S8.O.1

Gene regulatory architectures dissect the evolutionary dynamics of regulatory elements in humans and non-human primates

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Genes undergoing substantial evolutionary shifts in their expression profiles are often modulated by critical epigenomic changes that are among the primary targets of selection in evolution. Here, we investigate the evolution of epigenetic regulatory activities and their interplay with gene expression in human and non-human primate lineages. We extensively profiled a new panel of human and non-human primate lymphoblastoid cell lines using a variety of NGS techniques and integrated genome-wide chromatin contact maps to define gene regulatory architectures. We observe that epigenetic and sequence conservation are coupled in regulatory elements and reflect the impact of their activity on gene expression. The addition or removal of strong and poised promoters and intragenic enhancers is frequent in gene expression changes during recent primate evolution. In contrast, novel human-specific weak intragenic enhancers, dormant in our cell lines, have emerged in genes showing signals of recent adaptive selection, suggesting that they echo important regulatory innovations in other cell types. Among the genes targeted by these regulatory innovations, we find key candidate drivers of recently evolved human traits, such as FOXP2 or ROBO1 for speech and language acquisition, and PALMD for neocortex expansion, thus highlighting the importance of regulatory changes in human evolution.

S8.O.2

Deciphering a biological adaptation in the Andamanese people

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