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Emergence of novel CpG islands and genomic imprinting in Mammalian evolution

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Extended Abstract

Introduction:

Genomic imprinting is an epigenetic regulatory mechanism which induces parent of origin dependent expression to a subset of genes. In higher vertebrates, genomic imprinting has been observed only in viviparous mammals (eutherians and marsupials) and some imprinted genes have essential roles associated with fetal and placental development and control of post natal care as well as lactation, suggesting the correlation between the evolution of genomic imprinting and these characteristics of mammals. Therefore, to study how imprinted loci arose during mammalian evolution would be of great importance to understand how these mammalian traits evolved. Parent of origin dependent expression of imprinted genes is mostly controlled by parental allele-specific DNA methylation of the CpG islands called differentially methylated regions (DMRs). Although the essential role of DMRs for genomic imprinting mechanism has been well established, little is known about how they evolved. Comparative genome analysis of the SGCE-PEG10 imprinted domain revealed that PEG10, a retro transposon-derived imprinted gene essential for placental development, was acquired in the common ancestor of marsupials and eutherians.

Furthermore, in marsupials, both imprinting and differential DNA methylation were restricted to PEG10 unlike eutherians, suggesting that the insertion of PEG10 was the origin of imprinting in this imprinted domain. Also, comparative genome analyses in other imprinted domains showed that most DMRs have emerged as novel CpG islands during mammalian evolution. We presume that the emergence of novel CpG island consequent of retro transposon insertion was key genomic change for the acquisition of DMRs that evolved imprinted loci in mammalian genomes.

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