

THE HONG KONG UNIVERSITY OF SCIENCE & TECHNOLOGY

Department of Mathematics

PHD STUDENT SEMINAR

Identification of genetic signatures of human face by AI-Driven facial phenotyping

By

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<u>Abstract</u>

Traditional human face genome-wide association studies (GWASs) are limited by small sample size and inefficient landmarks-based phenotyping methods. By combining the state-ofthe-art deep face recognition (FR) network and training dataset contains millions of face images, we proposed an artificial intelligence (AI)-driven facial phenotyping approach for 2D face images, allowing for a robust and human-like perception of interpersonal facial variation in a low-dimensional latent space. In a sample of 8,016 research participants with self-reported face images from China direct-to-customer (DTC) genetic testing service company, we identified 25 independent loci associated with 128-dimensional AI-driven facial measurements, 13 (6 previously unreported) of which replicated in an independent 5,858 Chinese research participants. Two replicated loci show strong evidence of enrichment for active chromatin elements in human cranial neural crest cells, suggesting they involve in the early developmental origin of the facial variation. A few features of 128-dimensional facial measurements were identified to be genetically correlated with BMI and male-pattern baldness, partially interpreted the implications of black-box generated facial phenotypes. These results show that the integration of large DTC biobanks and AI-driven approach now promise to greatly improve the statistical power in GWAS research of complex and quantitative traits.

Date : 13 May 2020 (Wednesday) Time : 4:00pm – 5:00pm Zoom Meeting : <u>https://hkust.zoom.us/j/9656130237</u>

All are Welcome!