

أولاً.

Mandel 1865
Gene " " *Garrod* 1902 .ABO *Weilchtein* 1900
1909 *Johansson* . alkaptonurea

Huntington's disease sickle cell phenylketonuria
1953 .DNA 1944 *Avery* .cystic fibrosis
48 .DNA *Crick Watson*
.1958 46

Down's

[2].21 1959 syndrome

()

chromatin

.() chromosomes

.DNA (deoxyribonucleic acid)

RNA

23.000

blueprint DNA

14.000

46

23

Y X X X

Y X X

DNA -1-1

pentose : 3 DNA

.pyrimidines

thymine cytosine
guanine adenine

.purines

.(G A T C)

Crick Watson

(-1)

.5' 3'

DNA subunit

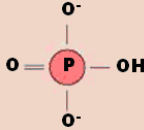
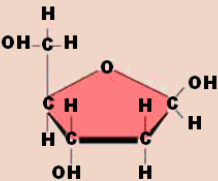
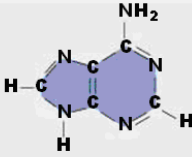
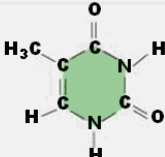
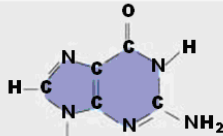
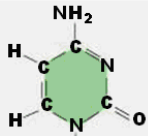
.nucleotide

.(TAACGT)

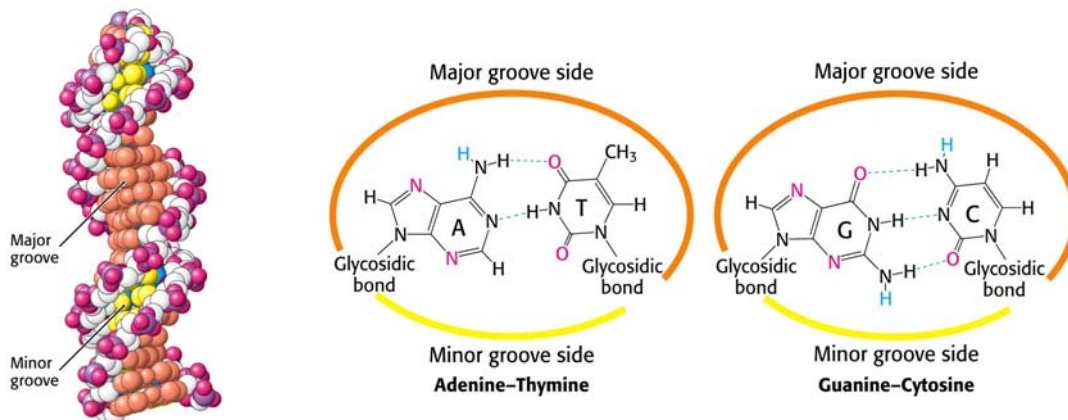
DNA

DNA

3

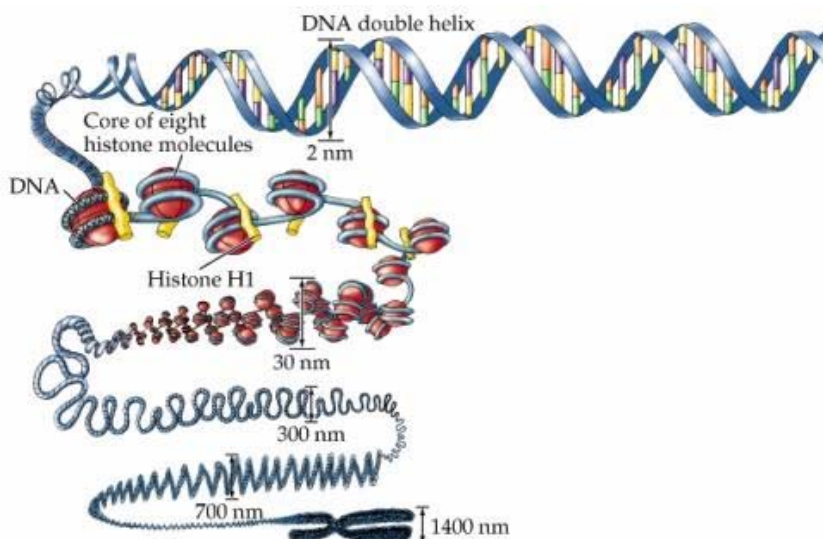
PHOSPHATE GROUP	SUGAR	BASES	
	 <p>Deoxyribose</p>	 <p>Adenine (A)</p>	 <p>Thymine (T)</p>
		 <p>Guanine (G)</p>	 <p>Cytosine (C)</p>

:DNA : -1



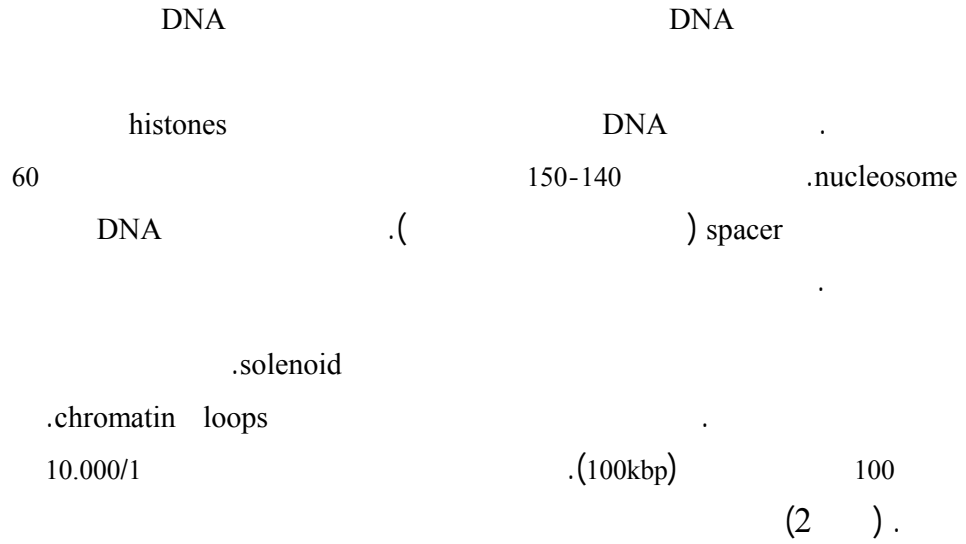
:DNA : -1
DNA

: : -1

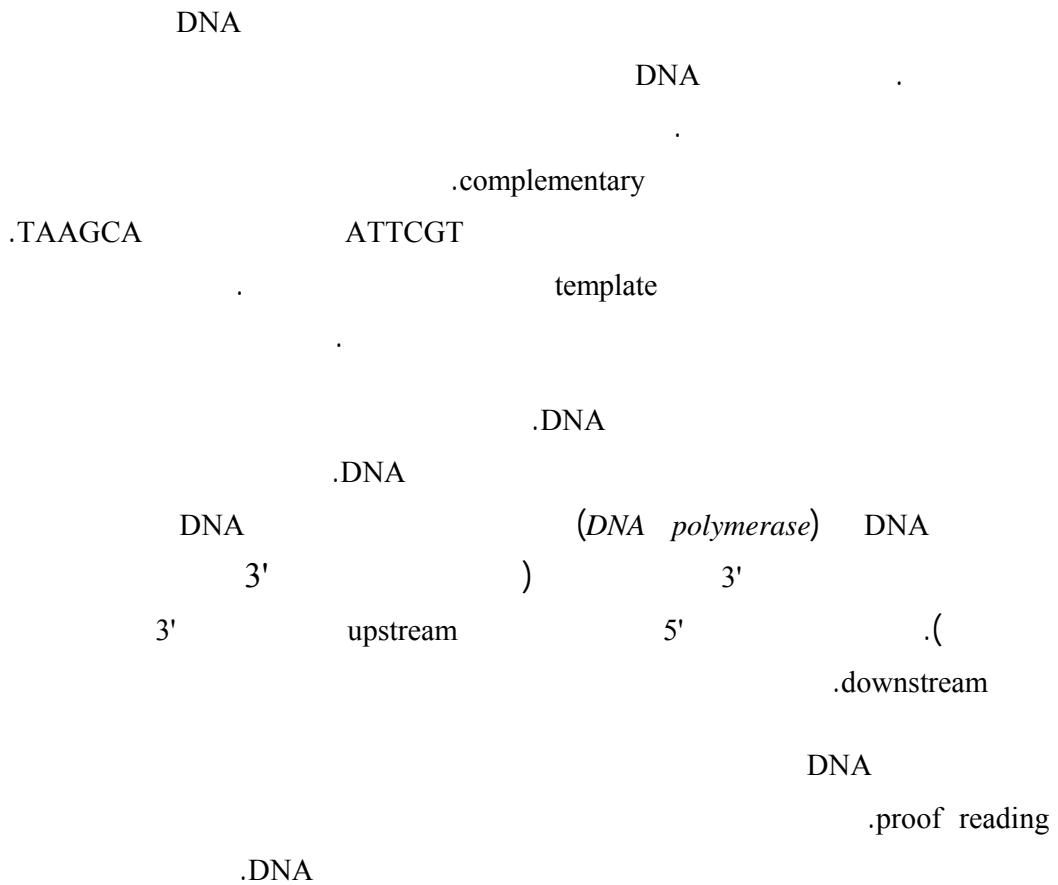


DNA :2

:DNA -2-1



:DNA -3-1



: _____ -4-1

()
 RNA
 DNA
 DNA
 RNA (ribonucleic acid)
 DNA mRNA
 mRNA DNA
 DNA RNA
 : DNA
 uracil RNA DNA
 RNA DNA RNA DNA

: _____ -5-1

.DNA RNA
 RNA RNA
 .promoter RNA polymerase II
 DNA RNA upstream
) RNA
 (. RNA
 .DNA RNA
 5' → 3' mRNA
 .antisense
 .5' cap mRNA
 RNA
 .termination sequence
 .poly-A tail mRNA 3' 200-100
 RNA mRNA
 mRNA DNA

DNA

mRNA

.primary translate

: _____ -6-1

house keeping genes "

variant

DNA

LDL

hematocytoblast

II -RNA

(50)

DNA

RNA

RNA

:Gene splicing -7-1

mRNA

mRNA

RNA

.DNA

.introns

mRNA

.exons

:Genetic code and translation

-8-1

64
.UAG UGA UAA stop codons

DNA

mitochondria

mRNA

(transporter RNA) tRNA

3' 80 RNA

tRNA .anticodon

(ribosomal RNA) rRNA RNA ribosome

tRNA mRNA

AUG

mRNA tRNA

post-translational modification

: _____ -9-1

cystic fibrosis

. neurofibromatosis

mutation

.DNA

.allels

.locus

homozygote

11

.heterozygote

DNA

%1

.Genotype

.polymorphic

: _____ -10-1

:

: _____ .1

(21)

.(X)

: _____ .2

: _____ .3

cleft palate

: _____ .4

.silent mutation

missense

mRNA

nonsense

.RNA

insertion deletion ()

3

.frameshift mutation

Carrot-Marry tooth

17

p22

.2500

.myelin

mRNA

mRNA

Alu repeats *SINE*

DNA

mobile elements

neurolymphomatosis

[2].B A

tandem repeated DNA

" X "

.extended repeats

DNA

.epigenetical mechanisms

RNA DNA DNA DNA

:

CH₃ :(DNA methylation) DNA .1

DNA .() 5-methylcytosin

.() CpG

DNA CpG

(de novo)

DNA

(DNA methyltransferase1) DNMT1

) 5-methylcytosin

) (

.(germ line

CpG

.X

()

:Chromatin modification .2

(histone acetyl-transferases) HATs

.DNA

.(histone deacetylases) HDACs

Chromatin rearrangement

DNA
 CpG
 %90-60
 CpG [5]
 DNA
Xenopus oocyte ()
 5-azacytidine
 CpG DNA [7,6] ()
 X
 [9,8]
 [10]
 [11]

-12-1

.Y X

.X Y

:X -1-12-1

X

Barr

(*Xist*) RNA

CpG

-2-12-1

:X

A X

Duchenne

()

X %50

Y

()

X

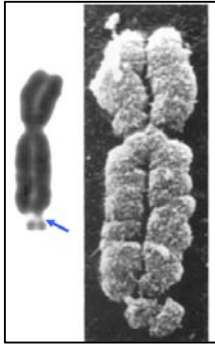
.X

10.000:1

(methyl CpG binding protein 2) *MECP2*

5' CG

X ثانياً.



X :3

%25

X (X-Linked Mental Retardation) (XLMR) X

(

Down

(Folate)

X

flourdeoxyuridine

[12].(3) thymidine

CGG

200

: -1-2

: -1-1-2



:4
[2] X

X

prognathism

[14,13]

X

[15]

%75
[16].X

[17].metacarpophalangeal joints

X
[14].

simian or Sydeny

) callus

[18].(

.palmar crease

sudden

premature growth
[19].

.mitral valve prolapse
[20].

[21].

: -2-1-2

X

%80

IQ .85 70 .50 IQ IQ

[22 ,1].

IQ

X

cluttered

(

)

(

) echolalia

[1].(

)

: -3-1-2

.hand-flapping ()
[12]

: -4-1-2

X
-
X (X)
X

[1]

: -5-1-2

X

X
X

X

[1]

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