



Figure 1. Evolution of the Y Chromosome

Early in evolution the two sexes may have differed at only a single, autosomal locus with one sex (designated proto-male) being heterozygous at this locus and the other sex (proto-female) being homozygous. The “male determining allele” is shown in yellow. If mating requires one member of each sex, then individuals homozygous for the male-determining allele cannot arise. At this early stage, physiological differences between the sexes will be subtle, comparable to those that distinguish the two mating types in yeast. To prevent the formation of intersex states, crossing-over will be suppressed within and around the male-determining locus (the suppressed area is shown as dark). Mutations will accumulate within this region because suppression of crossing-over will reduce their ability to spread through the population and, hence, the selection pressure against them. The degenerate region in which crossing-over is suppressed will gradually expand (“Muller’s ratchet”) until the chromosome has lost most of its active, functional genes. A small, active region must remain that is homologous to the X chromosome in order to allow pairing and crossing-over at meiosis. This is the pseudoautosomal region (PAR). The autosome originally homologous to the future Y (A in the diagram) will itself evolve, largely through translocations from other autosomes, eventually forming the distinctive X chromosome. The X, like other chromosomes, is a mosaic of DNA fragments put in place at different periods through evolution; some of these are ancient and some are relatively recent. This is illustrated by the differently patterned patches on the proto-X and X chromosomes. On the human X, the more recent arrivals are enriched in genes that escape X inactivation.