*British Journal of Applied Science & Technology*

*21(5): 1-10, 2017; Article no.BJAST.32463*
*ISSN: 2231-0843, NLM ID: 101664541*

# A Survey of Human Deoxyribonucleic Acid

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***Authors' contributions***

*This work was carried out in collaboration between authors HKB and KAG. The two authors read and approved the final manuscript.*

***Article Information***

DOI: 10.9734/BJAST/2017/32463
Editor(s):
(1) Ya-mei Gao, College of life science and technology,Heilongjiang Bayi Agariculture University, Daqing, Heilongjiang, China.
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(5) Elif Funda Sener, Erciyes University, Turkey.
Complete Peer review History: http://www.sciencedomain.org/review-history/19499

***Review Article***

***Received 27th February 2017***
***Accepted 16th May 2017***
***Published 13th June 2017***

## ABSTRACT

Deoxyribonucleic Acid (DNA) is a molecule that carries most of the genetic instructions used in the development, functioning, and reproduction of all living organisms. The *genes* in living organisms contain instructions responsible for their characteristics which are transferred from parent to offspring to structure his life. These instructions are encoded in DNA molecule. This Paper presents a survey on DNA molecules in passing instructions from parents to their offspring from one *gene*ration to the other.

*Keywords: DNA; gene; genetic; offspring; parents; living organism; generation.*

## 1. INTRODUCTION

Deoxyribonucleic Acid (DNA) is the primary genetic and hereditary material in all living organisms. It is a molecule composed of two complementary strands wrapped up in a double helix formation which is responsible for building, reproduction, and maintenance of life. It is the genetic code of life on Earth that contains the information for how organisms are structured, how they function and even how they behave [1]. It is also the genetic code in the body that

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transfers genetic message to the entire cells in the body. It is responsible for individual differences and habits. Thus, the scenarios of our lives conform to the design in our DNA. The answer to the question why goat gives birth to goat, lion to lion, man to man and so on is the DNA which contains the biological instructions and stores biological information [2] that makes each species unique. Genetic information is inherited by organisms through their parents.

DNA is made up of chemical building blocks called nucleotides [3] which contain nitrogen base adenine (A), thymine (T), cytosine (C) and guanine (G). The order of these nucleotides is called DNA sequence. Nucleotides can contain either a purine or a pyrimidine base. The purine bases are adenine (A) and guanine (G), while the pyrimidines are thymine (T) and cytosine (C) [4].

The genome of Human comprises of three billion nucleotides of which 99.9% are identical from one person to the other. The residual 0.1% allows us to differentiate one person to the other, in terms of appearance, character, etc. By this, Humans share 99.9% of their DNA, the parts that are different are a kind of “fingerprint” that is unique for everyone [5]. It contains the instruction for an organism to develop, survive and reproduce [6].

DNA is the blueprint of biological life from beginning to the end [7]. The 26 alphabets in the English language are used in the construction and expression during communication. Also, in a similar manner, the four nucleotides of DNA form a sequence of genetic instructions which direct the activity of the cells in human. The order of the sequence of these nucleotides determines the information available for the organism. Hundreds of nucleotides may link together in a DNA chain to form a sequence that spells out instruction for a single *gene*. e.g. the order ATCGTT might represent the instruction for blue and ATCGCT might be instruction for brown. DNA base pairs up together with each other to form a unit called base pair. The main purpose of DNA is to serve as a storage medium for hereditary information.

The rest of the article is organized as follows: Section 2 explains the literature review, section 3 discusses DNA and Chromosome, section 4 discusses the biomolecules of life, section 5 discusses the genetic code and mutation and finally, section 6 presents the conclusion of the article.

## 2. LITERATURE REVIEW

### 2.1 Discovery of DNA

For several years, scientists could not identify the exact molecule responsible for heredity in a living organism. In 1869 Friedrich Miescher [8] first discovered DNA, Scientists of that time believed that protein was the molecule that carries life biological instructions and DNA was too simple molecule to play such role [9]. He discovered DNA when he was studying white blood cells, then isolated molecules found to have traces of acid with the high percent of phosphorous and named this molecule ‘nuclein’ which is known as ‘nucleic acid’ and later called Deoxyribonucleic Acid’. Miescher did not attach much importance to his discovery because he assumed that it lacks all essential components. He believed like other Scientists of his generation that it was protein responsible for heredity.

### 2.2 Linking DNA to Heredity

Scientists believed for decades that protein was responsible for hereditary information, until 1944 when a biologist, Oswald Avery performed an experiment with bacteria that caused pneumonia. At that time, Scientists believed that some bacteria (called S-type) had capsules in their outer layer, but other types (called R-type) did not. By several experiments carried out, Avery and his colleagues found that only DNA could change R-type bacteria to S-type bacteria. This means the DNA allowed it to carry instruction from one cell to another which no other substance can perform, even the protein. This, therefore, identified DNA as the only substance responsible for heredity.

### 2.3 Confirmation of the Finding

Avery discovery was not popularly accepted by his fellow researchers until an independent and neutral experiment was carried out by other Scientists. In 1952, Alfred Hershey and Martha Chase carried out an experiment with bacteriophages [9], [10] (these are the viruses that infect bacteria) and confirmed that protein was not a hereditary material. Bacteriophages comprise of two substances (i) DNA and (ii) Protein. Radioactive label was used to integrate into either protein or DNA. The outcome of this experiment by Hershey and Chase confirmed that the virus infection found in the bacteria is caused by DNA transferred from bacteriophages. Additional research showed that DNA in a virus

can take over a bacteria cell, causing it to replicate the virus DNA and create a new virus.

Hershey and Chase presented experiments that clearly suggested and accepted that DNA controls the production of more DNA; DNA was itself the substance that directs the constructions and functions of living things. James Watson and Francis then put together how DNA carries hereditary information from one cell to another cell. In 1953 James Watson, Francis Crick, Maurice Wilkins and Rosalind Franklin described DNA as double helix shape which can carry biological information [11,12].

Watson, Crick, and Wilkins got Nobel Prize in Medicine in 1962 for their various discoveries on the molecular structure of nucleic acid and its significance to information transfer in the living material [6].

## 3. DNA AND CHROMOSOME

Deoxyribonucleic Acid carries genetic information which is transferred from one generation to another DNA is available in all cells in our body except the red blood cell, it contains information about our heritage [7].

A chromosome is the tight package of DNA into thread-like structures (Fig. 1).

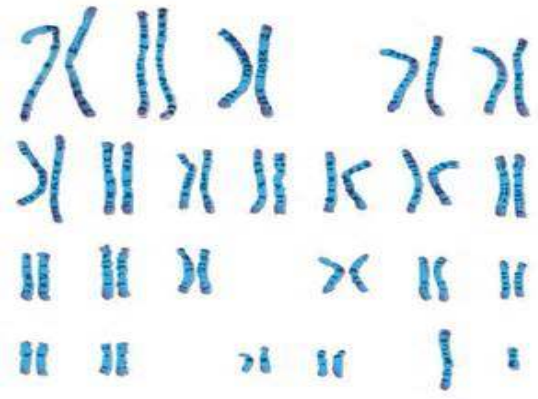

**Fig. 1. Chromosomes**
*Available:https://www.bing.com/images/search?q=chromosome&FORM=HDRSC2*

Every human has 23 pairs of chromosomes (Fig. 1) received from father and mother 23 each. These 23 pairs of chromosomes reside in the nucleus and are called nuclear DNA. 22 pairs (known as autosomal chromosomes) look alike in males and females. The last chromosome is the sex determinant chromosome (allosomal chromosome). The father and the mother donate one each, the father either donates X or Y-chromosome and the mother donate only X-chromosome. A Y-chromosome from the father will give a combination of XY which result gives male and XX combination will result in a female. Nuclear DNA is inherited from both father and mother. Y-chromosome can be used to trace paternal line.

### 3.1 Mitochondrial DNA

Mitochondrial DNA (mtDNA) was discovered in the 1960s by Margit M. K. Nass and Sylvan Nass [13]. In sexual reproduction, research showed that in a female, an egg contains an average of 200,000 mitochondrial DNA (mtDNA) and a sperm contains an average of 5 molecules [14], [15] of mtDNA. In sexual reproduction, the male mtDNA are usually destroyed by the egg cell after fertilization. This is because mtDNA are in the base of the sperm which is lost during fertilization. This makes mitochondria uniparental inheritance exclusively from the mother. Therefore, a mother and her offspring should have the same mtDNA. This can help out in a case where it is difficult to obtain a sample from the suspect, the sample from suspect relevant biological relative could be used. The culprit will be exonerated of the crime if the outcome of mtDNA fingerprint does not match.

### 3.2 Functions of Mitochondria

The function of mitochondria depends on the cell type in which they are found. However, the most important function is to produce Adenosine Triphosphate (ATP), which is the source of energy that keeps everything going in the cell. It also regulates metabolic activity in a cell and storage of calcium. The liver cells mitochondria have enzymes that detoxify ammonia. The mitochondria also help in building certain parts of blood and hormones like testosterone and estrogen. Mitochondrial DNA has been used in forensic science since the 1990s because of its high copy number and particular maternal inheritance [16]. Mitochondrial DNA has 37 genes which all play important role in mitochondrial function [17].

### 3.3 Mitochondrial Diseases

Mitochondria are structures which produce energy in almost all cells in the body. Mitochondrial diseases will cease the functions performed by mitochondria. Depends on which

cell of the body is affected, the disease affects brain heart and muscles with symptoms of the form: poor growth, visual and hearing problems, heart, liver or kidney diseases, diabetics, mental retardation, respiratory disorder, muscles weakness etc.

## 4. BIOMOLECULES OF LIFE

A biomolecule is any molecule that is present in living organisms. In addition to DNA, there are other biological molecules that make life possible, the Ribonucleic Acid (RNA) and Proteins. DNA and RNA look much similar except in the following areas as shown in Table 1.

### 4.1 The Proteins

Protein is an essential biological molecule in the body that facilitates a chemical reaction that takes place in the body like breaking down of food. It contains one or more chains of amino acid residues. The sequence of amino acid residues in a protein is defined by the sequence of the gene.

### 4.2 Ribonucleic Acid (RNA)

RNA and DNA play a vital role in the production of protein.

DNA ⟶ RNA ⟶ Protein
(Flow of genetic information)

Like DNA, RNA is also a vital molecule in synthesizing of proteins. It relays information encoded in DNA to produce protein.

### 4.3 Structures of DNA and RNA

DNA is usually a double helix with two strands in opposite direction. The strands are connected by base pairs that look like rungs in a ladder.

Each base pairs with one another such that A always pairs with T and G pairs with C (Fig. 2). The A-T base- pair has 2 hydrogen bonds while the G-C base pair has 3 hydrogen bonds. This makes the G-C intersection stronger with about 30 percent than the A-T base pair.

RNA also composed of four building blocks which are Adenine, (A), Cytosine (C), Guanine (G) and Uracil (U).

## 5. THE GENETIC CODE AND MUTATION

The Genetic code is the set of rules by which information encoded in genetic material (DNA or RNA sequences) is translated into proteins (amino acid sequences) by living cells. It is the primary source of transmission of hereditary information by the nucleic acid in all organisms. The "words" that comprise the code of life (the *genetic code*) communicate several levels of information [18].

**Table 1. Comparison between DNA and RNA**

| | DNA | RNA |
|---|---|---|
| Structural name | Deoxyribonucleic acid | Ribonucleic acid |
| Structure | A Double-stranded molecule with long chain of nucleotides | A single-stranded molecule with short chain of nucleotides |
| Reaction/stability | Deoxyribose sugar in DNA is less reactive because of C-H bond and stable under alkaline conditions | Ribose sugar in RNA is more reactive because of C-OH (hydroxyl) bond and not stable under alkaline conditions. |
| Action on enzyme | DNA cannot catalyze its own synthesis | RNA can catalyze its own synthesis |
| Functions perform in human | Storing and transferring genetic information | Directly codes for amino acids |
| Bases and sugar | DNA nucleotides are Thymine, Adenine, Cytosine and Guanine. Contains deoxyribose sugar | RNA nucleotides are Uralic, Adenine, Cytosine and Guanine. Contains ribose sugar |
| Base pairing | A-T(Adenine-Thymine) and G-C (Guanine-Cytosine) | A-U(Adenine-Uracil) and G-C (Guanine-Cytosine) |
| Ultraviolet damage | DNA can be damaged by exposure to Ultra-violet rays. | RNA is more resistant to damage by Ultra-violet rays. |
| Position at the nucleus | Found inside the nucleus | Present at the nucleus during transcription |

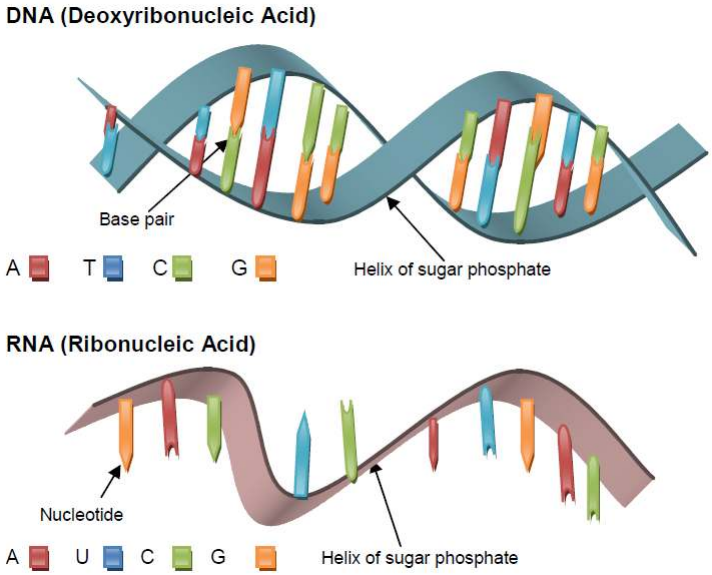


**Fig. 2. Structures of DNA and RNA**

The *genes* are multi-functional, which are capable of producing proteins [19]. In addition to coding for the correct protein, the letters that comprise the genetic code are organized in a way that minimizes errors in protein sequence and structure which help to regulate the amount of protein produced by the cell [20,21,22,10] and possibly assist proteins in folding into the correct functional shape [23,24]. The genetic code is tagged as "blueprint" because it holds the instructions needed by a cell in to maintain itself [25].

The DNA contains three-letter words; these letter words decide the nature and functions of life. The four letters A, C, G, T make the genetic code words Since there are *three* letters in each code word and only *four* letters to select from, the genetic code has 64 ($4^3$) words to imply the necessary information to make all the forms of life on our planet [18].

## 5.1 Reading the Codes

Three nucleotides form a group and referred to as codon where each codon represents one of the 20 different amino acids. Consider the sequence

CCTAAGCCGTTTAGAGAGATTCCT
is grouped as:

CCT-AAG–CCG-TTT–AGA-GAG-ATT-CCT
(where each group represents a codon)

## 5.2 Transcription and Translation

The genetic code can be expressed as either RNA codons or DNA codons. RNA codons occur in messenger RNA (mRNA). They are the codons read during the process of translation. RNA and DNA contain four bases of nucleotide each (A, G, C, U and A, G, C, T respectively), so there are 64 possible triplet codes (codons). 61 codons codes for 20 amino acids, 3 are stop codons (TAA, TAG, and TGA), one code for each of the 20 naturally occurring amino acid [5]. The Amino acid of DNA is shown in Fig. 2. The difference between an amino acid table of DNA and RNA is the substitution of T in DNA codon with U.

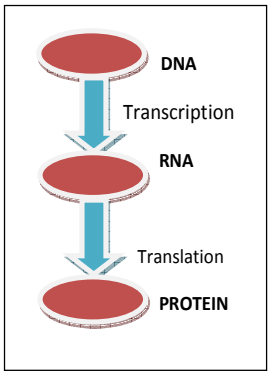


**Fig. 3. Central Dogma**

The Central Dogma of Molecular biology (Fig. 3.), describes the transcription and

translation processes where DNA produces RNA (transcription) and RNA produce Protein (translation). One strand of genomic DNA (strand A, coding strand) contains the following sequence reading from 5′- to 3′-:

The strand TCGTCGACGATGATCATCGGCTACTCG will give the duplex.

5′-TCGTCGACGATGATCATCGGCTACTCG-3'
3′-GCTCATCGGCTACTAGTAGCAGCTGCT-5'

The sequence of bases in the mRNA transcribed from strand A of DNA written 5′- to 3′- is

UCGAGUAGCCGAUGAUCAUCGUCGACG
UCG-AGU-AGC-CGA-UGA-UCA-UCG-UCG-ACG

The amino acid sequence coded by the above mRNA is Ser-Ser-Ser-Arg-STOP

## 5.3 DNA Applications

DNA profiling or DNA fingerprinting is a technique that analyzes the unique attributes of individual's DNA. The application of DNA fingerprinting has revolutionized maternity/ paternity testing, human family ancestry, crime scene investigation and identification of disaster victims [26].

DNA profiling is a forensic method used to identify individuals by characteristics of their DNA. Almost every cell in our body contains our DNA. On average, about 99.9% of the DNA between two humans is the same. The remaining 0.1% is about three million base pairs which are unique between two people (except in identical twin). These unique attributes make it a useful forensic tool to link an individual to a crime scene/criminal act, exonerate suspect by police and also can be used to identify victim of a disaster. Forensic DNA has played a vital role in the criminal justice system. It allows effective human identification [27,28,29]. Some other applications include the field of bioelectronics, photodynamic therapy, a clinical diagnosis which is based on the interaction of DNA with a different chemical probe [30].

## 5.4 DNA Methylation

DNA methylation refers to the addition of methyl group to the strand of DNA, more especially to the fifth carbon Cytosine. DNA methylation is important in growth and development which allows the expression of retroviral *genes.* DNA methylation plays an active role in the formation of chromatin structure which allows a single cell to grow into a complex multi-cellular organism with different tissues and organs [31]. Activities of DNA segment can be changed by methylation without affecting its sequence. Cells used various

**Table 2. Table of amino acids**

| Single- letter code | Three- letter code (Codon) | Amino acid name | DNA codon representation |
|---|---|---|---|
| A | Ala | Alanine | GCT, GCC, GCA, GCG |
| R | Arg | Arginine | CGT, CGC, CGA, CGG, AGA, AGG |
| D | Asp | Aspartic acid | GAT, GAC |
| N | Asn | Asparagine | AAT, AAC |
| C | Cys | Cysteine | TGT, TGC |
| Q | Gln | Glutamine | CAA, CAG |
| E | Glu | Glutamic acid | GAA, GAG |
| G | Gly | Glycine | GGT, GGC, GGA, GGG |
| H | His | Histidine | CAT, CAC |
| I | Ile | Isoleucine | ATT, ATC, ATA |
| L | Leu | Leucine | CTT, CTC, CTA, CTG, TTA, TTG |
| K | Lys | Lysine | AAA, AAG |
| M | Met | Methionine | ATG |
| F | Phe | Phenylalanine | TTT, TTC |
| P | Pro | Proline | CCT, CCC, CCA, CCG |
| S | Ser | Serine | TCT, TCC, TCA, TCG, AGT, AGC |
| T | Thr | Threonine | ACT, ACC, ACA, ACG |
| W | Trp | Tryptophan | TGG |
| Y | Tyr | Tyrosine | TAT, TAC |
| V | Val | Valine | GTT, GTC, GTA, GTG |

*Available: www.compoundchem.com*

techniques to control *gene* expression, but DNA methylation commonly uses epigenetic signaling tools that fix *genes* in the off position. Not all *genes* are active at all times. DNA methylation is one of the several epigenetic mechanisms that cells use to control *gene* expression [32]. Alteration in methylation has been associated with cancer and several other diseases [33].

## 5.5 Genetic Mutation

Any alteration in DNA is known as mutation [34]. If we consider the information in DNA as a series of sentences, then mutations are errors in spelling the word that make up those sentences. Example, MEAT and TEAM contain the same letters, but disarrangement of the letters results in a different meaning. Mutations are changes that occur in the nucleotide sequence of DNA, any alteration or change in DNA sequence can result in disease. Example, sickle cell anemia is the result of the change in a single nucleotide and represents just one class of mutations called point mutation [34].

### 5.5.1 How mutations occur

*5.5.1.1 Spontaneous mutations*

During cell division, an exact copy of its DNA is produced. DNA polymerase is the only enzyme that reads and copies the DNA [35]. They are cardinal enzymes which play a vital role in preserving and maintaining the blueprint of life in all living cells [36]. They are very accurate but not perfect. During the process to produce new DNA, a nucleotide might be wrongly attached to the chain [16].

**Example:**

Original sequence: A C G G T C
Copied sequence: A A G G T C

This type of mutation is called spontaneous because it occurred as part of the normal system. How a cell function is determined by the shape of the cell molecules. This shape is formed by the sequence of the nucleotides A, C, T and G. So, when genetic mutations occur, the structure and function of cells can change, which can lead to changes in the entire organism [1]. In addition, spontaneous mutation can also occur due to other mechanisms like another replication errors, depurination of DNA and damage of DNA by the generation of active-oxygen species.

However, spontaneous mutations cannot be avoided. It is hereditary in nature, it is inherited from our parents and remains with us throughout our lives in almost all available cells in the body.

*5.5.1.2 Induced mutations*

Mutation can also be caused by environmental factors, such as chemicals or ionizing radiation [e.g. UV rays from the sun] said Grace Boehkoff-Falk [37]. Nuclear DNA is the main target of ionizing radiation, exposure of which is followed by many types of DNA damage [38]. Also, mutation can be induced by radioactive exposures e.g. gamma radiation.

UV-B waves are responsible for causing mutations in a tumor suppressor *gene* called*p53.* The mutated *p53 gene* has been implicated in skin cancer, this is also called acquired mutation and it occurs one time during a person's life. It is present only in a certain cell (not in all cells) and has nothing to do with the egg/sperm from the parents, therefore cannot be transferred to the next generation.

Induced mutation can be minimized by avoiding body exposure to any chemical that can damage DNA e.g. chemical in tobacco, x-ray radiation, and Ultraviolet (UV) radiation.

## 5.6 Germinal versus Somatic Mutation

Mutations that take place in somatic tissue and germinal tissue of the *genes* and chromosomes are known as somatic mutations and germinal mutations respectively.

## 5.7 Effects of Genetic Mutations

If instead of TOP we then change the word to POT, these two words have the same letters but different meaning, so also any change in the sequence of DNA nucleotides can change the instructions for building a molecule. If there is any change in shape of a molecule, its functions become abnormal. This abnormality in the cell will change the characteristics of the organism. Human genetic diseases are probably the most dramatic examples of the possible consequences of genetic mutations. People with diseases like cystic fibrosis can suffer their entire lives and ultimately die as a result of a mistake in their DNA sequence. Cancer is another powerful example of how disease can result from genetic mutation.

**Table 3. Comparison table of somatic and germinal mutation**

| Somatic mutation | Germinal mutation |
|---|---|
| Develop after conception | Present at conception of offspring |
| Caused by environmental factor, example U-V radiation | Caused by radiation or mutagens |
| The mutation caused cancer and neurological diseases [39] | The mutation caused the diseases: sickle cell anemia, cystic fibrous, color blindness, albinism etc. |
| Does not occur in the germ cells | Mutation occurred in the germ cells (sperm and eggs) |
| Mutation in somatic cells are not heritable (not transmittable to the offspring) | Mutation in germinal cells are heritable (transmittable to the offspring) |

**Fig. 4. Effects of mutations**
*Available: https://www.bing.com/images/search?q=effects+of+mutations&FORM=HDRSC2*

DNA is found in almost all cells in our body, without the occurrence of a mutation, evolution may not be possible. The location of the DNA where the mutation occurs in our body differs from one person to another. Example, the albino people have a mutation in the *gene* which codes for skin colorations (tyrosinase).

Also, some people become sick if they consume milk, this is because they cannot absorb lactose while those who can absorb lactose in milk are able because of a mutation.

## 6. CONCLUSION

Deoxyribonucleic Acid (DNA) is often referred to as “the code of life” or “blueprint of life” because of the vital roles it plays in our existence. It is a molecule that encodes biological information of our lives and responsible for individual characters and uniqueness. Its knowledge led to the discovery of treatment of various diseases and its discovery in the twentieth century marked the beginning of vital events in medical research.

Research showed that there are over thirty trillion cells in human body, some of these cells carry genetic information [40] encoded in DNA and this genetic information can be transferred from parents to the offspring.

DNA comprises a sequence of nucleotides, any alteration in the sequence will result in mutation and may cause disease like sickle cells anemia. Mutation cannot be avoided (spontaneous mutations). On the other hand, we can run away from the risks by keeping off from radiation that can damage the DNA (induced mutations) such as chemicals found in tobacco, x-ray radiation, ultra violence radiation etc. However, mutation is very essential, because without it evolution will be impossible.

In conclusion, this survey has shown that DNA is essential molecules that play vital roles in our lives especially in growth, reproduction, crime scene investigation, criminal justice system etc.

## COMPETING INTERESTS

Authors have declared that no competing interests exist.

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