

专论与综述

Alu家族及其生物学意义Alu family and its biological significance

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摘要

Alu家族是灵长类基因组特有的含量丰富的短散在重复序列, 在基因组中的拷贝数现在已经达到了50万, 每个拷贝长度约300 bp。目前, 对于Alu序列的功能了解得还不透彻, 推测主要与基因调控有关, 如基因重排、CpG甲基化、hnRNA选择性剪切、结合转录因子和激素等。同时, Alu家族是人类群体遗传学、法医学、肿瘤学等的重要研究手段。Alu元件的插入、删除和重组导致了許多先天性遗传疾病和癌症。Abstract:The Alu family of short (~300bp) interspersed elements(SINEs) is one of the most successful mobile genetic elements, having arisen to a copy number in excess of 500,000 within primate genomes in the last 65 million years. The proliferation of these elements had a significant impact on the architecture of primate genomes. Now the functions of Alu elements are still unclear. It is speculated that Alu elements are involved in aspects of gene regulation, e.g. gene rearrangement, CpG methylation, alternative splicing of hnRNA, binding sites of transcription factors and hormone receptors, etc. At the same time, Alu family is an important research method in human population genetics, forensic, oncology, etc. Additionally, Alu insertion, deletion and recombination led to many ancestor genetic diseases and cancers.

关键词 [Alu家族](#) [基因调控](#) [疾病](#) [生物进化](#) Key words [Alu family](#) [gene regulation](#) [disease](#) [human evolution](#)

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