Facial Genetics

Science Circle Panel

30 January 2021

Faces

- We talk about beauty being skin deep.
- The architecture of your face is underlain by structures composed of bone, cartilage and muscle.
- The dimensions and growth patterns of the facial tissues are genetically determined.

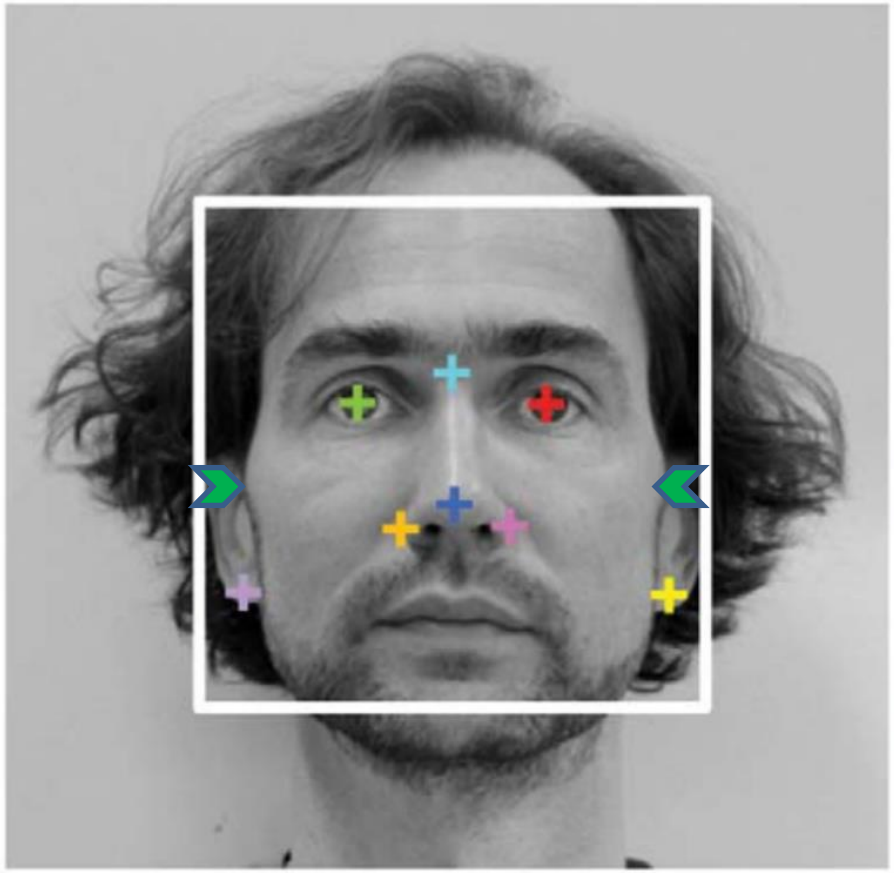
Image from
https://www.pinterest.com/pin/542824561328847735/

Identifying Facial Gene Markers: GWAS

- GWAS = Genome-Wide Association Studies
- DNA sequences of thousands of people studied.
- Polymorphisms identified as single nucleotide changes in a specific DNA tract.
- Genetic polymorphisms consistently associated with a given trait are identified.
- Each polymorphism is mapped and assigned an ID number, e.g. rs974448 in PAX3 (Paired box-3).
  Chromosome 2:222140595 A/G

First 5 Facial Genes 2012

Facial Reference Points



Candidate Genes

PRDM16

PAX3

TP63

C5orf50

COL17A1

Image from: doi:10.1371/journal.pgen.1002932 (2012)

First 5 Facial Genes 2012

SNPs associated with various facial regions

Gene	SNP	Chr	BP	Eff	Alt	FreqEff	Trait*
PRDM16	rs4648379	1p36.23-p33	3251376	T	C	0.28	AlrL-Prn
							AlrR-Prn
PAX3	rs974448	2q35	222713558	G	A	0.17	EyeR-Nsn
							EyeL-Nsn
TP63	rs17447439	3q28	191032117	G	A	0.04	EyeR-EyeL
C5orf50	rs6555969	5q35.1	171061069	T	C	0.33	ZygR-Nsn
							ZygL-Nsn
							EyeR-Nsn
							EyeL-Nsn
COL17A1	rs805722	10q24.3	105800390	T	C	0.19	EyeL-Nsn
							EyeR-Nsn

Eff = the allele whose action is being studied
Alt = the other (major) allele at the locus

Image from: doi:10.1371/journal.pgen.1002932 2012

Most Facial Genes encode Transcription Factors or Signalling Proteins

- Sonic hedgehog: Embryonic axis
- FGF: Fibroblast growth factors
- Homeobox genes: determine location of structures
- BMP: Bone morphogenetic proteins
- Pax3 (Paired-box 3): First associated with nervous system development on fruit flies. Pax3 is a transcription factor named for the “paired” DNA binding domain.

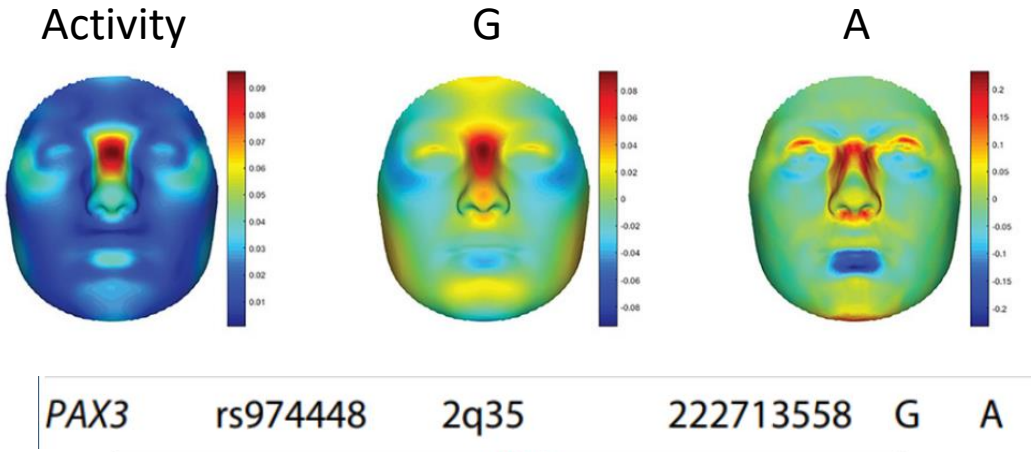
- >200 genes have been associated with facial features.

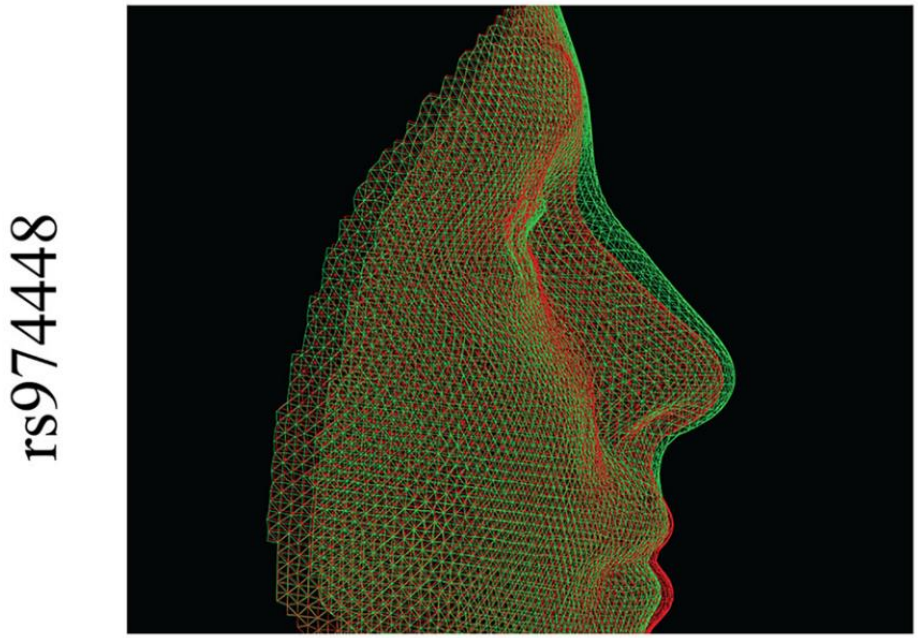
In faces, these genes control things like…

- Distance from eye to nose: PAX3
- Length of the nose: PRDM16
- Prominence of the nose: DCHS2
- Shape of the nose tip: SOX9
- Width of the nasal alae: SOX9
- Naso-labial angle: SUPT3H
- Eye width: C5orf50
- Length of the nose: PRDM16
- Distance from ear to nose: C5orf50
- Chin shape: ASPM

Several genes have effects on multiple facial traits

Pax3 Polymorphism rs974448



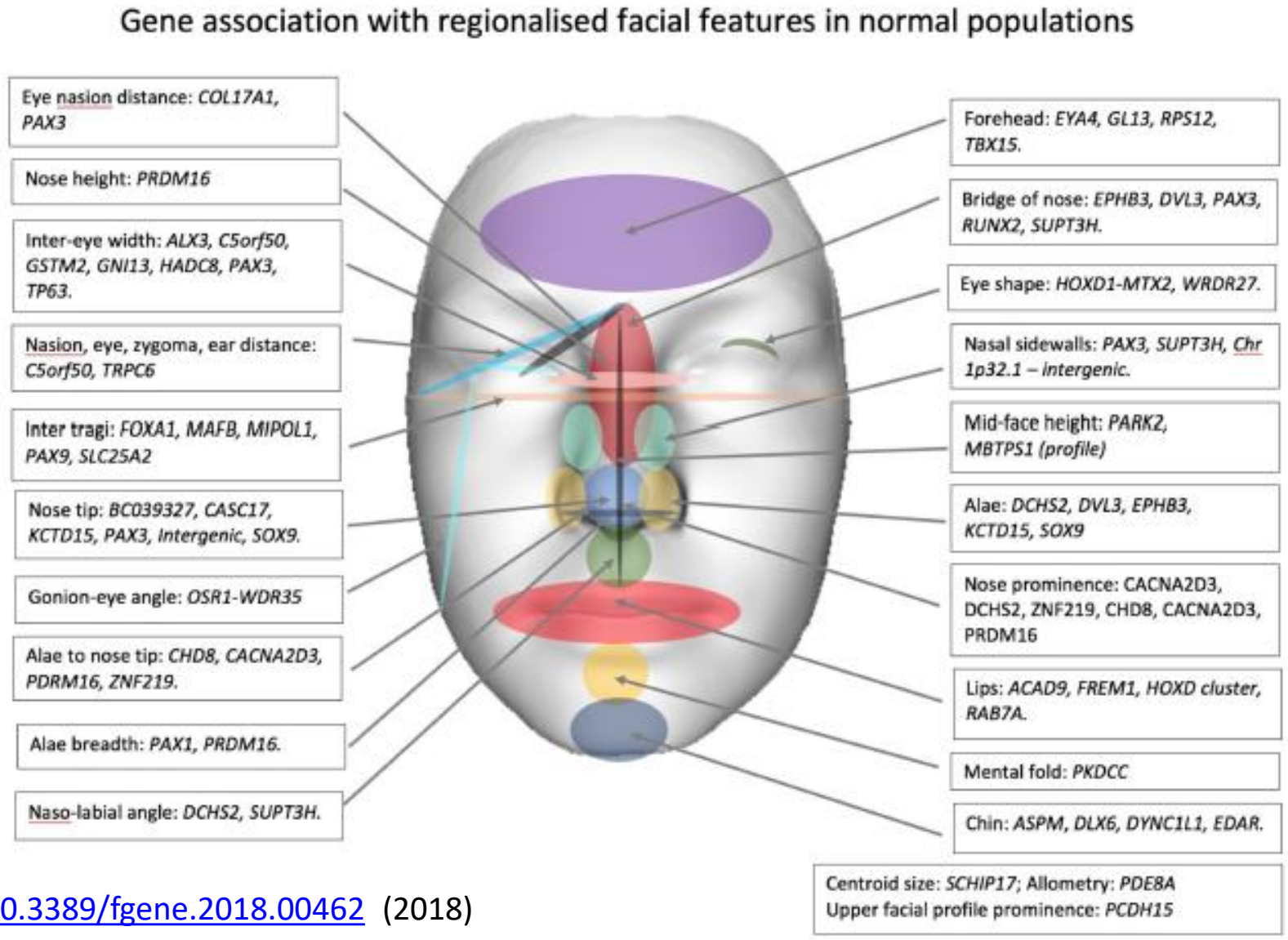


The variant effects are focused on the nose, especially nose ridge and bridge elevation. Polymorphism rs974448 also influences position of the nose relative to the eyes and lower orbits.

Wireframe represents the shape of the face with the two alleles: red = A, green = G.

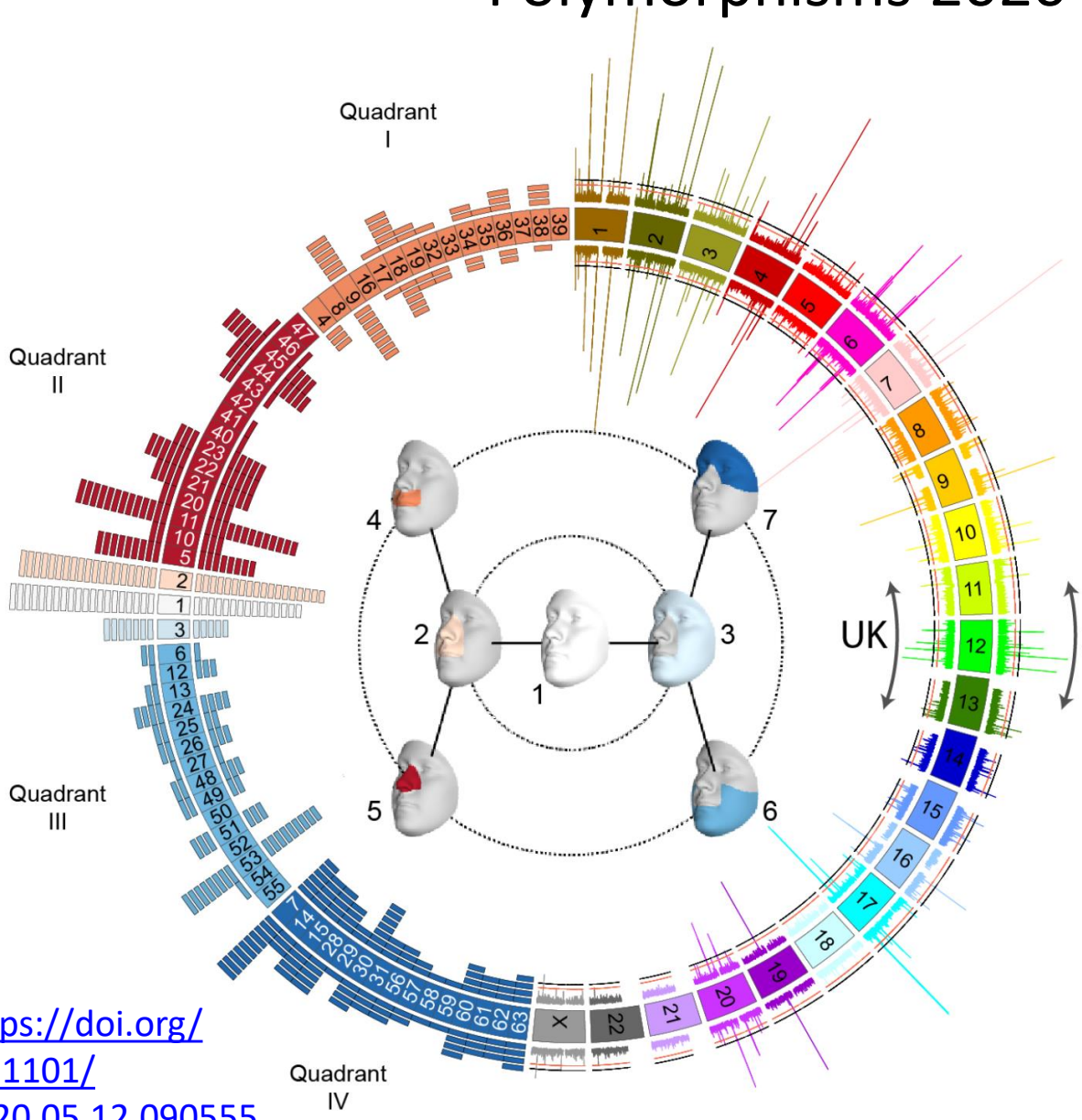
doi:10.1371/journal.pgen.1006250 (2016)

Genes associated with different facial features



doi: 10.3389/fgene.2018.00462 (2018)

Polymorphisms 2020



2020 study with >8000 US and UK participants.

Center: facial regions

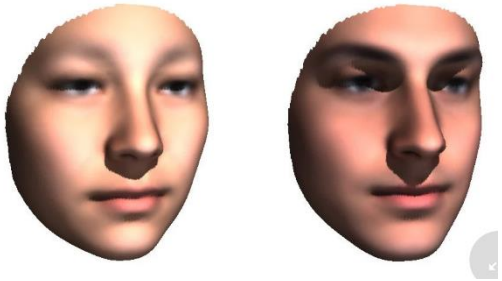
Left: SNP affecting each region
Blocks represent variability in US (outer) and UK (inner)

Right: distribution of SNPs along 23 chromosomes.
Peaks represent regions related to facial differences.

https://doi.org/10.1101/2020.05.12.090555

Can individuals be identified from images produced from DNA?

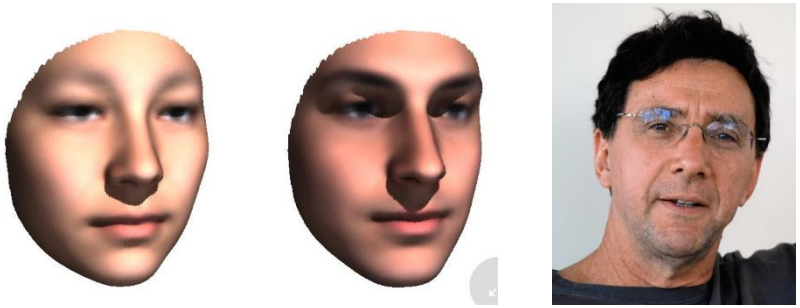
In 2015, two employees of the New York Times had their DNA sequenced and the sequences sent to Mark Shriver (Penn State), who has developed software for producing images from polymorphism information.



Images were produced for both individuals and given to their colleagues, who were asked to identified them. 50 colleagues responded.

Can individuals be identified from images produced from DNA?

In 2015, two employees of the New York Times had their DNA sequences and the sequences sent to Mark Shriver (Penn State), who has developed software for producing images from polymorphism information.



Images were produced for both individuals and given to their colleagues, who were asked to identified them. 50 colleagues responded.

The man was not correctly identified by any of his colleagues.

Can individuals be identified from images produced from DNA?

In 2015, two employees of the New York Times had their DNA sequences and the sequences sent to Mark Shriver (Penn State), who has developed software for producing images from polymorphism information.



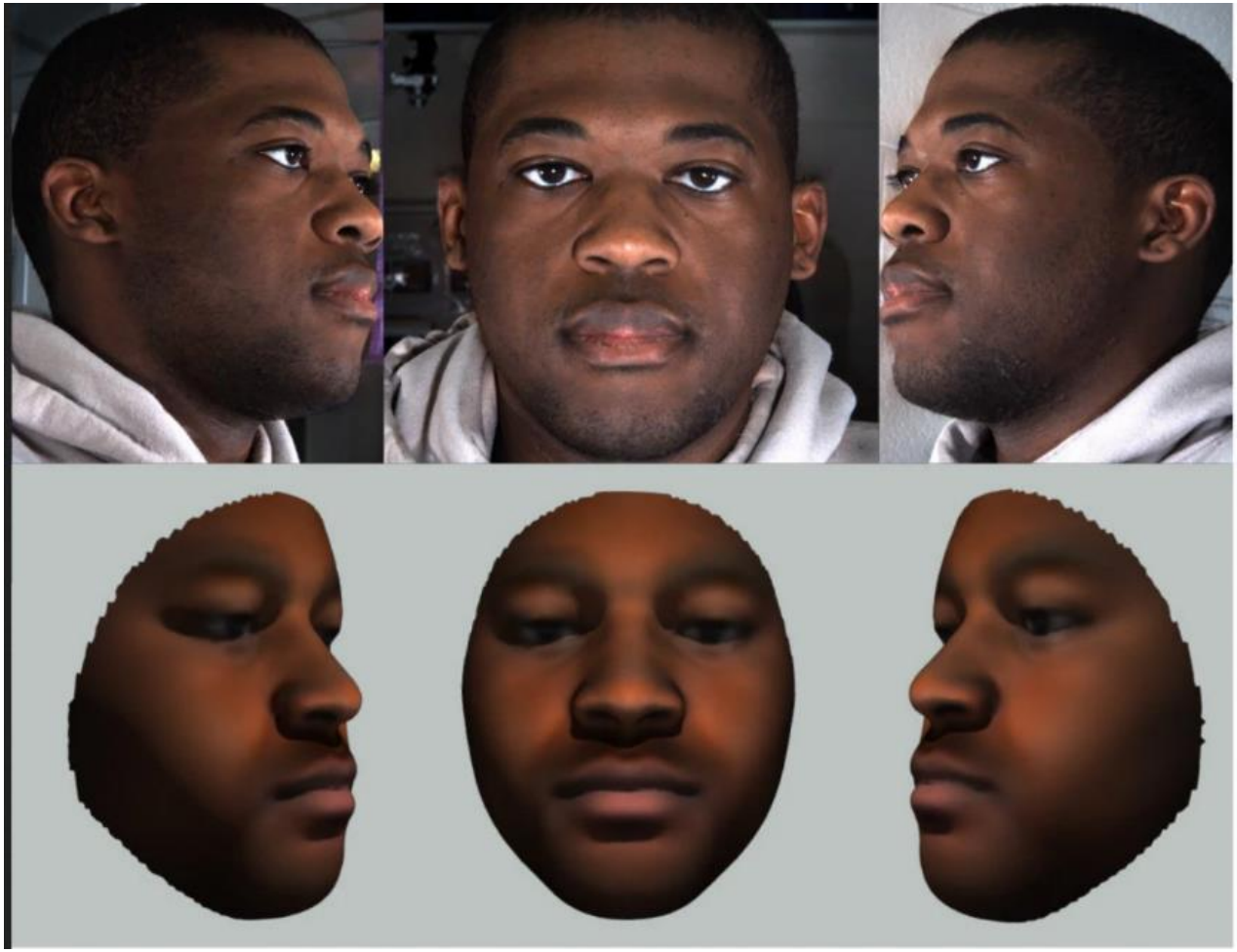

Images were produced for both individuals and given to their colleagues, who were asked to identified them. 50 colleagues responded.

The man was not correctly identified by any of his colleagues.

The woman was correctly identified by about 1/3 of her colleagues.

https://www.nytimes.com/2015/02/24/science/dna-generated-faces.html

Other examples of DNA profiles vs their subjects

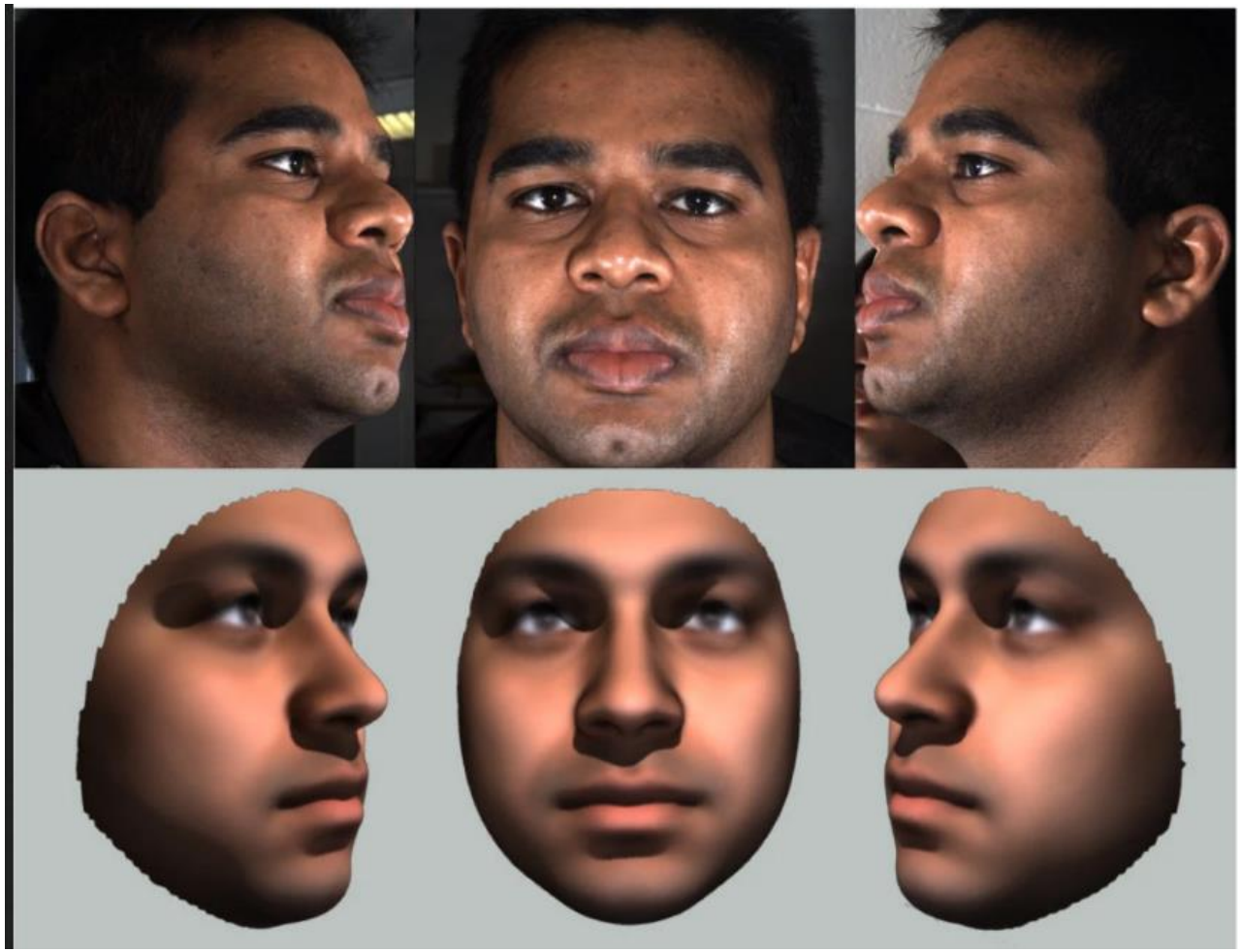


https://www.nytimes.com/2015/02/24/science/dna-generated-faces.html

Other examples of DNA profiles vs their subjects

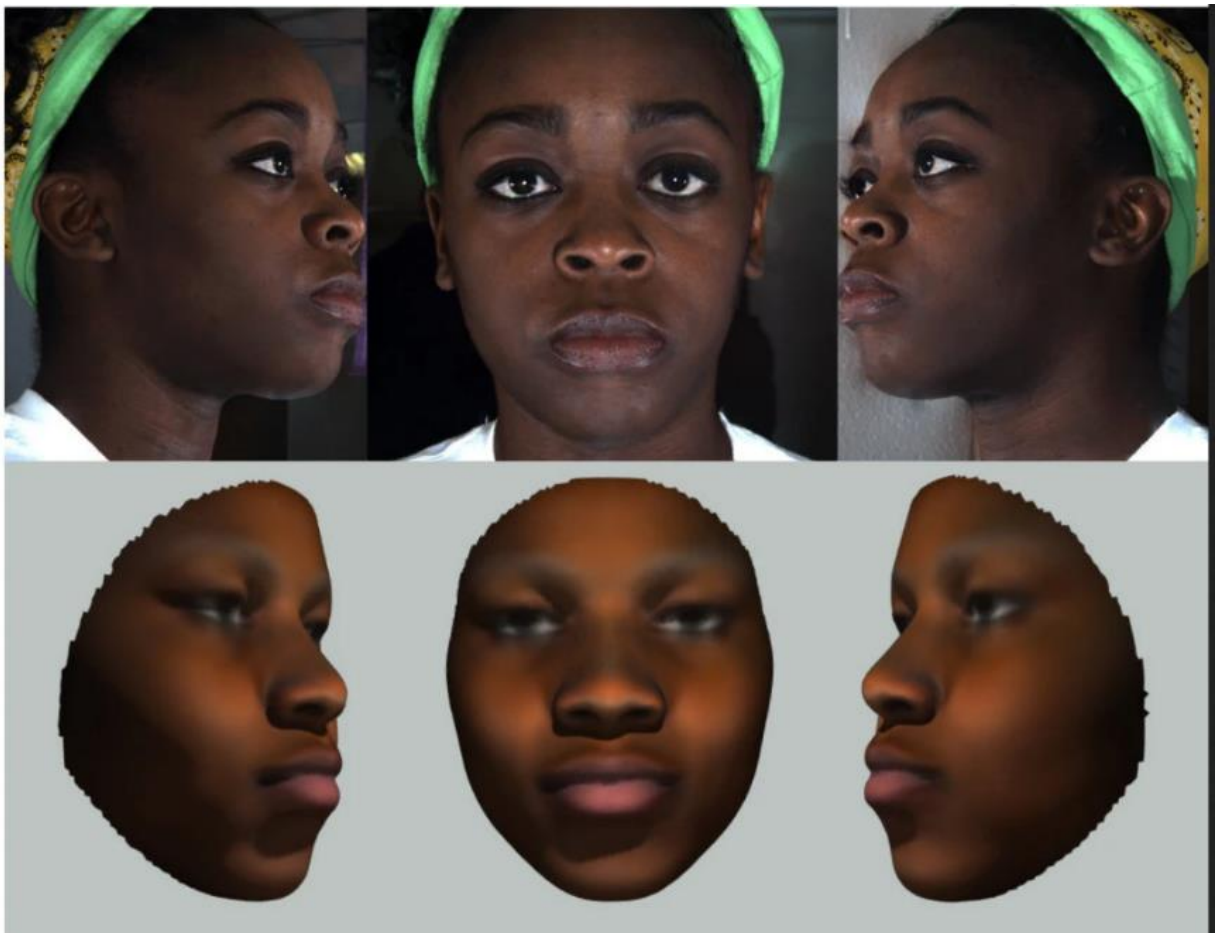
https://www.nytimes.com/2015/02/24/science/dna-generated-faces.html

Other examples of DNA profiles vs their subjects



https://www.nytimes.com/2015/02/24/science/dna-generated-faces.html

Other examples of DNA profiles vs their subjects



https://www.nytimes.com/2015/02/24/science/dna-generated-faces.html

References

- Claes P, Liberton DK, Daniels K, Rosana KM, Quillen EE, et al. (2014) Modeling 3D Facial Shape from DNA. PLoS Genet 10(3): e1004224. doi:10.1371/journal.pgen.1004224
- Claes P, Shriver MD (2016) New Entries in the Lottery of Facial GWAS Discovery. PLoS Genet 12(8): e1006250. doi:10.1371/journal.pgen.1006250
- Liu F, van der Lijn F, Schurmann C, Zhu G, Chakravarty MM, et al. (2012) A Genome-Wide Association Study Identifies Five Loci Influencing Facial Morphology in Europeans. PLoS Genet 8(9): e1002932. doi:10.1371/journal.pgen.1002932
- Richmond S, Howe LJ, Lewis S, Stergiakouli E and Zhurov A (2018) Facial Genetics: A Brief Overview. Front. Genet. 9:462. doi: 10.3389/fgene.2018.00462
- Stergiakouli E and Zhurov A (2018) Facial Genetics: A Brief Overview. Front. Genet. 9:462. doi: 10.3389/fgene.2018.00462
- https://www.nytimes.com/2015/02/24/science/dna-generated-faces.html
- https://www.nytimes.com/2019/12/03/business/china-dna-uighurs-xinjiang.html