Chapter 2

Genomic Rearrangements and Evolution

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Additional information is available at the end of the chapter

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1. Introduction

All genomes in living organisms can change under influence of internal or external factors. That is why genomic materials are commonly defined as dynamic entities and it is believed that they have been repeatedly altered and rearranged since the beginning of the life on the planet [1-4]. Understanding this dynamism is a valuable key to unlock the chest of the mysterious existence story in an evolutionary manner. Therefore, a lot of studies have been conducted on the dynamism of genomic materials in organisms and the count of related researches has gradually risen by the day. An enormous data from these studies call attention to recombinational, tranpositional and mutational processes as three main sources of genomic changes [1,2,5-18].

Recombinational changes of genomes are mainly dependent on internal factors which are closely associated with a great many of intracellular and intercellular interactions. Enzyme catalyzed pathways and predetermined timing are the most descriptive properties for many types of recombination events. For instance, usual meiotic crossing over, the best known recombinational event, always occurs under control of specified enzymatic reactions at a certain time period in the cell cycle [2,4,19-22].

Transpositional events are also important sources for sequential rearrangements in genomes and induced by external or internal genomic material pieces that are described as mobile or transposable elements. In mechanism of transposition, a transposable element changes its relative position within the genome. “Copy and Paste” or “Cut and Paste” postulates work in this process. A transpositional event occurring with the copy and paste mechanism is called as replicative transposition that a transposable element is duplicated during the process and copied sequence transferred into the target genomic sequence, and the other one with the cut and paste mechanism is called as non-replicative transposition that duplication of the trans-

posable element does not occur and the original sequence is transferred from one region into another [5,23-24]. In both cases, a transpositional event is commonly resulted in a mutational phenomenon and alteration in genomic sizes that makes them attractive for genomic evolution studies [6-7,23-26].

Mutations are described as sudden changes in genomic materials induced by internal and external factors [27]. They have importance in medicinal, agricultural and other related researches due to their deleterious, beneficial or functional effects on organisms [5,9,28]. Moreover, enormous potential for construction of novel genes and other types of genomic sequences, they are considered as the most attractive subject for genome evolution [2,29-32].

2. Recombinations

Genetic recombination is a process that is catalyzed by many different enzymes called as recombinases. It can take place in all living cells from bacteria to eukaryota as well as viral genomes. This process mainly results in DNA repair, genomic rearrangements, variations and evolutional forces. Genetic recombinations are assigned to one of two groups according to their mechanism, which can be described as either homologous or non-homologous recombination [2,4,20,22,33-35].

2.1. Homologous recombination

Homologous recombinational events are sequential changes that occur between similar or identical parts of genomic material. In the beginning of 20th century, initial descriptions of homologous recombinations were introduced by W. Bateson and R. Punnett to explain diversions from predicted Mendelian inheritance phenotypic ratios [4,36-37]. This process, which is commonly found in many organisms from bacteria to higher organized eukaryotes, plays a significant role in DNA repair mechanisms and genome evolution by producing variations [2,38-40].

In prokaryotic cellular organisms, the most known types of homologous recombinational events are transformation, conjugation and transduction [41]. All of these events are resulted in genomic variations that have great value for evolution [42].

Transformation was discovered by Frederick Griffith in the late 1920s. His transformation experiments are considered as the beginning mile stone of the molecular biology discipline [5]. In the mechanism of natural prokaryotic transformation, a naked DNA fragment released from a cell is taken up by another under appropriate conditions, thus an exogenous genetic material is introduced into a prokaryotic cell that result in genomic variation. Transformation occurs in several groups of Gram positive, Gram negative and Archaea. A healthy double strand DNA molecule with a homological property and specific size (mostly smaller than 1000 nucleotides) is the most fundamental requirement for transformation [2,41]. Figure 1 illustrates a summarized scheme for transformation.

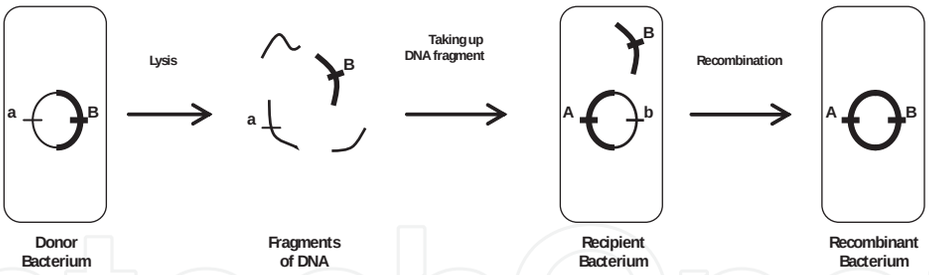

Figure 1. Simple mechanism of transformation

Bacterial conjugation, discovered in 1946 by Joshua Lederberg and Edward Tatum [43], is another process to transfer the genetic information in Prokaryotes. In its mechanism, the transfer of genetic material involves cell to cell contact and a plasmid encoded pathway. The process occurs between a donor cell, which includes a certain type of conjugative plasmid, and a recipient cell, which does not. In this process, the plasmid plays a key role by carrying all related genes on *tra* region. These genes encode the sex pilus (F pili) formation, which allow specific pairing to take place between the donor cell and the recipient cell. After generation of sex pilus mediated cell to cell contact, a copy of the plasmid is transferred to the recipient under control of various enzyme systems encoded by *tra* region. In most cases, this type of recombination does not cause genetic variation at high level because the transferred genetic information is restricted by sequential contents of the plasmid. However, in certain circumstances, conjugative plasmid may integrate into the main genomic material, resulting in the formation of Hfr (High Frequency Recombination) cells. These cells, commonly seen in Gram negative bacterial groups, have significant potential for recombination at higher levels due to leading transfer of genes from the host chromosome [2,41]. Figure 2 shows regular bacterial conjugation events and Hfr formation.

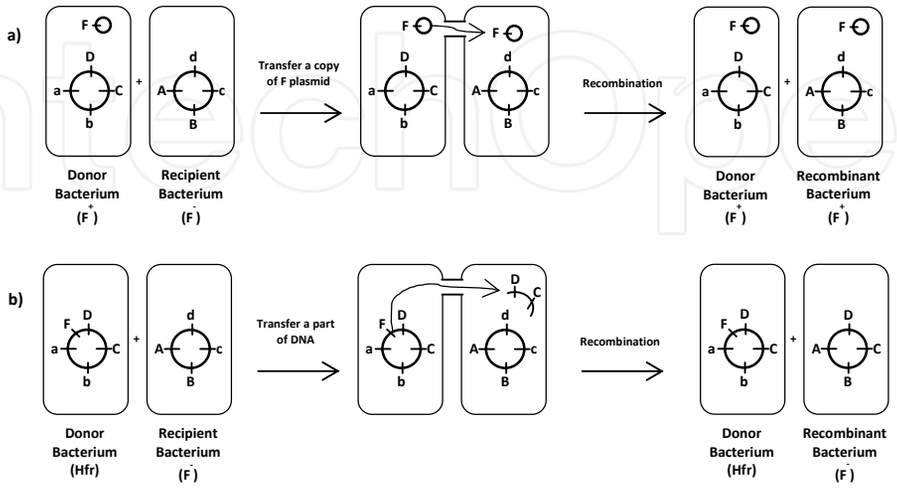

Figure 2. An illustrative scheme for bacterial conjugation of F^+ (a) and Hfr (b) cells

Transduction, initially discovered by Norton Zinder and Joshua Lederberg in 1951 [44], refers to virus-mediated transfers of genetic materials. There are two fundamental mechanisms as generalized and specialized transduction. In generalized transduction, any bacterial genomic sequence may be transferred to another bacterium via a modified bacteriophage that accidentally involves bacterial DNA instead of viral DNA. However, in specialized transduction, bacteriophage includes both bacterial and viral DNA at the same time [2,41]. Both types of transduction events are summarized at Figure 3.

In eukaryotic organisms, meiotic crossing over (chromosomal cross over) is the most well-known example for homologous recombination. This event occurs between homologous chromosomes at prophase I stage in meiosis and results in variation of genetic materials [2,5,45-46]. The scheme of meiotic crossing over is showed in Figure 4.

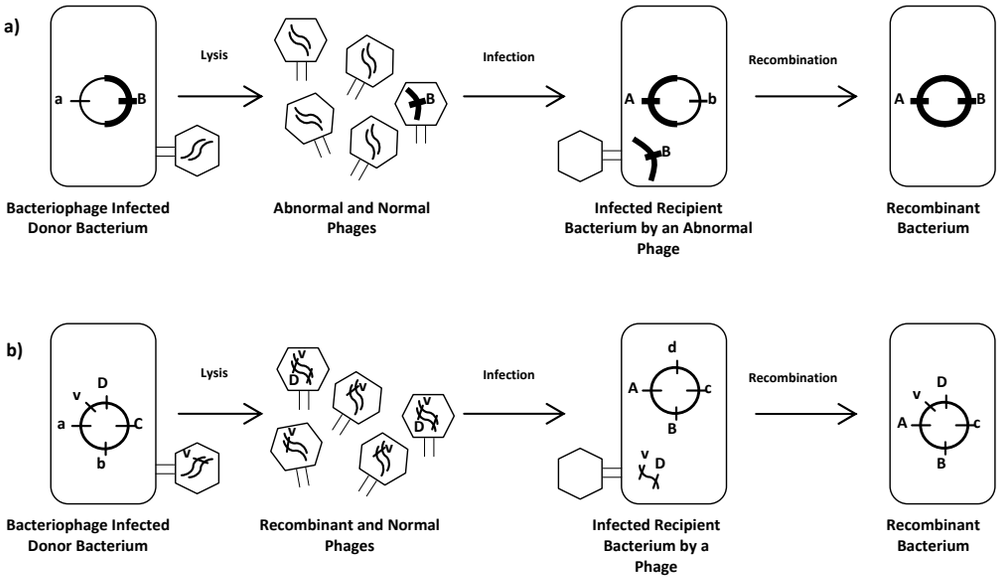

Figure 3. Mechanism of generalized (a) and specialized (b) transduction events

Homologous recombination also plays a significant role in DNA repair mechanisms in both prokaryotic and eukaryotic organisms. It is one of the major DNA repair processes in bacteria [2,46]. For example, double-strand breaks in bacteria are repaired by the RecBCD pathway of homologous recombination [42,47-49]. Moreover, it is well known that similar mechanisms work in eukaryotic organisms.

Homologous recombination also includes non-allelic ones that have been not well documented. These events occur between sequences arisen from duplications or deletions that show high homology, but are not alleles. It is believed that non-allelic homologous recombination has a great importance for evolution due to generating a decrease or an increase in copy number of sequences [50-52].

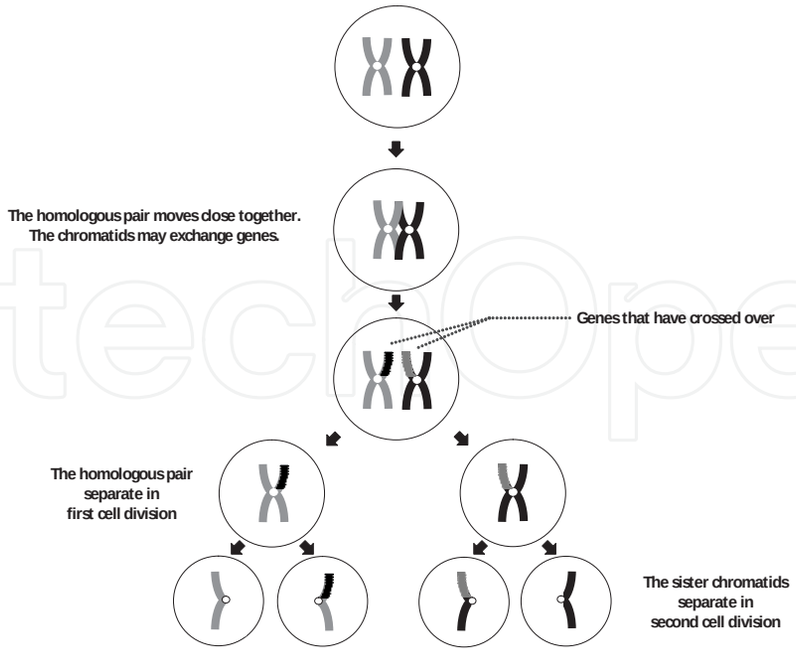

Figure 4. Mechanism of meiotic crossing over

2.2. Non-homologous recombination

Non-homologous recombination, also named as non-homologous end joining (NHEJ), is a pathway that mainly associated with DNA repair that especially works on double strand breaks. Contrary to the mechanisms of homologous recombination, it does not require sequential homology. However, this pathway has been identified in many groups of living organisms from bacteria to multicellular organisms, even in human being, recent studies have mainly focused on eukaryotes much more than bacteria. One reason for this is that prokaryotic DNA repair is heavily done by various processes of homologous recombination.

Nuclease, polymerase and ligase activities play the major role in NHEJ process. Despite its conservative mechanism, this process is generally resulting in variations of genetic materials [2,53-55].

3. Mobile genetic elements

Mobile genetic elements are described as DNA segments that can move within the genome. These include transposons, group II introns, plasmids and viral elements [56]. All these events result in genomic alterations that cause rising of evolutional forces [6,8,24-26,57-61].

3.1. Transposons

Transposons, also named as transposable elements, are major forces in the evolution and rearrangement of genomes [6,26,56]. Discovery of transposable elements was achieved in 1943 by Barbara McClintock who was awarded with a Nobel Prize after 40 years in 1983 [2,58]. Since

that time, the importance of transposons has been well established and much more attention has been given to their formation and consequences [62]. To get more easily comprehensive information, they are divided into three main groups as retrotransposons, DNA transposons and insertion sequences.

3.1.1. Retrotransposons

Retrotransposons can be considered as the biggest group of transposable elements due to their abundance in many eukaryotic genomes (i.e. 49-78% of the total genome in maize and 42% in human) [63-64]. The term "retrotransposon" is attributed to the transposition mechanism that involves via RNA intermediates. In the mechanism, a retrotransposon is initially copied to RNA (transcription), then converted to DNA (reversetranscription) and finally inserted to the genome (integration), and this process is mainly under control of the gene region of retrotransposons encoding reverse transcriptase. These elements can increase genome size and induce mutational events by disturbing genes [2,24,26,56,59,62,65].

Retrotransposons are divided into three main groups according to the operation mechanisms: long terminal repeats (LTRs) encode reverse transcriptase, similar to retroviruses; long interspersed elements (LINEs) do not have LTRs and encode reverse transcriptase and small interspersed elements (SINEs) do not encode reverse transcriptase. LINEs and SINEs are transcripted by RNA polymerase II and III, respectively [66-68].

3.1.2. DNA transposons

DNA transposons are the first discovered ones of transposable elements, initially named as "jumping genes" by Barbara McClintock in 1943 [69]. These are also called as Class II transposons, operate with a "cut and paste" mechanism. In this mechanism, transposition event mainly requires to transposase enzymes. Under control of the enzymatic processes, a DNA transposon is cut out of its location and inserted into a new location on the genome. Some transposases require a specific sequence as their target site; others can insert the transposon anywhere in the genomic material [2,24,41,62].

3.1.3. Insertion sequences

These are also known as IS elements. They are short DNA sequences that act as a simple form of transposable elements. Characterized properties of IS elements are that they have shorter sizes than other types of transposable elements (approximately 700 – 2500 bp), and carry some specific genes such as antibiotic resistance. Insertion sequences are usually flanked by inverted repeats [23,24,70].

3.2. Group II introns

Group II introns were discovered by Alexandre de Lencastre and his teammates in 2005 [71]. These elements, an important group of self-catalytic ribozymes, are generated during RNA splicing, and may cause genetic alterations [71].

3.3. Plasmids

Plasmids are circular and extra chromosomal genomic materials naturally found in bacteria, but rarely in several yeasts as eukaryotic organisms [41]. These elements show intracellular or intercellular mobility (see section 2.1.) that result in genomic alterations and evolutional forces.

3.4. Viral elements

Viral elements are genomic materials transferring between living organisms via virus infections. According to the mechanism of infection, viruses are divided into two categories as lytic and lysogenic. Lytic ones complete their eclipse phase in the cell and cause lysis of the host. However, lysogenic ones integrate their genomic materials into the host genome and directly cause genomic alterations [41]. For example, some retroviruses are common type of lysogenic viral elements and their effect mechanism is similar to retrotransposons.

4. Mutations

The "Mutation" term was initially used by Hugo de Vries in 1905 to describe the phenotypic changes in evening-primrose plant (*Oenothera lamarckiana*). However, it commonly describes any sequential change in the genomic material of living organisms in the present day. Their various effects resulting in genotypic and phenotypic alterations that cause diseases, gaining or loss of advantageous or deleterious properties, attract the scientific attention on mutation focused investigations. In these researches, mutations are generally classified according to the effect mechanisms and size of effected genomic sequences to perform more apparent and comprehensive evaluations [1-3,5,29-31,34].

4.1. Classification of mutations

Effect size of mutations on genomes is one of the most widely-accepted criteria for classification. According to this, mutations can be divided into two groups named as gene mutations and chromosome mutations [5,27].

4.1.1. Gene mutations

Gene mutations are small-scale mutations that effect one or few bases in a genome. However, they can induce many important phenomenon depend on properties of effected genomic sequences. For example, a gene mutation in a protein coding region of genomic material can result in synthesis of a non-functional protein that mostly causes deleterious effects for the organism. Gene mutations are also divide subcategories as base substitution and insertion/deletion [2,5,27,34].

Base Substitutions: They are also called as point mutations. These types of mutations are characterized by taking place of a different base instead of original one in the genome. When a purine base replaces with another purine or a pyrimidine base with another pyrimidine (A↔G

or C↔T), it is called as transition. On the other hand, if a purine base replaces with a pyrimidine or a pyrimidine base with a purine (A↔C, A↔T, G↔C or G↔T), then it is called as transversion.

Figure 5. Base substitutions type of gene mutations

Insertions/Deletions: The insertion term means addition of one or few bases into a genomic material. Contrary to this, deletions are defined as removing of one or few bases from a genome.

Figure 6. Insertion/Deletions type of gene mutations

4.1.2. Chromosome mutations

Chromosomal mutations are described as phenomenon that causes bigger sequence alterations than gene mutations. These are also called as macro-mutations due to their microscopically examination capabilities. There are two main subcategories as structural and numerical alterations in chromosomal mutations [5,9,27,34].

4.1.2.1. Numerical alterations

These types of mutations mainly cause alterations in chromosome numbers in the living cells. Euploidy and aneuploidy are two essential subgroups.

Euploidy: The word "euploidy" refers to cumulative alterations in chromosome numbers. For example, diploid (2n) chromosome number of an organism can be changed to tetraploid (4n) form after these kind of mutations.

Aneuploidy: The word "aneuploidy" refers to non-cumulative alterations in chromosome numbers. For example, diploid (2n) chromosome number of an organism can be changed to nullisomy (2n-2), monosomy (2n-1) or trisomy (2n+1) form after these kind of mutations.

4.1.2.2. Structural alterations

These types of mutations do not change chromosome numbers. However, their effects are mainly on chromosomal structure. According to their effect mechanisms, structural mutations are grouped in four subcategories including deletions, inversions, duplications and translocations [5,9,27,72].

Deletions: Chromosomal deletions include losing of chromosomal pieces resulting in gene losses from the genome.

Inversions: An inversion refers to a phenomenon in which a chromosome break following by 180° rotation and reattachment of the broken piece on the same chromosomal region. It does not cause gene losses, but results in an inverted genetic material.

Duplications: Duplication is a case having two or more copies of a chromosomal region.

Translocations: These types of alterations are arisen from non-homologues chromosomal piece exchanges.

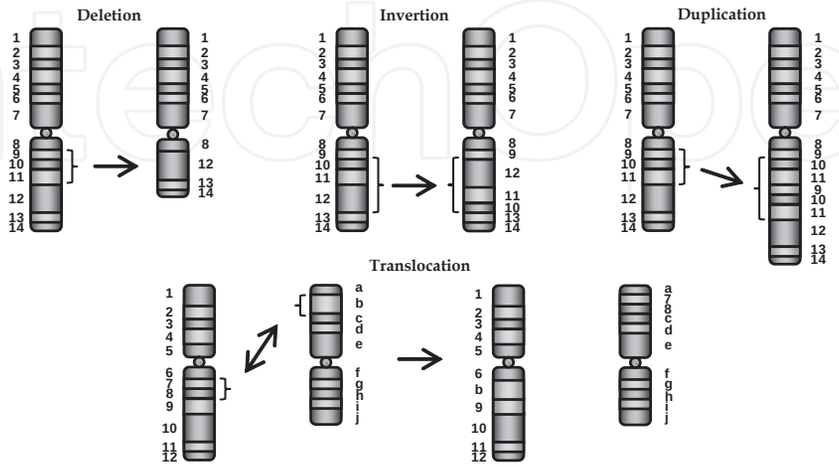

Figure 7. Structural chromosome mutations

5. Genome evolution

The origin of life on the earth has always been an attractive subject for all human beings. The question about formation of the first active biomolecule is one of the most important perspectives in this subject, and has been heavily researched for many years. Initial studies referred to proteins as first biomolecules due to their catalytic activities that operates various reactions for maintaining of life. Although this view was confirmed for a long time, their lack of potential to carry genetic information was the major handicap. In 1982, the commonly accepted thought about the first biomolecule was drastically changed by Thomas Cech and co-workers who published a paper that demonstrate the single intron of the large ribosomal RNA of *Tetrahymena thermophila* has self-splicing activity *in vitro*. This was the first report about catalytic RNA molecules. A year later, Sydney Altman and co-workers pointed out that the RNA component of ribonuclease P (RNase P) from *Escherichia coli* is able to carry out processing of pre-tRNA in the absence of its protein subunit *in vitro*. These studies lead to formation of "RNA world" perspective in genome evolution, and both scientists were awarded by Nobel Prize in 1989. In the recent view, the RNA world term means that ribonucleic acids have both the informational function of DNA and the catalytic function of proteins at the same time [2,12,73-78]. According to this concept, various types of RNAs can be proposed as initial genomes evolved on the planet. Major RNA types and their characteristic properties are given in Table 1.

Although the first genome has a potential to be ribonucleic acid form, instability and limited life of RNA molecules may have forced evolution of a more complex genomic material called as deoxyribonucleic acid (DNA). In this stage, there are several gaps and unanswered questions. However, the most discussed scenario about formation of DNA based genomes from initial RNA molecules (protogenome) proposes a phenomenon that is catalyzed by a reverse transcriptase [2,78,84].

Contrary to the high stability property, evolutional changes are continuously occurring in DNA based genomes that result in development of valuable features for adaptation. These changes have been mainly dependent on external forces since the beginning of the life on the planet (approximately 3.5 billion years ago) [2]. Understanding of this evolutional dynamism in genomic materials requires recognizing definitions of several important terms given in Table 2, prepared according to Eugene V. Koonin (2005) who is senior investigator at National Central of Biotechnology Information (NCBI) and studies on empirical comparative and evolutionary genomics [8].

Up to this point, all mentioned events cause changes in size and construction of genomic materials acting as evolutional forces. The genomic size is referred as "C value". Although the genomic size may reduce via deletions, it has generally intended to increase when compared to the first genome of universal common ancestor (UCA). This expansion is controlled by rearrangement forces, especially duplications and mobile genetic elements. There are two fundamental hypotheses for why genome sizes vary. According to the "Selfish-DNA hypothesis": genome size expansion is due to insertion and proliferation selfish genetic elements such as retrotransposons, and "Bulk-DNA hypothesis": having more genetic bulk can be adaptive because genome size effects nuclear volume, cell size, cell division rate in turn effecting developmental rate and size at maturity, thus it results in organisms with larger body size have larger cell sizes, and organisms with larger cells generally have larger genomes

[15,24-26,63,65,68,85-90]. In his paper, Zhang [88] underlined the positive correlation between duplicated gene amount and evolutional status of an organism. Table 3 represents prevalence of gene duplications in all three domains of life.

Type	Features	References
mRNA (Messenger RNA)	- responsible for coding - represents 4% of whole RNA amount in a cell - called as hnRNA or pre-mRNA before processing in eukaryotes	[2]
rRNA (Ribosomal RNA)	- composes ribosomes - the most abundant RNA in a cell (over 80%) - named as pre-rRNA before processing in all living organisms	[2]
tRNA (Transfer RNA)	- responsible for carrying amino acids to ribosomal complexes - specific for each amino acid - named as pre-tRNA before processing and modification in all living organisms	[2]
snRNA (Small Nuclear RNA)	- responsible for operation of splicing mechanism - found in nuclei of eukaryotes - also called as U-RNA - has a lot of sub-types with various catalytic activities	[2]
snoRNA (Small Nucleolar RNA)	- responsible for chemical modification of rRNA - found in nucleolar region of eukaryotic nuclei - shows catalytic activities	[2]
miRNA (MicroRNA)	responsible for regulation of gene expression double strand molecule intracellular origin (nucleus)	[2]
siRNA (Short Interfering RNA)	- responsible for regulation of gene expression - double strand molecule - extracellular origin (commonly synthetic) - called as small interfering or silencing RNA	[2]
piRNA (Piwi-interacting RNA)	- interacts with piwi proteins - the largest class of small non-coding RNA molecules	[76]
gRNA (Guide RNA)	- acts in mitochondrial mRNA processing - guides insertional or deletional events in mitochondrion	[77]
tmRNA (Transfer-messenger RNA)	- have tRNA and mRNA properties - also known as 10Sa RNA - found in bacterial genomes	[78]
shRNA (Small hairpin RNA)	- responsible for regulation of gene expression - makes a tight hairpin - extracellular origin	[79]
stRNA (Small Temporal RNA)	- regulates gene expression (down regulation)	[80]

Table 1. Major RNA types and their features

Homologs	Genes sharing a common origin
Orthologs	Genes originating from a single ancestral gene in the last common ancestor of the compared genomes.
Pseudoorthologs	Genes that actually are paralogs but appeared to be orthologous due to differential, linage-specific gene loss.
Xenologs	Homologous genes acquired via xenologous gene displacement (XGD) by one or both of the compared species but appearing to be orthologous in pairwise genome comparisons.
Co-orthologs	Two or more genes in one lineage that are, collectively, orthologous to one or more genes in another lineage due to a lineage-specific duplication(s). Members of a co-orthologous gene set are inparalogs relative to the respective speciation event.
Paralogs	**Genes related by duplication**
Inparalogs (Symparalogs)	Paralogs genes resulting from a lineage-specific duplication(s) subsequent to a given speciation event (defined only relative to a speciation event, no absolute meaning).
Outparalogs (Alloparalogs)	Paralogs genes resulting from a duplication(s) preceding a given speciation event (defined only relative to a speciation event, no absolute meaning)
Pseudoparalogs	Homologous genes that come out as paralogs in a single-genome analysis but actually ended up in the given genome as a result of a combination of vertical inheritance and horizontal gene transfer.

Table 2. Homology: terms and definitions from Koonin 2005 [8].

	Total number of genes	Number of duplicate genes (% of duplicate genes)
Bacteria		
Mycoplasma pneumoniae	677	298 (44)
Helicobacter pylori	1590	266 (17)
Haemophilus influenzae	1709	284 (17)
Archaea		
Archaeoglobus fulgidus	2436	719 (30)
Eukarya		
Saccharomyces cerevisiae	6241	1858 (30)
Caenorhabditis elegans	18424	8971 (49)
Drosophila melanogaster	13601	5536 (41)
Arabidopsis thaliana	25498	16574 (65)
Homo sapiens	40580[a]	15343 (38)

[a] The most recent estimate is ~30000.

[b] Use of different computational methods or criteria results in slightly different estimates of the number of duplicated genes.

Table 3. Prevalence of gene duplications in all three domains of life[b] from Zhang 2003 [88].

Besides, Xue et al. [91] laid emphasis on the roles of duplications in genomic size and compositional changes in their studies via exploring the evolution of segmental gene duplication in haploid and diploid populations by analytical and simulation approaches. The result of this study highlighted that duplications do not only cause alterations in genome size but they are also result in many recombinational events that closely related to formation of variations that have value in rising evolutional forces. In another paper, Force et al. [92] focused on the DDC (duplication-degeneration-complementation) model for the alternative fates (nonfunctionalization, neofuctionalization and subfuctionalization) of duplicate genes, and underlined their roles in genome evolution.

Mobile genetic elements also affect genome size. For example, horizontal transfer of transposable elements plays a key role in genome evolution. In their "copy-and-paste" operation mechanisms, retrotransposons, as common examples of mobile genetic elements that may cause horizontal gene transfer, transpose via an RNA-intermediated process, and this increases genomic material size [26,93-94]. Furthermore, all advanced biology sources covering microbial genetic title mention the role of other types of mobile genetic elements including plasmids and viral genomes in formation of variations in genomic size and structure [41].

On the other hand, reduction of genomic size in certain periods is an inevitable fact for genome evolution. In this manner, smaller genomes are more advantageous for selection than bigger ones due to their high replication potentials and metabolic inexpensiveness. Deletions can be given as the main force to diminish genomic size that causes gene losses [95-96]. In a recent paper, Pettersson and co-workers emphasized the role of deletions in regulation of genomic size and its coding density by using a mathematical model to determine the evolutionary fate [97].

A genomic material may accept deletions and reduce its size up to reach minimal genome limits that have the smallest number of genetic elements sufficient to build a modern-type free-living cellular organism. In addition, under some exceptional conditions, genomic materials of several endo-symbionts and co-symbionts carry much less genes than predicted minimal genome rates. For example, although *Pelagibacter ubique* (α-Proteobacteria) is known as a free-living organism with the smallest genome (only 1308 Kb in size and potentially contains 1354 genes), endo-symbiont *Hodgkinia cicadicola* (α-Proteobacteria) has the smallest genome (only 144 Kb in size and potentially contains 188 genes) among known-living organisms [98-102]. According to Juhas and co-workers' study [102], the extremely small genomes of endosymbionts usually encode only the most fundamental process, suggesting that some of their genes might have been transferred into the host cell genome. The endosymbiont *Wolbachia* strains that transfer ~1 Mb fragments of its genomic material to the host genome can be given as a good example for this phenomenon [98-102].

Contrary to the genomic material of *P. ubique* in which there is no pseudogenes, introns, transposons, or extrachromosomal elements, modern-type organism genomes need some or all of these differentiated genetic parts [97]. In this regard, genomic rearrangements have a critical potential via causing structural changes, especially new alleles and new regulatory regions in the genomes can be created by only mutations. There is a huge data giving information about the roles of mutations in evolution in the scientific literature

[1-3,5,8,9,11,12,29-33]. For instance, Halligan and Keightley [103] reviewed the relationship between mutagenesis and its role in genome evolution, and introduced mutational events as the ultimate source of genetic variation.

6. Conclusion

Recent attention of evolutionary studies has shifted to genetics, molecular and cellular biology as a result of finding out principles of genetics and DNA is the main molecule responsible for inheritance. Thus, the popularity of genome-wide studies has increased. In this regard, genomic rearrangement mechanisms (recombinations, mutations or mobility of several genetic elements) are major research topics for evolution of genomes because any change in the DNA molecule of the organisms may cause a valuable process for evolution when it has inheritable potential.

Thus, aim of the present study was conducted to emphasize potential value of genomic rearrangements for evolution, and therefore, basic rearrangement mechanisms were explained in detail, and their evolutionary effects on genomes were briefly discussed via giving important samples in this chapter.

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