Textos Científicos

www.revista-nanociencia.ece.buap.mx

ISSN 0188-6150

Inter*net* Electronic Journal
Nanociencia et Moletrónica
Octubre 2004, Vol. 2; N°2, (2004)

EL ADN LA MOLECULA DE LA VIDA: Tutorial

Jaime DESCAILLEAUX M. Sc.

PROFESOR PRINCIPAL D.E. LABORATORIO DE GENETICA HUMANA, INSTITUTO DE CIENCIAS BIOLOGICAS

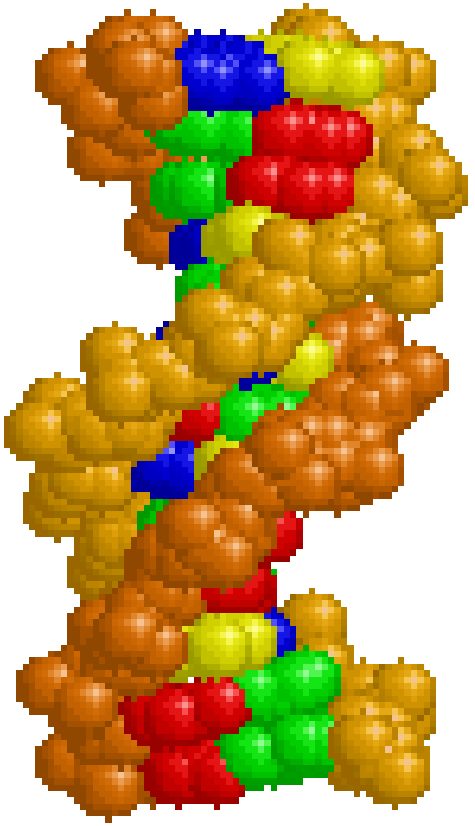
ANTONIO RAIMONDI

INSTITUTO NACIONAL DE BIOLOGIA ANDINA UNMSM.

Jdescailleaux@unmsm.edu.pe

http://www.revista-nanociencia.ece.buap.mx

EL ADN LA MOLECULA DE LA VIDA



Jaime DESCAILLEAUX M. Sc.

PROFESOR PRINCIPAL D.E.

LABORATORIO DE GENETICA HUMANA

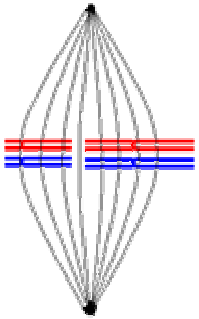
INSTITUTO DE CIENCIAS BIOLOGICAS ANTONIO RAIMONDI

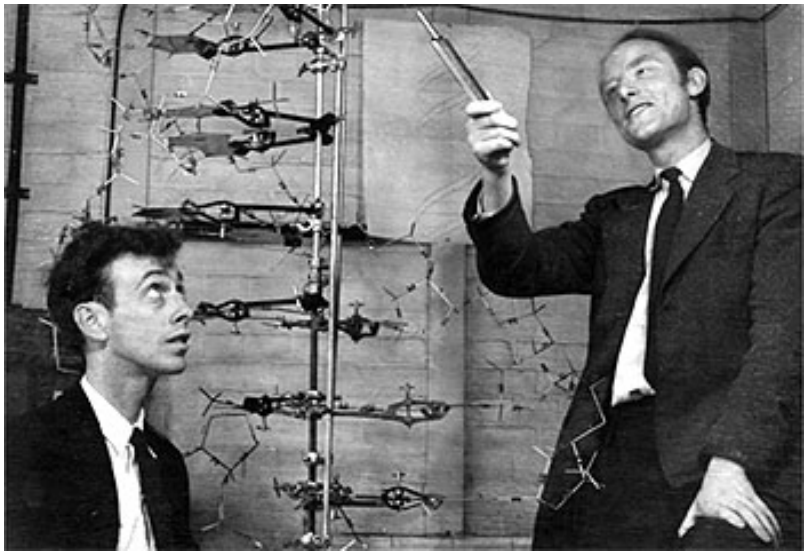
INSTITUTO NACIONAL DE BIOLOGIA ANDINA

UNMSM.

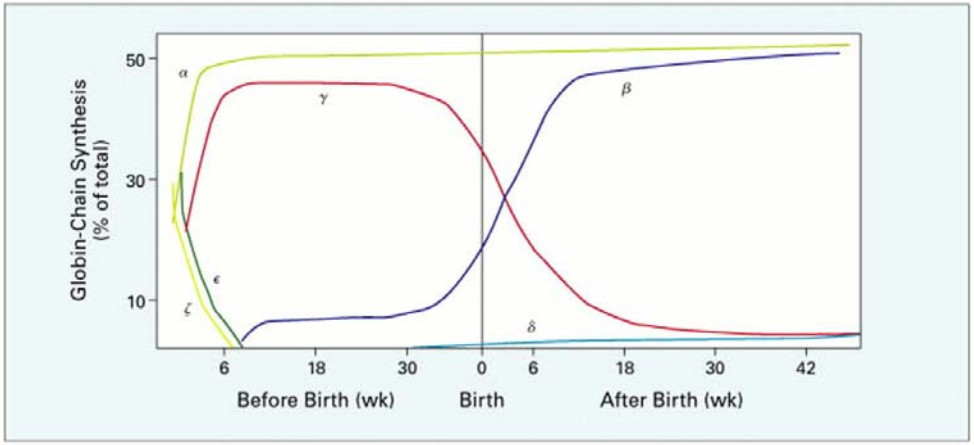
Jdescailleaux@unmsm.edu.pe

QUE ESTUDIA LA GENETICA?





- Como se transmite el material hereditario?
- Cuál es la naturaleza química del material genético?
- Cuál o cuales son los mecanismos que regulan la expresión génica?

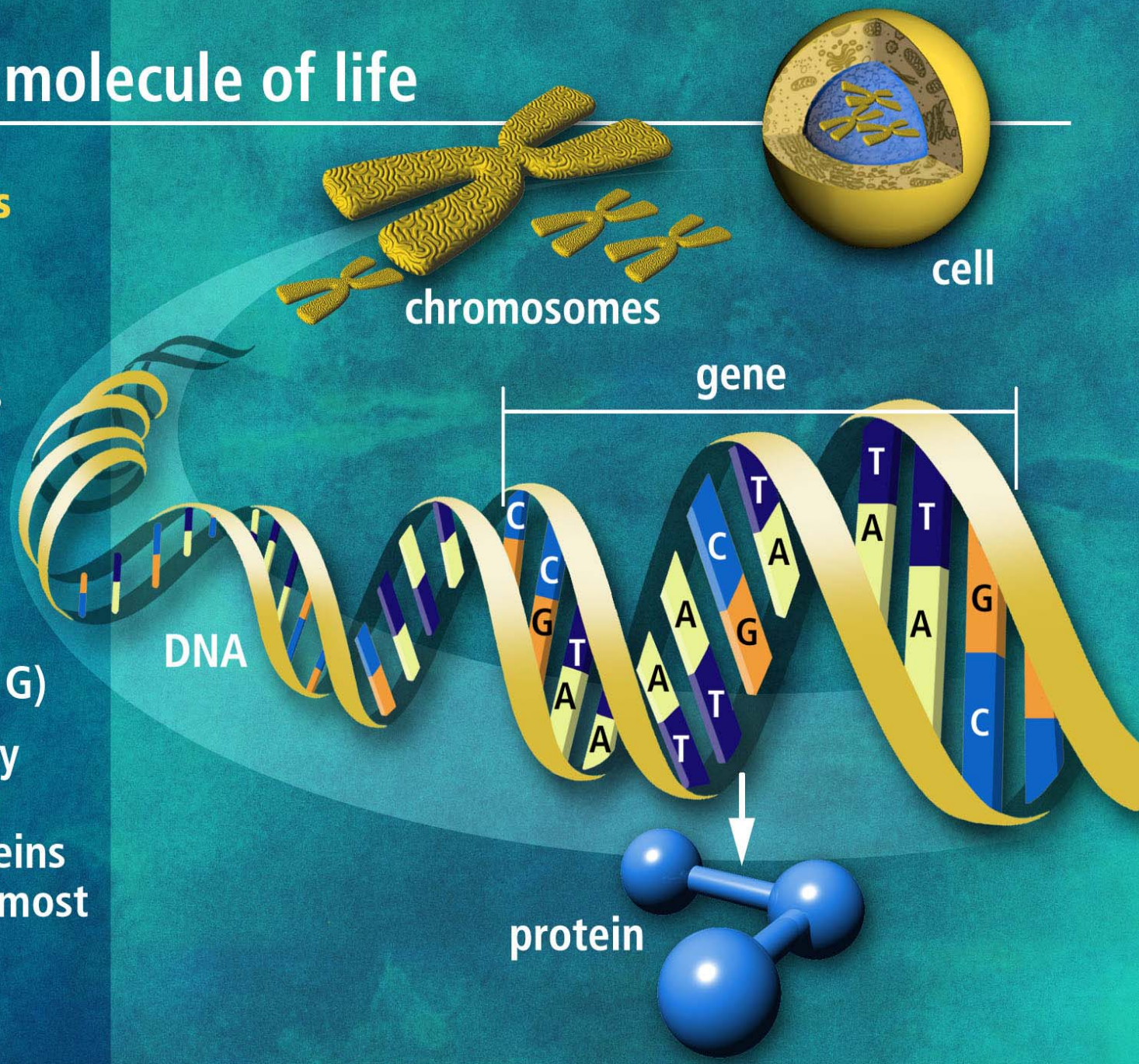


DNA the molecule of life

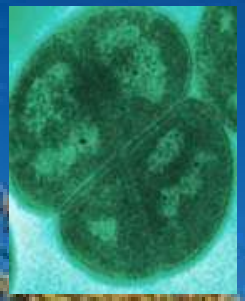
Trillions of cells

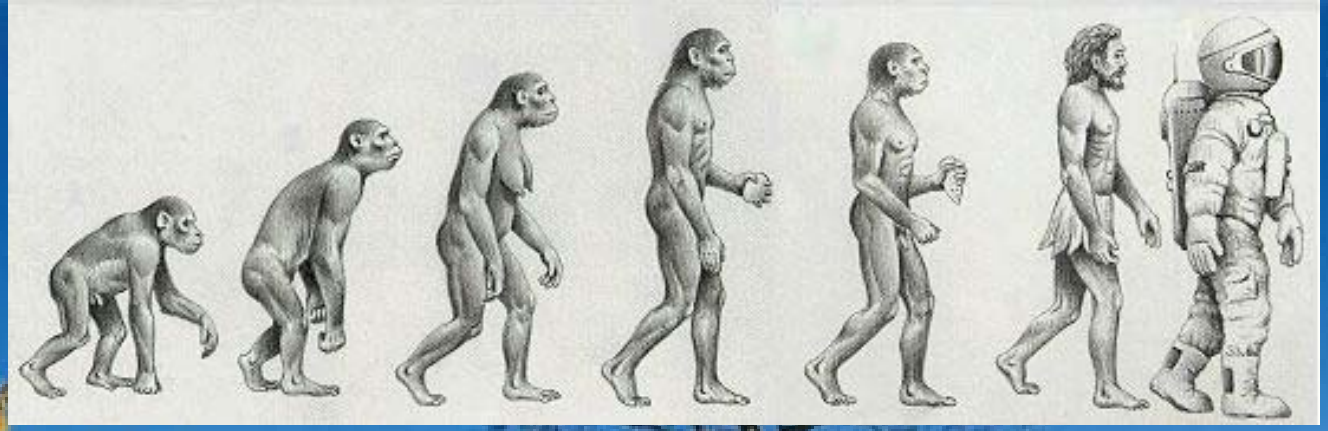
Each cell:

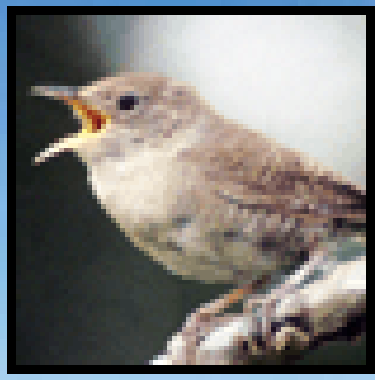
- 46 human chromosomes
- 2 meters of DNA
- 3 billion DNA subunits (the bases: A, T, C, G)
- Approximately 30,000 genes code for proteins that perform most life functions



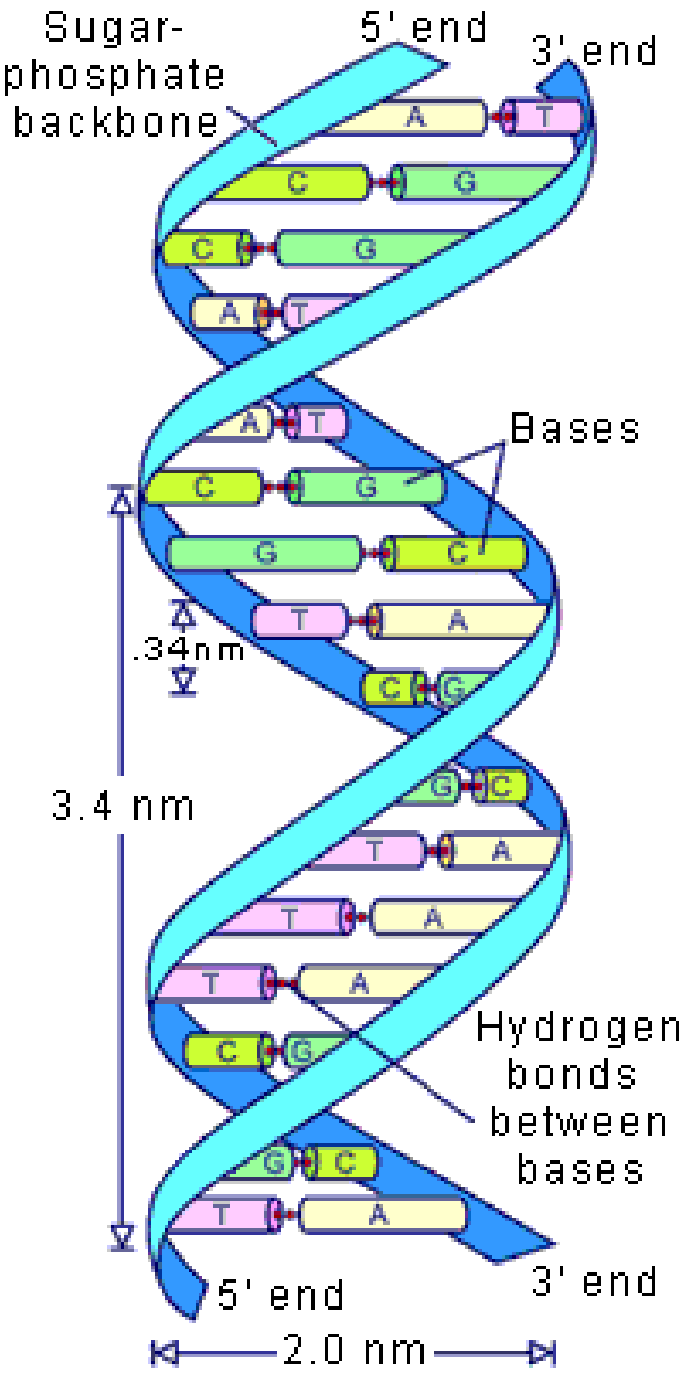
Y-GG 01-0085



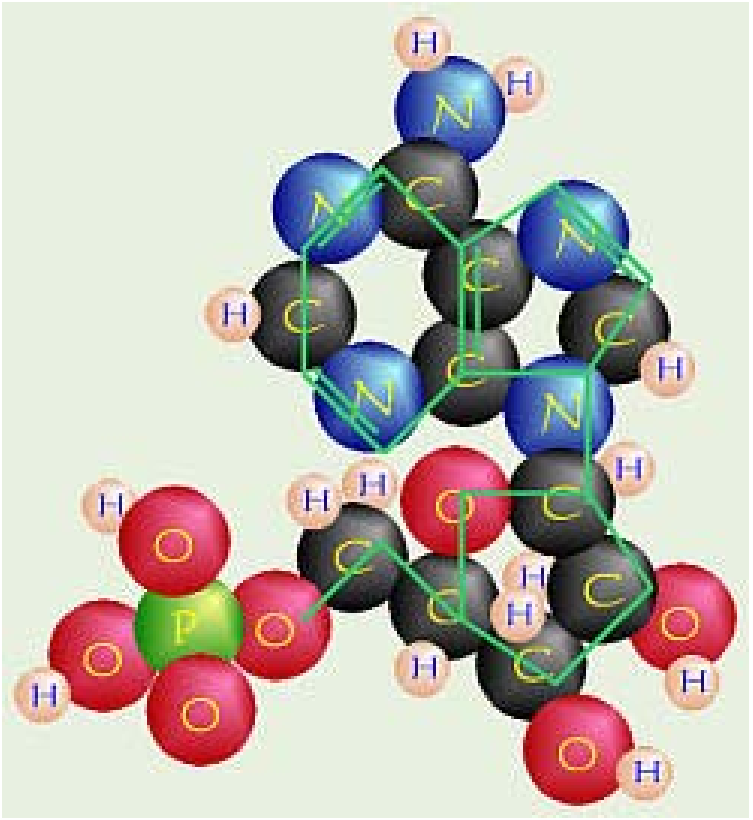


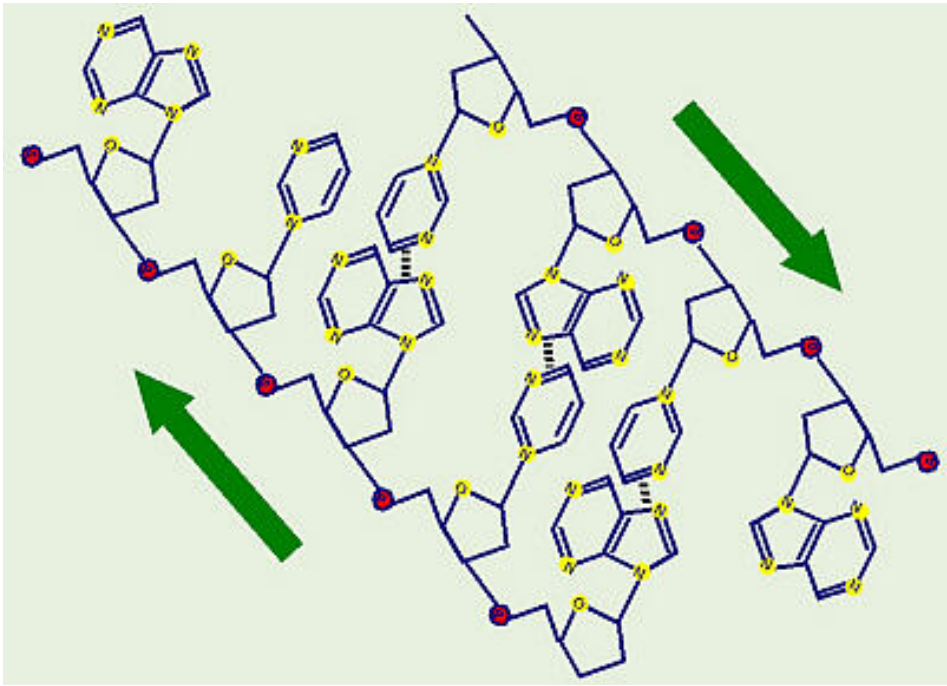


2 CADENAS ANTI PARALELAS EN DOBLE HELICE

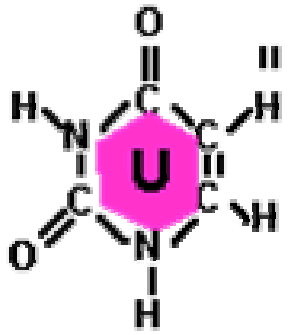


CADA CADENA ES UN POLI NUCLEOTIDO

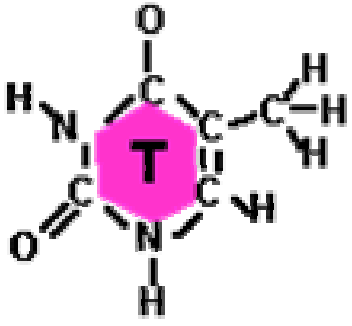




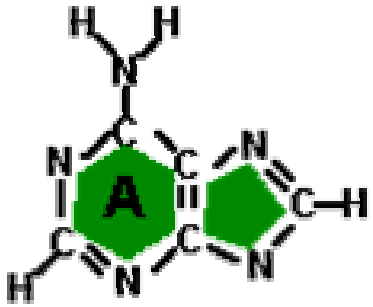
BASES NITROGENADAS: PIRIMIDINICAS Y PURICAS



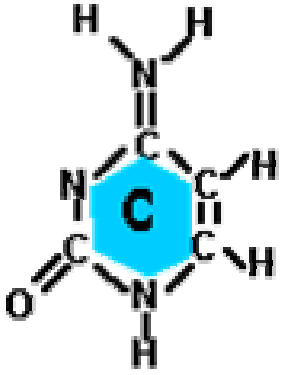
uracile



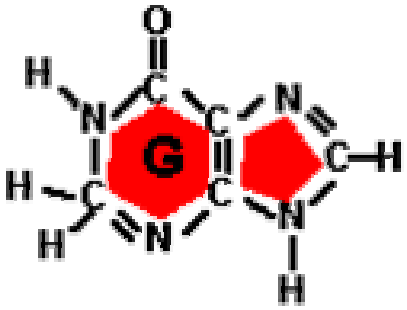
thymine



adénine

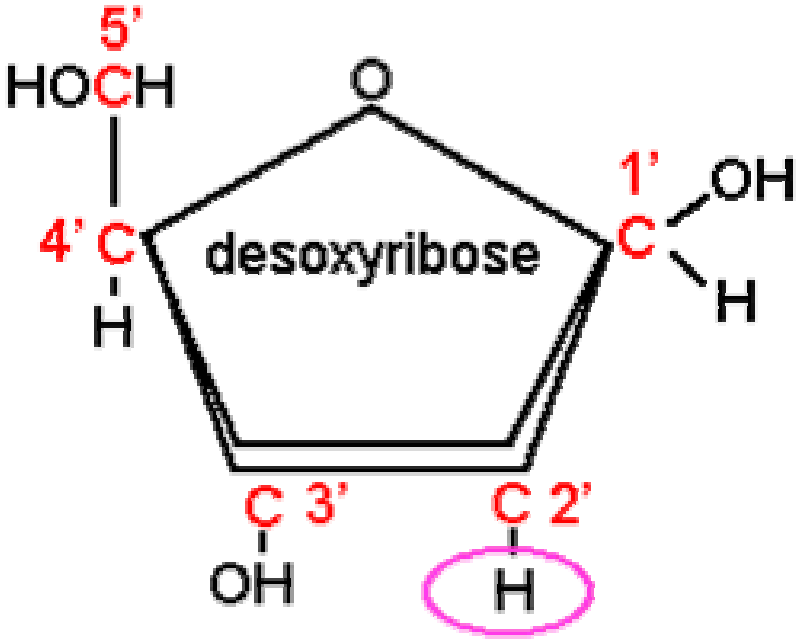


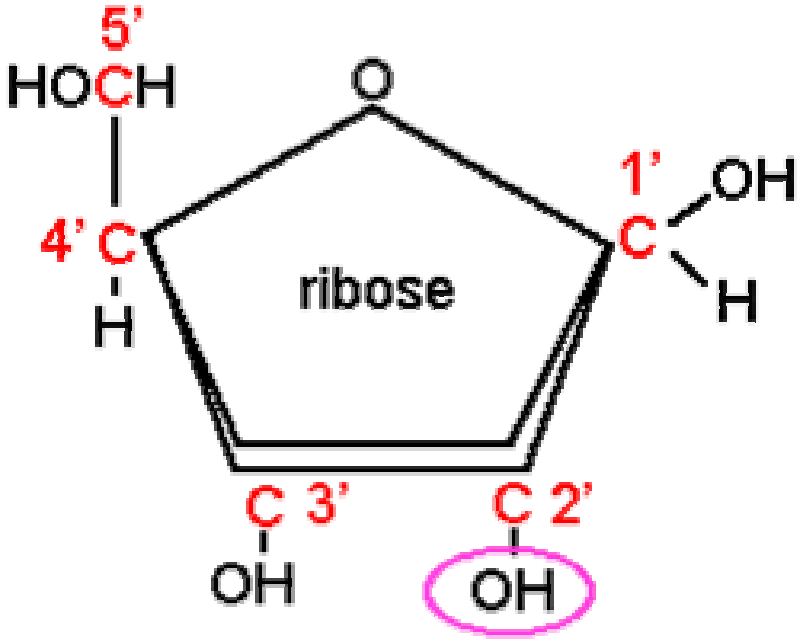
cytosine



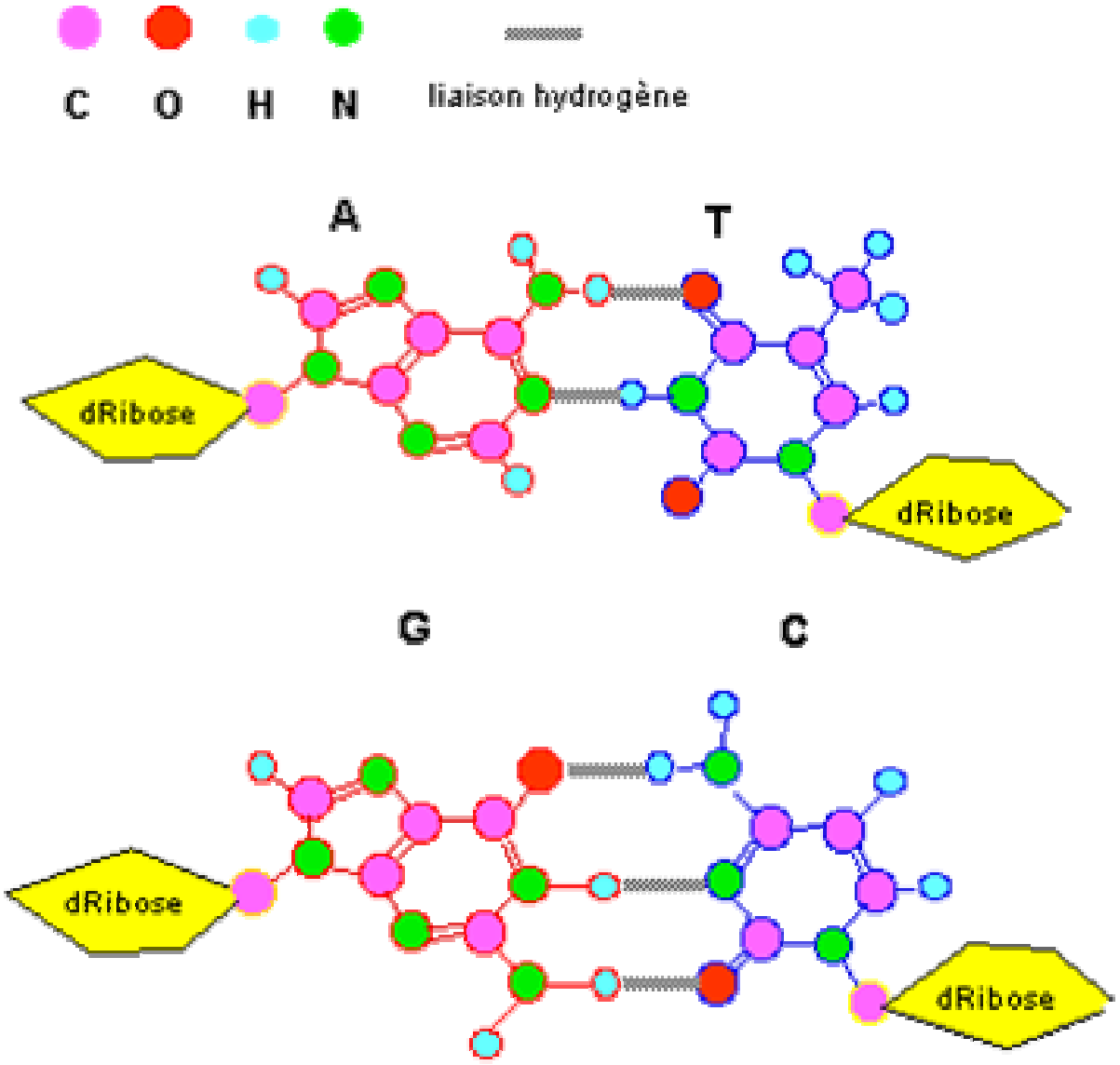
guanine

LAS PENTOSAS

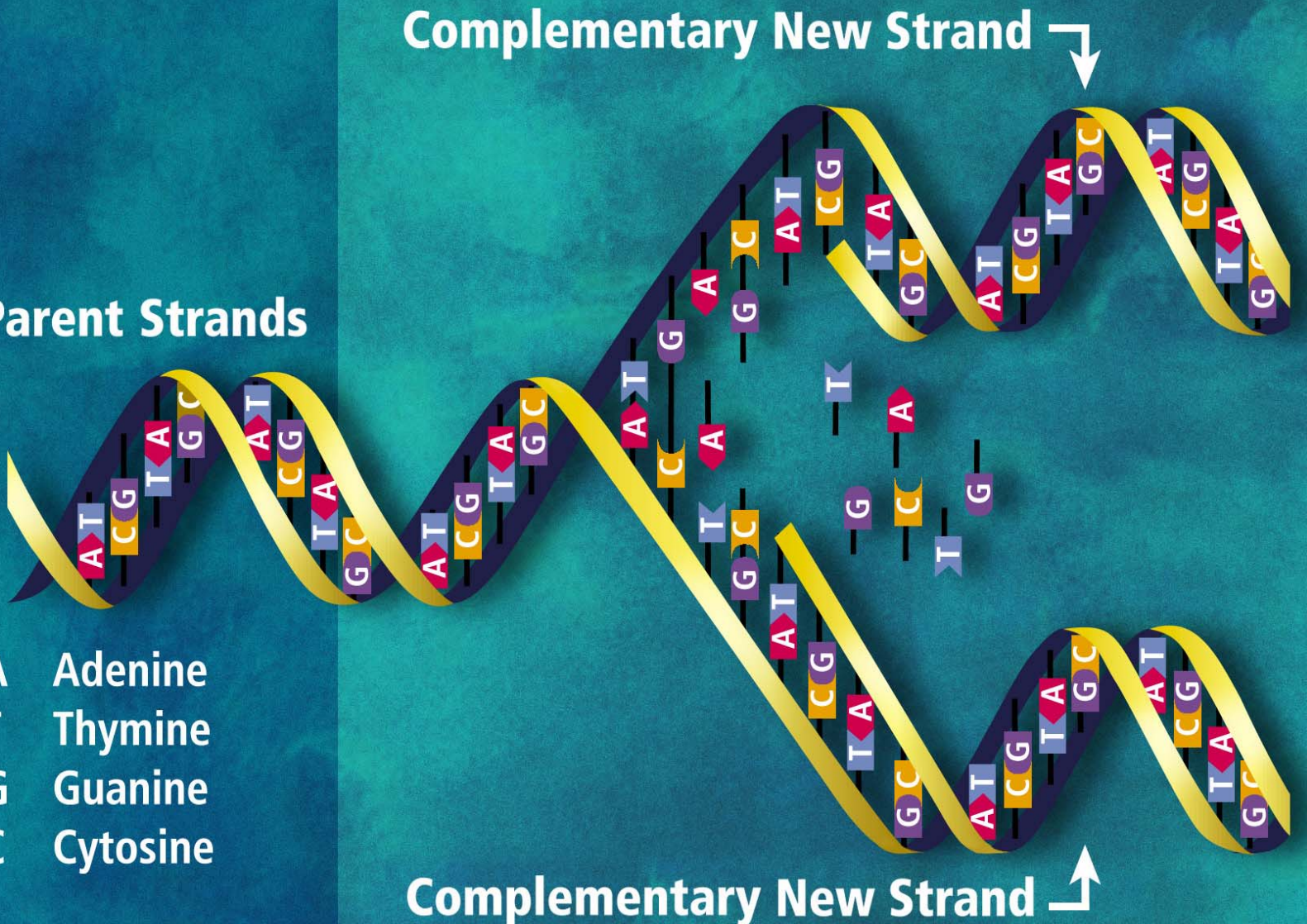




ENLACES DE H ENTRE LAS BASES N

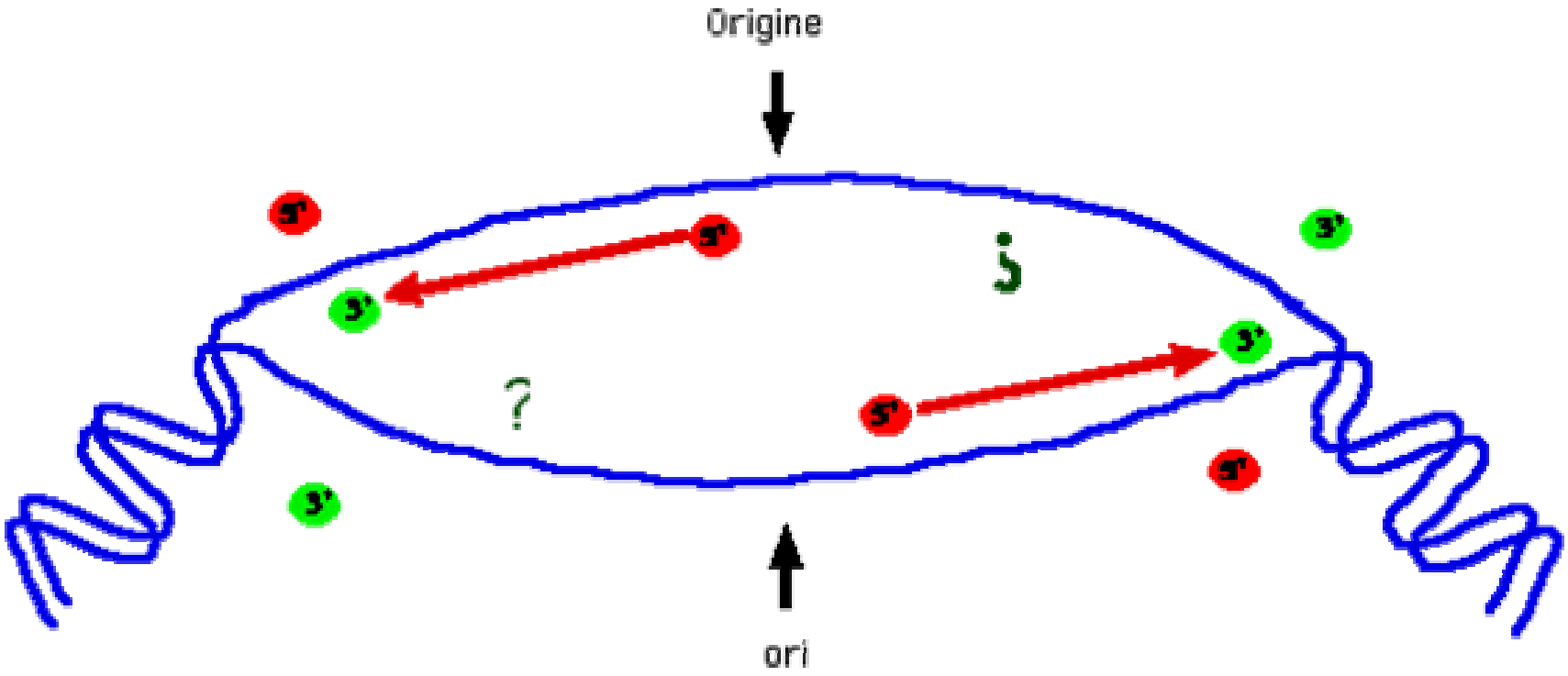


DNA Replication Prior to Cell Division

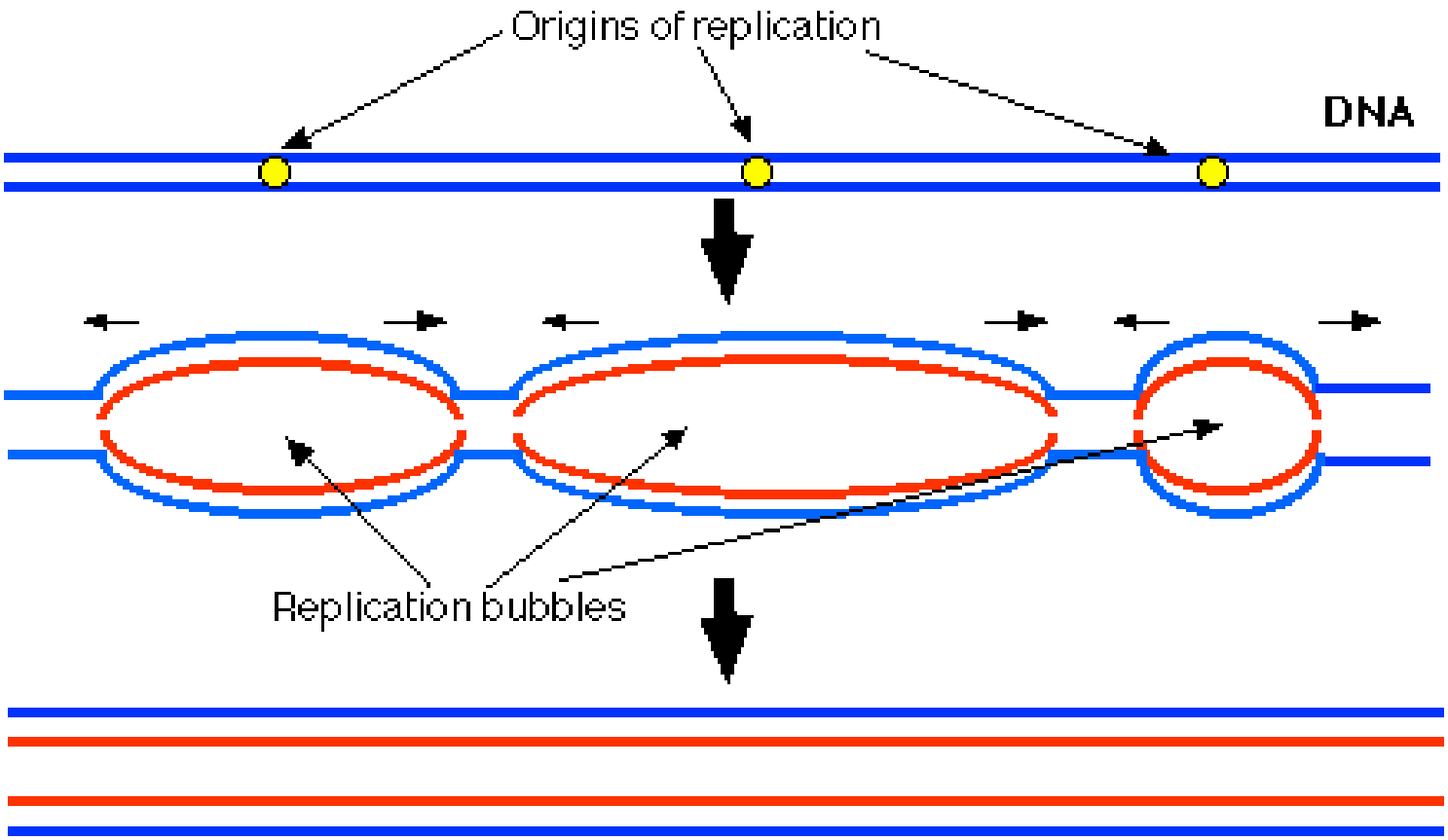


Y-GA 98-647

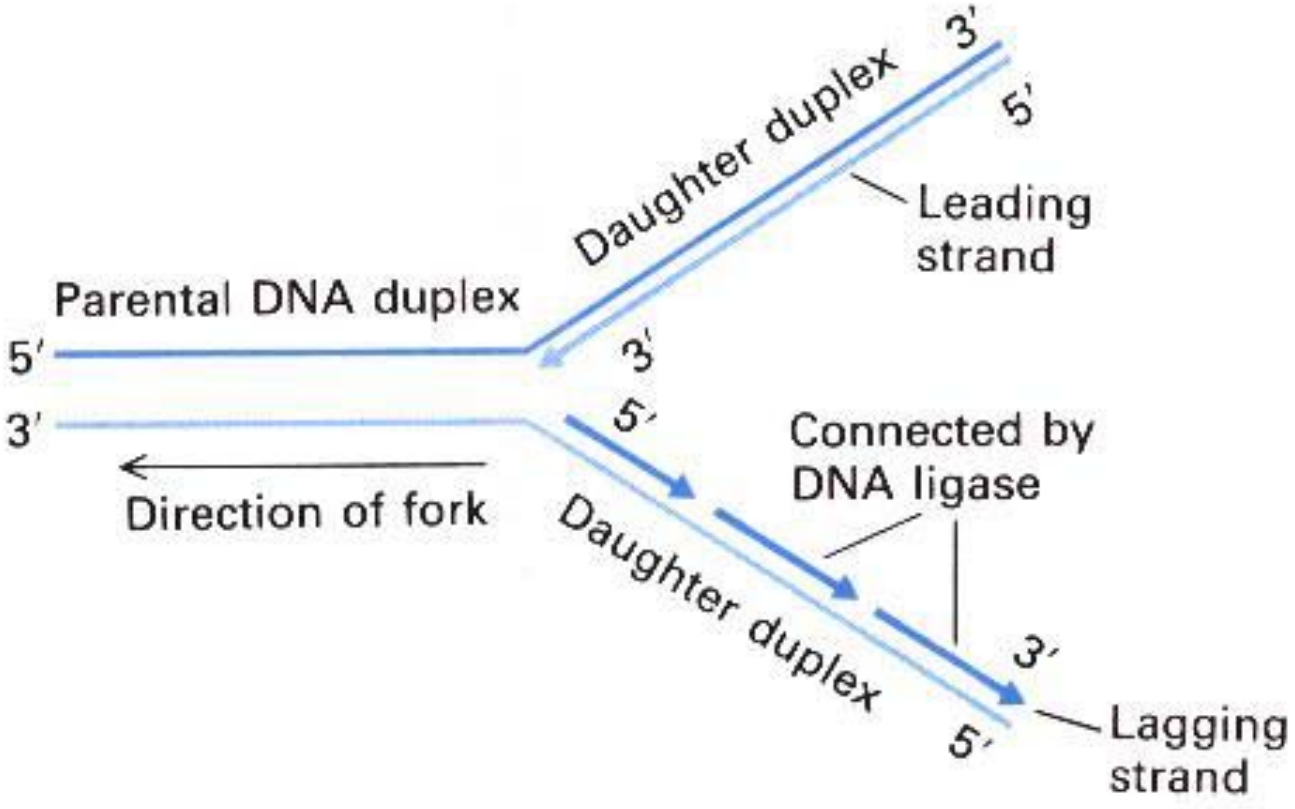
INICIO DE LA REPLICACION



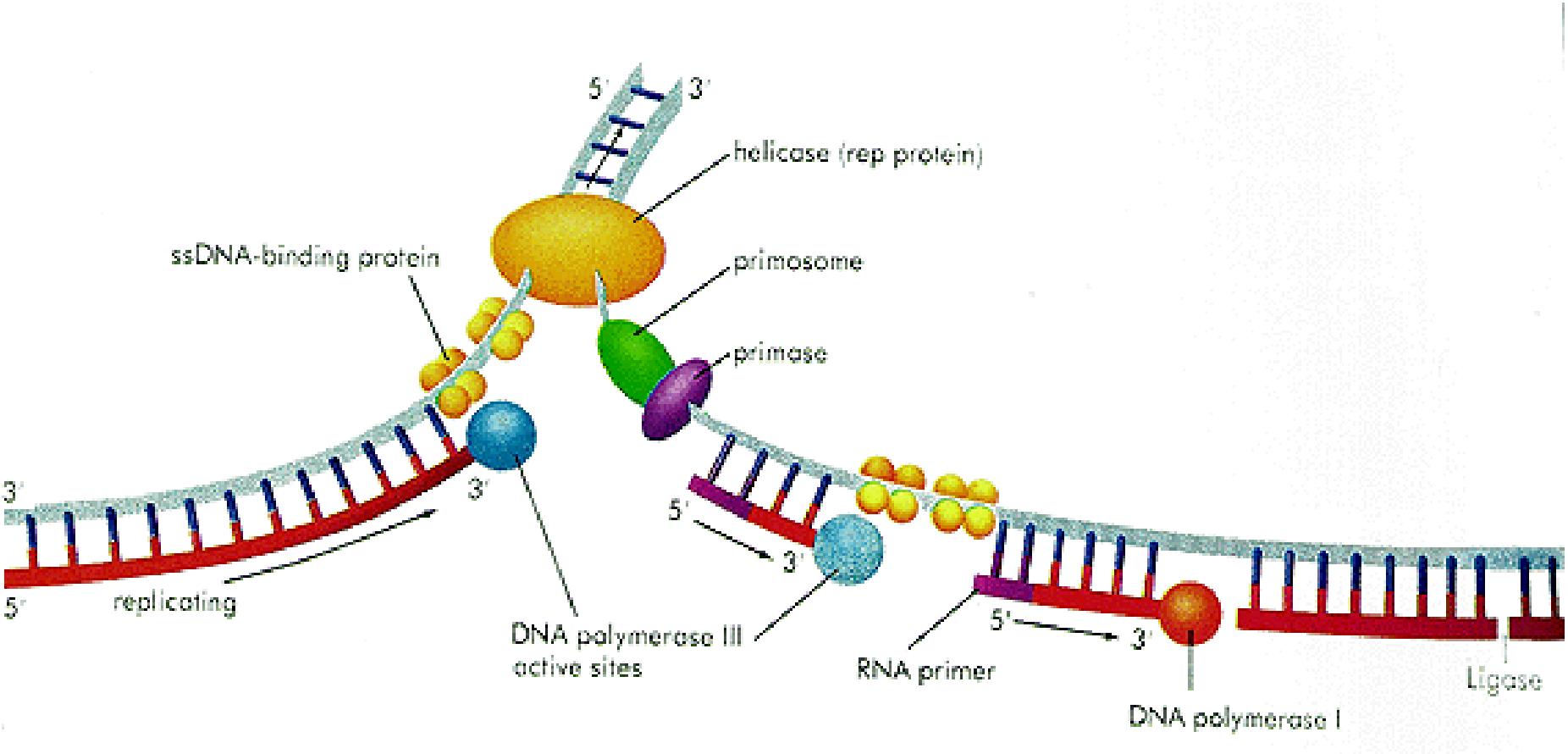
VARIOS SITIOS DE INICIO

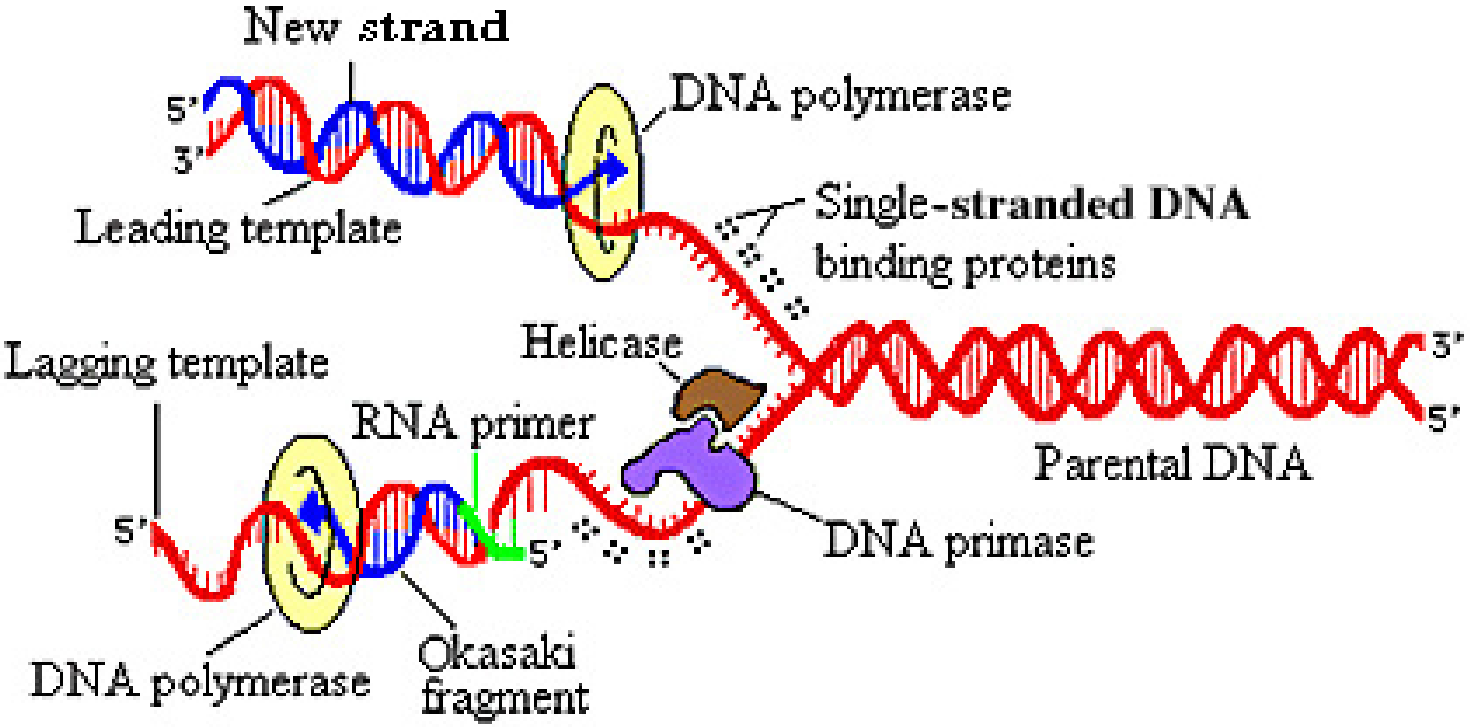


DIRECCION DE LA AUTODUPLICACION



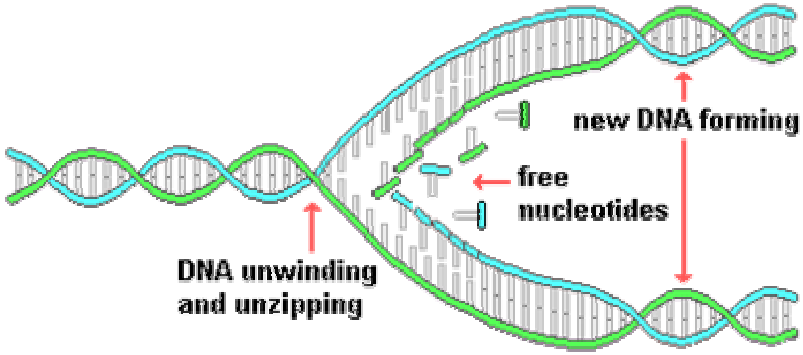
MOLECULAS QUE PARTICIPAN EN LA REPLICACION

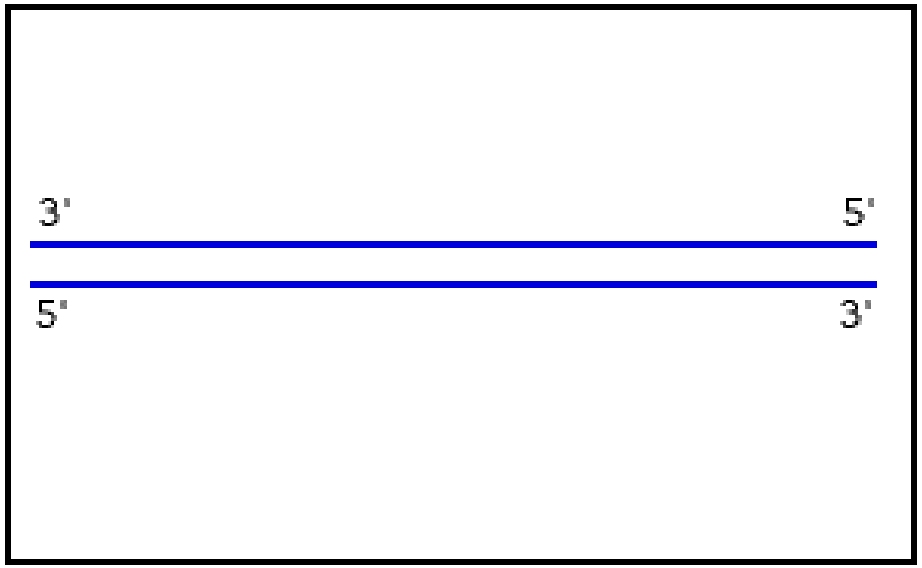


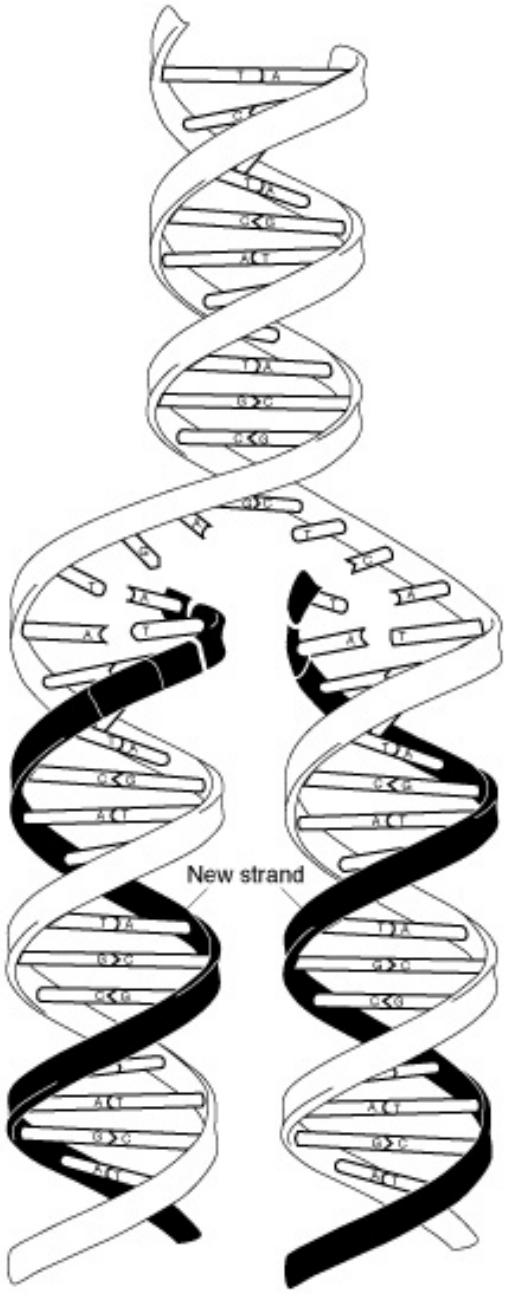


Collaboration of Proteins at the Replication Fork

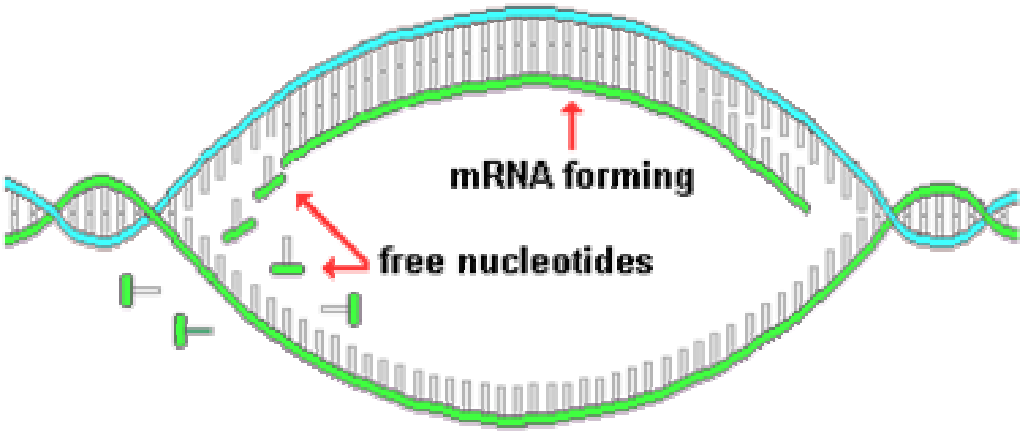
LA AUTODUPLICACION DEL ADN FINALIZA CON ⇒ 2 MOL IDENTICAS

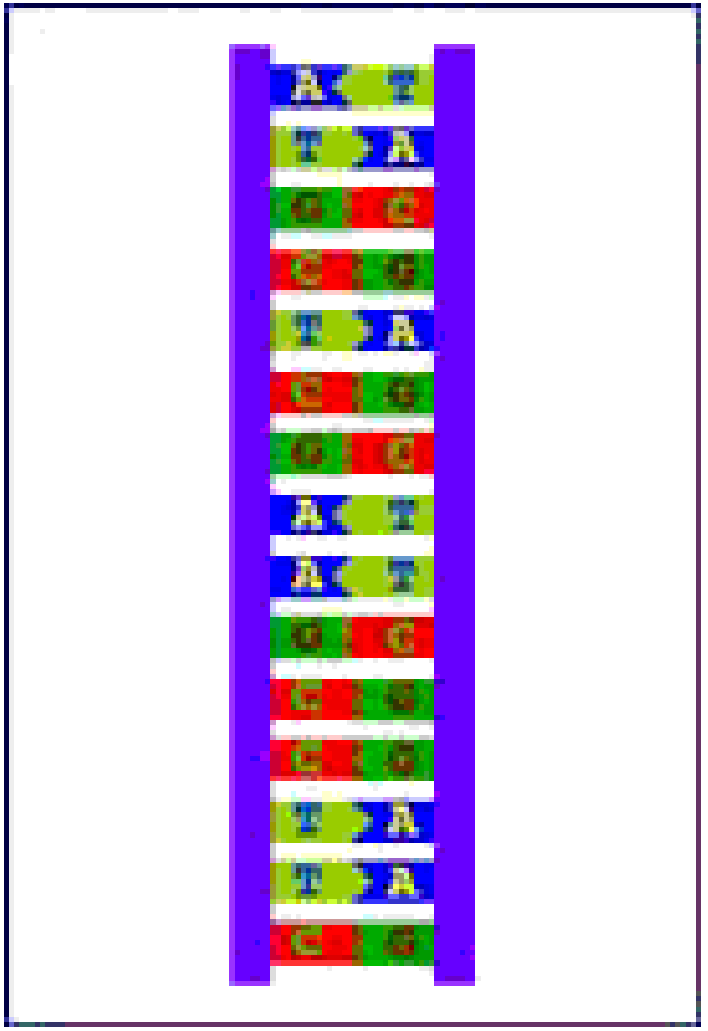




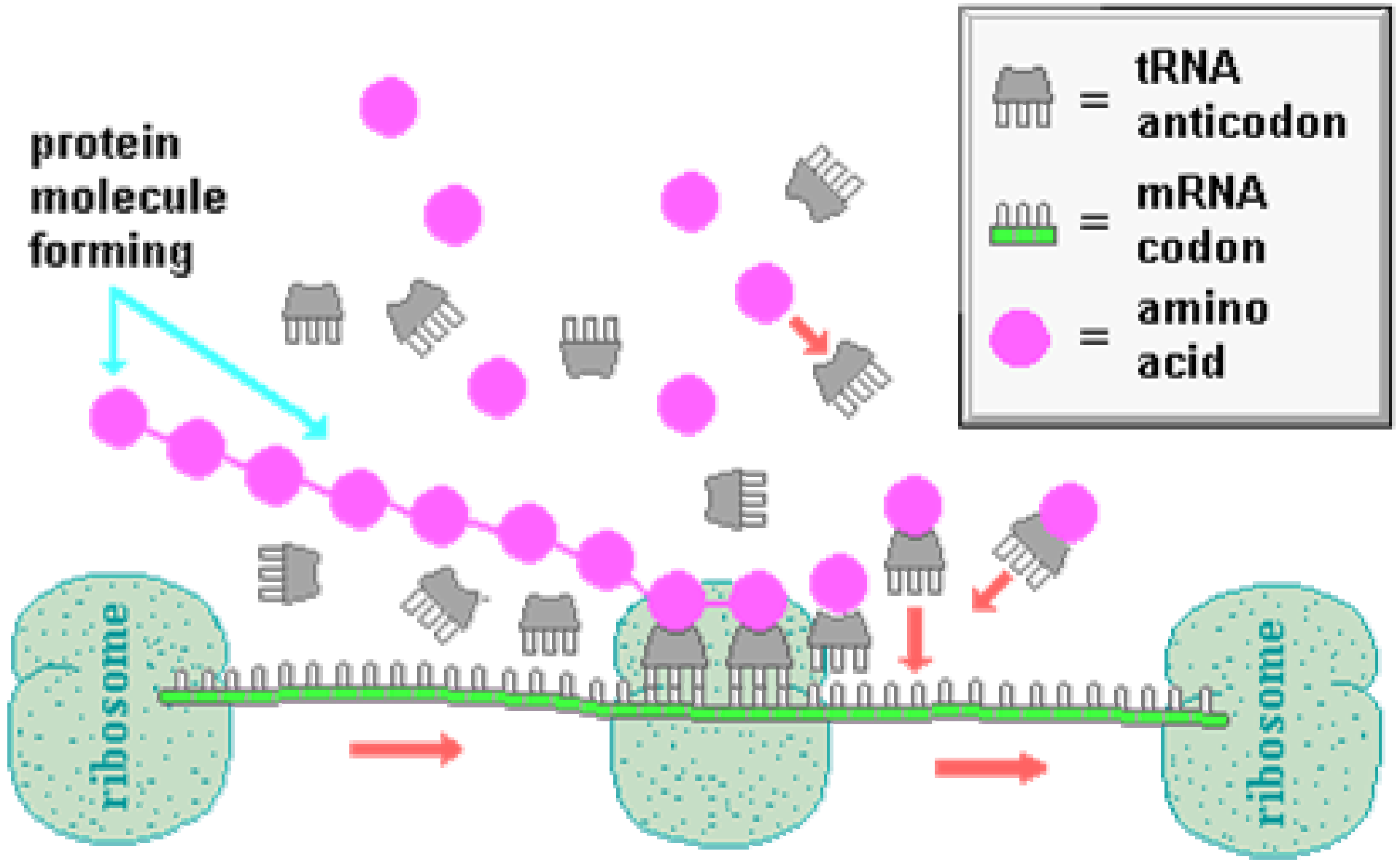


TRANSCRIPCION (SINTESIS DE ARN)

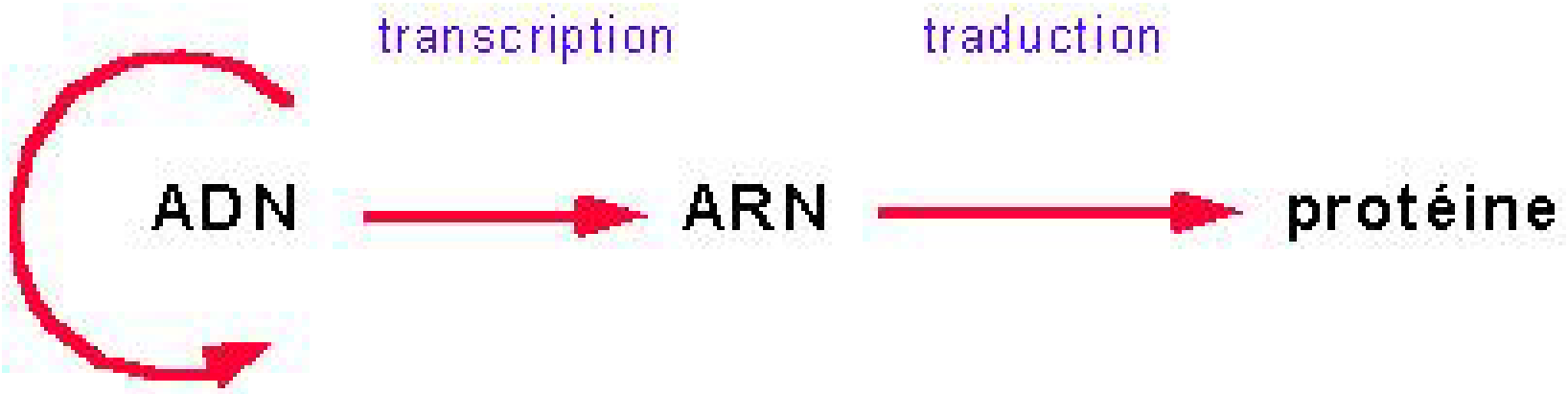




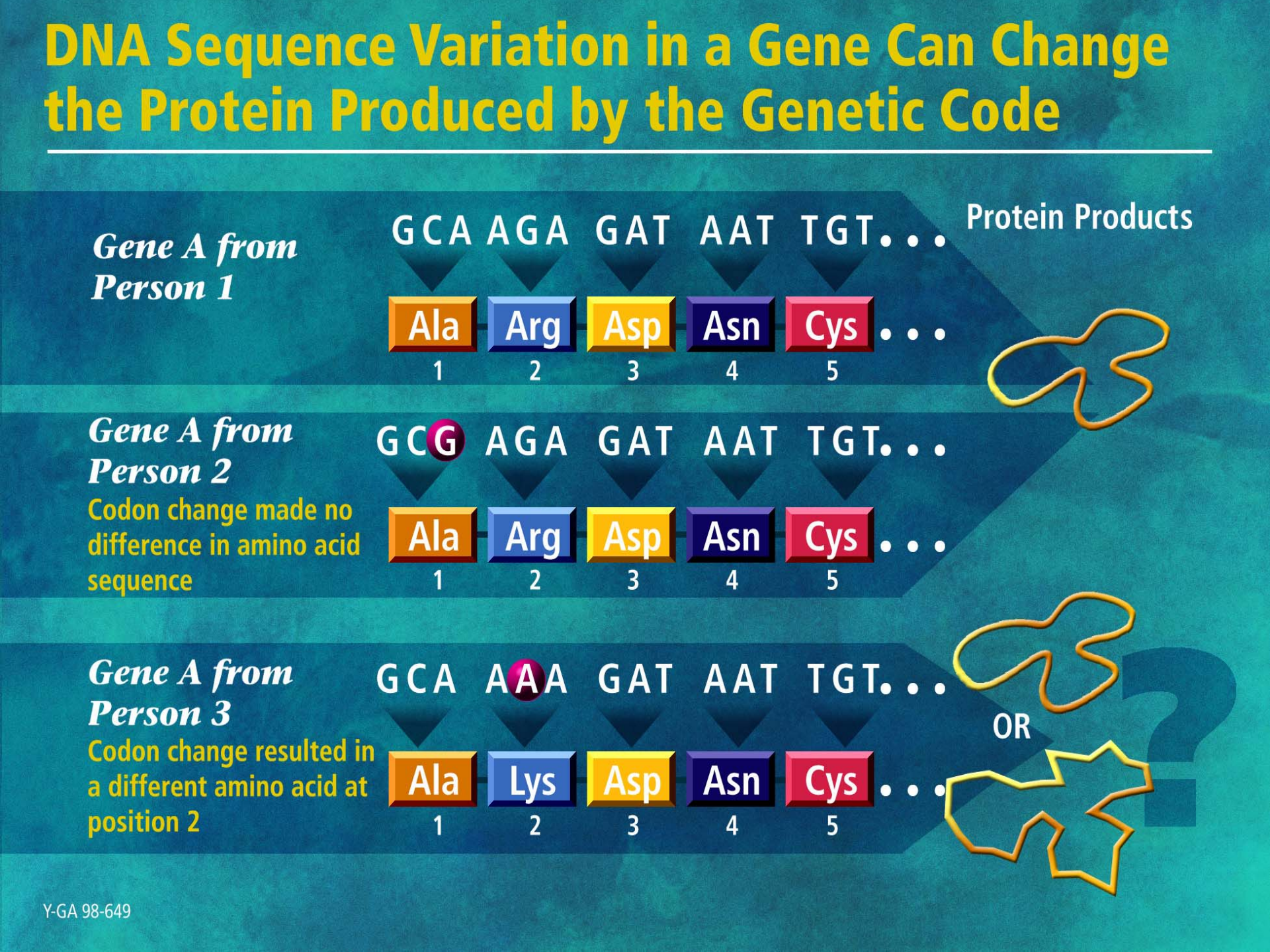
SINTESIS DE PROTEINAS



EL FLUJO DE LA INFORMACION GENETICA



DNA Sequence Variation in a Gene Can Change the Protein Produced by the Genetic Code



Y-GA 98-649

Health or Disease?

DNA Sequence

Person 1	A A A T T T	Normal protein
Person 2	A A T T T T	Some DNA variations have no negative effects
Person 3	A A C T T T	Low or nonfunctioning protein

Other variations lead to disease (e.g., sickle cell) or increased susceptibility to disease (e.g., lung cancer)

YGG-00-0480

ANEMIA FALCIFORME

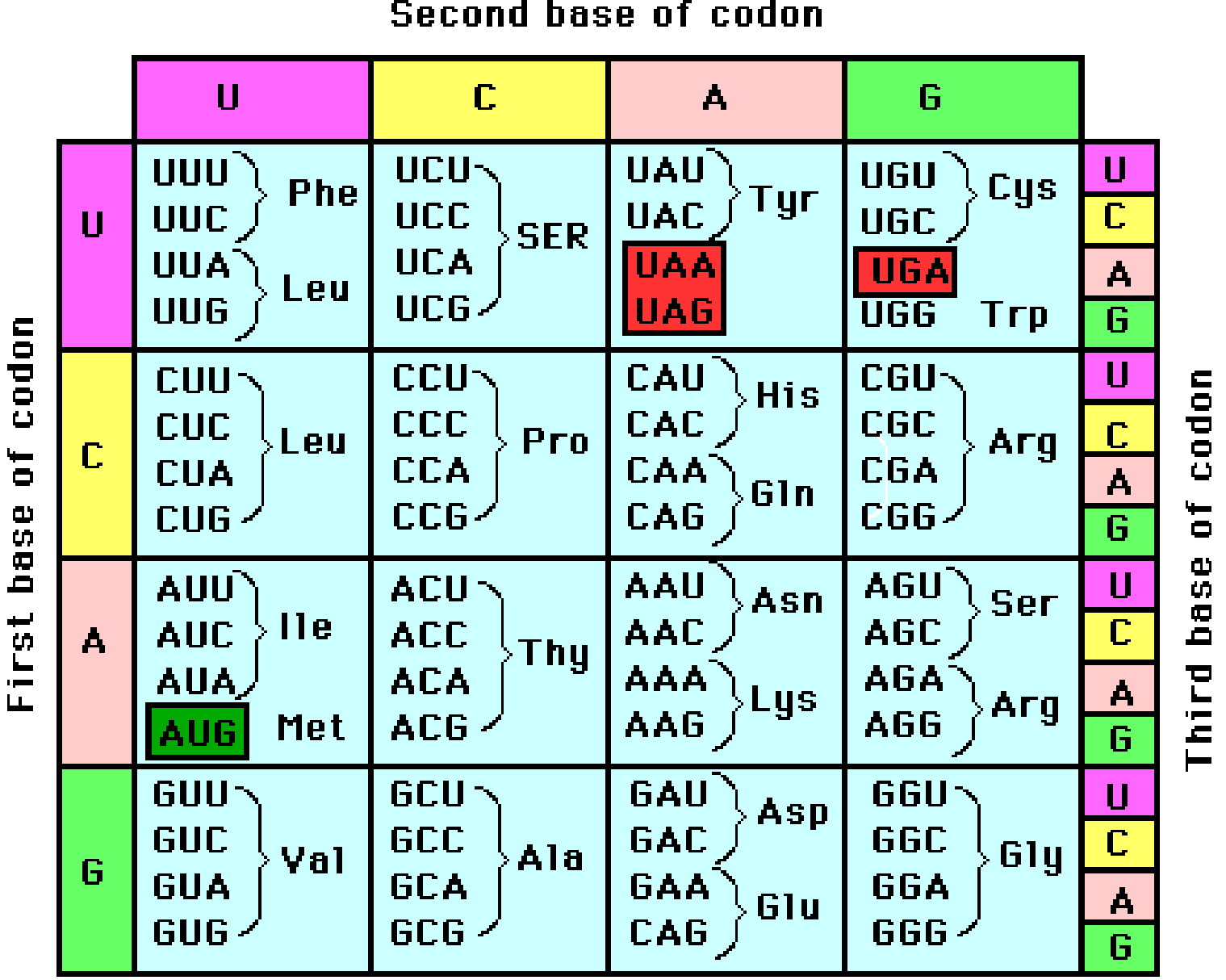
HBB Sequence in Normal Adult Hemoglobin (Hb A):

Nucleotide	CTG	ACT	CCT	GAG	GAG	AAG	TCT
Amino Acid	Leu	Thr	Pro	Glu	Glu	Lys	Ser
	3			6			9

HBB Sequence in Mutant Adult Hemoglobin (Hb S):

Nucleotide	CTG	ACT	CCT	GTG	GAG	AAG	TCT
Amino Acid	Leu	Thr	Pro	Val	Glu	Lys	Ser
	3			6			9

EL CODIGO GENE TICO



The genetic code, written by convention in the form in which the Codons appear in mRNA. The three terminato codons, UAA, UAG, and UGA, are boxed in red; the AUG initiator codon is shown in green

The genetic code arranged according to the pattern of degeneracy

Amino Acids with one Codon

AUG	UGG
Met	Trp

Amino Acids with two codons

AAA AAG	AAC AAU	CAA CAG	CAC CAU	GAA GAG	GAC GAU	UAC UAU	UGC UGU	UUC UUU
Lys	Asn	Gln	His	Glu	Asp	Tyr	Cys	Phe

Amino acid with three codons

AUA AUC AUU
Ile

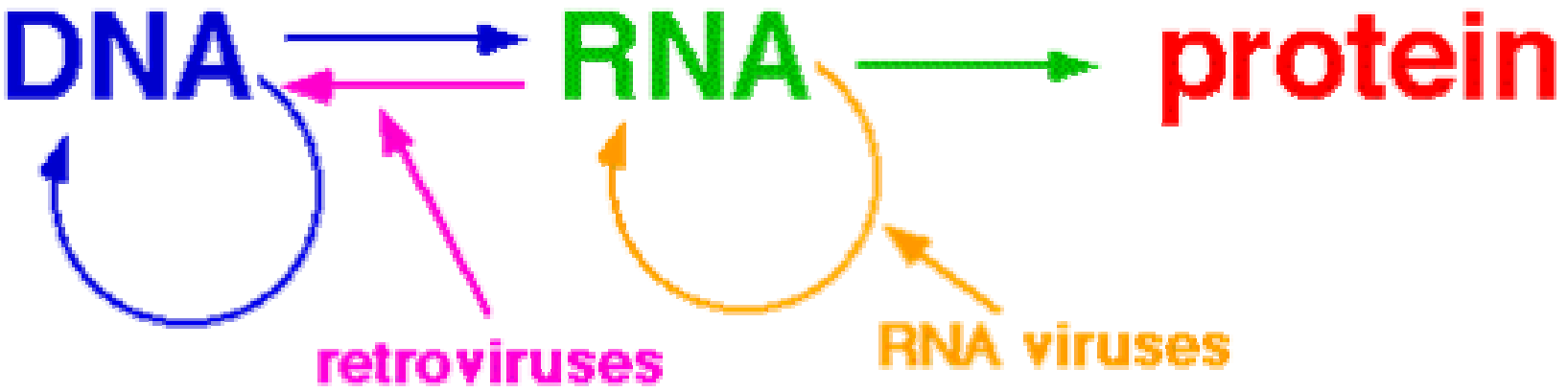
Amino Acids with four codons

ACA ACC ACG ACU	CCA CCC CCG CCU	GCA GCC GCG GCU	GGA GGC GGG GGU	GUA GUA GUG GUU
Thr	Pro	Ala	Gly	Val

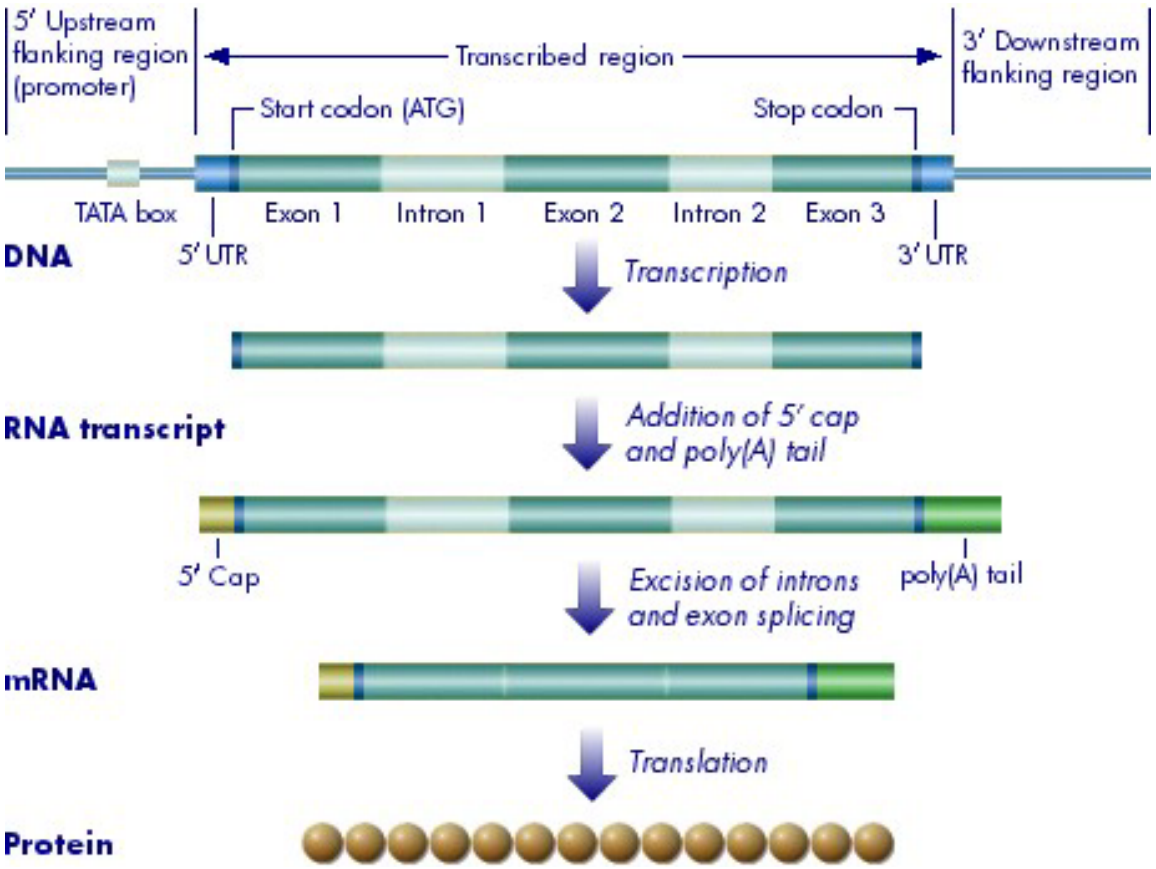
Aminio Acid with four codons

CGA CGC CGG CGU AGA AGG	GUA CUC CUG CUU UUA UUG	UCA UCC UCG UCU AGC AGU
Arg	Leu	Ser

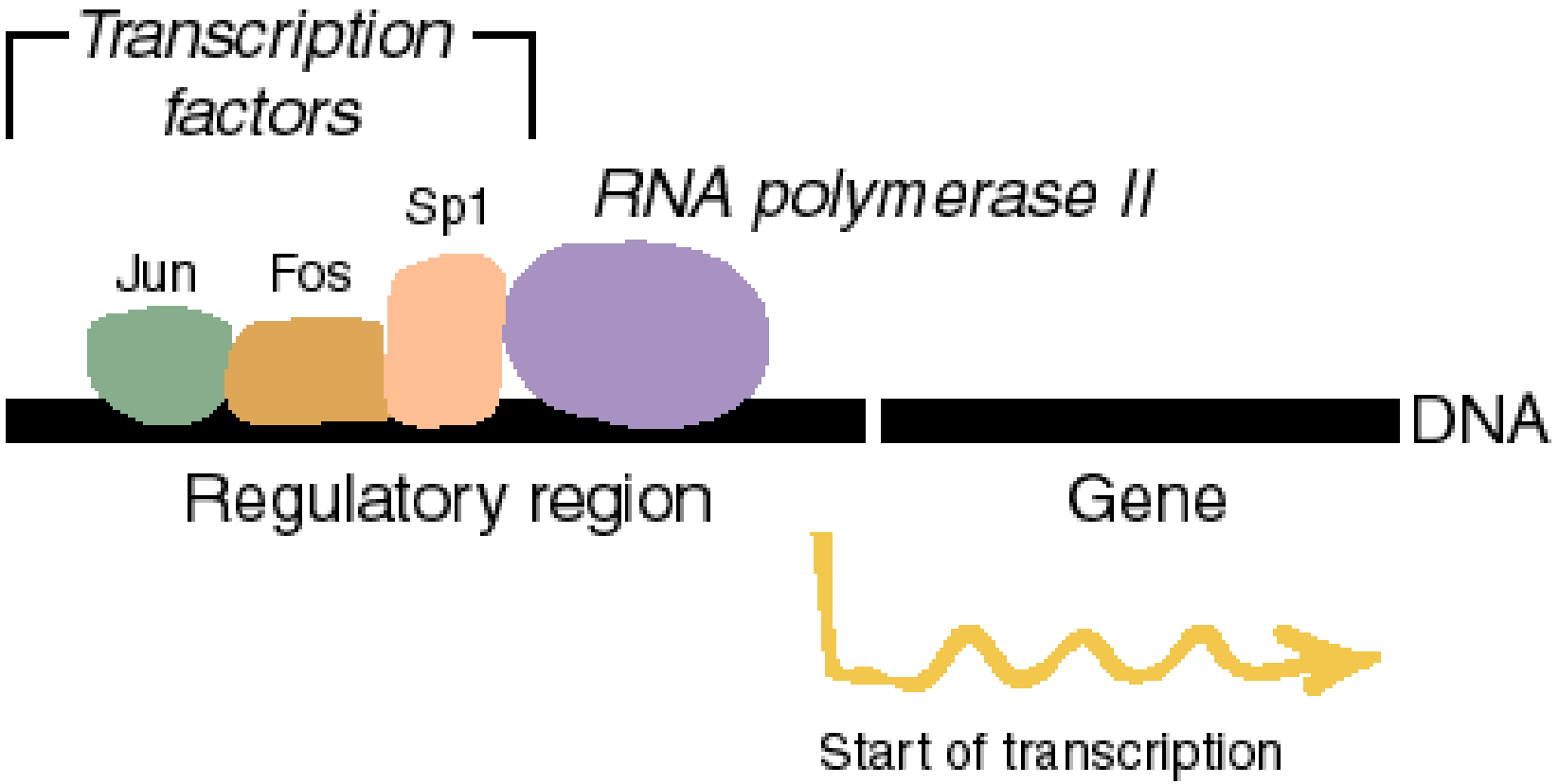
MODIFICACION DEL DOGMA CENTRAL



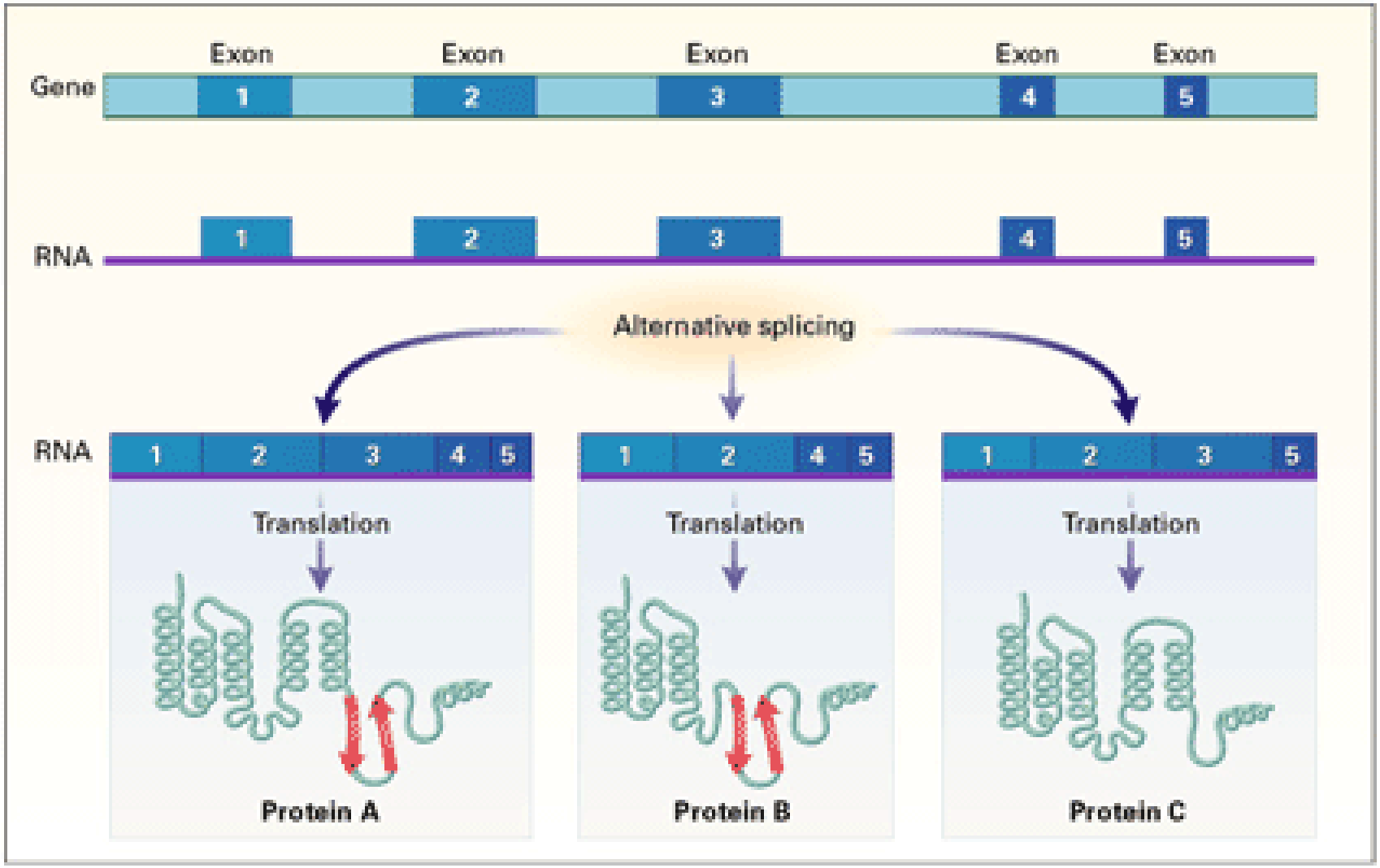
ESTRUCTURA GENICA



REGULACION DE LA EXPRESION GENICA

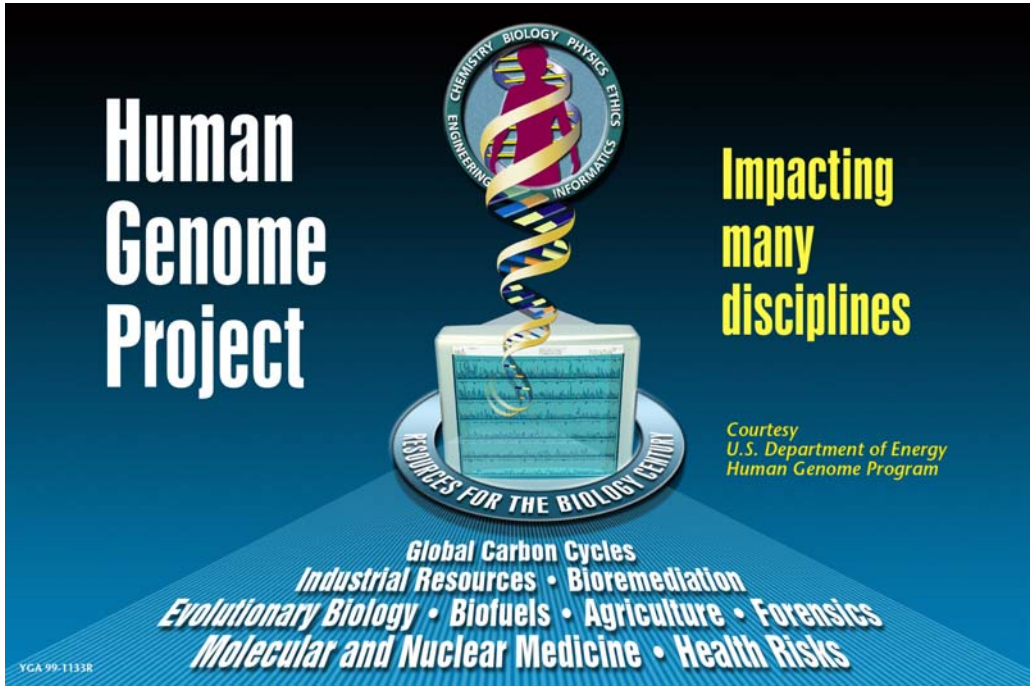


SPLICING ALTERNATIVO



EL GRAN PROYECTO DEL GENOMA HUMANO

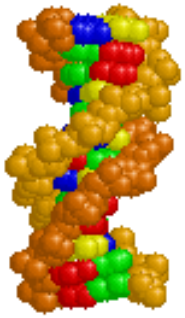
- Nació oficialmente en 1990
- Objetivos iniciales a ser alcanzados en 15 años.
- Creado, coordinado y financiado por el DOE y los NHI de los EE. UU.



OBJETIVOS INICIALES

- ***Determinación de la secuencia nucleotídica completa.***
- ***Identificación de todos los genes humanos. (30 000 actualmente)***
- ***Organizar la información resultante en bases de datos de fácil acceso.***
- ***Mejorar las metodologías del análisis genético.***
- ***Transferir las nuevas tecnologías al sector privado.***
- ***Dirigir la política legal, ética y social surgida del proyecto (ELSI).***

OBJETIVOS ALCANZADOS ANTES DEL 2005



- 1999: Secuencia completa del cromosoma 22
- 2000: Secuencia completa del cromosoma 21
- 2001: Borrador de la secuencia total de bases N
- 2003: Secuencia completa en limpio
- 2003: Secuencia completa del cromosoma 6
- NACIMIENTO DE NUEVOS PROGRAMAS:

  GENOMES TO LIFE

  HUPO

  GENOMAS DE OTROS ORGANISMOS:

  Mus musculus, C. Elegans, A. thaliana, S. cerevisae, D. melanogaster

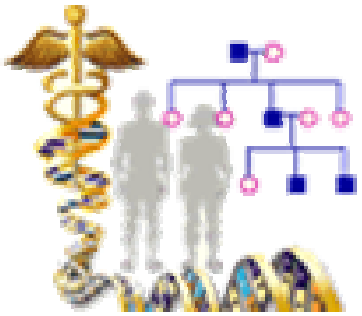
IMPLICANCIAS DEL PROYECTO DEL GENOMA HUMANO EN LA SALUD PUBLICA

LAS PRUEBAS GENETICAS. (ADN)

LA TERAPIA GENICA.

EL ACONSEJAMIENTO GENETICO.

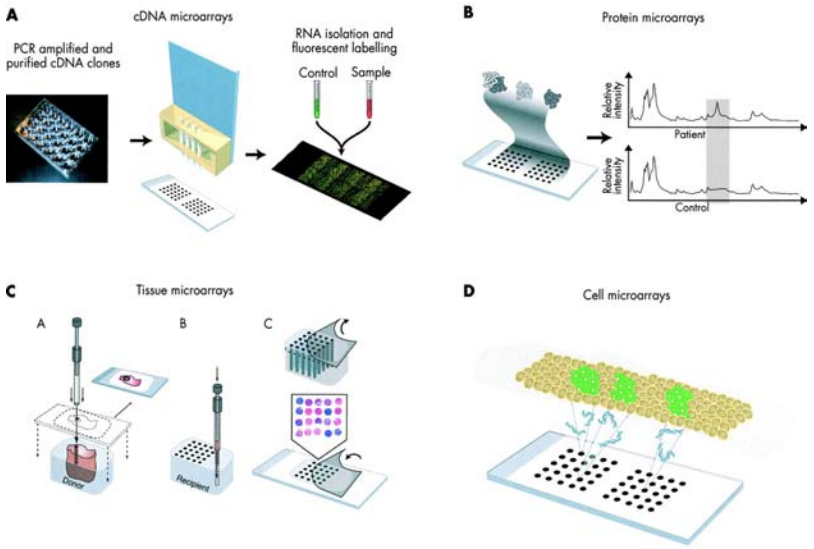
LA FARMACO – GENOMICA.



Genetics Home Reference

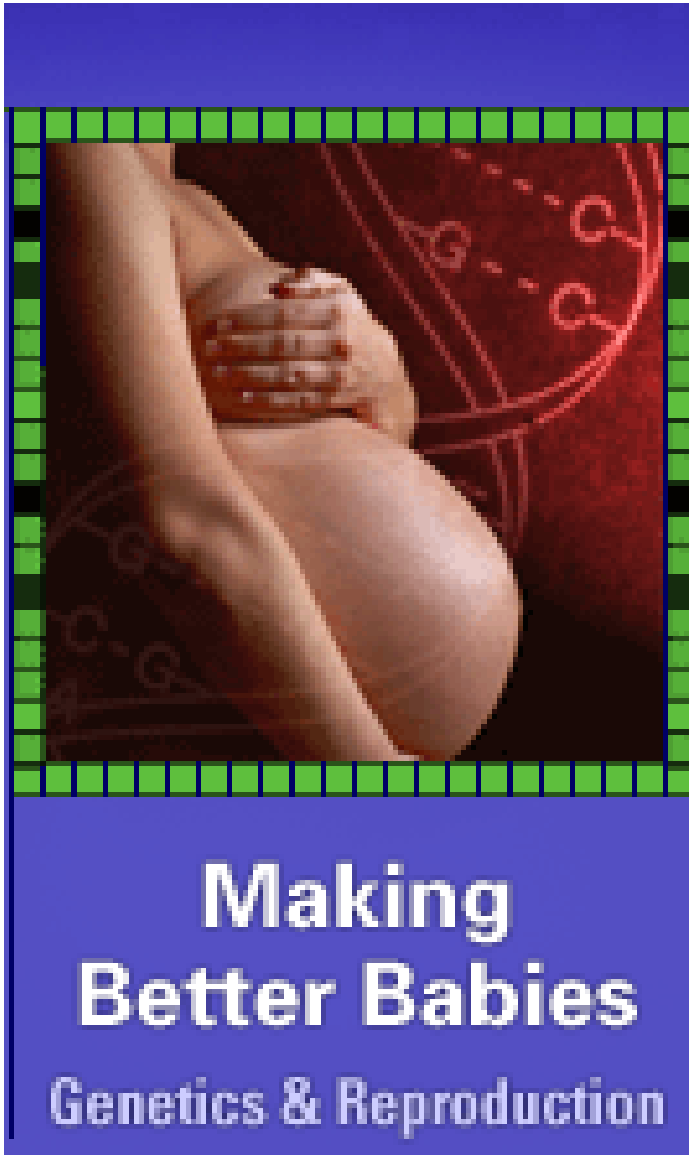
Your Guide to Understanding Genetic Conditions

PRUEBAS GENETICAS (ADN, ARN, PROTEINAS, CROMOSOMAS, METABOLITOS)



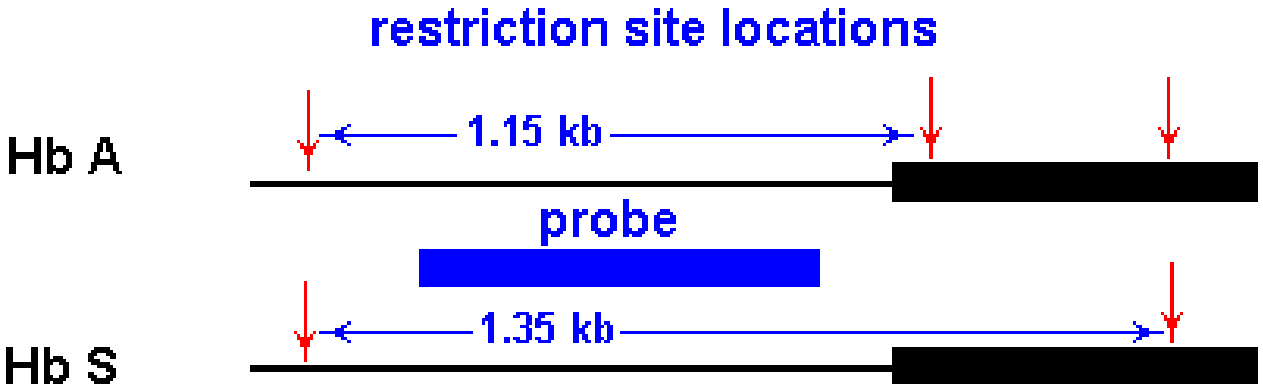
- **Screening de portadores sanos.**
- **Dx Pre natal.**
- **Screening recién nacidos**
- **Dx. en pre sintomáticos.**
- **Estimativa de riesgos.**
- **Confirmación de Dx.**
- **Pruebas forense/filiación**

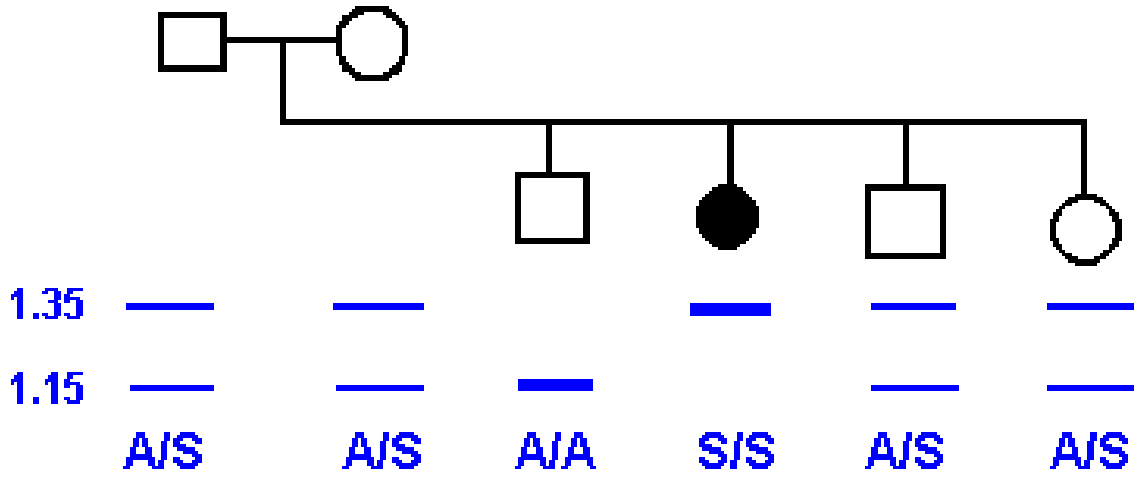
EL ACONSEJAMIENTO GENETICO



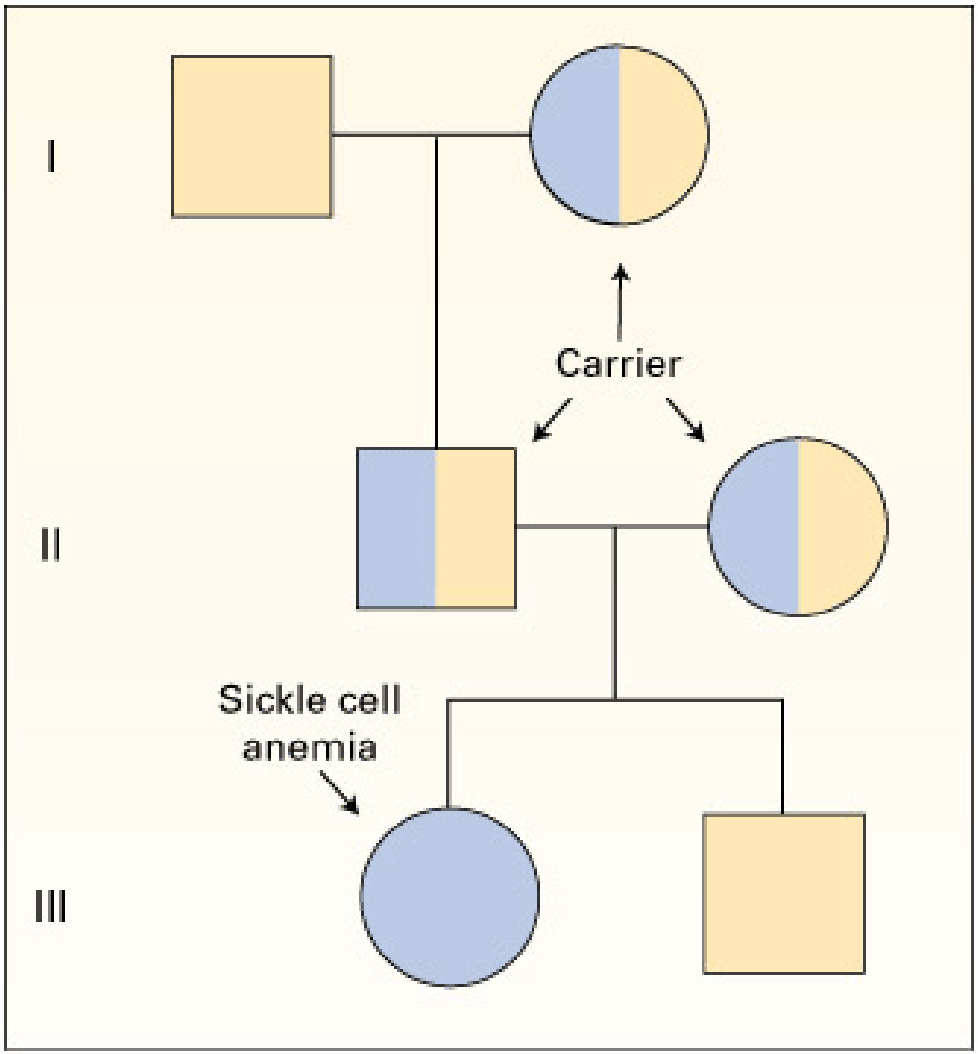
- Especialidad en pleno desarrollo:
- Prenatal.
- Clínica
- Comercial
- Educación Pública

DIAGNOSTICO MOLECULAR

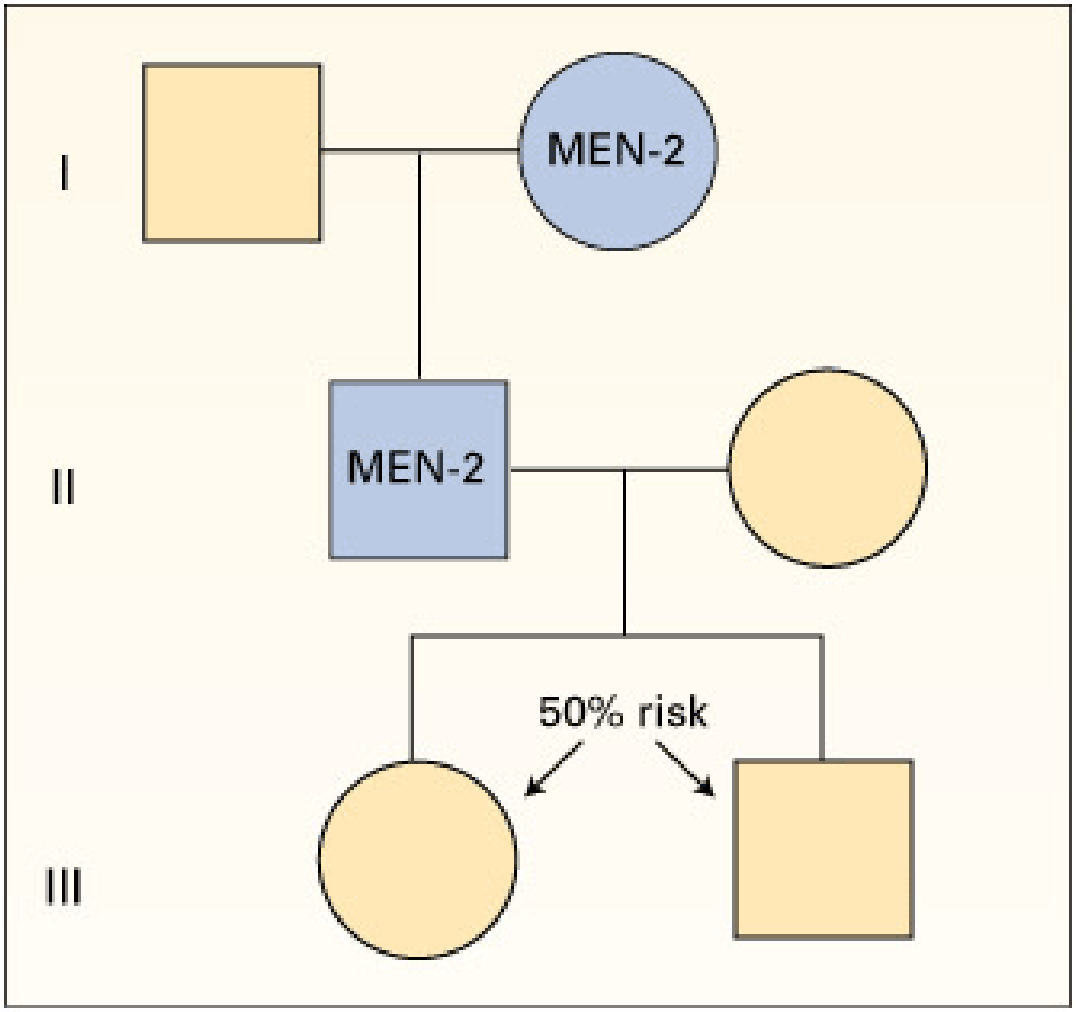




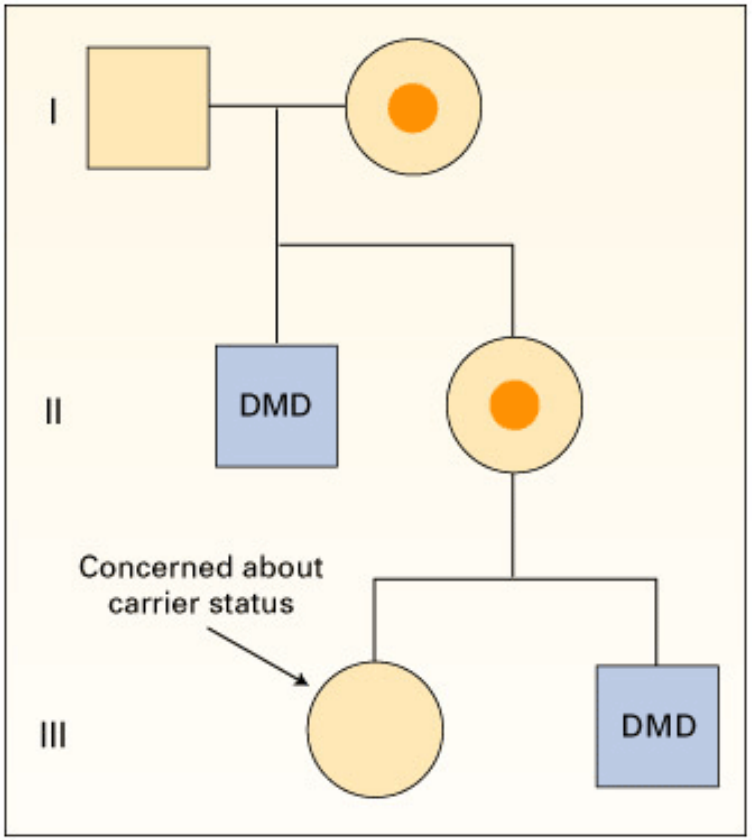
HERENCIA AUTOSOMICA RECESIVA

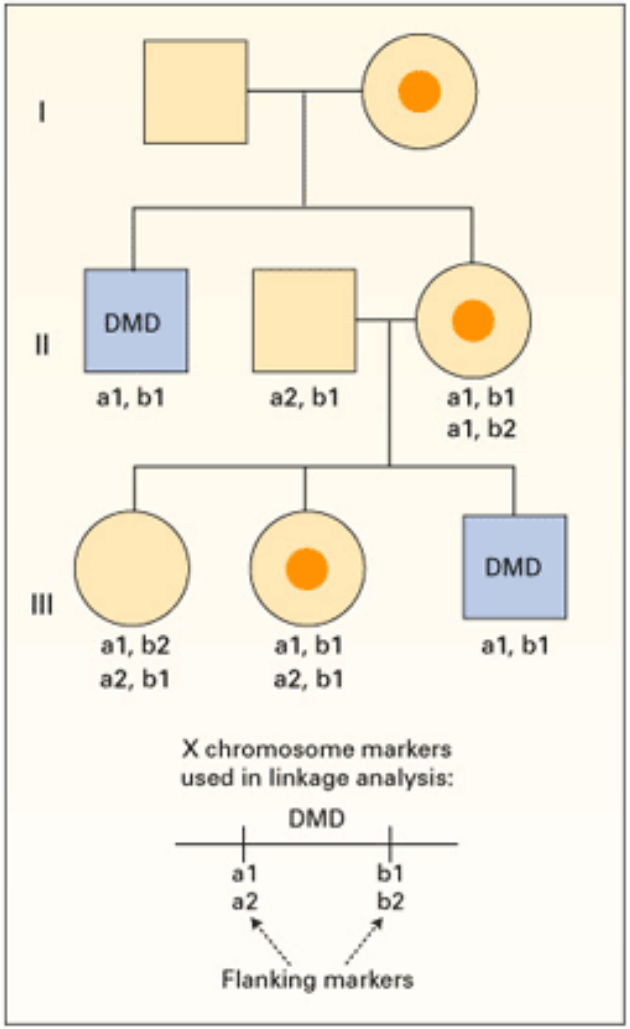


HERENCIA AUTOSOMICA DOMINANTE

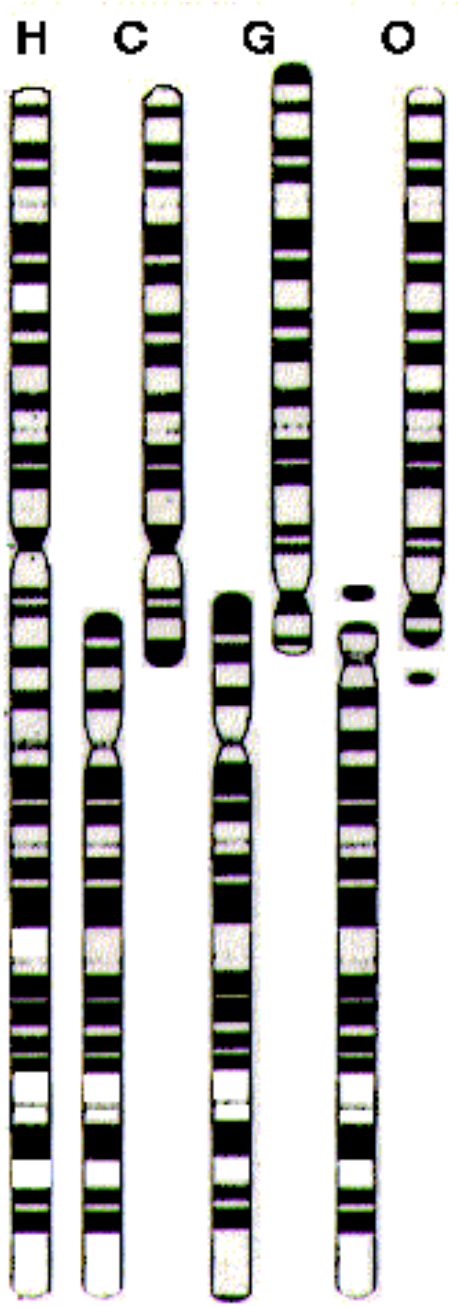


ESTIMATIVA DE RIESGO

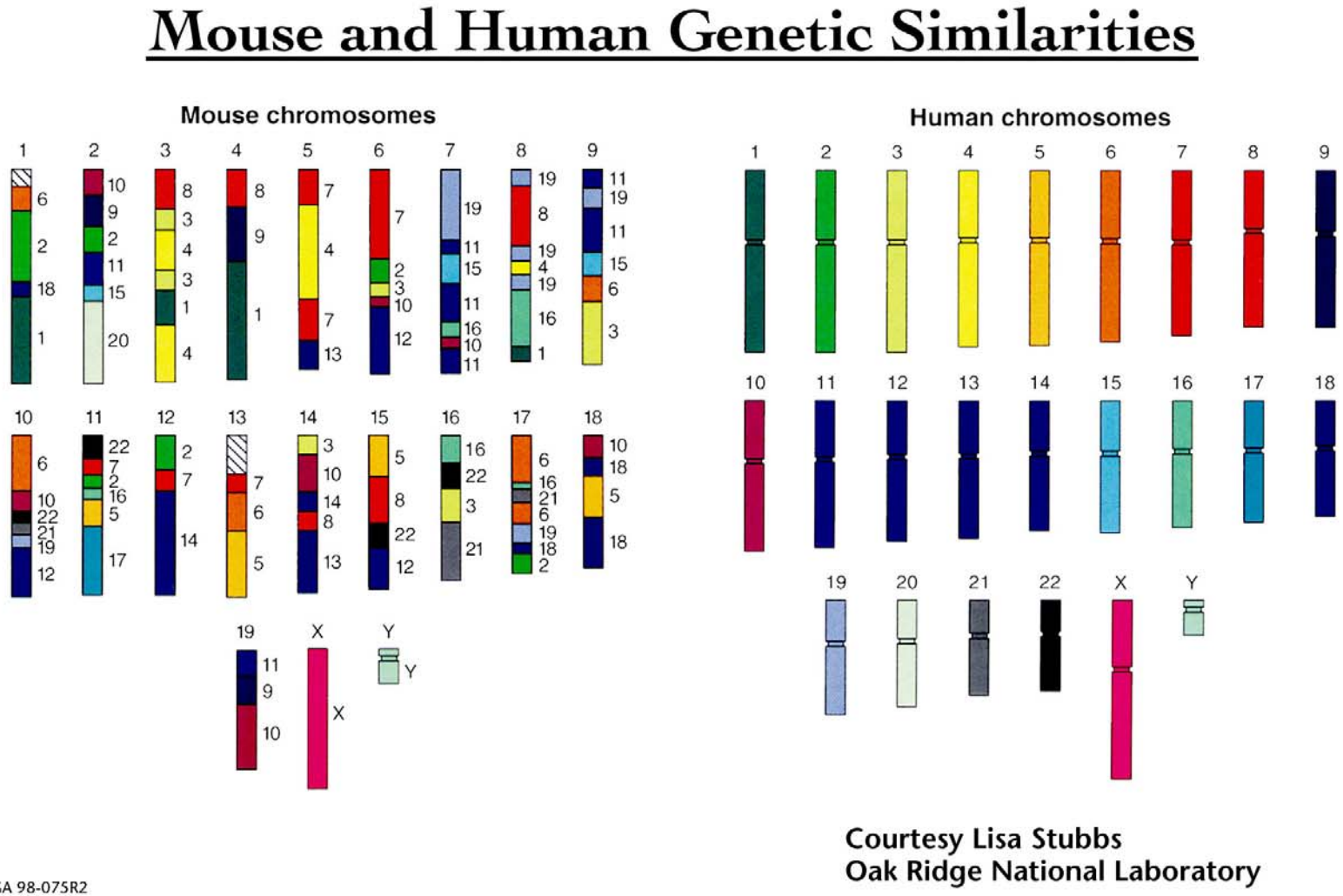




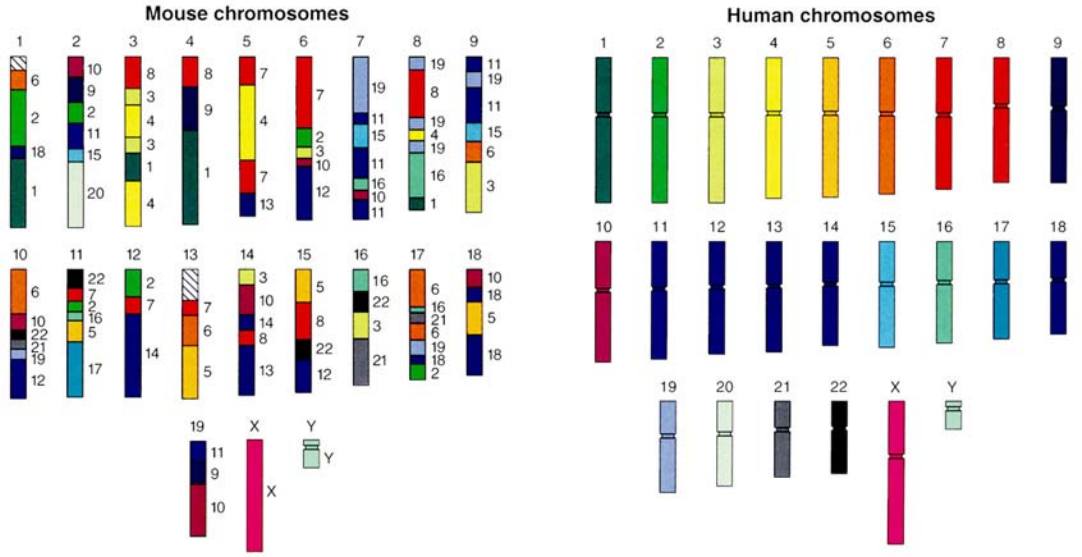
HOMOLOGIAS CROMOSOMICAS INTER ESPECIFICAS



HOMOLOGIAS CROMOSOMICAS (HOMBRE–RATON)



Mouse and Human Genetic Similarities



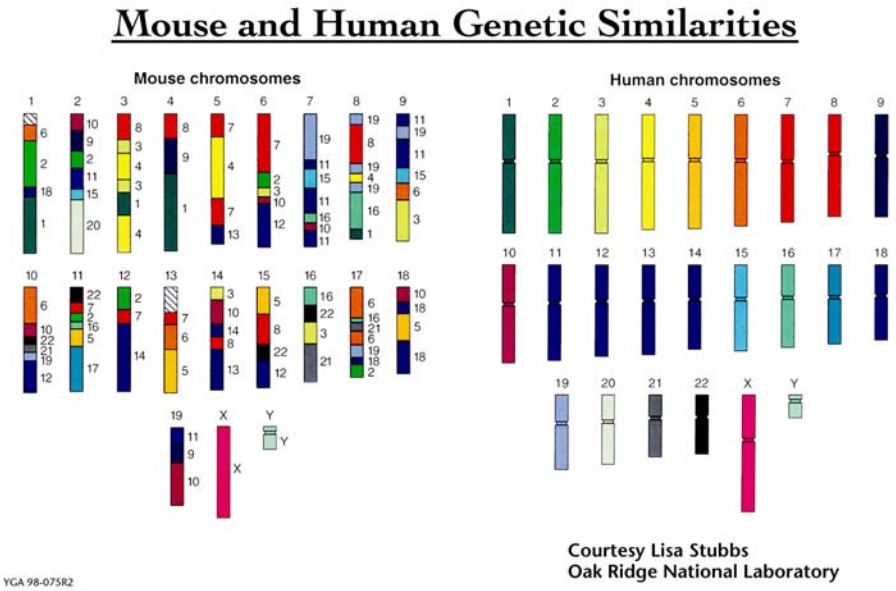
Courtesy Lisa Stubbs
Oak Ridge National Laboratory

YGA 98-075R2

OH, MY GOD!

QUE HORROR!

⇓


Mouse and Human Genetic Similarities
Mouse chromosomes
Human chromosomes
YGA 98-075R2
Courtesy Lisa Stubbs
Oak Ridge National Laboratory

MUCHAS GRACIAS!