

Applications of AI to dysmorphology / AIDY

Quentin Hennocq

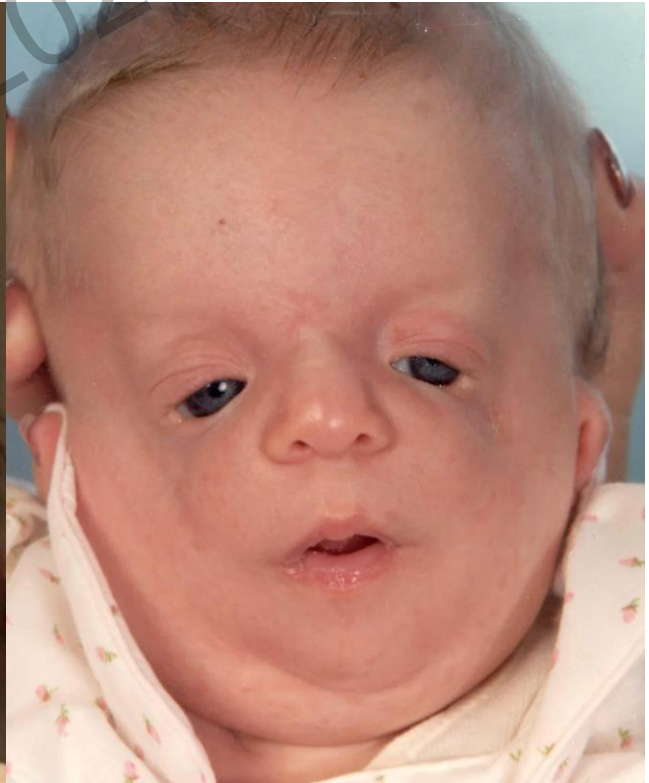
Imagine Institute – Hôpital Necker Enfants Malades / Laboratoire « Forme et croissance du crâne »

Supervised by Nicolas Garcelon & Roman Hossein Khonsari

Journée SOFFOET, 24/09/2021

Objectives are :

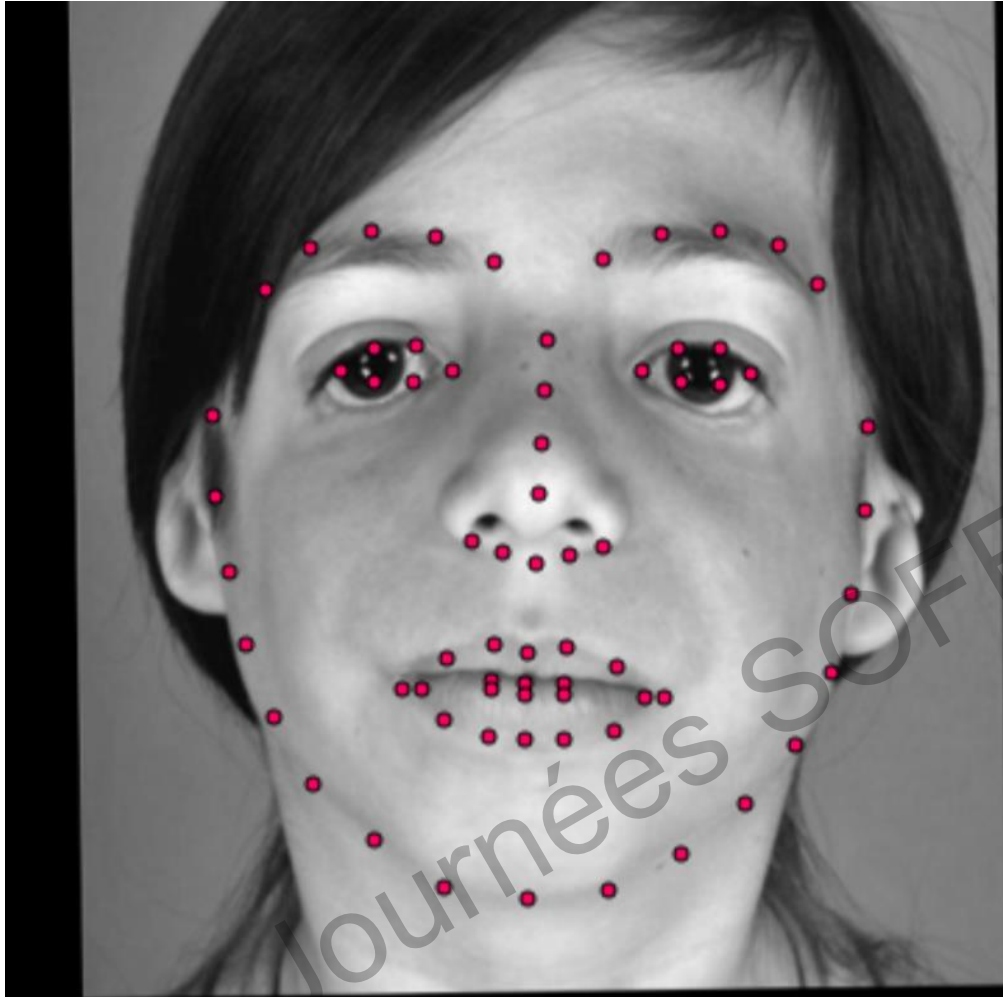
- 1) To **diagnose** syndromes with dysmorphic faces
- 2) To **understand** what are the recognized phenotypic features to diagnose a particular syndrome
- 3) To create **models** for each syndrome (as a function of age, ethnicity)
- 4) To **quantify the phenotypic differences** between syndromes or in one particular syndrome



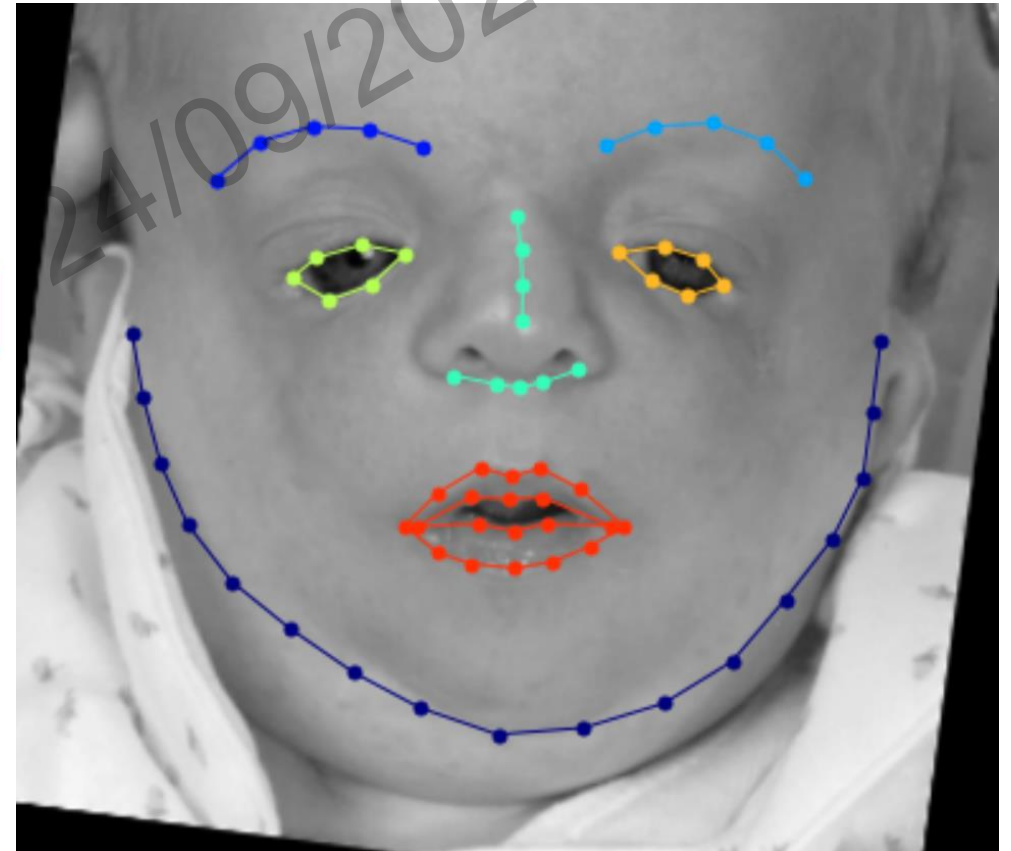
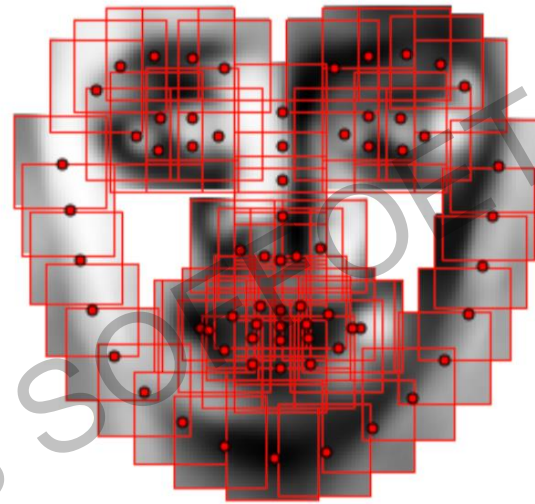
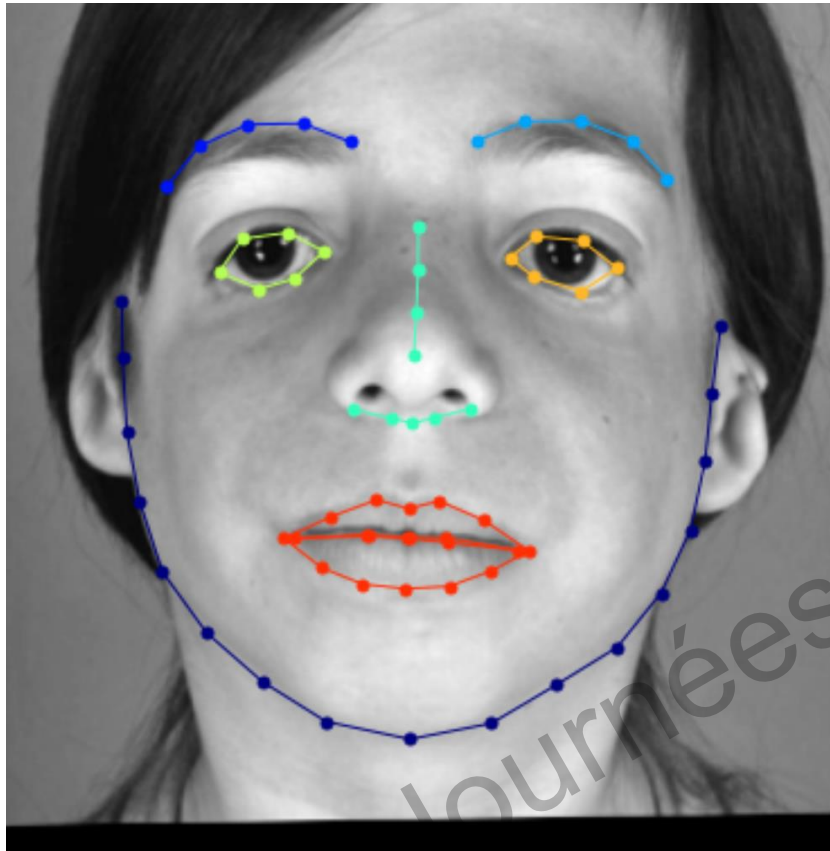
Jou...ees SOFF 09/2021



Belhumeur P et al. Localizing Parts of Faces Using a Consensus of Exemplars. Proceedings of the 24th IEEE Conference on Computer Vision and Pattern Recognition (CVPR), June 2011.



Active Appearance Models / Deep learning



Beichel R et al. Robust active appearance models and their application to medical image analysis. *IEEE Trans Med Imaging*. 2005 Sep;24(9):1151-69.

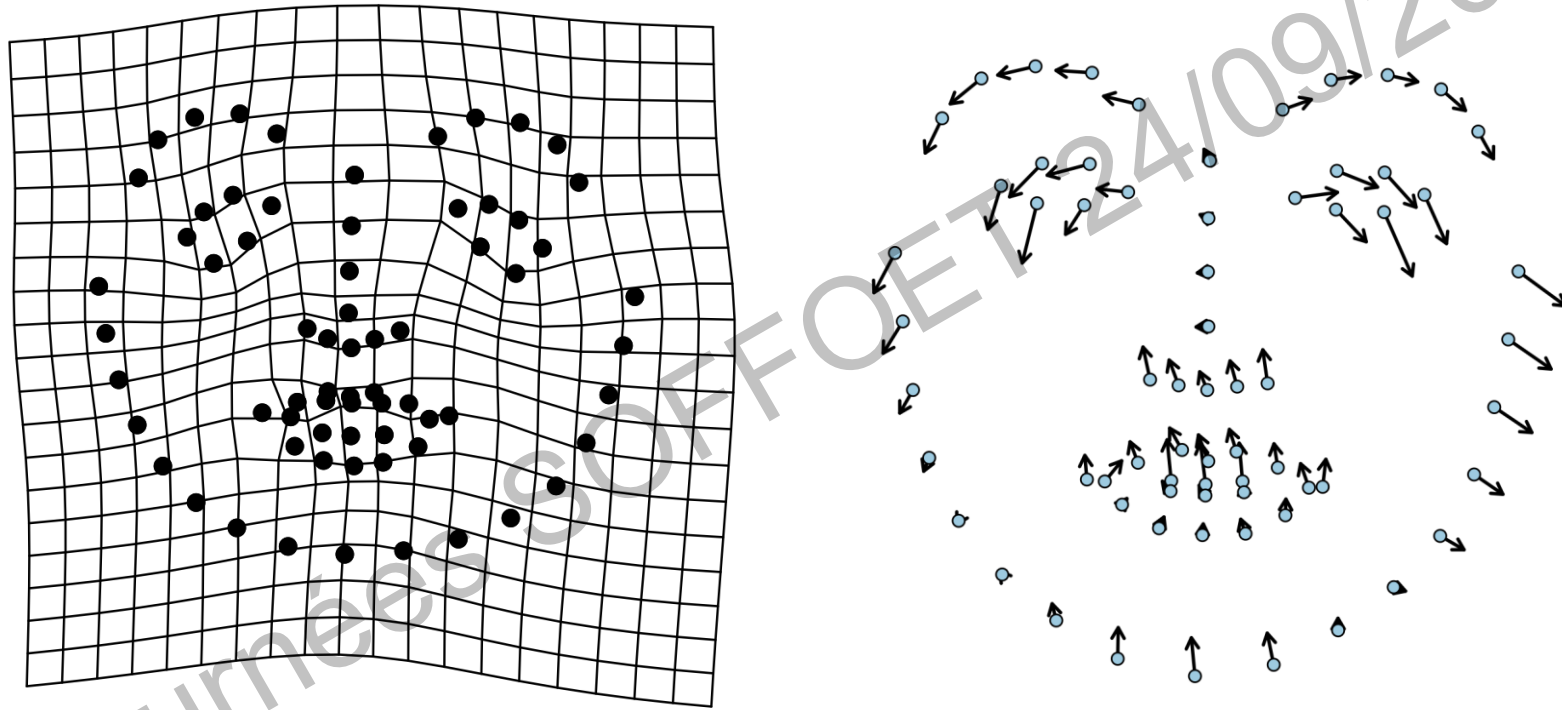
Procrustes analysis



Controls

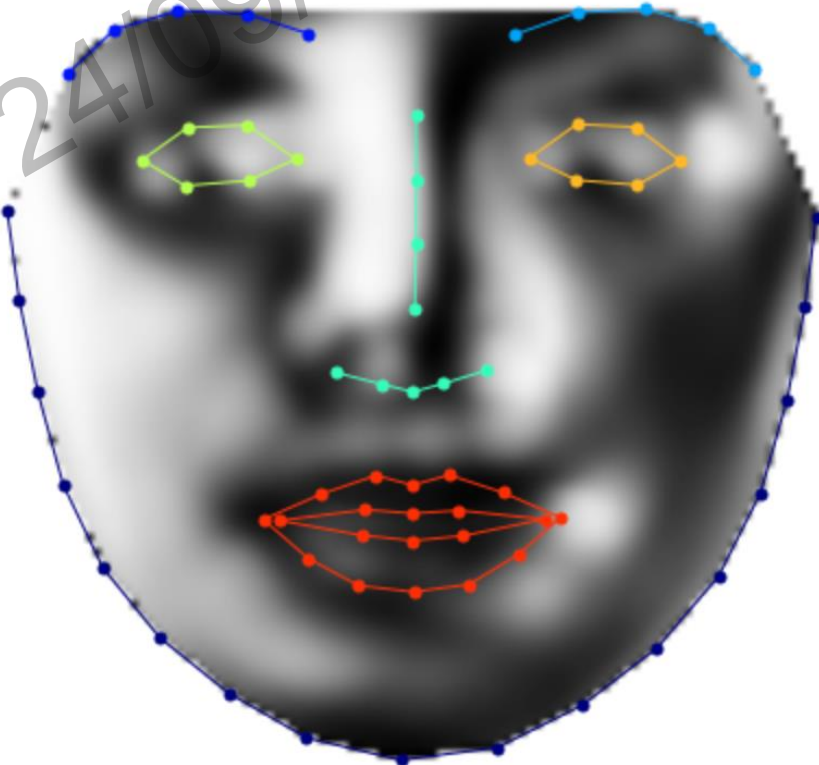
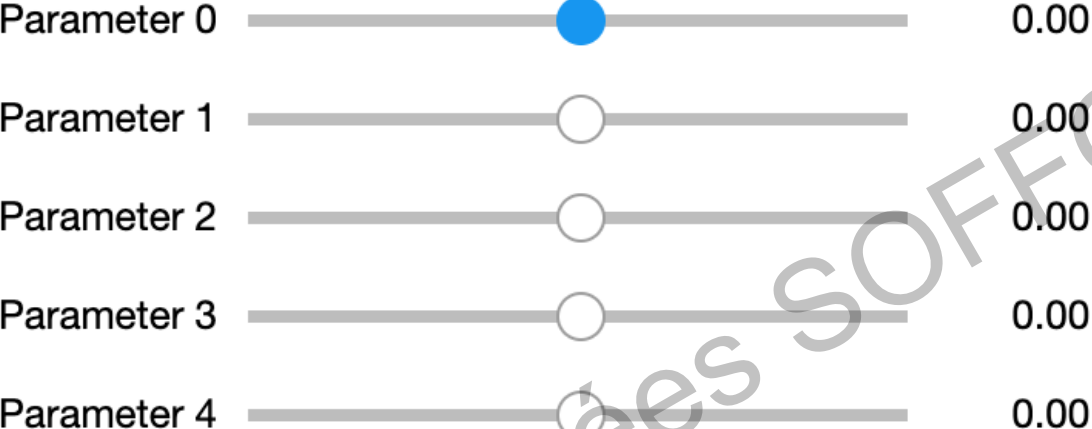
TC

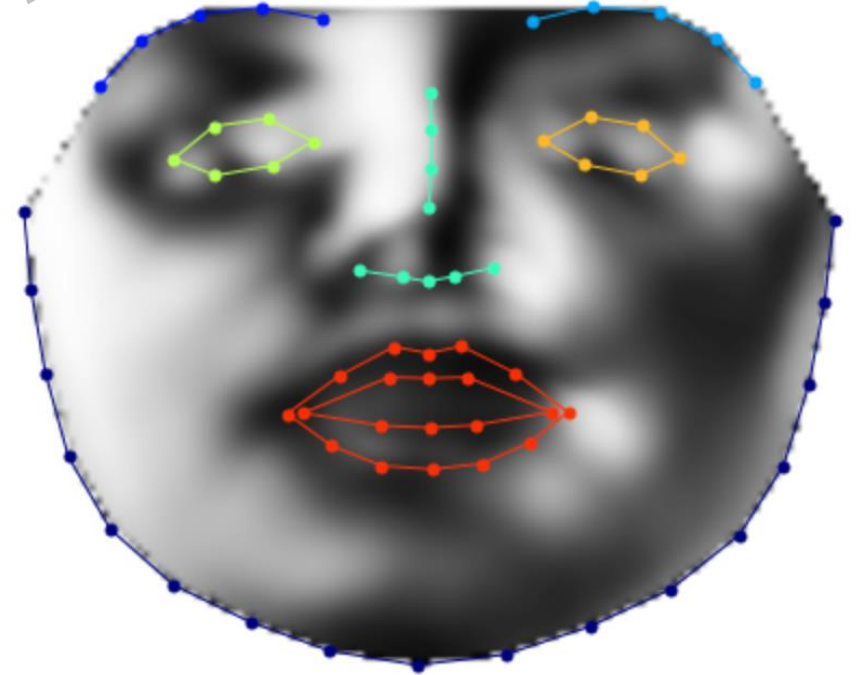
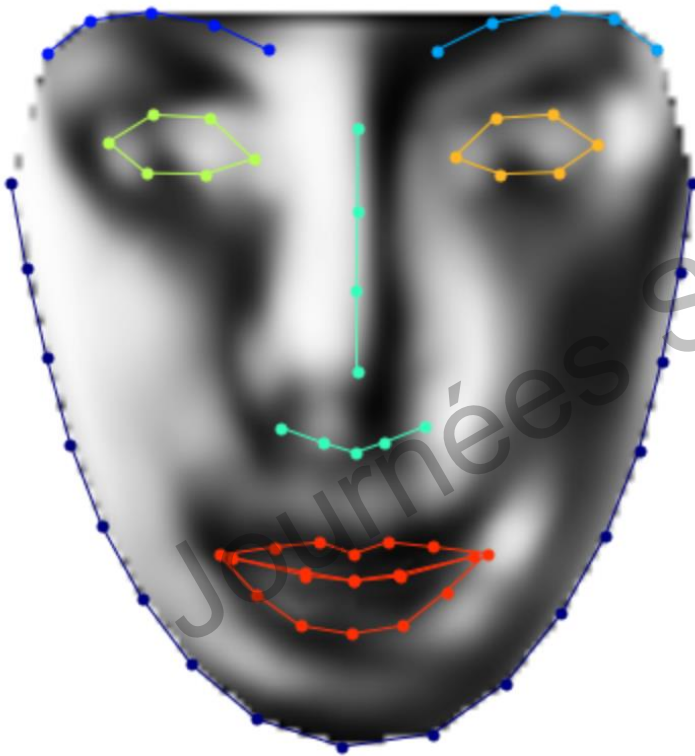
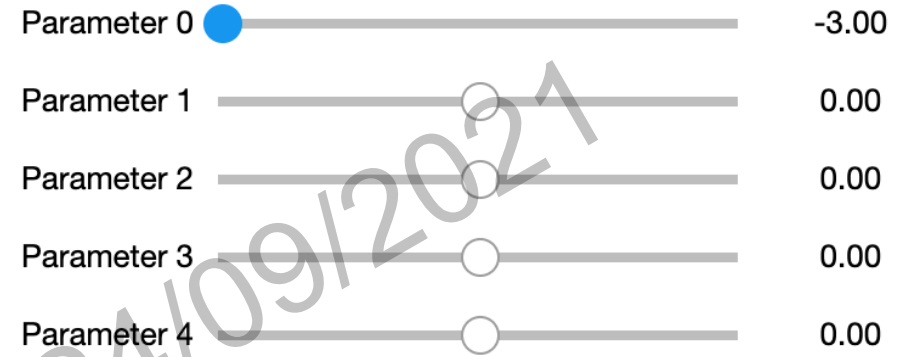
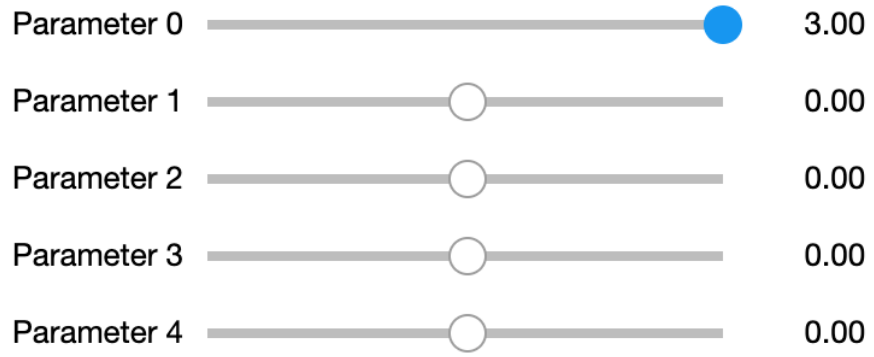
Procrustes analysis



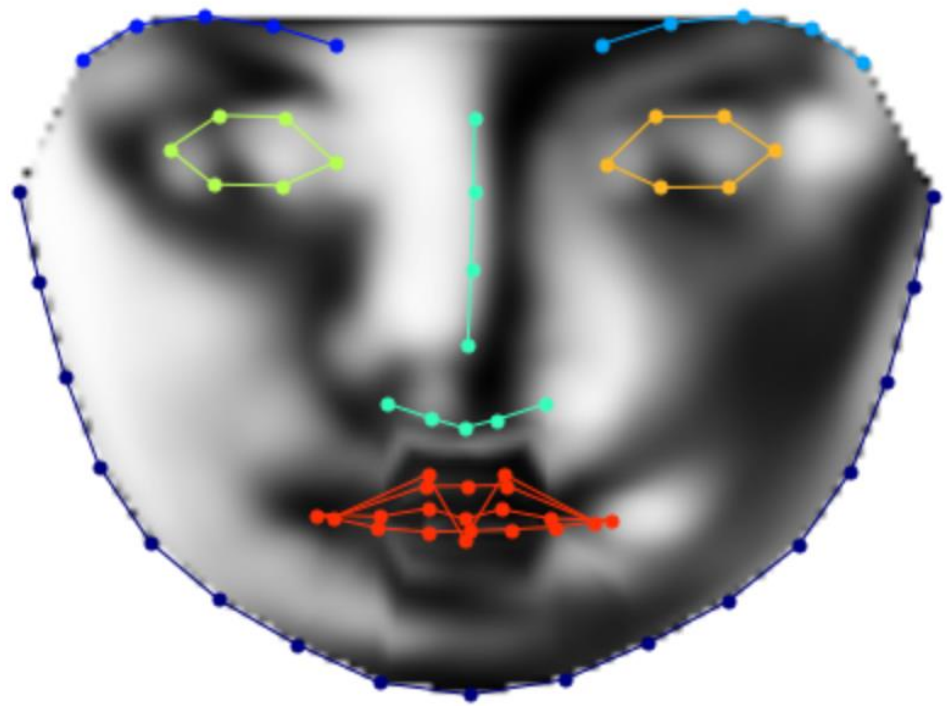
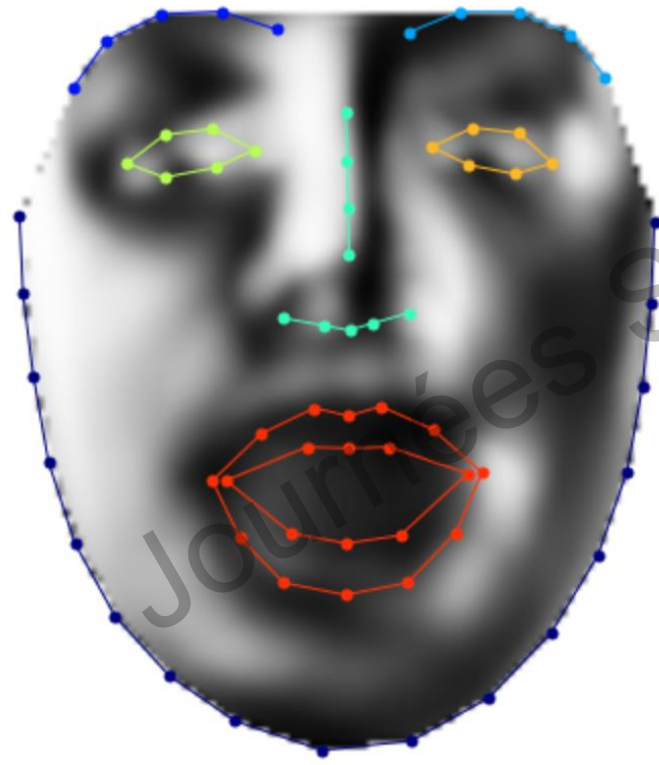
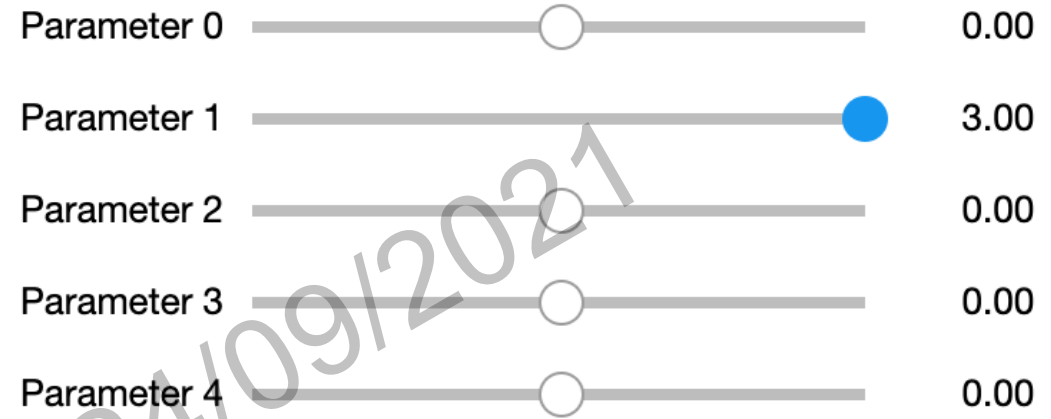
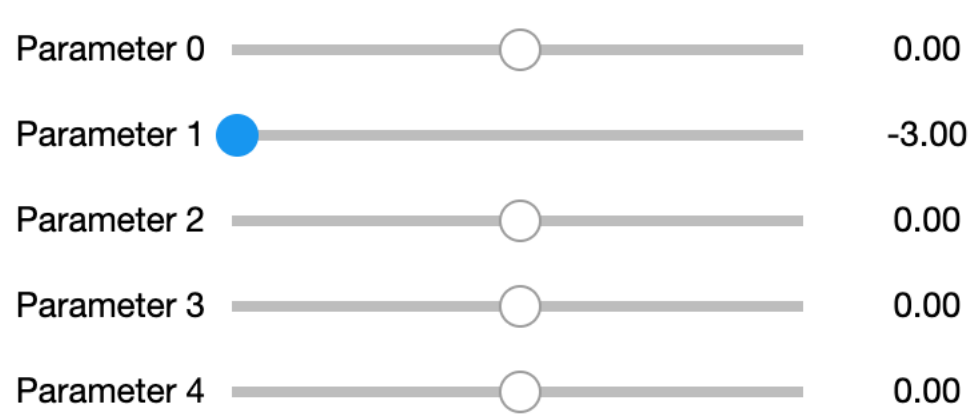
Controls vs. Treacher Collins syndrome

Dimension reduction



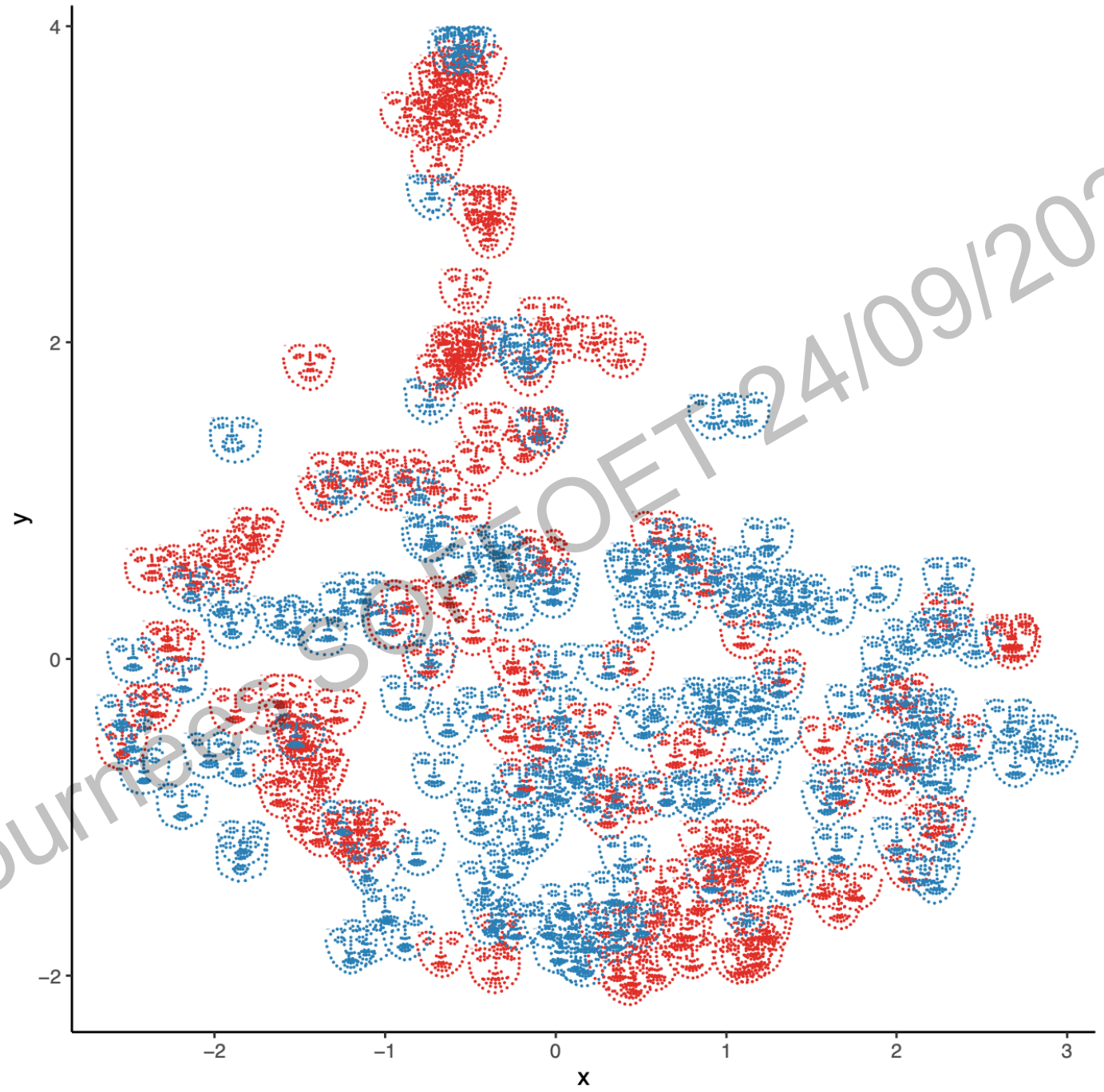


Journées SOFFOET 24/09/2021



Journal Pre-proof
SOFFOET 24/09/2021

Journals@SOET 24/09/2021



Machine learning

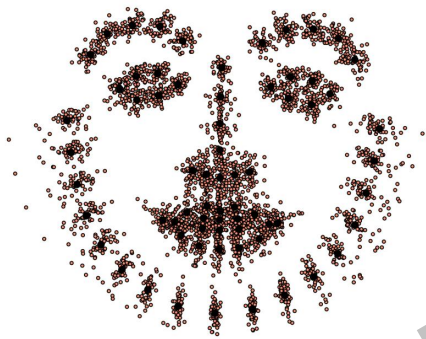
Coordinates



Journées SOFFOET 24/09/2021

Machine learning

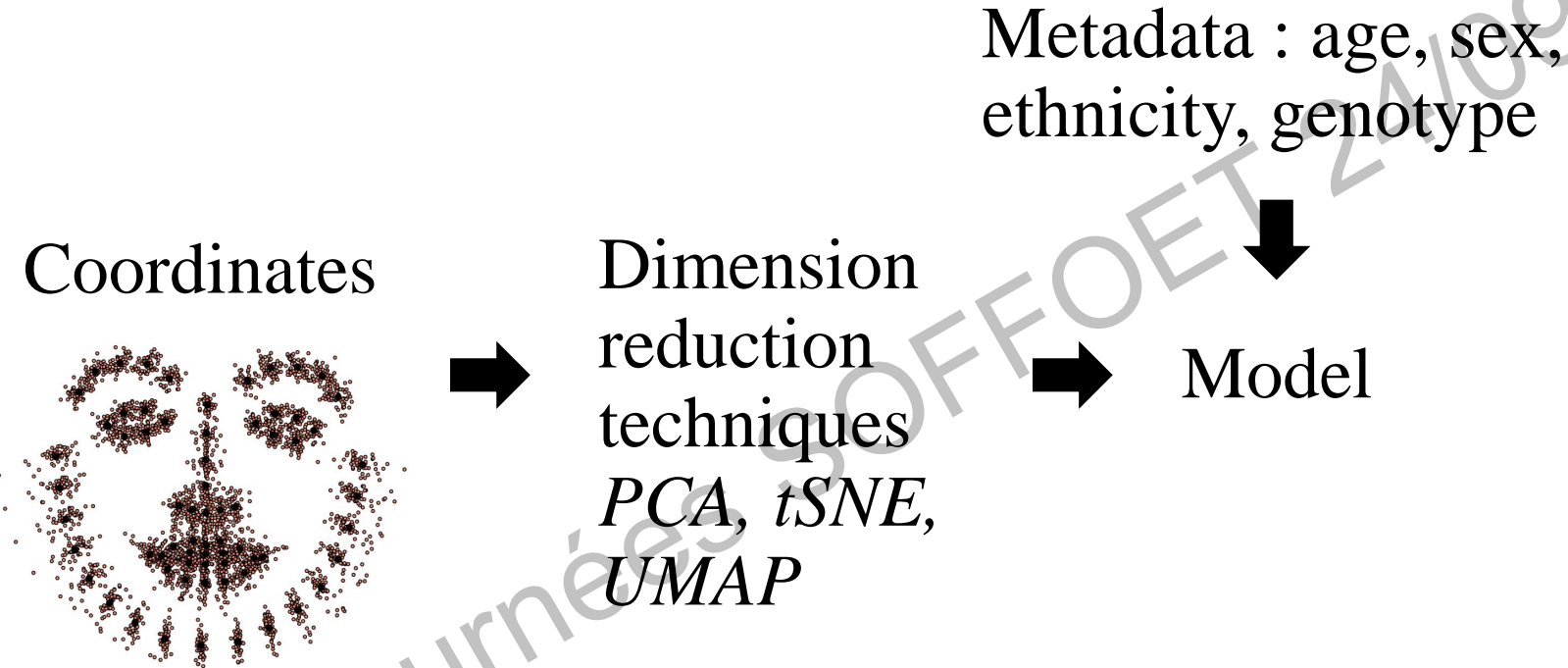
Coordinates



Dimension
reduction
techniques
PCA, tSNE,
UMAP

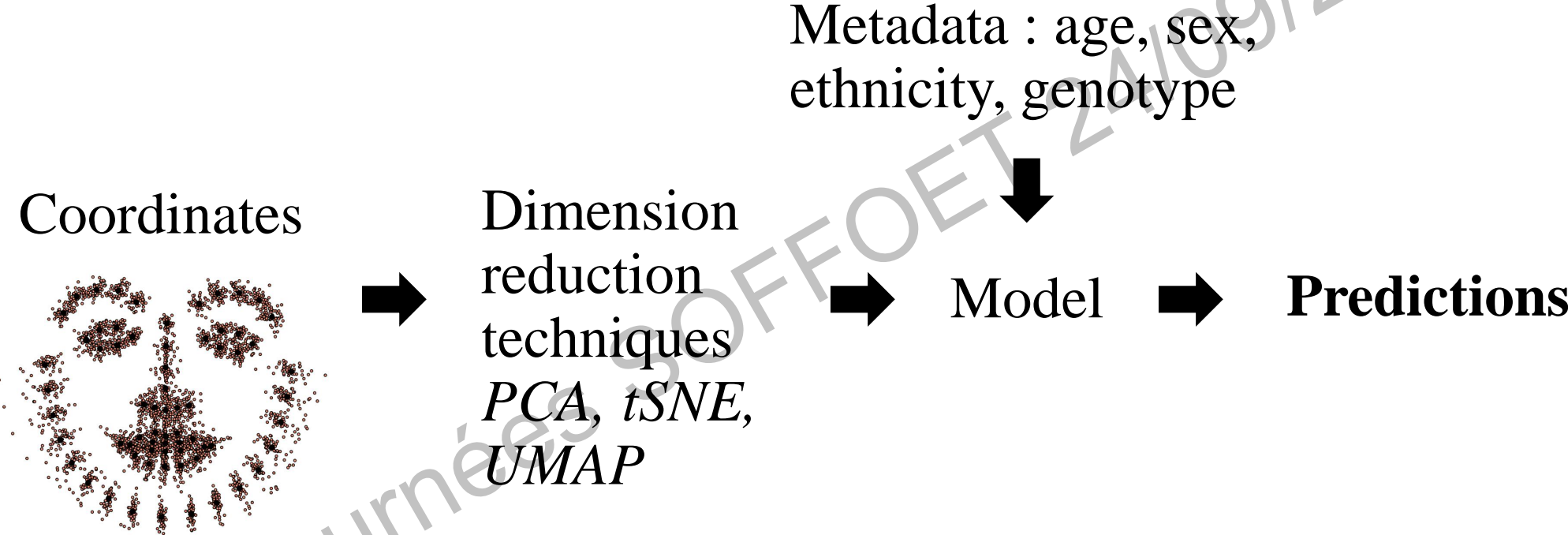
Journées OFFOET 24/09/2021

Machine learning



Journées OFFOET 24/09/2021

Machine learning





Journées OFFICIELLES 4/09/2021



Metadata :

3 y.o

Girl

Caucasian

TCOF1



Metadata :

3 y.o

Girl

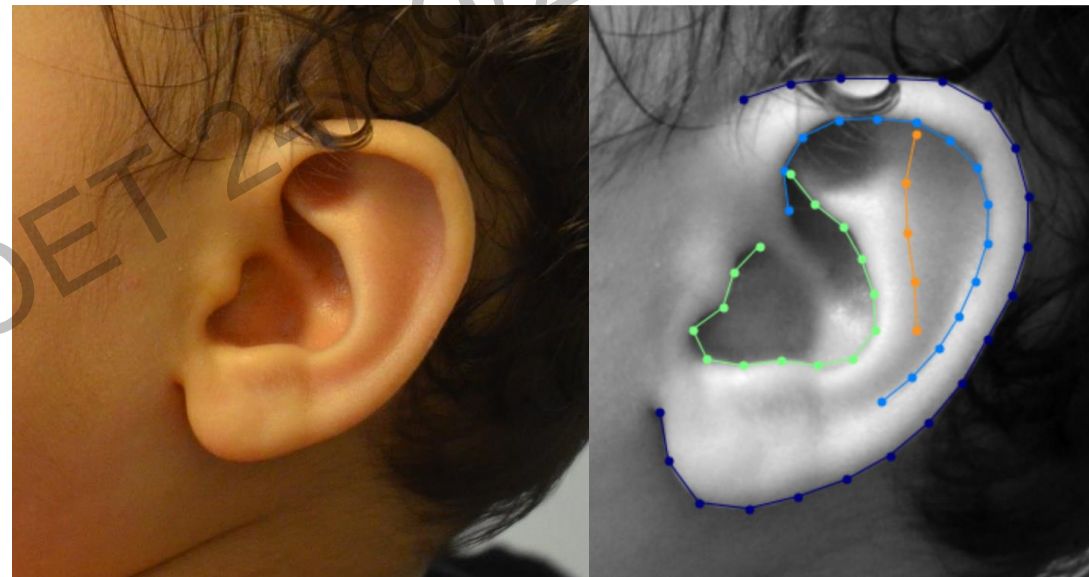
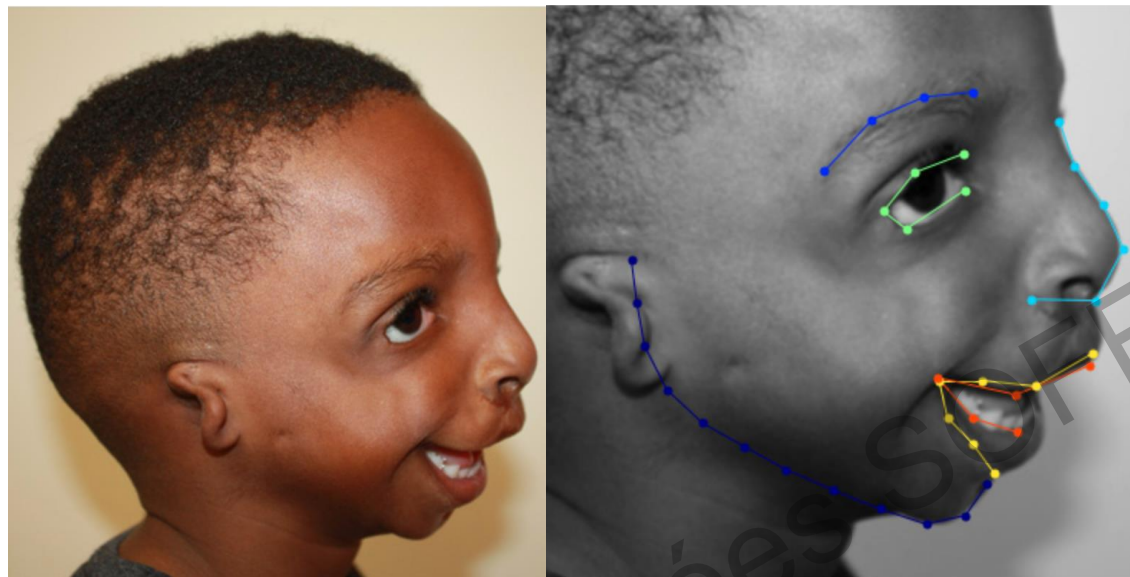
Caucasian

TCOF1



65 %

Profiles / Ears



Journées SFPOET 22-23/09/2021

Applications to foetopathology

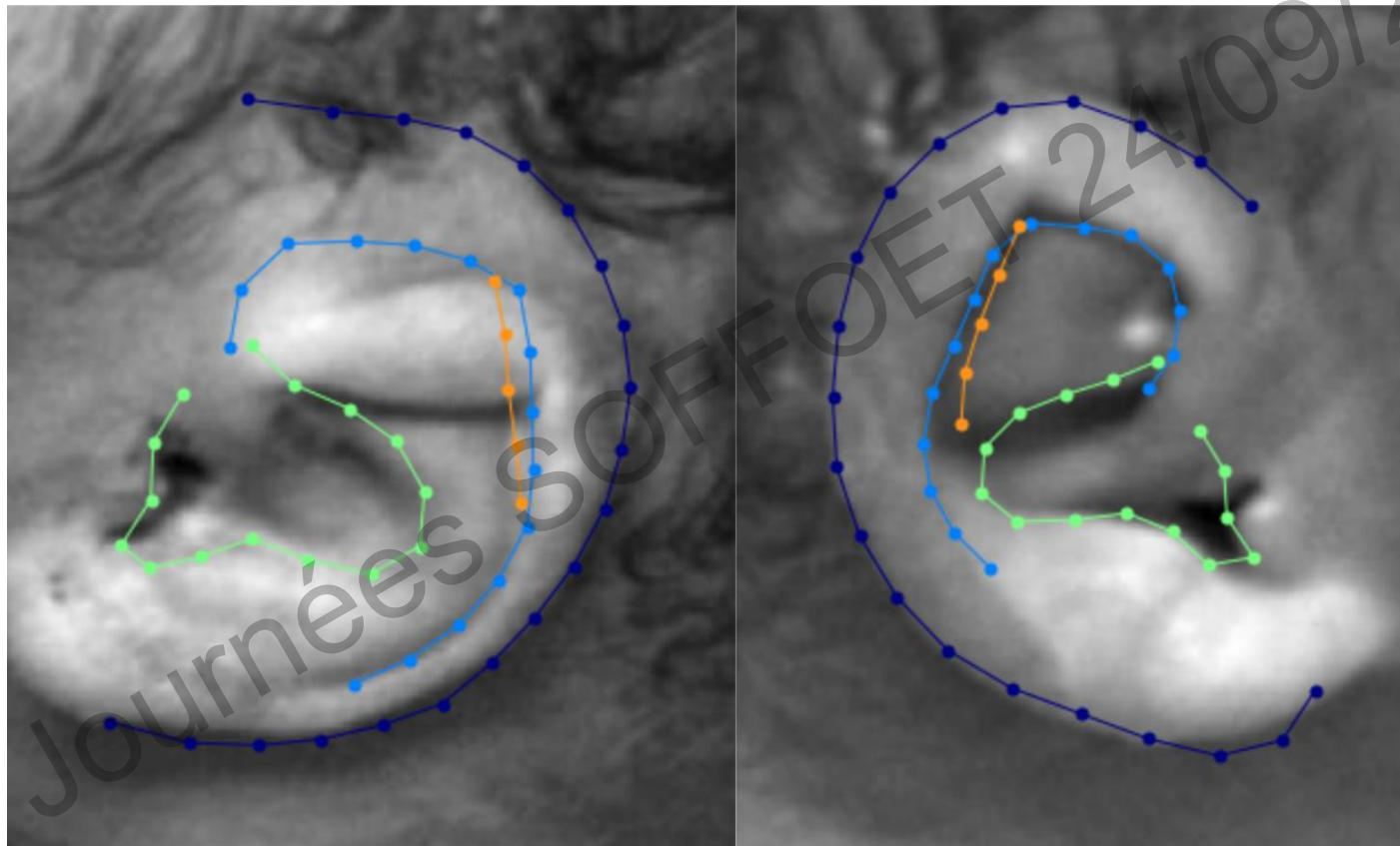


Foetopathology – Manual landmarking



Foetopathology – Automatic detection

Training = 180



Foetopathology – Applications

- 1) To **diagnose** syndromes
- 2) To **understand** what are the recognized phenotypic features to diagnose a particular syndrome
- 3) To create **growth models** for controls and for each syndrome as a function of term

Journées SOFFOET 24/09/2021

Thank you !

Journées SOFFOJET 24/09/2021

quentin.hennocq@aphp.fr