

표 1-1. 인체유전체 지표

염기쌍(base pair, bp)의 수 = 6×10^9 (이배수성 유전체)

이 중 1%에서 5%는 단백질 정보를 갖고 있는 코딩 염기 서열이며, 나머지는 조절 부위이거나 폐물(junk) DNA이다.

유전자의 수 = 이배수성 유전체에 35,000개의 유전자가 있다.

유전자의 크기 = 1,000에서 200,000 bp 사이로; 전형적인 유전자는 30,000 bp로 되어 있다.

유전체의 24%는 인트론으로 되어 있다.

유전체의 75%는 유전자 사이에 있다.

유전체의 50%는 반복되는 염기서열로 되어 있다.

개인 간의 유전체 차이는 전체 유전체의 0.1% 또는 1,000 bp 마다 1개씩이다.

단염기다형태의 수 = $2-2.5 \times 10^6$

인간과 침팬지의 유전체 차이: 2% 또는 1,000 bp 마다 20개씩이다.

염색체의 수 = 상동염색체 22쌍과 성염색체 XX 또는 XY 합해서 23쌍 46개.

염색체의 크기: 가장 큰 1번 염색체 = 279 Mb; 가장 작은 22번 염색체 = 48 Mb.

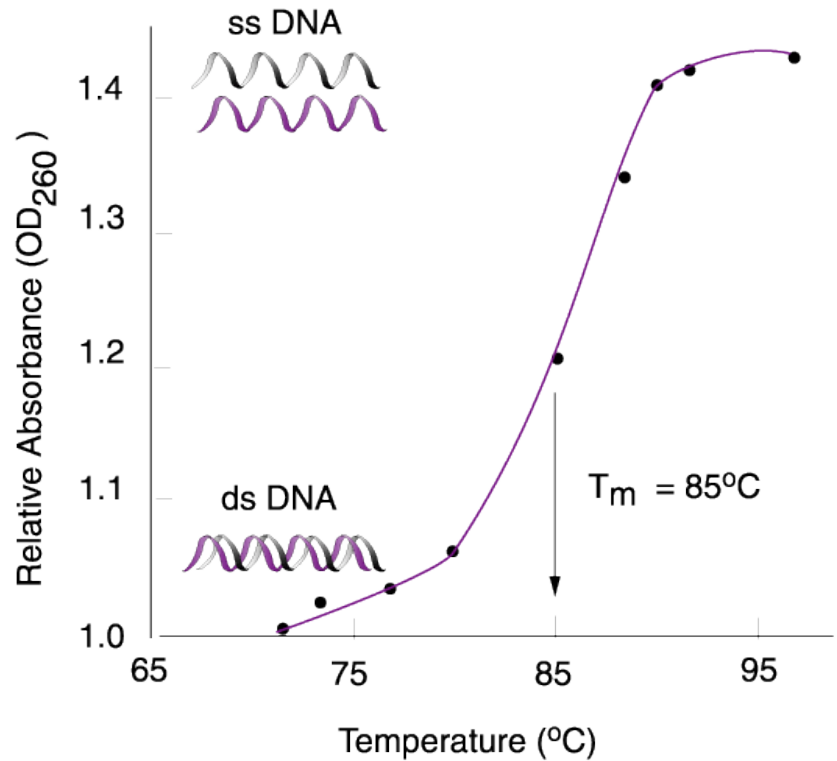
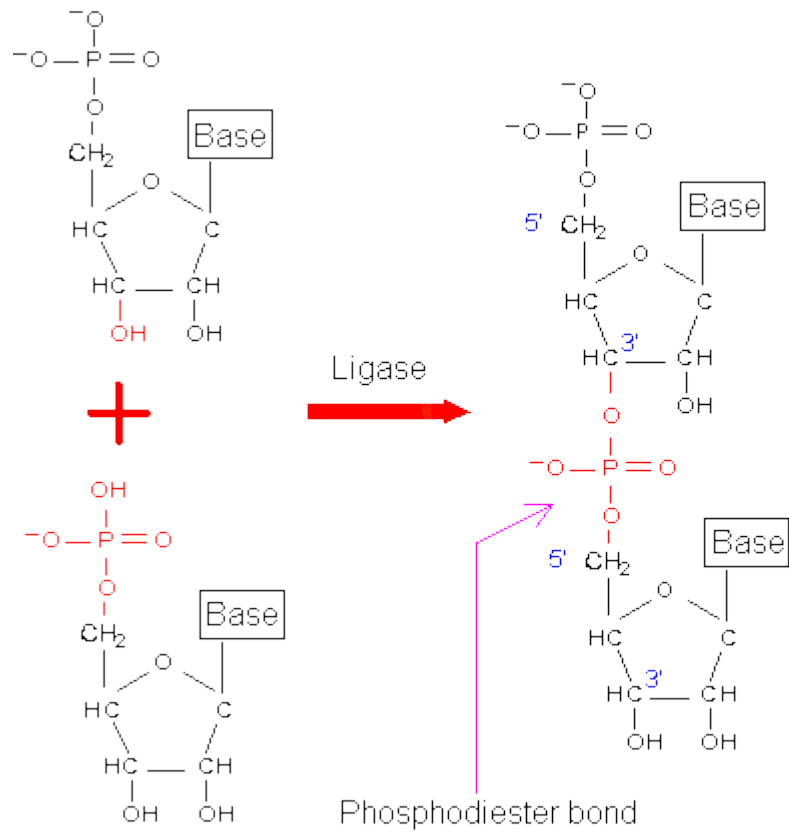
1 센티모르간 (cM): 약 백만 개의 bp (Mb).

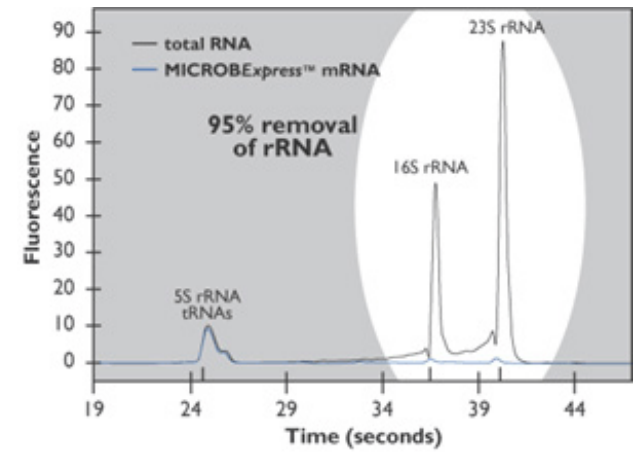
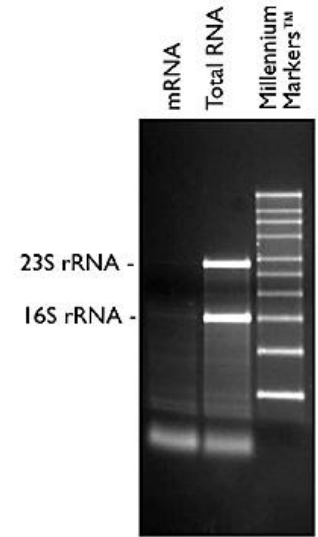
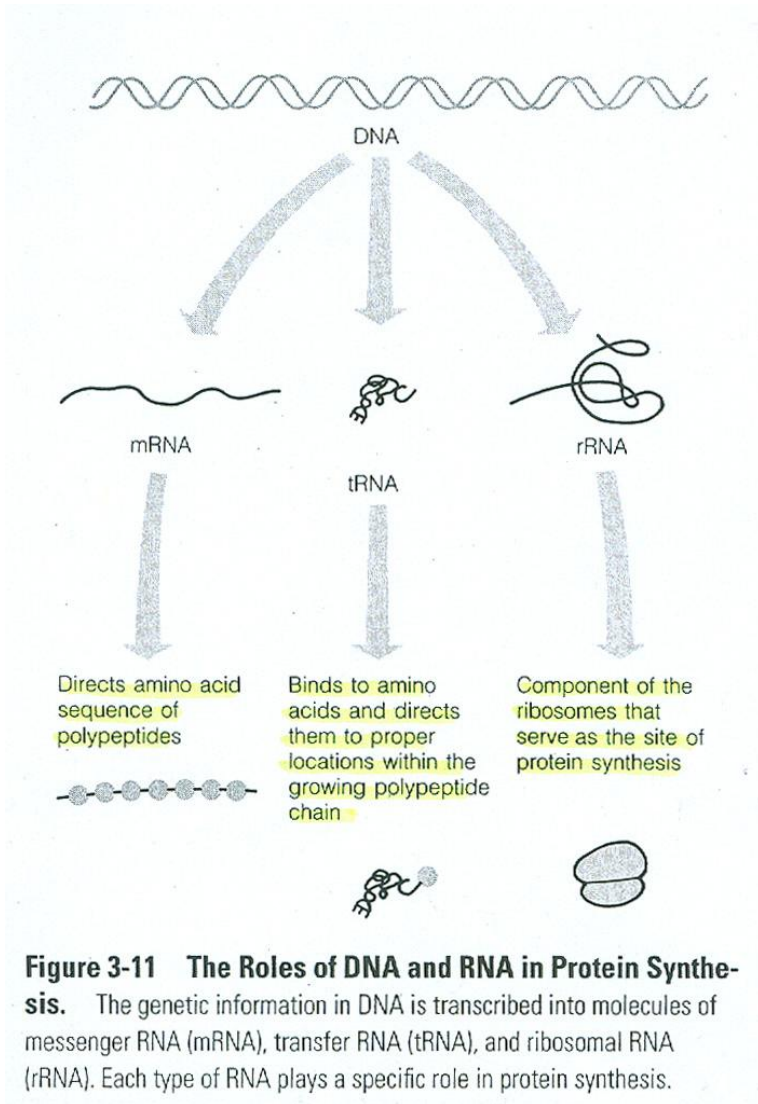
외가닥 DNA의 길이 = 2 m

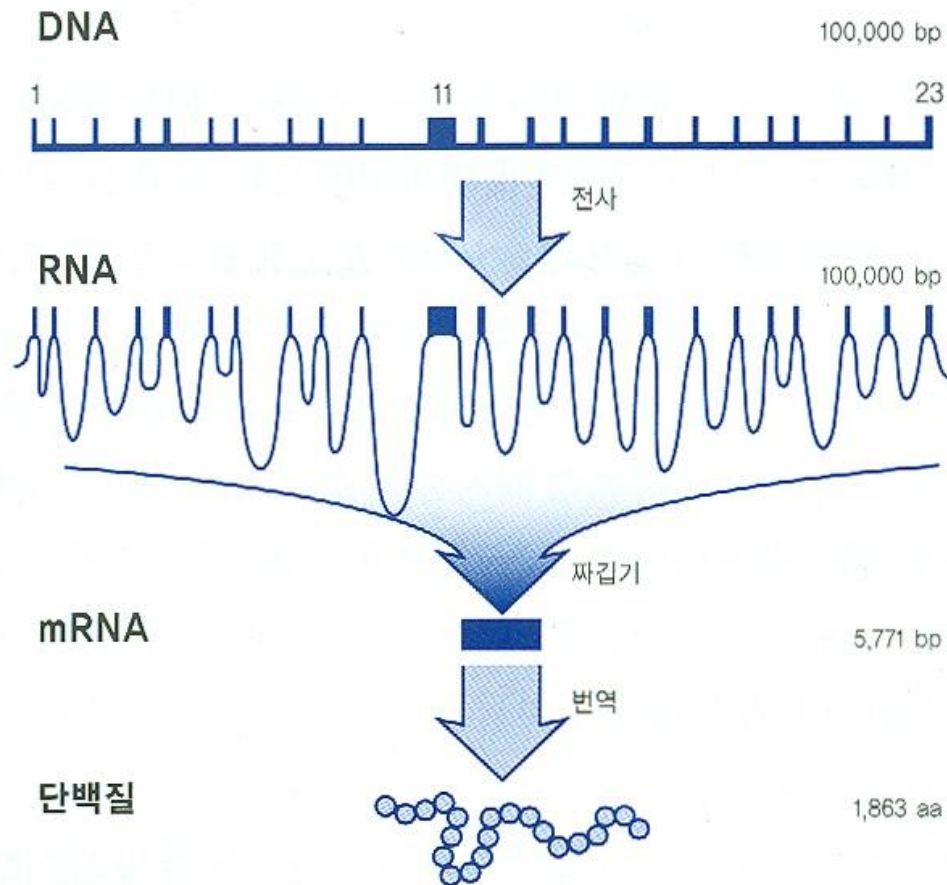
이중 나선 분자로서의 DNA 폭 = 2×10^{-9} m

매 10 bp 마다 360° 도는 나선의 길이 = 3.4×10^{-9} m

한 개의 세포 내에 있는 DNA의 질량 = 7.1×10^{-12} g







192 → aa

그림 1-3. BRCA1 유전자 DNA는 RNA로 전사된 후 인트론 부분은 떨어져 나가고 엑손에 해당되는 부분의 RNA가 붙어 전령 RNA를 형성한다. 전령 RNA는 단백질로 번역된다.

Search Nucleotide for

Display GenBank Show 5 Send to Hide: sequence all but gene, CDS and mdl

Range: from begin to end Reverse complemented strand Features: SNP STS

1: NM_000586. Reports Homo sapiens intc...[gi:125661059]

Comment Features Sequence

LOCUS NM_000586 822 bp mRNA linear PRI 24-AUG-2008

DEFINITION Homo sapiens interleukin 2 (IL2), mRNA.

ACCESSION NM_000586

VERSION NM_000586.3 GI:125661059

KEYWORDS

SOURCE Homo sapiens (human)

ORGANISM Homo sapiens
Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi; Mammalia; Eutheria; Euarchontoglires; Primates; Haplorrhini; Catarrhini; Hominidae; Homo.

REFERENCE 1 (bases 1 to 822)
AUTHORS Juremann, J.D., Jelcic, I., Roberts, S., Lutterotti, A., Taacken, B., Martin, R. and Kunz, C.
TITLE EBNA1-specific T cells from patients with multiple sclerosis cross react with myelin antigens and co-produce IFN-gamma and IL-2
JOURNAL J. Exp. Med. 205 (8), 1763-1773 (2008)
PUBMED 18663124
REMARK GeneRIF: Myelin cross-reactive T cells produced IFN-gamma, but differed from EBNA1-monospecific cells in their capability to produce interleukin-2.

REFERENCE 2 (bases 1 to 822)
AUTHORS Beretta, L., Cappiello, F., Moore, J.H., Barili, M., Greene, C.S. and Scorza, R.
TITLE Ability of epistatic interactions of cytokine single-nucleotide polymorphisms to predict susceptibility to disease subsets in systemic sclerosis patients
JOURNAL Arthritis Rheum. 59 (7), 974-983 (2008)
PUBMED 18576303
REMARK GeneRIF: Observational study of gene-disease association and gene-gene interaction. (HuGE Navigator)

REFERENCE 3 (bases 1 to 822)
AUTHORS Skinningsrud, E., Jusebye, E.S., Pearce, S.H., McDonald, D.O., Brandal, K., Bøe Wolff, A., Lovas, K., Egeland, T. and Indlien, D.F.
TITLE Polymorphisms in CLEC16A and CITA at 16p13 are associated with primary adrenal insufficiency
JOURNAL J. Clin. Endocrinol. Metab. (2008) In press
PUBMED 18593762
REMARK GeneRIF: Observational study of gene-disease association. (HuGE Navigator)

REFERENCE 4 (bases 1 to 822)
AUTHORS Hussain, S.K., Madeleine, M.M., Johnson, L.G., Du, Q., Malkki, M., Wilkerson, H.W., Farin, F.M., Carter, C.C., Galloway, D.A., Daling, J.R., Petersdorf, E.W. and Schwartz, S.M.
TITLE Cervical and vulvar cancer risk in relation to the joint effects of cigarette smoking and genetic variation in interleukin 2
JOURNAL Cancer Epidemiol. Biomarkers Prev. 17 (7), 1790-1799 (2008)
PUBMED 18628433
REMARK GeneRIF: Observational study of gene-disease association and gene-environment interaction. (HuGE Navigator)

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REFERENCE 5 (bases 1 to 822)
AUTHORS Amirzargar, A.A., Naroueynejad, M., Khosravi, F., Dianat, S., Rezaei, N., Mytilineos, J. and Nikdin, B.
TITLE Cytokine single nucleotide polymorphisms in Iranian populations
JOURNAL Eur. Cytokine Netw. 19 (2), 104-112 (2008)
PUBMED 18632425
REMARK GeneRIF: Observational study of genotype prevalence. (HuGE Navigator)

REFERENCE 6 (bases 1 to 822)
AUTHORS Purvis, S.F., Georges, D.L., Williams, T.M. and Lederman, M.M.
TITLE Suppression of interleukin-2 and interleukin-2 receptor expression in Jurkat cells stably expressing the human immunodeficiency virus Tat protein
JOURNAL Cell. Immunol. 144 (1), 32-42 (1992)
PUBMED 1394441

REFERENCE 7 (bases 1 to 822)
AUTHORS Mott, H.R., Driscoll, P.C., Boyd, J., Cooke, R.M., Weir, M.P. and Campbell, I.D.
TITLE Secondary structure of human interleukin 2 from 3D heteronuclear NMR experiments
JOURNAL Biochemistry 31 (33), 7741-7744 (1992)
PUBMED 1510960

REFERENCE 8 (bases 1 to 822)
AUTHORS Bazan, J.F.
TITLE Unraveling the structure of IL-2
JOURNAL Science 257 (5068), 410-413 (1992)
PUBMED 1631562

REFERENCE 9 (bases 1 to 822)
AUTHORS Brodie, C. and Gelfand, E.W.
TITLE Functional nerve growth factor receptors on human B lymphocytes. Interaction with IL-2
JOURNAL J. Immunol. 148 (11), 3492-3497 (1992)
PUBMED 1316918

REFERENCE 10 (bases 1 to 822)
AUTHORS Waldmann, T.A.
TITLE The interleukin-2 receptor
JOURNAL J. Biol. Chem. 266 (5), 2681-2684 (1991)
PUBMED 1993646
REMARK Review article

COMMENT REVIEWED REFSEQ: This record has been curated by NCBI staff. The reference sequence was derived from X01586.1 and BC070338.1. On Feb 14, 2007 this sequence version replaced gi:28178860.

Summary: The protein encoded by this gene is a secreted cytokine that is important for the proliferation of T and B lymphocytes. The receptor of this cytokine is a heterotrimeric protein complex whose gamma chain is also shared by interleukin 4 (IL4) and interleukin 7 (IL7). The expression of this gene in mature thymocytes is monoallelic, which represents an unusual regulatory mode for controlling the precise expression of a single gene. The targeted disruption of a similar gene in mice leads to ulcerative colitis-like disease, which suggests an essential role of this gene in the immune response to antigenic stimuli. [provided by RefSeq]

Publication Note: This RefSeq record includes a subset of the publications that are available for this gene. Please see the Entrez Gene record to access additional publications.
COMPLETENESS: full length.

PRIMARY	REFSEQ_SPAN	PRIMARY_IDENTIFIER	PRIMARY_SPAN	COMP
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FEATURES

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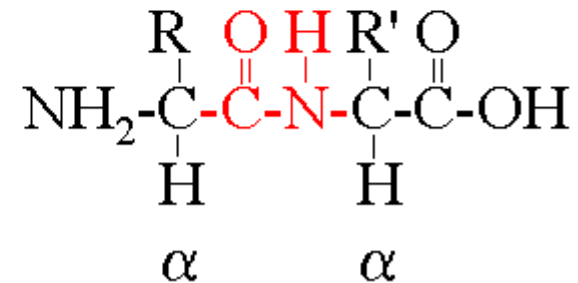
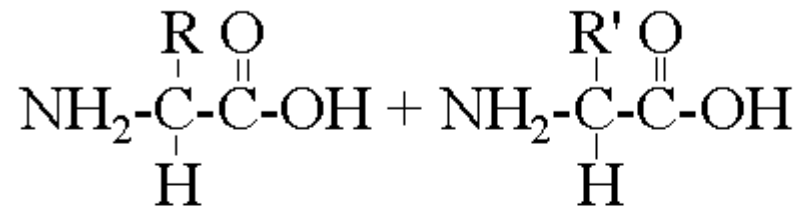
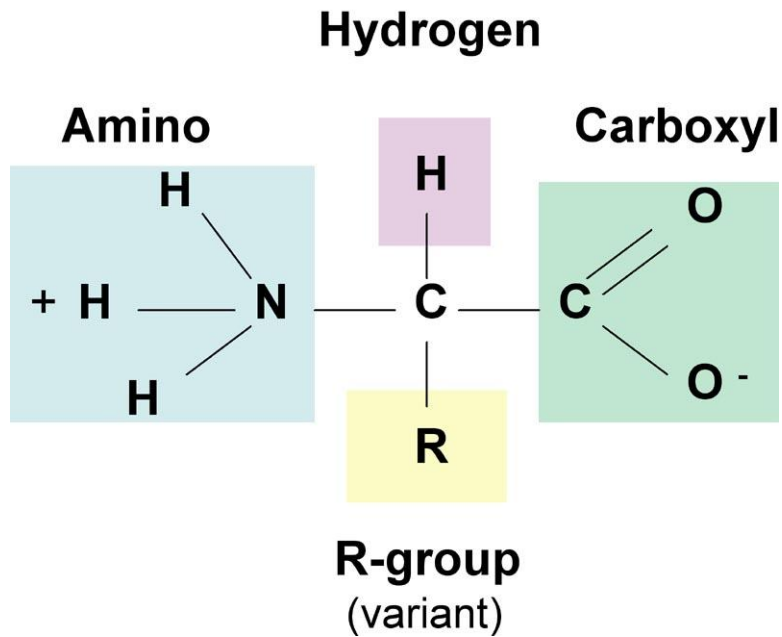
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Last update: Thu, 03 Jul 2008 Rev. 132917



Amino Acid Structure



Nonpolar, Non-ionizable

(소수성, 중성)

Alanine



Valine



Leucine



Isoleucine



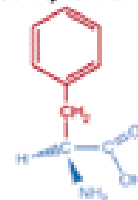
Proline



Tryptophan



Phenylalanine



Methionine



Polar, Non-ionizable

(친수성, 중성)

Glycine



Serine



Threonine



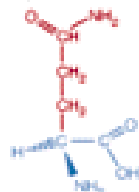
Tyrosine



Cysteine



Glutamine



Asparagine



Basic

Acidic

Lysine



Arginine



Histidine

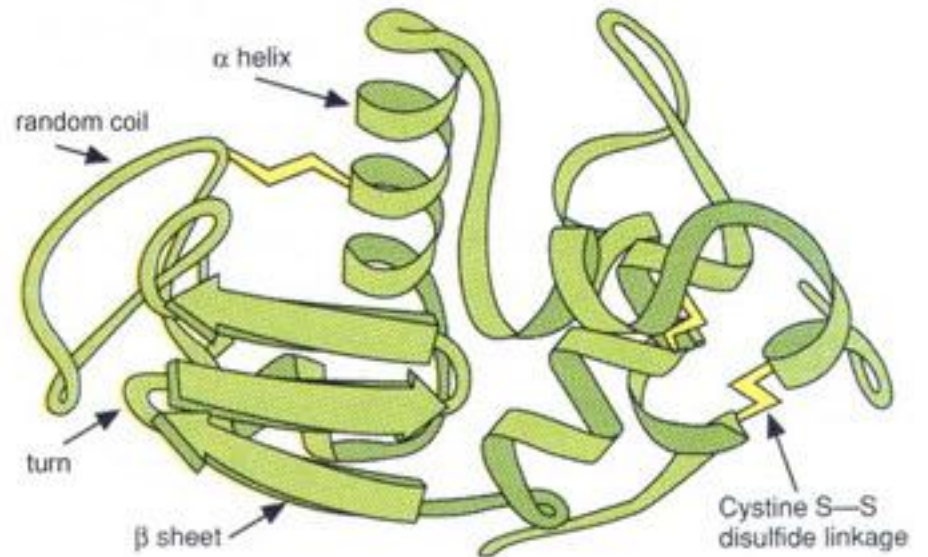
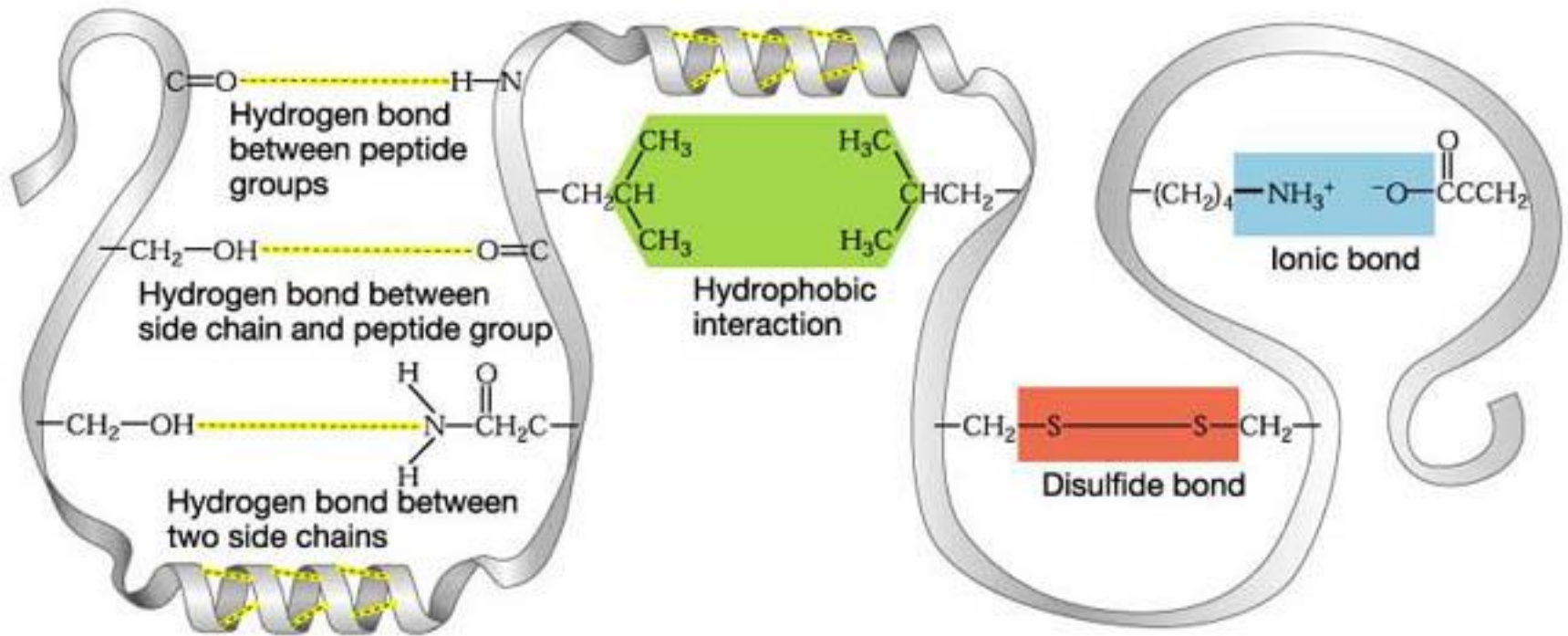


Aspartic acid



Glutamic acid







Primary protein structure is sequence of a chain of amino acids.

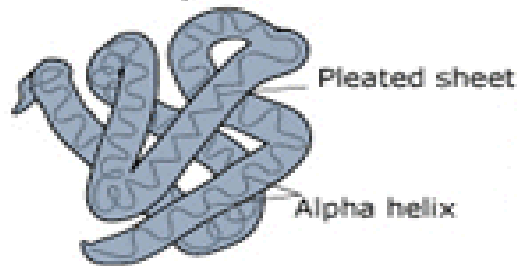
Amino Acids



Pleated sheet

Alpha helix

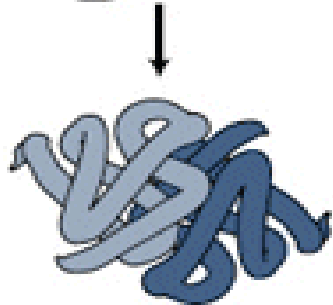
Secondary protein structure occurs when the sequence of amino acids are linked by hydrogen bonds.



Pleated sheet

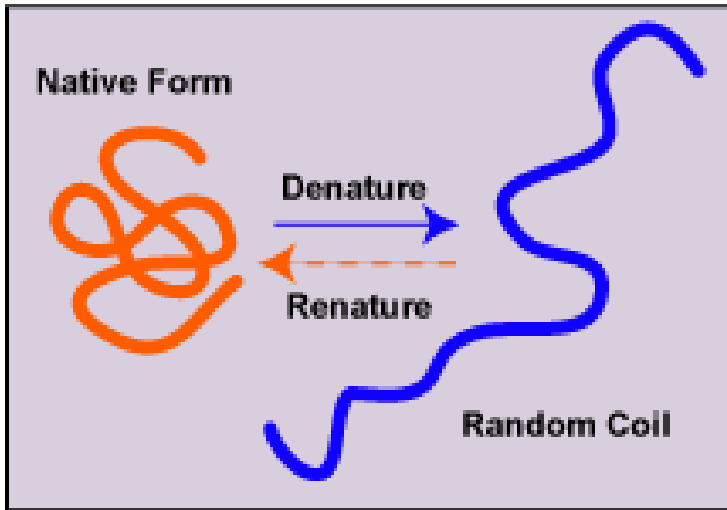
Alpha helix

Tertiary protein structure occurs when certain attractions are present between alpha helices and pleated sheets.

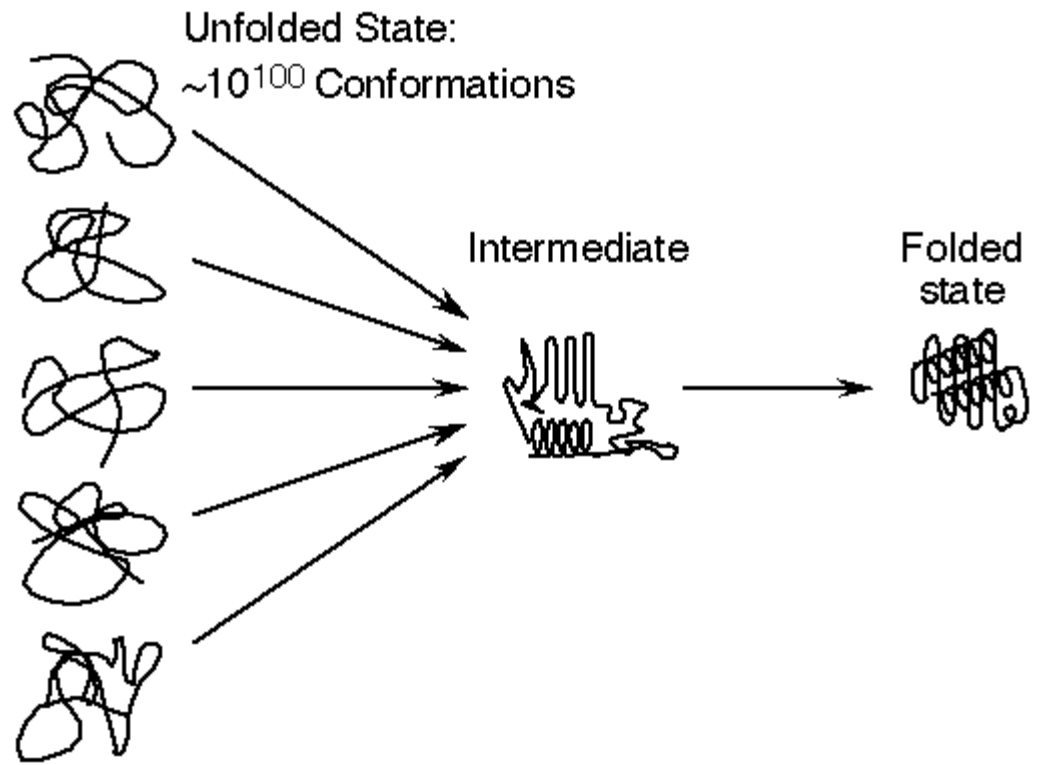


Quaternary protein structure is a protein consisting of more than one amino acid chain.

Image adapted from: National Human Genome Research Institute.



Refolding



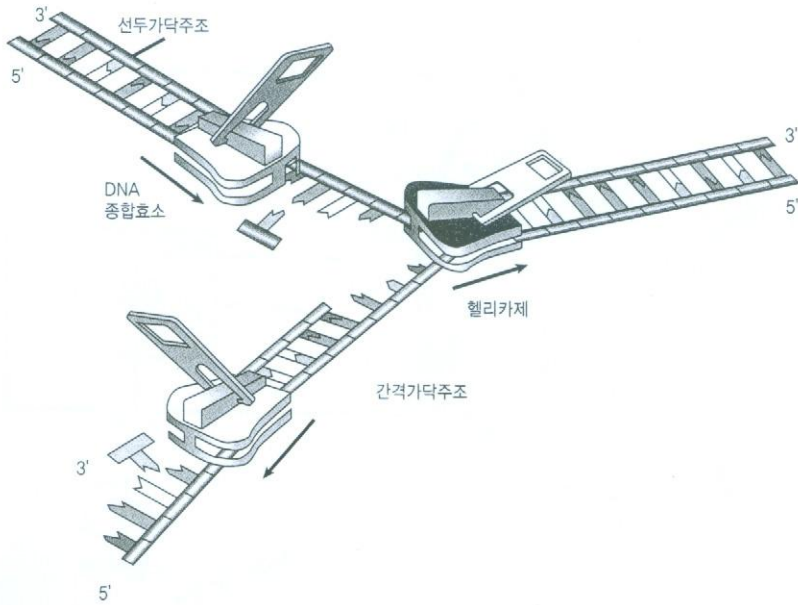
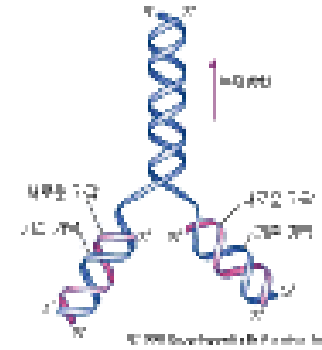
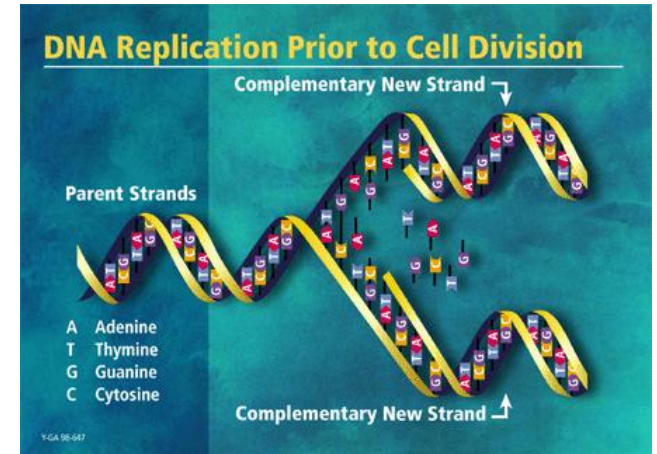


그림 2-1. DNA 복제는 두 가닥의 DNA가 풀려 한 가닥이 되면서 시작된다. 새로 만들어진 한 가닥의 DNA는 새로운 복사 DNA를 만들기 위해 주조로 이용된다.



by Watson & Crick

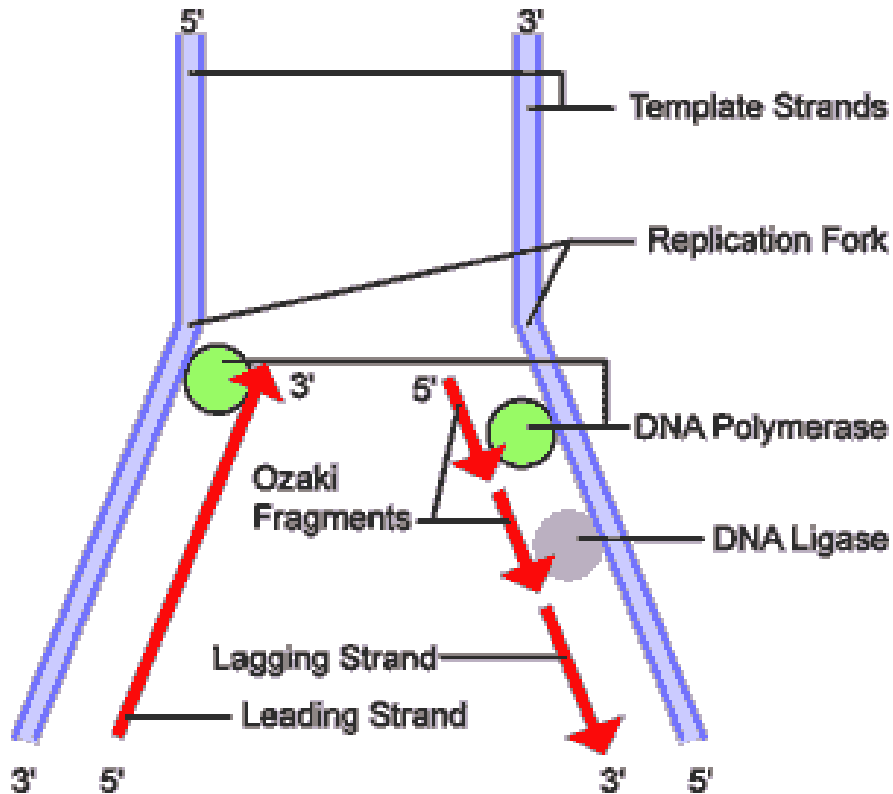


DNA replication의 특징

- : 반 보존적 복제 (semi-conservative replication)
- : by DNA-dependant DNA polymerase

DNA (RNA) 중합효소는 5'(P)-to-3'(OH) 방향으로만 작용함

- leading strand
- lagging strand : Okazaki fragment의 ligation



Enzymes for the replication process - I

Topoisomerase

: DNA untwisting

I. Topoisomerase I

: ahead of the replicating DNA

: ss cleavage

II. Gyrase (Topoisomerase II)

: ds cleavage

Helicase

: Unwinding of double helix

: ds 사이의 수소결합 절단

RNA pol.

- Rifampicin-sensitive

- 선도 가닥의 RNA primer 합성

- ori에서 한번만 작용

Primase

- Rifampicin-insensitive

- 지연가닥의 RNA primer 합성

- 오카자키 단편 마다 작용

DNA ligase

- 5'-일인산만을 기질로 사용함

- (DNA-DNA)만 가능

Enzymes for the replication process - II

DNA pol. I

- : DNA dep. DNA polymerization
- : 3'-to-5' exonuclease
- : 5'-to-3' exonuclease

Trypsin digestion of DNA pol. I

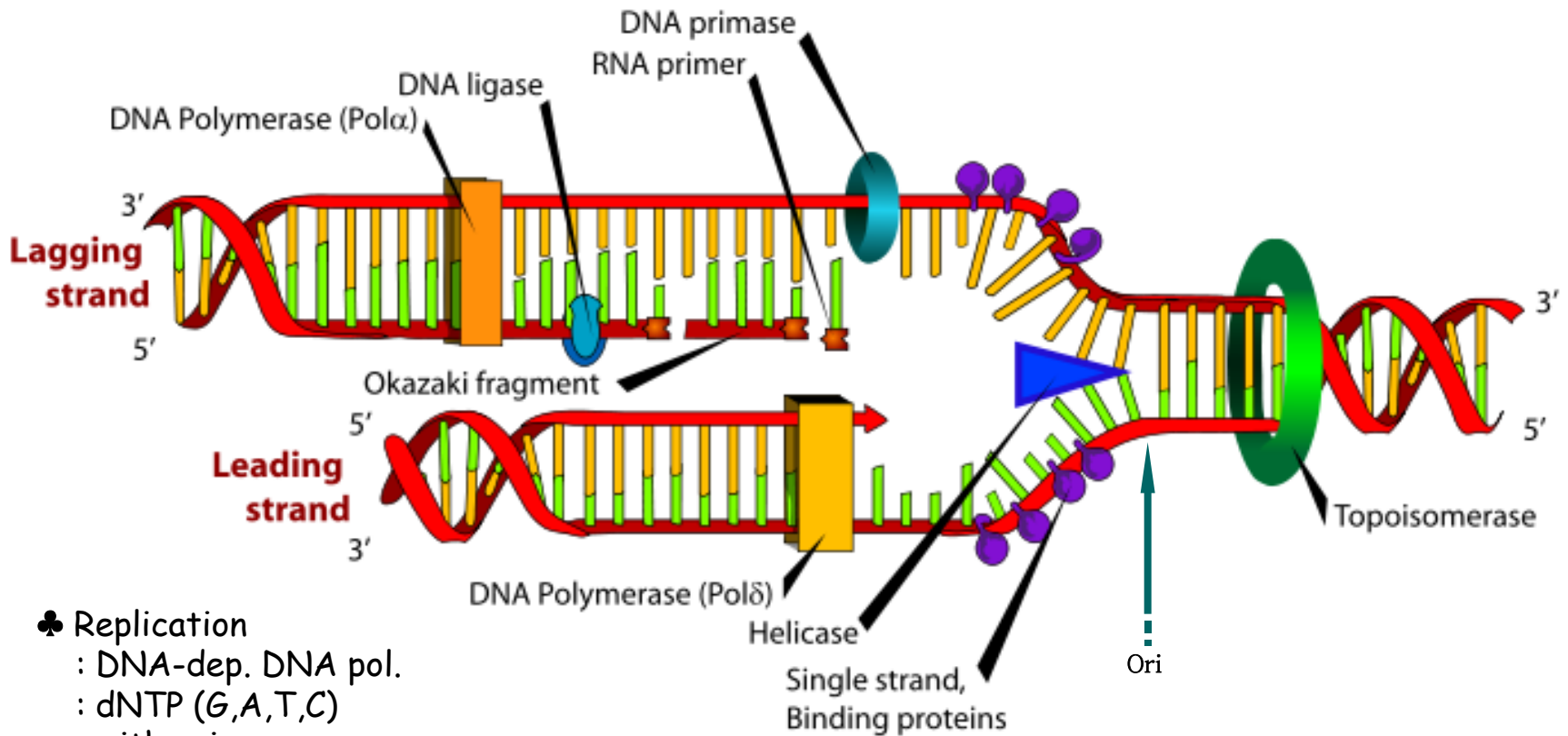
- Large carboxy terminal fragment
(Klenow fragment)
- DNA dep. DNA polymerization
 - 3'-to-5' exonuclease

- Small amino terminal fragment
- 5'-to-3' exonuclease

DNA pol. III

- : holoenzyme (10개의 subunit으로 구성)
- : α subunit - *dna E* gene product
 - DNA polymerization
- : ϵ subunit - *dna Q* gene product
 - 3'-to-5' exonuclease

- 전효소 (holoenzyme) : 여러 subunit으로 구성
일부 subunit 제거 시에도 활성 유지
- 핵심효소 (core enzyme) : 활성을 지닌 최소의 단위 집합체



- ♣ Replication
- : DNA-dep. DNA pol.
- : dNTP (G,A,T,C)
- : with primer
- : 5'-P
- : ds DNA to (ds DNA)₂
- : 역평행식
- : at origin
- : with various enz.