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Genetic Mutations and DNA Repair

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ABSTRACT: Reactive species within cells and external agents constantly attack cellular DNA. Different repair pathways reduce toxic and mutagenic effects, and this article describes 130 known human DNA repair genes. There is currently only one system to remove the primary DNA lesions caused by ultraviolet light, despite notable features such as four enzymes that can remove uracil from DNA, seven recombination genes linked to RAD51, and numerous recently identified DNA polymerases that avoid damage. By comparing with model organisms and identifying common folds in three-dimensional protein structures, more human DNA repair genes will be discovered. Clinical applications such as better radiotherapy, anticancer medication treatment, and a deeper comprehension of the forces that cause cellular ageing should result from modification to DNA repair.

Keywords: genetic factors, DNA repair, mutation, and crispr

1. INTRODUCTION

Any alteration to the quantity or arrangement of nucleotides in DNA that produces new nucleotide sequences whose effects are transferred to offspring with particular characteristics is known as a genetic mutation. The smallest genetic unit that can result in a genetic mutation is called a muton, which is the smallest number of mobile nucleotides that can cause phenotypic changes [1]. A muton can be as small as a nucleotide. The majority of mutations alter the number of chromosomes or the structure of a single chromosome. These changes may occur spontaneously or may be triggered by radiation, chemicals, or mutagens. A gene may become inactive and stop working for a specific process, such as producing an enzyme or hormone, or it may produce less or more of the gene [2]. This could be detrimental if a change at the gene level results in a change in the gene's image, i.e., a change to a different state. The study aims to identify genetic mutations, their sources, types, effects, and pathways of transmission to the gene, in addition to assessing the likelihood of mutation occurrence and its prerequisites.

2. EXPLAINING THE METHODS OF REPAIRING MUTATIONS THROUGH DNA

Repairing damaged DNA cells at the molecular level, and protecting genetic information. DNA is an unstable molecule that slowly degrades over time and can be damaged during daily activity. For life to continue, there must be self-repair mechanisms to carry out this process. Failure to repair defects in genetic material leads to the possibility of a person developing cancer, neurological disorders, immune deficiency, and premature aging [3]. The enzyme topoisomerase contributes to modifying the topology of DNA," Al-Khamisi told Al-Ilm. "The team focused on

understanding the mechanism by which cells repair defects in genetic material. If this process does not occur properly and quickly, it may lead to diseases, most notably senile dementia and atrophy of the nervous system [4]. It may also cause cancer. He added that most of the studies conducted before were done on cells grown in the laboratory, but the team was able for the first time to perform this process in a complete living organism [5]. We had two choices to conduct our research: either laboratory mice or zebrafish, but the team chose the latter. Al-Khamisi explained that it has several advantages, most notably that it is 99% similar to humans in its genetic preservation mechanism. Furthermore, the genes responsible for human disease are fully present in zebrafish. Furthermore, these fish are transparent at a young age, so you can easily observe the changes that occur [3].

3. MUTAGENIC FACTORS

Mutagens are numerous chemicals capable of causing genetic mutations or chromosomal changes. In recent years, dozens of these substances have been discovered to induce chromosomal changes when exposed to cells or tissues at specific concentrations over a specific period of time. These chemicals, such as mustard gas, nitrous acid (HNO2), hydroxylamine, and alkylating agents, interact with specific regions of the genetic material within the chromosome, altering its genetic structure. Their effects are more dangerous than ionizing radiation, causing qualitative and quantitative changes in the genetic material that can lead to genetic mutations. This is due to their ability to penetrate the drug and interact with the genetic material [6].

4. CRISPR TECHNOLOGY

Regarding the technology used in the study, Al-Khamisi explained that the team used CRISPR technology, a laboratory tool for modifying DNA to repair genetic defects or improve certain traits [7]. Al-Khamisi spoke about the causes of DNA deficiencies, attributing them to chemical reactions that occur naturally within the human body, such as metabolic reactions during the conversion of food into energy in the body, and the resulting free radicals that attack DNA. He stressed that this is the largest source of genetic deficiencies, in addition to genetic diseases, as some newborns suffer from changes in certain proteins [8]. He added that there are other reasons related to people's behavior, including excessive exposure to ultraviolet (UV) light, pollutants in the air, such as smoke, in addition to unhealthy eating habits such as eating overcooked meat, and eating a lot of foods that contain preservatives and color stabilizers [9]. Regarding the steps Al-Khamisi pointed the team is currently conducting experiments on young zebrafish to test a group of food-derived compounds to monitor their effects on improving DNA repair in young individuals. This could be a prelude to the possibility of developing effective treatments to delay and reduce the severity of age-related diseases [10]. Regarding the effectiveness of CRISPR technology in treating diseases in humans, he noted that the team is currently working on other projects related to gene therapy, and has achieved impressive results. He also noted that another research team has achieved significant results in treating spinal muscular atrophy (ALS) using gene therapy, with a success rate exceeding 85%. This is the disease that afflicted Al-Ahly and Egypt national team player, Moamen Zakaria, and which also afflicts children [11].

5. MUTATIONS

In genetics, a mutation is a mistake in gene copying that happens during a cell's self-replication. In other words, it describes any sudden change that affects the genetic material. Scientists have been interested in studying mutations because of their importance in medicine, as they can lead to diseases like cancer and others[3]. Chromosome mutations are mutations that can happen at the chromosome level. These mutations can result in a change in the number of chromosomes in an organism or in chromosomal mutations, which are changes to a single chromosome. By altering a pair of nucleotides or their location on the strand, mutations can happen at the DNA level. We refer to this kind of mutation as genetic or point mutation. A number of genes in E. coli can lead to excessive mutations in the entire genetic population if they are exposed to mutations. Point mutations are the term used to describe these genes [12].

6. CAUSES OF MUTATION

The genetic code, which is composed of tiny chemical components arranged side by side, like the letters of the Arabic language, is the language in which the instructions for creating a protein from a gene are written. These ordered molecules are known as nucleic acids, and the arrangement of these nucleic acids determines how different proteins are from one another. Therefore, any flaw in this configuration results in a mutation, which is a flaw in the protein's formation [8].

Thus, a genetic mutation is defined as a flaw in the nucleic acid arrangement of a gene (i.e., a flaw in the arrangement). A mutation can affect all of the body's cells or just one of them. When it is found in every cell, it indicates that it happened early in life, when our body's cell count was low [13].

Since every cell in our body is cloned from a single cell (the egg fertilised by the sperm), the mutation may also have existed in the egg or sperm from which we were created. Our cells may therefore acquire new mutations that weren't present in our parents' cells, and we may inherit a mutation from our biological parents because they happened

after our conception or birth. Consequently, mutations may or may not be passed on by one of the parents. Even though an organism may have many gene mutations, these do not necessarily lead to health issues [14].

If all mutations were bad, we would have suffered from diseases from the moment our mothers gave birth to us. A mutation may be harmful or harmless. If a mutation follows these two guidelines, it is not dangerous:

The first rule states that a mutation shouldn't occur inside a gene but rather outside of its boundaries, that is, next to the gene in the gap between genes.

The second rule, that a mutation is usually present if it affects only one copy of the gene (for instance, the copy you inherited from your father) and not the other copy you inherited from your mother, has a few exceptions. [5].

For instance, a protein called frax protein is required by the cell in a specific quantity (111 units), and the healthy gene is unable to produce the hundred units on its own because it can only produce 51 units. A deficient quantity occurs inside the cell, which is where the disease occurs, when the amount produced by the healthy gene is insufficient to fill the void created by boxes in the second gene. Instead of transferring the quantity, the mutation may have the opposite effect on the gene. In contrast to what is permitted inside the cell, the opposite occurred, and the amount produced by the gene impacted by the mutation increased. It goes without saying that this increase harms the cell and causes [15].

7. MUTATION EFFECT

There are the same number of genes in every cell in the body. Does this imply that illness in every organ results from a faulty gene present in every cell in the body? Not always, the answer is no. The existence of this mutation has no effect whatsoever on a cell that does not require the protein that this gene produces because it does not require this protein.

For instance, suppose a man has a gene that is damaged. His eye could get sick because this gene is especially crucial for eye cells. However, since the other organs do not require the substance produced by this damaged gene, it is not a given that they will be impacted by this damage. On the other hand, a gene may occasionally be crucial for multiple organs rather than just one. This material may be crucial for the brain, heart, and eyes, which causes the cells in these organs to deteriorate and become diseased, resulting in illnesses in all three [16].

As a result, some people may have a genetic disease in some organs due to a mutation (damage) in a single gene, which can lead to multiple issues in various body organs that may not initially appear to be connected. For instance, in hereditary leukaemia, the eyes are blue, the skin is extremely white, and the hair is light in colour. It is brought on by a lack of the red protein, which is necessary for the skin to produce pigment but is also vital for the eyes and hair. For instance, the disease has no effect on the liver or heart because these organs do not require this protein. even though this faulty gene is present in every liver and heart cell [10].

8. TYPES OF MUTATIONS

Mutations that occur in DNA can be classified according to the type of damage they cause to the DNA.

• Substitution mutation: (missense or substitution mutation) These mutations occur by replacing a specific nucleotide from the strand, which leads to a change in the nucleotide of the transcribed mRNA. It may lead to a change in one amino acid. The amino acids resulting from the process of assembling them before forming the protein, which changes the properties of this protein. This picture is a comparison between healthy DNA and DNA in which a substitution mutation occurred [17] .

• Frameshift mutation: This mutation occurs by deleting or adding a nucleotide from DNA, which leads to a change in the amino acids from the missing or added nucleotide to the end of the copied strand. The harm of this type of mutation lies in the fact that it causes a continuous change in the code, the transcribed RNA... As you know, the RNA code is divided in the form of RNA is divided in the form of triplets, each triplet is concerned with a specific amino acid. When a nucleotide is added or deleted, they will all recognize each other to produce a completely different protein. This sentence is an example to clarify.Our sentence THEBIGCATATETHERAT: After dividing it into threes, it becomes as follows: THE BIG CAT ATE THE RAT. By deleting a letter, the sentence becomes THEIGCATA TETHERAT. After dividing it into threes, it becomes as follows: THB IGCC ATA TET HER AT. Thus we see that deleting one letter affects the formation of the rest of the words... and this is what happens in the bar when deleting or adding ne, kle, and ted [18].

• Neutral Mutation: Here in this case we see that a change occurred in the DNA chain resulting in the replacement of glycosylated acid with alanine acid.... The change will not produce major effects as happened in the previous case because the alanine acid does not differ chemically enough to cause a major difference in the resulting protein.. Both acids are non-medical and this is what makes this mutation ineffective. When scientists established this type of mutation they were astonished because the possibility of its occurrence was higher than they expected and it is one of the most common mutations [19].

• Silent mutations: This type of mutation does not affect the amino acid sequence at all. When a nucleotide is changed, it produces another triplet code that produces the same amino acid. Therefore, this type is called a silent mutation.

• onsense mutations: This type of mutation is a change in the DNA sequence that results in a stop codon when the mRNA is copied. In this case, we have a comb protein that has no function. There are other cases in which mutations occur, such as duplication mutations, which occur during the replication of one of the gene segments, resulting in an additional copy of one of the genes. This type of mutation has characteristics that make it useful because:Over time, one of these mutations could be the basis for the emergence of a new, distinct function and thus the basis for natural selection.When two genes remain in parallel between the same genes and the same function, this remains a reserve of changes. This explains the principle of the dominant gene and the draft gene, where the draft gene is on the parallel gene [20].

9. THE POSSIBILITY OF MUTATION AND ITS HARMS

A mutation must occur in the genetic material involved in the sexual process of reproduction, so that it can be passed on to subsequent generations and remain in the gene pool. In contrast to natural selection, in which reduces the amount of the diversity in the gene pool, mutations increase diversity by introducing new genes into the gene pool. The process of DNA replication is highly accurate, and errors in replication range from one error in a billion, to one in a billion letters. However, the probability of a gene being mutated does not exceed 1111111 to 1111111. Since humans have 31111 genes, it is expected that at least one mutation will affect each of them, which makes mutation a common natural phenomenon according to what is known today. Large parts of the DNA strand are genetically "idle", meaning that they are copied from one genome to another, but they are not "active" [21].

A large proportion of mutations occur in the functional parts of the genetic material that do not contain active genes. Therefore, most mutations are considered "neutral" with respect to natural selection, meaning that they neither increase nor decrease the organism's chances of survival, nor do they increase the likelihood of the emergence of new traits. The number of mutations is the primary determinant of the rate of evolution, because mutations are what introduce diversity into the gene pool. However, in the short term, the gene pool can evolve relatively quickly from mutations "stored" in the inactive genetic material. However, the path for a mutation to become a vector for natural selection is still long [22] .

Genetic mutations and their effects on reproduction and natural selection:

• Many mutations occur in the idle parts of the genetic material in organisms that reproduce by mating. Half of the genetic material comes from one of the spouses, and thus the mutation may remain in the part that was not used.

• Many hereditary traits consist of a pair of traits, one dominant and the other weak. The mutation is often in the weak trait. Therefore, the mutation is not activated during the organism's lifetime in this generation. The mutation remains complete until a sufficient number of individuals carry the weak trait before individuals carrying the recessive trait are formed.

• Most mutations that appear in the form of active genes lead to errors in the functioning of the genetic material (diseases or genetic abnormalities), and thus individuals who carry these traits are eliminated through natural selection, and they die at an early age without passing on the mutation to future generations [3].

10. CAUSES OF MUTATIONS

A virus, radiation, or chemical exposure can all cause errors in the cell division process. When one of the many cells in a multicellular organism is cloned, a mutation may result. This indicates that a greater number of cells raises the possibility of mutation, which could impair an organism's ability to function and result in illness or death. In any event, it becomes a part of the pool of total genetic alterations. The theory of punctuated equilibria, which was misrepresented in Darwin's theory, includes neutral mutations. Its main ideas can be summed up in the presentation that follows: In the future, when changes occur that test the organism, mutations that are retained in the entire mutation pool may be used [3].

11. 11. DNA repair

The DNA in the cell is subject to many chemical changes, and in order for the mutations encoded in the DNA strand to remain effective, these chemical changes must be corrected, and any failure in this correction leads to the appearance of the mutation. The latest publication of the human genome showed the presence of 131 genes whose products contribute to the process of DNA repair inside the cell [21].

First: Factors that lead to DNA damage

Radiation with a specific wavelength such as ionizing radiation (gamma rays, x-rays, UV-C rays with a wavelength of -260 nm) that strongly penetrates DNA, and also the longer wavelength UV-B rays that also affect the ozone layer. A highly chemically active oxygen radical produced by biological processes. Intracellular Chemicals present in the environment, such as some hydrocarbons, such as those found in cigarettes, and Some microbial or plant products, such as aflatoxins. Some chemicals used to treat diseases such as cancer [19].

Second: Types of DNA damage

• Damage to the four bases in DNA (A, T, C, G) can be mutated or covalently placed in different places, or a mismatch can occur in the binding of opposite dinucleotides, and the most common is: The most common cases are the exchange of an amino group (i.e., the process of (“deamination”) that leads to changing a nucleotide to 1,

for example. The incompatibility between the dinucleotides can be due to the failure of interpreting and reading the DNA during manufacturing. The most common example: the incorporation of the pyrimidine U, which is found naturally in RNA, instead of C from [23].

• Base strand breakage: It can be limited to a break in one strand of DNA, called a single-stranded break (SS), or it can break both strands, called a double-stranded break (DSB). It can be caused by radioactivity, and ionic products do it better.

• Crosslinks: that can form between bases. It may occur between the bases of one strand, or in the opposite bases of both strands. It is caused by chemical drugs [24].

The most common cause of DNA damage and mutations in humans is the spontaneous addition of a methyl group (CH3-) as an example of alkylation (removal of an alkyl group). Fortunately, most of these changes are repaired by enzymes called glycosylases. When a mismatch occurs as a result of the exchange of C to T following alkylation, these enzymes repair the resulting mismatch by returning the C to C. This is done without the need to break the DNA and break the strands. Some drugs used to treat cancer (chemo) cause DNA damage through alkylation. Some alkyl groups can be removed by a protein produced by the MGMT gene. This protein is capable of performing this process only once, so removing all groups requires additional protein molecules. Direct reversal mechanisms eliminate some problems, the most important of which is that these mechanisms are considered very promising!!! The changes that occur in DNA are countless, and each individual change requires a specialized mechanism. Therefore, the cell needs more general mechanisms capable of correcting all types of chemical damage with several Specific work [25].

12. Mechanisms for repairing damaged parts

The missing or incompatible bases can be repaired in several ways: direct chemical reversal (the opposite of the reaction that led to them). Repair of damaged segments: the damaged base or bases are removed and replaced with a correct base in the place of the defect that occurred in the DNA. There are 3 methods for repairing damaged parts, and each method requires a specific set of enzymes Base Excision Repair (BER), Nucleotide Excision Repair (NER), Mismatch Repair (MMR), Breaks SSingle-Single SSBs [26].

• Base Excision Repair (BER): The process can be summarized as follows:

Removal of the damaged base occurs approximately 21,111 times in our body cells daily (via the enzyme). Leaving a gap we have two genes responsible for coding the ribosomes for this function. This process is carried out by a different DNA enzyme, and each enzyme is responsible for removing a specific error from the damage that affects the bases. Then the same enzyme removes the ribose and phosphate related to this base from the DNA strand. Glycosylase. We have at least 8 genes that encode DNA glycosylase. AP endonuclease in E. coli bacteria. The term “AP” refers to the infected site. Coded in our genes are polymerases, one of the 11 beta DNA polymerases. DNA polymerases re-sequence the correct nucleotide. This process depends on DNA. Then the break is re-packaged into the DNA chain. There are two enzymes that can do this process in E.coli bacteria have DNA polymerase I and DNA ligase, both of which require ATP to provide the necessary energy [27].

• Nucleotide Excision Repair (NER): NER differs from the first method in several points: it uses different enzymes. If only one base is damaged, this process removes a large area around it. The damaged area, even if the surrounding dinucleotides are intact, is removed. This method removes a large patch around the damage. This method include recognized the damage by a group of protein factors (or just one factor)....and all these factors gather around the affected area. DNA is unwound and disintegrated in the laboratory and the group of enzymes that do this work are: Transcription factors: IIH, TFIIH, which also perform a function in the production of Disintegrations and cuts occur at the ends of the damaged area (3 and 5)... and thus DNAJ The area containing the sparks can be removed and the secrets are repaired by replacing the nucleotides with the enzyme delta polymerase epsilon. DNA ligase rejoins DNA fragments by covalently joining them at each strand. In E.Coli bacteria, the proteins UvrA, UvrB, and Uvr are responsible for removing the labeled dinucleotides and their surroundings. This is done with the help of DNA polymerase ligase1 [28]. In yeast, the proteins are similar to the r-radiation proteins. Examples of diseases that occur due to failure of this process(Xeroderma Pigmentosum XP), this disease is a genetic disease that affects humans. It is rare. This disease causes skin problems such as wounds and spots with colors ranging from brown to red and sometimes pink when exposed to the sun. It is similar to a widespread case of skin cancer. This disease is caused by a mutation in a few genes, all of which play a role in nucleotide. Which senses the protein that binds to the damaged sites and helps assemble another protein that plays a role in the same repair process: XPA of which is Excision Repair XPD and XPB and some mutations in TFIIH are part of XPD and XPB. Causes premature aging Which cuts the chain from the 5th side of the image, XPA Who cuts the chain from the end of 3 damage, XPG [29].

• Mismutch Repair MMR

This repair occurs in structurally sound nucleotides that suffer from pairing errors, which contradicts what Watson and Crick established regarding nucleotide pairing. Also, the use of other enzymes that were not involved in the two processes, specialized for these (NER) and (BER). This method may use enzymes used in the two previous processes [30].

The function of recognizing the location of the mismatch requires some proteins, including a protein encoded in the MSH2 gene. Removing the mismatch requires other different proteins, including the protein encoded in the MLHI gene. The deficiency of the MLHI and MSH2 genes leads to hereditary colon cancer, as these two genes are considered tumor suppressors. The specific mechanism for recognizing mismatches in human DNA has not yet been discovered. However, in the bacteria Ecoli, recognition is achieved by a methylase (a demethylase) called "Dam methylase", which can methylate all adenine that falls within the 5' GATC chain. After DNA is synthesized, the copied strand is methylated [31]. The copied (i.e., the new) strand is not methylated, so a difference can occur between the original and the copied strand The manufacturing process begins with the Muts protein, which binds to the incompatible dimers. Then the proteins Helicase and SSB protein, with the help of excmucleuse, then the cleaved part of the mismatched chain is removed, activating the enzyme Abdus Salamiyah DNA from which the GATC was copied, resulting in the cleavage of the MutH chain to perform the same task. Activation of MutkH binds this complex and activates the work of another protein, MutL.If the cleavage occurs at the 13′ end of the mismatch, this step will activate the exonuclease enzyme, which lowers only the single strand on the 3′ side towards the 5′ side. If the crack occurs at the 5th end of the mismatch, the process occurs due to the action of the zym. The point of mismatch may exceed 1111 base pairs, so the repair of the mismatch is considered ineffective. GATC fills the gap left by the previous steps. The distance between the end of the DNA ligase and polymerase III is done by the enzymes Recl or exonuclease VII [32].

• Fractions in a single series: Breaks SSingle-Single SSBs

These breaks are repaired using the same enzymes used in the BER system. Double Strand DSBs There are two mechanisms by which the cell attempts to repair complete breaks in the DNA molecule, called kku, to recognize the broken regions and bind to the broken ends and join them together again. This will happen better if complementary nucleotides are present, but it can happen without them. This type of joining is also called... Non-homologous End-Joining (NHEJ) meaning non-homologous end joining. A link error may cause translation errors, which can lead to a lot of problems [33].

13. Conclusions

• Genetic mutations may be inherited or acquired.
• Mutations can decrease or abnormally increase gene function.
• Only mutations in germ cells are heritable.
• Most mutations disrupt genetic function and cause disease.
• Natural selection removes harmful mutations.
• Mutations may result from genetic errors or environmental factors.

Recommendations:

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CONFLICTS OF INTEREST

The authors declare no conflict of interest

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