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The Evolutionary and Molecular impacts on Human Genetics; Review

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Abstract: The human genome holds a record of the evolutionary forces that have shaped our species. In human genome evolution, DNA sequences-genomics analysis, molecular-based analysis, population genetics, molecular genetics, functional genomics, genome function identification/profiling, and genomics modeling, and simulation have deepened our understanding of human genome evolutionary history, natural selection, and other studies. We study, some factors that influence the evolution of the human genome are the functional modification of DNA, post-translational modification (PTM), gene regulation, analysis of chromatin conservation, modification of histone, structural variations, mutational biases, single nucleotide polymorphism (SNPs) and gene-phenotypes interaction along the genome. By using the natural selection and evolutionary theory as study, these some phenomena will play a vital role in the understanding of the human genome and also breakthroughs in *in-vivo* and *in-silico* studies.

Keywords: Human Genome, Evolution, SNPs, DNA

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1.Introduction

The study in which genes change and change over time, molecular evolution. Is the study of DNA sequence knowledge an integral part of bioinformatics? The variation in cellular, molecular sequence composition like DNA, RNA, and protein. The field of molecular evolution is used in evolutionary biology theories and population genetics to illustrate the patterns of these changes. The human genome is a complete set of human genome sequences, encoded in DNA, into the cell nuclei or a small genome molecule located within each of 23 chromosomes pairs. The size of the human genome is 3,234 Mbp (Gibson, 2012). The human genomics is a basic goal to understand its evolution by decoding the biological program coded into the human genome accurately will reveal answers to the molecular human origin and biological specific characteristics issues. Studying the history of evolution and ancestry of our species also reveals how and why people of today get sick. Evolutionary forces have formed the genome of humans, that often no longer control the circumstances of most humans and disease can be caused by a mismatch between our genes and our environmental factors. The knowledge of human genomics has been provided by functional and comparative genomics, but we still know very little about their spatial mechanisms. What is the 3-dimensional genome structure? How is this system special and when? The structural organization of the genome has long been suspected of placing constraints on its growth. The dynamic physical environment of the human genome has been discovered recently through genome-wide data on chromatin structure, which gains unpredictable insights into chromosome composition, gene structure, mechanisms of mutation and selection (Lee, Abecasis, Boehnke, & Lin, 2014). The recent development and sequence by thousands of human genomes from various populations in the field of genetics, from the dynamics selection through populations and near related species to identify differences in the rate of mutations in the human population, revealed significant complexity in conventional topics, some Specific genetic factors and demographic factors are impacting the evolution of the human genomes (Gluckman, Beedle, Buklijas, Low, & Hanson, 2016; Henn, Botigué, Bustamante, Clark, & Gravel, 2015; Prado-Martinez et al., 2013).

2.The gene regulatory mechanism affects the production of the Human genome

There are some morphological variations in the human genome between closely associated species through gene regulatory changes like as cis-regulatory elements (CREs), such as promotor and enhancers in humans-diseases evolution with change of SNPs. The ability to map the gene, transcript factor sites histone modifications in several organisms and tissues in the genome. During gene expression, organisms like CREs are experiencing rapid turnover within similar tissue. Among other more than 20 mammalian species, liver promoters and enhancers found that 25% of the genes, enhancers, and approximately 10% of the promoters were highly conserved and unique among sequences, and different conserved findings have been observed in humans and mouse for similar organs (Lynch et al., 2015). The evolution and dynamics of the regulatory sequence across body tissues, organisms and groups of species by CREs. For example, transposable elements (TEs) have helped reprogram gene regulatory networks in tissues of mammals, humans and other evolution in different species. The rapid turnover of CREs and retained gene expression are typical features of the development of the mammalian genome with various development and tissue pressures. maintenance, alteration, and effect on genome variation of certain regulatory processes. Integrating genome-wide maps of CREs and expression in techniques for determining *in-vivo* chromatin conformation of DNA may provide a framework for modeling the influence of gene regulation on genome evolution. A recent study on chromatin-looping analysis of the function of genes with topological conserved domains in many human and mouse tissues through cells and species. Integrating genome structure and CREs data over several populations would possibly result in better modeling of the regulatory sequence evolution and acts with time and tissue on gene expression and regulation (Melé et al., 2015; Sakabe, Savic, & Nobrega, 2012; Siepel & Arbiza, 2014; Weirauch & Hughes, 2010).

3.Evolution of Human genetics with chemical modifications to DNA

The human body is made up of numerous different cell types, each containing the same genome, but play a different function in different locations of the human body by PTM. The roles of gene expression systems observed through cell types in diverse genomic organisms in which DNA and histone modifications as well as post-translational modifications (PTM) including methylation and acetylation (Brunet & Berger, 2014; Ptashne, 2013). These modifications can be influenced by environmental factors are inherited across generations. In extensive work, modifying all growth,

disease, aging processes and influencing genetically modified sequence. For example, the chemical modification potentials impose constraints on the patterns of DNA sequence, the conservation degree of DNA, and histone changes among humans and closely related organisms. Paralogous regions are typical between closely related species by changing the status of orthologous for DNA acetylation and DNA methylation. There is a strong link between changes in acetylation-methylation promoters and changes in sequence. Understanding the evolution of these chemical modifications in future debates or other processes like transcription and many other types of post-translational modification (PTM) processes (Heard & Martienssen, 2014; Zhou, Goren, & Bernstein, 2011).

4.Mechanism of interaction between genes and phenotypes

The diversity of human clinical and developmental phenotypes is detected in the human genome by a variety of loci. Developing models that account for relationships between gene and phenotype and multiple genetic variations will be critical to fully dissecting the evolution and complex genetic human traits. Furthermore, histone modifications are a non-additive relationship between genetic variation, because a variety of technological and biological factors its effect on human traits was controversial. For these areas, new biostatistical methods are required to upgrade the models to allow full use of genotype and phenotype data complexity (Sivakumaran et al., 2011; Solovieff, Cotsapas, Lee, Purcell, & Smoller, 2013; Wood et al., 2014).

5.Mutational biases in Human genome

The most potential influential mutational biases are a recombination association process called GC-biased gene conversion (gBGC). The gBGC results from a slight preference for G-C alleles in the mismatch repair machinery that has the potential to promote the maintenance of deleterious alleles (alternative form, expression). The action of gBGC is widespread across humans and across diverse ecosystems (Lachance & Tishkoff, 2014; Wood et al., 2014). The gBGC's genome-wide modeling has shown variation in intensity around the heritage of humans and chimpanzees, and among humans. The origin and effect of gBGC are closely linked to the recombination dynamics,

which differ greatly in the rate along the genome within the human population and between closely related species. The recombination patterns were affecting many genome evolution drivers including mutation rate, selection rate, and accumulation of de novo mutations (Coop & Przeworski, 2007; Torres, Szpiech, & Hernandez, 2018). We must develop high-resolution mutation rate maps for different populations, better models of how selection and recombination interact., and a deeper understanding of their impact on organismic performance in nature.

6.Functional impact of structural variation

The Indels and SVs make more variations in nucleotide between humans and chimpanzees than SNPs, and they have the genome restructured. Examples are the indels and SVs de novo levels of systemic changes observed in the human population including replication of variations and other rearrangements in genomics are more likely to have caused certain diseases. The calculated rate of de novo single nucleotide mutation was approximately 95 % per generation. SNPs are the ability to appears in different forms like as (AA**G**C-AA**T**C) difference as one nucleotide. These SNPs are in the form harmful, harmless and latent occur in the human genome. The effect of SNPs (silent) in the form directly and indirectly (Chuzhanova, Anassis, Ball, Krawczak, & Cooper, 2003; Montgomery et al., 2013; O'bleness, Searles, Varki, Gagneux, & Sikela, 2012; Stankiewicz & Lupski, 2010). For example, are the SNPs in cancer, in which DNA repairs, drug metabolize-enzymes are responsible for the metabolism/detoxification of carcinogenic acts as a cancer particular gene are involved? The potential significance in the genetic modeling and research relationship with SNPs of indels and SVs and new genes of phenotypically distinctive human and closely related species. Enough accurate maps of developmental events are possible in humans, and these data findings will help to establish new modeling approaches.

7.Conclusion

In human genome evolution, we understand the processes of evolutionary events occurs in human species and developmental advance programs in the human genome are encoded. The history of evolution is directly involved in our ability to treat the human genome fluctuation in basic and clinical science. In this study different research domains have highlighted, that have acts as a potential role in human genome evolution. We are also seeing the working of recent technical and

research advancement in analysis of genome evolution, human genome, diagnostic disease profiling, analysis of gene sequencing, functional genomics, single-cell evolution analysis, experimental omics analysis will alter our understanding of the human evolutionary process and study design. Ultimately, the human genome analysis in an evolutionary framework for the origins of human-specific biological systems.

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