

Mammalian karyotype evolution

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Abstract | The chromosome complements (karyotypes) of animals display a great diversity in number and morphology. Against this background, the genomes of all species are remarkably conserved, not only in transcribed sequences, but also in some chromosome-specific non-coding sequences and in gene order. A close examination with chromosome painting shows that this conservation can be resolved into small numbers of large chromosomal segments. Rearrangement of these segments into different combinations explains much of the observed diversity in species karyotypes. Here we discuss how these rearrangements come about, and show how their analysis can determine the evolutionary relationships of all mammals and their descent from a common ancestor.

The Origin of Species (1859) introduced biologists to the concept that allied species are descended from a common ancestor and, indeed, that all forms of life arise from early progenitors. Darwin saw the Natural System as a genealogical arrangement with various grades of difference marked by the terms varieties, species, genera, families and so on. He realized that in order to understand the relationships between species it was necessary to determine “the lines of descent by the most permanent characters whatever they may be”¹. He followed the classifications that were introduced by the *Systema Naturae* of Linnaeus (Tenth edition, 1758 (REF. 2)), which were mostly based on anatomical features of living organisms and their fossil progenitors. But using morphological characters as the basis for classification does not always lead to dependable results as they are often far removed from the genotype.

Fortunately, comparative genomics now provides Darwin's successors with the more appropriate DNA-based characters with which to investigate these relationships. The most productive genetic techniques, in order of increasing resolution, have been chromosome analysis, gene mapping and gene sequencing. In recent years, modern cytogenetics has contributed a large share of information about evolutionary relationships between a great number of mammalian species³. Each species has a characteristic chromosome complement — the species karyotype — that consists of pairs of chromosomes that can be arranged in order of size. One pair are the sex chromosomes, XY in male mammals and XX in females; the remaining pairs are autosomes. By comparing the chromosomes of mammalian species, much can be learned not only about karyotype

evolution, but also about the mechanisms involved and their significance for speciation and, indeed, about some of the genetic factors that distinguish between closely related organisms.

Here we focus on the mechanisms and events that have contributed to mammalian karyotype evolution, and the insights that this examination provides into evolutionary relationships among mammals. We begin this overview by describing the extent of genome conservation and how genome variation within this conserved structure can lead to the modifications in development that are observed in all organisms. We briefly introduce the modern molecular cytogenetic methods that are used in evolutionary studies. These methods are comparatively recent, and their use in phylogenomics is not always appreciated by evolutionary biologists³. We describe chromosome homology mapping on which evolutionary trees of the main groups of mammals can be based. Finally, we show how such studies have led to the derivation of ancestral karyotypes of extinct species. The results presented here complement and extend phylogenies based on gene sequencing. A full discussion of the latter is outside the scope of this Review, and the reader is referred to the literature for further details⁴.

Genome conservation

Genome sequencing of an increasing number of organisms reveals that the transcribed sequences of genomes of all species are highly conserved. This conservation is seen at several levels and includes homology of genetic linkage groups, and even large regions of chromosomes. Groups of genes that are linked together and in similar order can be found in species as disparate

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doi:10.1038/nrg2199

