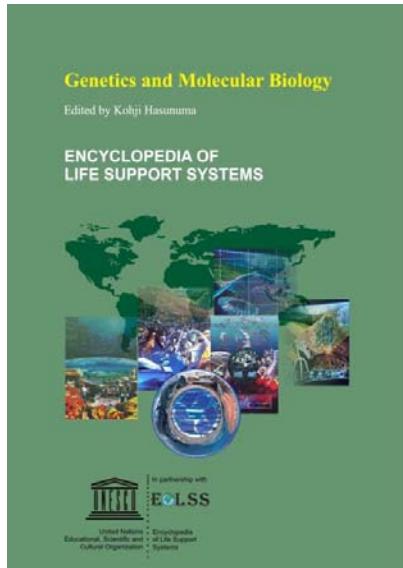


CONTENTS

GENETICS AND MOLECULAR BIOLOGY



Genetics and Molecular Biology - Volume 1

No. of Pages: 526

ISBN: 978-1-84826-123-5 (eBook)

ISBN: 978-1-84826-573-8 (Print Volume)

For more information of e-book and Print Volume(s) order, please [click here](#)

Or contact : eolssunesco@gmail.com

CONTENTS

- | | |
|---------------------------------------------------------------------------------------------------------------------------------------------------|----------|
| Genetics and Molecular Biology | 1 |
| <i>Kohji Hasunuma, Yokohama City University, Kihara Institute for Biological Research, Graduate School of Integrated Science, Yokohama, Japan</i> | |
-
1. Introduction
 2. Signal Transduction via Plasma Membrane to Gene Expression
 - 2.1. Signal Transduction via Hormone Receptors in Plasma Membrane
 - 2.2. Growth Factor Receptors and MAP Kinase Cascades
 - 2.3. Signal Transduction Driven by ROIs and Oxidative Stress
 - 2.3.1. The Production and Turning-Over of ROIs
 - 2.3.2. Signal Transduction Controlled by ROIs and Oxidative Stress
 - 2.3.3. Regulation of the MAP Kinase Cascade by Src Tyrosine Kinase through ROIs and NO (Nitric Oxide)
 - 2.3.4. The Action of H₂O₂ in Controlling MAP Kinase Cascade
 - 2.3.5. The Function of NO in Controlling the MAP Kinase Cascade
 - 2.4. Transcription Regulation by Reactive Oxygen Intermediates and NO
 - 2.4.1. Redox Regulation of Transcription Factor NF-κB
 - 2.4.2. AP-1 and Redox Control
 - 2.4.3. The Activation of Src Tyrosine Kinase
 - 2.5. Signal Transduction via JAK and STAT
 3. Signal Transduction of Light in Animals (Vertebrates), *Neurospora crassa*, and Plants
 - 3.1. Light Signal Transduction in Animals
 - 3.2. Light Induced Responses in *Neurospora crassa*
 - 3.3. Circadian Rhythm
 - 3.4. Molecular Analysis of Light Signal Transduction
 - 3.5. Light Signal Transduction in Plants
 4. Epigenetic Regulation of Gene Expression in Eukaryotes
 - 4.1. RNA Interference (RNAi)
 - 4.2. Repeat Induced Point Mutation (RIP) in *Neurospora crassa*
 - 4.3. Gene Regulation by Polycomb Group (*PcG*) Genes and *trithorax* Group (*trxG*) Genes
 - 4.4. Gene Regulation Through the Methylation of DNA in *Arabidopsis thaliana*
 - 4.5. Developmental Regulation by DNA Methylation in Mammals
 5. Conclusion
-
- | | |
|-------------------------------------------------------------------------------------------------------------------------------------------------------------|-----------|
| History and Scope of Genetics; Epigenetic Regulation of Gene Expression | 41 |
| <i>Kohji Hasunuma, Yokohama City University, Kihara Institute for Biological Research, Graduate School of Integrated Science, Yokohama, 244-0813, Japan</i> | |
-
1. Introduction
 2. RNA interference
 - 2.1. Process of RNA interference (RNAi)
 - 2.2. The genes regulating the process of RNAi
 - 2.3. Putative function of small dsRNA in the process of RNA interference
 - 2.4. RNase activities from dsRNA to siRNA, Dicer
 3. Repeat induced point mutation (RIP)
 - 3.1. RIP in *Neurospora crassa*
 - 3.2. Methylation-induced premeiotically (MIP) in *Ascobolus immersus*
 - 3.3. "Quelling" triggered by introduction of DNA in *Neurospora crassa*
 4. Genetic regulation by Polycomb group (*PcG*) proteins
 - 4.1. The determination of cell fate and the mechanism of the maintenance of cell memory
 - 4.2. *PcG* gene groups
 - 4.3. Gene silencing by *PcG* proteins antagonized by the molecules controlling locus control region (LCR)
 - 4.4. Trithorax Group (*TrxG*) genes
 5. Epigenetics in *Arabidopsis thaliana*

- 5.1. DNA methyltransferase
- 5.2. Decrease in DNA methylation, *DDM1*
- 5.3. Regulation of homeotic gene expression by a P_cG gene in *Arabidopsis thaliana*
- 5.4. Maintenance of genomic imprinting at the medea (mea) locus is dependent on zygotic *DDM1* activity
- 6. Paramutation

Mendelian Genetics and its Development**56**

Hikoyuki Yamaguchi, Faculty of Agriculture, University of Tokyo, Japan

- 1. The Classical Theory of Direct Inheritance
 - 1.1. The Views of Hippocrates and Aristotle
 - 1.2. Darwin's Theory
 - 1.3. Experiments by Knight and by Goss
- 2. Mendelian Genetics
 - 2.1. Mendel's Law of Inheritance
 - 2.2. The Diversity of Mendelian Traits
 - 2.3. The Chromosome Theory of Inheritance
 - 2.4. Genetic Recombination and Mapping
 - 2.5. Physical Evidence for Recombination
- 3. Molecular Genetics
 - 3.1. The Discovery of DNA
 - 3.2. What Genes Are Made Of
 - 3.3. The Relationship between Genes and Proteins
 - 3.4. What Genes Do
 - 3.4.1. How Genes Replicate
 - 3.4.2. How Genes Direct the Production of Proteins
 - 3.4.3. How Genes Accumulate Mutations
 - 3.5. Gene Cloning and Manipulation
- 4. Population Genetics
 - 4.1. Types of Genetic Variation
 - 4.2. Measuring Genetic Variation
 - 4.3. The Hardy-Weinberg Principle
 - 4.4. Molecular Population Genetics
 - 4.4.1. Neutral Theory and Molecular Evolution
 - 4.4.2. The Molecular Clock
 - 4.4.3. Molecular Phylogenetics
 - 4.4.4. Mitochondrial and Chloroplast DNA Evolution
 - 4.4.5. Multigene Families

Classical to Modern Genetics**72**

Kohji Hasunuma, Yokohama City University, Kihara Institute for Biological Research, Graduate School

of Integrated Science, Yokohama, Japan

Naoto Yabe, Yokohama City University, Kihara Institute for Biological Research, Graduate School of
Integrated Science, Yokohama, Japan

- 1. Introduction
- 2. *Bacillus subtilis*
 - 2.1. Life cycle of *Bacillus subtilis*
- 3. *Agrobacterium tumefaciens*
- 4. *Neurospora crassa*
 - 4.1. Life cycle
 - 4.2. Effect of environmental cues
 - 4.3. Genetical information
 - 4.4. Transformation by DNA
 - 4.5. Gene silencing

- 4.6. Gene silencing by mutation; repeat induced point mutation (RIP)
- 4.7. Gene silencing by DNA methylation
- 4.8. Repeat induced gene silencing in vegetative cells; quelling
- 4.9. Gene regulation by meiotic pairing; transvection
5. Aspergillus nidulans
6. Lotus corniculatus L. var. japonicus Regel
 - 6.1. Nitrogen fixation equipped by *Rhizobium-Legume* symbiosis
 - 6.2. Rhizobia
 - 6.3. Legumes
 - 6.4. Development of nodules

Bacterial and Yeast Genetics - A Historical Account**84**

Toshio Fukasawa, Department of Microbiology, School of Medicine Keio University, Tokyo, Japan

1. Bacterial genetics
 - 1.1. Introduction - Bacterial Genetics
 - 1.2. *Escherichia coli* and its related bacterial species
 - 1.2.1. Cell structure, growth, and mutations
 - 1.2.2. Conjugation (Transfer of genetic information by cell-to-cell contact)
 - 1.2.3. Transduction (Transfer of genetic information mediated by phage)
 - 1.2.4. Transformation (Transfer of genetic information by free DNA)
 - 1.2.5. Gene expression and its regulation
 - 1.2.6. Bacteriophages
 - 1.2.7. Plasmids and transposable elements
 - 1.2.8. Cell division and the chromosome replication
 - 1.2.9. Birth of molecular genetics
 - 1.2.10. Recombinant DNA technology
 - 1.2.11. Genomics
 - 1.3. Other bacterial species used in bacterial genetics
2. Yeast genetics
 - 2.1. Introduction – Yeast genetics
 - 2.2. *Saccharomyces cerevisiae*
 - 2.2.1. Cell structure, cell division, and life cycle
 - 2.2.2. Classic genetic analysis
 - 2.2.3. Recombinant DNA technology and gene manipulation
 - 2.2.4. Extra-nuclear genetic elements
 - 2.2.5. Gene expression and its regulation.
 - 2.2.6. Genomics
 - 2.3. *Schizosaccharomyces pombe*
 - 2.3.1. Cell structure, growth, and life cycle
 - 2.3.2. Classic genetic analysis
 - 2.3.3. Recombinant DNA technology
 - 2.3.4. Genomics
 - 2.4. *Candida albicans*
 - 2.4.1. Cell morphology, growth, and life cycle
 - 2.4.2. Classical genetics
 - 2.4.3. Recombinant DNA technology
 - 2.4.4. Genomics

Diploid and Haploid Genetics and Recombination Mechanisms**184**

Yasuo Hotta, Department of Health and Nutrition, Niigata University of Health and Welfare, Niigata-shi, Japan

1. General Recombination
2. Recombination and Pairing of Homologous Chromosomes
3. Induction of Recombination

4. Establishment of Recombination
5. Structure and Function of the Synaptonema Complex (SC):
6. Transcription and Recombination
7. Somatic recombination/ Diploid Genetics
8. Genes and Proteins in Meiotic Cells / Haploid Cell Genetics
9. Meiosis and Environment
10. Conclusion

Non-Mendelian Inheritance**203**

Yoshiki Nishimura, *University of Tokyo, Japan*
 Tsuneyoshi Kuroiwa, *University of Tokyo, Japan*

1. Chloroplast Inheritance
 - 1.1. Early Cytological Observations of Chloroplast Inheritance
 - 1.2. Chloroplast Inheritance in Isogamous Organisms
 - 1.2.1. Unicellular Green Algae: *Chlamydomonas reinhardtii*
 - 1.2.2. Discovery of Chloroplast DNA
 - 1.2.3. Molecular Mechanism of Non-Mendelian Inheritance of cpDNA
 - 1.2.4. Transformation of cpDNA
 - 1.2.5. Active Digestion of mt- cpDNA Revealed in a Single Zygote by Using the Optical Tweezer in *C.reinhardtii*
 - 1.2.6. Chloroplast Inheritance in Other Isogamous Algae
 - 1.3. Chloroplast Inheritance in Anisogamous and Oogamous Organisms
 - 1.3.1. Anisogamous and Oogamous Algae, and Ferns
 - 1.3.2. Malarial Parasites: *Plasmodium falciparum*
 - 1.4. Chloroplast Inheritance in Higher Plants
 - 1.4.1. Angiosperms
 - 1.4.2. Gymnosperms
2. Mitochondrial Inheritance
 - 2.1. Early Genetic Analysis of Mitochondrial Inheritance
 - 2.2. Discovery of Mitochondrial DNA
 - 2.3. Mitochondrial Inheritance in Isogamous Organisms
 - 2.3.1. Yeast-*Saccharomyces cerevisiae*
 - 2.3.2. Unicellular Green Algae: *Chlamydomonas reinhardtii*
 - 2.3.3. True Slime Mold: *Physarum polycephalum*
 - 2.4. Mitochondrial Inheritance in Oogamous Organisms
 - 2.4.1. Mammals
 - 2.4.2. Humans
 - 2.4.3. Higher Plants

Genome Analysis of Cyanobacteria**220**

Satoshi Tabata, *Kazusa DNA Research Institute, Chiba, Japan*

1. Introduction
2. Sequence Features of the Synechocystis sp. PCC6803 Genome
3. Assignment of RNA and Protein-Coding Genes in the Synechocystis Genome
 - 3.1. Potential Structural RNA Genes in the Genome
 - 3.2. Potential Protein-Coding Genes in the Genome
4. Characteristic Features of Synechocystis Genes
 - 4.1. Genes for Photosynthesis
 - 4.2. Relationship to Plant Plastids
 - 4.3. Genes for Two-Component Signal Transduction Systems
5. Functional Genomics in Synechocystis
 - 5.1. Systematic Gene Disruption
 - 5.2. Transcriptome Analysis
 - 5.3. Proteome Analysis

6. Databases Supporting Synechocystis Research
7. Genome Analysis of Other Cyanobacteria

Genomic Analysis of *Arabidopsis thaliana***231**Takayuki Kohchi,*Nara Institute of Science and Technology, Japan*

1. Introduction: Why is *Arabidopsis* a Model Plant Species?
2. Small Size and Simple Organization of the *Arabidopsis* Genome
3. Chromosomes and Maps
4. Analysis of the Complete Genome Sequence
5. Chromosomal Elements: Centromeres, Telomeres, and rDNA Repeats
6. *Arabidopsis* Plastid and Mitochondrial Genomes
7. Functional Genomics of *Arabidopsis*
8. Perspectives

Genome Science of Invertebrates: The Nematode *C.elegans***242**Yuji Kohara,*Center for Genetic Resource Information, National Institute of Genetics, Yata 1111, Mishima, Shizuoka 411-8540, Japan.*

1. What is a nematode?
2. What is *C.elegans* ?
3. Genetic resources of *C.elegans*: CGC
4. Genomic resources
5. Resources for gene expression and function
6. Future directions

Genome Science of the Rat**249**Takashi Kuramoto,*National Cancer Center Research Institute, Tokyo, Japan*
Toshikazu Ushijima,*National Cancer Center Research Institute, Tokyo, Japan*

1. Introduction
2. Genome Resources
 - 2.1. Genetic Markers
 - 2.1.1. Microsatellite Markers
 - 2.1.2. RDA-Related Markers
 - 2.2. Large-Insert Library
 - 2.3. Radiation Hybrid Panel and Radiation Hybrid Map
 - 2.4. Comparative Map
 - 2.5. Rat EST Project
 - 2.6. Rat Genome Sequencing Project
 - 2.7. DNA Array
 - 2.8. Databases
3. Genetic Engineering
4. Positional Cloning of Monogenic Traits
5. QTL Analysis
 - 5.1. Hypertension
 - 5.2. Diabetes
 - 5.3. Cancer

Genome Science of Vertebrate**264**Naoki Osato, *RIKEN Yokohama Institute, Japan*
Hidenori Kiyosawa, *RIKEN Yokohama Institute, Japan*
Jun Kawai, *RIKEN Yokohama Institute, Japan*
Yoshihide Hayashizaki, *RIKEN Yokohama Institute, Japan*

1. The Importance of the Mouse Genome and Transcriptome Study
2. The History of Mouse Genetics
 - 2.1. The Beginning of Mouse Genetics and the Establishment of Inbred Strains
3. The Determination of Mouse Genome Sequences
4. A Full-Length Mouse cDNA Project
 - 4.1. From the EST Project to the Full-Length cDNA Project
 - 4.2. Findings from Genome and cDNA Sequences
 - 4.3. Functional Annotation of Mouse cDNA Sequences
5. Positional Cloning
6. cDNA Encyclopedia and the Positional Candidate Approach
7. Functional Genomics
 - 7.1. DNA Microarray
 - 7.2. PPI and PDI (Protein–Protein Interaction, Protein–DNA Interaction)
 - 7.3. Structural Genomics

DNA Repair

279

Takahiko Taguchi,*Tokyo Metropolitan Institute of Gerontology, Japan*

1. DNA Damage
 - 1.1. Spontaneous Alterations of DNA (by Mutator Genes)
 - 1.1.1. DNA Polymerase
 - 1.1.2. Proofreading Activity
 - 1.2. Environmental Damage to DNA
2. DNA Repair by Reversal of Damage Without Excision
 - 2.1. Photoreactivation
 - 2.2. Repair of O6-Alkylguanine and Alkylthymine Without DNA Strand Excision
3. Base Excision Repair in Non-Mammalian Cells
 - 3.1. DNA Glycosylase in Non-Mammalian Cells
4. Base Excision Repair in Mammalian Cells
 - 4.1. DNA Glycosylases in Mammalian Cells
 - 4.2. AP Endonuclease
 - 4.3. DNA Deoxyribophosphodiesterase, DNA Polymerase, and DNA Ligase
5. Nucleotide Excision Repair in Non-Mammalian Cells
 - 5.1. Nucleotide Excision Repair by UvrABC
 - 5.2. Transcription Coupling Nucleotide Excision Repair in *E.coli*
6. Nucleotide Excision Repair in Mammalian Cells
 - 6.1. Nucleotide Excision Repair by Endonucleases and Exonucleases
 - 6.2. Transcription Coupling Nucleotide Excision Repair in Mammalian Cells
 - 6.3. Gap Filling by DNA Polymerases
7. Mismatch Repair
 - 7.1. Long-Patch Mismatch Repair in Non-Mammalian Cells
 - 7.2. Short-Patch Mismatch Repair in Non-Mammalian Cells
 - 7.3. Long-Patch Mismatch Repair in Mammalian Cells
 - 7.4. Short-Patch Mismatch Repair in Mammalian Cells
8. Repair Enzymes in Mitochondria
9. SOS Responses
10. Recombination Repair
11. Human Hereditary Diseases with Defective Processing of DNA Damage

Recombination - Transformation, Transduction and Conjugation

302

Yasuo Hotta,*Niigata University of Health and Welfare, Japan*

1. Introduction
2. Recombination
3. Involvement of RNA in DNA Recombination
4. Recombination of RNA

5. Inverted Repeats
6. Conjugation
7. Transduction
8. Transformation

Reverse Transcriptase and cDNA SynthesisKunitada Shimotohno,*Kyoto University, Japan***316**

1. Introduction
2. Discovery of Reverse Transcriptase
3. Characteristic Features of Retroviral Reverse Transcriptases
 - 3.1. Template and Primer Requirements for Reverse Transcriptase
 - 3.2. Substrates for the Enzyme
 - 3.3. Other Factors Required for Reverse Transcription
 - 3.4. Natural Primers for cDNA Synthesis of Retrovirus Genome
 - 3.5. Inhibitors of Reverse Transcriptases
4. Reverse Transcriptase Activity in Other Organisms
 - 4.1. Reverse Transcriptase Activity in Retroposons
 - 4.2. Reverse Transcriptase Activity in Pararetroviruses
 - 4.2.1. Reverse Transcriptase Activity in Hepatitis B Virus (HBV)
 - 4.2.2. Reverse Transcriptase Activity in Cauliflower Mosaic Virus (CaMV)
 - 4.3. Reverse Transcriptase Activity and Telomerase
 - 4.4. Reverse Transcriptase Activity in Thermus Thermophilus DNA Polymerase
5. Conserved Amino Acid Residues in Putative Reverse Transcriptase
6. Structure of Retroviral Reverse Transcriptases
7. cDNA Synthesis by Reverse Transcription

Horizontal and Vertical Gene TransferMichio Imai,*Jichi Medical School, Saitama, Japan***329**

1. Gene Transfer
2. Horizontal Gene Transfer
3. Vertical Gene Transfer

Patterns of Heredity and Genetic Alteration; Epigenetics of MammalsKohji Hasunuma,*Yokohama City University, Japan***331**

1. Introduction
2. DNA Methyltransferase in Mammals
 - 2.1. Dnmt1
 - 2.2. Dnmt3a and Dnmt3b
3. DNA Methyltransferase Defective Mice
4. Genomic Imprinting and DNA Methylation
5. X-Chromosome Inactivation and DNA Methylation
6. Methyl-CpG-Binding Proteins
 - 6.1. MeCP2
 - 6.2. MBD1
 - 6.3. MBD2
 - 6.4. MBD3
 - 6.5. MBD4
7. Chromatin Formation
8. Remodeling of Nucleosomes
9. Histone Acetylation and Nucleosome Remodeling
10. Histone Deacetylation
11. SIN3 Complex
12. Mi2-NuRD Complex

13. Transition Regulation Under the Control of Chromatin Remodeling Factors

Gene Action in InheritanceSatoru Kobayashi,*Okazaki National Research Institute, Japan***350**

1. Germline Development
2. Polar Granules, the Distinctive Organelles of Germ Plasm
3. Maternal Genes Required for Germ Plasm Assembly
4. Maternal Factors Required for Pole Cell Formation
 - 4.1. mtlrRNA
 - 4.2. Transport of mtrRNAs from Mitochondria to Polar Granules
 - 4.3. Role of mtrRNAs in Pole Cell Formation
 - 4.4. Germ Cell-less mRNA
5. Maternal Factors Required for Pole Cell Differentiation
 - 5.1. Nanos Protein Required for Pole Cell Migration
 - 5.2. Role of Nos Protein in Gene Regulation in Pole Cells
 - 5.3. Pgc RNA
6. Germ Plasm Components in Other Animals

Genotypic and Phenotypic VariationsHiroaki Kagawa,*Okayama University, Japan.***361**

1. Brief Introduction to Historical Background
2. Brief Introduction to the Genetics of the Nematode *Caenorhabditis*
 - 2.1. Cell Lineage and Genome Sequence
 - 2.2. Genetics and Mutants
3. Uncoordinated Mutants of *C. elegans*
 - 3.1. General Introduction to Uncoordinated Mutants
 - 3.2. Structural Protein of Muscle
 - 3.2.1. Thick Filament (Myosin Heavy Chain, Paramyosin, and Twitchin)
 - 3.2.2. Thin Filament (Actin and Tropomyosin, Troponins)
 - 3.3. Transcription Factors
 - 3.4. Receptors and Channels
 - 3.5. Kinases and Enzymes
 - 3.6. Neuron-Related Proteins
4. Other Mutants
 - 4.1. Vulva Formation
 - 4.2. Sex Determination and Dauer Formation
 - 4.3. Chemical Response and Signal Transduction
 - 4.4. Cell Death and Life Span
5. Future Scope
 - 5.1. Gene Duplication and Genome Organization
 - 5.2. Interaction Among Different Genes
 - 5.3. Economic Estimate

Molecular Genetics of Inherited DisordersYasuyoshi Nishida,*Division of Biological Science, Graduate School of Science, Nagoya University, Japan.***379**

1. Introduction
2. Mutations
 - 2.1. Causal agents of mutations
 - 2.2. Types of mutations
3. Effects of mutations on gene function
 - 3.1. Gene structure and function
 - 3.2. Alleles may have different phenotypes

4. Molecular pathology of the human β -globin gene.
 - 4.1. The structure of the human β -globin gene cluster
 - 4.2. Structure of the β -globin gene and protein
 - 4.3. Mutations in the β -globin gene
 - 4.3.1. Sickle cell anemia and other hemoglobin variants
 - 4.3.2. β -Thalassemias
5. Patterns of inheritance
 - 5.1. Autosomal inheritance
 - 5.2. Sex-linked inheritance
 - 5.3. Some cases of unusual inheritance
 - 5.3.1. Mutations in genes subjected to genomic imprinting
 - 5.3.2. Tumor suppressor genes
 - 5.4. Epigenetic effects of mutations: position effect
6. Concluding remarks

Heredity and Environment; Light Signal Transduction in Plants and Fungi 390

Kohji Hasunuma, *Yokohama City University, Kihara Institute for Biological Research, Totsuka-ku, Yokohama, Japan*

Naoto Yabe, *Yokohama City University, Kihara Institute for Biological Research, Totsuka-ku, Yokohama, Japan*

1. Introduction
2. Historical aspects of analysis of respond to light
3. Plants; *Arabidopsis thaliana*
 - 3.1. Photoreceptors
 - 3.1.1. Phytochromes
 - 3.1.2. Cryptochromes
 - 3.1.3. Phototropin
 - 3.1.4. Putative photoreceptors for UV-A, UV-B and UV-C
 - 3.2. Signal transduction
4. Fungi; *Neurospora crassa*
 - 4.1. Photoreceptors
 - 4.2. Signal transduction
 - 4.3. Circadian rhythm

The Human Genome 406

Hikoyuki Yamaguchi, *University of Tokyo, Japan*

1. Introduction
2. Human Genome
3. DNA Variation
 - 3.1. Mutations
 - 3.2. Polymorphisms
4. Physical Maps of Human Chromosomes
 - 4.1. Human Chromosomes
 - 4.2. Human Repetitive Elements
 - 4.2.1. Tandem Arrays of Repeats
 - 4.2.2. DNA Fingerprint Analysis
 - 4.2.3. Telomeric and Subtelomeric Repeats
 - 4.3. Retroposons
 - 4.4. Endogenous Retroviruses
 - 4.5. Human Disease Gene Mapping
 - 4.5.1. Genetic Architecture of Human Diseases
5. Body Expression Map of the Human Genome
6. Applications of Gene Expression Analysis in Biomedical Research
 - 6.1. Pharmacogenomics

- 6.2. Genetic Therapy
- 7. Mitochondrial DNA
- 8. The Origin of Modern Humans
 - 8.1. mtDNA
 - 8.2. Y Chromosome Markers

Human Genetics**421**Ichiro Matsuda, *Ezuko Institution for Developmental Disabilities, Japan*

- 1. Human Genetics and Medical Genetics
- 2. Historical Aspects of Human Genetics
- 3. Genetic Diseases
 - 3.1. Chromosomal disorders
 - 3.2. Inherited Single-gene (Monogenic) Disorders
 - 3.3. Non-Mendelian Inherited Disease
 - 3.4. Multifactorial Inheritance or Polygenic Disorders
 - 3.5. Birth Defects
 - 3.6. Cancer Genetics
- 4. Genetic Services
 - 4.1. Genetic Counseling
 - 4.2. Genetic Testing and Screening
 - 4.3. Principle of Genetic Testing
 - 4.4. Prenatal Testing and Pre-Implantation Diagnosis
 - 4.5. Pre-Symptomatic Testing
 - 4.6. Susceptibility Testing (Cancer Susceptibility)
 - 4.7. Genetic Testing During Childhood
 - 4.8. Newborn Screening
- 5. Gene Therapy
 - 5.1. Practice of Gene Therapy
 - 5.2. Ethical Issues Related to Gene Therapy

Gene Expression and Embryogenesis in Amphibians**445**Horst Grunz, *Department of Zoophysiology, University Essen, Universitätstr. 5, 45117 Essen, Germany*

- 1. Introduction
- 2. Historical background
- 3. The search for inducing factors
 - 3.1. Isolation of the first embryonic inducing factor from chicken embryos – a milestone for our understanding of evolutionary conserved proteins
 - 3.1.1. The vegetalizing factor (a homologue of activin) – the first embryonic inducing factor available in highly purified form
 - 3.1.2. The mechanism of action of the vegetalizing factor
 - 3.1.3. Importance of different threshold concentrations for the determination of different cell types
 - 3.2. The search for neuralizing factors
- 4. The Responding Tissue
- 5. Autoneuralization and the Neural Default Hypothesis
- 6. Organizer and Antiorganizers
- 7. The importance of gradients for the anterior-posterior organization of the embryo
- 8. Germ layer determination – the three-signal (cascade)-model
- 9. Planar versus vertical signal during neural induction
- 10. Amphibians – a model system for organ engineering
- 11. Conclusions and Prospects in the 21 st Century

Index**463****About EOLSS****477**