

PRIMATE EVOLUTION AND HUMAN DISEASES

The Premise is that evolution is the study of the past to understand the present. Nevertheless, more often than not, it is merely a matter of curiosity and lacks practical significance. This post shows that the study of evolution occasionally also has relevant practical implications.

Recently, Science published two papers on the analysis of 233 primate genomes (Kuderna et al.¹; Gao et al.²).

Evan Eichler's comment³ stresses the importance of these two papers for the interpretation of human variants in protein-coding genes with respect to human diseases. This is because of the relatively low degree of genetic diversity within our species as compared to the diversity in primates. Indeed, Gao et al.² created a primate population variant database of 4.3 million common missense variants that is 50 times larger than the clinical variant database (ClinVar) generated from human data. From this primate database they derived a semi-supervised 3D convolutional neural network, PrimateAI-3D, which outperforms 15 machine-learning classifiers bases on human variation only.

The last sentence by Eichler is thought provoking: "There is an irony in that the genetic information present in nonhuman primates—facing extinction due to human interference—may improve our understanding of our own species and the health of our children".

1- <https://www.science.org/doi/10.1126/science.abn7829>

2- <https://www.science.org/doi/10.1126/science.abn8197>

3- <https://www.sciencedirect.com/science/article/pii/S2666979X23001416?via%3Dihub>