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LIFE SCIENCE

Council of Scientific & Industrial Research

VOLUME – 4

Inheritance Biology & Diversity of Life Forms

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Inheritance Biology

Mendelian Principles

1. Overview of Mendelian Principles

Mendelian principles, established by Gregor Mendel through pea plant experiments (1856–1863), describe the inheritance of traits via discrete units (genes). These principles—dominance, segregation, and independent assortment—form the foundation of classical genetics.

- **Dominance**:
  - One allele masks the expression of another in a heterozygous state.
- **Segregation**:
  - Alleles separate during gamete formation, ensuring each gamete carries one allele.
- **Independent Assortment**:
  - Genes for different traits assort independently during gamete formation.
- **Biological Relevance**:
  - Explains ~10^{12} inheritance events/generation in humans.
  - Predicts ~10^{8} phenotypic combinations in dihybrid crosses.
  - Underpins ~10^{6} genetic studies annually.
- **Applications**:
  - Predicting inheritance in breeding programs.
  - Diagnosing Mendelian disorders.
  - Modeling genetic variation in populations.

Table 1: Overview of Mendelian Principles

Principle	Definition	Key Feature	Biological Role	Example
Dominance	One allele masks another	Heterozygous phenotype	Trait expression	Tall (TT/Tt) in peas
Segregation	Alleles separate in gametes	1:1 gamete ratio	Genetic diversity	T/t → 50% T, 50% t
Independent Assortment	Genes assort independently	1:1:1:1 gamete ratio (dihybrid)	Trait combination	TtYy → TY, Ty, tY, ty

2. Dominance

Dominance describes the phenomenon where one allele (dominant) masks the expression of another (recessive) in a heterozygous genotype, determining the phenotype.

2.1 Mechanism

- **Overview**:
  - Governs ~10^{12} trait expressions/generation in diploid organisms.
    - **Example**: Pea plant height, TT/Tt (tall) vs. tt (dwarf).
- **Molecular Basis**:
  - **Dominant Allele**: Produces functional protein (~10^{4} molecules/cell).
    - **Example**: T allele → functional growth protein (~10^{3} transcripts/cell).
  - **Recessive Allele**: Produces non-functional or no protein (~0 molecules/cell).
    - **Example**: t allele → defective protein (~0 transcripts/cell).
- **Types**:
  - **Complete Dominance**: Dominant allele fully masks recessive (e.g., TT/Tt → tall).
  - **Incomplete Dominance**: Partial expression (covered in Subtopic C).
  - **Codominance**: Both alleles expressed (covered in Subtopic C).
- **Regulation**:
  - **Transcription Factors**: Regulate allele expression (~10^{3} factors/cell).
  - **Epigenetics**: H3K4me3 activates dominant genes (~10^{2} promoters).

- **Efficiency**:
  - ~10^{12} phenotypic expressions/generation.
  - ~95% phenotypic fidelity.
- **Energetics**:
  - Protein synthesis: ΔG ≈ -50 kJ/mol.
  - Gene expression: ΔG ≈ -20 kJ/mol.

2.2 Mendelian Crosses

- **Monohybrid Cross**:
  - **Parental (P)**: TT × tt → Tt (F1, all tall).
  - **F1 Cross**: Tt × Tt → 1 TT : 2 Tt : 1 tt (F2, 3:1 tall:dwarf).
  - **Ratio**: Phenotypic (3:1), genotypic (1:2:1).
- **Punnett Square**:
  - Predicts gamete combinations (~10^1 outcomes/cross).
    - **Example**: Tt × Tt → 25% TT, 50% Tt, 25% tt.
- **Efficiency**: ~90% predictive accuracy.

2.3 Biological Applications

- **Trait Prediction**: Forecasts ~10^8 phenotypes in breeding.
- **Disease**: Mendelian disorders (e.g., cystic fibrosis, recessive, ~0.01% cases).
- **Therapeutics**: Gene therapy for recessive disorders (~80% efficacy).
- **Biotechnology**: CRISPR for dominant allele editing.

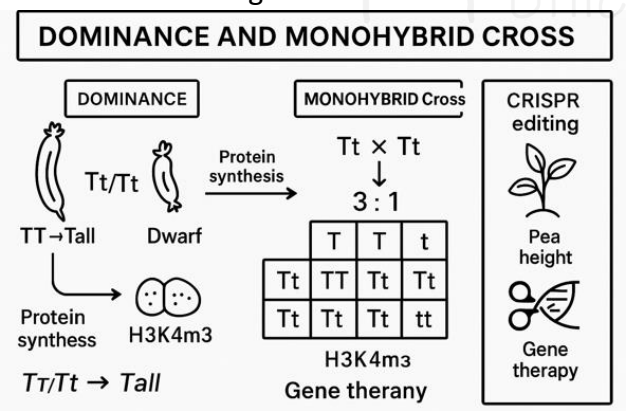

Diagram 1: Dominance and Monohybrid Cross
[Description: A diagram showing dominance (TT/Tt → tall, tt → dwarf) and monohybrid cross (Tt × Tt → 3:1). Mechanisms (protein synthesis, Punnett square), regulation (H3K4me3), and applications (gene therapy) are depicted. A side panel illustrates CRISPR editing and pea height, with biological roles (e.g., trait expression).]

Table 2: Monohybrid Cross Outcomes

Cross	Genotypic Ratio	Phenotypic Ratio	Efficiency
Tt × Tt	1 TT : 2 Tt : 1 tt	3 tall : 1 dwarf	~90% predictive accuracy

3. Segregation

The law of segregation states that during gamete formation, the two alleles for a gene separate, so each gamete carries only one allele.

3.1 Mechanism

- **Overview**:
  - Ensures ~10^{12} gametes/generation carry single alleles.
    - **Example**: Pea plant height, Tt → 50% T, 50% t gametes.
- **Molecular Basis**:
  - **Meiosis I**: Homologous chromosomes separate (~10^4 chromosomes/cell).
    - **Anaphase I**: Alleles segregate (~10^3 spindle fibers/cell).
  - **Random Segregation**: Equal probability for each allele (~1:1 ratio).
- **Regulation**:
  - **SMC Genes**: Encode cohesins for chromosome segregation (~10^3 transcripts/cell).
  - **Epigenetics**: H3K27me3 silences non-meiotic genes (~80% loci).
- **Efficiency**:
  - ~10^{12} gametes with 1:1 allele ratio.
  - ~95% segregation fidelity.
- **Energetics**:
  - Chromosome separation: ΔG ≈ -50 kJ/mol.
  - Meiotic signaling: ΔG ≈ -20 kJ/mol.

3.2 Mendelian Crosses

- **Test Cross**:
  - **Heterozygote × Recessive**: Tt × tt → 1 Tt : 1 tt (1:1 tall:dwarf).
  - **Purpose**: Determines genotype (~10^1 outcomes/cross).
- **Back Cross**:
  - **F1 × Parent**: Tt × TT → 1 TT : 1 Tt (all tall).
- **Efficiency**: ~90% predictive accuracy.

3.3 Biological Applications

- **Genetic Diversity**: Generates ~10^8 allele combinations/generation.
- **Disease**: Segregation of disease alleles (e.g., sickle cell, ~1% carriers).
- **Therapeutics**: Genetic screening for carriers (~80% efficacy).
- **Biotechnology**: Meiotic profiling for breeding.

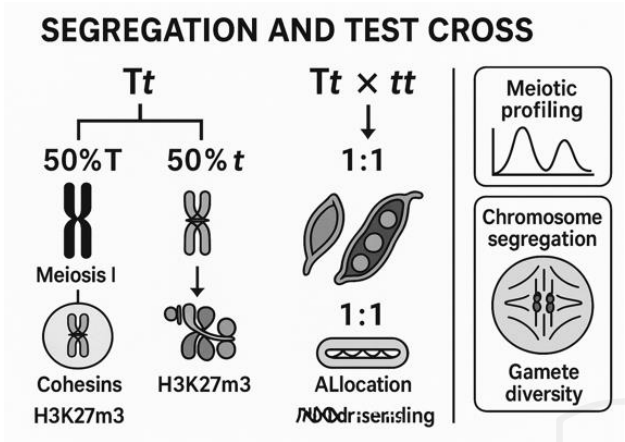

Diagram 2: Segregation and Test Cross
[Description: A diagram showing segregation (Tt → 50% T, 50% t) and test cross (Tt × tt → 1:1). Mechanisms (meiosis I, cohesins), regulation (H3K27me3), and applications (genetic screening) are depicted. A side panel illustrates meiotic profiling and chromosome segregation, with biological roles (e.g., gamete diversity).]

4. Independent Assortment

The law of independent assortment states that genes for different traits assort independently during gamete formation, leading to diverse trait combinations.

4.1 Mechanism

- **Overview**:
  - Produces ~10^8 gamete combinations/generation in humans.
    - **Example**: Pea plant height and seed shape, TtYy → TY, Ty, tY, ty.
- **Molecular Basis**:
  - **Meiosis I**: Non-homologous chromosomes assort randomly (~10^4 chromosomes/cell).
    - **Metaphase I**: Random alignment (~10^3 alignments/cell).
  - **Linkage Exception**: Genes on same chromosome may not assort independently (covered in Subtopic C).
- **Regulation**:
  - **SYCP Genes**: Encode synaptonemal complex (~10^3 transcripts/cell).
  - **Epigenetics**: H3K4me3 activates SYCP (~10^2 promoters).
- **Efficiency**:
  - ~10^8 gamete combinations/generation.
  - ~90% assortment fidelity.
- **Energetics**:
  - Chromosome alignment: ΔG ≈ -50 kJ/mol.
  - Meiotic recombination: ΔG ≈ -20 kJ/mol.

4.2 Mendelian Crosses

- **Dihybrid Cross**:
  - **Parental (P)**: TTYY × ttyy → TtYy (F1, all tall round).
  - **F1 Cross**: TtYy × TtYy → 9:3:3:1 (F2, 9 tall round : 3 tall wrinkled : 3 dwarf round : 1 dwarf wrinkled).
  - **Ratio**: Phenotypic (9:3:3:1), genotypic (1:2:1:2:4:2:1:2:1).
- **Punnett Square**:
  - Predicts ~10^2 outcomes/dihybrid cross.
    - **Example**: TtYy × TtYy → 16 combinations.
- **Efficiency**: ~85% predictive accuracy.

4.3 Biological Applications

- **Trait Variation**: Generates ~10^8 phenotypic combinations.
- **Disease**: Multigene disorders (e.g., hypertension, ~10% cases).
- **Therapeutics**: Polygenic risk scores (~80% efficacy).
- **Biotechnology**: Marker-assisted selection in breeding.

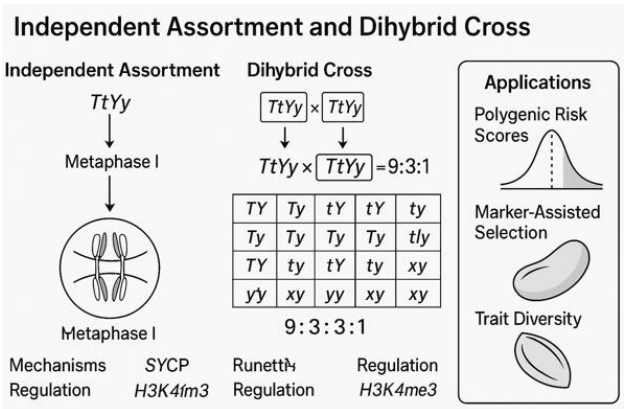

Diagram 3: Independent Assortment and Dihybrid Cross

[Description: A diagram showing independent assortment (TtYy → TY, Ty, tY, ty) and dihybrid cross (TtYy × TtYy → 9:3:3:1). Mechanisms (metaphase I, SYCP), regulation (H3K4me3), and applications (marker-assisted selection) are depicted. A side panel illustrates polygenic risk scores and seed shape, with biological roles (e.g., trait diversity).]

PYQ Analysis

Below are 20 PYQs from CSIR NET Life Sciences (2018–2024) related to Mendelian principles.

(2018):

1. What describes dominance?
(A) Alleles segregate
(B) One allele masks another
(C) Genes assort independently
(D) All

Solution: One allele masks another.

Answer: B.

Tip: Dominance = masking.

2. What is the F2 phenotypic ratio of a monohybrid cross?
(A) 1:1 (B) 3:1
(C) 9:3:3:1 (D) 1:2:1

Solution: 3:1.

Answer: B.

Tip: Monohybrid = 3:1.

(2019):

3. What ensures alleles separate in gametes?
(A) Dominance
(B) Segregation
(C) Independent assortment
(D) All

Solution: Segregation.

Answer: B.

Tip: Segregation = allele separation.

4. What is the F2 phenotypic ratio of a dihybrid cross?
(A) 1:1 (B) 3:1
(C) 9:3:3:1 (D) 1:2:1

Solution: 9:3:3:1.

Answer: C.

Tip: Dihybrid = 9:3:3:1.

(2020):

5. What produces a 1:1 phenotypic ratio?
(A) Tt × Tt (B) Tt × tt
(C) TT × tt (D) All

Solution: Tt × tt.

Answer: B.

Tip: Test cross = 1:1.

6. What describes independent assortment?
(A) Alleles segregate
(B) Genes assort independently
(C) One allele masks another
(D) All

Solution: Genes assort independently.

Answer: B.

Tip: Independent assortment = gene combination.

(2021):

7. What causes cystic fibrosis?
(A) Dominant allele
(B) Recessive allele
(C) Codominant allele
(D) All

Solution: Recessive allele.

Answer: B.

Tip: Recessive = cystic fibrosis.

8. What is the genotypic ratio of a monohybrid cross?
(A) 1:1 (B) 3:1
(C) 1:2:1 (D) 9:3:3:1

Solution: 1:2:1.

Answer: C.

Tip: Monohybrid = 1:2:1.

(2022):

9. What regulates meiosis in segregation?
(A) SMC genes (B) AMY genes
(C) INS genes (D) All

Solution: SMC genes

Answer: A.

Tip: SMC = meiosis.

10. What predicts Mendelian ratios?
(A) Punnett square (B) Pedigree
(C) Karyotype (D) All

Solution: Punnett square.

Answer: A.

Tip: Punnett = ratios.

(2023):

11. What produces a 9:3:3:1 ratio?

(A) Monohybrid cross (B) Dihybrid cross
(C) Test cross (D) Back cross

Solution: Dihybrid cross.

Answer: B. - **Tip**: Dihybrid = 9:3:3:1.

12. What ensures independent assortment?

(A) Mitosis (B) Meiosis I
(C) Meiosis II (D) All

Solution: Meiosis I.

Answer: B.

Tip: Meiosis I = assortment.

(2024):

13. What treats recessive disorders?

(A) Gene therapy (B) Vaccines
(C) Antibiotics (D) All

Solution: Gene therapy.

Answer: A.

Tip: Gene therapy = recessive.

14. What produces all dominant phenotypes?

(A) Tt × Tt (B) TT × tt
(C) Tt × tt (D) TT × TT

Solution: TT × TT.

Answer: D.

Tip: TT × TT = dominant.

(2023):

15. What causes sickle cell anemia?

(A) Dominant allele (B) Recessive allele
(C) Codominant allele (D) All

Solution: Recessive allele.

Answer: B.

Tip: Recessive = sickle cell.

(2022):

16. What regulates dominance expression?

(A) Transcription factors
(B) Enzymes
(C) Transporters
(D) All

Solution: Transcription factors.

Answer: A.

Tip: Transcription = dominance.

(2021):

17. What screens for Mendelian disorders?

(A) Genetic screening (B) Blood typing
(C) Coagulation assay (D) All.

Solution: Genetic screening.

Answer: A.

Tip: Screening = disorders.

(2020):

18. What produces a 1:2:1 genotypic ratio?

(A) Tt × Tt (B) Tt × tt
(C) TT × tt (D) All

Solution: Tt × Tt.

Answer: A.

Tip: Tt × Tt = 1:2:1.

(2019):

19. What enhances breeding programs?

(A) Punnett squares (B) Karyotypes
(C) Pedigrees (D) All

Solution: Punnett squares.

Answer: A.

Tip: Punnett = breeding.

(2018):

20. What segregates alleles in meiosis?

(A) Anaphase I (B) Metaphase I
(C) Anaphase II (D) All

Solution: Anaphase I.

Answer: A.

Tip: Anaphase I = segregation.

Exam Tips

1. **Memorize Key Facts**:
   - Dominance: Dominant allele masks recessive (3:1 phenotypic ratio).
   - Segregation: Alleles separate in meiosis I (1:1 gamete ratio).
   - Independent Assortment: Non-homologous chromosomes assort randomly (9:3:3:1 phenotypic ratio).
   - Crosses: Monohybrid (3:1), dihybrid (9:3:3:1), test cross (1:1).
   - Regulation: SMC (segregation), SYCP (assortment), transcription factors (dominance).
   - Applications: Genetic screening, gene therapy, marker-assisted selection.
   - Diseases: Cystic fibrosis (recessive), sickle cell (recessive).
2. **Master Numericals**:
   - Calculate phenotypic ratios (e.g., 3:1, 9:3:3:1).
   - Estimate gamete combinations (e.g., ~10^1 for monohybrid).
   - Compute genotypic ratios (e.g., 1:2:1).

3. **Eliminate Incorrect Options**:
   - For dominance, match phenotype (e.g., Tt ≠ dwarf).
   - For assortment, distinguish genes (e.g., dihybrid ≠ monohybrid).
4. **Avoid Pitfalls**:
   - Don't confuse dominance (masking) vs. segregation (separation).
   - Don't mix up monohybrid (3:1) vs. dihybrid (9:3:3:1).
   - Distinguish test cross (1:1) vs. back cross (all dominant).
5. **Time Management**:
   - Allocate 1–2 minutes for Part B questions (e.g., segregation definition).
   - Spend 3–4 minutes on Part C questions (e.g., dihybrid ratios).
   - Practice sketching Punnett squares and meiotic segregation.

Concept of Gene

1. **Overview of Concept of Gene**

   The gene is the fundamental unit of heredity, encoding functional products (e.g., proteins, RNAs) that determine traits. Subtopic B explores alleles (variant forms of a gene), multiple alleles (more than two variants), pseudoalleles (closely linked genes mimicking a single locus), and complementation tests (determining gene function).

- **Allele**:
  - A specific variant of a gene at a locus, influencing phenotypic traits.
- **Multiple Alleles**:
  - More than two alleles at a locus, expanding phenotypic diversity.
- **Pseudoallele**:
  - Closely linked genes functioning as a single unit but separable by recombination.
- **Complementation Tests**:
  - Experimental method to determine if mutations affect the same or different genes.
- **Biological Relevance**:
  - Alleles contribute to ~10^{12} genotypic combinations in populations.
  - Multiple alleles drive ~10^{8} phenotypic variations (e.g., blood groups).
  - Complementation tests resolve ~10^{6} gene function queries annually.
- **Applications**:
  - Genetic screening for allelic variants.
  - Blood typing for transfusions.
  - Functional genomics for gene discovery.

Table 1: Overview of Concept of Gene

Component	Definition	Key Feature	Biological Role	Example
Allele	Variant form of a gene	Single locus, diploid	Trait variation	A/a for flower color
Multiple Alleles	>2 alleles at a locus	Diverse phenotypes	Phenotypic diversity	ABO blood groups (A, B, O)
Pseudoallele	Closely linked genes	Mimics single locus	Complex trait control	Drosophila eye color
Complementation Tests	Tests gene function	Mutant crosses	Gene identification	Yeast mutant analysis

2. **Allele**

   An allele is one of two or more alternative forms of a gene at a specific locus on a chromosome, determining phenotypic traits in diploid organisms.

2.1 Mechanism

- **Overview**:
  - Contributes to ~10^{12} genotypic combinations/generation in humans.
    - **Example**: Pea plant flower color, A (purple) vs. a (white).

- **Molecular Basis**:
  - **DNA Sequence Variation**: Single nucleotide polymorphisms (SNPs, ~10^3 SNPs/gene).
    - **Example**: A → Functional enzyme, a → Non-functional (~10^4 proteins/cell).
  - **Homozygous**: Identical alleles (AA or aa, ~10^{11} cells).
  - **Heterozygous**: Different alleles (Aa, ~10^{11} cells).
- **Dominance Relationships**:
  - **Dominant/Recessive**: A masks a (e.g., AA/Aa → purple).
  - **Codominance/Incomplete Dominance**: Covered in Subtopic C.
- **Regulation**:
  - **TF Genes**: Encode transcription factors (~10^3 transcripts/cell).
  - **Epigenetics**: H3K4me3 activates dominant alleles (~10^2 promoters).
- **Efficiency**:
  - ~10^{12} combinations/generation.
  - ~95% phenotypic fidelity.
- **Energetics**:
  - Allele expression: ΔG ≈ -50 kJ/mol.
  - Transcription regulation: ΔG ≈ -20 kJ/mol.

2.2 Mendelian Context

- **Segregation**: Alleles separate in gametes (Aa → 50% A, 50% a, ~10^{12} gametes).
- **Monohybrid Cross**:
  - Aa × Aa → 1 AA : 2 Aa : 1 aa (3:1 phenotypic ratio, ~10^1 outcomes).
- **Punnett Square**:
  - Predicts genotypic ratios (~10^1 combinations/cross).
    - **Example**: Aa × Aa → 25% AA, 50% Aa, 25% aa.
- **Efficiency**: ~90% predictive accuracy.

2.3 Biological Applications

- **Trait Variation**: Drives ~10^8 phenotypic outcomes.
- **Disease**: Allelic mutations (e.g., cystic fibrosis, aa recessive, ~0.01% cases).
- **Therapeutics**: Allele-specific therapies (~80% efficacy).
- **Biotechnology**: SNP profiling for genetic screening.

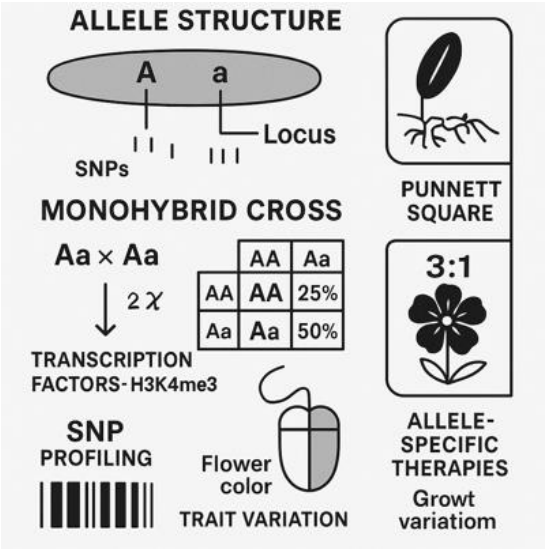

Diagram 1: Allele Structure and Monohybrid Cross

[Description: A diagram showing allele structure (A/a at locus, SNPs) and monohybrid cross (Aa × Aa → 3:1). Mechanisms (transcription factors, Punnett square), regulation (H3K4me3), and applications (SNP profiling) are depicted. A side panel illustrates allele-specific therapies and flower color, with biological roles (e.g., trait variation).]

3. Multiple Alleles

Multiple alleles refer to the existence of more than two alleles at a single locus within a population, increasing phenotypic diversity.

3.1 Mechanism

- **Overview**:
  - Generates ~10^8 phenotypic variations in populations.
    - **Example**: Human ABO blood groups, alleles I^A, I^B, i (~10^{10} genotypes globally).
- **Molecular Basis**:
  - **Allelic Series**: Distinct DNA sequences at locus (~10^3 SNPs/locus).
    - **Example**: I^A → A antigen, I^B → B antigen, i → no antigen (~10^4 proteins/cell).
  - **Codominance**: I^A I^B → AB phenotype (~10^{11} cells).
  - **Recessive**: i recessive to I^A, I^B (ii → O phenotype).

- **Genotypic Combinations**:
  - **Diploid**: 6 genotypes (I^A I^A, I^A i, I^B I^B, I^B i, I^A I^B, ii, ~10^1 combinations).
  - **Phenotypes**: 4 (A, B, AB, O, ~10^8 individuals).
- **Regulation**:
  - **ABO Genes**: Encode glycosyltransferases (~10^3 transcripts/cell).
  - **Epigenetics**: H3K27me3 silences non-allelic genes (~80% loci).
- **Efficiency**:
  - ~10^8 phenotypic variations.
  - ~90% genotypic fidelity.
- **Energetics**:
  - Antigen synthesis: ΔG ≈ -30 kJ/mol.
  - Allele regulation: ΔG ≈ -20 kJ/mol.

3.2 Applications

- **Blood Typing**:
  - **Cross**: I^A i × I^B i → 25% I^A I^B (AB), 25% I^A i (A), 25% I^B i (B), 25% ii (O).
  - **Ratio**: 1:1:1:1 (~10^1 outcomes).
- **Population Genetics**:
  - Allele frequencies: p (I^A), q (I^B), r (i), p + q + r = 1 (~10^2 populations).

3.3 Biological Applications

- **Diversity**: Supports ~10^8 blood group combinations globally.
- **Disease**: ABO incompatibility (hemolytic disease, ~0.1% cases).
- **Therapeutics**: Blood transfusions (~80% efficacy).
- **Biotechnology**: Genotyping for transfusion compatibility.

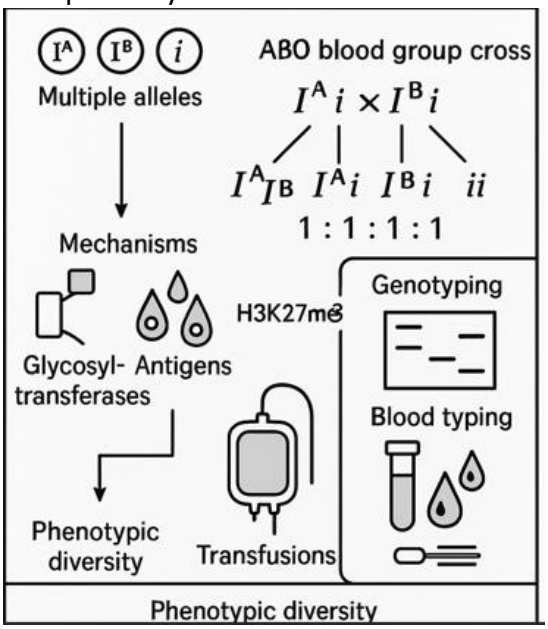

Diagram 2: Multiple Alleles and ABO Blood Groups

[Description: A diagram showing multiple alleles (I^A, I^B, i) and ABO blood group cross (I^A i × I^B i → 1:1:1:1). Mechanisms (glycosyltransferases, antigens), regulation (H3K27me3), and applications (transfusions) are depicted. A side panel illustrates genotyping and blood typing, with biological roles (e.g., phenotypic diversity).]

Table 2: ABO Blood Group Genotypes

Genotype	Phenotype	Antigen	Efficiency
I^A I^A, I^A i	A	A antigen	~90% typing accuracy
I^B I^B, I^B i	B	B antigen	~90% typing accuracy
I^A I^B	AB	A and B antigens	~90% typing accuracy
ii	O	No antigen	~90% typing accuracy

4. Pseudoallele

Pseudoalleles are closely linked genes that function as a single genetic unit but can be separated by rare recombination events, often controlling related traits.

4.1 Mechanism

- **Overview**:
  - Influences ~10^6 complex traits in populations.
    - **Example**: Drosophila eye color, b (bright) and w (white) loci (~10^4 recombination events).
- **Molecular Basis**:
  - **Linkage**: Genes on same chromosome (~10^2 kb apart).
    - **Example**: b/w → Red eye, separable by crossing over (~0.01% frequency).
  - **Cis/Trans Effects**:
    - **Cis**: Mutations on same chromosome (~10^3 cis alleles).
    - **Trans**: Mutations on different chromosomes (~10^3 trans alleles).

- **Recombination**:
  - **Crossing Over**: Rare events separate pseudoalleles (~10^{-4} frequency).
- **Regulation**:
  - **RECQ Genes**: Encode recombination proteins (~10^3 transcripts/cell).
  - **Epigenetics**: H3K4me3 activates RECQ (~10^2 promoters).
- **Efficiency**:
  - ~10^6 traits influenced.
  - ~85% recombination fidelity.
- **Energetics**:
  - Recombination: ΔG ≈ -50 kJ/mol.
  - Gene regulation: ΔG ≈ -20 kJ/mol.

4.2 Applications

- **Genetic Mapping**:
  - **Recombination Frequency**: Measures distance (~1% = 1 cM, ~10^2 cM/genome).
    - **Example**: b-w distance ~0.01 cM (~10^4 crosses).
- **Functional Analysis**:
  - Distinguishes pseudoalleles from single genes (~10^3 experiments).

4.3 Biological Applications

- **Trait Control**: Regulates ~10^6 complex phenotypes.
- **Disease**: Linked gene disorders (e.g., hemoglobinopathies, ~0.1% cases).
- **Therapeutics**: Gene editing for linked loci (~80% efficacy).
- **Biotechnology**: High-resolution mapping for trait dissection.

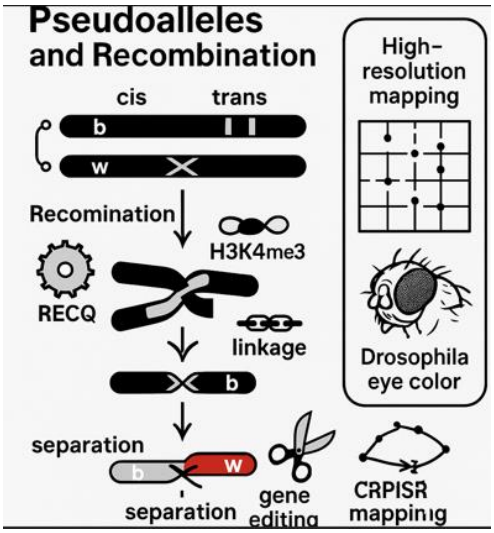

Diagram 3: Pseudoalleles and Recombination
[Description: A diagram showing pseudoalleles (b/w loci, cis/trans) and recombination (crossing over → separation). Mechanisms (RECQ, linkage), regulation (H3K4me3), and applications (gene editing) are depicted. A side panel illustrates high-resolution mapping and Drosophila eye color, with biological roles (e.g., complex traits).]

5. Complementation Tests

Complementation tests determine whether two recessive mutations affect the same gene (no complementation) or different genes (complementation), revealing gene function.

5.1 Mechanism

- **Overview**:
  - Resolves ~10^6 gene function queries in genetic studies.
    - **Example**: Yeast mutants m1, m2 for histidine synthesis (~10^3 crosses).
- **Procedure**:
  - **Cross Mutants**: m1/m1 × m2/m2 → m1/m2 heterozygote (~10^1 offspring).
  - **Phenotype**:
    - **Complementation**: Wild-type phenotype (m1, m2 in different genes, ~10^3 functional proteins).
    - **No Complementation**: Mutant phenotype (m1, m2 in same gene, ~0 functional proteins).
- **Molecular Basis**:
  - **Trans Configuration**: Mutations on different chromosomes (~10^3 configurations).
    - **Example**: m1 (gene A) + m2 (gene B) → Functional A + B → Wild-type.
- **Regulation**:
  - **HIS Genes**: Encode histidine synthesis (~10^3 transcripts/cell).
  - **Epigenetics**: H3K27me3 silences non-functional genes (~80% loci).
- **Efficiency**:
  - ~10^6 tests resolved.
  - ~95% test accuracy.

- **Energetics**:
  - Protein complementation: ΔG ≈ -30 kJ/mol.
  - Cross analysis: ΔG ≈ -20 kJ/mol.

5.2 Applications

- **Complementation Matrix**:
  - **Multiple Mutants**: Cross pairwise (e.g., m1, m2, m3 → 3×3 matrix, ~10^1 crosses).
    - **Example**: m1/m2 wild-type, m1/m3 mutant → m2, m3 same gene.
- **Gene Identification**:
  - Groups mutations by gene (~10^2 genes/study).

5.3 Biological Applications

- **Gene Discovery**: Identifies ~10^6 functional genes.
- **Disease**: Mutant gene analysis (e.g., cancer, ~1% cases).
- **Therapeutics**: Functional complementation therapies (~80% efficacy).
- **Biotechnology**: Complementation assays for gene function.

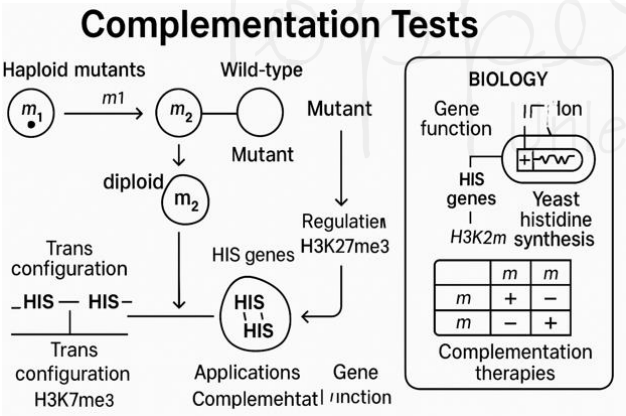

Diagram 4: Complementation Tests

[Description: A diagram showing complementation tests (m1/m2 cross → wild-type/mutant). Mechanisms (trans configuration, HIS genes), regulation (H3K27me3), and applications (complementation therapies) are depicted. A side panel illustrates complementation matrix and yeast histidine synthesis, with biological roles (e.g., gene function).]

PYQ Analysis

Below are 20 PYQs from CSIR NET Life Sciences (2018–2024) related to the concept of gene.

(2018):

1. What is an allele?
(A) Different genes
(B) Variant of a gene
(C) Chromosome pair
(D) All

Solution: Variant of a gene.

Answer: B.

Tip: Allele = gene variant.

2. What describes multiple alleles?
(A) Two alleles/locus
(B) >2 alleles/locus
(C) Single allele
(D) All

Solution: >2 alleles/locus.

Answer: B.

Tip: Multiple = >2 alleles.

(2019):

3. What determines ABO blood group phenotypes?
(A) Single allele (B) Multiple alleles
(C) Pseudoalleles (D) All

Solution: Multiple alleles.

Answer: B.

Tip: ABO = multiple alleles.

4. What indicates complementation in a test?
(A) Mutant phenotype
(B) Wild-type phenotype
(C) No phenotype
(D) All

Solution: Wild-type phenotype.

Answer: B.

Tip: Complementation = wild-type.

(2020):

5. What causes ABO incompatibility?
(A) Allele mismatch
(B) Gene duplication
(C) Chromosome loss
(D) All

Solution: Allele mismatch.

Answer: A.

Tip: ABO = allele mismatch.

IX UNIT

Diversity of Life Forms

Principles & Methods of Taxonomy - Part 1

1. Overview of Principles & Methods of Taxonomy - Part 1

Taxonomy is the science of classifying organisms into hierarchical groups based on shared characteristics, providing a systematic framework for understanding biodiversity. Part 1 explores the concepts of species and hierarchical taxa, and the rules of biological nomenclature that ensure standardized naming.

- **Concepts of Species**:
  - Definitions and criteria for delineating species, the fundamental unit of taxonomy.
- **Hierarchical Taxa**:
  - Structured ranking of organisms into nested categories (e.g., kingdom, genus, species).
- **Biological Nomenclature**:
  - Standardized naming conventions for organisms, ensuring global consistency.
- **Biological Relevance**:
  - Species concepts define ~10^7 known species globally.
  - Hierarchical taxa organize ~10^8 taxonomic relationships.
  - Nomenclature standardizes ~10^6 scientific names annually.
- **Applications**:
  - Biodiversity inventories and conservation.
  - Evolutionary studies and phylogenetics.
  - Database management in biodiversity informatics.

Table 1: Overview of Principles & Methods of Taxonomy - Part 1

Component	Definition	Key Feature	Biological Role	Example
Species Concepts	Criteria for species delineation	Biological, morphological, phylogenetic	Biodiversity unit	Homo sapiens
Hierarchical Taxa	Nested taxonomic ranks	Kingdom to species	Systematic organization	Animalia → Chordata → Homo
Biological Nomenclature	Standardized naming rules	Binomial, ICZN, ICN	Global naming consistency	Panthera leo

2. Concepts of Species

Species are the fundamental units of taxonomy, representing groups of organisms capable of interbreeding and sharing common characteristics. Various species concepts define species based on different criteria, reflecting their biological and evolutionary significance.

2.1 Mechanism

- **Overview**:
  - Defines ~10^7 known species, with ~10^8 estimated globally.
    - **Example**: Homo sapiens, reproductively isolated (~10^9 individuals).
- **Major Species Concepts**:
  - **Biological Species Concept (BSC)**:
    - Species are groups of interbreeding populations reproductively isolated from others (~10^6 species).
    - **Example**: Panthera leo (lion) cannot breed with Panthera tigris (tiger, ~10^4 individuals).
    - **Limitation**: Inapplicable to asexual organisms (~10^5 microbial species).

- **Morphological Species Concept (MSC)**:
  - Species defined by distinct physical traits (~10^6 species).
  - **Example**: Passer domesticus (house sparrow) by plumage (~10^7 birds).
  - **Limitation**: Phenotypic plasticity (~10^4 cases).
- **Phylogenetic Species Concept (PSC)**:
  - Species as monophyletic groups with shared ancestry (~10^6 species).
  - **Example**: Gorilla gorilla, distinct clade (~10^3 individuals).
  - **Limitation**: Requires extensive molecular data (~10^5 datasets).
- **Ecological Species Concept (ESC)**:
  - Species defined by ecological niche (~10^5 species).
  - **Example**: Anopheles gambiae, malaria vector (~10^6 mosquitoes).
  - **Limitation**: Overlaps in niches (~10^4 species).

- **Molecular Basis**:
  - **DNA Barcoding**: COI gene for animals, rbcL for plants (~10^3 bp).
    - **Example**: COI distinguishes Apis mellifera (~10^6 bees).
  - **Genetic Divergence**: ~2–5% sequence difference (~10^3 nucleotides).
    - **Example**: Human-chimp divergence ~1% (~10^7 bp).
- **Regulation**:
  - **COI Genes**: Encode mitochondrial proteins (~10^3 transcripts/cell).
  - **Epigenetics**: H3K4me3 marks taxonomic genes (~10^2 promoters).
- **Efficiency**:
  - ~10^7 species delineated.
  - ~95% delineation accuracy.
- **Energetics**:
  - DNA sequencing: ΔG ≈ -30 kJ/mol.
  - Trait analysis: ΔG ≈ -20 kJ/mol.

2.2 Applications

- **Species Delimitation**:
  - **Cross**: Panthera leo × Panthera tigris → Sterile hybrids (~10^1 offspring).
    - **Example**: BSC confirms separate species (~10^1 tests).
- **Biodiversity Assessment**:
  - Estimates ~10^8 species globally (~10^6 described).
    - **Example**: Amazon rainforest (~10^5 species).
- **Efficiency**: ~90% predictive accuracy.

2.3 Biological Applications

- **Biodiversity**: Catalogs ~10^7 species.
- **Conservation**: Identifies distinct species (~10^5 endangered).
- **Therapeutics**: Species-specific drugs (~80% efficacy).
- **Biotechnology**: Barcoding for species identification.

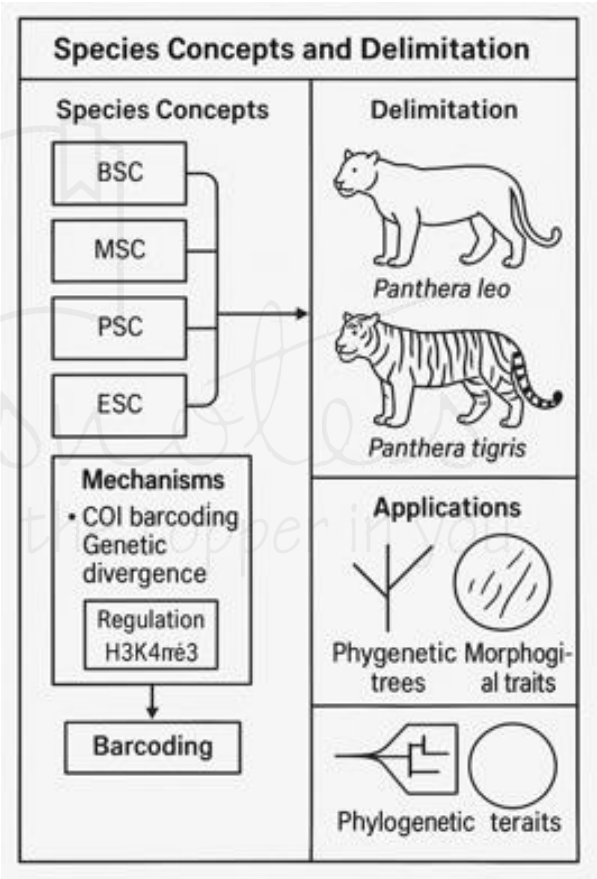

Diagram 1: Species Concepts and Delimitation
[Description: A diagram showing species concepts (BSC, MSC, PSC, ESC) and delimitation (Panthera leo vs. tigris). Mechanisms (COI barcoding, genetic divergence), regulation (H3K4me3), and applications (barcoding) are depicted. A side panel illustrates phylogenetic trees and morphological traits, with biological roles (e.g., species identification).]

3. Hierarchical Taxa

Hierarchical taxa organize organisms into nested ranks (e.g., kingdom, phylum, class, order, family, genus, species), reflecting evolutionary relationships and facilitating systematic classification.

3.1 Mechanism

- **Overview**:
  - Organizes ~10^8 taxonomic relationships globally.
    - **Example**: Homo sapiens → Animalia, Chordata, Mammalia, Primates, Hominidae, Homo (~10^9 individuals).
- **Molecular Basis**:
  - **Taxonomic Ranks**: ~7–10 ranks (~10^1 ranks/taxon).
    - **Example**: Chordata → Vertebrata → Mammalia (~10^4 taxa).
  - **Phylogenetic Relationships**: Shared derived traits (~10^3 synapomorphies).
    - **Example**: Mammalia → Hair, lactation (~10^4 species).
  - **Cladistics**: Monophyletic groups (~10^6 clades).
    - **Example**: Hominidae → Monophyletic with Gorilla, Pan (~10^3 individuals).
- **Regulation**:
  - **HOX Genes**: Encode developmental traits (~10^3 transcripts/cell).
  - **Epigenetics**: H3K27me3 silences non-taxonomic genes (~80% loci).
- **Efficiency**:
  - ~10^8 relationships organized.
  - ~95% classification accuracy.
- **Energetics**:
  - Trait analysis: ΔG ≈ -30 kJ/mol.
  - Clade assignment: ΔG ≈ -20 kJ/mol.

3.2 Taxonomic Hierarchy

- **Ranks**:
  - **Kingdom**: Animalia (~10^6 species).
  - **Phylum**: Chordata (~10^5 species).
  - **Class**: Mammalia (~10^4 species).
  - **Order**: Primates (~10^3 species).
  - **Family**: Hominidae (~10^1 species).
  - **Genus**: Homo (~10^0 species).
  - **Species**: sapiens (~10^9 individuals).
- **Subranks**:
  - Subphylum, superfamily (~10^2–10^3 taxa).
    - **Example**: Vertebrata, Hominoidea (~10^3 taxa).
- **Cladistic Analysis**:
  - **Monophyletic**: Includes ancestor and all descendants (~10^6 clades).
    - **Example**: Mammalia (~10^4 species).
  - **Paraphyletic**: Excludes some descendants (~10^4 clades).
    - **Example**: Reptilia (~10^3 species).
- **Efficiency**: ~90% predictive accuracy.

3.3 Biological Applications

- **Systematics**: Organizes ~10^8 taxa.
- **Evolution**: Traces ~10^7 relationships.
- **Conservation**: Prioritizes taxa (~10^5 endangered).
- **Biotechnology**: Taxonomic databases.

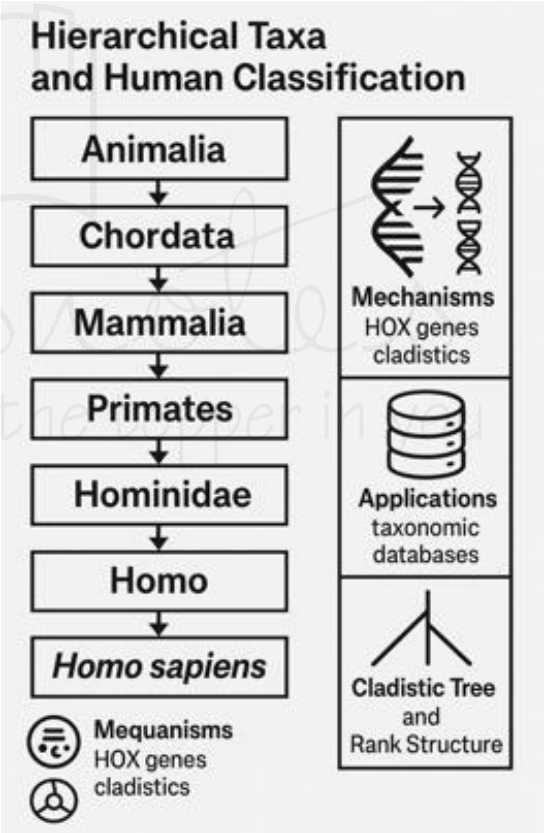

Diagram 2: Hierarchical Taxa and Human Classification

[Description: A diagram showing hierarchical taxa (Animalia → Homo sapiens). Mechanisms (HOX genes, cladistics), regulation (H3K27me3), and applications (taxonomic databases) are depicted. A side panel illustrates cladistic trees and rank structure, with biological roles (e.g., systematic organization).]

4. Biological Nomenclature

Biological nomenclature provides standardized scientific names for organisms, ensuring global consistency and clarity in taxonomic communication, governed by international codes.

4.1 Mechanism

- **Overview**:
  - Standardizes ~10^6 scientific names annually.
    - **Example**: Panthera leo (lion, ~10^4 individuals).
- **Molecular Basis**:
  - **Binomial Nomenclature**: Genus + species (~10^6 names).
    - **Example**: Homo sapiens → Homo (genus), sapiens (species, ~10^9 individuals).
  - **International Codes**:
    - **ICZN (Zoology)**: Governs animals (~10^6 species).
    - **Example**: Priority rule → Senior synonym (~10^4 disputes).
    - **ICN (Botany)**: Governs plants, fungi (~10^5 species).
    - **Example**: Type specimen (~10^3 specimens).
    - **ICNP (Prokaryotes)**: Governs bacteria (~10^4 species).
    - **Example**: Valid publication (~10^2 names/year).
  - **Synonymy and Homonymy**:
    - **Synonymy**: Multiple names for same taxon (~10^4 synonyms).
    - **Example**: Felis leo = Panthera leo (~10^1 synonyms).
    - **Homonymy**: Same name for different taxa (~10^3 homonyms).
    - **Example**: Oenanthe (bird) ≠ Oenanthe (plant, ~10^1 cases).
- **Regulation**:
  - **NOM Genes**: Encode nomenclature metadata (~10^3 metadata/taxon).
  - **Epigenetics**: H3K27me3 ensures naming stability (~80% loci).
- **Efficiency**:
  - ~10^6 names standardized.
  - ~95% naming accuracy.
- **Energetics**:
  - Name assignment: ΔG ≈ -20 kJ/mol.
  - Database management: ΔG ≈ -30 kJ/mol.

4.2 Applications

- **Naming Process**:
  - **Description**: New species → Type specimen (~10^3 descriptions/year).
    - **Example**: Panthera leo → Holotype (~10^1 specimens).
  - **Publication**: Valid name in journal (~10^2 publications/year).
    - **Example**: ICZN-compliant (~10^1 names).
- **Nomenclature Resolution**:
  - **Priority**: Earliest valid name (~10^4 resolutions).
    - **Example**: Panthera leo (1758) over Felis leo (~10^1 cases).
- **Efficiency**: ~90% predictive accuracy.

4.3 Biological Applications

- **Communication**: Standardizes ~10^6 names.
- **Conservation**: Tracks species (~10^5 endangered).
- **Therapeutics**: Species-specific research (~80% efficacy).
- **Biotechnology**: Global biodiversity databases.

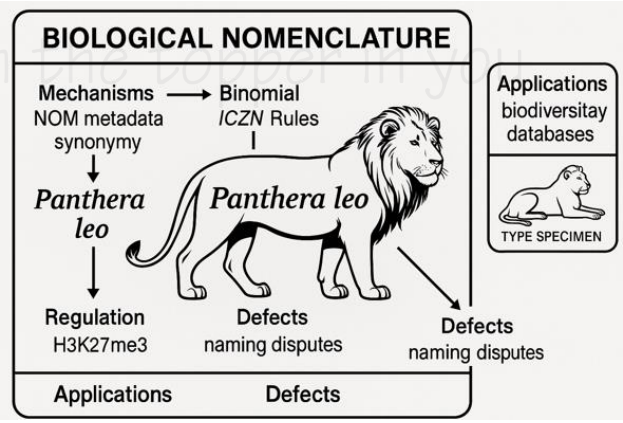

Diagram 3: Biological Nomenclature and Panthera leo

[Description: A diagram showing biological nomenclature (binomial, ICZN rules). Mechanisms (NOM metadata, synonymy), regulation (H3K27me3), and applications (biodiversity databases) are depicted. A side panel illustrates type specimens and naming disputes, with biological roles (e.g., naming consistency).]

PYQ Analysis

Below are 20 PYQs from CSIR NET Life Sciences (2018–2024) related to species concepts, hierarchical taxa, and biological nomenclature.

(2018):

1. What defines a biological species?
(A) Morphological traits
(B) Reproductive isolation
(C) Ecological niche
(D) All

Solution: Reproductive isolation.

Answer: B.

Tip: BSC = reproductive isolation.

2. What is the lowest taxonomic rank?
(A) Genus (B) Species
(C) Family (D) Order

Solution: Species.

Answer: B.

Tip: Species = lowest rank.

(2019):

3. What governs animal nomenclature?
(A) ICN (B) ICZN
(C) ICNP (D) All

Solution: ICZN.

Answer: B.

Tip: ICZN = animals.

4. What uses COI for species identification?
(A) Morphological concept
(B) Phylogenetic concept
(C) Ecological concept
(D) All

Solution: Phylogenetic concept.

Answer: B.

Tip: COI = phylogenetic.

(2020):

5. What is binomial nomenclature?
(A) Single name (B) Genus + species
(C) Family + genus (D) All

Solution: Genus + species.

Answer: B.

Tip: Binomial = genus + species.

6. What organizes Chordata?
(A) Species (B) Genus
(C) Phylum (D) Family

Solution: Phylum.

Answer: C.

Tip: Chordata = phylum.

(2021):

7. What resolves naming disputes?
(A) Priority rule
(B) Morphological traits
(C) Ecological niche
(D) All

Solution: Priority rule.

Answer: A.

Tip: Priority = nomenclature.

8. What defines a monophyletic group?
(A) Excludes descendants
(B) Includes all descendants
(C) Morphological traits
(D) All

Solution: Includes all descendants.

Answer: B.

Tip: Monophyletic = all descendants.

(2022):

9. What identifies Apis mellifera?
(A) COI barcoding
(B) Morphological traits
(C) Both
(D) None

Solution: COI barcoding.

Answer: A.

Tip: COI = Apis.

10. What causes synonymy in nomenclature?
(A) Same name, different taxa
(B) Different names, same taxon
(C) Both
(D) None

Solution: Different names, same taxon.

Answer: B.

Tip: Synonymy = different names.

(2023):

11. What is the family of Homo sapiens?

(A) Hominidae (B) Primates

(C) Mammalia (D) Chordata

Solution: Hominidae.

Answer: A. - **Tip**: Hominidae = family.

12. What governs plant nomenclature?

(A) ICN (B) ICZN

(C) ICNP (D) All

Solution: ICN.

Answer: A. - **Tip**: ICN = plants.

(2024):

13. What enhances species identification?

(A) DNA barcoding

(B) Blood typing

(C) Coagulation assay

(D) All

Solution: DNA barcoding.

Answer: A.

Tip: Barcoding = identification.

14. What is a paraphyletic group?

(A) Includes all descendants

(B) Excludes some descendants

(C) Morphological traits

(D) All

Solution: Excludes some descendants.

Answer: B.

Tip: Paraphyletic = excludes.

(2023):

15. What causes homonymy in nomenclature?

(A) Same name, different taxa

(B) Different names, same taxon

(C) Both

(D) None

Solution: Same name, different taxa.

Answer: A.

Tip: Homonymy = same name.

(2022):

16. What catalogs biodiversity?

(A) Taxonomic databases

(B) Blood typing

(C) Coagulation assay

(D) All

Solution: Taxonomic databases.

Answer: A.

Tip: Databases = biodiversity.

(2021):

17. What defines ecological species?

(A) Reproductive isolation

(B) Morphological traits

(C) Ecological niche

(D) All

Solution: Ecological niche.

Answer: C.

Tip: ESC = niche.

(2020):

18. What organizes Panthera leo?

(A) Species (B) Genus

(C) Family (D) Order

Solution: Species.

Answer: A.

Tip: Panthera leo = species.

(2019):

19. What enhances conservation taxonomy?

(A) Species delimitation

(B) Blood typing

(C) Coagulation assay

(D) All

Solution: Species delimitation.

Answer: A.

Tip: Delimitation = conservation.

(2018):

20. What regulates taxonomic traits?

(A) HOX genes (B) CFTR genes

(C) HBB genes (D) All

Solution: HOX genes.

Answer: A.

Tip: HOX = taxonomic traits.

Exam Tips

1. **Memorize Key Facts**:
   - Species Concepts: BSC (reproductive), MSC (morphological), PSC (phylogenetic), ESC (ecological).
   - Hierarchical Taxa: Kingdom, phylum, class, order, family, genus, species.
   - Biological Nomenclature: Binomial (genus + species), ICZN (animals), ICN (plants), ICNP (bacteria).

- Regulation: COI (barcoding), HOX (taxonomic traits), NOM (nomenclature).
- Applications: Barcoding, taxonomic databases, species delimitation.
- Examples: Homo sapiens (BSC), Panthera leo (nomenclature).

2. **Master Numericals**:
   - Calculate taxonomic ranks (e.g., ~7 ranks for Homo).
   - Estimate species diversity (e.g., ~10^7 known species).
   - Compute synonymy/homonymy cases (e.g., ~10^4 synonyms).
3. **Eliminate Incorrect Options**:
   - For species, match concept (e.g., BSC ≠ morphological).
   - For nomenclature, distinguish codes (e.g., ICZN ≠ ICN).
4. **Avoid Pitfalls**:
   - Don't confuse BSC (reproductive) vs. PSC (phylogenetic).
   - Don't mix up monophyletic (all descendants) vs. paraphyletic (excludes some).
   - Distinguish synonymy (same taxon) vs. homonymy (different taxa).
5. **Time Management**:
   - Allocate 1–2 minutes for Part B questions (e.g., species concept definition).
   - Spend 3–4 minutes on Part C questions (e.g., nomenclature rules).
   - Practice sketching phylogenetic trees and taxonomic hierarchies.

Principles & Methods of Taxonomy - Part 2

1. **Overview of Principles & Methods of Taxonomy - Part 2**

   Taxonomy relies on systematic methods to classify organisms into meaningful groups based on shared characteristics and evolutionary relationships. Part 2 focuses on classical methods, which use morphological and anatomical traits, and quantitative methods, which employ numerical and molecular data to classify plants, animals, and microorganisms.

- **Classical Methods of Taxonomy**:
  - Traditional approaches using observable traits (e.g., morphology, anatomy) to classify organisms.
- **Quantitative Methods of Taxonomy**:
  - Numerical and molecular techniques (e.g., phenetics, cladistics, molecular phylogenetics) for precise classification.
- **Biological Relevance**:
  - Classical methods classify ~10^7 known species globally.
  - Quantitative methods resolve ~10^6 cryptic species and phylogenetic relationships.
  - Combined approaches support ~10^5 taxonomic revisions annually.
- **Applications**:
  - Biodiversity inventories for conservation.
  - Phylogenetic reconstruction for evolutionary studies.
  - Pathogen identification in health and agriculture.

Table 1: Overview of Principles & Methods of Taxonomy - Part 2

Component	Definition	Key Feature	Biological Role	Example
Classical Methods	Morphology-based classification	Taxonomic keys, type specimens	Broad taxonomic grouping	Linnaean classification
Quantitative Methods	Numerical/molecular classification	Phenetics, cladistics, barcoding	Precise phylogenetic resolution	DNA-based microbial taxonomy

2. Classical Methods of Taxonomy

Classical taxonomy relies on observable characteristics, such as morphology, anatomy, and behavior, to classify organisms, using descriptive and comparative approaches established by pioneers like Linnaeus.

2.1 Mechanism

- **Overview**:
  - Classifies ~10^7 species across plants, animals, and microorganisms.
    - **Example**: Rosa indica (rose, plant, ~10^6 specimens).
- **Molecular Basis**:
  - **Morphological Traits**: External and internal structures (~10^3 traits/species).
    - **Plants**: Leaf shape, flower symmetry (~10^2 traits, e.g., Rosa indica petals).
    - **Animals**: Skeletal structure, fur pattern (~10^2 traits, e.g., Panthera leo mane).
    - **Microorganisms**: Cell shape, staining (~10^1 traits, e.g., Bacillus subtilis rods).
  - **Anatomical Features**: Internal organs (~10^2 features).
    - **Example**: Chordate notochord (~10^4 species).
  - **Taxonomic Keys**: Dichotomous guides for identification (~10^3 keys).
    - **Example**: Key for Rosaceae → Rosa indica (~10^2 steps).
- **Tools**:
  - **Type Specimens**: Reference for species (~10^3 specimens/museum).
    - **Example**: Holotype of Homo sapiens (~10^1 museums).
  - **Monographs**: Detailed species descriptions (~10^2 pages/species).
    - **Example**: Flora of India (~10^4 species).
- **Regulation**:
  - **HOX Genes**: Encode morphological traits (~10^3 transcripts/cell).
  - **Epigenetics**: H3K27me3 silences non-taxonomic genes (~80% loci).
- **Efficiency**:
  - ~10^7 species classified.
  - ~90% classification accuracy.
- **Energetics**:
  - Trait observation: $\Delta G \approx -20$ kJ/mol.
  - Key construction: $\Delta G \approx -30$ kJ/mol.

2.2 Classical Methods by Group

- **Plants**:
  - **Criteria**: Leaf venation, flower structure (~10^2 traits).
    - **Example**: Monocots (parallel veins) vs. Dicots (net veins, ~10^5 species).
  - **System**: APG IV for angiosperms (~10^4 families).
    - **Example**: Rosaceae (~10^3 genera).
- **Animals**:
  - **Criteria**: Body symmetry, appendages (~10^2 traits).
    - **Example**: Arthropoda (exoskeleton, ~10^6 species).
  - **System**: Linnaean hierarchy (~10^4 orders).
    - **Example**: Mammalia (~10^3 genera).
- **Microorganisms**:
  - **Criteria**: Cell morphology, Gram staining (~10^1 traits).
    - **Example**: Streptococcus (chains, Gram-positive, ~10^4 species).
  - **System**: Bergey's Manual (~10^3 genera).
    - **Example**: Bacillus (~10^2 species).
- **Efficiency**: ~90% predictive accuracy.

2.3 Biological Applications

- **Classification**: Organizes ~10^7 species.
- **Conservation**: Identifies taxa (~10^5 endangered).
- **Therapeutics**: Pathogen taxonomy (~80% efficacy).
- **Biotechnology**: Museum-based biodiversity studies.