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Chapter

Chromosome Banding and Mechanism of Chromosome Aberrations

Sanjay Kumar, Asikho Kiso and N. Abenthung Kithan

Abstract

Chromosome identification depends on the morphological features of the chromosome and therefore karyotype and its banding pattern analyses are the most suitable technique to identify each and every chromosome of a chromosome complement. Moreover, aberrations caused by breaks play an important role in the evolution of a chromosome set and chromosome complement by decreasing or increasing the chromosome number. Therefore, both the aspects are discussed in detail in the present chapter. At present, the chapter will highlight the karyotype and its components, karyotype trends, evolution and its role in speciation, banding pattern and techniques, chromosome differentiation and linearization, banding applications and their uses, detection and analysis of chromosomal aberrations, chromosome and chromatid types of aberrations and mechanism of the formation of chromosome aberrations and breaks for karyotype evolutionary trends.

Keywords: chromatin, karyotypes, karyoype trend, karyotype evolution, chromosome banding techniques and pattern, chromosome aberrations and detection

1. Introduction

Chromatin is a mixture of DNA, RNA and proteins could be easily visible during the interphase and prophase of the cell division cycle. Chromatin from interphase (loose mixture of DNA, RNA Protein) to thick mitotic structure (tightly packed or compressed mixture of DNA, RNA Protein) packed through Nucleosome model of DNA packing. Chromatin is divided into euchromatin and heterochromatin. Euchromatin is a part of chromatin which takes less stain, loosely packed, genetically active, involved in active transcription, dispersed appearance with more DNA content than RNA. On the other hand, heterochromatin is slightly opposite to the euchromatin with dark stained region, tightly packed, genetically inactive, not involved in the active transcription, thick appearance with more RNA content than DNA. Heterochromatin could be of two type's constitutive and facultative heterochromatin. Constitutive heterochromatins are permanently conserved or condensed and in stable form i.e. not changed from heterochromatin to euchromatin and vice-versa. It consists of multiple repeats of DNA sequences with quite less density of genes in this region which are transcriptionally inactive. Most probably, thick and condensed state of the constitutive heterochromatin, replicates late in S-phase with reduced frequency of genetic recombination.

Facultative heterochromatins are not permanently conserved or condensed and in unstable form i.e. easily changed from euchromatin to heterochromatin and vice-versa [1–3].

Heterochromatic regions could be easily recognized on chromosome structure in the form of chromomeres, chromocentres and knobs. Chromomeres are regular features of all prophase chromosomes but their number, size, distribution and arrangements are specific for a particular species at a particular stage of development. Chromocentres are the regions with varying size near the centromere in the proximal regions of chromosome arms. Some of the Chromocentres could be resolved into large number of strings of chromomeres which are much larger in size as compared to the chromomeres found in the distal region of the chromosmes arms during the mid-prophase. The relative distribution of the chromocentres on the chromosome structure, sometimes considered to be of significant evolutionary value. Knobs are considered to be a spherical bodies or regions with spherical in shape and sometimes diameter of these spherical bodies is equal in width to chromosome arm, but the size may vary i.e. less or more than the diameter of chromosome arm. For example, a very distinct such type of chromosome knob could be observed in maize (*Zea mays*) at pachytene stage of meiosis I. It could be considered as a valuable chromosome marker for distinguishing chromosome of related species and races [4–6].

2. Concept of karyotype and its components

Karyotype may be defined as the study of chromosome morphology of a chromosome complement in the form of size, shape, position of primary constriction or centromere, secondary constriction, satellite, definite individuality of the somatic chromosomes and any other additional features. Karyotype highlights closely or distantly related species based on the similarity or dissimilarity of the karyotypes. For example, a group of species resemble each other in the number, size and form of their chromosomes. There may be 12 different types of karyotype categories depending on increasing asymmetry in chromosome complement [7]. The degree of asymmetry of chromosome complement depends on the four arm ratios (1 to 4) and the size of the smallest and largest chromosome and three different proportions of the metacentric chromosomes (ABC) of a given chromosome complement. Arm ratio 1 being the most symmetrical and 4 is the most asymmetrical. There are various quantitative karyotypic ratios to observe the karyotype variations and precise description of the karyotype such as relative length, centromeric index, total form percent, disprsin index, disparity index, coefficient of variation, volume of chromosomes, value of relative chromatin and so on. Asymmetric karyotype may be defined as the huge difference between the largest and smallest chromosome as well as less number of metacentric chromosomes in a chromosome complement. Similarly, symmetric karyotype may be defined as the small difference between the largest and smallest chromosome as well as more number of metacentric chromosomes in a chromosome complement [8, 9].

The principle ways in which karyotypes differ from each other are (i) basic chromosome number, (ii) form and relative size (V➔J or L➔I) of different chromosomes of the same set, (iii) number and size of satellites (related to those positions of the chromosome which form nucleoli) and secondary constrictions (NOR region of chromoosmes), (iv) absolute size of the chromosomes, (v) distribution of material with different staining properties i.e. euchromatin and heterochromatin [10, 11].

2.1 Karyotype trend

Karyotypic trend may be defined as the evolutionary changes in the chromosome complement by increasing or decreasing its base chromosome number which showed a definite direction of movement or pattern of its movement either from polyploidy to diploid or vice versa. For example, *Luzula* species (Juncaceae), also called wood rush, a monocot with holocentric chromosomes showed huge variation in genome and pattern or direction of chromosome movement from diploid to polyploidy or vice versa through symploidy and agmatoploidy phenomenon. The phenomenon could be related to the ascending or descending dysploidy which is also known as pseudoaneuploidy where chromosomes rearrange themselves within or between the chromosomes to decrease or increase the chromosome number in the chromosome complement of a particular species. Simploidy is the phenomenon of fusion of chromosomes together to reduce the chromosome number while the agmatoploidy breaks the chromosomes (fission) to increase the chromosome number for a particular species (**Figure 1**). The trend of *Luzula* species are as follows, L. purpureo-splendens (2n = 2x = 6; chromosome length 6.66 μm), L.elegans (2n = 2x = 6; chromosome length 4.62 μm), *L. alpinopilosa* (2n = 2x = 12 ± 1; chromosome length 2.55 μm), L.nivea (2n = 2x = 12; chromosome length 1.70 μm), L. sylvetica (2n = 2x = 12; chromosome length 1.48 μm), *L. multiflora* (2n = 6x = 36; chromosome length 1.32 μm), and *L. sudetica* (2n = 8x = 48; chromosome length 0.52 μm). The trend could be explained to understand that species with chromosome number 12 has merged their chromosomes together through a process of symploidy to occur speciation of a new diploid with 2n = 6. This could be possible because the size of the chromosomes increasing in L elegans and L. purpurosplendans. Similarly, there is a possibility of agmatoploidy phenomenon has been occurred and the size the chromosomes decreased in *L. sudetica*. Moreover, it clearly

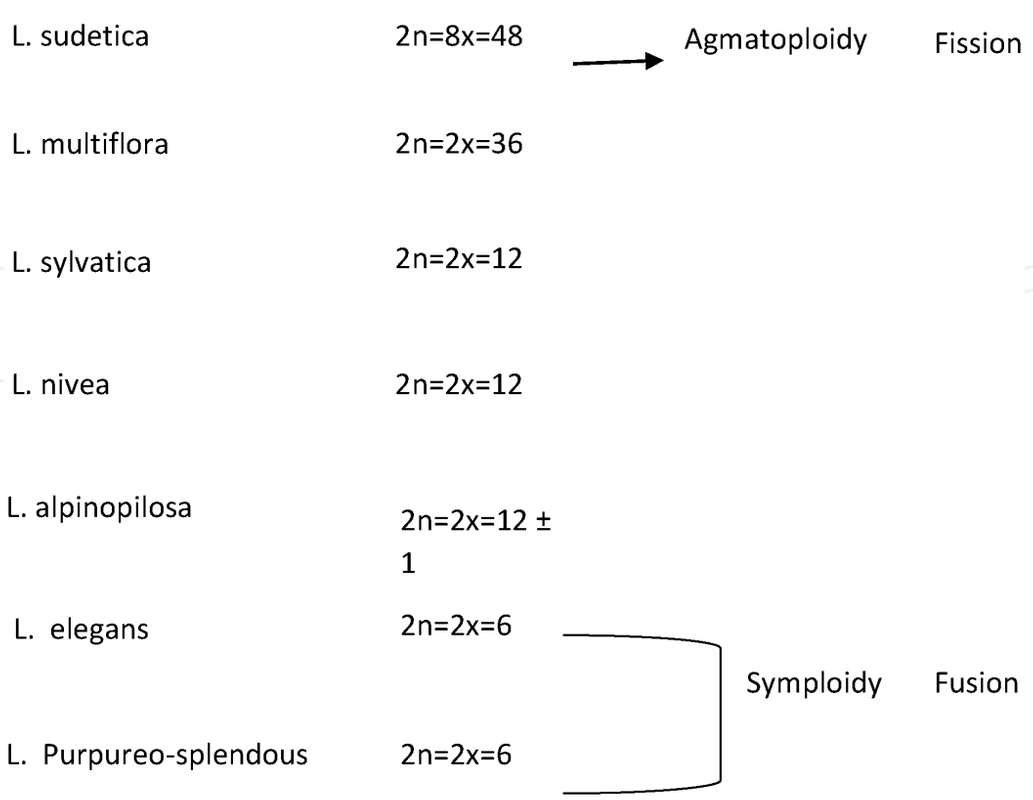


Figure 1.
Karyotype trend in Luzula *species by fission and fusion of the chromosomes.*

suggests the trend of chromosome size decrease from diploid species towards polyploidy species or vice-versa [12, 13].

2.2 Karyotype evolution and speciation

Karyotype evolution may be defined as a phenomenon of change in chromosome number with time and space where fusion or fission and rearrangement may take place among chromosomes to decrease or increase its chromosome complement as well as to adapt themselves in available climatic conditions at that particular place and for their survival over a period of time (**Figure 2**). For example, triticeae genome with a basic chromosome number 7 had undergone 5 centric and 7 nested fusion to reach the present 5 chromosome structure karyotype in *Brachypodium distachyon*. The fusion in the genome of triticeae and *B. distachyon* involved different combinations of ancestral chromosomes and therefore they were independent of each other [14]. When triticeae genome was crossed with *B. distachyon* genome [triticeae, 2n = 2x = 14 (TT) × *B. distachyon*, 2n = 2x = 10 (BB) = hybrid, 2n = 2x = 12 (TB)], a hybrid of 2n = 2x = 12 chromosomes was considered to be the ancestor of present day *B. distachyon* (2n = 2x = 10). It was considered that approximately 7 nested fusion (large number of breaks in the chromosomes and then repositioning of the fragments) suggest descending dysploidy in the ancestor to reach the present *B. distachyon* (2n = 2x = 10). The evolution of eudicot and monocot lineages is driven by two counteracting processes i.e. whole genome duplication (WGD) and diplodization. It is inferred that allthe present grass genomes evolved from an intermediate ancestor with 12 chromosomes which itself arose from 5 or 7 chromosome ancestor through WGD and two reciprocal translocations (**Figure 3**). Although this particular rearrangements is common in grasses, it rarely occurs in eudicots in which end to end fusion are mostly responsible for reduction in chromosome number [15, 16].

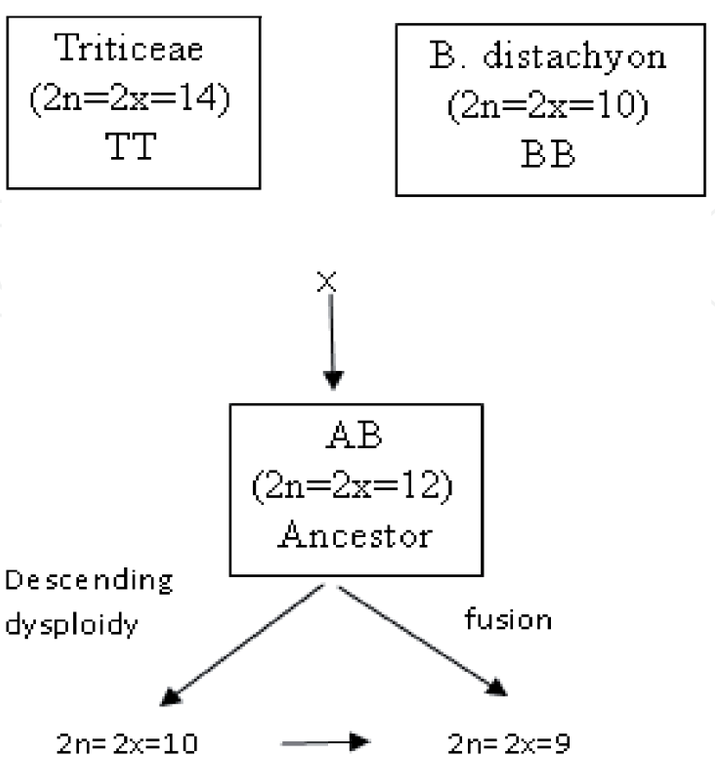


Figure 2.
Karyotype evolution in Brachypodium *by fusion or descending dysploidy from the ancestor.*

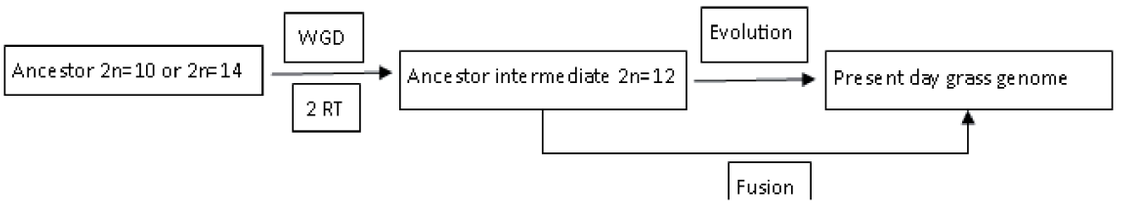


Figure 3.
Schematic diagram to show the karyotype evolution in grasses.

Similarly, evolutionary history of a karyotype is oftenly difficult to trace for older events and with time the accumulation of chromosome rearrangements remove the exact identity, number and order of the events occurred along the lineages leads to an extant karyotype. There are techniques to reconstruct the Extant karyotypes by extracting the information's from the extinct and its close relatives to get the hint for the direction of evolution of karyotype under paleogenomics [17].

3. Banding techniques

This is a technique for the identification of chromosomes and its structural abnormalities in the chromosome complement. Chromosome identification depends on their morphological characteristics such as relative length, arm ratio, presence and absence of secondary constrictions on the chromosome arms. Therefore, it is an additional and useful tool for the identification of individual chromosome within the chromosome complement. Further, it could be used for identification of chromosome segments that predominantly consist of either GC or AT rich regions or constitutive heterochromatin. The technique which involves denaturation of DNA followed by slow renaturation permits identification of constitutive heterochromatin as it mainly consists of repetitive DNA. On banded chromosome, darkly stained or brightly fluorescent transverse bands (positive bands) alternate with the lightly stained or less fluorescent (negative bands). The bands are consistent, reproducible and are specific for each species and each pair of homologous chromosomes. Banding techniques also revealed the extensive genetic polymorphism manifested as inter-individual differences in the size and stain ability of certain chromosomal segments. Initially four basic types of banding techniques were recognized for the identification of Human chromosome complement (Q, C, G and R bands) and later on two additional major type of bands were developed (N and T bands) for complete identification of the chromosome complement (**Figure 4**). Now present bands and newly developed bands or molecular

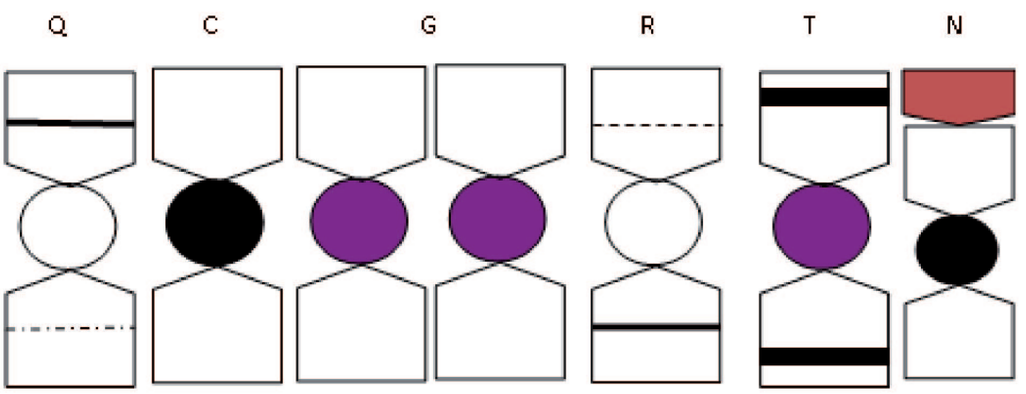


Figure 4.
Characteristics of various basic banding techniques.

bands are widely used in animals and plants for the identification of chromosome complement, chromosome aberrations as well as traces of phylogeny [18, 19].

3.1 Banding pattern of Q, G, R and C bands on Human chromosome complement

Chromosome band C and G clearly identifies the secondary constrictions of chromosome number 1, 9 and 16 sometimes slight or occasional staining were found for secondary constrictions of chromosome 9. C-band clearly stains and identifies peri-centromeric region on the chromosomes, while band Q slightly stains peri-centromeric region of chromosome 3. Both C and Q bands are equally important for staining the distal part of long arm of Y chromosome but for both the bands partial staining was recorded for satellites. Partial Q band staining was reported for chromosome 3, 13 and 21 while other chromosomes were recorded with intense staining. The C-band was found suitable to stain important regions and structures of the Human chromosome complement and widely used band. The G band is also known as golden band for the identification of the homologous pair within complement and could be considered a basic band before application of any sophisticated and molecular approach for further investigation [20].

3.2 Code system for banding pattern

There were 3 letter coding system for the banding procedure, for example, first letter codes for the type of banding to be done; second letter codes for the general technique to be used and third letter codes for the stain to be used. For instance, code QFQ indicates the Q-band to be done, fluorescence technique to be used and quinacrin mustard stain to be used during banding procedure. Similarly, other codes may be QFH, QFA, GTG, GTL, GAG, CBG, RFA, RHG, RBG, RBA, THG and THA depending on the bands, techniques and stains [21].

4. Chromosome bands

4.1 Q (quinacrine) band

The band stains the chromosome with fluorochromequinacrine mustard or quinacrinedihydrochloride (atebrin), observed under fluorescence microscope, and shows a specific banding pattern [22]. The fluorescence intensity is determined by the distribution of DNA bases along the chromosomal DNA with which the dyes interacts. The AT-rich regions enhance the fluorescence while GC-rich regions quench the fluorescence. The brightly fluorescent Q bands show high degree of genetic polymorphism but the fluorescence of Q band is not permanent and fades rapidly, therefore, the banding must be observed on fresh preparation and selected metaphases photographed immediately for further analysis. The disadvantage of the technique is the application of an expensive fluorescent microscope.

Q banding could also be achieved by fluorochromes other than quinacrine or its derivatives e.g. daunomycin, hoechst33258, BrdUetc which enhances AT-rich regions and quenches GC-rich regions. Acridineornage stains AT-rich regions red and GC rich regions green.

4.2 C (constitutive heterochromatin) banding

C banding was developed as a by product of in situ hybridization experiments on the localization of the mouse satellite DNA [23]. Centromeric regions with

constitutive hterochromatin where satellite DNA was located stained more deeply with Geimsa than the rest of the chromosome [24]. The C banding technique is based on the denaturation and renaturation of DNA and the regions containing constitutive heterochromatin stain dark (C band) and could be visible near the centromere of each chromosome. The C bands are polymorphic in size which is believed to correspond to the content of the satellite DNA in those regions. C banding allows precise analysis of abnormalities in the centromeric regions and detection of isochromosomes. The C banding in combination with simultaneous T-banding in particular, extends to easy detection of dicentric rings [25]. Sometimes, C banding also permits to ascertain the parental origin of foetal chromosomes and distinguish between maternal and foetal cells in amniotic fluid cell culture.

4.3 G (Giemsa) banding

The banding could also be recognised as the modification of C banding procedure [26]. The technique permits the accurate identification of each pair of the chromosomal complement as well as recognition of the specific chromosomal rearrangements within complement. The preparations are permanent after staining with giemsa. A number of modifications for G bands have been developed and proposed such as pre-treatment with trypsin, urea, enzymes and salts, even though original ASG method (G like bands) as well as trypsin method often slightly modified and most widely used.

G bands correspond exactly to chromomeres of meiotic chromosomes but the mechanism leading to the visualization of the basic chromosome pattern is still unclear. The process is believed to be associated with denaturation and distributin of non-histone proteins and rearrangements of chromatin fibres from G negative to G positive bands.

4.4 R (Reverse) banding

R banding patterns are based on the thermal treatment of chromosomes and in general the reverse of the Q and G bands developed and proposed by Dutrillaux and Lejeune [27]. The ends of the R banded chromosomes are almost or always found positive and the centromeric regions are easily distinguished. This permits the observation of minor abnormalities in the terminal regions of chromosomes and the precise determination of chromosomal lengths. The technique is performed on a fixed chromosomal preparation and is based on heat denaturation of chromosomal DNA. R bands (GC rich regions) are more sensitive to DNA denaturation than Q and G bands (AT-rich regions). Giemsa stained R bands can be observed under phase contrast microscope while acridine orange stained R bands require fluorescence microscope.

4.5 T (Telomeric) banding

T bands are, in fact, the segments of the R bands that are most resistant to the heat treatment and the transition patterns between the R and T bands could be obtained by gradual treatments. Therefore, it may be regarded as the modifications of the R banding technique [28]. The clear marking of telomeric regions of chromosome with T banding enables the detailed analyses of the structural rearrangements at the ends of chromosomes. It also allows the detection of human chromosome 22 and its involvement in translocation. The usefulness of this method is for the detection of dicentric rings that were undetectable by other procedures. T bands can be observed either after giemsa or acridine orange staining.

4.6 N (Nucleolar organizing regions) banding

The NOR regions could be selectively stained by techniques involving either giemsa or silver staining. The giemsa technique developed by Matsui and Sasaki [29] allows the staining NOR after extraction of nucleic acids and histones. The technique N banding was improved by Funaki et al. [30]. The silver staining technique fall into two categories; a) Ag-As method: the method is based on the staining with combined silver nitrate and ammoniacal silver solutions; b) Ag method: in this method, staining with ammoniacal silver is omitted. NOR banding stains only those regions that were active as nucleolus organizers in the preceding interphase as well as useful for visualization and study of satellite associations.

4.7 Sequential banding

In routine cytogenetic diagnosis, a single banding technique is usually sufficient for the detection of chromosomal abnormalities e.g. G banding or R banding, but sometimes, more complicated chromosomal rearrangements often require sequential staining of the same metaphase by several banding techniques and the process is known as sequential banding. The quality of chromosomes in sequential banding deteriorates with each staining therefore; it restricts the sequential banding up to 3 or 4 different staining techniques (**Figure 5**). For example, single metaphase ➔ First procedure, Q banding ➔ Second procedure, G banding ➔ Third procedure, C banding ➔ deteriorates the chromosome quality ➔ therefore, restricts up to 3 or 4 staining procedures [31].

4.8 Simultaneous banding

This is the technique that produces simultaneously two types of banding on the same metaphase or on one slide but different metaphases (**Figure 5**). For example, same metaphase ➔ first procedure, G banding ➔ second procedure, C banding OR single slide with different metaphases ➔ first procedure, C banding ➔ second procedure, T banding. Simultaneous banding restricts up to two staining procedures in different or single metaphase and results in precise estimation of chromosomal aberrations [32].

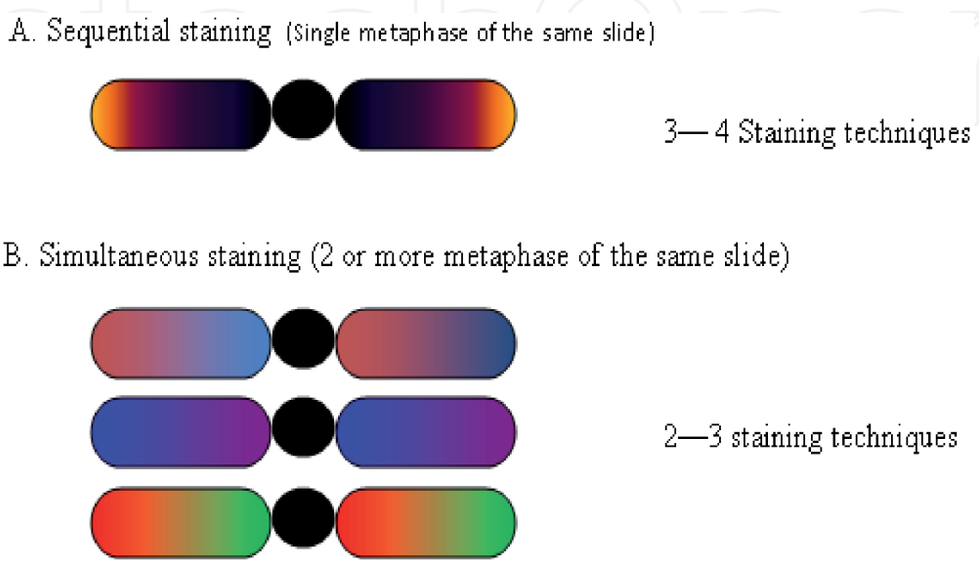


Figure 5.
Banding techniques for sequential and simultaneous staining.

4.9 High resolution banding

The general trend is to pick highly thick and contracted metaphase chromosomes are chosen for banding procedures which gives enough banding patterns for prediction of any kind of aberrations or arrangements within complement, but sometimes it fails also. Therefore, elongated chromosomes (earlier stages of mitotic divisions before reaching metaphase stage) are standardized for banding patterns and prediction of aberrations or arrangements called high resolution banding. Elongated chromosome standardization and preparation could be obtained through synchronization of cell cycle or use of various chromosome anti-contraction reagents through a procedure known as cell cycle synchronization technique. Cell synchronization is a process by which mitotic cells at different stages of cell cycle in culture are brought to the same phase through physical fractionation or chemical blockage or inhibition of DNA synthesis during S-phase. Cell synchrony may be defined as the progression of cells through cell cycle. The possible procedure for high resolution banding pattern are described. Mitotic stages (cultured cells) ➔ cell synchronization by adding chemical reagents ➔ Amethopterin or methotrexate or thymidine or fluorouracil ➔ cultured cells ➔ Blocks DNA synthesis in cultured cells ➔accumulation of cells in S-phase of cell cycle ➔ block released ➔ cell synchrony (large quantities of cells continue their cycle from the same level) ➔ prophase to mid-metaphase range ➔ high number of bands (gives more information as compared to the compact metaphase banding) [33, 34].

The technique allows precise localization of break points in chromosomal rearrangements and detection of minute chromosomal alterations that are undetectable by the mid-metaphase banding techniques [35].

5. Chromosome differentiation

Conventional chromosome banding techniques help us in understanding the patterns associated with chromosomal evolution of a species, speciation processes (formation of a new species) and generation of genetic variability among the species. Similarly, molecular banding techniques such as FISH and GISH provide little more and specific information by causing more differentiation in banding pattern of a chromosome of a particular species. Now, computational analyses (software packages) of the chromosome bands provide maximum information on banding pattern by increasing number of bands which helps to predict the specific and precise result on chromosomal aberrations and arrangements (**Figure 6**). For example, conventional chromosome bands ➔ enough bands for analysis ➔ molecular banding techniques ➔ more bands, more differentiation, more information ➔ computational techniques (software packages) ➔ still more bands, still more differentiation, still more information [36].

6. Chromosome linearization

Chromosome linearization is an important tool under computational analyses of the chromosome bands which suggest that linear chromosomes will provide maximum number of bands as well as information regarding aberrations or arrangements as compared to the twisted, rounded, curved and overlapped chromosomes. The information obtained from the straight chromosomes will be

larger or maximum in quantity and quality. The tool of image linearization enables a better visualization technique which ultimately extends and refines the information that can be extracted from the chromosomes (**Figure 6**). For example, conventional chromosome bands ➔ enough bands for analysis ➔ molecular banding techniques ➔ more bands, more differentiation, more information ➔ computational techniques (software packages) ➔ better organization of cytogenetic data ➔ tool of image linearization ➔ still more bands, still more differentiation, still more information [37].

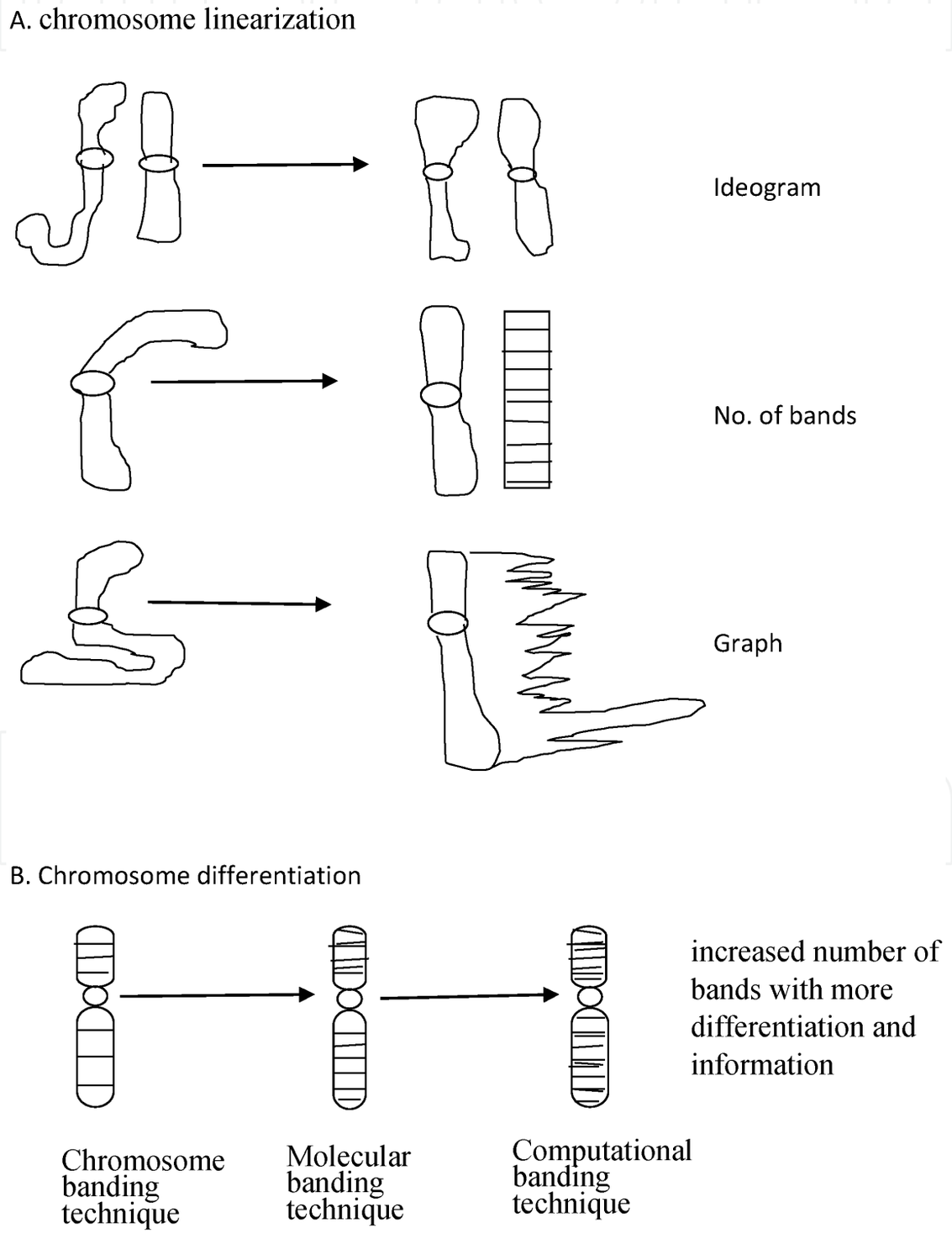


Figure 6.
Characteristics of chromosome linearization and differentiation banding technique.

7. Chromosome banding application

The chromosome banding primarily could be used for the detection and recognition of nature and type of the aberrations, identification of the chromosome involved and most importantly, the location of the presumptive positions of the lesions (usually termed as break points) involved in the structural changes in chromosome complements [38].

The basic requirement for the detection of chromosomal structural changes using the banding methods is disruption in the normal sequential band pattern of a chromosome arm region. The disruption in the chromosomal arm may take several forms and the most common forms are presence of an additional band, absence of an expected band, unexpected change in banding pattern and reversion of a part of banding pattern. The basic requirements for the chromosomal structural changes are possible by the existence of consistent and fixed pattern of chromosomal bands on chromosomal arm region as well as existence of good sequential differentiation between the chromosomal arm bands. Chromosome condensation is a process that occurs as the cell progresses towards metaphase and continues till the cells are held at metaphase by the action of spindle inhibiting chemicals. Consequently, number of bands, banding pattern, clarity and differentiation among the bands would be a function of state of chromosome contraction and possible provides the existence of consistent and fixed pattern of chromosomal bands at metaphase on chromosomal arm region. The good sequential differentiation between the chromosomal arm bands may cause differences in size, staining intensity, longitudinal spacing, numerical concentration of dark and light bands which forms the basis for the identification of individual chromosomes, but sometimes, the quality of differentiation varies between treatments and also between the cells on the same slide by application of different staining and banding procedures [39].

8. Detection and analysis of preliminary chromosomal aberrations

The primary chromosome aberrations could be detected and analysed in three forms such as achromatic lesions, chromosome type and chromatid type structural aberrations. The detection and characterization of aberrations or breaks which falls in the light banded region or euchromatic heterochromatin region which generally takes light stain could be identified as achromatic lesion. Chromosome type structural aberrations could be detected and identified in the form of asymmetrical interchanges (two translocations in the same arm), symmetrical interchanges (reciprocal translocations), inter arm intra changes (centric ring, pericentric inversions), intra arm intra changes (interstitial deletions, paracentric inversions), and breaks [(i) fragment compound (usually contains two terminal regions) and suspects an incomplete dicentric or centric ring; compound fragment with a terminal and unrelated non-terminal region suspects an incomplete complex interchanges, (ii) fragment simple (genetic material from one arm only) but with altered banding sequencing, suggest that sequence alteration is most frequently cause the inversion of a chromosomal proximal segment which is most probably indicates an incomplete paracentric inversion, (iii) fragment simple but the normal band sequence; (a) short arm with normal band sequences, if fragment present and not related to short arm might be incomplete reciprocal translocation or pericentric inversion; if fragment present and related to short arm may be true terminal deletion; (b) short arm with abnormal band sequences, first, observe for a distal inversion or incomplete paracentric inversion, second, observe for distal genetic

material missing or for incomplete interstitial deletions, third, observe for presence of any minutes and fourth, if any extra additional distal segment is present but not related to short arm with abnormal band sequences, it may be regarded as incomplete complex changes and look for the origin of additional segment, (iv) fragment simple but non terminal; perhaps represents incomplete acentric ring or large interstitial deletion]. Chromatid type structural aberrations could be detected and identified in the form of interchanges (involve the interchanges of chromatids), interarm intra changes (requires two breaks and causes pericentric inversions, double duplications-deletions, dicentric and centric rings), intra arm intra changes (two breaks involved and caused the formation of isochromatid deletion, duplication-deletion, chromatid minutes and paracentric inversions), and breaks [{causes chromatid terminal deletions and may be of various types; (i) a tandem duplication may be present in the complete chromatid, (ii) a tandem duplication may be present either in centric or acentric portion of the incomplete chromatid, (iii) the origin of acentric fragment may be of intercalary type and the sister chromatid has a normal pattern but it may show a bending opposite to the site of deletion, (iv) a paracentric inversion could be possible adjacent to the break in the acentric fragment but other sister chromatid may be normal, (v) a paracentric inversion could be possible adjacent to the break in the centric region or portion but other sister chromatid may be normal}].Additional chromatid type structural aberrations could be possible in the form of as isochromatid (isochromatid exchanges), insertions and additional dark bands in one of the chromatid at the exchange point. It is important to locate the position of aberration or break points at some stage in the production of breakage and rejoining of chromatid threads as this is the event that causes disruption and could be observed using different techniques. A detectable point of breakage is generally referred to as 'break point' but 'presumptive break point' would be more appropriate in light of consideration [40].

9. Mechanism of formation of chromosome aberration

DNA breaks (especially dsDNA) is a serious threat for cell when unrepaired or misrepaired, as they can result in genomic instability or later on may lead to chromosomal alterations and even cell death. The chromosomal aberrations formation is one of the major alteration formed during dsDNA breaks. Moreover, it has been reported that during each cell division approximately 5000 ssDNA breaks were generated per nucleus and approximately 1% of total ssDNA breaks converted in dsDNA breaks. There were two theories forwarded for explanation of how chromosomal aberrations and its formation take place i.e. breakage and reunion theory and exchange theory. The breakage and reunion theory explains that breaks in chromosome may rejoin and form the original structure through restitution and the exchange theory might lead to exchange the aberration by rejoining another type of breaks. dsDNA breaks could be repaired by possibly three pathways i.e. homologous recombination repairing (HRR) which restores the original sequences, non-homologous DNA end joining (NHEJ) which usually generates and repairs small alterations such as base pair substitution, insertions, deletions at break sites etc., and single strand annealing (SSA) which may lead to the formation of interstitial deletions. HRR (requires one break) and NHEJ (requires two breaks) and SSA are important pathways for repairing of dsDNA breaks with one or two breaks in eukaryotic cells as well as mammalian cells [41].

Chromosomal aberrations may be caused by various physical and chemical factors such as ionizing radiations, chemicals and spontaneous dsDNA breaks e.g. endogenous reactive oxygen species, topoisomerases and replication errors.

There are various methods for the detection of chromosomal aberrations such as cytogentic testing [{(a) chromosomal aberration test, (b) micronucleus test; (i) giemsa staining method, (ii) cytokinesis-blocked micronucleus assay method, and (iii) flow-cytometry micronucleus assay method, (c) karyotyping}], molecular cytogentic testing [(a) fluorescence in situhybridization (FISH), (b) microarray comparative genomic hybridization (mCGH)], and prenatal screening to detect foetal abnormalities [42].

The clinical symptoms of chromosomal aberrations includes, a) chromosomal aberrations and spontaneous aberrations (e.g. congenital malformations, heart and renal malformations), b) chromosomal aberrations and cancer (e.g. chronic myeloid leukemia), c) behaviour peculiarities associated with chromosomal aberrations (e. g. turner syndrome), d) changes in course of adolescence and fertility (e.g. premature aging), e) pattern of dysmorphic signs in chromosomal aberrations (e.g. down syndrome, cri-du-cat syndrome) and f) congenital malformations and chromosome aberrations (e.g. dandy-walker malformation, gastrointestinal malformations, CNS-spina bifida) [43].

10. Conclusion

At present, most of the studies are surrounded towards the human health and disease caused by the interaction of genetic and environmental factors. Sometimes it is difficult to understand the genetic constitution or mechanism of chronic diseases. Therefore, it is very important to understand the mechanism behind the abnormal cells either genetic or environmental or both to solve the problems completely. The identification of a particular abnormality at the initial stage is crucial and banding techniques conventional or molecular provide such an opportunity. Banding and chromosome aberrations are played an important key role in the assessment of various risks faced by the genetic constitution of eukaryotic cells. Therefore, it may continue further to assess the risk of various kinds of ailments, diseases, and geno-toxicity induced by the radiations, pharmaceuticals, environmental and synthetic chemicals.

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