

F-box基因拷贝数目变异的机制研究：以12种果蝇为例

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Mechanisms underlying copy number variation in F-box genes: evidence from comparison of 12 *Drosophila* species

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摘要

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摘要 拷贝数目变异是一种对表型变异和生物进化具有重要意义的基因组结构变异。以前的研究表明不同物种中F-box基因的拷贝数目差异较大。为了深入探索拷贝数目变异的式样和机制,我们以12个果蝇近缘种为研究对象,分析了F-box基因的系统发育关系、进化式样以及它们在染色体上的位置。结果发现,虽然各个物种中F-box基因的拷贝数目差别不大(42-47个),但是仍然存在着很多引起拷贝数目变异的基因获得和丢失事件。这说明表面上变化不大的拷贝数目在一定程度上掩盖了频繁发生的基因获得和丢失事件。通过比较这些基因在染色体上的位置,发现只有在亲缘关系很近的物种之间才能鉴定出有明显微共线性关系的基因组区段。我们还发现,造成F-box基因拷贝数目增加的主要机制是散在重复和串联重复,而反转录转座和新基因的非编码区起源也是两种值得注意的机制。此外,序列变异导致的外显子边界变化以及外显子丢失是引起拷贝数目减少的两种机制。在12种果蝇的最近共同祖先中,F-box基因的拷贝数目与现存物种基本相似,但是基因的获得和丢失事件使得现存物种中的F-box基因在构成上已经有了明显的差别。对数目变异的式样及其与基因功能的关系的研究表明,拷贝数目变异是F-box基因家族“生与死”的进化在基因组层面的系统反映,并有可能为表型变异提供了原始材料。

关键词: 拷贝数目变异 F-box基因 果蝇 直系同源基因

Abstract: Copy number variation (CNV) is a special type of mutational change that plays important roles in phenotypic variation and organismal evolution. To explore the mechanisms underlying copy number variation and to understand its biological significance, we analyzed the phylogenetic relationships, evolutionary patterns and chromosomal locations of F-box genes in 12 closely-related *Drosophila* species. A total of 541 F-box genes were identified and phylogenetic analyses suggested that they are members of 48 gene clusters (or orthologous groups). Although we observed no drastic changes in the total numbers of F-box genes among the 12 extant *Drosophila* species (42-47), we found many gene gain and loss events that have caused copy number variation. These results demonstrated that the similarity in the total numbers of F-box genes among different species has, to a certain degree, masked the frequent and independent gain and loss events. Comparisons of the chromosomal locations of orthologous genes showed that extensive microsynteny could be detected only between very closely-related sibling species. We also found that the main mechanisms that caused the increase in gene number were dispersed duplication and tandem duplication, while retroposition and *de novo* origination from non-coding sequences were two other noteworthy mechanisms. Mutations that caused shifts in exon-intron boundaries and/or losses of exons seemed to be the main mechanisms that underlie decreases in copy number. Although the most recent common ancestor (MRCA) of the 12 *Drosophila* species had a similar number of genes as the extant species we studied, gains of new genes and losses of existing ones have caused changes in the makeup of F-box genes in descendent species. Our study suggested that variations in the numbers of gene copies is a reflection of “birth-and-death” evolution at the genomic level and have provided raw materials for phenotypic and physiological diversification.

Keywords: copy number variation F-box gene *Drosophila* orthologs

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