Contents
Volume 1

SECTION 1
Basic Principles

1. Nature and incidence of genetic disease 3
A. E. H. Emery, D. L. Rimoin
2. Gene structure and function in eukaryotic organisms 7
Gregory S. Barsh, Charles J. Epstein
3. Molecular biology in relation to medical genetics 33
R. F. Mueller
4. Mutation in man 53
F. Vogel
5. Chromosomal basis of inheritance 77
O. J. Miller
6. Unifactorial inheritance 95
R. Skinner
7. Bayesian methods in medical genetics 107
A. E. H. Emery
8. Segregation analysis 115
M. Anne Spence
9. Population genetics (Hardy-Weinberg equilibrium and factors affecting it) 121
J. A. Sofaer
10. Gene mapping 133
U. Francke
11. Analysis of genetic linkage 149
Jean-Marc Lalouel, Ray White
12. Multifactorial inheritance 165
D. T. Bishop
13. Twins 175
J. A. Sofaer
14. Teratogenic agents 183
James W. Hanson
15. A clinical approach to the dysmorphic child 215
K. L. Jones, M. C. Jones
16. Short stature 225
David L. Rimoin, John M. Graham, Jr

SECTION 2
Chromosome Disorders

17. Human cytogenetic nomenclature 237
H. A. Lubs, P. S. Ing
18. Autosomal disorders 247
J. de Grouchy, C. Turleau
19. Sex chromosome abnormalities 273
A. de la Chapelle
20. X-linked mental retardation and the fragile X 301
H. A. Lubs
21. Infertility and recurrent abortion 313
A. C. Chandley

SECTION 3
Systemic Disorders

A: NEUROLOGICAL DISORDERS

22. The genetics and prevention of neural tube defects and 'uncomplicated' hydrocephalus 323
K. M. Laurence
23. The convulsive disorders 347
G. C. Sutton
24. Hereditary disorders of the basal ganglia 357
R. Eldridge
25. Huntington disease 373
P. Michael Conneally
26. The hereditary ataxias and paraplegias 383
A. E. Harding

27. Autonomic and sensory disorders 397
F. Axelrod

28. Peripheral neuropathies 413
P. K. Thomas, A. E. Harding

29. The phakomatoses 435
Vincent M. Riccardi

30. Demyelinating disorders 447
Sarah Bundey

B: MENTAL DISORDERS

31. Schizophrenia and major mood disorders (Manic-depressive illness) 457
D. K. Kinney

32. The presenile dementias 473
D. Vassilopoulos

33. Addictive disorders 481
D. P. Agarwal, H. W. Goedde

34. Mental retardation 495
Hugo W. Moser, Craig T. Ramey, Claire O. Leonard

35. Congenital myopathies (including glycogenoses) 513
J. Z. Heckmatt, V. Dubowitz

C: NEUROMUSCULAR DISORDERS

36. The muscular dystrophies 539
A. E. H. Emery

37. Spinal muscular atrophies 565
J. Pearn

38. Myotonic dystrophy and related disorders 579
P. S. Harper

39. Myasthenia gravis 599
Gerald M. Fenichel

40. Motor neuronal diseases 609
J. Kelemen, W. G. Bradley

41. The periodic paralyses 621
I. Gamstorp

D: OPHTHALMOLOGICAL DISORDERS

42. Congenital blindness 631
M. Warburg

43. Optic atrophy 637
L. N. Went

44. Glaucoma: congenital and later onset 645
Sherwin J. Isenberg, John R. Heckenlively

45. Defects of the cornea 655
J. Sugar

46. Anomalies of the lens 669
H.-R. Koch, A. Wegener, E. Roth

47. Hereditary retinal and choroidal degenerations 683
John R. Heckenlively

48. Retinoblastoma 705
D. F. Roberts, G. E. S. Aherne

49. Genetic aspects of strabismus 723
J. Bronwyn Bateman, Sherwin J. Isenberg

E: HEREDITARY DEAFNESS

50. Hereditary deafness 733
P. Beighton

F: CRANIOFACIAL DISORDERS

51. Craniofacial disorders 749
M. Michael Cohen Jr, F. Clarke Fraser, Robert J. Gorlin

G: DERMATOLOGICAL DISORDERS

52. Abnormalities of pigmentation 797
Carl J. Witkop, Jr

53. Ichthyosiform dermatoses 835
Howard P. Baden, P. Hooker

54. Epidermolysis bullosa 855
T. Gedde-Dahl Jr, I. Anton-Lamprecht

55. Other genetic disorders of the skin 877
L. A. Goldsmith

INDEX (Volumes 1 and 2)

Contents
Volume 2

H: SKELETAL DISORDERS

56. The chondrodysplasias 895
D. L. Rimoin, R. S. Lachman

57. Disorders of bone density, volume and mineralisation 933
D. O. Sillence

58. Abnormalities of bone structure 953
William A. Horton

59. Dysostoses 967
J. G. Hall

60. Arthrogryposes (multiple congenital contractures) 989
J. G. Hall

61. Common skeletal deformities 1037
William A. Horton

I: CONNECTIVE TISSUE DISORDERS

62. Marfan syndrome 1047
R. E. Pyeritz

63. Ehlers-Danlos syndrome 1065
P. H. Byers, K. A. Holbrook

64. Pseudoxanthoma elasticum and related disorders 1083
R. M. Goodman

J: GASTROINTESTINAL DISORDERS

65. Peptic ulcer 1097
J. I. Rotter, T. Shohat

66. Developmental defects of the gastrointestinal tract 1117
E. Passarge

67. The polyposes 1125
A. M. O. Veale

68. Inherited disorders of bilirubin metabolism 1135
Jayanta Roy Chowdhury, Pulak Lahiri, Namita Roy Chowdhury

K: RESPIRATORY SYSTEM DISORDERS

69. Cystic fibrosis 1165
W. M. McCrae, R. Williamson

70. Asthma and other allergic conditions 1173
J. A. Raeburn

71. Alpha$_1$-antitrypsin deficiency and related disorders 1179
Jack Lieberman

L: CARDIOVASCULAR DISORDERS

72. Congenital heart defects 1207
Virginia V. Michels, Vincent M. Riccardi

73. Coronary heart disease 1239
Gerd Utermann

74. The cardiomyopathies 1263
R. Emanuel, R. Withers

M: RENAL DISORDERS

75. Congenital and hereditary urinary tract disorders 1273
J. Zonana, J. H. DiLiberti

76. Renal cystic diseases 1291
Stanley C. Jordan, Hooshang Kangarloo

77. The nephrotic syndromes 1305
R. Norio

N: HAEMATOLOGICAL DISORDERS

78. Haemoglobinopathies and thalassaemias 1315
J. A. Phillips, H. H. Kazazian, Jr

79. Hereditary red blood cell disorders (Excluding haemoglobinopathies and thalassaemias) 1343
Bertil E. Glader

80. Congenital disorders of haemostasis 1371
C. A. Ludlam

81. Leukaemias, lymphomas and related disorders 1391
Janet D. Rowley

O: IMMUNOLOGICAL DISORDERS

82. Immunodeficiency disorders 1411
R. Hirschhorn, K. Hirschhorn

83. Complement defects 1431
F. S. Rosen, C. A. Alper

84. Disorders of leucocyte function 1439
M. E. Miller, H. R. Hill

85. The HLA system 1453
T. Strachan, R. Harris

P: ENDOCRINOLOGICAL DISORDERS

86. Genetic disorders of the pituitary gland 1461
D. L. Rimoin

87. Thyroid disorders 1489
D. A. Fisher

88. Parathyroid disorders 1503
C. E. Jackson

89. Diabetes mellitus 1521
C. M. Vadheim, D. L. Rimoin, J. I. Rotter

90. Congenital adrenal hyperplasia 1559
Maria I. New, Perrin C. White, Phyllis W. Speiser, Christopher Crawford, Bo Dupont

91. Disorders of gonads and internal reproductive ducts 1593
Joe Leigh Simpson

92. Disorders of vitamin D metabolism or action 1619
Stephen J. Marx

Q: METABOLIC DISORDERS

93. Disorders of amino acid metabolism 1639
C. R. Scott, S. D. Cederbaum

94. Disorders of carbohydrate metabolism 1675
Won G. Ng, Thomas F. Roe, George N. Donnell

95. Disorders of purine and pyrimidine metabolism 1697
J. E. Seegmiller

96. Disorders of organic acid metabolism 1723
S. I. Goodman

97. Renal tubular disorders 1739
R. Hillman

98. The inherited porphyrias 1747
Robert J. Desnick, Andrew G. Roberts, Karl E. Anderson

99. Disorders of copper metabolism 1771
D. M. Danks

100. Disorders of iron metabolism and related disorders 1783
Marcel Simon

101. The mucopolysaccharidoses 1797
J. Spranger

102. The oligosaccharidoses 1807
J. G. Leroy

103. Gangliosidoses and related lipid storage diseases 1827
Alan K. Percy

104. The peroxisomal disorders 1857
R. B. H. Schutgens, H. S. A. Heymans, R. J. A. Wanders

R: PHARMACOGENETICS

105. Pharmacogenetics 1869
David A. Price Evans

S: NEOPLASTIC DISORDERS

106. Cancer genetics 1881
R. Neil Schimke

107. Oncogenes 1909
James V. Watson, Karol Sikora

SECTION 4
Applied Genetics

108. Genetic counselling 1923
R. Skinner

109. Newborn genetic screening 1935
R. W. Erbe, G. R. Boss

110. Heterozygote screening 1951
M. M. Kaback

111. Prenatal diagnosis and therapy 1959
Joel Charrow, Henry L. Nadler, Mark I. Evans

112. Genetic registers 1995
Andrew P. Read

113. Treatment of inherited metabolic diseases 2001
Robert J. Desnick

114. Paternity testing 2011
Robert S. Sparkes, Susan E. Hodge

115. Legal considerations in the delivery of genetic care 2017
Margery W. Shaw

116. Challenges for the future 2025
H. Galjaard

INDEX (Volumes 1 and 2)