

Table of Contents

Foreword (A. G. MOTULSKY)	V
Preface (F. VOGEL and K. SPERLING))	VII
1 Human Genetics in a Wider Perspective	1
Presidential Address. Human and Medical Genetics: Past, Present, and Future (A. G. MOTULSKY)	3
Our Mutation Load (N. P. BOCHKOV)	14
Cultural Evolution and Genetics (L. L. CAVALLI-SFORZA)	24
The Ecumenical Future of Human Genetics (J. V. NEEL)	34
Human Genetics and the Responsibility of the Medical Doctor (F. VOGEL)	44
2 Structure of the Human Genome	55
Human Genomics 1986: Toward a Complete Gene Map and Nucleotide Sequence of the Human Genome (V. A. MCKUSICK)	57
Progress Toward an Accurate Human Linkage Map (K. K. KIDD)	99
HLA, Immune Response, and Disease (W. F. BODMER)	107
Molecular Genetics: An Approach Towards the Basic Defect Causing Cystic Fibrosis (E. K. WATSON, B. J. WAINWRIGHT, H.-Y. LAW, X. ESTIVILL, H. KRUYER, and R. WILLIAMSON)	114
A Molecular Analysis of X-Linked Disease (K. E. DAVIES, T. J. SMITH, S. M. FORREST, S. J. KENWRICK, M. N. PATTERSON, L. WILSON, K. PAULSEN, H. R. DORKINS, I. LAVENIR, A. W. KING, A. SPEER, and C. COUTELLE)	120
Molecular Techniques in Mammalian Genetics: A New Era in Genetic Analysis (A. CRAIG, F. MICHIELS, G. ZEHETNER, B. SPROAT, M. BURMEISTER, M. BUĆAN, A. POUSTKA, T. POHL, A.-M. FRISCHAUF, and H. LEHRACH)	126
Human Sex Chromosomes: Molecular Analyses Have Not Yet Provided All the Answers (H. H. ROPERS)	133
Homologies Between the Sex Chromosomes of Man (P. J. GOODFELLOW, S. M. DARLING, B. PYM, C. PRITCHARD, and P. N. GOODFELLOW)	145

Molecular Diagnosis of X-Linked Disorders (P. L. PEARSON, G. J. B. VAN OMMEN, E. BAKKER, A. BRÖCKER-VRIENDS, N. CARPENTER, and H. VEENEMA)	152
Mitochondrial Clans and the Age of Our Common Mother (A. C. WILSON, M. STONEKING, R. L. CANN, E. M. PRAGER, S. D. FERRIS, L. A. WRISCHNIK, and R. G. HIGUCHI)	158
Functions of the Proteins Encoded in Human Mitochondrial DNA (G. ATTARDI, A. CHOMYN, and P. MARIOTTINI)	165
Evolutionary Implications of Mitochondrial DNA Polymorphism in Human Populations (S. HORAI, T. GOJOBORI, and E. MATSUNAGA) . . .	177
Mammalian Genes and Islands on Non-Methylated CpG-Rich DNA (A. BIRD, P. LAVIA, D. MACLEOD, S. LINDSAY, M. TAGGART, and W. BROWN)	182
Antibodies and T-Cell Receptors: Jumping Genes and Supergene Families and New Instruments for Studying Them (L. E. HOOD) . . .	187
Detection and Estimation of Linkage, Especially Multipoint Mapping (J. OTT, C. ASTON, M. BAUR, T. BISHOP, A. CHAKRAVARTI, J. CLAYTON, J. H. EDWARDS, R. C. ELSTON, B. KEATS, M. LATHROP, M. NEUGEBAUER, and L. PASCOE)	188
Molecular Human Genetics – Strategies of Gene Identification (H. LEHRACH)	190
X Chromosome Inactivation (S. M. GARTLER)	192
Workshop on Repetitive DNA in Mammalian Genomes (T. SHARMA, H. J. COOKE, J. T. EPPLIN, P. VOGT, H. NEITZEL, and T. IGO-KEMENES)	196
3 Cytogenetics and Development	199
Microdeletions and Mendelian Phenotypes (U. FRANCKE)	201
Autosomal Disorders: New Frontiers of the Phenotype (J. DE GROUCHY and C. TURLEAU)	211
Contribution of High-Resolution Banding and In Situ Hybridization to Clinical Cytogenetic Diagnosis (J. F. MATTEI, M. G. MATTEI, N. PHILIPS, and F. GIRAUD)	216
Phenotypic-Karyotypic Correlations of Gonadal Determinants: Current Status and Relationship to Molecular Studies (J. L. SIMPSON) .	224
Chromosome Abnormalities: Origin and Etiology in Abortions and Livebirths (P. A. JACOBS and T. J. HASSOLD)	233
Androgenesis and Parthenogenesis in Humans (K. OHAMA)	245
AB0 and HLA Systems: Effects of Maternal Incompatibilities on the Conceptus (S. D. LAWLER)	250
Meiosis and Male Infertility (A. C. CHANDLEY)	257
Aneuploidy in Mouse and Man (C. J. EPSTEIN)	260
Chromosome Sorting (M. A. FERGUSON-SMITH and C. CREMER)	269

Genetics and Molecular Biology of Lethal Genes in the Mouse T/t Complex (H. UEHARA, K. ABE, C.-H. PARK, K. ARTZT, and D. BENNETT)	272
Two-Dimensional Electrophoretic Protein Analysis in Human Genetics (J. KLOSE)	276
4 Genetic Mechanisms in Hereditary Disease	281
<i>β</i> -Thalassemic Syndromes As a Model for the Study of the Molecular Basis of Human Inherited Disease (S. OTTOLENGHI, B. GIGLIONI, P. COMI, R. MANTOVANI, N. MALGARETTI, S. NICOLIS, C. CAMASCHIELLA, and G. SAGLIO)	283
Common Disease: From Phenotype to Genotype (J.-M. LALOUEL)	294
Taxonomy and Inheritance of Anxiety States (C. R. CLONINGER)	302
The Genetics of Hypertension: An Unsolved Puzzle With Many Pieces (R. R. WILLIAMS, S. C. HUNT, S. J. HASSTEDT, L. B. JORDE, L. L. WU, G. K. BARLOW, K. O. ASH, B. M. STULTS, and H. KUIDA)	311
Genetic Risk Factors for Atherosclerotic Disease (K. BERG)	326
Apolipoprotein Gene Mutations, Dyslipoproteinemia and Coronary Heart Disease (J. L. BRESLOW)	336
Peroxisomal Disorders (J. FRÉZAL and A. MUNNICH)	342
Clinical Approach to Inherited Peroxisomal Disorders (B. T. POLL-THE, J. M. SAUDUBRAY, H. OGIER, A. LOMBES, A. MUNNICH, and J. FRÉZAL)	345
Inherited Peroxisomal Disorders: Defects in the Oxidation of Very Long Chain Fatty Acids and Phytanic Acid (A. POULOS, N. DERWAS, A. J. FELLEBERG, D. W. JOHNSON, B. PATON, P. SHARP, and H. SINGH)	352
Peroxisomal Disorders: Biochemical Characteristics and Genetic Relationship Between the Different Diseases (R. B. H. SCHUTGENS, R. J. A. WANDERS, A. W. SCHRAM, H. VAN DEN BOSCH, H. S. A. HEYMANS, G. SCHRAKAMP, and J. M. TAGER)	360
Biogenesis of Peroxisomes: Implications for Zellweger Syndrome (P. B. LAZAROW)	369
Transgenic Mice in the Study of Human Disease (P. LEDER)	377
Workshop: Nosology of the Inherited Disorders of Connective Tissue (P. BEIGHTON)	378
Nosologic Grouping in Birth Defects (J. M. OPITZ, A. CZEIZEL, J. A. EVANS, J. G. HALL, M. S. LUBINSKY, and J. W. SPRANGER)	382
Summary of the Workshop on DNA Repair Defects (D. BOOTSMA, M. VAN DUIN, R. T. JOHNSON, L. H. F. MULLENDERS, M. S. MEYN, C. J. R. CURRY, and R. A. GATTI)	386
Summary of the Workshop on New Approaches in Twin Research: Applications in Genetic Epidemiology (W. E. NANCE and P. PARISI)	390
The Temporal Dimension of the Gene (L. GEDDA)	394
Summary of the Workshop on Clinical and Genetic Heterogeneity of Fanconi's Anemia (A. D. AUERBACH, T. M. SCHROEDER-KURTH, A. ROGATKO, and N. E. MORTON)	396

5 Single Cell and Cancer Genetics	399
Chromosome Abnormalities and Oncogenes in Human Leukemia and Lymphoma (J. D. ROWLEY)	401
Comparative Genetic Analysis of Homeo-Box Genes in Mouse and Man (F. H. RUDDLE, C. P. HART, M. RABIN, A. C. FERGUSON-SMITH, and D. PRAVTCHEVA)	419
The Role of the Ph Chromosome in Chronic Myelocytic Leukemia (G. GROSVELD, A. DE KLEIN, A. HERMANS, L. HOEFSLOOT, M. VON LINDERN, N. HEISTERKAMP, J. GROFFEN, and D. BOOTSMA)	428
Somatic Cell and Molecular Genetics of Growth Factor Receptors (N. SHIMIZU)	435
6 Psychobiological and Behavior Genetics; Pharmacogenetics	439
Sex-Linked Mental Retardation (M. MIKKELSEN)	441
Introduction to the Symposium on Psychobiological Genetics (P. PROPPING)	450
Single Gene Effects in Psychiatric Disorders (P. PROPPING)	452
Genetics and Schizophrenia – Current State of Negotiations (I. I. GOTTESMAN, P. MCGUFFIN, A. FARMER, M. MCGUE, G. VOGLER, A. BERTELSEN, and D. C. RAO)	458
Inbreeding and the Epidemiology of Schizophrenia (L. SAUGSTAD and Ø. ØDEGÅRD)	466
Gene Activity in the CNS, a Tool for Understanding Brain Function and Dysfunction (J. G. SUTCLIFFE, M. KIEL, F. E. BLOOM, and R. J. MILNER)	474
Pharmacogenetic Approaches to the Study of Brain Function (J. I. NURNBERGER, JR.)	484
Introduction to the Symposium on Pharmacogenetics (H. W. GOEDDE)	492
Genetic Variation and Basis of Alcohol Metabolism and Response (H. W. GOEDDE and D. P. AGARWAL)	497
Genetic Variation in the Hepatic Cytochrome P-450 System (W. KALOW)	507
Genetic Variations in Hydrolytic Enzymes: Human Serum Cholinesterase and Paraoxonase (B. N. LA DU)	517
The N-Acetyl Transferase Polymorphism and Its Clinical Relevance (D. A. PRICE EVANS)	525
Workshop on the Rett Syndrome (F. HANEFELD, J. SCHMIDTKE, J. M. OPITZ, and J. WAHLSTRÖM)	533
7 Genes and Birth Defects in Populations	537
Hereditary Diseases in Asia: Chairman's Introduction (E. MATSUNAGA)	539
Incidence of Genetic Disease in Japan (E. MATSUNAGA)	540

Medical Genetic Survey of Soviet Middle Asian Populations (E. K. GINTER)	547
Anthropogenetic Study of Variation in the Craniospinal Joint (A. K. KALLA, I. P. SINGH, S. KHANNA, S. SHARMA, and F. VOGEL)	557
Genetic Diseases in China – Some Epidemiological Features (W. HWEI-YUEN LO)	569
Workshop on Population Genetics: Field Studies (G. FLATZ and DU RUOFU)	578
Workshop on Human Genetics Problems in the Third World (N. HASHEM)	580
Workshop on the Epidemiology of Congenital Anomalies (A. CZEIZEL)	586
Workshop on the Genetic Aspects of Human Adaptation (F. ROTHHAMMER, P. MAJUMDER, K. OMOTO, E. J. E. SZATHMARY, and F. M. SALZANO)	590

8 Genetic Counseling and Prenatal Diagnosis 595

Chromosomal Findings in Chorionic Villi: A Collaborative Study (M. MIKKELSEN and S. AYMÉ)	597
Chromosome Study of Chorionic Villi After Short-Term Incubation: Diagnostic and Experimental Applications (G. SIMONI)	607
Worldwide Experience with First-Trimester Fetal Diagnosis by Molecular Analysis (H. GALJAARD)	611
Workshop on Genetic Counseling (E. SEEMANOVÁ, R. WITKOWSKI, and H. W. RÜDIGER)	622
Workshop on Gene Mapping and Genetic Prediction for Autosomal Dominant Disorders (P. S. HARPER)	626
Workshop on the Prevention of Inherited Hemoglobinopathies (D. LOUKOPOULOS, A. CAO, ZENG YITAO, and O. O. AKINYANJU)	628
Workshop on the Prenatal Diagnosis of Cystic Fibrosis (D. J. H. BROCK)	631
Workshop on Training in Medical Genetics (W. FUHRMANN)	633

9 Genetic Engineering and Gene Therapy 637

Adenosine Deaminase Gene Transfer (J. W. BELMONT, J. HENKEL-TIGGES, K. WAGER-SMITH, S. M. W. CHANG, and C. TH. CASKEY)	639
Open Fetal Surgery 1986 (M. S. GOLBUS and M. R. HARRISON)	644
Medical In-Utero Therapy (J. D. SCHULMAN)	648

10 Ethical and Legal Issues 655

Ethics and Human Genetics: A Cross-Cultural Study in 17 Nations
(J. C. FLETCHER, D. C. WERTZ, J. R. SORENSON, and K. BERG) 657

Comparative Law and Legislation on Eugenic Sterilization and Selective
Abortion (B. M. DICKENS) 673

Ethical Issues in Reproductive Alternatives for Genetic Indications
(H. KUHSE and P. SINGER) 683

Permanent Committee for the International Congresses of Human
Genetics 692

Subject Index 695