Subject - M.Sc Botany, (Sem-IV)

MBOTEC – 1, Paper : Cytogenetics and Crop Improvement

Topic – Evolution of Karyotype (UNIT II)

Dr. Punam Ranjan
Assistant Professor
Department of Botany
Patna Science College, Patna.
e-mail gayatripunam@gmail.com

Chromosome and Karyotype

Nuclear genomes are contained within chromosomes representing **genetic linkage** groups. The number, size and shape of chromosomes, constitute the **karyotype** of an organism, and it vary considerably among groups of eukaryotes.

- **What is the importance of studying Evolution of Karyotype?**

Chromosomes are not only structures which result as end products of a series of gene-controlled developmental processes; they are themselves the bearers of the genes or hereditary factors. This at once puts them into a different category from all other structures of the body. White (1945) has observed that, chromosomes are not merely aggregates of discrete genic units. But they are units of karyotype in themselves. Changes in the chromosomes bear a direct relationship to genetic-evolutionary processes.

Variation in Karyotype

Genome size and chromosome numbers are generally stable in some phylogenetic clades, but may vary considerably in others. Several mechanisms responsible for the variation in size, shape and number of chromosomes, as well as in DNA content between organisms, are recognized.

Cause of variation in Karyotype -

- Change in size and shape by gain (i.e. insertion or duplication) or loss (i.e. deletion) of DNA
- Through rearrangements within or between chromosomes
- Ascending or descending dysploidy respectively
- By ploidy related mutations involving the entire complement (polyploidy) or individual chromosomes (aneuploidy).

Karyotype evolution can be studied by using comparative genetic and genomic analyses. Palaeogenomics, which is an integrated approach combining genetic and cytogenetic maps, EST data sets and whole-genome sequences, enables researchers to trace the evolutionary history of genomes within a phylogenetic framework. It

also helps to elucidate whole-genome duplications and genome reshuffling that has led to extant chromosome complements. **Figure 1**

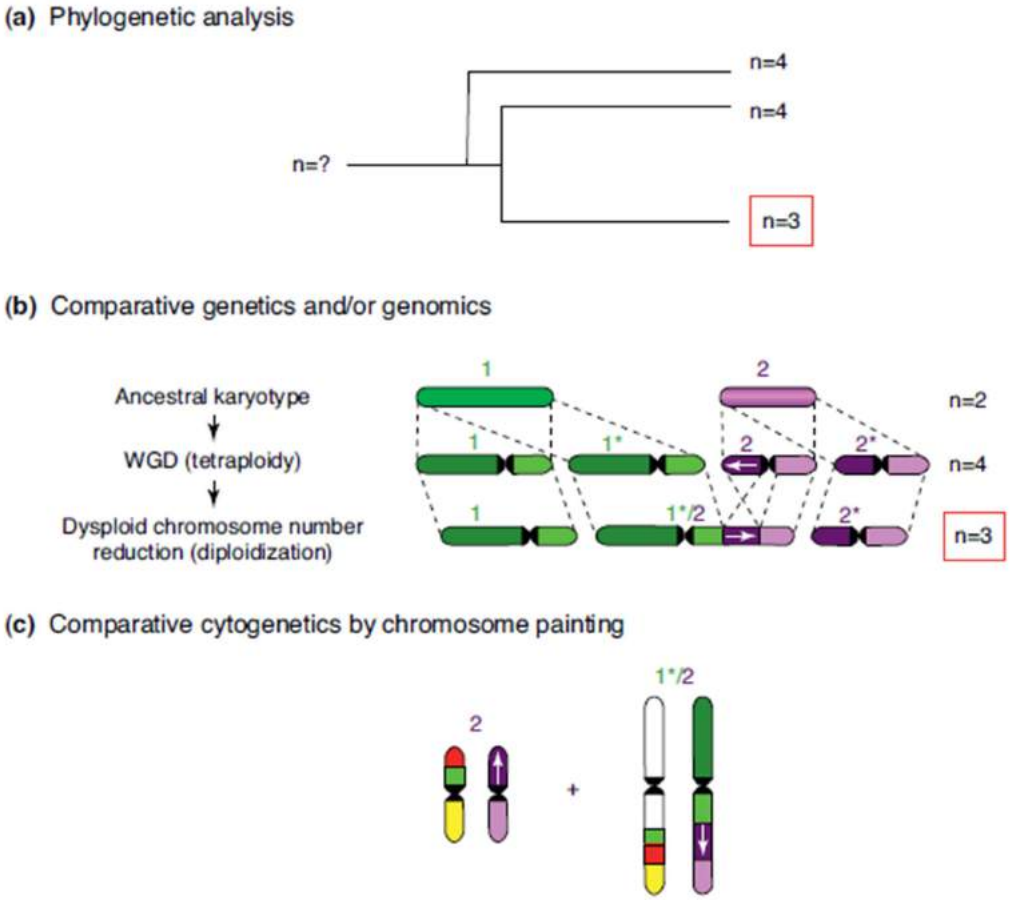


Figure 1. Showing, how to study evolutionary history of an extanct genotype to know its most probable origin by redescribing its karyotype. (Figure from The trends in Genetics mentioned at Reference no. 2)

<u>Evolution of Karyotype</u> (Mechanism for variation in Karyotype)

The evolutionary history of a karyotype is often difficult to determine, especially for older events. With time, the accumulation of chromosome rearrangements will obscure the exact identity, number and order of events that have occurred along a lineage leading to extant karyotypes. There are, however, techniques to help reconstruct this blurred history. Comparative chromosome painting has proved to be useful for tracing karyotype evolution in mammals and in plants (e.g. in Brassicaceae).

Karyotype evolution has proceeded to different degrees in the different groups since they diverged from the common ancestor. There has been ample opportunity for chromosomal rearrangements to occur during the evolution of different species, but there has evidently been strong selection against total genome scrambling.
As a result of karyotype evolution, each species has a unique arrangement of homologous chromosome segments known as evolutionarily conserved chromosome segments (ECCS) (Langford and Breen, 2003)

Chromosome Rearrangements

Various intra- and inter-chromosomal rearrangement types (explained below and illustrated in **Figure 2,** that cause karyotype evolution:
1 Intra-chromosomal inversions
2 Non-homologous inter-chromosomal translocations
3 Centromere-centromere or telomere-telomere fusions

Inversions

Inversions involve the detachment of a chromosome segment, its rotation through 180 degrees and its subsequent reattachment. As a result the order of the genes in that segment is reversed with respect to the rest of the chromosome. Intra-chromosomal pericentric (including the centromere) or paracentric (not including the centromere) inversions of chromosome blocks do not affect the overall size of the chromosome but they do affect the arrangement of segments within it and may well change the relative lengths of the two arms. For example, if an acrocentric chromosome acquires a pericentric inversion, it can be transformed into a metacentric chromosome, whereas if an acrocentric or metacentric chromosome acquires a paracentric inversion, the morphology of the chromosome will not be changed. Such reorganisations may increase or decrease the number of evolutionarily conserved chromosome segments in a karyotype as well as change their arrangement. Inversions are also produced through the activity of transposable elements. Segmental duplications occurring as a result of the insertion of transposable elements could sponsor chromosomal inversions by the process of recombination.

Translocations

Translocations involve the detachment of a segment from one chromosome and its attachment to a different (non-homologous) chromosome. The significance of this

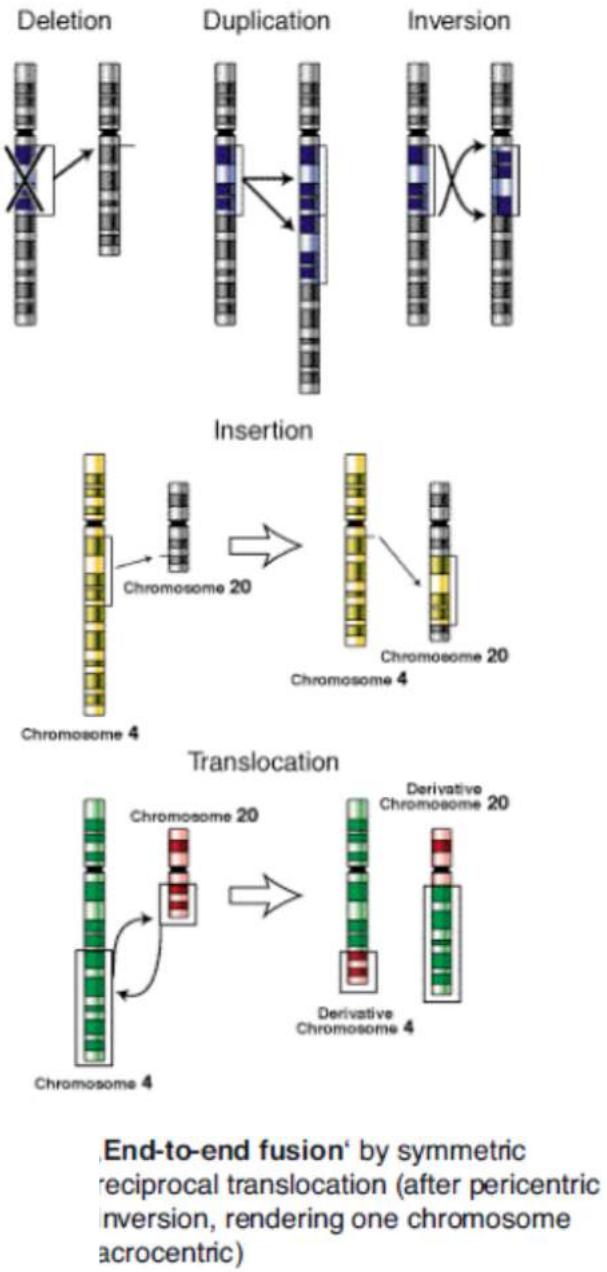
is that genes from one chromosome are transferred to another chromosome and their linkage relationships are altered. When pieces of two non-homologous chromosomes are interchanged without any net loss of genetic material, the event is referred to as a reciprocal translocation. Segmental duplications caused by the activity of transposable elements may cause translocations by recombination. During meiosis, heterozygous translocated chromosomes could be expected to pair with their non-translocated homologues in a cross-like pattern. This configuration is diagnostic of a translocation heterozygote. Cells in which the translocated chromosomes are homozygous do not form crosses. Instead, each of the translocated chromosomes pairs smoothly with its structurally identical partner.

Fusions

Non-homologous chromosomes can fuse at their centromeres, creating structures called Robertsonian translocation chromosomes. For example, if two acrocentric chromosomes fuse, they will produce a metacentric chromosome; the tiny short arms of the participating chromosomes are lost in this process. Such chromosome fusions have apparently occurred quite often in the course of karyotype evolution (Ward, et al., 1987). For example, G-banding studies suggest that each of the large chromosomes of the Indian muntjac deer evolved by the fusion of numerous small ancestral acrocentric chromosomes.

This mechanism could explain how chromosomal variants become fixed in populations and how non-random segregation could affect karyotype evolution across a broad phylogenetic range. Chromosomes can also fuse end-to-end (a telomere-telomere fusion) to form a structure with two centromeres. If one of these is subsequently inactivated, the chromosome fusion will be stable. Such a fusion evidently occurred in the evolution of our own species. Human chromosome 2 (Homo sapiens (HSA) 2), which is metacentric.

2 A



End-to-end fusion' by symmetric reciprocal translocation (after pericentric inversion, rendering one chromosome acrocentric)

2 B

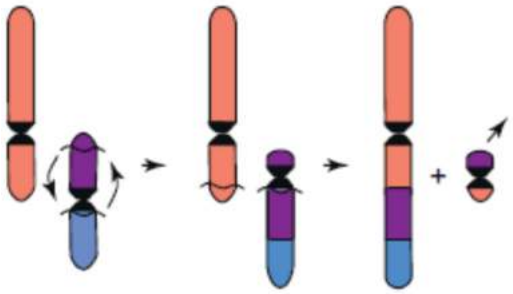


Figure 2: Some examples of Chromosomal rearrangements detected in Human that causes Change in Karyotype. Source for Figure 2 A: CHAPTER 1 Introduction ftp.sanger.ac.uk › pub › resources › theses › Langford. Source for Figure 2 B: Figure from The trends in Genetics mentioned at Reference no. 2.

Phenotypic Effects of Germline Chromosome Rearrangements

Homozygous segmental deletions that remove several genes are usually lethal because at least some of the missing genes are likely to be essential for life. Duplications, in contrast, may be viable in the homozygous condition, provided they are not too large. In the heterozygous condition, deletions and duplications could affect the phenotype by altering the dosage of groups of genes. Usually, the larger the chromosome segment involved, the greater the phenotypic effect. In fact, aneuploidy for very large chromosome segments typically is lethal. However, sometimes small heterozygous deletions or duplications can have a lethal effect, indicating that the aneuploid region contains at least one gene with a strict requirement for proper dosage. For example the loss of one copy of some developmental genes can cause severe problems because of **haploinsufficiency**, where a single copy of a gene cannot produce enough protein.

Inversions and translocations may also affect the phenotype. Sometimes the rearrangement breakpoints disrupt genes, rendering them mutant. The mutant phenotype appears if the rearrangements then become homozygous. It is also possible to get the mutant phenotype where the translocation is heterozygous, for example where parts of two separate genes fused to create a gene whose product is damaging and/or inappropriately expressed. In other cases, the breakpoints are not themselves disruptive, but the genes near them are put into a different chromosome environment, where they may not function normally. Such a gene is influenced by chromosome position effect. If an euchromatic gene is juxtaposed near heterochromatin, the heterochromatin could exert a repressing effect on the gene function.

❖ Methods of studying Karyotypic evolution

- Comparative Banding
- Comparative Genome mapping

❖ Approaches for constructing comparative map

- Genetic linkage analysis

- Somatic hybrid nanlysis
- Comparative sequence analysis
- In situ hybridization analysis

Example - Karyotype evolution study in Crepis

Chromosomes which resemble each other in outward appearance are not necessarily alike in genic content or in hereditary potentiality. The changes in the chromosomes produced by gene mutations are by definition invisible even under the most powerful microscope. In some genera, like Pinus and Quercus differentiation of species appears to have been entirely by this process, since in their gross structure the chromosomes of all the species are so similar as to be indistinguishable. This is shown both by comparative karyology of somatic chromosomes and by the regular pairing of the chromosomes at meiosis in species hybrids. Furthermore, the chromosomes of different species may look exactly alike as to size and form, but may nevertheless possess many differences in gross structure, such as translocations and inversions, which become evident only when they pair with each other in species hybrids. For example- genus Paeonia (Stebbins1938)

On the other hand, the superficial appearance of the chromosomes may be completely altered in two entirely different ways, without comparable changes in the genotype. One way is by means of large unequal reciprocal translocations of chromosomal segments. Such changes have little or no effect on the external morphology or the physiological reactions of the plant. They have been induced artificially in *Crepis tectorum* (Gerassimova 1939) and in several other examples.

Darlington (1937*)* has shown how conditions favoring the loss or gain of a chromosome can be produced by means of unequal translocations. If two different nonhomologous chromosomes both have sub terminal or nearly terminal centromeres (the "acrocentric" chromosomes of White, 1945), then a segmental interchange involving the long arm of one chromosome and the short arm of the other will produce one long chromosome with a median centromere ("metacentric," White 1945) and one very short, fragment type of chromosome.
Figure 3

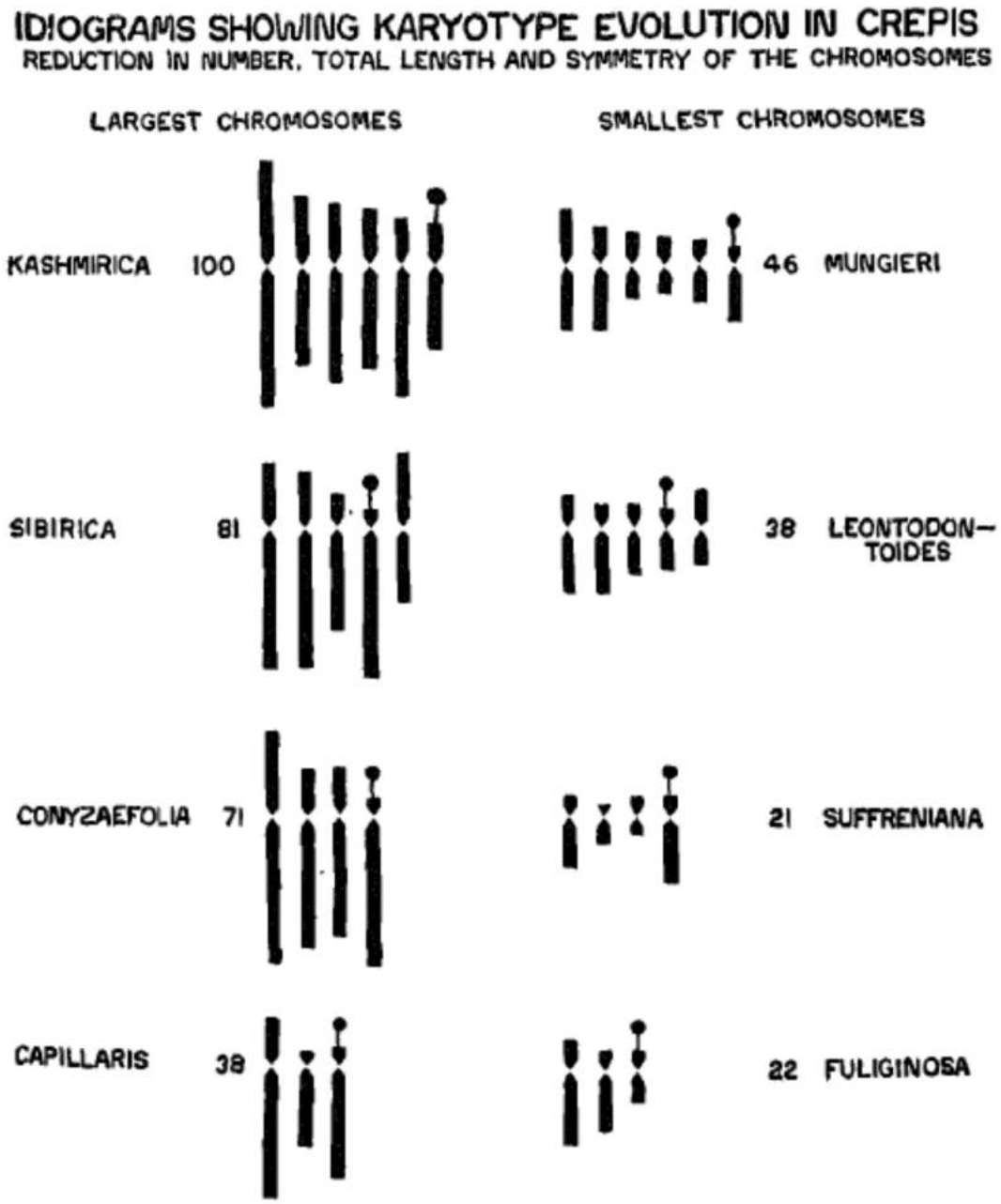


Figure 3. Ideogram showing the basic haploid chromosome complements of various species of Crepis and illustrating the phylogenetic progression in the reduction of chromosome number and size. (Babcock 1947, University of California Press) Source: Evolutionary Trends I- The Karyotype. Reference no. 3

Studies suggested that that the most primitive species have the highest basic numbers and that the trend has been toward reduction. In the genus Dubyaea,

which in both vegetative and floral characteristics is more, primitive and generalized than Crepis, and which forms a connecting link between the large genera Crepis, Lactuca, Prenanthes, and Hieracium (Stebbins 1940), all the species counted have the haploid number x = 8, while the basic numbers of most of the genera related to Crepis, namely, Youngia, Prenanthes, Hieracium, Lactuca, Sonchus, Launaea, and Taraxacum, are either x = 8 or x = 9. In Crepis itself, the species which in one way or another approach one of these other genera have x = 7, x = 6, or x = 5, while those with x = 4 and x = 3 are the most typical of the genus and the farthest removed from the other genera. In addition, the reduction in basic number has been accompanied by certain definite trends of specialization in external morphology. The most primitive species of Crepis are those, with x = 6 (the seven-paired species are a specialized offshoot, perhaps forming a transition toward the related genus Ixeris). Their primitiveness consists in the perennial habit, the presence of shallow rooting rhizomes, entire or shallowly dissected leaves, relatively large involucres in a few-headed inflorescence, more or less imbricated, unspecialized involucral bracts, and unspecialized, unbeaked fruits or achenes. The species with x = 5, x = 4, and x = 3 possess to an increasing degree some of the following specializations: annual habit; deep taproots; deeply pinnatifid leaves; smaller and more numerous involucres; specialized involucral bracts in 'two series, the outer reduced and the inner variously thickened and otherwise modified; and beaked, sometimes strongly dimorphic, achenes. This correlation between reduction in basic number and increasing morphological specialization is not complete; some species with x = 5 (*C. foetida*) are highly specialized in nearly all of their characteristics, while at least one three-paired species, C. capillaris is less specialized in most respects than are some species with x = 4. But when all the facts are considered, the series can be read in only one way. The three-paired species are related only to four-paired species of Crepis; all the latter are typical of the genus and show clear connections with the former and with the five-paired species, while it is only in those species with six and five pairs that we can see the evolutionary connection between Crepis and other genera.

Both morphological and genetic evidence show that the lower numbers have arisen independently several times. The three species with x = 3, *C. capillaris, C. fuliginosa,* and *C. zacintha*, are all more closely related to various unconnected

four-paired species than they are to each other. Similar independent connections can be traced between various four-paired species and related ones with x = 5.

Darlington's postulates concerning the mechanism by which basic chromosome numbers are reduced, is therefore fully confirmed for Crepis.

Suggested Books and References

1. Principles of Genetics 6th Edition by Snustad and Simmons.
2. Interpretation of karyotype evolution should consider chromosome structural constraints. Ingo Schubert and Martin A. Lysak. Cell Press. Trends in Genetics, June 2011, Vol. 27, No. 6.
3. Evolutionary Trends I- The Karyotype. (Read this free available online, it has very detailed description of all types of basics involved in evolution of Karyotype, with examples)

   https://courses.botany.wisc.edu/botany_940/15Stebbins/Chapters/Chapt12.pdf

4. Pdf. CHAPTER 1 Introduction ftp.sanger.ac.uk › pub › resources › theses › Langford