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THE GENETIC BASIS FOR PRIMATE EVOLUTION AT *CIS*-REGULATORY SYSTEM LEVEL

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Understanding human and other primate phenotypic evolution from genetic point of view is tempting, however only little is known about its mechanism so far. Good candidates as responsible genetic elements facilitating body plan alterations are developmental controlling genes such as transcription factors and signal transduction molecules known as “toolkit genes”. But recent comparative genomics suggested that the toolkit genes likely to be largely similar in every mammalian species. Therefore cis-element changes that altered gene expression might be more plausible candidates accounting for the evolution of lineage specific morphological traits. In 1975 Mary-Claire King and Allan Wilson already recognized that gene regulatory mechanism in human-primate evolution is important, and they argued that regulatory mutations might account for the genetic basis of the difference between human and other great apes, rather than macromolecule (protein coding gene) mutations. It is becoming realistic to test the regulatory mutation hypothesis by studying functional aspect of evolution of the cis-regulatory mechanism in mammals and primates, due to both accumulation of genome data and advance in transgenic mouse technology. To show recent technologies in cis-element evaluation, I will introduce our studies with regard to function and evolution of cis-regulatory elements in the developmental toolkit genes, including Dlx and ParaHox homeobox genes in mammals. I will also discuss about the origin of novel function in cis-element evolution, especially about a possibility of gain-of-function type evolution in the cis-regulatory mechanism of toolkit genes by showing our recent data about retroposon exaptations and a human specific mutation in cis-element.

Keywords: cis-element, toolkit gene, transgenic mouse, homeobox