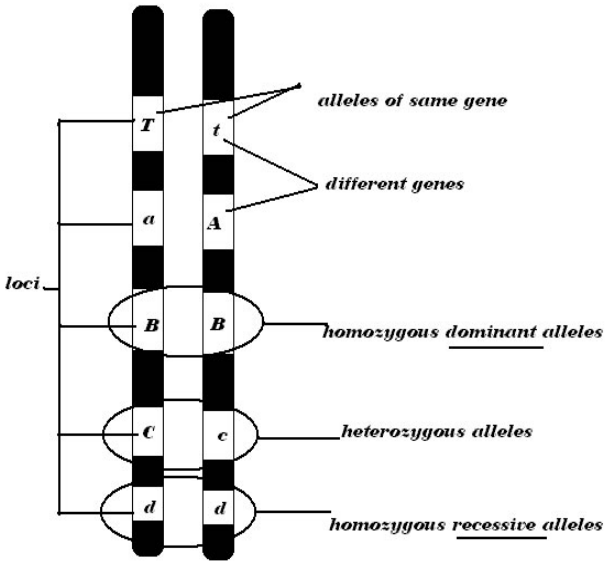
Chapter 5 Genetics

Relationship between genes and chromosome of diploid organism and the terms used to describe them



Know the terms

Terms	Meaning	E ×ample
Locus	Address/ location of a gene in a chromosome	T,A.b,d etc
Allele	Allelomorphs= alternative form of a gene	T and t OR A and a etc
Homozygous	Both alleles of a gene at a locus similar	AA or aa
Heterozygous	Both alleles of a gene at a locus dissimilar	Aa or Tt etc
Homozygous Dominant	Both alleles of a gene at a locus similar & dominant	AA
Homozygous recessive	Both alleles of a gene at a locus similar & recessive	aa

Mendel's first law

(Law of dominance)characters are controlled by discrete units called genes (allele) which occur in pair. In heterozygous condition only one gene that is dominant can e ×press itself. (Can be e ×plained by monohybrid cross)

Mendel's second law

(Law of segregation): The two alleles received, one from each parent, segregate independently in gamete formation, so that each gamete receives one or the other with equal probability. (Can be e ×plained by monohybrid cross)

Mendel's third law

(Law of recombination): Two characters determined by two unlinked genes are recombined at random in gamete formation, so that they segregate independently of each other,

each

according to the first law (note that recombination here is not used to mean crossing-over in meiosis).

(Can be e ×plained by dihybrid cross)

This is what Mendel said (summary) :

1) **Dominant** alleles overpower recessive alleles. Dominant traits overpower recessive traits.

2) **Rule of segregation (Separation)**: Gametes (se × cells) only receive one allele from the original gene.

3) **Rule of Independent assortment**: One trait will not determine the random selection of another.

Incomplete dominance: When one allele of a gene is not completely dominant over the other and the

F1 hybrids are intermediate between two parents. The phenotypic and genotypic ratio is same.1:2:1 in

F2 generation. E.g. Snapdragon or Antirrhinum majus

Co dominance: Two alleles of a gene are equally e ×pressive and dominant in a generation eg. **Human blood group**

(**Note** : Human blood group is also an e ×ample for multiple allelisim i.e when a gene e ×ists in more

than two allelic forms IA , IB and i)

Basic outline of Mendels cross

1. Pure breeding parents for a pair of contrasting character (allelic pair) is taken

Eg.Tall pure-bred pea plants (TT) & short pure-bred pea plants (tt)

↓

2. Gamete formation (Meiosis)

↓

3. Hybridization (crossing is done)

↓

4. F1 generation - the product of the above cross (are called hybrids)

↓

5. Selfing (allowed to self fertilize / self breeding)

↓

6. Gamete formation (Meiosis)

↓

7. F2 generation - the product of the above selfing

↓

8. Analysis of result (Phenotype and Genotype)

Linkage

Tendency of genes on same chromosome to remain together Such genes are called – linked genes.

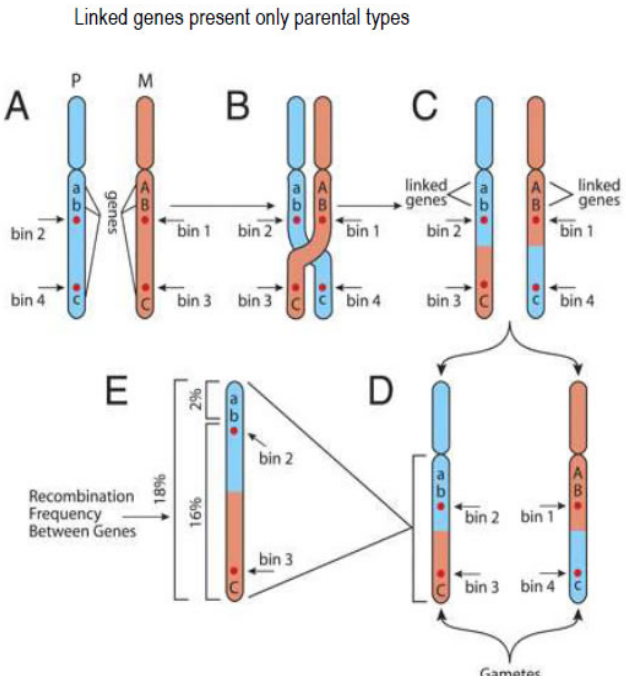


Figure Schematic of Genetic Linkage and Recombination

(A) Two homologous chromosomes: blue (paternal) and orange (maternal). Three genes with separate alleles and linkage " noted (A,a; B,b; C,c;).

(B) Crossing over during meiosis. (chiasma formation)

(C) Two alleles and their linked genes have switched locations via recombination. Four additional alleles and their associated (A, a; B, b;) have not switched and are considered linked.

(D) Recombined haploid chromosomes segregate separately during meiosis as gametes before fertilization.

(E) Sample recombination frequencies between genes demonstrating higher rates of recombination for genes further apart.

Cross	Result of F2 generation	
	Phenotypic ratio	Genotypic ratio
Monohybrid Tt × Tt	3:1	1:2:1
Dihybrid cross	9:3:3:1	1:2:1:2:4:2:1:2:1
YyRr × YyRr		
Incomplete dominance Rr × Rr	1:2:1	12:1

Co Dominance and multiple allelisim	
Blood group	Possible genotype
A	I^AI^A OR I^Ai
B	I^BI^B OR $^B I^Bi$
AB	I^AI^B
O	ii

IAi × IAi A ; O

Crosses of blood group (CO DOMINANCE		
Blood group	Possible genotype	Possible phenotype
A × A	IAIA × IAIA	A
	IAIA × IAi	A
	IAi × IAi	A;O
B × B	IBIB × IBIB	B
	IBIB × IBi	B
	IBi × IBi	B;O
AB × AB	IBIB × IBIB	AB: A;B
O × O	ii × ii	O

POSSIBLE BLOOD GROUP OF PROGENY WITH RESPECT TO THE BLOOD GROUP OF PARENTS

Parent	Progeny			
	A	B	AB	O
AXA	+	-	-	+
AXO	+	-	-	+
AXB	+	+	+	+
BXB	-	-	-	+
BXO	-	-	-	+
ABXA	+	+	+	-
ABXB	+		+	-
ABXO	+	+		-
ABXAB	+	+	+	-
OXO	-	-	-	+
KEY	+ = POSSIBLE		- = NOT POSSIBLE	

Sex determination and sex chromosome

Organism	Male	Female
Human beings	XY	XX
Birds	ZZ	ZW
Insects	XO	XX

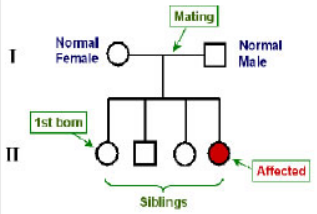
Pedigree Analysis

Pedigree is a chart of graphic representation of record of inheritance of a trait through several generations in a family Symbols used:- refer NCERT Text Book

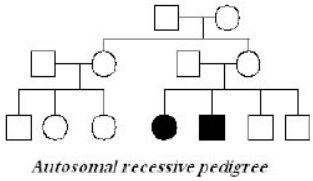
Four patterns of inheritance

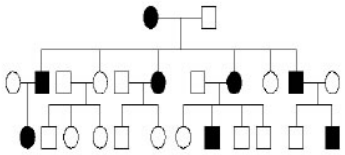
AUTOSOMAL DOMINANT	AUTOSOMAL RECESSIVE
1. Traits are controlled by dominant genes 2. Both males and females are equally affected 3. **Traits do not skip generations** 4. e.g. polydactyly, tongue rolling ability etc	1. Traits controlled by recessive genes and appear only when homozygous 2. Both male and female equally affected 3. **Traits may skip generations** 4. 3:1ratio between normal and affected. 5. Appearance of affected children from

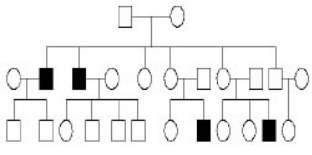
normal parents (heterozygous)
6. All children of affected parents are also affected.
7. e.g.- Albinism, sickle cell anaemia etc



a model of pedigree chart


Autosomal recessive pedigree


A 'typical' autosomal dominant pedigree


x-chromosome linked pedigree

Now try to answer

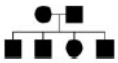
1. Is it possible that this pedigree is for an autosomal dominant trait?

2. Can two individuals that have an autosomal dominant trait have unaffected children?

3. Is it possible that this pedigree is for an autosomal dominant trait?

4. Is it possible that this pedigree is for an autosomal dominant trait?

5. Is it possible that the pedigree above is for an autosomal recessive trait?
6. Assuming that the trait is recessive, write the genotype of each individual next to the symbol
   A = normal
   a = the trait (a genetic disease or abnormality)



7. Is it possible that the pedigree above is for an autosomal recessive trait?
8. Write the genotype of each individual next to the symbol

9. Is it possible that the pedigree above is for an autosomal recessive trait?

10. Is it possible that the pedigree above is for an X-linked recessive trait?
11. Write the genotype next to the symbol for each person in the pedigree

12. Is it possible that the pedigree above is for an X-linked recessive trait?

13. Is it possible that the pedigree above is for an X-linked recessive trait?

14. Is it possible that the pedigree above is for an X-linked recessive trait?

15. Is it possible that the pedigree above is for an X-linked recessive trait?

16. Is it possible that the pedigree above is for an X-linked recessive trait?

Clues

	Affected	Unaffected
Autosomal Dominant	AA Aa	aa
Autosomal Recessive	aa	AA Aa
X- chromosome linked recessive	$X^{-}X^{-}$ $X^{-}Y$	XX XX^{-} XY

TERMINOLOGIES

Allele = A factor or letter that makes up a gene. 2 alleles make up one gene. Alternative forms of a genetic locus; a single allele for each locus is inherited separately from each parent (eg., at a locus for eye color the allele might result in blue or brown eyes).

Alleles = "B" and "b" are different alleles.

Autosomal = refers to genes that are not found on the sex chromosomes.Autosomal chromosomes are ones that are not XX and XY. A chromosome not involved in sex determination. The diploid human genome consists of 46 chromosomes, 22 pairs of autosomes, and 1 pair of sex chromosomes (the X and Y chromosomes).

Carrier = a person who has a defective gene and a dominant normal gene and therefore, is normal. (Nn)

Centimorgan (cM): A unit of measure of recombination frequency. One centimorgan is equal to a 1% chance that a marker at one genetic locus will be separated from a marker at a second locus due to crossing over in a single generation. In human beings, 1 centimorgan is equivalent, on average, to 1 million base pairs

Chromosomes = 46 are found in human cells. Genes are carried among chromosomes.

Clones: A group of cells derived from a single ancestor.

Cystic Fibrosis = Autosomal recessive. Mucous in lungs.Death in the 20's.notices that pure tall plants bred to pure short plants resulted in tall hybrid plants. Tallness was dominant over shortness

Dominant = an allele that overpowers another is dominant.

Down's Syndrome = due to an extra chromosome in (21st pair).

Gamete = sperm or egg. Germ Cell. In humans, germ cell contains 23 chromosomes

Genetics: The study of the patterns of inheritance of specific traits

Gene = Every trait is controlled by a gene. A human has 20,000 genes. Genes are controlled by 2 factors called "alleles" Each allele comes from a parent.

Genotype = All the genes of a beastie equal the genotype of the beastie. (Genes an organismpossesses)

Genome: All the genetic material in the chromosomes of a particular organism; size generally given as its total number of base pairs.

Germ Cell- An egg or sperm cell.A gamete. In humans, a germ cell contains 23 chromosomes

Haploid= A single set of chromosomes (half the full set of genetic material), present in the egg and sperm cells of animals and in the egg and pollen cells of plants. Human beings have 23 chromosomes in their reproductive cells

Hemophilia = sex-linked recessive. Males get it most often.

Heterozygous= alleles of a gene are "different"

Heterozygosity= presence of different alleles at one or more loci on homologous chromosomes.

Homozygous = alleles of a gene are "the same"

Homologous chromosomes: A pair of chromosomes containing the same linear gene sequences, each derived from one parent

Huntington's Chorea = Autosomal Dominant. People die at 40 +... Jerky muscular motions

Hybrid = alleles of a gene are "different" (Hh). See heterozygous Independent Assortment: Johann Gregor Mendel's 2nd principle. States that alleles of one gene separate independently from alleles of another gene. In other words, eye color does not affect a person's ability to roll his or her tongue. In vitro: outside a living organism.

Karyotype : Photomicrograph of an individuals chromosomes arranged in a standard format showing the number, size, and shape of each chromosome type. Linkage: Proximity of two or more genes on a chromosome.The closer together the genes, the lower the probability that they will be separated during meiosis and hence the greater the probability that they will be inherited together. Linkage map: relative positions of genetic loci on a chromosome, determined on the basis of how often the loci are inherited together. Distance is measured in centimorgans (cM). Locus (pl. loci): The position on a chromosome of a gene or other chromosome marker; also, the DNA at that position. The use of locus is sometimes restricted to mean regions of DNA that are expressed.

Meiosis: kind of cell division that produces sperm and egg. Meiosis cuts the number of chromosomes in half. In humans, for instance, the nuclei of body cells contain chromosomes. Due to meiosis, sex cells carry only chromosomes – one chromosome from each original homologous pair.

Mendel, Johann Gregor = The father of genetics (said that traits are controlled by 2 factors etc...)

Mutation = Change in the DNA instructions. Change in DNA sequence. Change can be beneficial, detrimental or neutral. Ultimately results in change in protein. For instance, random genetic mutation gave rise to the dark phenotype of the peppered moth. Non-Disjunction: When homologous chromosomes fail to segregate properly during meiosis. Down syndrome, Turner syndrome and Klinefelter syndrome result from non-disjunction.

Phenotype : the way an organism looks.(EXTERNAL CHARACTERISTICS)

Recessive : A small, weaker allele is recessive. (CANNOT EXPRESS ITSELF IN HETEROZYGOUS CONDITION)

Segregation : One of Mendel's principles. Mendel said all genes are comprised of 2 factors, one

from each parent. Chromosomes segregate during meiosis. These factors (alleles) of a gene separate during the formation of gametes (sperm and egg). This ensures that each parent contributes 50% of their genetic information.

Sex chromosomes : chromosomes that determine sex (XY and XX) Somatic Cell : Body cell that contains 46 chromosomes in humans.

Tay Sachs : Autosomal recessive. Children die young. Head enlarges.... Trait : feature of an organism.