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## **The structure and evolution of mitochondrial genomes on the example of selected orders of birds**

### **SUMMARY**

Due to its small size, constant gene composition, and the absence of introns, it is believed that the mitochondrial genome of vertebrates during evolution tends to be as compact as possible. Therefore, gene rearrangements and duplications should rarely occur in it, and if they do occur, they should be quickly eliminated. However, the sequences of the mitogenomes of various groups of vertebrates obtained in recent years have revealed that many of them contain duplicated regions. The most interesting group are birds (Aves) because their mitochondrial genomes differ from the other vertebrates in their gene rearrangement. The most common avian gene order, first identified in the chicken (*Gallus gallus*), was considered ancestral to all birds. However, in representatives of multiple orders, other rearrangements containing duplications of the control region and/or adjacent genes have been reported. Repeated regions can be easily overlooked due to inadequate DNA amplification and genome sequencing strategies. This raises the question of the actual mitogenomic duplication frequency and the validity of the current view on the evolution of the avian mitochondrial genome.

Thanks to the new duplication detection strategy used in this study, previously undetected duplicate regions and their associated different gene orders were identified in the mitogenomes of 261 species out of the 338 tested, representing 24 orders of birds. The obtained results made it possible to determine how the duplicated regions evolved and showed that duplications are much more common than previously thought. The distribution of mitogenomes with duplications on the avian phylogenetic tree suggests that the last common ancestors of paleognathic birds (Palaeognathae) and major groups of neognaths (Neognathae) had a tandem duplication involving the control region and adjacent mitochondrial genes. This duplication was probably inherited by many modern phylogenetic lines of birds and has been independently evolved by them by randomly losing extra copies and generating many new gene patterns. Only in some lines the same gene orders appeared independently. The analyses also revealed that the control regions of the same mitochondrial genome in many parrot (Psittaciformes) and passerine (Passeriformes) species are very similar to each other, which indicates that they are homogenized during genome replication. Such homogenization may occur at a frequency that

is greater than the rate at which new species are formed. However, the control regions of falcons (Falconiformes) duplicated before the separation of the analyzed taxa and evolved independently while accumulating mutations. The complicated evolution of the control regions makes them unreliable molecular markers for reproducing the species tree and inferring taxonomic relationships.

This study is the first one to report a new type of heteroplasmy, which was found in 45 taxa representing 12 orders of birds. It consists in the presence of mitogenomes differing in the number of genes, i.e., variants with and without duplication. A variant with a single gene order may arise as a result of a replication slippage, intrachromosomal crossover and deletion, or unequal crossing-over.

Duplication, especially of the control region, appears to have some benefits, e.g., increasing energy production by the mitochondria. It has also been found that birds with duplications in the mitogenome live longer. All of these findings shed new light on the evolution of the avian mitochondrial genome and the importance of duplications within it. They indicate that bird mitogens are still a dynamically evolving genetic system.