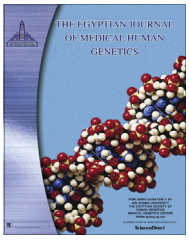
The Egyptian Journal of Medical Human Genetics (2014) **15**, 1–5

Ain Shams University

The Egyptian Journal of Medical Human Genetics

www.ejmhg.eg.net
www.sciencedirect.com



REVIEW

Biological evolution: Some genetic considerations

Mohammad Saad Zaghloul Salem

Faculty of Medicine, Ain-Shams University, Cairo, Egypt

Received 11 November 2013; accepted 15 November 2013
Available online 8 December 2013

KEYWORDS

Evolution;
Creation;
Natural selection;
Creative selection;
Biological adaptation;
Evolutionary diversity;
Biomolecules;
Human genome;
Human proteome;
Phylogenetic divergence

Abstract *Background:* The concept of biological evolution has long been accepted as a palatable theory aiming at explaining how life began and how creatures diverged so widely along the life span of the earth. Meticulous analysis and criticism of the different postulations of this concept, however, reveals that evolution is an illogic concept based on theoretical hypotheses that can never be tested. Creation, on the other hand, represents the other side of the coin, and up till now debates confronting creation versus evolution are still occupying much interest of atheist as well as of believer biologists.

Aim of this article: The motive for accepting the concept of evolution by most biologists, stems solely from their atheism and their saying that creation can neither be experimented nor validated, the same criticism directed against their assumptions regarding the basic aspects of evolution. This article, through analysis, criticism and reevaluation of some relevant genetic considerations that have long been traditionally considered as observations in support of the concept of evolution, viz. genetic memory and evolutionary variations, genomic adaptations to stress and evolution, comparative genomics and natural versus targeted selection, tries to elucidate and reveal some insensible assumptions embodied within the core ideas of evolution that stand in direct controversy with many well-known facts regarding the structure, function and behavior of living matter.

Conclusion: Natural selection might be observed in nature but not in life. The concept of biological evolution is an illogic and insensible hypothesis since it stands in direct contradiction with our current knowledge regarding the behavior as well as the structural and functional characteristics of the human genome and human proteome. Additionally, almost all basic postulations of this concept can neither be tested nor imitated for experimentation, which is a prerequisite for acceptance and validation of any scientific hypotheses.

E-mail addresses: mszsalem@hotmail.com, mszsalem@yahoo.com

Peer review under responsibility of Ain Shams University.

Production and hosting by Elsevier

1110-8630 © 2014 Production and hosting by Elsevier B.V. on behalf of Ain Shams University.
http://dx.doi.org/10.1016/j.ejmhg.2013.11.004

Contents

Introduction

The concept of biological evolution needs reevaluation as regards most of its basic assumptions. There is no logic in postulating that performing a particular physiological function in human cells necessitates more complicated genetic systems or more complex metabolic pathways than corresponding requirements for performing the same function in lower or any other species. Also, the presence of endogenous retroviral elements in the human genome and their roles, not only as a subset of transposable elements but also as regulators of gene expression, could be interpreted in an equally plausible different contrasting way. Acquired elements, following viral infection and insertional mutagenesis, might have been used by the host genome for its own benefit to perform the functions attributable to these elements. Obviously, this explanation seems to be more plausible than nonsense postulations attributing super capability and dominance of inserted viral genomes over infected human genome that enable it to induce evolutionary changes or directed adaptations in the human genome. Presence of similar, even identical, genetic sequences in different organisms irrespective of their phylogenetic positions, humans and drosophila for instance, need not be an indication of anything other than that similar biological functions in living organisms are expected to be regulated by similar genetic sequences and mediated by similar proteomic networks. Phylogenetic taxonomy of living organisms clearly discriminates between human beings, Homo sapiens, on one side and all other living organisms, viz. animals–plants–microorganisms including archaea, and reveals the uniqueness of humans as distinct single species compared to thousands of species comprising each of the three other kingdoms of living organisms. The persistence of this characteristic phylogenetic distinction between humans and all other creatures withstands as coherent theoretical and logical obstacle against the core concept of evolution, and points to the existence and persistence of the human genome as a unique bio-system all through human life on earth.

In fact, logical analysis of the concept of evolution reveals, in a very obvious manner, that evolution is an insensible idea. Postulations regarding final chance occurrence of self-assembly of biomolecules that started life activities, i.e. nucleic acids and proteins, after innumerable random reactions ignore the simple and firm fact that chance, by definition, has no memory. The Regular recurrence of biological phenomena in the same persistent, strict and repetitive pattern as seen in growth and development of living organisms, as well as in all their life activities, can never be attributed to chance and demolish these postulations. Regular recurrence of specific behavior patterns of biomolecules is a solid indication of their being pre-programed to behave in the same way under similar environmental effectors. Similarly, the construction of pre-programed structured systems, like the genome, the transcriptome and the proteome, that obey definite physical laws and behave in accord to strict regulatory principles applicable to solid and living matter, as well, nullifies allegations as regards the ability of biomolecules to evolve, depending on innate self-assembly, in a selective pathway away from these laws. If we accept the idea that deeply seated roots of evolution began by chance event, then stochastic behavior of components undergoing evolution would be the rule in view of the continuous and persistent external effectors and stimuli they are exposed to. The exquisite control of structures and functions of biomolecules, living matter and living organisms throughout their life span invalidates any significance to these postulations regarding origin of life and evolution.

Genetic memory and evolutionary variation

In spite of the exquisite ability of the protein translation system to recognize and decode the mRNA transcript and to harmonize the actions of tens to hundreds of factors involved in protein synthesis, it cannot recognize changes of the original genetic code embodied within the codon sequence of the mRNA transcript. It decodes and recognizes triplets of bases, or codons, along the transcript without giving attention to whether they are complementary to the original sequences of the gene or not. It seems that the protein translation system has no prior memory to predict the validity of the codon sequence of the mRNA transcript with respect to both the gene sequence and the amino acid sequence of the protein. Though this apparent defect might be considered a prerequisite for evolutionary variation of protein phenotypes necessary for acquisition of new functional abilities, e.g. formulation and construction of new metabolic pathways or acquisition of favorable selective advantages, since it allows for synthesis of different new proteins, it is a major cause of pathogenesis of genetic defects due to the absence of a translation proofreading and repair system comparable to those of DNA and mRNA repair systems.

Absence of genetic memory necessary for proofreading of translated proteins is enigmatic and bewildering in view of the prime and critical significance of the translation process, since the majority of genetic diseases result from synthesis of defective proteins or deficient synthesis of required proteins. Though it might be considered as a genomic regulatory mechanism allowing for selective pressure to proceed in favor of evolutionary variation, it results in marked pathological effects on

the organism if newly synthesized proteins are structurally defective. This obvious contradiction between potential, possibly, favorable effects and actual degrading consequences on the genome poses many fundamental queries as regards putative epigenomic regulatory mechanisms responsible for maintaining integrity and stability of the genome, both of which are pivotal conservative features mandatory for keeping species-specific genomic identity of the organism. On the other hand, it raises many inquiries as regards the actual significance and true purpose of evolution since one of the most plausible and considerable definitions of evolution entail improving potentials, performance, capabilities and persistence of biological systems and living organisms. Absence of proofreading mechanisms of translation of the proteome might, accidentally, allow for evolutionary changes to take place, but it mostly causes damage to the proteome with consequent deterioration of biological capabilities and survival fitness of living organisms.

Genomic adaptations to stress and evolution

Reconsidering the proteome as being a separate and independent biological system participating in defining life framework of living cells, not merely a tributary structured system synthesized under regulatory control of the genome, might help in interpreting some perplexing aspects of evolutionary adaptation. Duplication of the genetic material, either on gene level or chromosome level, is looked at as evolutionary solution to stress and evidence of their occurrence in yeast has been revealed in many studies [1]. Increased genome size in response to stressful conditions, nevertheless, should be induced first by deficiency in proteome functions because stressful environmental stimuli exert their unfavorable impact first on the metabolic networks in the cytoplasm or in other cellular components. They do not affect the genome in a straightforward way except under certain extraordinary conditions, e.g. direct damage to genes. Neither qualitative nor quantitative changes of the proteome have direct or lasting effects on constitution of the genome in a manner capable of compelling the genome to respond in an adaptive way leading to duplication of some of its components. Compensatory increase of gene product in response to stressful conditions can be attained via increasing gene expression by many known mechanisms, e.g. enhancing transcription through promoter activation, increasing stabilization of mRNA and multiple translation rounds of the same transcript, without the need to increase genome size by gene or chromosome duplication. Modifications of DNA-associated histones, for instance, have both enhancing and silencing regulatory effects on concerned genes but they do not result in, or induce, genetic duplications or increase in genome size.

The concept of biological evolution stands in direct contradiction with our current knowledge regarding the structural and functional characteristics of both the genome and proteome. Stressful environmental conditions, traditionally considered as the main triggers of evolutionary adaptations, affect the proteome of the cell first. Accordingly, if assumptions regarding genomic adaptations as evolutionary processes are postulated, they have to reveal first how primary disturbances in structure or function of the proteome can lead to adaptive changes in structure and function of the genome. Though the possibility of formulating novel, meaningful metabolically active networks composed of, and mediated by, new functioning proteins synthesized as a result of defective translation, is extremely difficult to accept or to interpret by the basic concepts of randomness and/or coincidence, its occurrence will be temporally limited by the life cycle of the cell. Unless adaptive genomic changes creating new genes coding for the new proteins occur, newly formulated networks cannot be maintained, fixed or inherited. Some sort of interactive feedback mechanism between the proteome and the genome, similar to classic stimulus–response pathways, might exist and represent the missing link mediating this proteome–genome interaction. However, these assumptions rely, basically, on the hypothesis of participation of the proteome, as a separate and independent regulatory structured system, in conducting life processes in the cell and in inducing adaptive and sustained changes of the genome.

Alternatively, supporters of the theoretical hypothesis which postulates that evolution and divergence could be initiated, mediated and maintained, primarily, by genomic adaptation in response to stressful or demanding environmental conditions have to reveal induced mutagenic beneficial effects capable of causing increases in genome size. Currently known mutagens, e.g. irradiation–chemicals–viruses, are damaging agents and result in detrimental effects and pathogenetic consequences leading, in most instances, to disease. The main support to this hypothesis comes from findings indicating that a significant part of the human genome, making up nearly 8% of its size, is composed of endogenous retrovirus elements and fragments [2].

The mere presence of endogenous viral sequences in the genomes of higher species including humans, however, cannot be considered neither as an indication of genomic evolution nor as a causative factor participating in its initiation or progression for many reasons:

- First: infection of germinal cells involved in reproduction by the virus genome, a prerequisite step for transmitting the viral genome to offspring and fixing the new host–virus genomic recombination, is a rare event.
- Second: efficient protective proofreading and anti-mutation mechanisms of the genome against unrecognized, or unregistered, sequences do exist and result in successful recognition of inserted strange viral genomes followed by their excision and deletion during genomic recombination stage via a specific mechanism known as recombinational deletion [3].
- Third: with one exception only, HERV-K (HML2) gene family, no human endogenous retrovirus elements capable of replication have been identified, all studied elements appear to be structurally defective due to major deletions and/or nonsense point mutations [4].
- Fourth: non-pathological insertional mutagenesis induced by viral sequences are expected to occur in intergenic, intronic or non-functional segments of the genome otherwise, they would result in decadence rather than evolution of the genome if they are inserted within functional sequences, e.g. exons, thus leading to their disruption. Complete physical mapping of all assumed endogenous retroviral elements is necessary to disclose this aspect of genomic adaptation if any roles in genomic evolution are attributed to these elements.

- Fifth: the many regulatory roles attributed to these elements in controlling some of the most critical aspects of life activities of infected host genome, e.g. reproduction and immune competence, poses many questions regarding integrity and stability of the host genome before the acquisition of these elements. It is hard to accept the idea that a complex genome, like the human genome, would be pawned and dependent on external processes, like accidental insertion of endogenous retrovirus elements, to maintain its integrity, preservation and continuation through reproduction.

Comparative genomics

The human genome is unique in being characteristic of one species only: Homo sapiens or humans. All human beings irrespective of their ethnic background have the same genome, albeit with minor differences. Conversely, families, genera and species of animals, plants and microbes have different genomes characteristic of each species and shared, in many instances, by other subspecies of the same genus. The presence of innumerable numbers of genetically distinct living species, other than humans, without any genetic evidence of changes attributable to evolutionary adaptation should compel us to reconsider the unjustified tendency for applying results of research and experimentation on these species to humans. What might be considered as evidence of evolutionary adaptations, e.g. development of novel metabolic circuits by some microorganisms in response to environmental effects, might be due to activation of already existing, still undefined and unrecognized, genes or other functional components of the genome responsible for mediating these novel functions under the influence of the new environmental conditions. The frequent delineation of new adaptive metabolic networks without defining new genes responsible for their establishment stands in favor of this interpretation. Obviously, unless new genes responsible for synthesis of new proteins mediating these novel networks are mapped, defined and characterized for sure, these metabolic adaptations could never be considered within the context of evolution.

Although the development and appearance of new biological functions secondary to environmental effectors or endogenous mutational events reflects the concept of adaptation, a fundamental and universal biological principle shared by living organisms and indispensable for any biological systems existing in a continuously varying and stressful environment, adaptation needs not be considered as modifications secondary to genomic alterations. As referred to previously, the concept of evolution is hard to consider unless solid evidence of progressive and appreciable increase in proteome size accompanied by parallel increase in genome size from simple to more complex biological systems, e.g. from unicellular to multicellular organisms, is documented. Actually, this is not the case as many simple organisms have larger genomes than more complex organisms. This might be interpreted by more liability of simple organisms, which have simple cellular architecture and deficient protective mechanisms, to infection by invading viruses capable of amalgamating their DNA with host DNA with a consequent increase of the size of host genome, compared to more complex organisms that have protective and anti-mutation mechanisms against infecting viruses.

Natural versus directed selection

The concept of natural selection might be applicable in nature but not in life. The continuing occurrence and persistence of genetic defects deleteriously affecting biological fitness, as measured by survival and reproduction, among humans contradicts assumed roles attributed to natural selection as major effectors of adaptation and evolution. Mutations that cause these genetic defects occur spontaneously at a more or less constant rate among most populations. Eradication or lessening their harmful burden can be achieved only by targeted, rather than by natural, selection, e.g. through pre-implantation diagnosis, prenatal detection and termination of pregnancy and other similar prophylactic genetic measures. In the animal kingdom, for instance, there is no chance for survival or reproduction of sick or weak animals, similarly in the plant kingdom, plants incapable of tolerating conditions of dehydration due to accidental lack of water would perish quickly, but this is not the case in human life where deeply rooted spiritual considerations rule and control the way we manipulate and control our genomes, not the reverse. There is no place for the concept of natural selection in human life or in human genetics. Instead of that, the concept of creative or artificial selection, e.g. cloning and trials to construct new creatures with new genomes, in spite of moral protestation and ethical expostulation against its applications, will probably accomplish the major role in inducing true evolutionary changes of the human genome through targeted reproductive mechanisms like pre-marital, pre-implantation and prenatal diagnosis.

Assumptions based on the ability of solid matter and biomolecules, nucleic acids and proteins for instance, to decide their own destinations are quite irrational. They are more close to science fiction imagination than to scientific thinking. The concept of evolution comprises within the details of its wide spectrum of some of these illogic ideas. Conferring discernment that enable biomolecules to think, choose and behave like rational creatures, whether motivated by its own benefit or in response to external stresses, deserves no attention and ideas attributing biological diversities of organisms to perceptive behavior of their constituting biomolecules can never be considered seriously. Similarly, trends aiming at imposing the concepts of biological evolution, irrespective of its discrepancies, in most, if not all, aspects of biology cannot be accepted unless their validity is proved beyond doubt. Neither experimentation nor logically interpretable observations are in support of these concepts. Ideas postulating that events that lead to adaptation–natural selection and evolution of simple organisms to more complex creatures at some point of their phylogenetic divergence in response to internal or external effectors, are responsible for most of the biological diversities of present era's living creatures, including humans, can neither be tested nor simulated for validation since supporters of these ideas date the timing of these events back to a few billion years ago. Unfortunately, no human beings, including evolutionary scientists, can live for such a long time to reveal the truth in this regard.

The human genome, like genomes of other living creatures, is programed in a very strict way to behave in a very defined manner under specific recurring circumstances, for example replication during cell division and differential mass suppression and activation of many of its components during embryogenesis. Success in discovering and revealing underlying

epigenomic, rather than epigenetic, factors and mechanisms responsible for regulating and maintaining the three fundamental aspects of the genome, viz. integrity–stability–identity, would, surely, have revolutionary impact on all aspects of human genetics and in particular medical genetics. However, the benefits of such an achievement are conditioned by their applications in different fields of medical genetics, otherwise they will probably lead researches to nothing except getting things back to old futilitarian nonsense discussions concerning adaptation and evolution. Within the context of medical genetics, researches aiming at revealing how things happen, in order to direct their happening for the welfare of patients, are much more important and cost-effective than wasting efforts, time and money in research studies trying to know why they happen.

Conflict of interest

The author declares that there is no conflict of interest.

References

[1] Yonaa Avihu, Manor Yair, Herbs Rebecca, Romano Gal, Mitchell Amir, Kupiec Martin, et al. Chromosomal duplication is a transient evolutionary solution to stress. Proc Natl Acad Sci 2012;109(51):21010–5.

[2] Belshaw R, Pereira V, Katzourakis A, Talbot G, Paces J, Burt A, et al. Long-term reinfection of the human genome by endogenous retroviruses. Proc Natl Acad Sci 2004;101(14):4894–9.

[3] Belshaw Robert, Watson Jason, Katzourakis Aris, Howe Alexis, Woolven-Allen John, Burt Austin, et al. Rate of recombinational deletion among human endogenous retroviruses. J Virol 2007;81(17):9437–42.

[4] Ruggieri Alessia, Maldener Esther, Sauter Marlies, Mueller-Lantzsch Nikolaus, Meese Eckart, Fackler Oliver, et al. Human endogenous retrovirus HERV-K (HML-2) encodes a stable signal peptide with biological properties distinct from Rec. Retrovirology 2009;6:17.

Other sources

[5] Pierce givennameBenjamin. Genetics. A conceptual approach. 2nd ed. Gordonsville, VA, USA: Freeman & company; 2005.

[6] Dale Jeremy givennameW, Park Simon givennameF. Molecular genetics of bacteria. 4th ed. John Wiley & Sons Ltd; 2004.

[7] Anthony givennameGriffiths JF et al. Introduction to genetic analysis. 9th ed. Gordonsville, VA, USA: W.H. Freeman & company; 2007.

[8] Pasternak givennameJJ. An introduction to human molecular genetics. second ed. Hoboken, NJ, USA: Wiley-Liss, John Wiley & Sons, Inc; 2005.

[9] Rédei givennameGP. Encyclopedia of genetics, genomics, proteomics and informatics. third ed. New York, USA: Springer; 2008.

[10] James givennameWatson, Baker givennameTania, Bell givennameStephen, Gann givennameAlexander, Levine givennameMichael, Losick givennameRichard. Molecular biology of the gene. fifth ed. Menlo Park, California, USA: Benjamin Cummings; 2004.