

Part I Overview

1. Genetically Determined Immunodeficiency Diseases:
A Perspective 3
C. I. Edvard Smith, Hans D. Ochs, and Jennifer M. Puck
2. Genetic Principles and Technologies in the Study
of Immune Disorders 16
Jennifer M. Puck and Robert L. Nussbaum
3. Mammalian Hematopoietic Development
and Function 27
Gerald J. Spangrude
4. T Cell Development 39
*Rae S. M. Yeung, Pamela S. Ohashi, Mary E. Saunders,
and Tak W. Mak*
5. Molecular Mechanisms Guiding B Cell Development 61
*Antonius G. Rolink, Jan Andersson, Ulf Grawunder,
and Fritz Melchers*
6. Signal Transduction by T and B Lymphocyte
Antigen Receptors 71
Neetu Gupta, Anthony L. DeFranco, and Arthur Weiss
7. Lymphoid Organ Development, Cell Trafficking,
and Lymphocyte Responses 93
Sirpa Jalkanen and Marko Salmi
8. Phagocytic System 103
Kuender D. Yang, Paul G. Quie, and Harry R. Hill

Part II Syndromes

9. X-Linked Severe Combined Immunodeficiency 123
Jennifer M. Puck
10. Autosomal Recessive Severe Combined Immunodeficiency
Due to Defects in Cytokine Signaling Pathways 137
Fabio Candotti and Luigi Notarangelo

11. V(D)J Recombination Defects 153
Jean-Pierre de Villartay, Klaus Schwarz, and Anna Villa
12. Immunodeficiency Due to Defects of Purine Metabolism 169
Rochelle Hirschhorn and Fabio Candotti
13. Severe Combined Immunodeficiency Due to Mutations in the CD45 Gene 197
Talal A. Chatila and Markku Heikinheimo
14. Severe Combined Immunodeficiency Due to Defects in T Cell Receptor-Associated Protein Kinases 203
Melissa E. Elder
15. Human Interleukin-2 Receptor α Deficiency 212
Chaim M. Roifman
16. CD3 and CD8 Deficiencies 216
José R. Regueiro and Teresa Espanol
17. Molecular Basis of Major Histocompatibility Complex Class II Deficiency 227
Walter Reith, Barbara Lisowska-Groszpierré, and Alain Fischer
18. Peptide Transporter Defects in Human Leukocyte Antigen Class I Deficiency 242
Henri de la Salle, Lionel Donato, and Daniel Hanau
19. CD40, CD40 Ligand, and the Hyper-IgM Syndrome 251
Raif S. Geha, Alessandro Plebani, and Luigi D. Notarangelo
20. Autosomal Hyper-IgM Syndromes Caused by an Intrinsic B Cell Defect 269
Anne Durandy, Patrick Revy, and Alain Fischer
21. X-linked Agammaglobulinemia: A Disease of Btk Tyrosine Kinase 279
C. I. Edvard Smith, Anne B. Satterthwaite, and Owen N. Witte
22. Autosomal Recessive Agammaglobulinemia 304
Mary Ellen Conley

23. Genetic Approach to Common Variable Immunodeficiency and IgA Deficiency 313
Lennart Hammarström and C. I. Edvard Smith
24. Autoimmune Lymphoproliferative Syndrome 326
Jennifer M. Puck, Frederic Rieux-Laucat, Françoise Le Deist, and Stephen E. Straus
25. Autoimmune Polyendocrinopathy, Candidiasis, Ectodermal Dystrophy 342
Leena Peltonen-Palotie, Maria Halonen, and Jaakko Perheentupa
26. Immune Dysregulation, Polyendocrinopathy, Enteropathy, and X-Linked Inheritance 355
Troy R. Torgerson, Eleonora Gambineri, Steven F. Ziegler, and Hans D. Ochs
27. Periodic Fever Syndromes 367
Daniel L. Kastner, Susannah Brydges, and Keith M. Hull
28. Inherited Disorders of the Interleukin-12/23–Interferon Gamma Axis 390
Melanie J. Newport, Steven M. Holland, Michael Levin, and Jean-Laurent Casanova
29. Ataxia-Telangiectasia 402
Martin F. Lavin and Yosef Shiloh
30. Chromosomal Instability Syndromes Other Than Ataxia-Telangiectasia 427
Rolf-Dieter Wegner, James J. German, Krystyna H. Chrzanowska, Martin Digweed, and Markus Stumm
31. Wiskott-Aldrich Syndrome 454
Hans D. Ochs and Fred S. Rosen
32. X-Linked Lymphoproliferative Disease Due to Defects of *SH2D1A* 470
Volker Schuster and Cox Terhorst
33. DiGeorge Syndrome: A Chromosome 22q11.2 Deletion Syndrome 485
Deborah A. Driscoll and Kathleen E. Sullivan
34. Hyper-IgE Recurrent Infection Syndromes 496
Bodo Grimbacher, Jennifer M. Puck, and Steven M. Holland
35. Immunodeficiency with Centromere Instability and Facial Anomalies 505
R. Scott Hansen, Corry Weemaes, and Cisca Wijmenga

36. Immunodeficiencies with Associated Manifestations of Skin, Hair, Teeth, and Skeleton 513
Mario Abinun, Ilkka Kaitila, and Jean-Laurent Casanova
37. Chronic Granulomatous Disease 525
Dirk Roos, Taco W. Kuijpers, and John T. Curnutte
38. Cell Adhesion and Leukocyte Adhesion Defects 550
Amos Etzioni and John M. Harlan
39. Cyclic and Congenital Neutropenia Due to Defects in Neutrophil Elastase 565
David C. Dale and Andrew G. Aprikyan
40. Chediak-Higashi Syndrome 570
Richard A. Spritz
41. Inherited Hemophagocytic Syndromes 578
Geneviève de Saint Basile
42. Genetically Determined Deficiencies of the Complement System 589
Kathleen E. Sullivan and Jerry A. Winkelstein

Part III Assessment and Treatment

43. Assessment of the Immune System 611
Helen M. Chapel, Siraj Misbah, and A. David B. Webster
44. Genetic Aspects of Primary Immunodeficiencies 633
Jennifer M. Puck
45. Immunodeficiency Information Services 644
Jouni Väliäho, Crina Samarghitean, Hilikka Piirilä, Marianne Pusa, and Mauno Vihinen
46. Conventional Therapy of Primary Immunodeficiency Diseases 655
E. Richard Stiehm and Helen M. Chapel
47. Bone Marrow Transplantation for Primary Immunodeficiency Diseases 669
Rebecca H. Buckley and Alain Fischer
48. Gene Therapy 688
Fabio Candotti and Alain Fischer