

Contents

Preface

vii

PART 1: MENDELIAN GENETICS

1. Introduction to Genetics

3

1.1 Biological Essence of Species

1.2 Two main Tendencies of a Species

1.3 Resemblance and differences among Relatives

1.4 Mechanisms Involved for Genetic differences among Relatives

1.5 Science of Genetics

1.6 Levels of Genetics

1. First Level of Genetics - At Individual Level

2. Second Level of Genetics - At Population Level

1.7 Physical and Chemical Basis of Inheritance

1.8 Functions of Genetic Material

1.9 Applications of Genetics

2. Variation in Populations

15

2.1 Types of Variation

2.2 Genetic Variation in Natural Populations

2.3 Sources of Genetic Variation

2.4 Chromosomal Variation	
2.5 Extra-chromosomal Variations	
Causes of Maternal Effects	
3. Heredity and Environment	33
3.1 Relative Role of Heredity and Environment	
3.2 Environmental Factors and Expression of Major Genes	
(I) External Environmental Factors	
(II) Internal Environmental Factors	
3.3 Phenotypic Stability (Canalization) and Environment	
3.4 Phenocopy: Environmentally Induced Phenotype	
3.5 Variability in Gene Expression	
3.5.1 Penetrance	
3.5.2 Expressivity	
4. Mendel's Laws of Heredity	47
4.1 Mendel's Experiment	
4.2 Monohybrid and Dihybrid Crosses and Mendel's Laws	
4.2.1 Monohybrid Crosses and Mendel's Laws	
4.2.2 Dihybrid Cross and Law of Independent Assortment of Genes	
4.3 Tests of Homozygous and Heterozygous Dominant	
4.4 Applications of Mendel's Laws of Heredity	
4.5 Reasons for Remaining Unnoticed the Mendel's Laws for 35 Years	
4.6 Basic Monohybrid Crosses	
Exercises	
5. Chromosome Theory of Heredity	65
5.1 Genes are Located on Chromosomes	
5.2 Evidence of Chromosome Theory of Heredity	
5.3 Mendel's Principles Based on Chromosome Theory	
6. Multiple Alleles	71
6.1 Causes of Modification in Mendel's Genetic Ratios	
6.2 Multiple Alleles	
6.3 Characteristics of Multiple Alleles	
6.4 Theory of Multiple Alleles	
6.5 Examples of Multiple Alleles	

6.6 Multiple Alleles in Humans	
6.7 Importance of Multiple Alleles	
Numerical Exercises	
7. Gene Action I. Allelic Interaction (Additive Gene Action)	87
7.1 Additive Gene Action (AGA) - Lack of Gene Interaction	
7.2 Non-Additive Gene Action (NAGA)	
7.3 Allelic Interaction	
Numerical Exercises	
8. Gene Action II. Non-Allelic Interaction (Epistasis)	101
8.1 Epistasis and its Types	
8.2 Simple Epistasis	
8.4 Duplicate Epistasis	
8.4 No Modification of Ratios by Epistasis (Complementary or Collaborative Gene Action)	
8.5 Modifying Genes	
8.6 Inheritance of Morphological Characters in Poultry	
8.7 Pleiotropic Gene Action	
Exercise	
9. Linkage and Crossing Over	121
9.1 Theories of Linkage	
9.2 Linkage	
9.2.1 Linkage Causes Deviation in Dihybrid F_2 Cross	
9.2.2 Linkage Causes Deviation in Dihybrid Test Cross	
9.2.3 Conclusion of the Results of F_2 Cross and Test Cross	
9.3 Recombination and Crossing Over	
9.4 Illustration of Crossing Over	
9.5 Cytological Basis of Crossing Over	
9.6 Meiotic Stage to Occur Crossing Over	
9.7 Expected Ratios with Independent Assortment and Linkage	
9.8 Linkage Detection	
9.9 Importance of Crossing Over and Linkage	
9.10 Effect of Linkage	
9.11 Chromosome Mapping	
9.12 Methods to Estimate the Gene Order	

9.13	<i>Interference and Coincidence</i>	
9.14	<i>Discrepancy between Recombinant Frequency and Genetic Map Distance</i>	
9.15	<i>Cytogenetic Mapping</i>	
9.16	<i>Correlation of Physical Map with Genetic and Cytological Map</i>	
9.17	<i>Uses of Chromosome Mapping</i>	
	<i>Numerical Examples</i>	
10.	Sex Determination	157
10.1	<i>Sexuality in Organisms</i>	
10.2	<i>Sex Determination Mechanism</i>	
10.3	<i>Chromosome Theory of Sex Determination</i>	
10.3.1	<i>XO Type Mechanism</i>	
10.3.2	<i>XY Type Mechanism</i>	
10.4	<i>Sex Chromosomes</i>	
10.5	<i>Gene Theory of Sex Determination</i>	
10.6	<i>Gynandrisim (Sex Mosaic)</i>	
10.7	<i>Hermaphroditism</i>	
10.8	<i>Environmental Sex Determination</i>	
10.9	<i>Hormones and Sexual Expression (Sex Reversion)</i>	
11.	Sex Controlled Inheritance	171
11.1	<i>Sex Influenced Characters</i>	
11.2	<i>Sex Limited Inheritance</i>	
11.3	<i>Similarities and Differences in Sex Influenced and Sex Limited Characters</i>	
11.4	<i>Sex-Linked Characters</i>	
11.4.1	<i>Inheritance Pattern of Sex-Linked Characters</i>	
11.4.2	<i>Complete vs Incomplete Sex-linked Genes</i>	
11.4.3	<i>Ways of Expression of a Sex Linked Recessive Gene (character)</i>	
11.4.4	<i>Sex-linked Inheritance in different Species</i>	
11.5	<i>Contribution of Sex Linkage</i>	
11.6	<i>Use of Knowledge of Sex Linkage</i>	
11.7	<i>Y-Chromosome heredity</i>	
11.8	<i>Pseudo-autosomal Genes</i>	
	<i>Numerical Exercises</i>	

PART 2: MOLECULAR GENETICS

12. Microbial Genetics: Bacteria and Viruses	191
12.1 Place of Bacteria in Plant Kingdom	
12.2 Importance of Bacteria	
12.3 Importance of Bacteria in Genetic Studies	
12.4 Advantages of using Bacteria in Genetic Studies	
12.5 Disadvantages of using Bacteria in Genetic Studies	
12.6 Bacterial Cell: Prokaryotic Cell	
12.7 Bacterial Genetic: Genetic Material of Bacteria	
12.8 E.coli – most Intensively Studied Bacterium	
12.9 Mutation in Bacteria	
12.10 Genetic Exchange Mechanism in Bacteria	
12.10.1 Transformation – Uptake of Free DNA by Bacteria	
12.10.2 Conjugation – Contact of Donor Cell to a Recipient Cell	
12.10.3 Transduction	
12.11 Distinction among Three Para-Sexual Processes	
12.12 Enzymes of Bacteria	
12.13 Viruses	
12.14 Viral Chromosome	
12.15 Bacteriophages	
13. Structure of Genetic Material: Nucleic Acids (DNA and RNA)	215
13.1 Evidence of Genetic Material (Nucleic Acids)	
13.1.1 Indirect Evidences from Comparisons	
13.1.2 Direct Evidences from Experiments	
13.2 Molecular Structure of Genetic Material: The Nucleic Acids	
13.3 Deoxyribonucleic Acid (DNA)	
13.4 RNA Structure and its Types	
13.5 Differences and Similarities between DNA and RNA	
13.6 Gene Concept	
14. Mutation: General Aspects	255
14.1 Definition and Concept of Mutation	
14.2 Types of Mutations	
14.3 Phenotypic Effects of Mutations	
14.4 Mutation Rate	
14.5 Detection of Mutation	

14.6 Metabolic Pathway of Mutation	
14.7 Molecular Basis of Mutation	
14.8 Mutation in Human β -globin Chain	
14.9 Significance of Mutation	
Exercises - Solved	
15. Induction of Mutation	281
15.1 Radiation Induced Mutations	
15.2 Chemically Induced Mutations	
15.3 Transposable Genetic Elements Inducing Mutations	
15.4 Repeated Trinucleotide Induced Mutations	
15.5 Cancer: Disease of Cell Division	
15.6 DNA Repair Mechanisms	
15.7 Defects in DNA Repair: Inherited Diseases (Human)	
16. DNA Replication	297
16.1 Importance and Basis of DNA Replication	
16.2 Main Features (Nature) of DNA Replication	
16.3 Enzymes Involved in DNA Replication	
16.4 Similarities and Differences in DNA Replication of Prokaryotes and Eukaryotic Chromosomes	
16.5 Watson and Crick (1953) Model of Replication	
16.6 Replication of Eukaryotic Chromosome: Special Aspects	
17. Gene Expression – Molecular Mechanism (Protein Synthesis)	
I. Transcription (RNA Production)	317
17.1 Gene Expression (Protein Synthesis): A Two stage Phenomenon	
17.2 The Central DOGMA: Transfer of Genetic Information	
17.3 Transcription (mRNA Synthesis)	
17.4 Transcription in Prokaryotes	
17.4.1 RNA Polymerase (Prokaryotes)	
17.4.2 Stages of Transcription (Prokaryotes)	
17.5 Transcription in Eukaryotes	
17.5.1 RNA Polymerase (Eukaryotes)	
17.5.2 Stages of Transcription (Eukaryotes)	
17.6 Primary Transcripts and mRNAs Structures	
17.7 RNA Editing and Alteration of Information	
17.8 Transcription of Introns in Eukaryotes	

18. Genetic Code	335
18.1 Base and Importance of Genetic Code	
18.2 Essential Features of Genetic Code	
18.3 Triplet Codes of Nitrogen Bases for 20 Amino Acids	
18.4 Triplet Codons of Amino Acids	
18.5 Codon – Anticodon Interaction	
Exercises - Solved	
19. Gene Expression – Molecular Mechanism (Protein Synthesis)	
II. Translation (Polypeptide Production)	345
19.1 Relation between Genes and their Polypeptide Products	
19.2 Components Required for Protein Synthesis	
19.3 Stages of Translation (Synthesis of Polypeptides)	
19.4 Common Features of Synthesis of DNA, RNA, and Polypeptides	
19.5 Simultaneous Occurrence of Transcription, Translation and mRNA Degradation	
19.6 Overview of Protein Synthesis	
20. Gene Expression – Regulation I. Prokaryotes	351
20.1 Mechanism of Control of Gene Expression	
20.2 Regulatory Mechanism of Transcription in Prokaryotes	
20.3 Constitutive, Inducible and Repressible Gene Expression	
20.4 Positive and Negative Control of Gene Expression in Prokaryotes	
20.5 Operon Models of Gene Expression in Prokaryotes	
20.6 Two Operon in <i>E. coli</i>	
20.6.1 Lactose (Lac) Operon in <i>E. coli</i>	
20.6.2 Tryptophan (trp) Operon in <i>E. coli</i>	
20.7 Relation of Gene Expression at Transcription Level	
20.8 Posttranscriptional Regulatory Mechanisms in Prokaryotes	
21. Gene Expression – Regulation II. Eukaryotes	367
21.1 Occurrence of Phenomenon in Regulation of Gene Expression	
21.2 Structural Complexities in Control of Transcription in Eukaryotes	
21.3 Regulation of Eukaryotic Gene Expression in Relation to Chromatin Organization	
21.3.1 Density of Chromatin Region (Euchromatin and Heterochromatin)	
21.3.2 Sensitivity of Chromatin to Nuclease Enzyme	
21.3.3 Nucleosome Structure – Chromatin Remodeling	

21.3.4 DNA Methylation	
21.3.5 Imprinting – Sex of Parent Transmitting the Gene	
21.4 Regulation of Eukaryotic Gene Expression in Relation to Sex Chromosomes: Dosage Compensation Mechanisms	
21.4.1 Inactivation of X-chromosome in Mammals	
21.4.2 Hyperactivation of X-chromosome in <i>Drosophila</i>	
21.4.3 Hypoactivation of X-chromosome in Nematode	
21.5 Control of Eukaryotic Gene Action in Relation to RNA Related Factors	
21.5.1 RNA Processing: Modification in RNA	
21.5.2 Differential Splicing Pattern of RNA – Removal of Introns	
21.5.3 Differential Stability of mRNA	
21.6 Gene Regulation in Relation to Environmental Factors	
21.6.1 Temperature Effect: Heat Induced Transcription	
21.6.2 Hormones Induced Signals	
21.7 Regulation of Eukaryotic Genes at Transcription Level	
21.7.1 DNA Sequences Involved in Transcription	
21.7.2 Transcription Factors Involved in Transcription	
21.8 Regulation of Eukaryotic Genes at Postranscription Level	
22. Gene Expression – Biochemical Pathway (Biochemical Genetics)	387
22.1 Biochemical Pathway of Gene Action	
22.2 Human Biochemical Genetics	
22.3 Biochemical Genetics of other Animals	
23. Techniques of Molecular Genetics – Recombinant DNA Technology	397
23.1 Various Technologies	
23.2 Biotechnology: Recombinant DNA Technology (Genetic Engineering)	
23.3 Biotechnology in India	
23.4 Molecular Analysis	
23.5 Important Discoveries and Techniques of Molecular Genetics	
23.6 Landmarks in Molecular Techniques	
23.7 Progress of DNA Sequencing	

23.8 Genetic Engineering: Recombinant DNA Technology	
23.8.1 Production of Recombinant DNA Products	
23.8.2 Procedure of Gene Cloning: Amplification of Recombinant DNA	
23.8.3 Somatic Cell Hybridization or Protoplast Fusion	
23.9 Southern Blot Hybridization: Molecular Analysis of DNA	
23.10 Northern Blot Hybridization: Molecular Analysis of RNA	
23.11 Western Blot Hybridization: Molecular Analysis of Proteins	
23.12 Molecular Analysis (Mapping) of Genes and Chromosomes	
23.12.1 Restriction Enzyme Cleavage Sites – As the Physical Map	
23.12.2 Nucleotide-Pair Sequence – As the Physical Map	
24. Applications of Molecular Genetics – Genome Manipulation	423
24.1 Areas of Animal Improvement	
24.2 DNA Profiling/Typing or Genotyping	
24.3 Transgenesis – Gene Transfer Technology	
24.4 Animal Cloning - Nuclear Transfer Technique or Reproductive Cloning	
24.4.1 Types of Animal Cloning	
24.4.2 Production of Cloned Calves/Kids in India	
24.4.3 Applications of Livestock Cloning	
24.5 Production to Eukaryotic Proteins in Bacteria: Nutrition and Health	
24.6 Methods of Genetic Dissection of Biological Processes	
24.6.1 Classical Genetic Approach	
24.6.2 Reverse Genetic Approaches	
24.7 Reproductive Biotechniques	
24.8 Cell Culture Products	
24.9 Genome Maps	
25. Markers and Genome Maps	439
25.1 Genome Projects	
25.1.1 Genome Mapping	
25.1.2 Sequencing	
25.1.3 Functional Analysis	
25.2 Types of Markers	
25.3 Genetic Markers	

- 25.4 Types of Molecular Markers
 - 25.4.1 Defined and Anonymous Markers
 - 25.4.2 Other Classifications of Molecular Markers
- 25.5 Selection of Ideal Marker
- 25.6 Applications of Molecular Markers
- 25.7 Types of Chromosome Maps
 - 25.7.1 Genetic Maps
 - 25.7.2 Physical Maps
 - 25.7.3 Cytological Maps
- 25.8 Map Position-Based Cloning of Genes
- 25.9 Chromosome Walking and Jumping
 - 25.9.1 Chromosome Walking
 - 25.9.2 Chromosome Jumping
- 25.10 FISH: Fluorescence in situ Hybridization
- 25.11 Microsatellite (STR) Mapping
 - 25.11.1 Somatic Cell Hybridization
 - 25.11.2 Individual Chromosome Preparation

PART 3: GENETIC STATISTICS

26. Probability Rules

461

- 26.1 Events and their Probabilities
- 26.2 Types of Events
- 26.3 Probability Defined
- 26.4 Characteristics of Probability
- 26.5 Rules of Calculating Probability
- 26.6 Laws of Probability
 - 26.6.1 Additive Theorem of Probability
 - 26.6.2 Multiplication Theorem of Probability
- 26.7 Discrete Probability Distribution
- 26.8 Binomial Distribution
- 26.9 Characteristics of Binomial Theorem
- 26.10 Mean (Expectation) and Variance of Binomial Distribution
- 26.11 Effect of n , p , and r on Probability
- 26.12 Multinomial Distribution
- 26.13 Use of Probability

26.14 Applications of Binomial Theorem in Genetics

Solved Exercises - Probability Rules

27. Chi-Square (χ^2) Test

493

27.1 Hypothesis and Level of Significance

27.2 Test of Measurement - Chi Square (χ^2) Test27.3 Use of χ^2 Test

27.4 Comparison with Expectation

27.4.1 Distribution following Binomial Theorem

27.4.2 Testing Genetic Ratios

27.4.3 Testing Homogeneity among Classes - or Experimental Conditions

27.4.4 Detection of Linkage: Two Factor Segregation

27.5 Testing the Independence (Association or Correlation)

27.6 Correct Use or Limitations of Chi-square (χ^2)**ANNEXURES**

A. 14.1 Mechanisms Involved in Transition Mutation	511
A. 14.2 Mechanism of Transposition	519
A. 15.1 Causes of Cancer: Genetic Disease of Cell Division	528
A. 19.1 Amino Acids: Structure and Polymerization	536
A. 19.2 Proteins Structure: Three-Dimensional Structure of Complex Molecule	540
A. 19.3 One Gene-One Polypeptide Concepts	543
A. 19.4 Roles of different Components in Protein Synthesis	545
A. 23.1 Biological Tools Used in Recombinant DNA Technology	550
A. 23.2 Restriction Endonuclease	554
A. 23.3 Plasmid, Phage or Viral Chromosome	559
A. 23.4 Sources and Procedures to obtain Isolated Gene	562
A. 23.5 Techniques of DNA Transfer (Integration)	570
A. 23.6 <i>In situ</i> Colony or Plaque Hybridization	573
A. 23.7 Amplification of DNA Sequence or Desired Gene <i>In vitro</i> : Polymerase Chain Reaction	576
A. 23.8 PROBE: Suitably Labelled Nucleic Acid Molecule of 15-30 Bases Long	581
A. 23.9 Autoradiography	585

A. 23.10 Electrophoresis	586
A. 23.11 Somatic Cell Hybridization	587
A. 23.12 Enzyme Linked Immune- Sorbent Assay (ELISA)	588
A. 23.13 Nick Translation	590
A. 23.14 Dot Blot Technique	591
Subject Index	593