

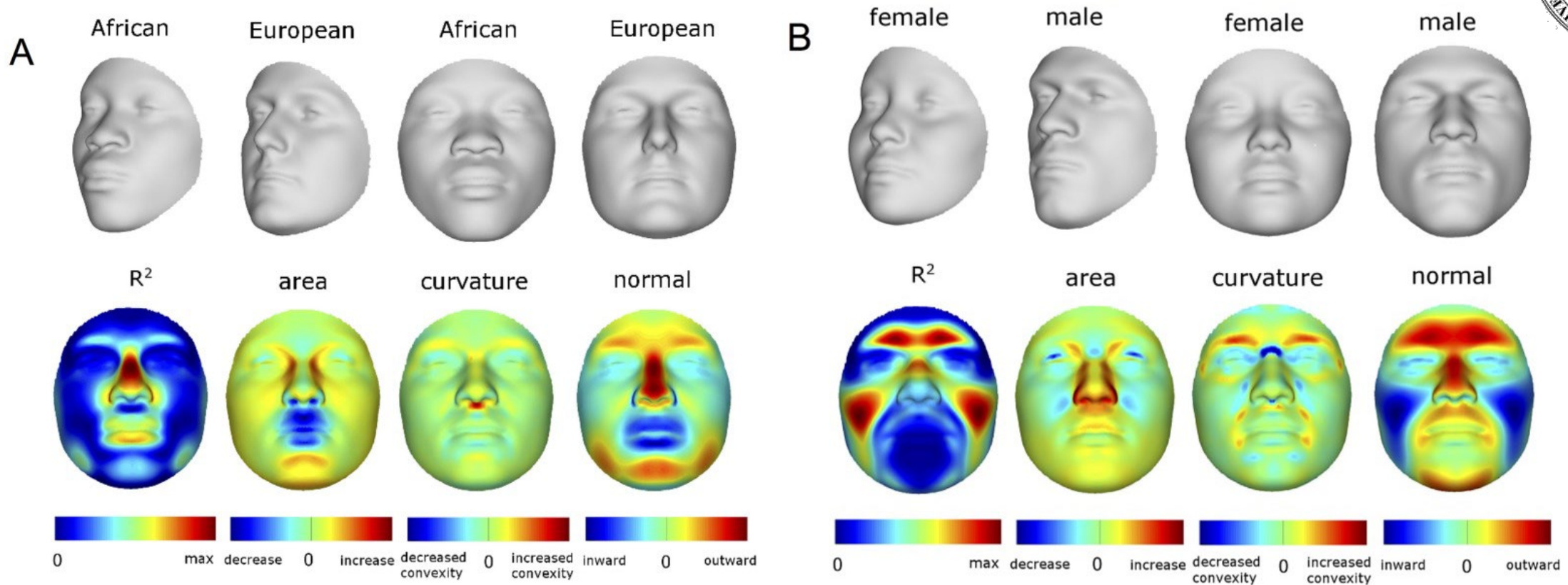
Három perc alatt 3D arcrekonstrukció a génállomány alapján

2021.11.02.

- Az arc morfológiai sajátosságainak öröklődő háttere
- Jellegzetes arc eltérés → célzott génvizsgálat
- Arcunk DNS-ünk tükré?
- Mesterséges intelligencia (Face2Gene) platform
- Igazságügyi szakértők: DNS-alapú arcfelismerés

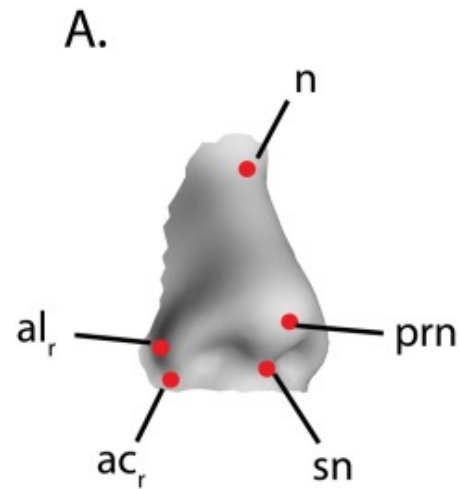


ALAPÍTVÁNY



SKI, LMNA, SIL1, EDN1, RSPO2, TRPS1, POLR1D, MAP2K1, ADAMTS10, TBX1, PEX14, HSPG2, CAV3, CTNND2, TFAP2A, PEX6, PEX3, MEOX2, RELN, ROR2, NEBL, CHUK, FGFR2, WT1, PEX16, BMP4, FANCA, RAI1, FOXA2, ECE1, DPYD, ZEB2, SATB2, FGFR3, NIPBL, NSD1, ENPP1, GLI3, COL1A2, BRAF, ASPH, FREM2, SNRPN, FBN1, MAP2K2, RPS19, DNMT3B, GDF5, és UFD1L

AZ ORR-MORFOLÓGIA GENETIKAI HÁTTERE

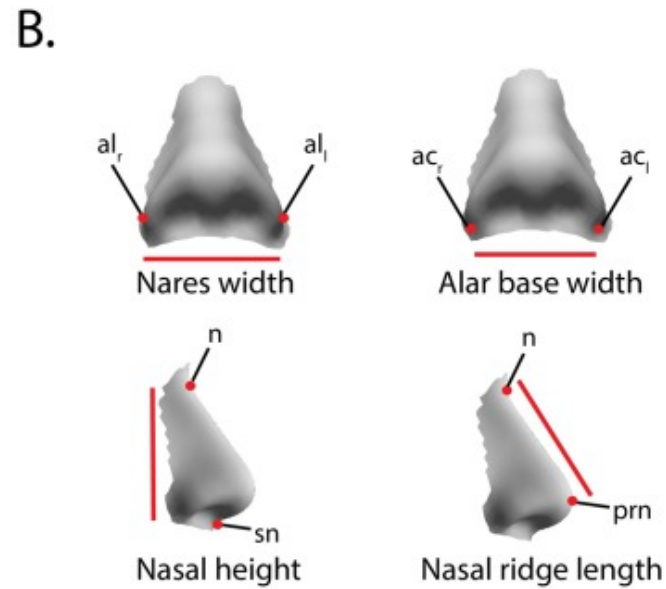


Midline Landmarks:

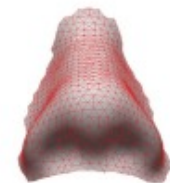
1. Nasion (n)
2. Pronasale (prn)
3. Subnasale (sn)

Paired Landmarks:

1. Alare (al)
2. Alar curvature (ac)



Distances



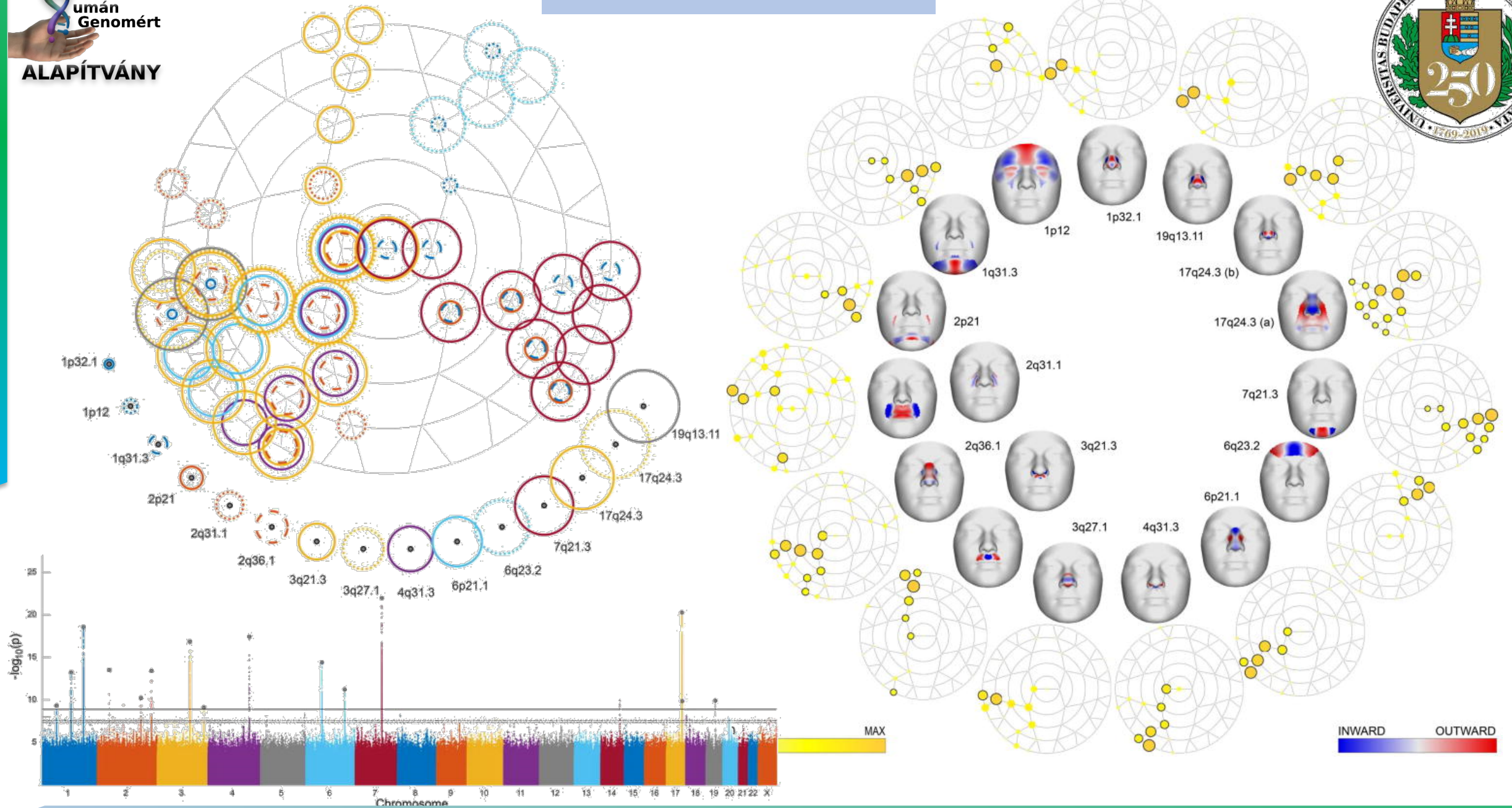
Overall area



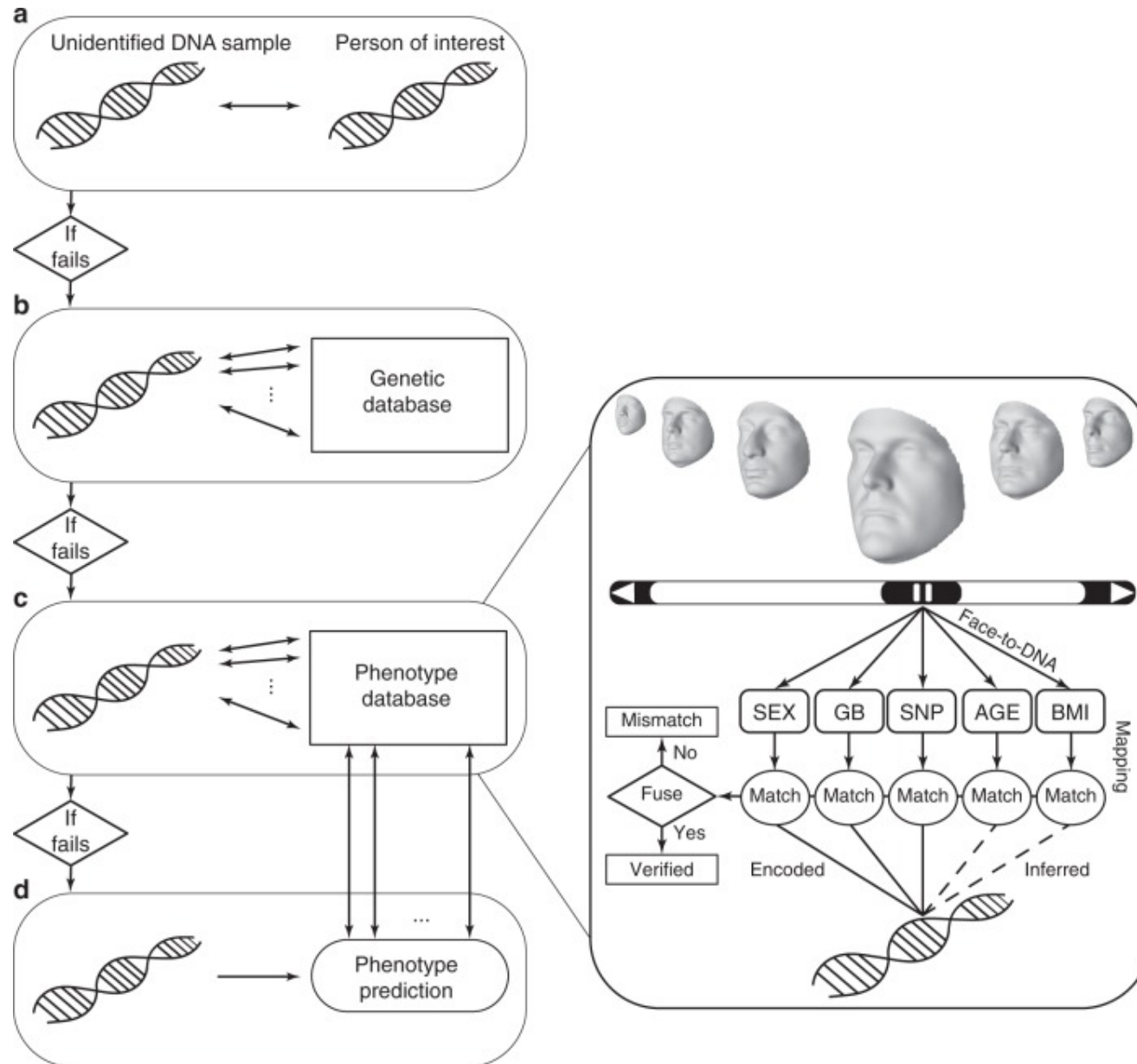
Nostril area

Areas

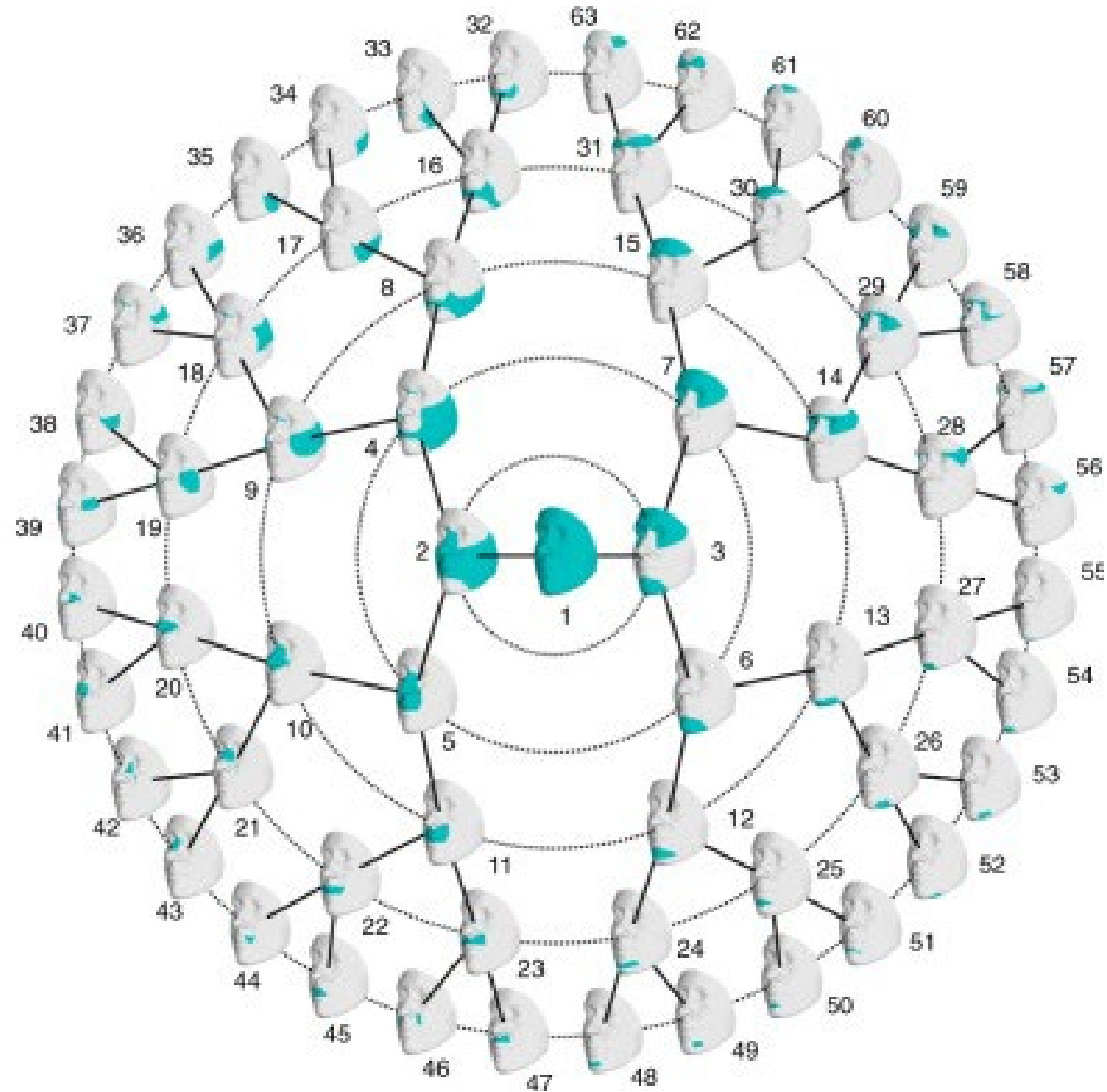
GÉNSZAKASZ VARIÁNS-HATÁS



Claes, Peter et al. "Genome-wide mapping of global-to-local genetic effects on human facial shape." *Nature genetics* vol. 50,3 (2018):



ARCUNK GENETIKAI SZEGMENTUMAI

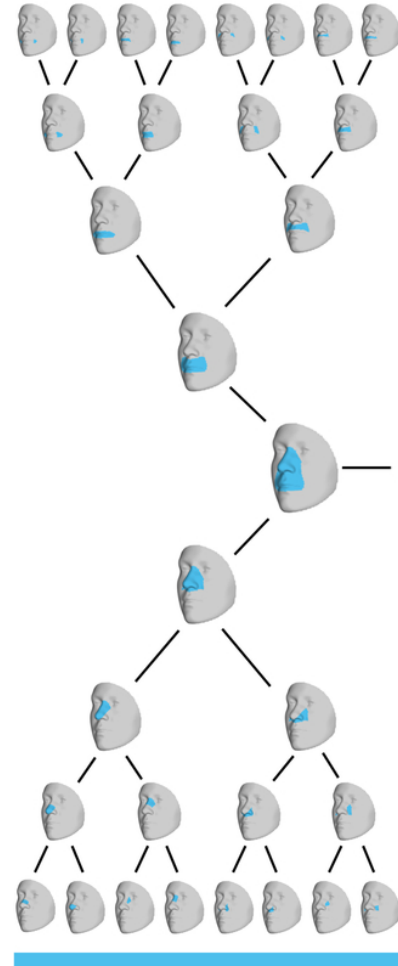


ARCUNK GENETIKAI KVADRÁNSAI



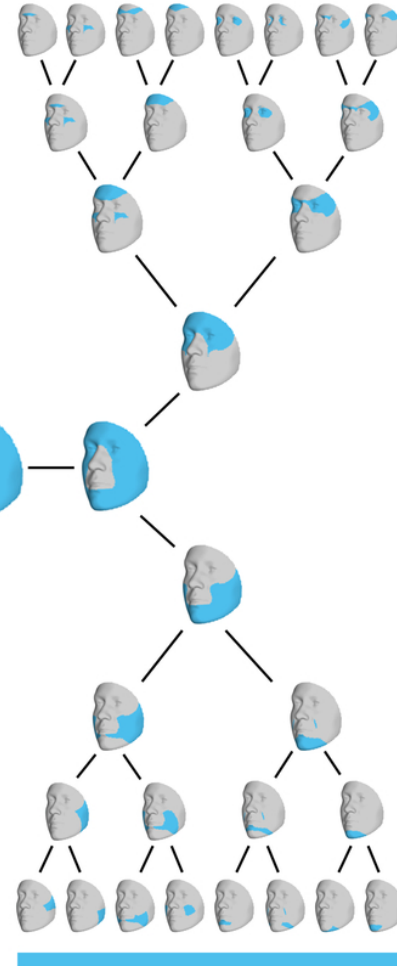
HOXD; RAB7A/ACAD9

Lip Modules



TBX15;RPS12/EYA4

Upper Face and Orbit Modules

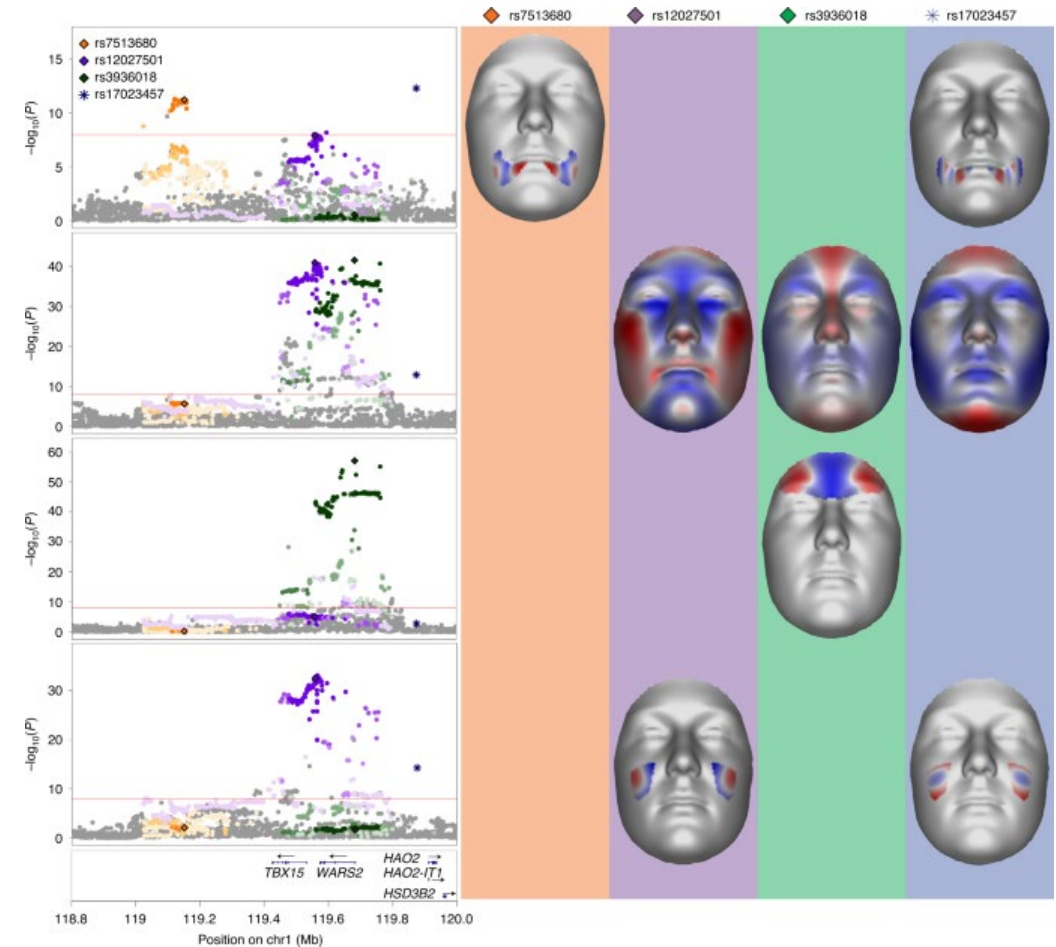
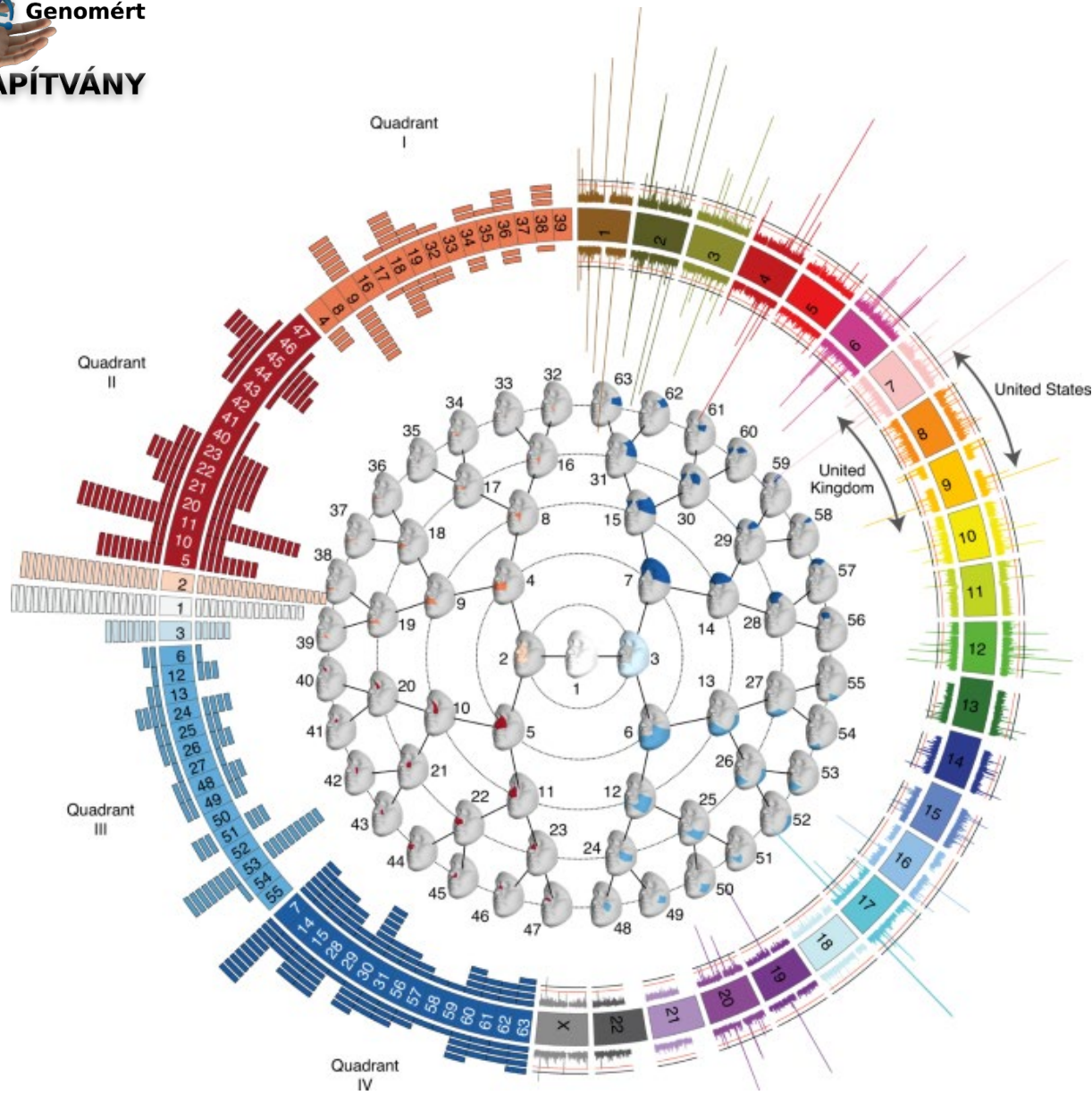


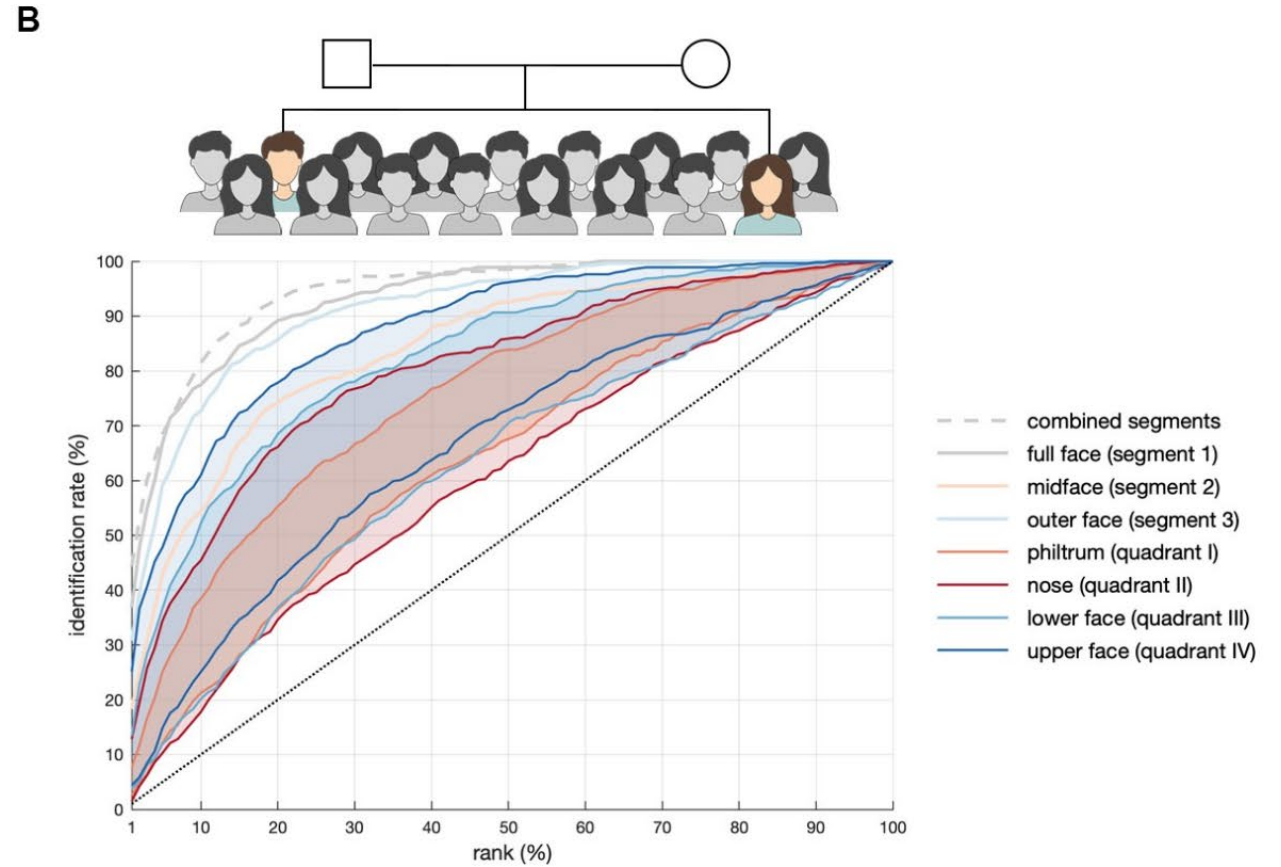
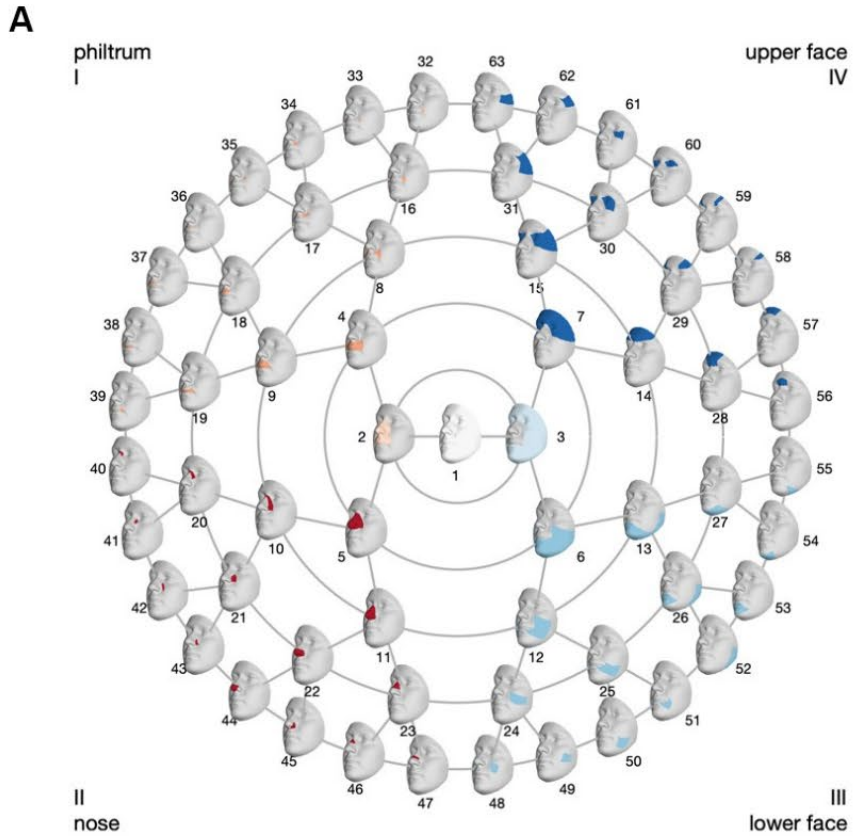
Nose Modules

1p32.1; HOXD; PAX3; RAB7A/ACAD9; EPHB3/DVL3; DCHS2 SUPT3H; BC039327; SOX9; KCTD15; CDH18; PAX1**

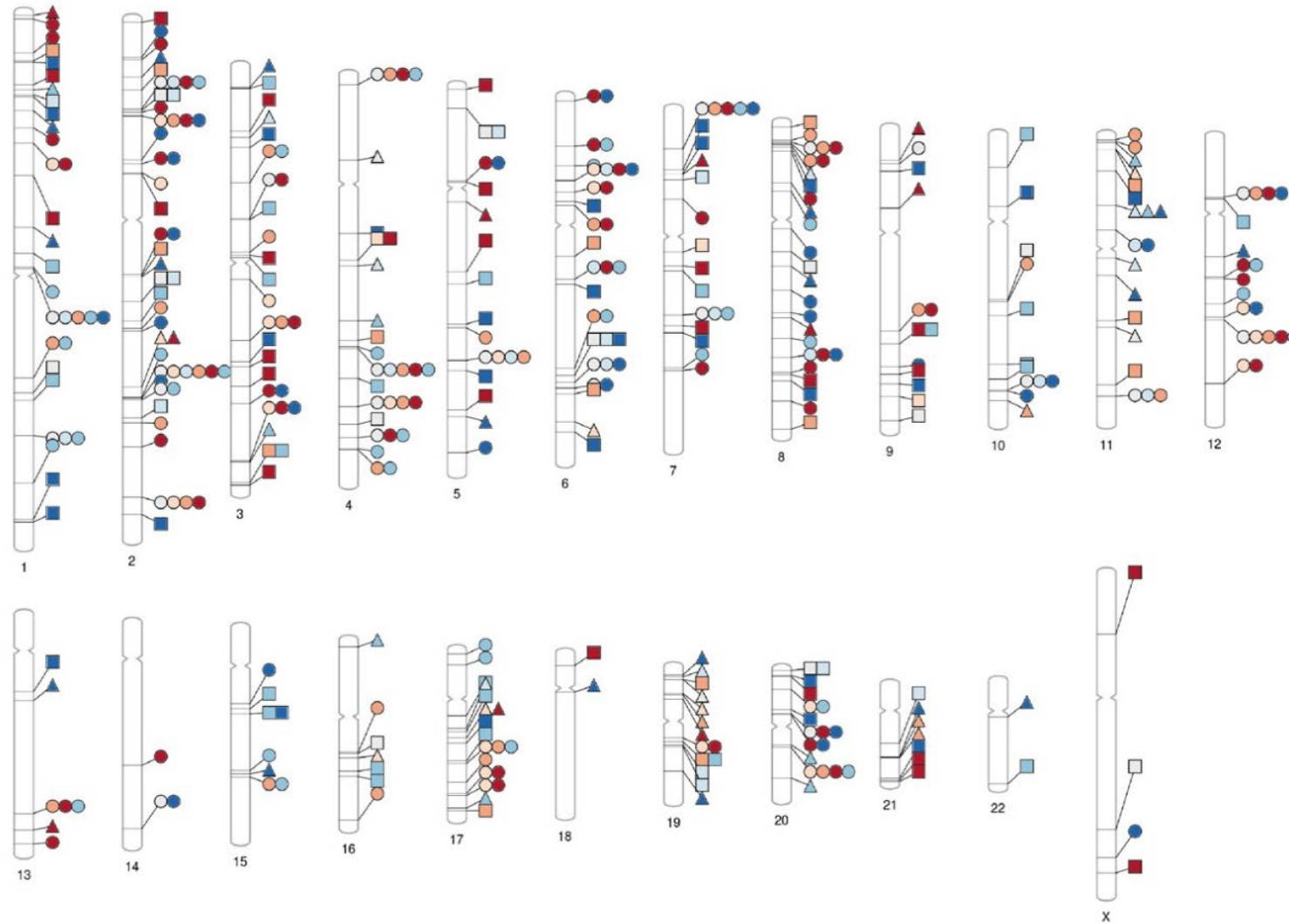
Mandible Modules

*ASPM; PKDCC; DLX6; NHP2/ZNF354a**



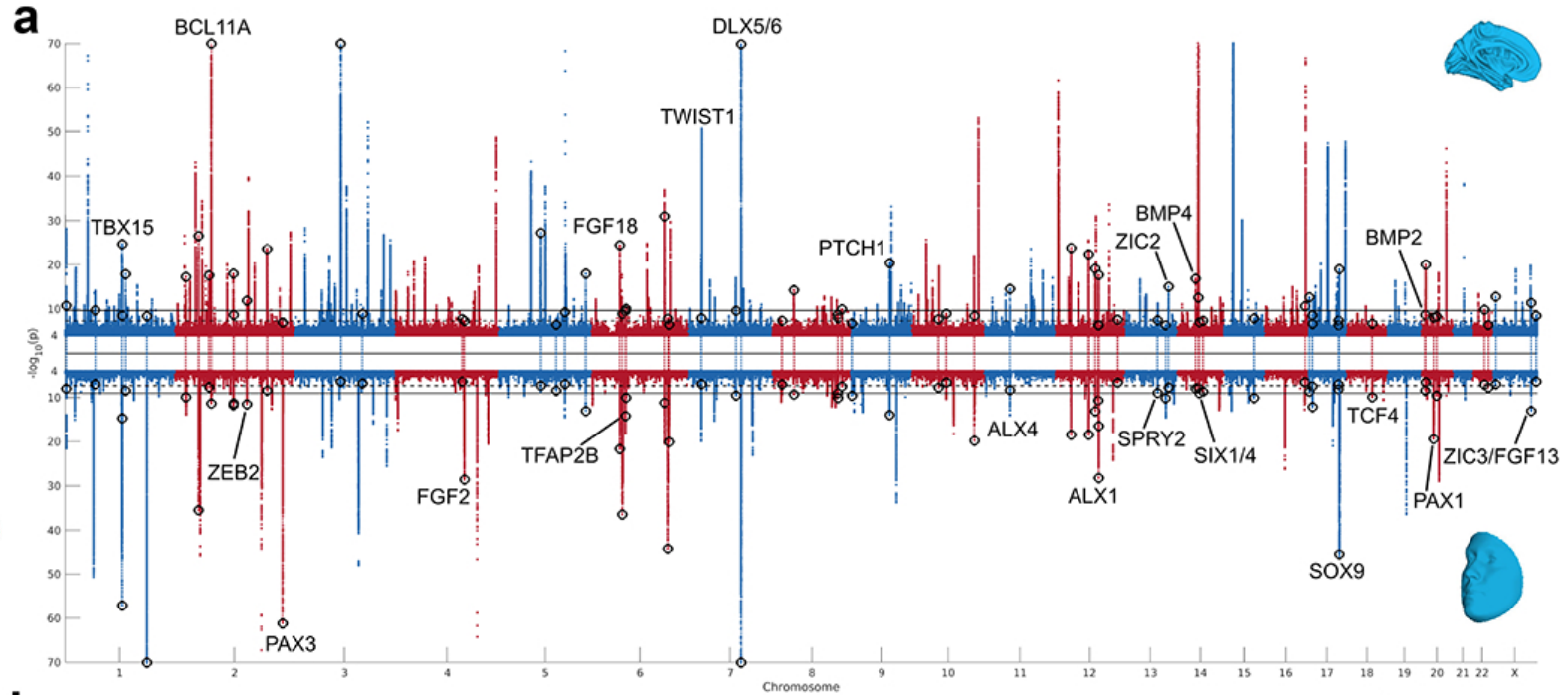
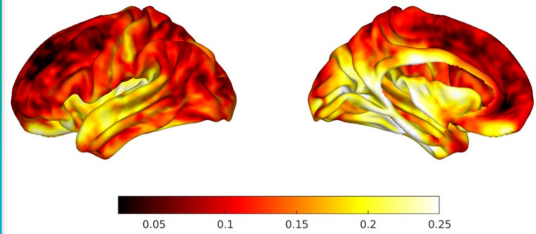


203 GENETIKAI LÓKUSZ, MINT ARCMORFOLÓGIAI MEGHATÁROZÓ



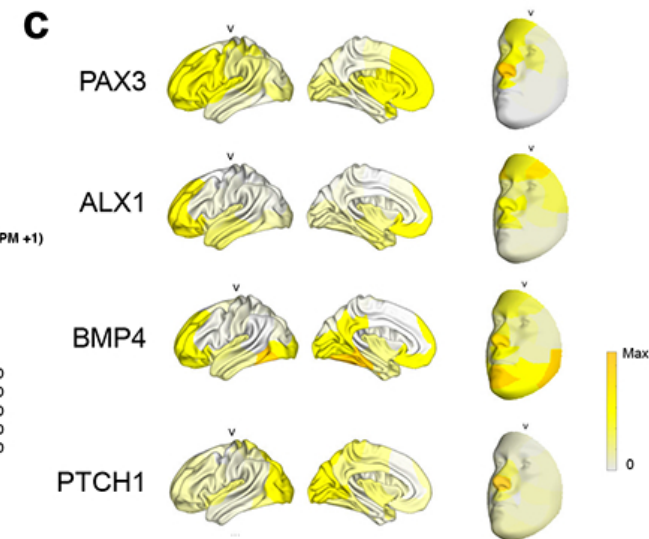
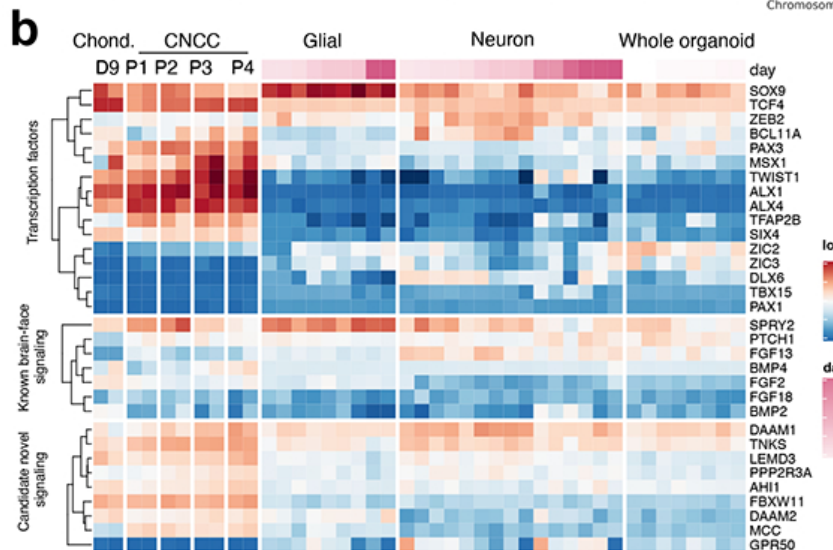
- | | | |
|-------------------------|-----------------------|----------------------------------|
| ○ GWAS (overlap) | □ GWAS (novel) | △ low-frequency variant analysis |
| □ full face (segment 1) | □ midface (segment 2) | □ outer face (segment 3) |
| ■ philtrum (quadrant I) | ■ nose (quadrant II) | ■ lower face (quadrant III) |
| | | ■ upper face (quadrant IV) |

ARC ÉS AGY SZERKEZETÉNEK GENETIKAI ÁTFEDÉSE



HIPOTÉZIS:

DeMyer W, **THE FACE PREDICTS THE BRAIN: DIAGNOSTIC SIGNIFICANCE OF MEDIAN FACIAL ANOMALIES FOR HOLOPROSENCEPHALY (ARHINENCEPHALY).** *Pediatrics* 34, 256 LP-263 (1964)



KÖSZÖNÖM SZÉPEN A FIGYELMET!

