

LECTURE 35

EVOLUTIONARY EFFECTS OF CHROMOSOMAL REARRANGEMENTS

A. Six basic types of chromosomal rearrangements:

1. Robertsonian fusions/dissociations

- a) major mode of chromosome number change
- b) no known (or empirically demonstrated) fertility effects
- c) impact is decrease/increase in number of linkage groups

2. Heterochromatin additions/deletions

- a) most frequent type of chromosomal alteration
- b) no known (or empirically demonstrated) fertility effects
- c) effects on crossing over (recombination) could impact maintenance of co-adapted gene complexes

3. Duplications/deficiencies

- a) Duplications – impacts could relate to coding structural genes (e.g., rRNA genes) or to *forbidden mutations* and divergence to new (but related) genes
- b) Deficiencies – most should be highly deleterious or selectively neutral

4. Inversions and translocations: these will be the main focus of discussion (below).

B. Net effects of major chromosomal rearrangements: inversions and translocations.

1. Both types of rearrangement alter gene position.

- a) Both change the position(s) of genes relative to other genes or chromosomal regions (e.g., heterochromatin). This can lead to “position effects.”
- b) Both change linkage relationships: inversions (same chromosome) increase or decrease the distance between genes, whereas translocations (different chromosome) change linkage relationships completely.

2. Both types of rearrangement permit the build-up and maintenance of co-adapted gene complexes:

- a) By “suppressing” recombination and/or crossing over, “blocks” of alleles can be generated and maintained in specific arrays/

3. Both effect marked reductions in fertility. There also is the possibility that chromosomal “breaks” may occur in functionally important sequences.

C. Two critical questions:

1. How frequent in nature are major chromosomal rearrangements?
2. How are the deleterious effects on fertility and viability overcome?

Answer to Question #1: Very!

- a) Most species, even very closely related ones, differ by one or more major chromosomal alterations (rearrangements). This indicates the past occurrence of chromosomal rearrangements in the history of most species.
 - (i) Apparent exceptions are the *homosequential* species of *Drosophila* where different, reproductively isolated species are identical polytene band for polytene band. It is worth noting, however, that some of the homosequential species of *Drosophila* differ markedly in the composition, amount, and location of constitutive heterochromatin (satellite DNA sequences).
- b) Intraspecific chromosomal polymorphisms (especially inversions) are numerous in many animal and plant species groups (e.g., paracentric inversions in continental *Drosophila*, pericentric inversions in *Peromyscus*).
- c) The estimated rates of *de novo* occurrence appear high. In humans, for example, estimates of the embryonic-birth incidence of all chromosomal alteration (including aneuploidy, polyploidy) ranges from 1/500 to 1/1,500.

Answer to Question #2: Good question!

- a) The issue must be considered from a population perspective. Each rearrangement arises in a single individual within a population, i.e., the individual becomes heterozygous for the rearrangement (e.g., *st/in*). The remainder of the population is homozygous for the other arrangement (*st/st*).
- b) Most rearrangements (certainly inversions and translocations) are probably “lost” at inception, either because of effects of chromosome breakage or because of fertility problems in the chromosomal heterozygote (remember there is only a single chromosomal heterozygote in the population at inception). However, the occurrence of both chromosomal polymorphisms *within* species and chromosomal differences *between* species indicates that not all major chromosomal alterations are not lost at inception.
- c) What must happen for the *in* sequence to increase in frequency to the point where the population becomes polymorphic for the two sequences is that the chromosomal heterozygote (*st/in*) must leave more gametes than *st/st* individuals. Note that of the three possible genotypes in this situation (i.e., *st/st*, *st/in*, and *in/in*), the chromosomal heterozygote (*st/in*) is expected to have *reduced fertility* relative to the chromosomal homozygotes.

- (i) The only possibilities whereby the *in* sequence could increase in frequency *despite* the (expected) effects on fertility are:
- (a) genetic drift acting on the population (small effective population size),
 - (b) natural selection, where the advantage of being a structural heterozygote outweighs the disadvantage, and
 - (c) meiotic drive, where (for some reason) the *in* sequence is inherited disproportionately to its frequency in the population.
- d) Regardless, once the *in* sequence increases in frequency (and passes through the bottleneck”), the population becomes polymorphic, with three chromosomal genotypes existing in the population. These chromosomal genotypes are: *st/st*, *st/in*, and *in/in*.
- D. What happens after the population passes through “bottleneck” when both chromosomal sequences occur in the population in detectable frequencies?
1. Once the population is in the “polymorphic” state, the situation is *dynamic*, with one of several potential outcomes
 - a) The population returns to fixation for the *st* sequence (i.e., all individuals become *st/st*).
 - b) The population goes to fixation for the *in* sequence (i.e., all individuals become *in/in*).
 - c) The population “splits” into two chromosomal “races” that are fixed for the two different sequences, i.e., one subpopulation becomes fixed *st/st*, while the other subpopulation becomes fixed *in/in*. This typically represents the beginning of incipient speciation, where the chromosomal difference serves as a primary (postmating) reproductive isolating mechanism.
 - d) The populations remain polymorphic for the chromosomal alteration.
 2. Alternatives (a) – (c) can occur via genetic drift, natural selection, or meiotic drive; in most cases, either genetic drift or selection are invoked as the primary mechanism(s).
 3. Alternative (d) can only be explained under a model of selection.
 - a) Genetic drift (ultimately) should lead to increasing homozygosity (fixation of a chromosomal sequence) because of associated inbreeding effects in small populations.
 - b) Similarly, meiotic drive (if operational) also should result ultimately in fixation of one of the chromosomal sequences (if only because of a possible interaction between meiotic drive and genetic drift).

E. Models of selection operating to maintain (sustained) chromosomal polymorphism.

1. Overdominant selection: The chromosomal heterozygote more heterozygous for genes than either chromosomal homozygote.
 - a) This is a simple model, where rearrangement heterozygosity maximizes and maintains heterozygosity for genes.
 - b) It would be highly effective in species with historically small effective population sizes where genetic heterozygosity would be eroded continually by genetic drift.
2. Frequency-dependent selection: Different (homozygous) combinations of chromosomal genotypes are favored under varying habitats (e.g., seasonal differences).
3. Both models insofar as large chromosomal sequences are concerned are very difficult to test empirically.