

Chromosome-level genome assembly of tree sparrow reveals a burst of new genes driven by segmental duplications

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Abstract

The creation of new genes is a major force of evolution. Despite as an important mechanism that generated new genes, segmental duplication (SD) has yet to be accurately identified and fully characterized in birds because the repetitive complexity leads to misassignment and misassembly of sequence. In addition, SD may lead to new gene copies, which makes it possible to test the “out of testis” hypothesis which suggests genes are frequently born with testis-specific expression. Using a high-quality chromosome-level assembly, we performed a systematic analysis and presented a comprehensive landscape of SDs in tree sparrow (*Passer montanus*). We detected co-localization of newly expanded genes and long terminal repeat retrotransposons (LTR-RTs), both of which are derived from SDs and enriched in microchromosomes. The newly expanded genes are mostly found in eight families including *C2H2ZNF*, *OR*, *PIM*, *PAK*, *MROH*, *HYDIN*, *HSF* and *ITPRIPL*. The large majority of new members of these eight families have evolved to pseudogenes, whereas there still some new copies preserved transcriptional activity. Among the transcriptionally active new members, new genes from different families with diverse structures and functions shared a similar testis-biased expression pattern, which is consistent with the “out of testis” hypothesis. Through a case analysis of the high-quality genome assembly of tree sparrow, we reveal that the SDs contribute to the formation of new genes. Our study provides a comprehensive understanding of the emergence, expression and fate of duplicated genes and how the SDs might participate in these processes and shape genome evolution.

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