Open chromatin regions in primate genomes catch recent exogenous DNA insertions

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In the course of evolution, copies of mitochondrial DNA (mtDNA), called NUMTs (NUclear MiTochondrial DNAs), are known to insert into the nuclear genome via the repair of DNA double-strand breaks [1,2]. In our recent work, we newly found a strong correlation between NUMT insertion events and open chromatin regions in the human genome [3]. Interestingly the correlation between human NUMTs and open chromatin regions drops sharply when examining NUMTs older that the split between human and chimpanzee. This suggests that open chromatin regions where NUMTs insert has shifted dramatically in the human line during relatively short evolutionary time scales, and at the same time, this raises a question whether species-specific NUMT insertion sites tend to co-occur with species-specific (i.e. recently created) open chromatin regions.

Fortunately, DNase-seq open chromatin datasets in human, chimpanzee, and macaque have been published recently [4]. In response to this occasion, we analyzed 598 NUMTs in human and 600 NUMTs in chimpanzee to verify the correlation between species-specific open chromatin regions and NUMT integration sites appearing after the species diverged. In both the human and chimpanzee nuclear genomes, we observed that species-specific NUMT insertion sites are enriched in species-specific open chromatin regions ($p \approx 6.41 \times 10^{-4}$ in human and $p \approx 2.31 \times 10^{-5}$ in chimpanzee). This highlights the possibility of the utility of NUMTs as open chromatin markers. In addition to this, we also show analysis results of the relation between recent retortransposon insertions and species-specific open chromatin regions.

Reference

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