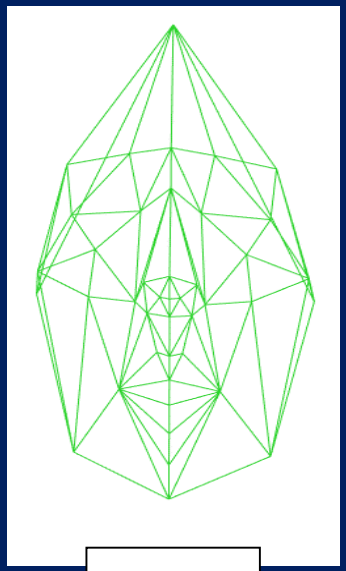
Glut1: more homogenous faces

PC1 – control subjects

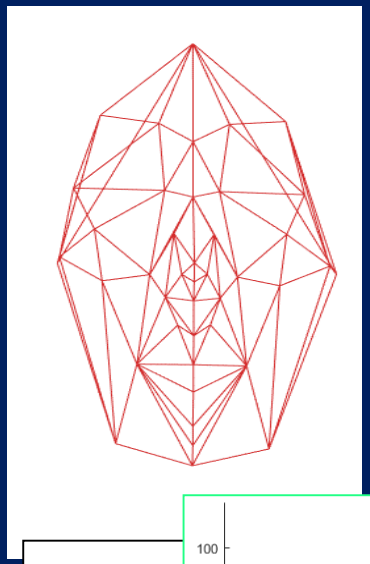
explained variance: 43.6%



Mean -2 SD

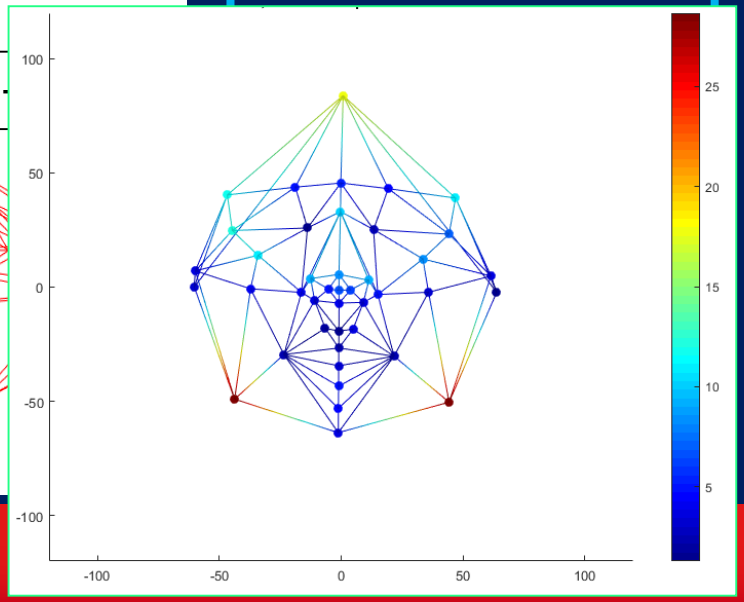


Mean



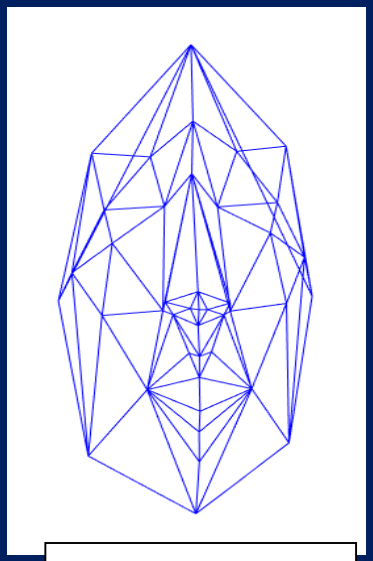
Mean -

Vertical vs. anterior-posterior development of face;

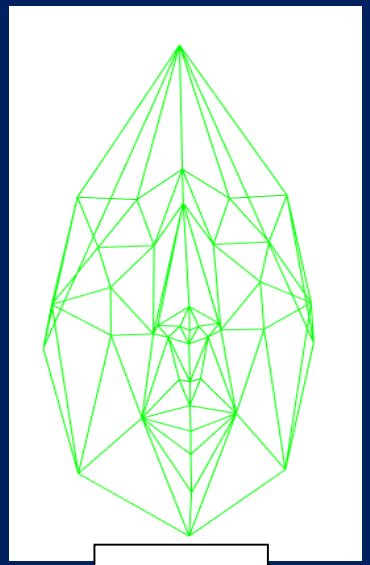


PC1 – Glut1 subjects

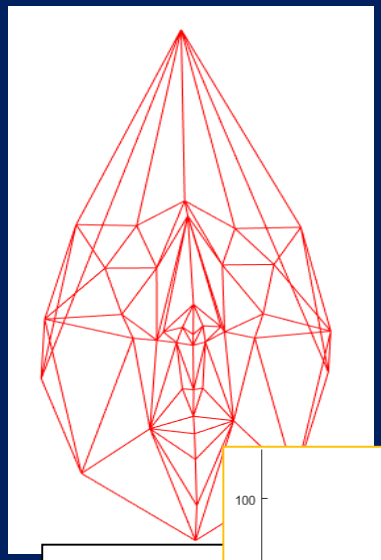
explained variance: 24.9%



Mean -2 SD

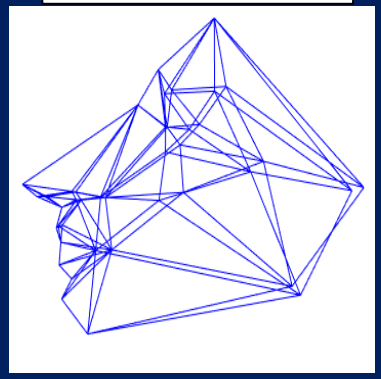


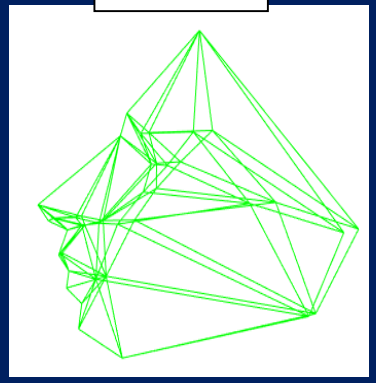
Mean

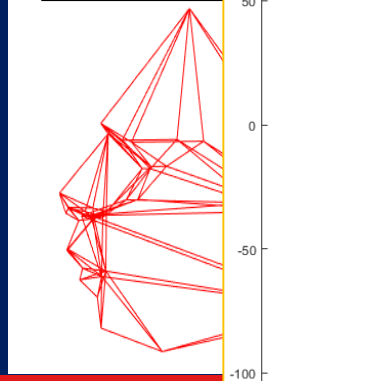


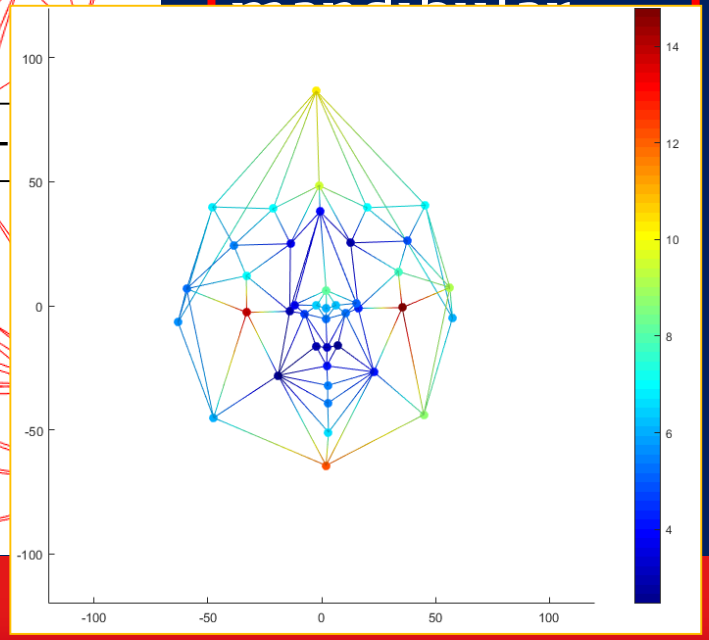
Mean +

Forehead;
nose
prominence;
facial width;
mandibular









mm

Glut1 vs. Control subjects

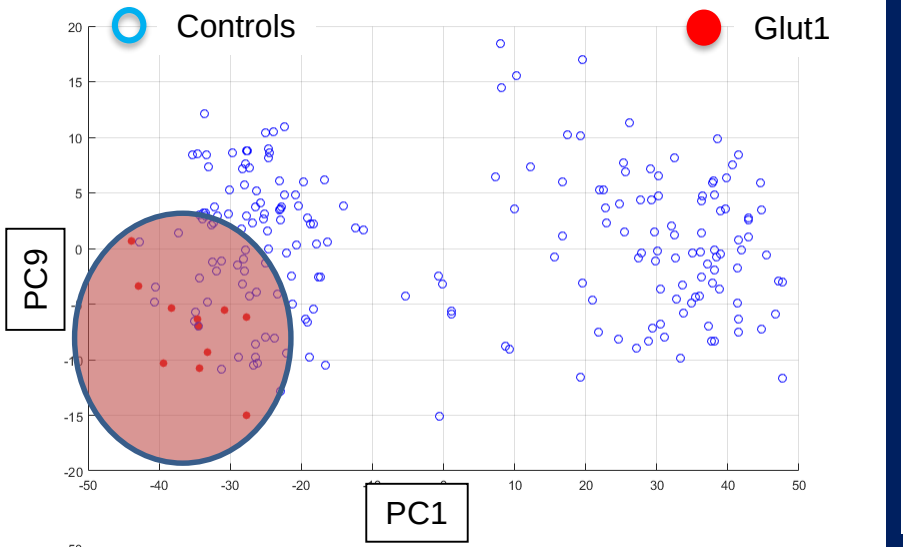
PCA:

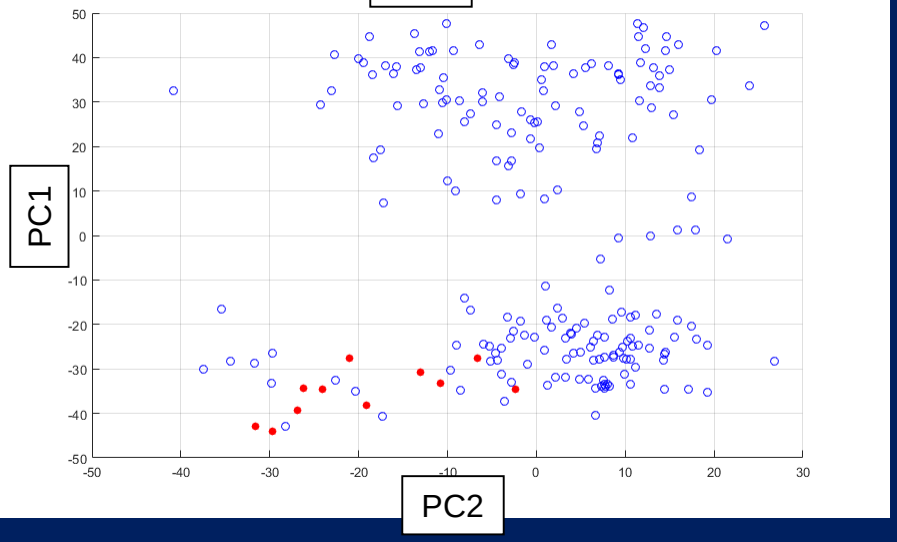
- 1st principal component: 43% of face variability
- 1st-71st components: 99% of face variability

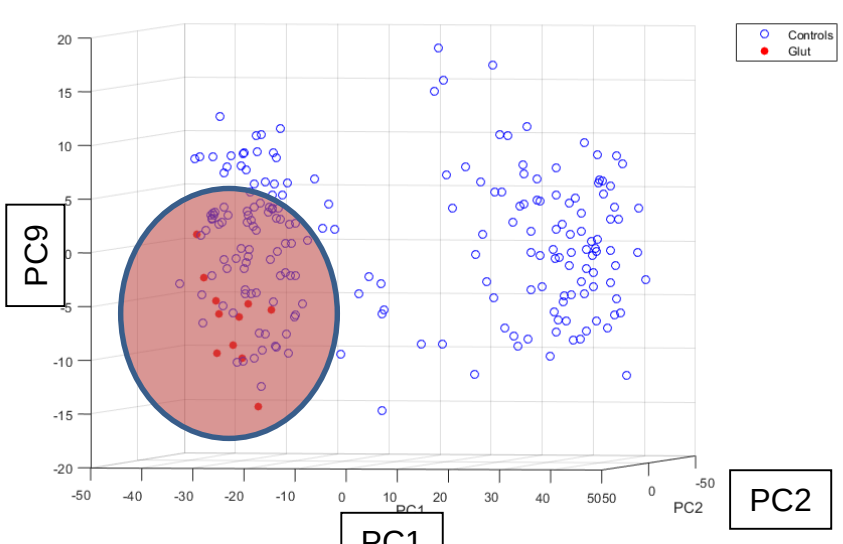
Stepwise linear regression: PC1, PC2, PC9

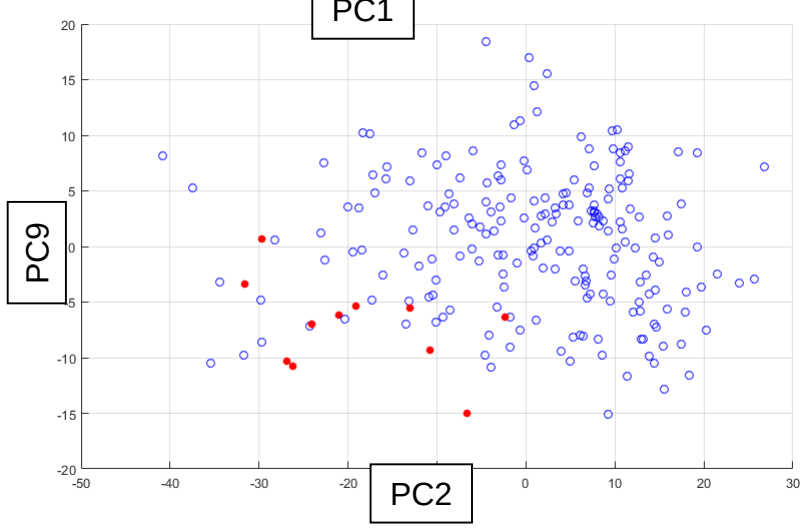
PC1 p < 0.000	*PC13 p = 0.004*
PC2 p < 0.000	*PC14 p = 0.010*
PC3 p = 0.009	*PC21 p < 0.000*
PC6 p < 0.000	*PC22 p = 0.005*
PC9 p < 0.000	*PC23 p = 0.028*
PC11 p= 0.012	*PC36 p < 0.000*

Stepwise linear regression



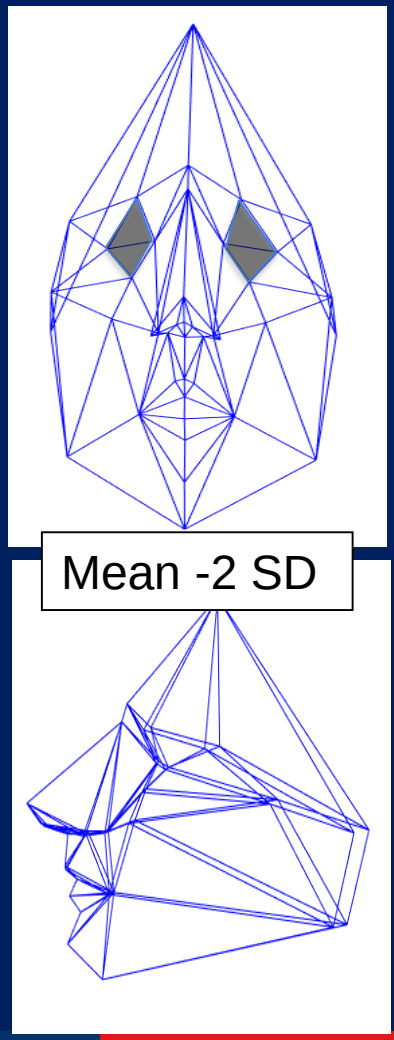


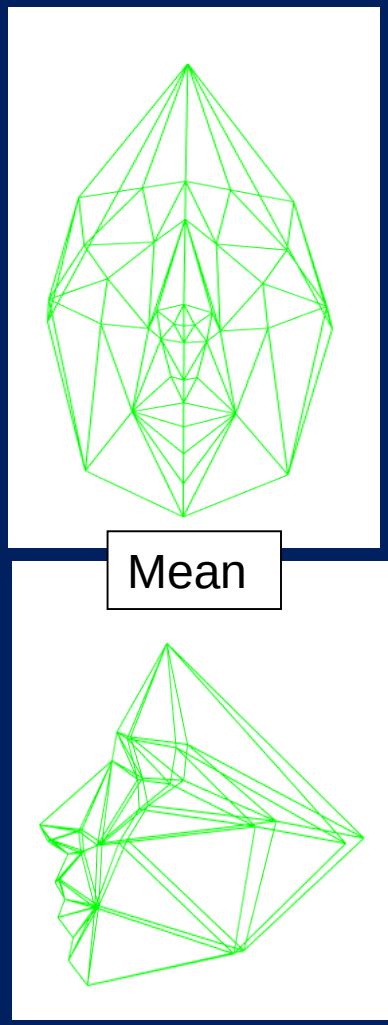


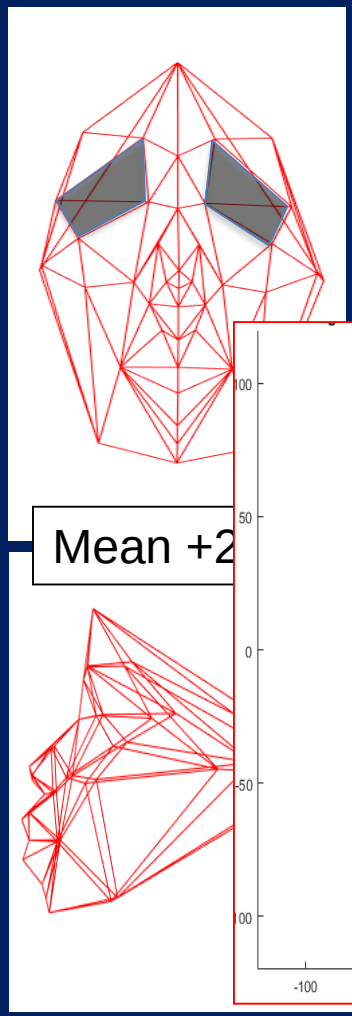


PC1 – Glut1 vs control subjects

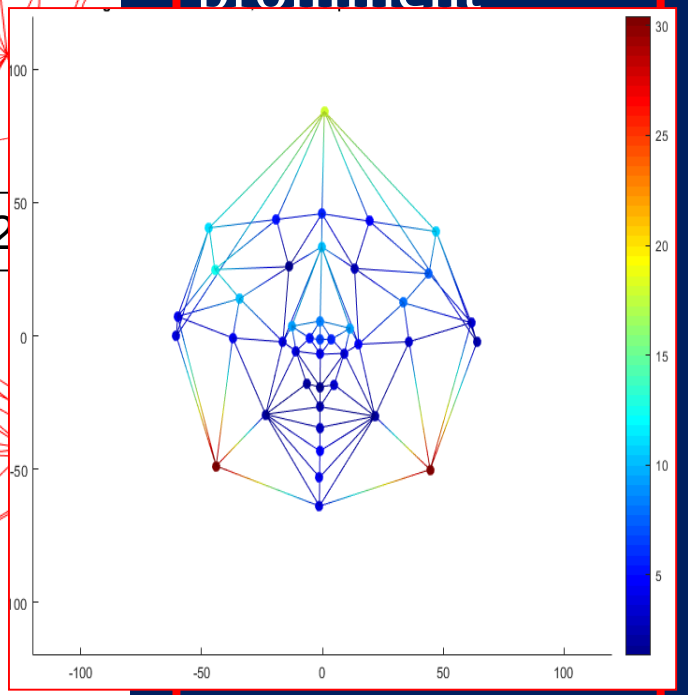
explained variance: 43.6%





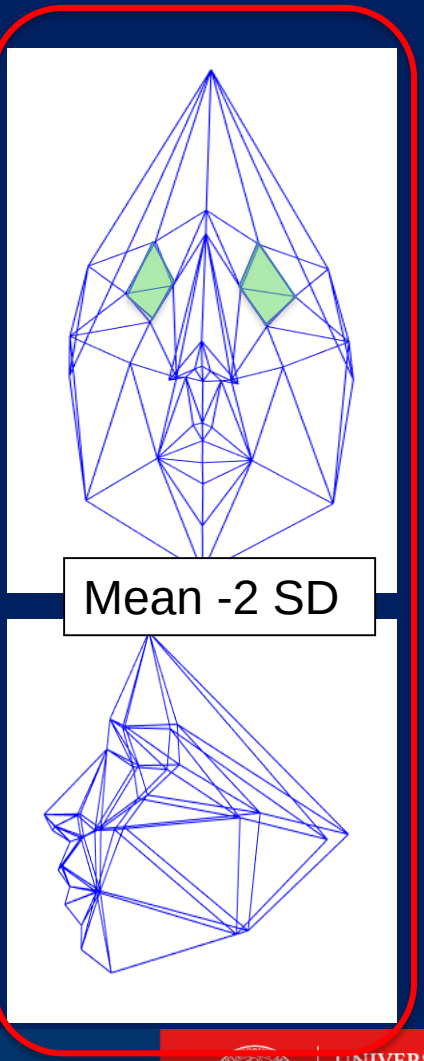


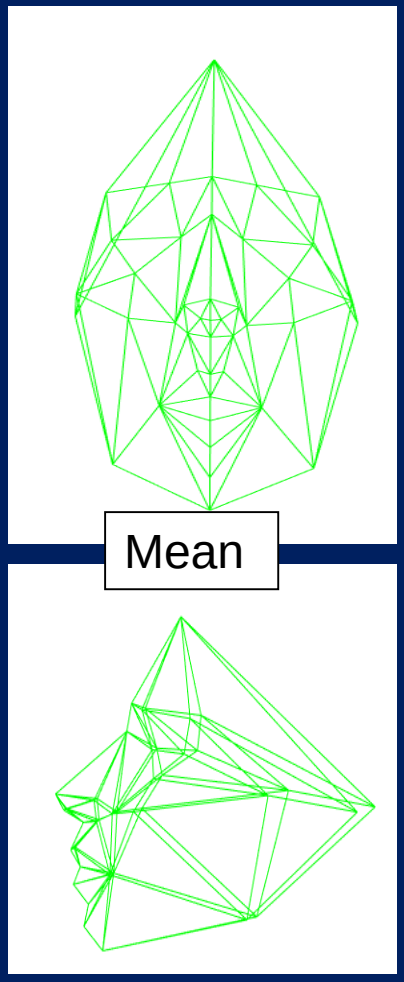
Glut1: high forehead; smaller eyes; prominent

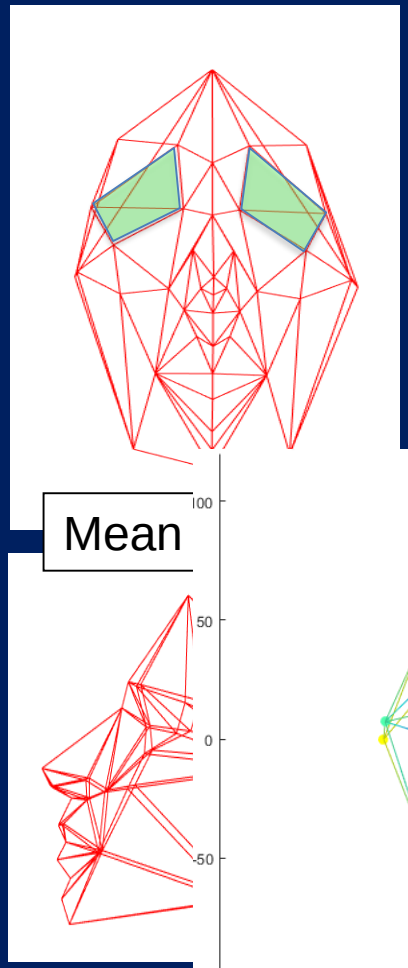


PC2 – Glut1 vs control subjects

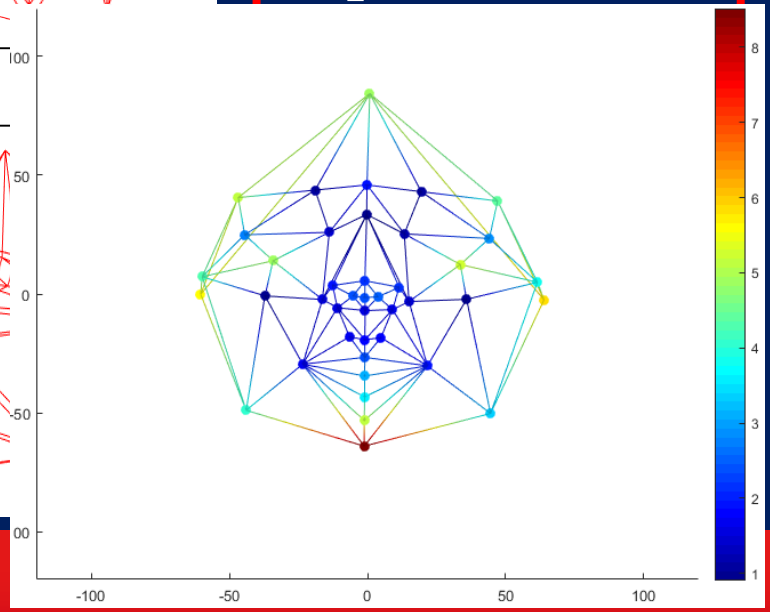
explained variance: 8.6%





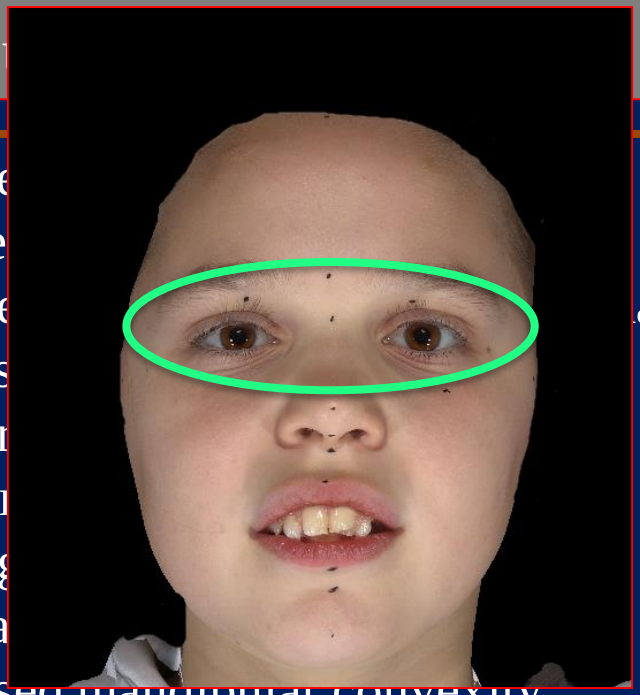


Glut1: high forehead; smaller eyes; reduced facial depth;



Glut1 vs control subjects
3D facial features

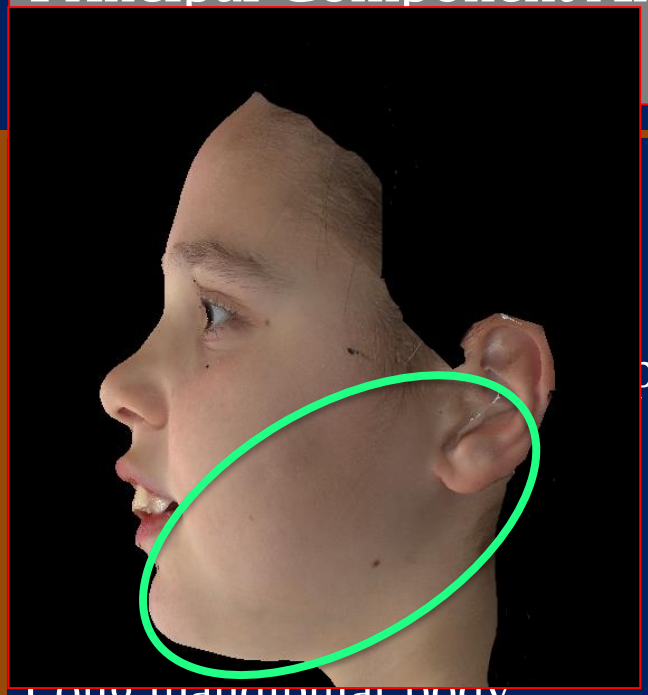
Landmark to landmark meas



Reduce
Smalle
Reduce axilla)
Increas
Longer
Shorter
Small g
Decrea
Increased mandibular convexity

Mandibular prognathism:
1p36; 1p22.3; 1p22.1; 1q32.2
Glut1DS: 1p35-31.3

Principal Component Analysis



oth
Long mandibular body
Small gonial angle
Reduced chin prominence

Am J Orthod Dentofacial Orthop 2014;145:757-62; J Dent Res 2009;88:56-60; J Dent Res 2015;94:569-76

Laboratorio di Anatomia Funzionale dell'Apparato Stomatognatico (1989)

- Dr. Daniela Ciprandi
- Dr. Marina Codari
- Dr. Claudia Dolci
- Prof. Virgilio F. Ferrario
- Dr. Daniele M. Gibelli
- Giulia Andrea Guidugli
- Dr. Luca Pisoni
- Dr. Valentina Pucciarelli
- Francesca M.E. Rusconi
- Dr. Matteo Zago

- Prof. Alberto Battezzati, UniMI
- Dr. Simona Bertoli, UniMI
- Dr. Valentina De Giorgis, UniPV
- Prof. Pierangelo Veggiotti, UniPV

Photo: Vaclav Sedy, 2007

UNIVERSITÀ DEGLI STUDI DI MILANO
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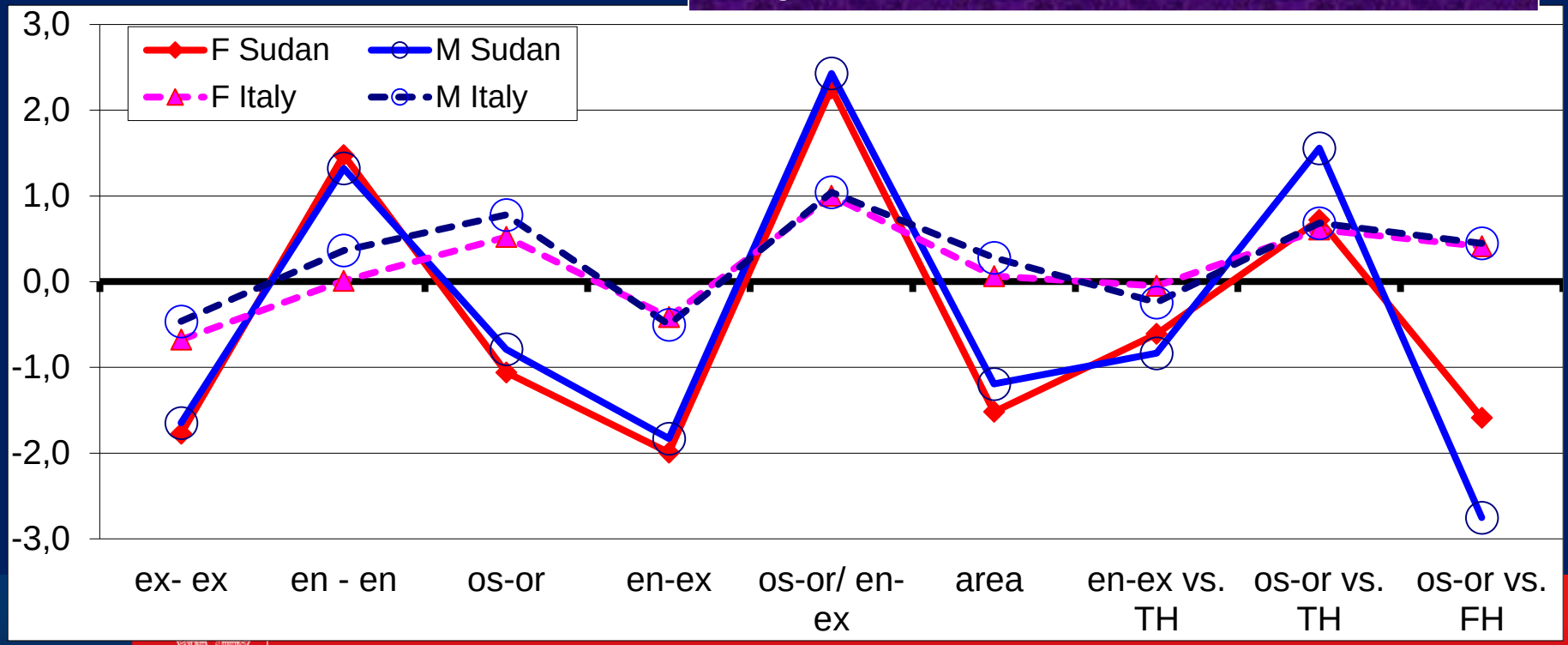
Down syndrome

Morphometry of the Orbital Region Soft Tissues in Down Syndrome

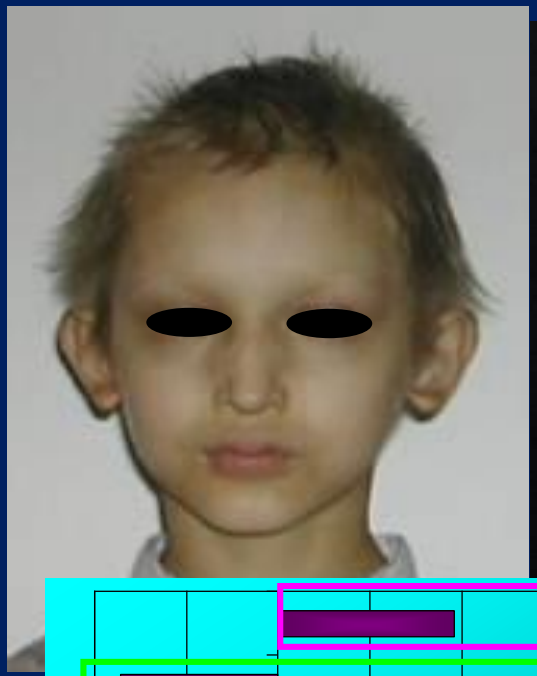
The Journal of Craniofacial Surgery • Volume 23, Number 1, January 2012

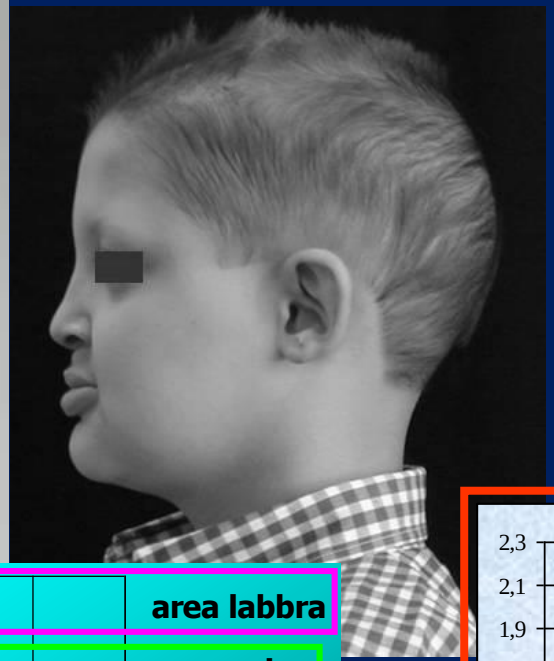
Chiarella Sforza, MD, PhD, Fadil Elamin, MSc, BSc, BDS, MOrthEd, FFDRCSI,† Claudia Dellavia, DDS, PhD,* Riccardo Rosati, DDS, PhD,* Gianluigi Lodetti, DDS,*‡ Andrea Mapelli, MBioEng,* and Virgilio Ferruccio Ferrario, MD, PhD**

Larger deviations in Sudan than in Italy; no sex differences.

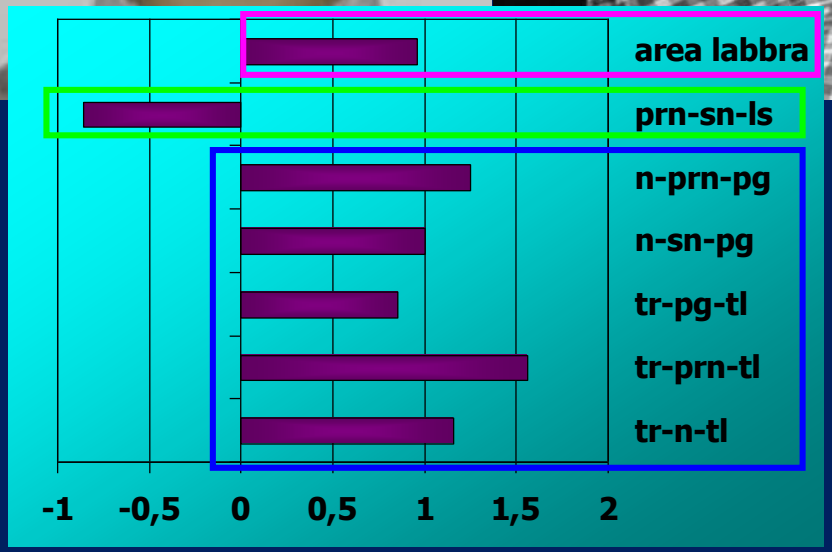


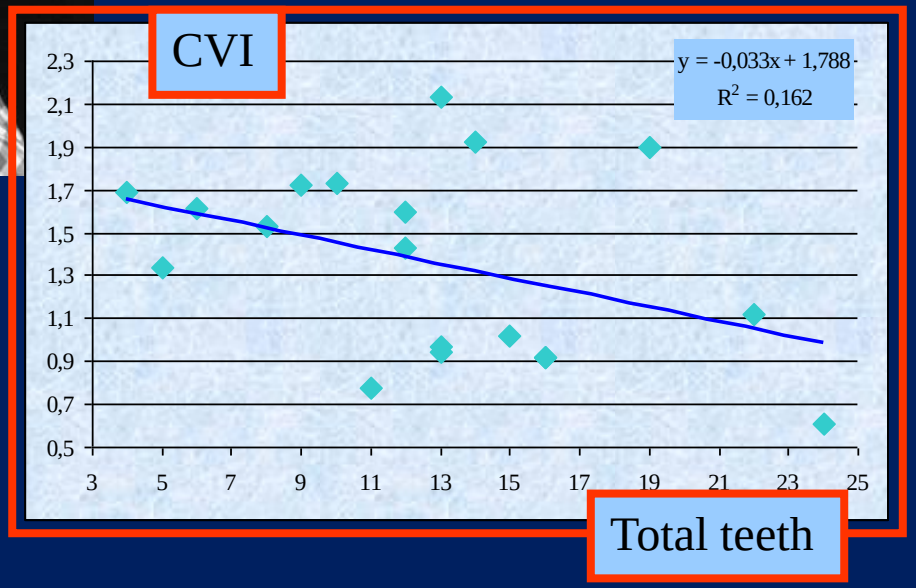
Hypohydrotic ectodermal dysplasia (OMIM #305100)





Flatter face, more acute nasolabial angle, prominent lips; less deviations with more teeth.

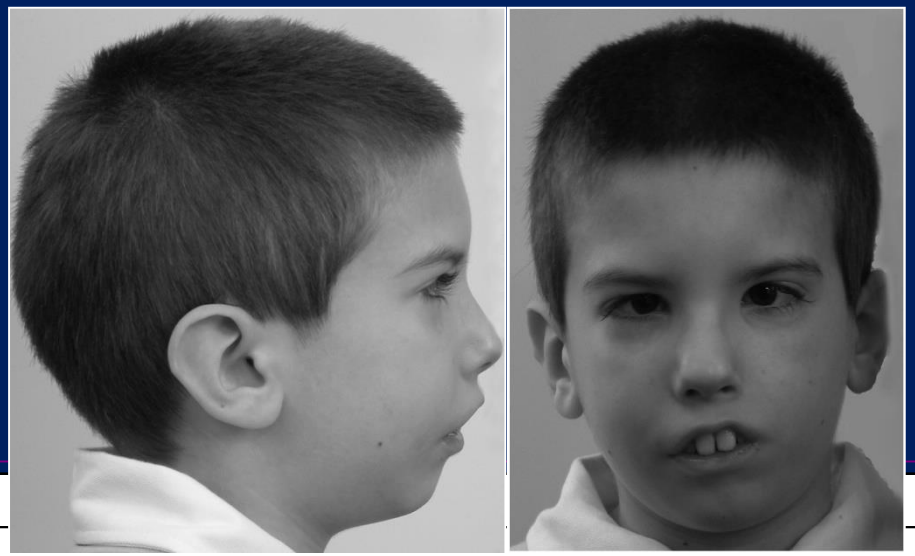


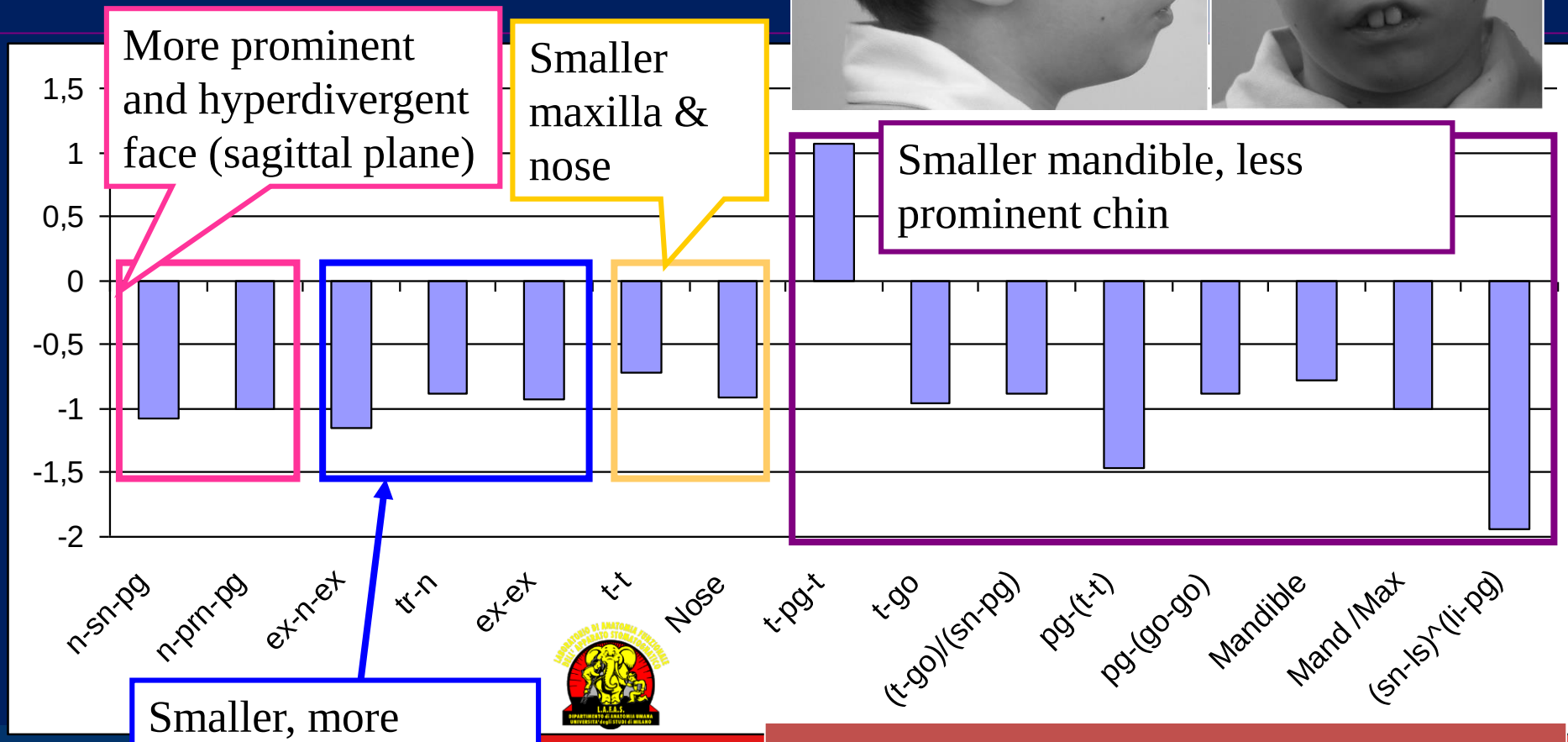


Ferrario et al. Am J Med Genet 2004; Sforza et al. Cleft Palate CJ 2004, 2006
Dellavia et al. Angle Orthod 2006, 2010; Eur J Oral Sci 2008

Moebius syndrome

Congenital facial palsy (facial nerve) & impairment of ocular abduction (abducens nerve) (OMIM %157900)





Sforza et al. Eur J Oral Sci 2009;117:695–703; It J Maxillofac Surg 2014;25:1-7

Photo: Vaclav Sedy, 2007