Faculty of Medicine
AIN-SHAMS UNIVERSITY
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GENETIC DETERMINANTS OF UROLOGIC DISEASES

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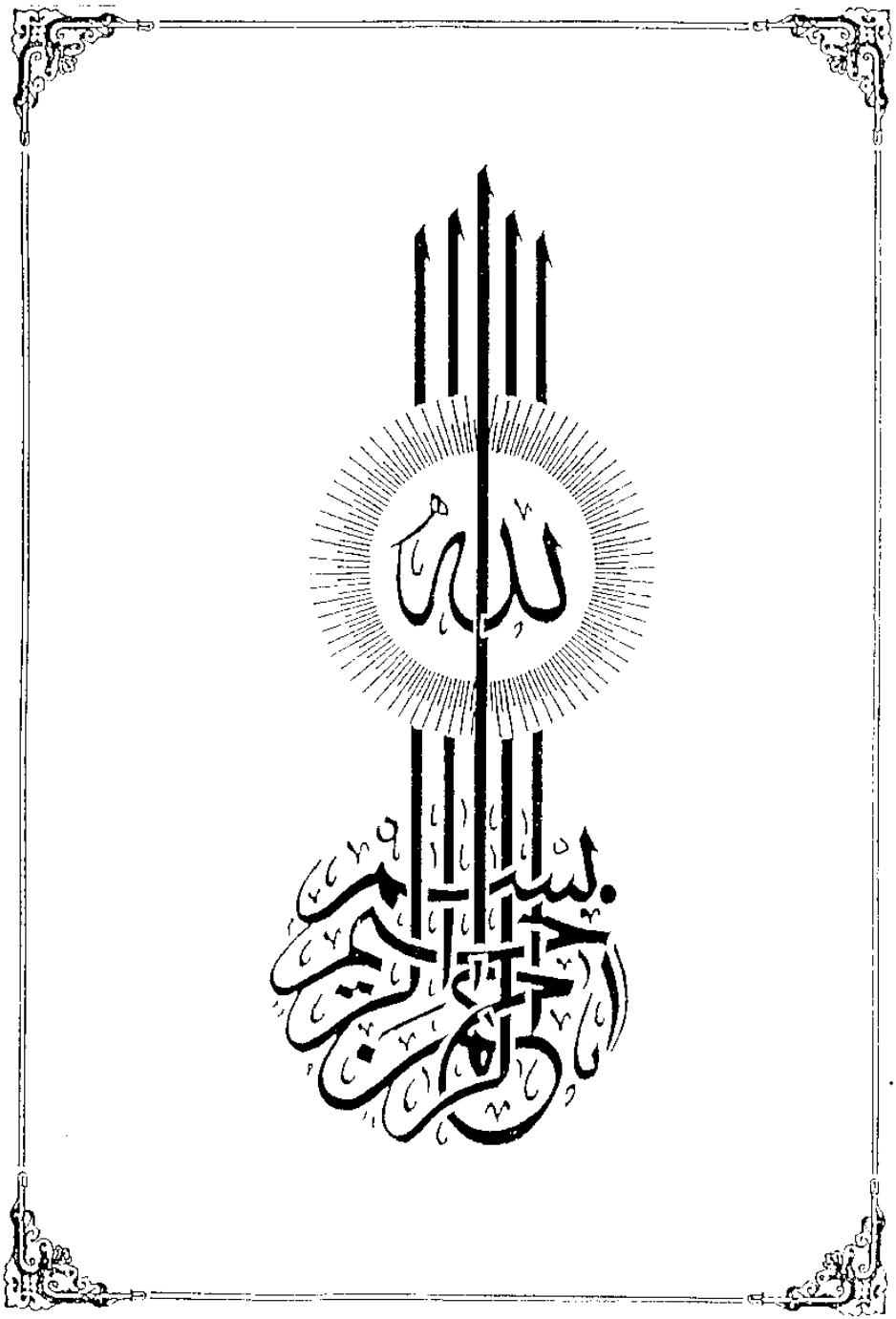
بسم الله الرحمن الرحيم

ربى اشرح لى صدرى ويسر لى امرى
واحلل العقدة من لسانى يفقهوا قولى

صدق الله العظيم



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Hany Hamed, 1995.

TO MY

PARENTS

&

COLLEAGUE

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GENETIC DETERMINANTS OF UROLOGIC DISEASES

PROTOCOAL FOR M.S. ESSAY IN UROLOGY

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GENETIC DETERMINANTS OF UROLOGIC DISEASES

BASIC GENETICS

GENETIC DETERMINANTS OF UROLOGIC DISEASES

All of us must know that **DNA** is the molecule of life . As it contains within its elegantly simple structure that code informations that ultimately dictates all structure and its function.

The last decade has provided the scientific community with the tools and techniques to extract valuable information from this code in such a way that now the Urologic community may for the first time begin to understand the basis for disease and ultimately treatment.

To understand the pathogenic message of the disease we must know some information about **BASIC GENETICS** and **Molecular Basis of gene expression.** **DNA** is poly deoxy ribonucleatides linked together by 3' 5' phospho diesteric linkage **(COVALENT BOND).** It is formed of two strands arranged in double helical antiparallel manner. Both strands are attached by weak hidrogen bond A=T C≡G **"Non Covalent Bond"**. This manner = **Base Piaring** or **Base Complimentary** (Fig. 1).

* The **NEUCLEOTIDE** = Phosphote - Ribose - Base attached by 5' 3' suger phosphate links.

* The **DNA BASES** are Adenine "**A**", Guaunin "**G**", Cytosine "**C**", Thymine "**T**".

* The **GENETIC CODE** = The relation ship between the neucleotide sequence and the protein polypeptide formed "**mRNA**".

* The **GENE** = Series of triplets in the DNA strand "**3 Neucleotides**" e.g. A.C.G. "each triplet = **Codon**" that specifies the amino acid sequence of asingle polypeptide chain of aprotein molecule.

* **N.B.** only one of two strands of the DNA codes for the protein.

* **GENETICS,** It is a term coined by William Bateson (an Englishman) in 1905 and derived from the Greek verb *gennan*, i.e., to generate. Mendel's discovery, expressed in modern terms, is that discrete hereditary traits, as defined by color, shape, and others, are transmitted by units "***genes***" that always go in pairs, or "**alleles**" (Greek allos, other) of which one is provided by the male parent and the other by the female parent. The "***phenotype***" (or character) of the offspring depends on which alleles, dominant or recessive, are received from the two parents. Two individuals with the same two alleles are designated "***homozygous***"; individuals with different alleles are *heterozygous* and express the dominant trait. This is what "**Mendel**" found more than a centure ago [Mendel, 1866]. Inevitably, however, newer exception have been recognized. For example, it is now known that genes may not exist as independent units but are inheritted in sets, a phenomenon termed "***linkage***", or coupling between genes.

To understand the genetics and "**citogenesis**" "Union of cells which found in sexual cells only" we must first understand the folowing :-

A) Function of DNA

The functions of **DNA** can be summarized as :

I) REPLICATION :

Definition :

Formation of a copy of **DNA** carrying the same genetic characters.

Steps :

1) Separation of the parent DNA strands

→ 2 templets 3' → 5' & 5' → 3'

2) The replication of two strands, first the leading strand and as the replication always occur from 5' → 3' so, the 3' → 5' strand templet equal the leading strand, second replication of the trailing strand .

a) Replication of the leading strand by formation of ten **RNA** by primase RNA primer. Then by the DNA polymerase → complimentary **DNA** Then removale of the primer by **ribounlease** enzyme and **DNA**

polymerase fill the gap by DNA. The tow fragments ligated by the **ligase** enzyme.

b) Replication of the trailing stand is a discontinues manner

i.e. primase → ten **RNA**

DNA polymerase as → one hundred **DNA**

→ one hundred ten neucleotides called → **OKAZAKI** piece

Then removal of primers and fill the gap by ligase

II) TRANSCRIPTION :

Formation of **mRNA** from **DNA** strand as a model to prescripe it as a message.

In The mRNA

* The ribonucleotides arranged in a complementary manner to that in DNA gene in triplets or genetic codes. This is by DNA dependent RNA polymerase.

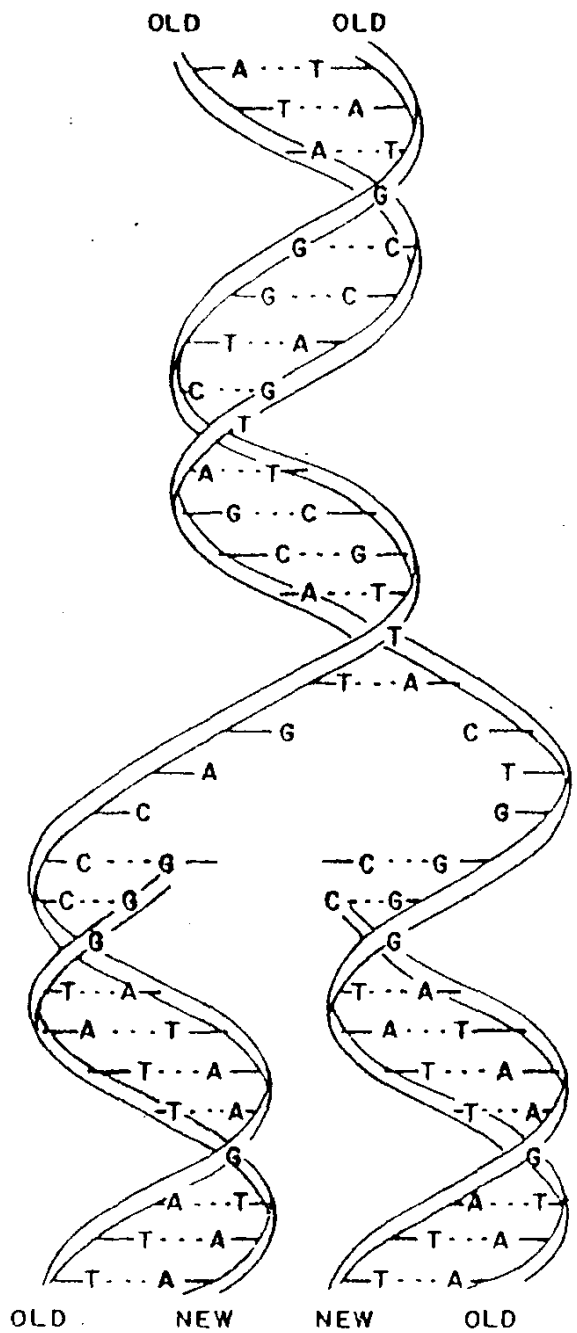

Figure 1 The double-stranded structure of DNA and the template function of each old strand on which a new complementary strand (shaded) is synthesized. (From James D Watson, *Molecular Biology of the Gene*, 3rd ed. Copyright © 1976, 1970, 1965, by W.A. Benjamin, Inc., Menlo Park, Calif.)