

GENETIC DISORDERS & STRUCTURE OF DNA,

BY:-

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GENETIC DISORDERS

TYPES: MENDELIAN DISORDERS & CHROMOSOMAL DISORDERS

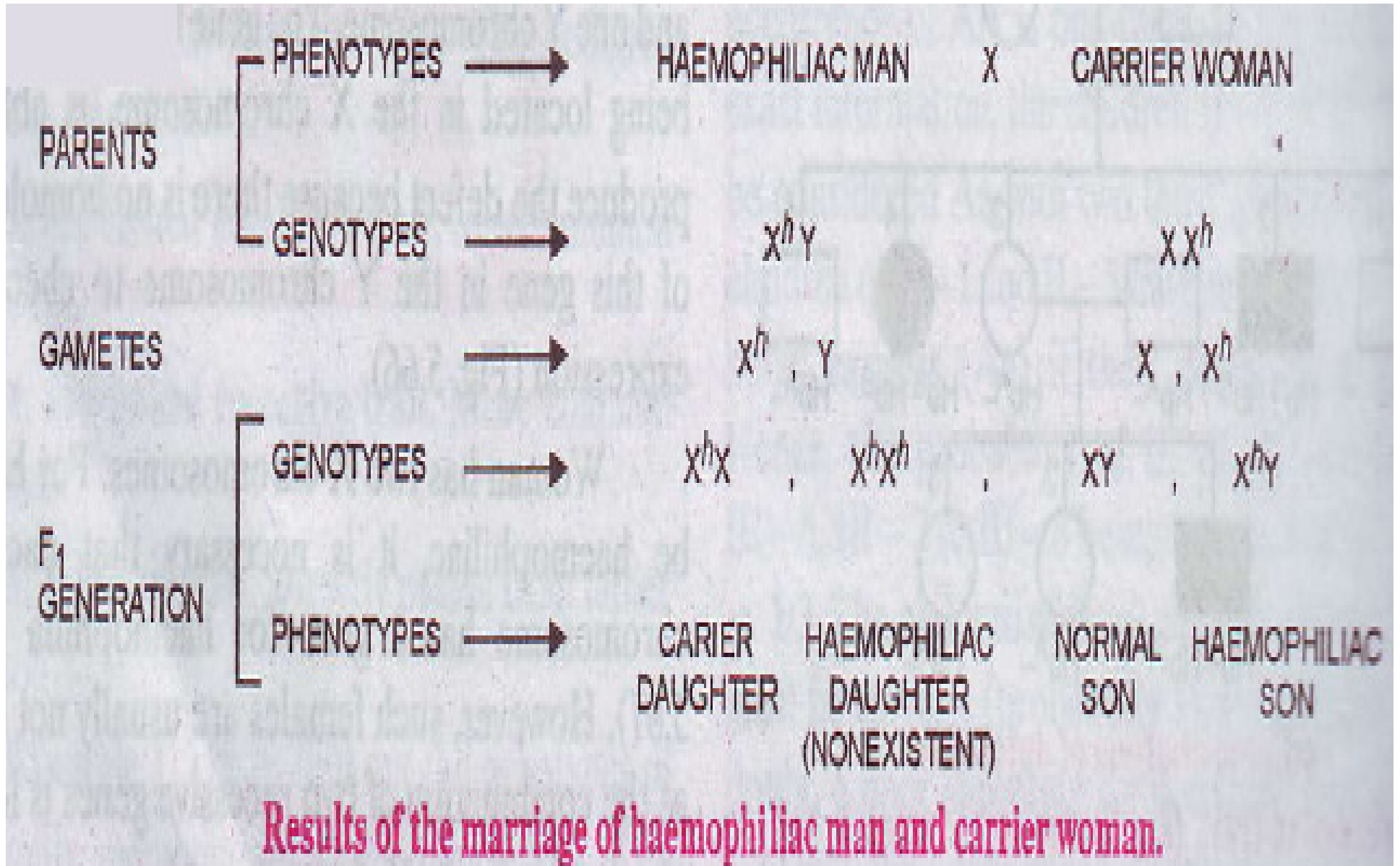
MENDELIAN DISORDERS:

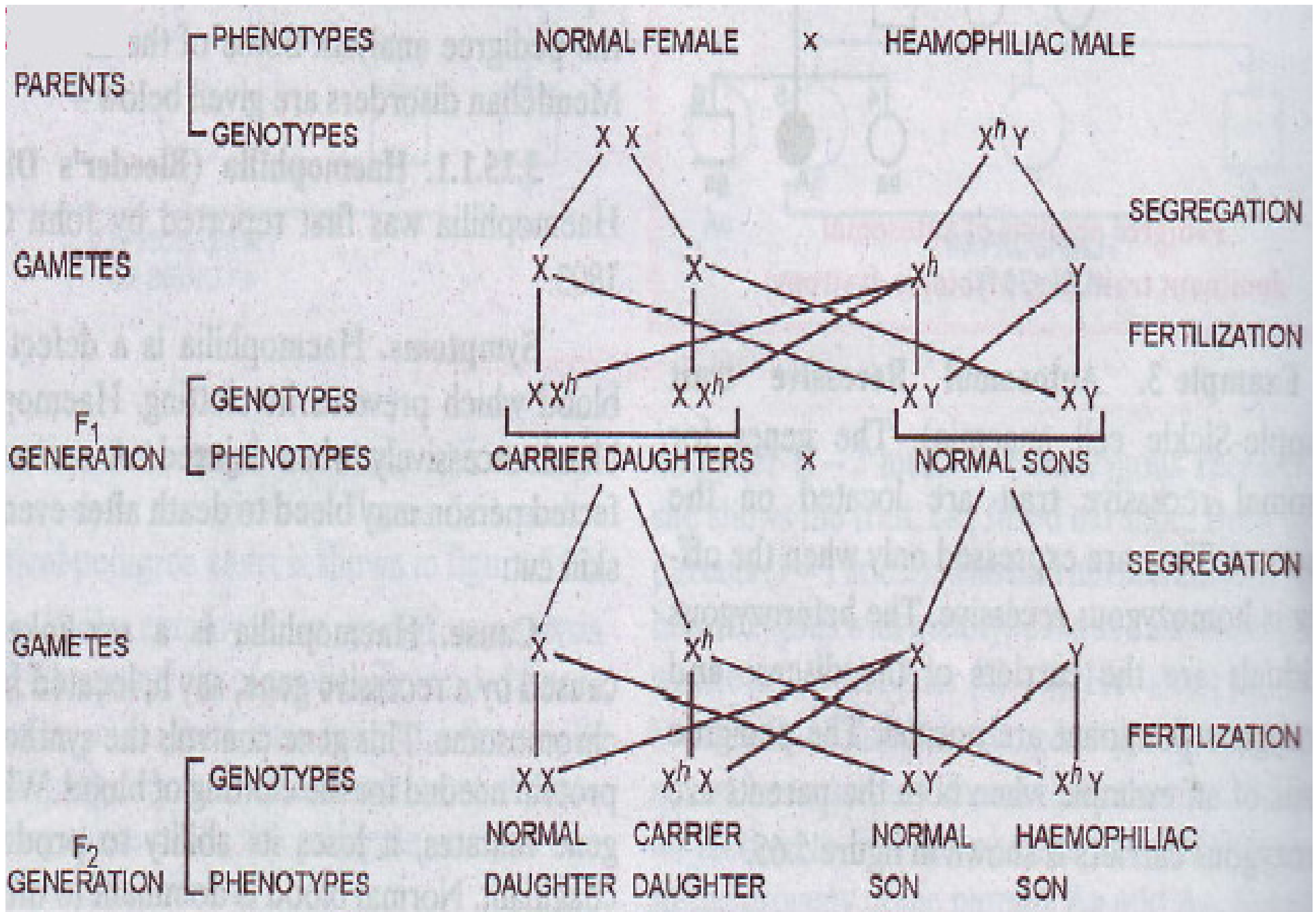
GENETIC DISORDER DUE TO GENE MUTATIONS.

HAEMOPHILIA: DEFECT OF BLOOD WHICH PREVENTS ITS CLOTTING.

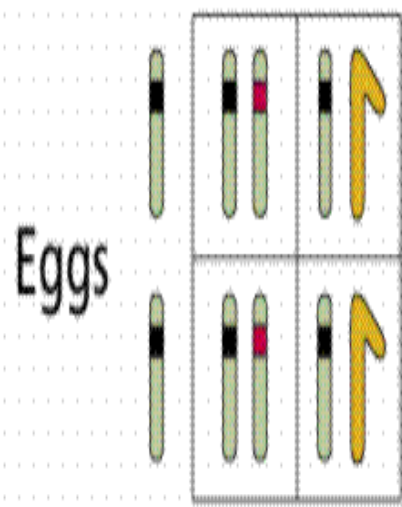
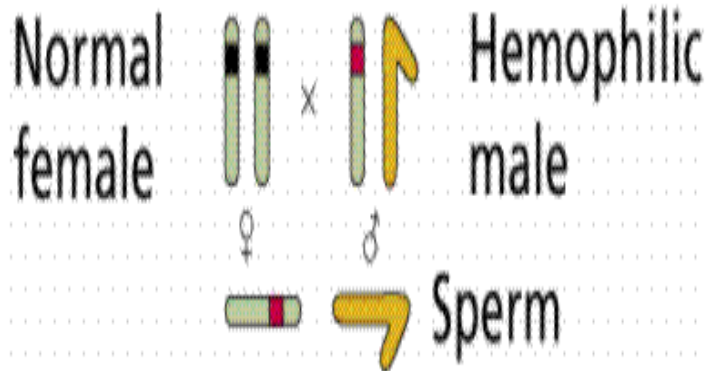
SEX-LINKED TRAIT CAUSED BY RECESSIVE GENE 'h' ON X CHROMOSOME. THIS GENE CONTROLS SYNTHESIS OF PROTEIN NEEDED FOR CLOTTING OF BLOOD.

INHERITANCE

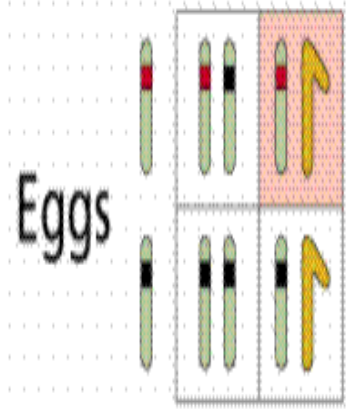
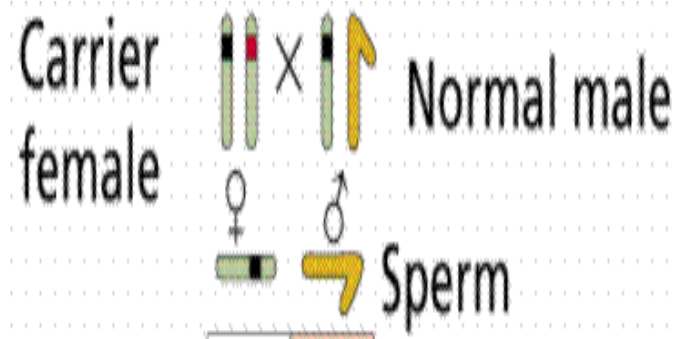




Inheritance of haemophilia in humans.



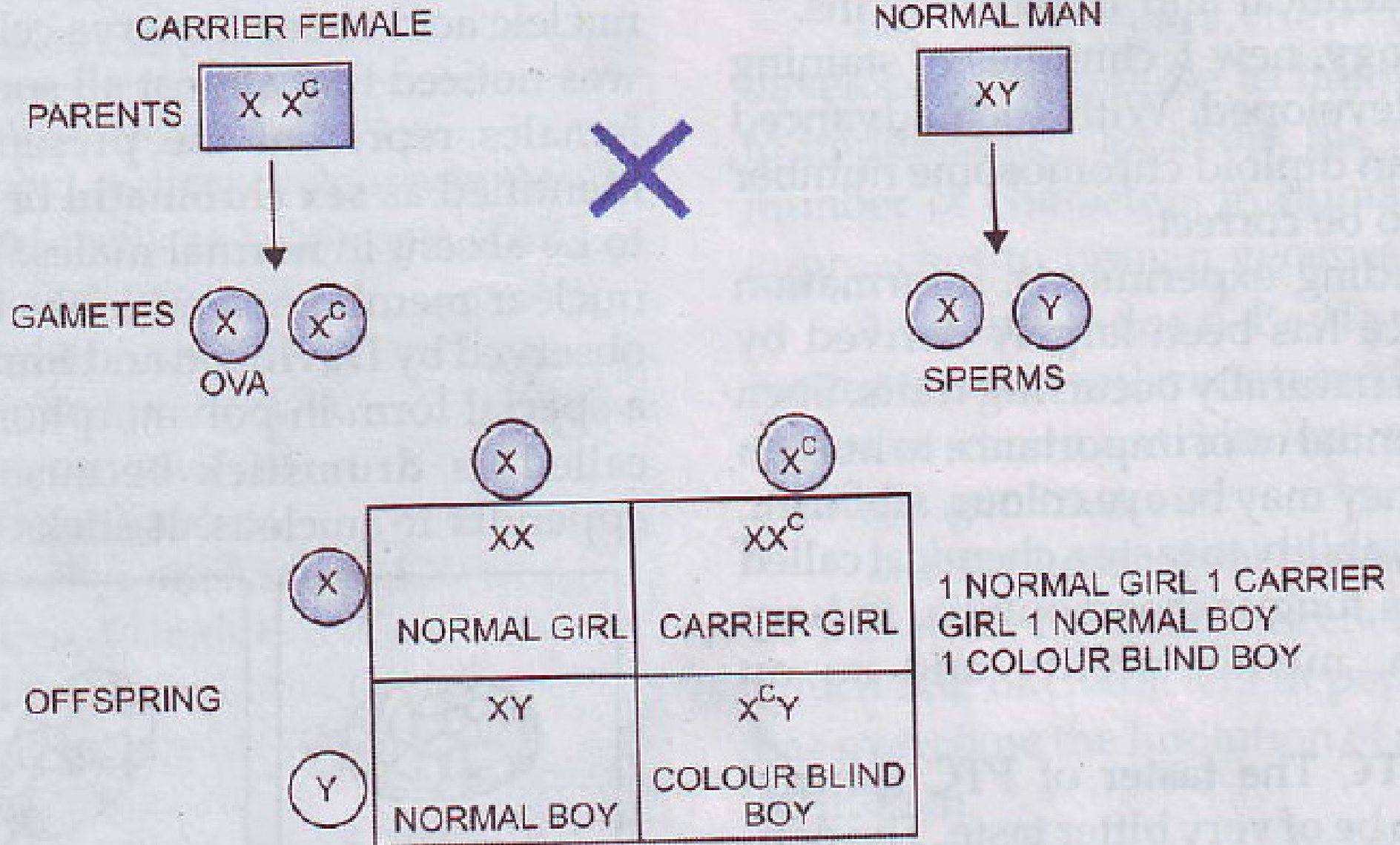
All daughters are carriers All sons are normal



$\frac{1}{2}$ of daughters are normal $\frac{1}{2}$ of sons are hemophilic

$\frac{1}{2}$ of daughters are carriers $\frac{1}{2}$ of sons are normal

- **COLOUR BLINDNESS:** SEX LINKED TRAIT.
- CAUSED BY X-LINKED RECESSIVE GENE BY CAUSING LACK OF CONE PIGMENTS OF RETINA.



PARENTS → COLOUR-BLIND MAN × CARRIER WOMAN

GENOTYPES → $X^c Y$ $X X^c$

GAMETES → X^c, Y X, X^c

CHILDREN →		X	X^c	
	X^c	XX^c	X^cX^c	1 CARRIER ♀ 1 COLOUR-BLIND ♀
	Y	XY	X^cY	1 NORMAL ♂ 1 COLOUR-BLIND ♂

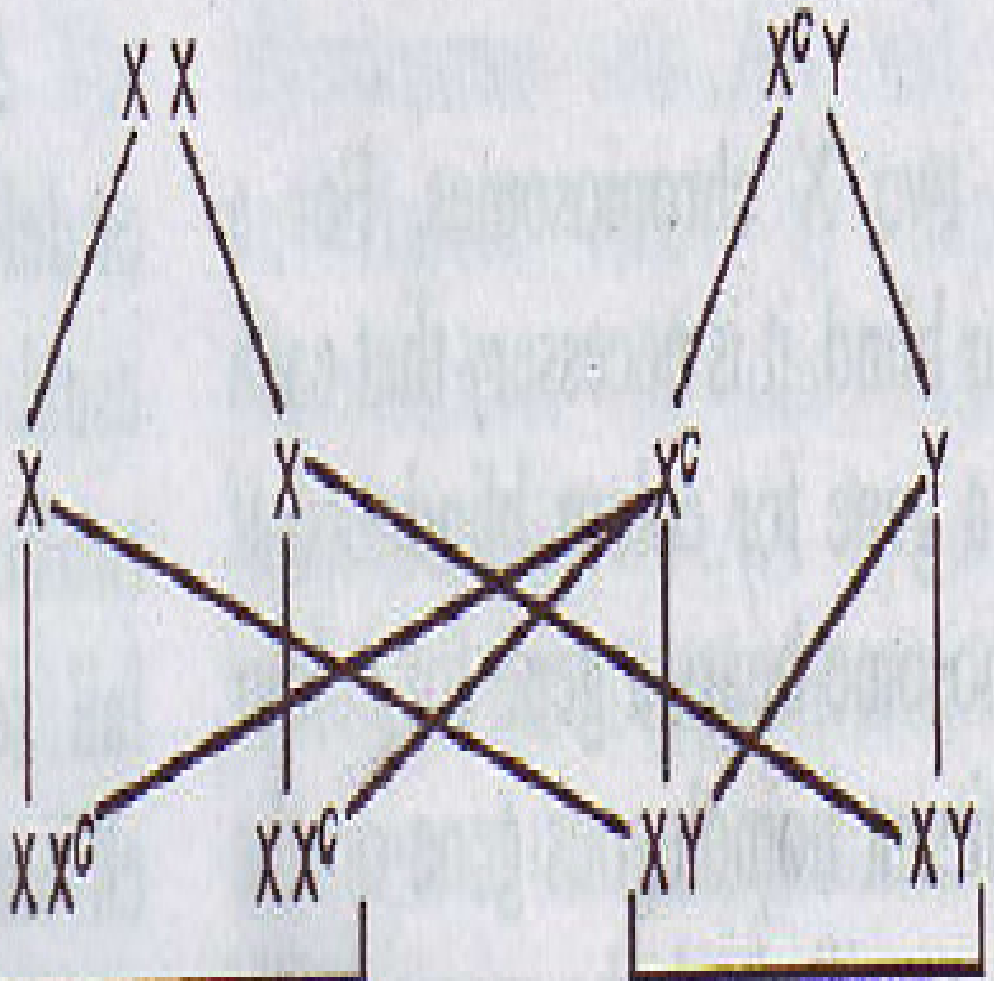
Result of the marriage of a colour-blind man and a carrier woman.

NORMAL FEMALE X COLOUR BLIND MALE

PARENTS

PHENOTYPES
GENOTYPES

GAMETES



F₁ GENERATION

GENOTYPES
PHENOTYPES

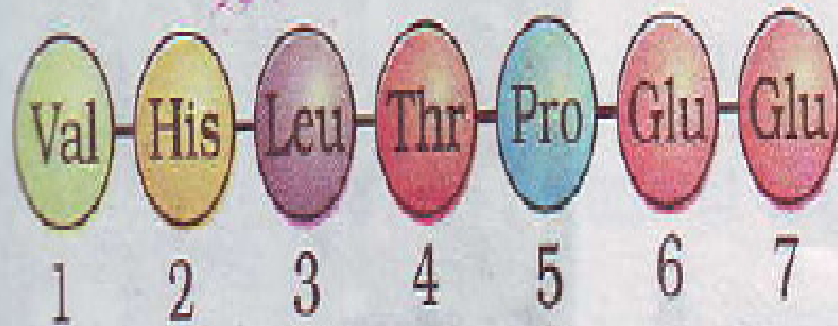
CARRIER DAUGHTERS X NORMAL SONS

SICKLE CELL ANAEMIA

- AUTOSOMAL HEREDITARY DISEASE.
- CHARACTERIZED BY SICKLE SHAPED RED BLOOD CORPUSCLES
- DEFECT CAUSED BY SUBSTITUTION OF GLUTAMIC ACID BY VALINE OF BETA GLOBIN CHAIN OF HAEMOGLOBIN MOLECULE
- MUTANT HAEMBOGLOBIN UNDERGOES POLYMERISATION CAUSING CHANGE IN SHAPE OF RBC'S

Normal Hb (A) gene ...GAG...
...CTC...

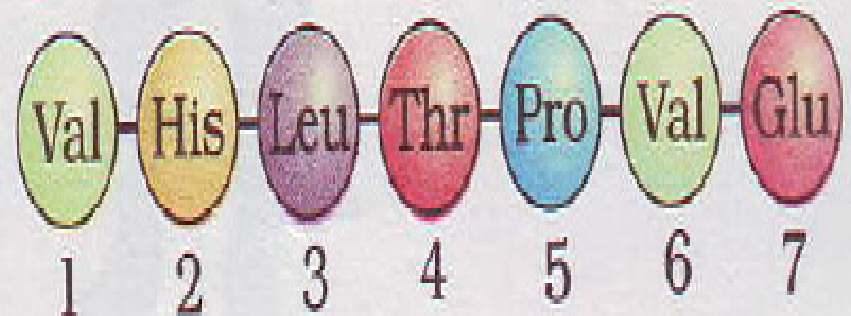
mRNA ...GAG...



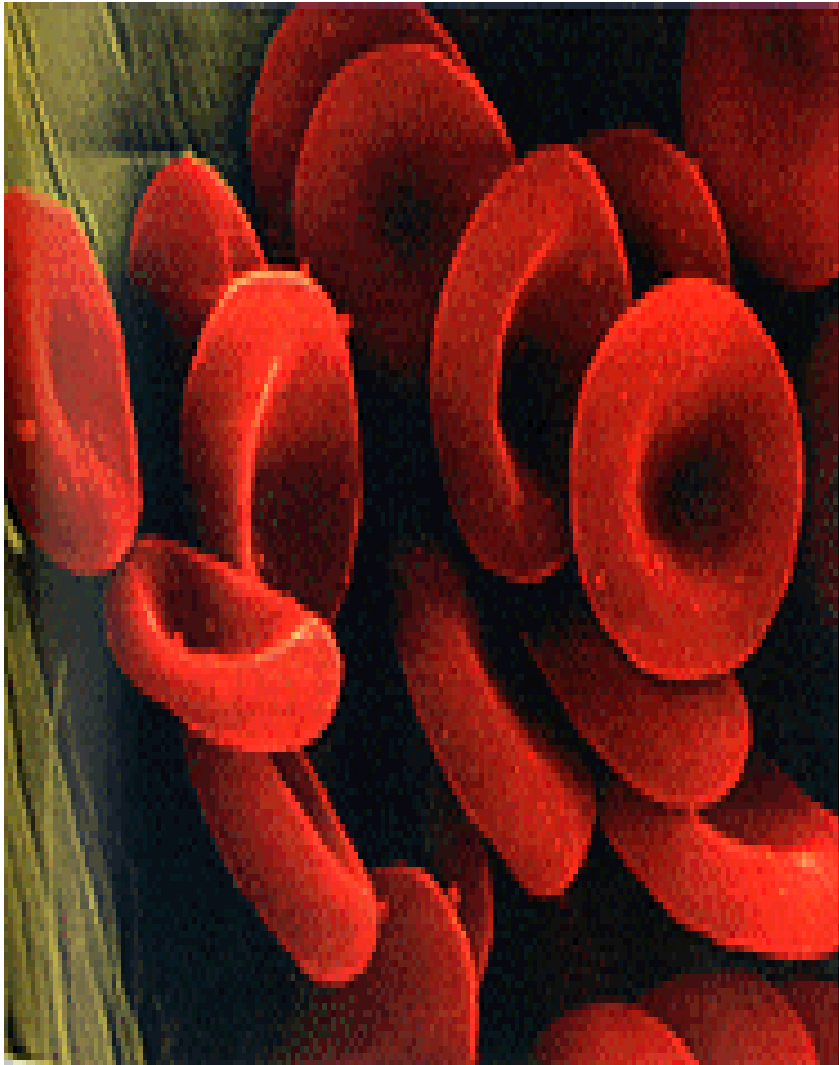
HbA peptide

Sickle-cell Hb(S) gene ...GTG...
...CAC...

mRNA ...GUG...



HbS peptide

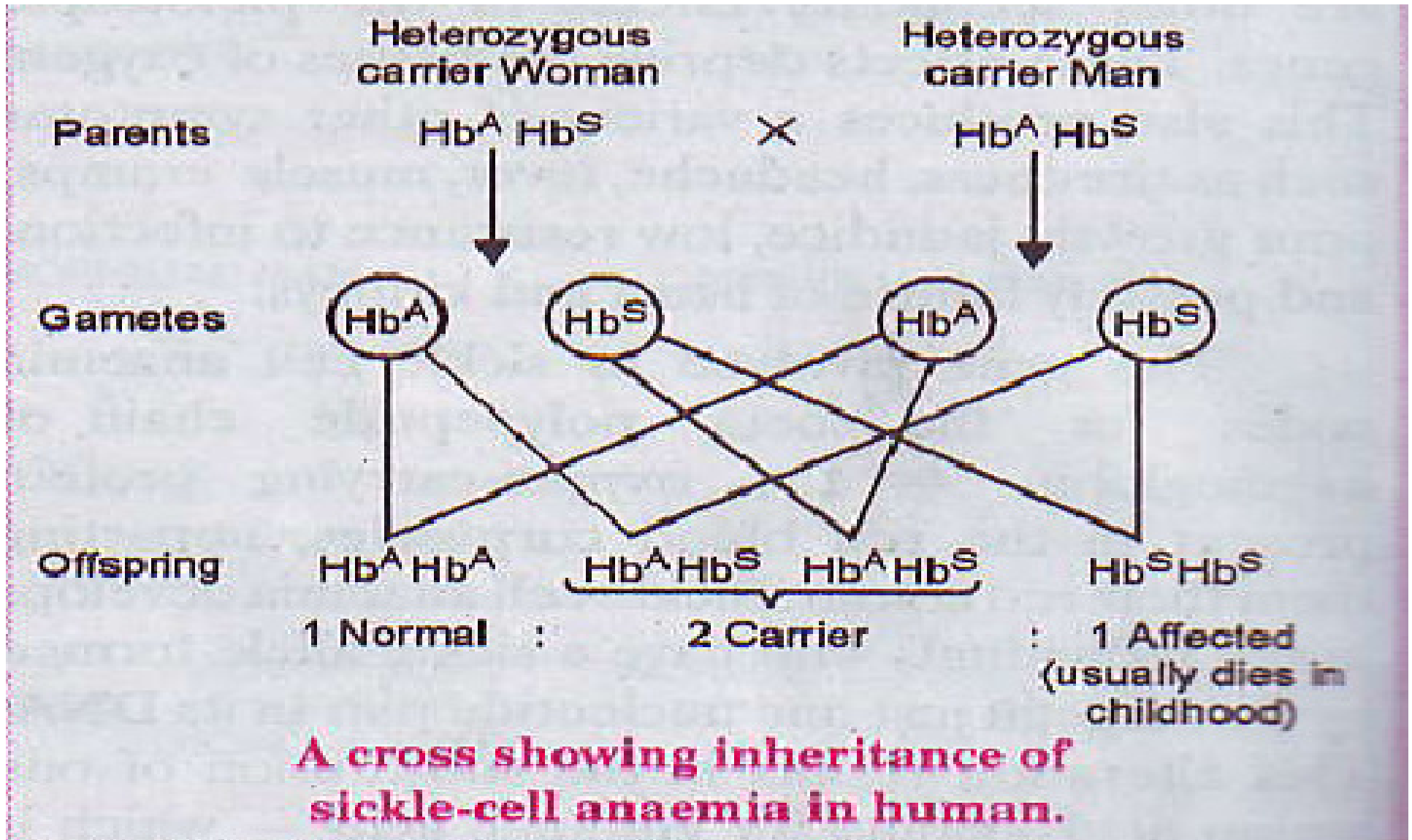


Normal RBCs



Anemic RBCs
(Sickle cell)

- SICKLE CELL TRAIT:** PERSON MAY HAVE ONE GENE FOR NORMAL HAEMOGLOBIN & ONE GENE FOR SICKLE CELL HAEMOGLOBIN [Hb^A, Hb^S] THIS HETEROZYGOUS CONDITION IS SICKLE CELL TRAIT



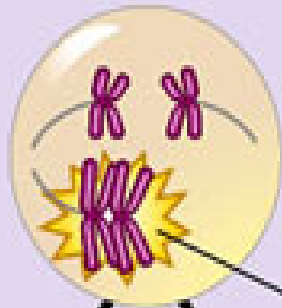
PHENYLKETONURIA

- INBORN ERROR OF METABOLISM ALSO INHERITED AS AUTOSOMAL RECESSIVE TRAIT.
- CHARACTERISED BY ABNORMAL INCREASE IN LEVEL OF PHENYLALANINE IN BLOOD DUE TO ABSENCE OF ENZYME PHENYLALANINE HYDROXYLASE.
- PHENYLALANINE DOESN'T CHANGE INTO TYROSINE ,GETS ACCUMULATED
- RESULTS IN SEVERE MENTAL RETARDATION

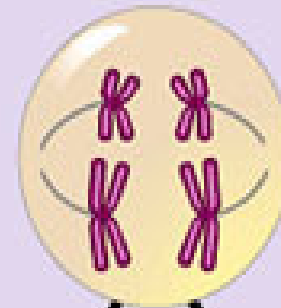
CHROMOSOMAL DISORDERS

- CAUSED DUE TO ABSENCE OR EXCESS OR ABNORMAL ARRANGEMENT OF ONE OR MORE CHROMOSOMES.
- **ANEUPLOIDY:** FAILURE OF SEGREGATION OF CHROMATIDS DURING CELL DIVISION RESULTS IN GAIN OR LOSS OF A CHROMOSOME.

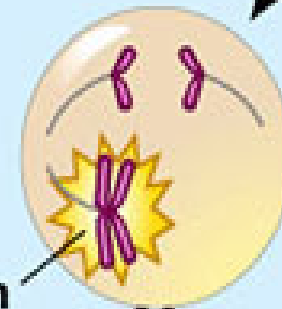
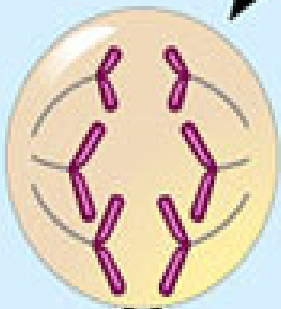
Meiosis I



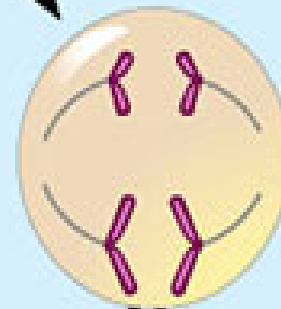
Nondisjunction



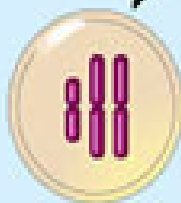
Meiosis II



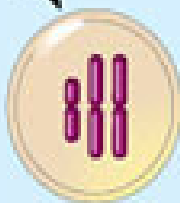
Nondisjunction



Gametes



$n + 1$



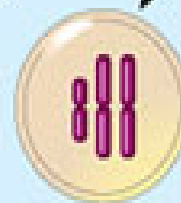
$n + 1$



$n - 1$



$n - 1$



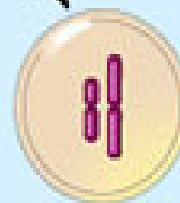
$n + 1$



$n - 1$



n



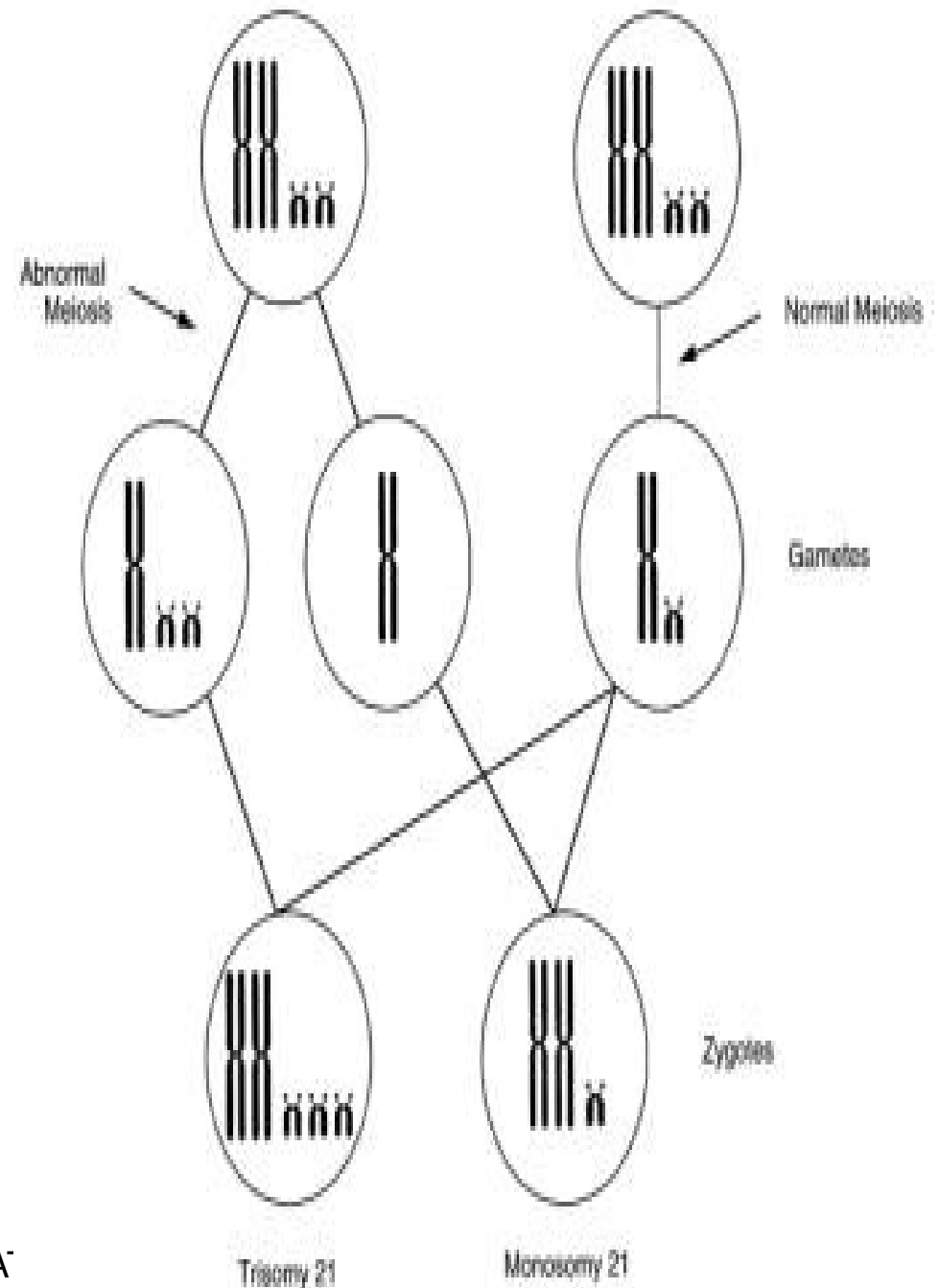
n

Number of chromosomes

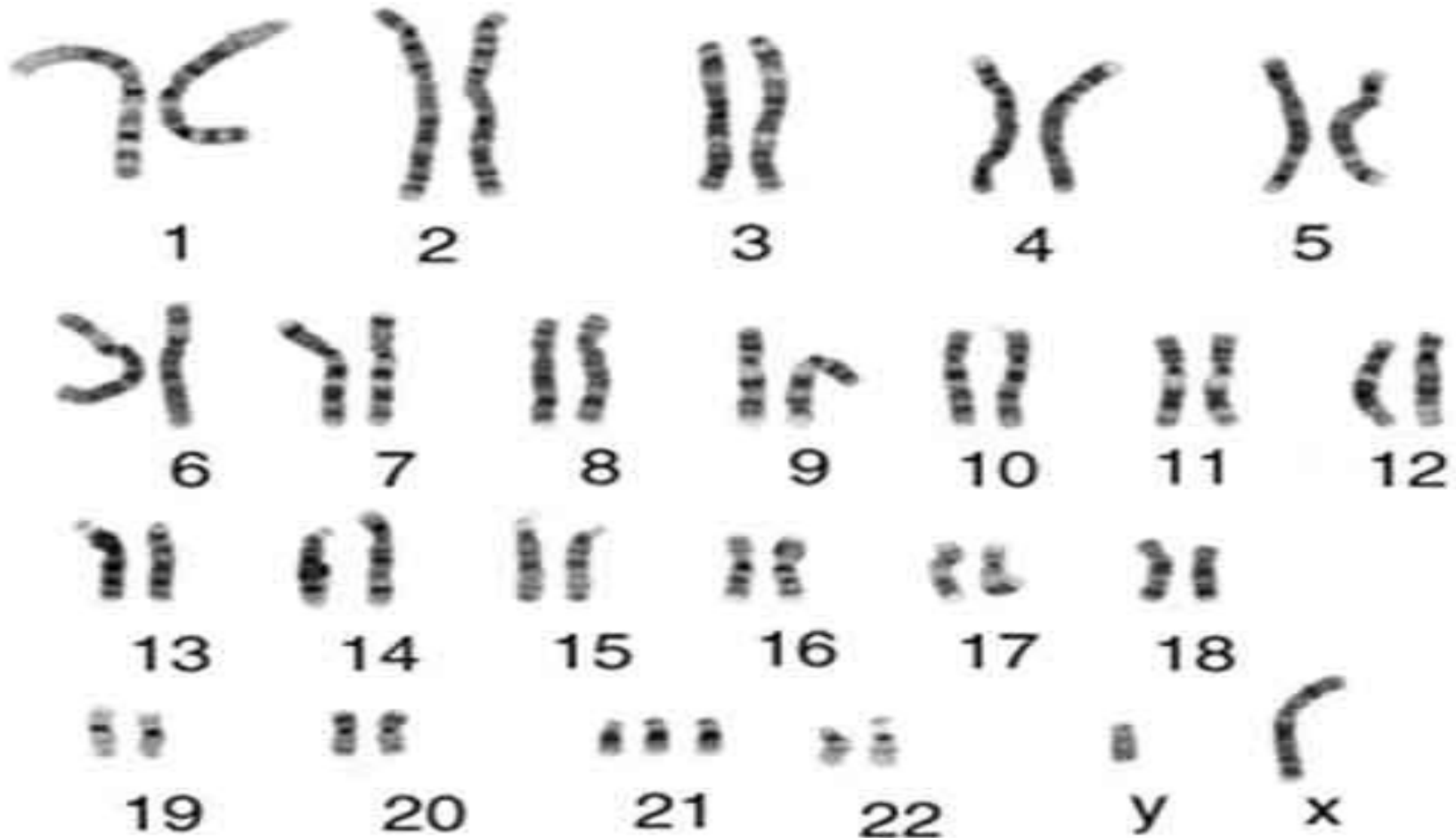
(a) Nondisjunction of homologous chromosomes in meiosis I

(b) Nondisjunction of sister chromatids in meiosis II

- **DOWN'S SYNDROME:**
AN ANEUPLOID
TRISOMIC DISEASE.
- **BOTH**
CHROMOSOMES NO.
21 PASS INTO
SINGLE EGG DUE TO
NONDISJUNCTION
DURING OOGENESIS
IN OVARY
- **EGG HAS 24**
CHROMOSOMES &
OFFSPRING HAS 47
CHROMOSOMES



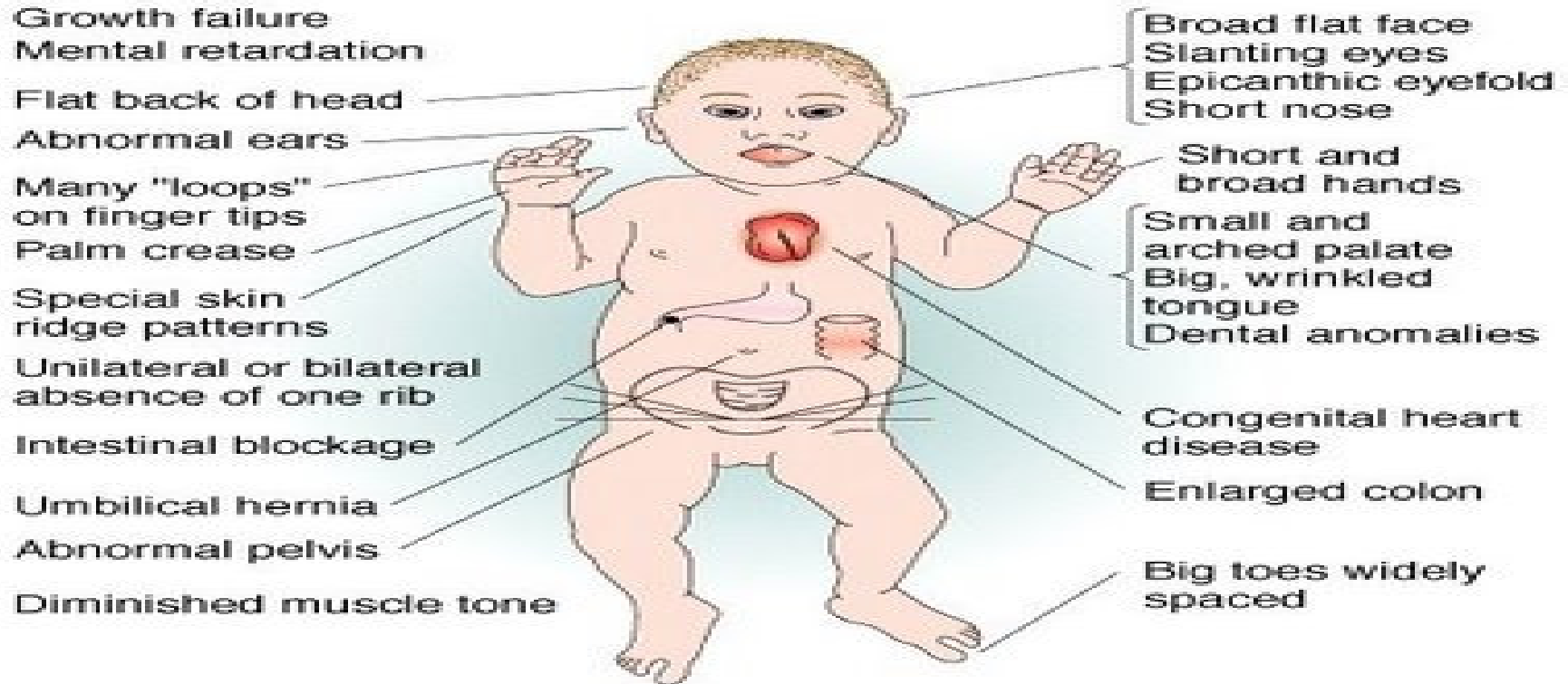
TRISOMY -21



47,XY,+21

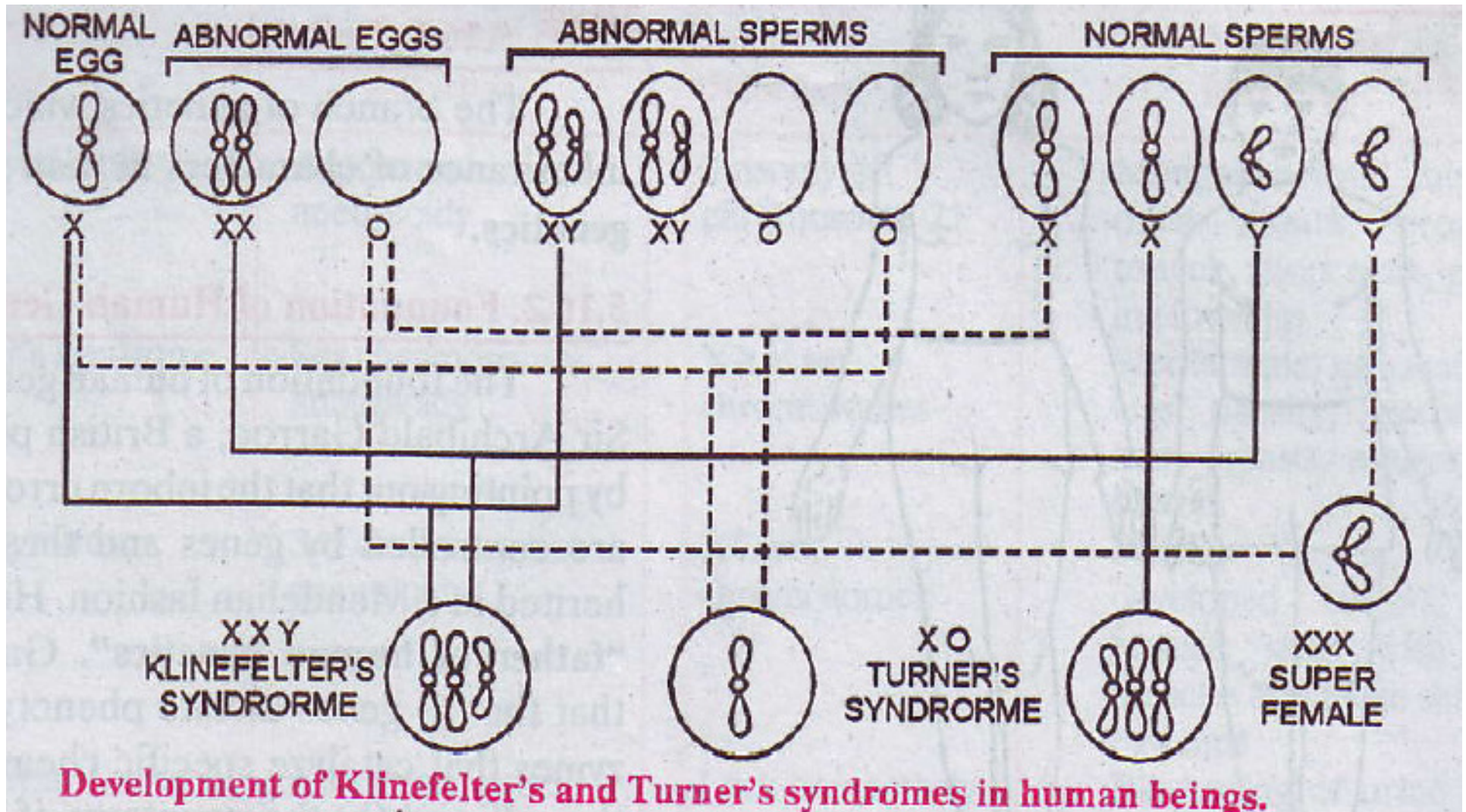
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- **ALSO KNOWN AS 21- TRISOMY.**
- **CHARACTERISED BY ROUNDED FACE ,BROAD FOREHEAD , PERMANENT OPEN MOUTH,PRITUDING TONGUE,FLAT HANDS.**
- **VICTIM HAS LITTLE INTELLIGENCE**



(a)

- **KLINEFELTER'S SYNDROME:**
- ANEUPLOID CONDITION WITH 3 SEX CHROMOSOMES.



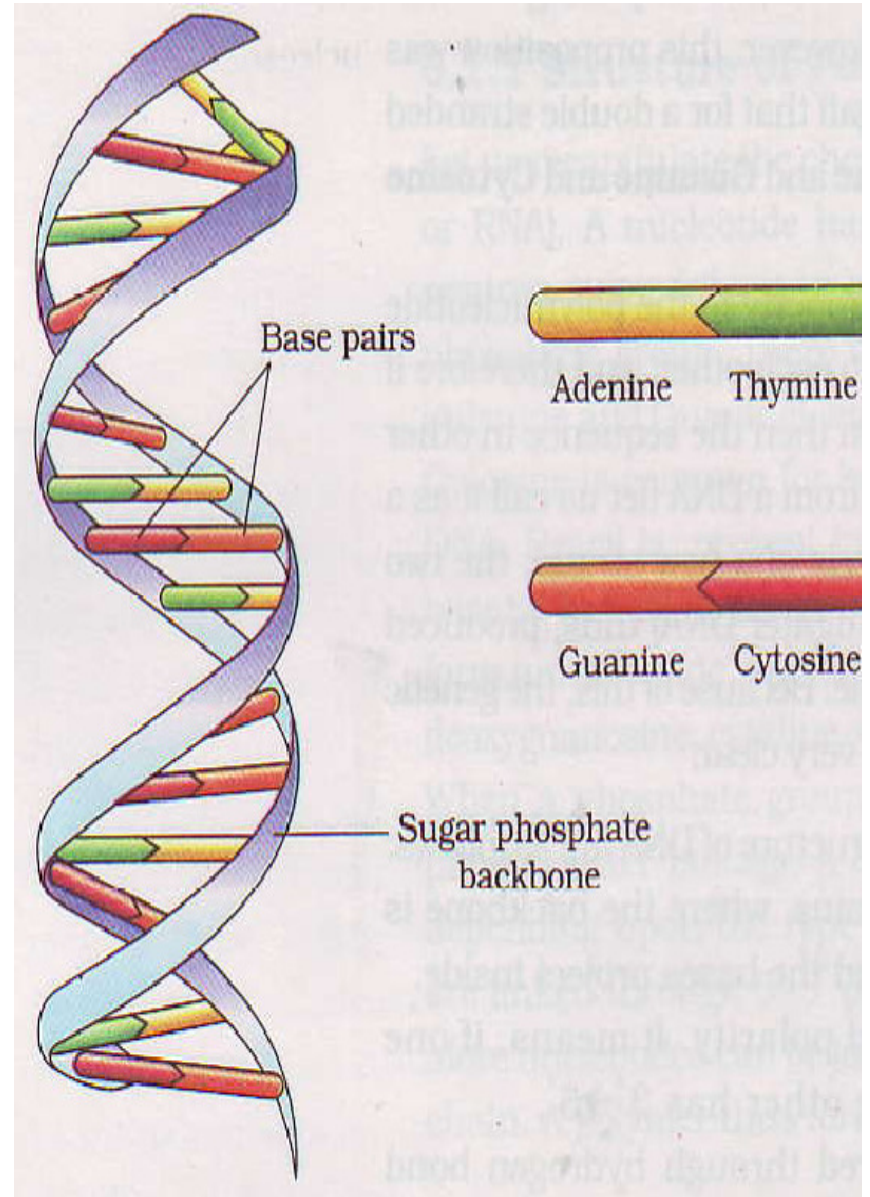
- CAUSED BY UNION OF NONDISJUNCT XX EGG & NORMAL Y CHM.OR NORMAL X EGG & ABNORMAL XY SPERM.
- PERSON IS STERILE MALE WITH UNUSUALLY LONG LEGS, SPARSE BODY HAIR & MANY FEMALE CHARACTERISTICS

- **TURNER'S SYNDROME:** AN ANEUPLOID CONDITION WITH A SINGLE SEX CHROMOSOME.
- PRODUCED BY FUSION OF AN ABNORMAL O EGG WITH X SPERM OR A NORMAL X EGG & ABNORMAL O SPERM
- HAS 45 CHROMOSOMES.
- INDIVIDUAL IS STERILE FEMALE WITH UNDERDEVELOPED BREASTS, REDUCED OVARIES SHORT STATURE & MANY MALE CHARACTERISTICS LIKE HEAVY NECK MUSCLES.

STRUCTURE OF DNA

- GENETIC MATERIAL IN ALL ORGANISMS.
- IN **PROKARYOTIC CELLS**, DNA IS CIRCULAR & OCCURS IN CYTOPLASM.
- IN **EUKARYOTIC ORGANISMS**, DNA LINEAR & LARGELY CONFINED TO NUCLEUS & SMALL QUANTITY IN MITOCHONDRIA.

- CHEMICAL STRUCTURE EXPLAINED BY **P.A.LEVENE**.
- LARGEST MOLECULE COMPOSED OF LONG DOUBLE CHAIN OF **DEOXYRIBONUCLEOTIDE UNITS**.



STRUCTURE OF DNA

- EACH DEOXYRIBONUCLEOTIDE UNIT HAS:
- DEOXYRIBOSE SUGAR
- NITROGEN BASES:
- PURINES: ADENINE & GUANINE.
- PYRIMIDINES: THYMINE & CYTOSINE.
- PHOSPHORIC ACID
- IN EACH DEOXYRIBONUCLEOTIDE UNIT, NITROGEN BASE IS JOINED TO DEOXYRIBOSE SUGAR AT 1' CARBON ATOM.

5' phosphate

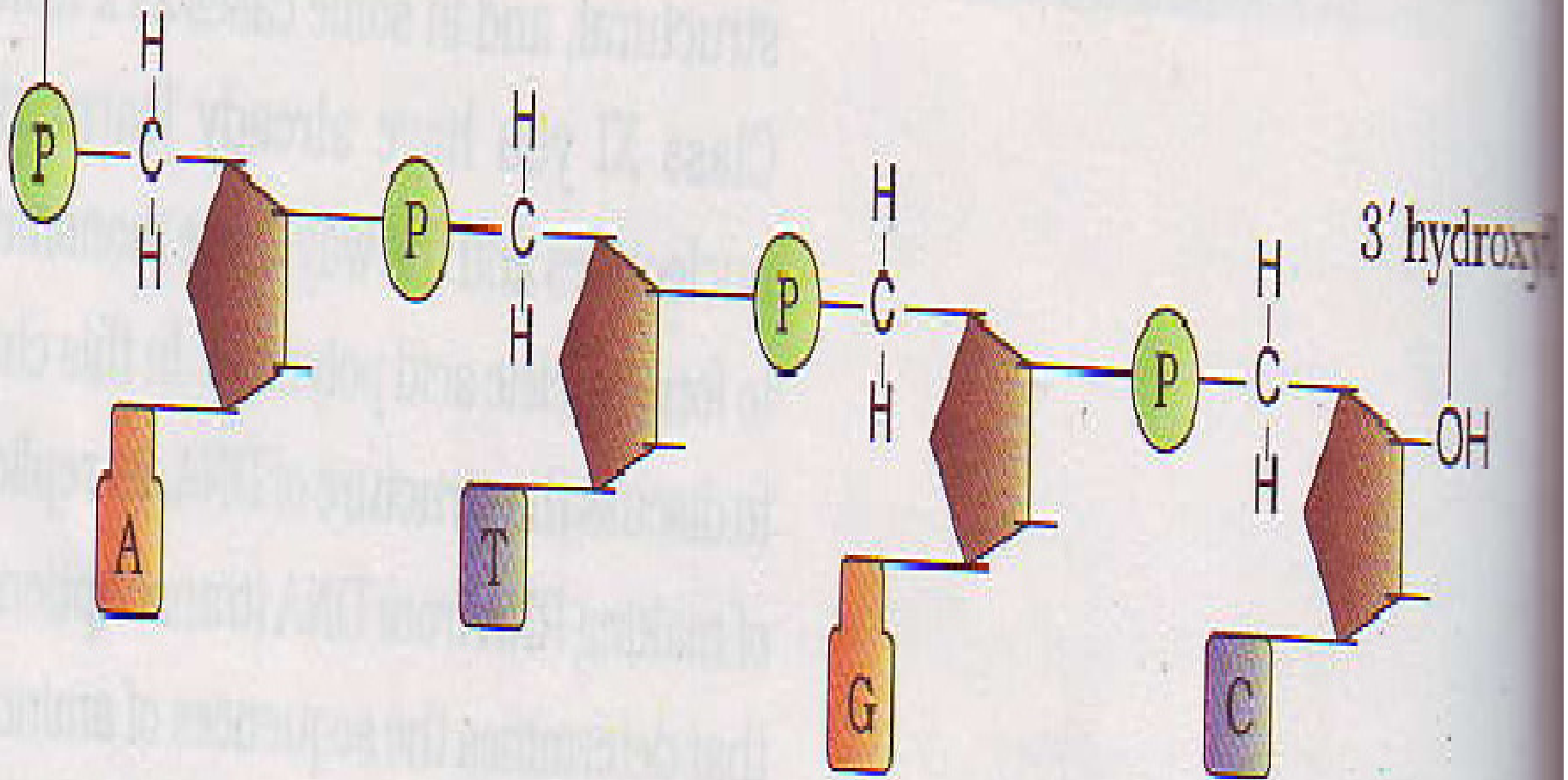
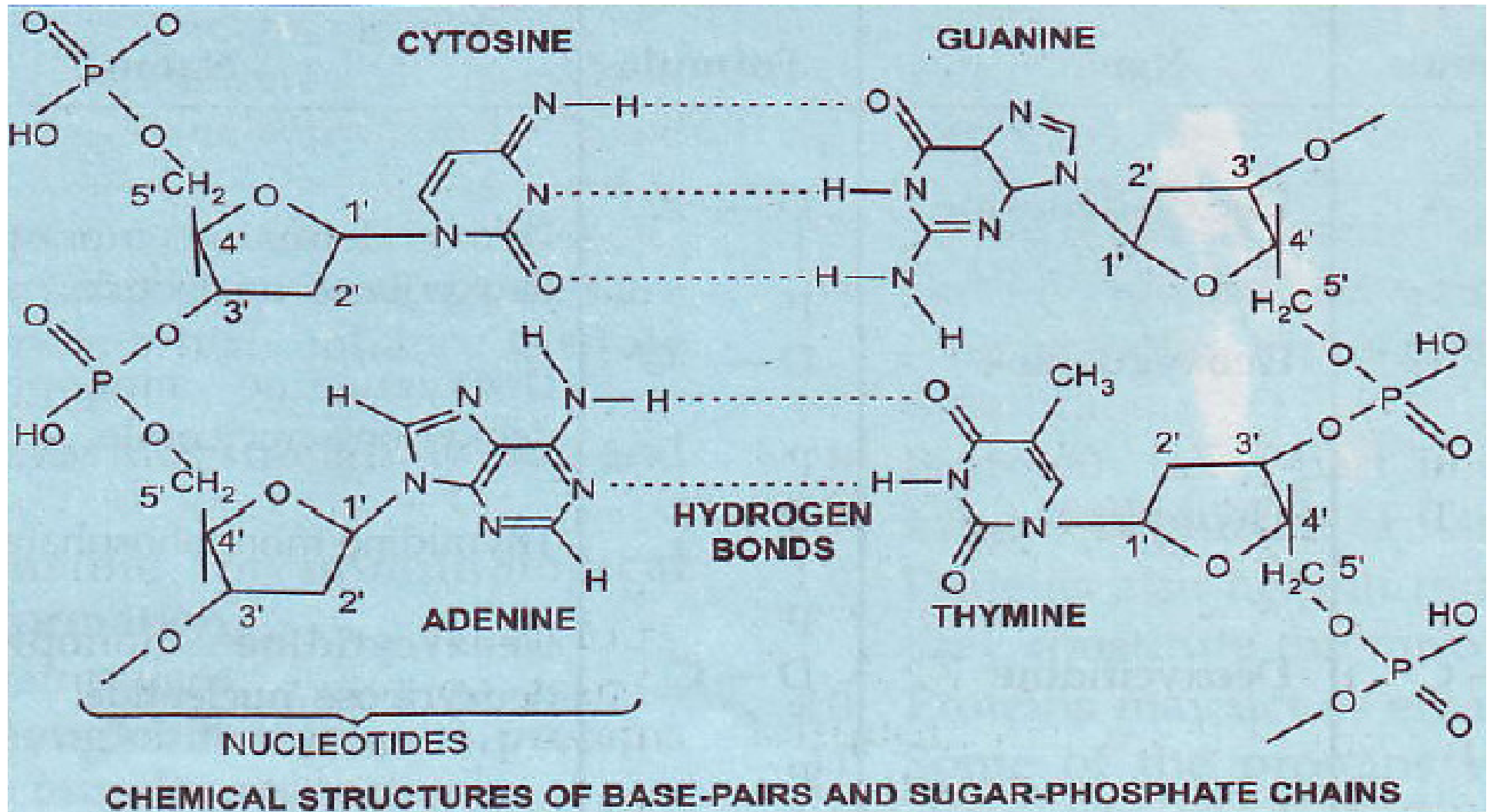


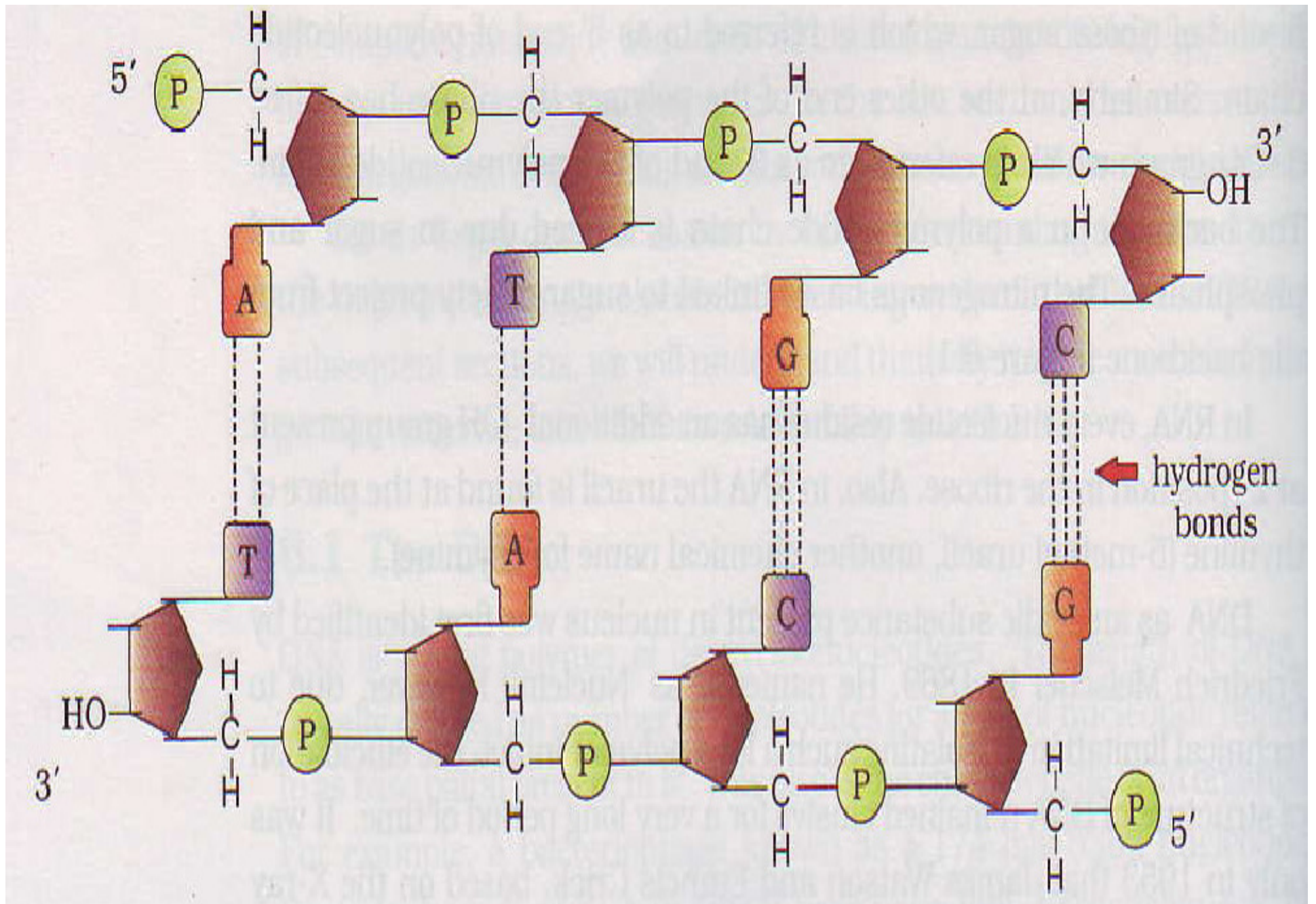
Figure 6.1 A Polynucleotide chain

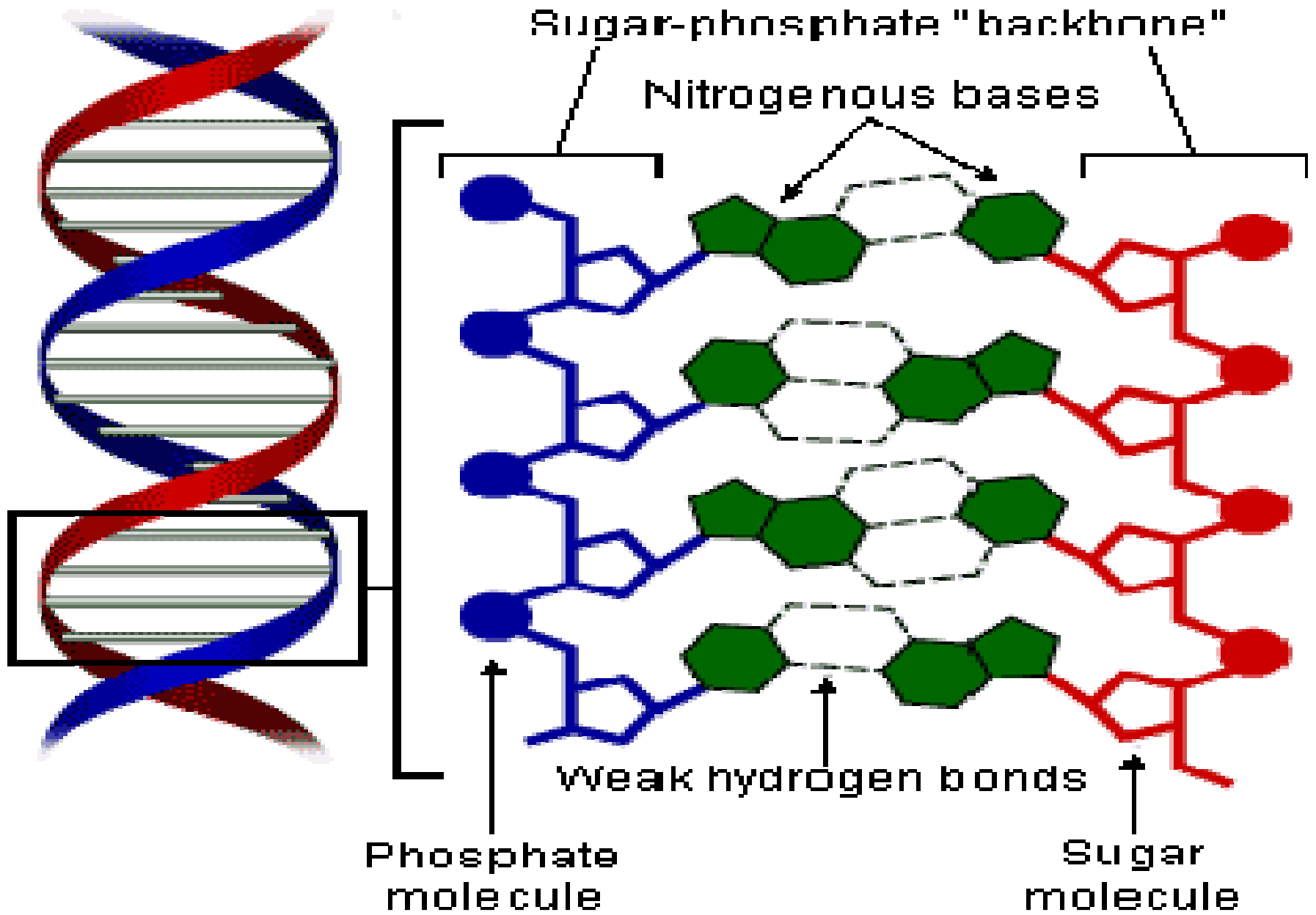
- IN EACH CHAIN PHOSPHATE CARRIED BY CARBON ATOM AT 5' OF SUGAR IN ONE NUCLEOTIDE UNIT JOINED BY PHOSPHO DIESTER BOND TO HYDROXYL OF CARBON AT 3' OF SUGAR IN NEXT NUCLEOTIDE UNIT.



SALIENT FEATURES OF DOUBLE-HELIX ST. OF DNA

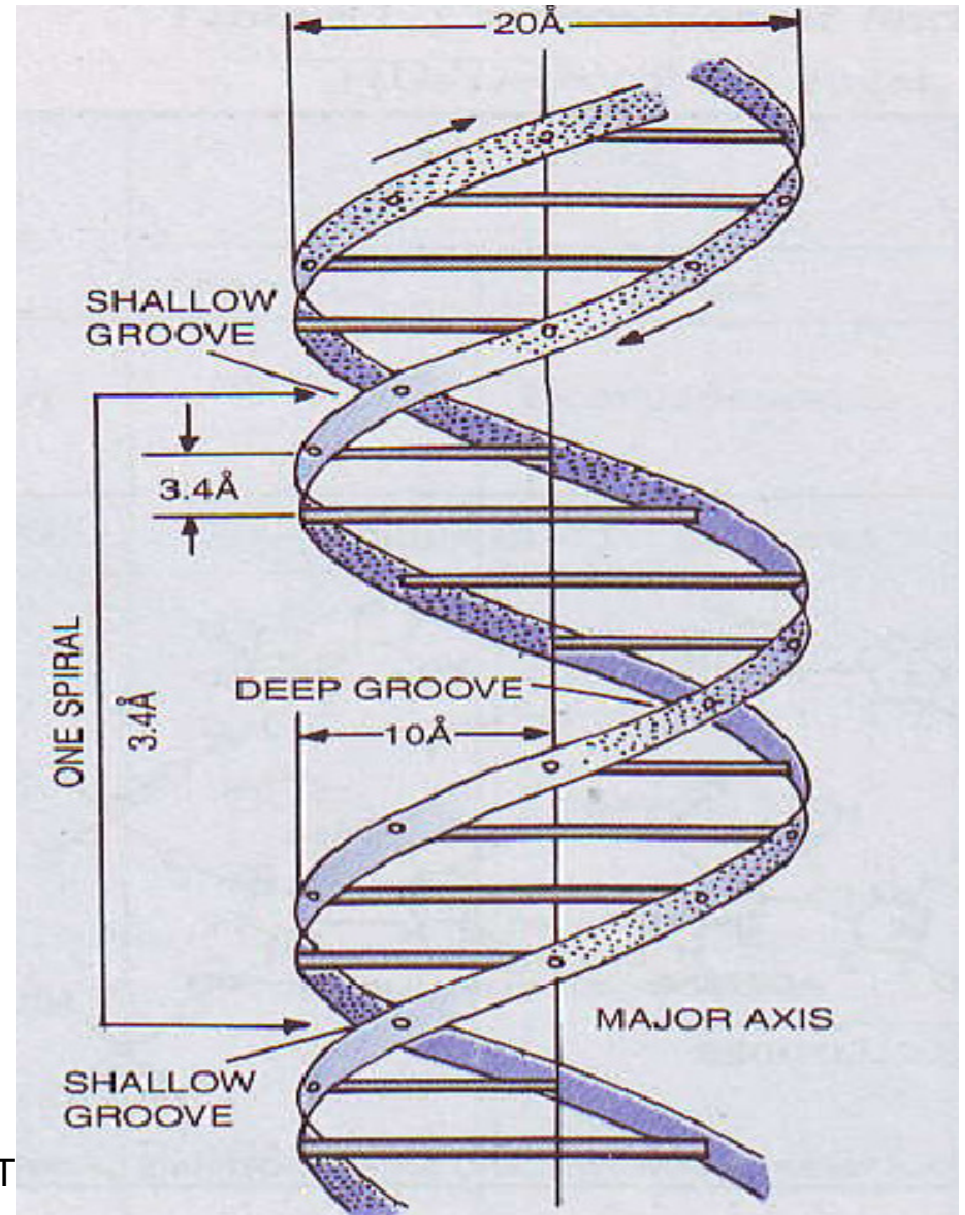
- TWO POLYNUCLEOTIDE CHAINS HAVING BACKBONE CONSTITUTED BY SUGAR- PHOSPHATE & BASES PROJECT INSIDE.
- ANTI PARALLEL POLARITY OF TWO CHAINS.
- BASES OF TWO STRANDS PAIRED BY H- BONDING.
- TWO CHAINS COILED IN RIGHT HANDED FASHION





WATSON AND CRICK MODEL

- J.D. WATSON & F.H.C CRICK DESIGNED THE MODEL OF DNA FOR WHICH THEY GOT THE NOBEL PRIZE IN 1962.
- IT CONSISTS OF TWO LONG PARALLEL CHAINS WHICH ARE JOINED TOGETHER BY SHORT CROSSBARS AT REGULAR INTERVALS.
- TWO CHAINS SPIRALLY COILD AROUND A COMMON AXIS TO FORM DOUBLE HELIX.
- DIAMETER OF DOUBLE HELIX IS 20 Å & ONE COMPLETE SPIRAL OF HELIX IS 34 Å HAS 10 BASE PAIRS



CHARGAFF'S RULES

- DNA MOLECULE ALWAYS HAS $A-T = G-C$ BASE PAIRS.
- PURINES ALWAYS IN EQUAL AMOUNT:
- $A+G=T+C$
- $A=T$ & $G=C$
- DEOXYRIBOSE SUGAR & PHOSPHATE COMPONENTS OCCUR IN EQUAL PROPORTIONS.

TYPES OF DNA

- **LINEAR & CIRCULAR DNA:**

- LINEAR DNA HAS FREE ENDS & PRESENT IN EUKARYOTES.
- CIRCULAR DNA HAS ENDS JOINED TOGETHER & PRESENT IN PROKARYOTES.

- **TROPHIC & GENETIC DNA:**

- TROPHIC DNA PRESENT IN MACRONUCLEUS OF CILIATES
- GENETIC DNA PRESENT IN MICRONUCLEUS OF CILIATES

A-,B-,C-,D-,Z-DNA:

S. No.	Character	A-DNA	B-DNA	C-DNA	D-DNA	Z-DNA
1.	Direction of coiling	Right-handed	Right-handed	Right-handed	Right-handed	Left-handed
2.	Base Pairs per turn	11	10	9	8	12
3.	S-P Backbone	Smooth	Smooth	Smooth	Smooth	Zigzag
4.	Length of Turn		34 Å long			45 Å long

THANKS

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