

If two species are closely related, we may expect that their genetic maps will reveal similar gene orders and relative locations. More distantly related organisms may also display similarities in their genetic maps, at least over short distances. This was illustrated in Fig. 5.1, where contents of mouse chromosomes and a human chromosome were compared. We also saw in Chapter 5 how alterations in gene orders could serve as measures of the evolutionary distance between organisms.

14.3.1 Conservation of Synteny and Segmental Duplication

As indicated in Section 1.3.2, two or more genes are said to be **syntenic** if they reside on the same chromosome. A set of genes g_1, \dots, g_n that are syntenic in organism A and also syntenic in organism B represents a **conserved synteny**. Note that the chromosomes in the respective organisms may not be related to each other in a simple way. A group of genes that display conserved synteny *and* that appear in the same order and relative map positions in the two genomes constitutes a **conserved segment** (also called a *conserved linkage* or *syntenic segment*; see Fig. 1.4B). As was illustrated in Fig. 5.1, there are multiple instances of conserved segments shared by the human and mouse genomes, even though these two lineages diverged from each other more than 83 million years ago. The existence of conserved segments indicates that analysis and annotation of such segments in one genome can guide analysis in a related genome. For example, the human genome sequence provided a useful framework for generating a high-resolution physical map for the mouse genome (Gregory et al., 2002). This was done by matching end-sequence reads from inserts in a mouse BAC library with the assembled human genome sequence. The tiling could be checked by comparing fingerprints (patterns of insert restriction fragments) of overlapping BACs.

Once conserved segments between genomes have been identified by approaches to be described below, the rearrangements that have occurred since the two organisms diverged from the common ancestor can be inferred. A particularly simple example is the X chromosome in humans (HsaX) and mice (MmuX). The plot in Fig. 5.3 showed that runs of contiguous loci in MmuX have homologs in HsaX. These runs in the two organisms could be related by a series of reversals. Other mouse-human chromosome comparisons (Fig. 14.6) show much more complicated relationships involving many chromosomes. For example, Hsa3 has conserved segments corresponding to six different mouse chromosomes. Of course, both mouse and human contain DNA unique to each organism (white regions, Fig. 14.6). Moreover, each organism contains organism-specific transposable elements.

Within-genome comparisons can reveal large and small genomic regions that appear repeatedly, even though they are not transposable elements. **Segmental duplication** refers to duplicated regions that encompass only a fraction of the genome. The *Arabidopsis* genome provides a striking example of