Ch 4 · Principles of Inheritance & Variation

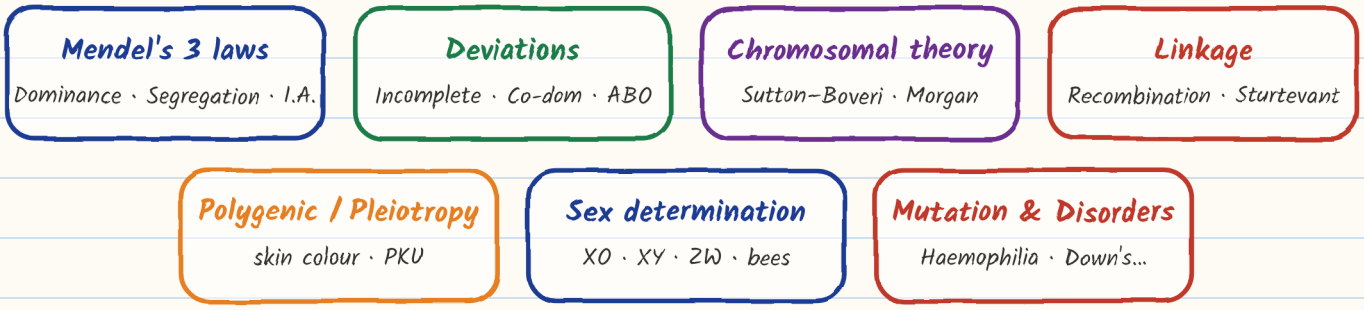
अध्याय 4 · वंशागति तथा विविधता के सिद्धांत

Class 12 Biology · NCERT based · English + हिंदी · ✍️ revision notes

Index / विषय-सूची

1. Basic terms
2. Mendel's experiments
3. Monohybrid cross, Laws of Dominance & Segregation
4. Incomplete dominance, Co-dominance, Multiple alleles
5. Dihybrid cross & Law of Independent Assortment
6. Chromosomal theory, Linkage & Recombination
7. Polygenic inheritance & Pleiotropy
8. Sex determination
9. Mutation
10. Genetic disorders – Pedigree, Mendelian, Chromosomal
11. ⭐ Exam Corner (Q&A)

Chapter map · अध्याय मानचित्र



Infographic: chapter map

1. Basic Terms / मूल शब्दावली

- **Genetics** (आनुवंशिकी) – branch of biology dealing with inheritance & variation.
- **Inheritance** (वंशागति) – process by which characters are passed from parent to progeny.
- **Variation** (विविधता) – degree by which progeny differ from their parents.

Term	हिंदी	Meaning
Gene / factor	जीन / कारक	Unit of inheritance
Allele	विकल्पी	Slightly different forms of the same gene (T & t)
Homozygous	समयुग्मजी	Identical alleles – TT, tt
Heterozygous	विषमयुग्मजी	Different alleles – Tt
Genotype	जीनप्ररूप	Genetic make-up (TT, Tt, tt)
Phenotype	लक्षणप्ररूप	Observable character (tall/dwarf)
True breeding	शुद्ध प्रजनन	Shows stable trait inheritance for several generations (continuous self-pollination)

Humans knew about inheritance since 8000–1000 BC – **artificial selection & domestication**; e.g. **Sahiwal cows** in Punjab.

2. Mendel's Experiments / मेंडल के प्रयोग

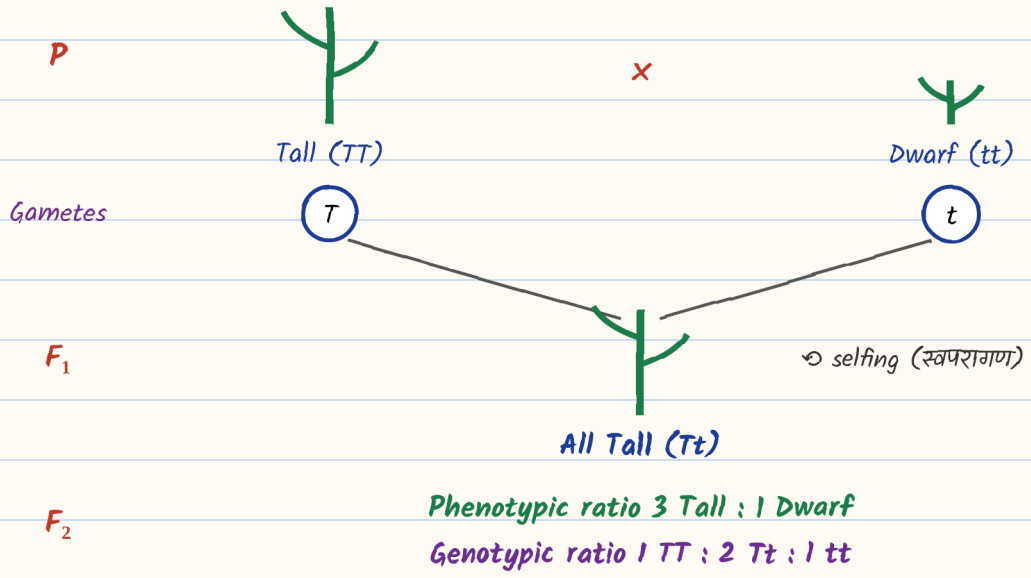
- **Gregor Mendel** – "Father of Genetics"; hybridisation experiments on **garden pea** (*Pisum sativum*) for 7 years (1856–1863).
- First to apply **statistical analysis & mathematical logic** to biology; large sample size → credible data.
- Selected 14 true-breeding pea varieties = **7 pairs** of contrasting traits.

📌 **Why pea? (2–3M)** · Many contrasting traits · Naturally self-pollinating, true-breeding lines · Easy artificial cross-pollination (bisexual flowers) · Short life cycle · Large number of seeds.

#	Character	Dominant ✔	Recessive
1	Stem height (तने की ऊँचाई)	**Tall**	Dwarf
2	Flower colour (पुष्प रंग)	**Violet**	White
3	Flower position (पुष्प स्थिति)	**Axial**	Terminal
4	Pod shape (फली आकार)	**Inflated**	Constricted
5	Pod colour (फली रंग)	**Green**	Yellow
6	Seed shape (बीज आकार)	**Round**	Wrinkled
7	Seed colour (बीज रंग)	**Yellow**	Green

📌 **Trick:** Pod is **green** (dominant) but seed is **yellow** (dominant) – students often mix these up! "**G**reen **P**od, **Y**ellow **S**eed" → GP-YS.

3. Inheritance of One Gene – Monohybrid Cross / एकसंकर संकरण



Infographic: Monohybrid cross in pea plant · मटर में एकसंकर संकरण

F_2 Punnett square (by **Reginald C. Punnett**) (पनेट वर्ग):

♂ \ ♀	**T**	**t**
T	TT Tall	Tt Tall
t	Tt Tall	tt Dwarf

Mendel's observations निष्कर्ष

- F_1 resembled only **one parent** – no blending; the other trait **disappeared** but **reappeared in F_2** unchanged.
- Proposed something is passed unchanged from parent to offspring via gametes = "**factors**" (now called **genes**).
- Factors occur in **pairs**; in a dissimilar pair, one **dominates** the other.

Test cross परीक्षार्थ संकरण

Organism showing dominant phenotype (genotype unknown) is crossed with the recessive parent to find its genotype.

If tall plant is...	× tt gives...	Ratio
TT (homozygous)	all Tall (Tt)	100% tall
Tt (heterozygous)	Tall (Tt) : Dwarf (tt)	1 : 1

Mendel's first two laws मेंडल के नियम

① **Law of Dominance** (प्रभाविता का नियम):

(i) Characters are controlled by discrete units called factors. (ii) Factors occur in pairs. (iii) In a dissimilar pair, one member **dominates** (dominant) and the other is **recessive**. → Explains **3:1** in F_2 and expression of only one parental character in F_1.

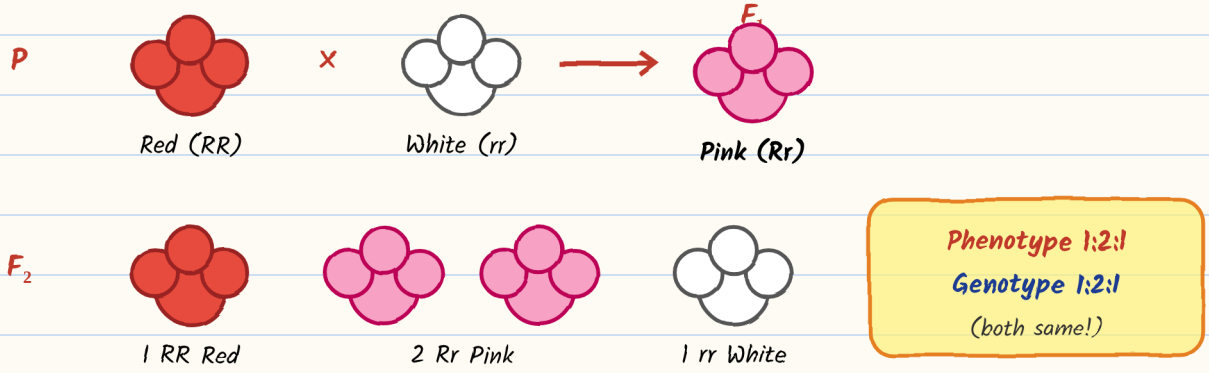
② **Law of Segregation** (पृथक्करण का नियम):

Alleles do not show blending; both characters are recovered as such in F_2. During gamete formation, the **two alleles of a pair separate** so that each gamete gets **only one** of the two factors. Also called "Law of purity of gametes".

4. Deviations from Mendelism / मेंडलवाद से विचलन

(a) Incomplete dominance अपूर्ण प्रभाविता

F_1 has a phenotype **in between** the two parents. e.g. Snapdragon / Dog flower (*Antirrhinum* sp.) flower colour.



Infographic: Incomplete dominance in snapdragon · अपूर्ण प्रभाविता

📌 **Concept of dominance:** Dominance is not an autonomous feature of a gene – it depends on the **gene product & the phenotype** we choose to examine. Example: **pea seed shape** – gene for starch synthesis:

BB → large starch grains, **round** · **bb** → less efficient, **wrinkled** · **Bb** → round seeds but **intermediate** starch grain size.

→ If seed shape is the phenotype: **dominance**; if starch grain size: **incomplete dominance**.

(b) Co-dominance सहप्रभाविता

F_1 resembles **both parents** – both alleles express **fully**. e.g. ABO blood groups in humans.

- Controlled by gene **I** (isoagglutinin); RBC plasma membrane has sugar polymers whose type is controlled by this gene.
- Three alleles: I^A, I^B, **i**. I^A & I^B produce slightly different sugar; **i** produces none.
- I^A and I^B are completely dominant over **i**; but I^A & I^B are co-dominant to each other.
- 3 alleles → 6 genotypes, 4 phenotypes.

Allele from parent 1	Allele from parent 2	Genotype	Blood group
I^A	I^A	I^AI^A	**A**
I^A	i	I^Ai	**A**
I^B	I^B	I^BI^B	**B**
I^B	i	I^Bi	**B**
I^A	I^B	I^AI^B	**AB** (co-dominance)
i	i	ii	**O**

(c) Multiple allelism बहुविकल्पता

More than two alleles governing the same character, e.g. ABO (I^A, I^B, i). Since an individual has only two alleles, multiple alleles can be found only in a **population**.

5. Inheritance of Two Genes – Dihybrid Cross / द्विसंकर संकरण

P: Round Yellow **RRYY** × Wrinkled Green **rryy** → Gametes: **RY** & **ry** →

F_1: all Round Yellow **RrYy** → **selfing** → F_1 gametes: RY, Ry, rY, ry (¼ each)

♂ \ ♀	RY	Ry	rY	ry
RY	RRYY	RRYy	RrYY	RrYy
Ry	RRYy	RRyy	RrYy	Rryy
rY	RrYY	RrYy	rrYY	rrYy
ry	RrYy	Rryy	rrYy	rryy

Round Yellow 9 | Round Green 3 | Wrinkled Yellow 3 | Wrinkled Green 1

F_2 phenotypic ratio = 9 : 3 : 3 : 1

Each pair separately still gives 3:1 → Round : Wrinkled = 12 : 4 = 3:1; Yellow : Green = 3:1. So 9:3:3:1 = (3:1) × (3:1).

Dihybrid test cross (RrYy × rryy) → 1 : 1 : 1 : 1.

③ **Law of Independent Assortment** (स्वतंत्र अपव्यूहन का नियम):
When two pairs of traits are combined in a hybrid, segregation of one pair of characters is **independent** of the other pair of characters.
संकर में जब दो जोड़ी लक्षण साथ हों, तो एक जोड़ी लक्षण का पृथक्करण दूसरी जोड़ी से स्वतंत्र होता है।

📌 **History:** Mendel published his work in **1865**, but it remained unrecognised till **1900** (reasons: lack of communication, "factors" concept not accepted, mathematical approach new, no proof for existence of factors). Rediscovered independently in 1900 by **de Vries, Correns & von Tschermak**.

6. Chromosomal Theory of Inheritance / वंशागति का गुणसूत्रीय सिद्धांत

- Proposed by Walter Sutton & Theodore Boveri (1902).
- Chromosomes & genes both occur in **pairs**; the two alleles of a gene pair are located on **homologous sites** on homologous chromosomes.
- Sutton united chromosomal segregation with Mendelian principles → chromosomal theory.
- Experimental verification by Thomas Hunt Morgan using fruit fly ***Drosophila melanogaster***.

Gene behaviour (Mendel)	Chromosome behaviour (meiosis)
Occur in pairs	Occur in pairs
Segregate at gamete formation; only one of each pair transmitted	Segregate at gamete formation; only one of each pair transmitted
Independent pairs segregate independently	One pair segregates independently of another

📌 **Why Drosophila? (2–3M)** · Grown on simple synthetic medium in lab · Completes life cycle in about **2 weeks** · Single mating → large number of progeny · Clear **sexual differentiation** (male & female easily distinguishable) · Many types of hereditary variations seen with low-power microscopes.

Linkage & Recombination सहलग्नता व पुनर्योजन

- Morgan did dihybrid crosses in Drosophila for **sex-linked genes** (yellow body/brown body; white eye/red eye; miniature wing).
- F_2 ratios **deviated significantly** from 9:3:3:1, because genes were on the **same (X) chromosome**.
- Linkage (term by Morgan) = physical association of genes on a chromosome.
- Recombination = generation of non-parental gene combinations.
- **Tightly linked** genes → very low recombination; **loosely linked** → higher recombination.
- **Alfred Sturtevant** (Morgan's student) used **frequency of recombination** between gene pairs as a measure of distance → genetic maps (chromosome maps).

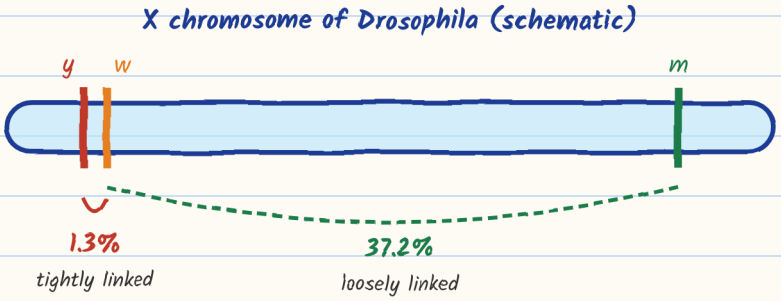


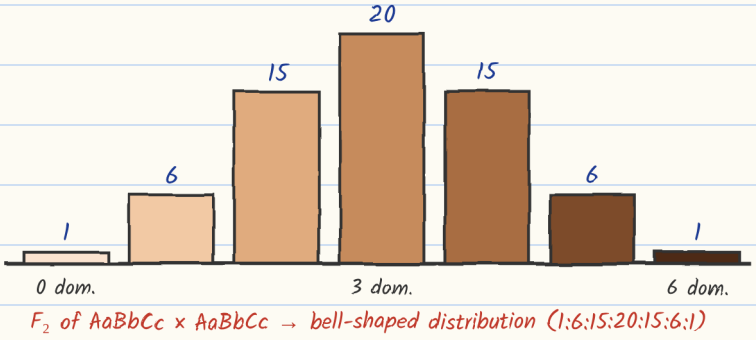
Fig: y = yellow body, w = white eye, m = miniature wing · recombination %

📌 **Remember:** Recombination between **yellow–white = 1.3%**; **white–miniature = 37.2%**. Closer genes → less recombination.

7. Polygenic Inheritance & Pleiotropy / बहुजीनी वंशागति व बहुप्रभाविता

(a) Polygenic inheritance बहुजीनी वंशागति

- Traits controlled by **three or more genes**; phenotype reflects **contribution of each allele** (additive effect) + influence of environment.
- e.g. **human skin colour**, **height** – show a gradient, not distinct categories.
- Skin colour: 3 genes A, B, C. **AABBCC** → darkest; **aabbcc** → lightest; **AaBbCc** → intermediate.



Infographic: Polygenic inheritance of skin colour

(b) Pleiotropy बहुप्रभाविता

- **Single gene** exhibits **multiple phenotypic expressions** (opposite of polygenic).
- Mechanism: effect of a gene on metabolic pathways contributing towards different phenotypes.
- e.g. Phenylketonuria (PKU) – mutation in gene for enzyme **phenylalanine hydroxylase** → mental retardation + reduced hair & skin pigmentation.

8. Sex Determination / लिंग निर्धारण

- **Henking (1891)** – observed a specific nuclear structure in 50% of sperms of some insects → named it **X body** (later = **X chromosome**).
- Chromosomes involved in sex determination = **sex chromosomes** (allosomes); rest = **autosomes**.

XX – XO	XX – XY	ZW – ZZ	Haplo-diploid
Grasshopper	Humans, Drosophila	Birds	Honey bee
♀ XX (autosomes+XX)	♀ XX	♀ ZW	♀ diploid 2n = 32 (fertilised egg)
♂ XO (only one X)	♂ XY	♂ ZZ	♂ drone haploid n = 16 (unfertilised – parthenogenesis)
♂ heterogametic	♂ heterogametic	♀ heterogametic	♂ sperms by mitosis
50% sperms X, 50% none	sperm X or Y decides sex	egg decides sex	

Infographic: Types of sex determination · लिंग निर्धारण के प्रकार

Sex determination in humans मनुष्य में लिंग निर्धारण

- 23 pairs: **22 pairs autosomes + 1 pair sex chromosomes**. ♀ = 44 + XX; ♂ = 44 + XY.

- Male produces two types of sperms: **22+X** and **22+Y** (50% each); female produces only **22+X** ova.
- Sex of the baby is determined by the father; each pregnancy has a 50% chance of male or female.

♀ ova \ ♂ sperm	X	Y
X	XX Girl	XY Boy

📌 **Honey bee facts:** Males (drones) have **no father** → cannot have sons, but have a **grandfather** and can have **grandsons**.

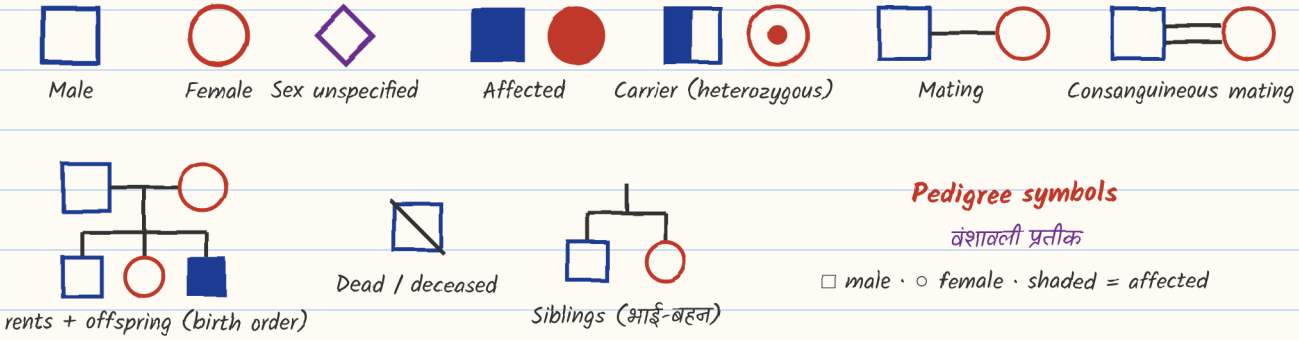
9. Mutation / उत्परिवर्तन

- Phenomenon resulting in **alteration of DNA sequences** → change in genotype & phenotype. Also a source of **variation**.
- **Chromosomal aberrations** (गुणसूत्रीय विपथन): loss (**deletion**) or gain (**insertion/duplication**) of a DNA segment → commonly seen in cancer cells.
- **Point mutation** (बिंदु उत्परिवर्तन): change in a **single base pair** – e.g. sickle-cell anaemia.
- **Frameshift mutations**: deletions & insertions of base pairs of DNA.
- **Mutagens** (उत्परिवर्तजन): chemical & physical factors inducing mutation, e.g. **UV radiation**.

10. Genetic Disorders / आनुवंशिक विकार

(a) Pedigree analysis वंशावली विश्लेषण

Analysis of traits in several generations of a family = **pedigree analysis**. Used to trace inheritance of a trait, abnormality or disease. Control crosses aren't possible in humans, so pedigrees are used.



Infographic: Symbols used in human pedigree analysis

(b) Mendelian disorders मेंडलीय विकार

Determined by alteration/mutation in a **single gene**; inherited following Mendel's principles; traced by pedigree. Can be **dominant or recessive**, **autosomal or sex-linked**.

Disorder	Type	Key points
Haemophilia हीमोफीलिया	Sex-linked (X) recessive	A protein in the clotting cascade is affected → simple cut → **non-stop bleeding**. Heterozygous ♀ = **carrier**. ♀ becomes haemophilic very rarely (mother must be carrier + father haemophilic; usually not survive later). Seen in descendants of **Queen Victoria** (she was a carrier) – "royal disease".
Sickle-cell anaemia दात्र कोशिका अरक्तता	Autosomal recessive	Disease when both parents are carriers (Hb^AHb^s). Hb^s Hb^s = diseased. **Glutamic acid (Glu) → Valine (Val)** at **6th position of β-globin** chain. Point mutation **GAG → GUG**. Under low O_2 tension, RBCs become **sickle-shaped**.
Phenylketonuria फीनाइलकीटोनूरिया	Autosomal recessive	Lacks enzyme **phenylalanine hydroxylase** → phenylalanine can't convert to tyrosine → accumulates → **phenylpyruvic acid** → mental retardation; excreted in urine (poor kidney absorption).
Thalassemia थैलेसीमिया	Autosomal recessive	Reduced synthesis of α or β globin → abnormal Hb → **anaemia**. **α-thal**: genes HBA1 & HBA2 on **chromosome 16**. **β-thal**: gene HBB on **chromosome 11**. Quantitative problem (too few globin).
Colour blindness वर्णांधता	Sex-linked (X) recessive	Defect in red or green cone of eye → can't distinguish **red & green**. **~8% males**, only **~0.4% females**. Son of carrier mother has 50% chance.

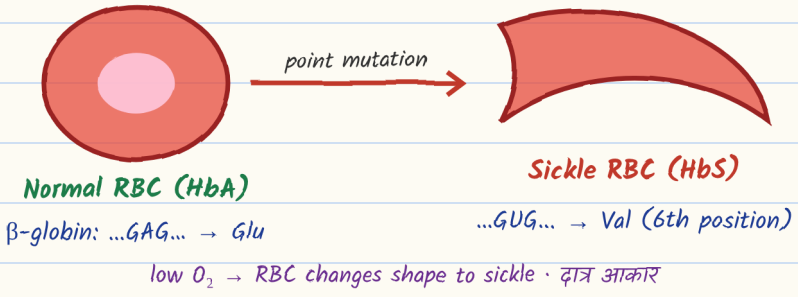


Fig: Normal vs sickle-shaped RBC

📌 **Sickle-cell vs Thalassemia:** Sickle-cell = **qualitative** problem (incorrectly functioning globin). Thalassemia = **quantitative** problem (too few globin molecules). Both autosomal recessive.

(c) Chromosomal disorders गुणसूत्रीय विकार

- Caused by **absence, excess or abnormal arrangement** of one or more chromosomes.

- **Aneuploidy** (असुगुणिता): gain or loss of chromosome(s) due to **failure of segregation of chromatids** during cell division.
- **Polyploidy** (बहुगुणिता): failure of cytokinesis after telophase → increase in a whole set of chromosomes; often seen in **plants**.
- **Trisomy** = additional copy (2n+1); **Monosomy** = lack of one chromosome (2n−1).

Down's syndrome
Trisomy 21 (47)

Langdon Down (1866)
- short stature, small round head
- furrowed tongue, partially open mouth
- broad palm with palm crease
- retarded physical, psychomotor & mental development

Klinefelter's syndrome
47, XXY

extra X chromosome in male
- overall masculine development
- but feminine development too – gynaecomastia (breast development)
- individuals are sterile

♂ (44 + XXY)

Turner's syndrome
45, XO

absence of one X in female
- sterile females
- ovaries rudimentary
- lack secondary sexual characters

♀ (44 + XO)

Infographic: Chromosomal disorders · गुणसूत्रीय विकार

⚡ Quick Revision – Ratios / अनुपात

Cross	Phenotypic ratio	Genotypic ratio
Monohybrid F_2	**3 : 1**	1 : 2 : 1
Monohybrid test cross	1 : 1	1 : 1
Incomplete dominance F_2	**1 : 2 : 1**	1 : 2 : 1
Co-dominance F_2	1 : 2 : 1	1 : 2 : 1
Dihybrid F_2	**9 : 3 : 3 : 1**	1:2:1:2:4:2:1:2:1 (9 genotypes)
Dihybrid test cross	1 : 1 : 1 : 1	1 : 1 : 1 : 1
Polygenic (3 genes) F_2	1:6:15:20:15:6:1	—

Scientist	Contribution
Gregor Mendel	Laws of inheritance (pea) – Father of Genetics
R.C. Punnett	Punnett square
de Vries, Correns, von Tschermak	Rediscovered Mendel (1900)
Sutton & Boveri	Chromosomal theory (1902)
T.H. Morgan	Linkage, recombination (Drosophila)
Alfred Sturtevant	Gene mapping (recombination frequency)
Henking	X body (1891)
Langdon Down	Down's syndrome (1866)

⭐ Exam Corner / परीक्षा उपयोगी प्रश्न

1-Mark Questions 1M

Q1. What is a test cross? / परीक्षार्थ संकरण क्या है?

→ Crossing an organism of dominant phenotype with the recessive parent to know its genotype.

Q2. Give the genotype of a person with blood group AB.

→ I^AI^B

Q3. Name the scientist who proposed the chromosomal theory of inheritance.

→ Sutton and Boveri.

Q4. What is the sex chromosome make-up of a male grasshopper?

→ XO.

Q5. Which amino acid substitution causes sickle-cell anaemia?

→ Glutamic acid → Valine at 6th position of β-globin chain.

Q6. Karyotype of Turner's syndrome?

→ 45, XO (44 + XO).

Q7. Name a disorder showing pleiotropy.

→ Phenylketonuria.

2/3-Mark Questions 2–3M

Q8. State the law of segregation. Why is it called the law of purity of gametes?

→ Alleles separate at gamete formation; each gamete gets only one allele, i.e., gametes are always "pure" for a trait.

Q9. Differentiate incomplete dominance and co-dominance with examples.

→ Incomplete: F_1 intermediate (pink snapdragon). Co-dominance: both alleles expressed fully (AB blood group).

Q10. A man with blood group A marries a woman with blood group B. Their child has blood group O. Give genotypes.

→ Father I^Ai, mother I^Bi, child ii.

Q11. Why did Morgan choose Drosophila? (any 3)

→ Simple medium, 2-week life cycle, many progeny, clear sex differences, visible hereditary variations.

Q12. Explain linkage and recombination with Morgan's example.

→ Genes on same chromosome stay together (linkage); new combinations = recombination; yellow–white 1.3%, white–miniature 37.2%.

Q13. Why is haemophilia more common in males than females?

→ X-linked recessive; males have only one X (hemizygous) so one recessive allele expresses; females need two copies.

Q14. Differentiate between sickle-cell anaemia and thalassemia.

→ Qualitative vs quantitative globin defect; GAG→GUG vs reduced α/β globin synthesis (chromosome 16 / 11).

Q15. Explain sex determination in honey bee.

→ Haplodiploid: fertilised eggs → females (2n=32); unfertilised → males (n=16) by parthenogenesis; males form sperms by mitosis.

5-Mark Questions SM ★★★

Q16. Explain the law of independent assortment with a dihybrid cross. Draw the Punnett square.

→ RRYY × rryy → F_1 RrYy → F_2 9:3:3:1 + Punnett square + statement of law.

Q17. With a monohybrid cross, explain Mendel's laws of dominance and segregation.

→ TT × tt → F_1 Tt → F_2 3:1 & 1:2:1 + laws.

Q18. What is pedigree analysis? Explain two Mendelian disorders and two chromosomal disorders.

→ Definition + symbols; haemophilia, sickle-cell; Down's, Klinefelter's/Turner's.

Q19. Explain ABO blood group inheritance as an example of co-dominance and multiple allelism.

→ Gene I, 3 alleles, 6 genotypes, 4 phenotypes; I^AI^B co-dominant.

🎯 **Last-minute tips:** Always show **gametes** in crosses (encircled) · Write both **phenotypic & genotypic** ratios · Use symbols ♂ ♀ and generation labels P, F_1, F_2 · In pedigree questions, check: affected only in males → likely X-linked; skips generations → recessive · Practise ABO blood group "possible child" problems – very common in boards!

~ All the best! / शुभकामनाएँ! ~