

DEFECTS OF SIGNALING

T-B+NK-SCID

- 1 γc
- 2 JAK3

T-B+NK+SCID

- 3 IL-7R α
- 4 CD45
- 5 CD3 ϵ/ζ
- 6 Corrin-1A

T+B+NK+SCID

- 7 Lck p56

Non-SCID defects

- 8 CD3 γ
- 9 ZAP70
- 10 CD8 α
- 11 Itk
- 12 STAT5b
- 13 CD25
- 14 CD40L
- 15 DOCK8
- 16 Ca Channels

DEFECTS in DNA

Rearrangement and Repair

Non-SCID defects

- 17 Winged helix deficiency (FOXN1)
- 18 DNA ligase IV
- 19 Cernunnos deficiency

T-B-NK+SCID

- 20 RAG 1/2 deficiency
- 21 DCLRE1C (Artemis) deficiency
- 22 DNA PK ζ deficiency

T-B(low)/NK+

- 23 Reticular dysgenesis (*ak2* mutation)

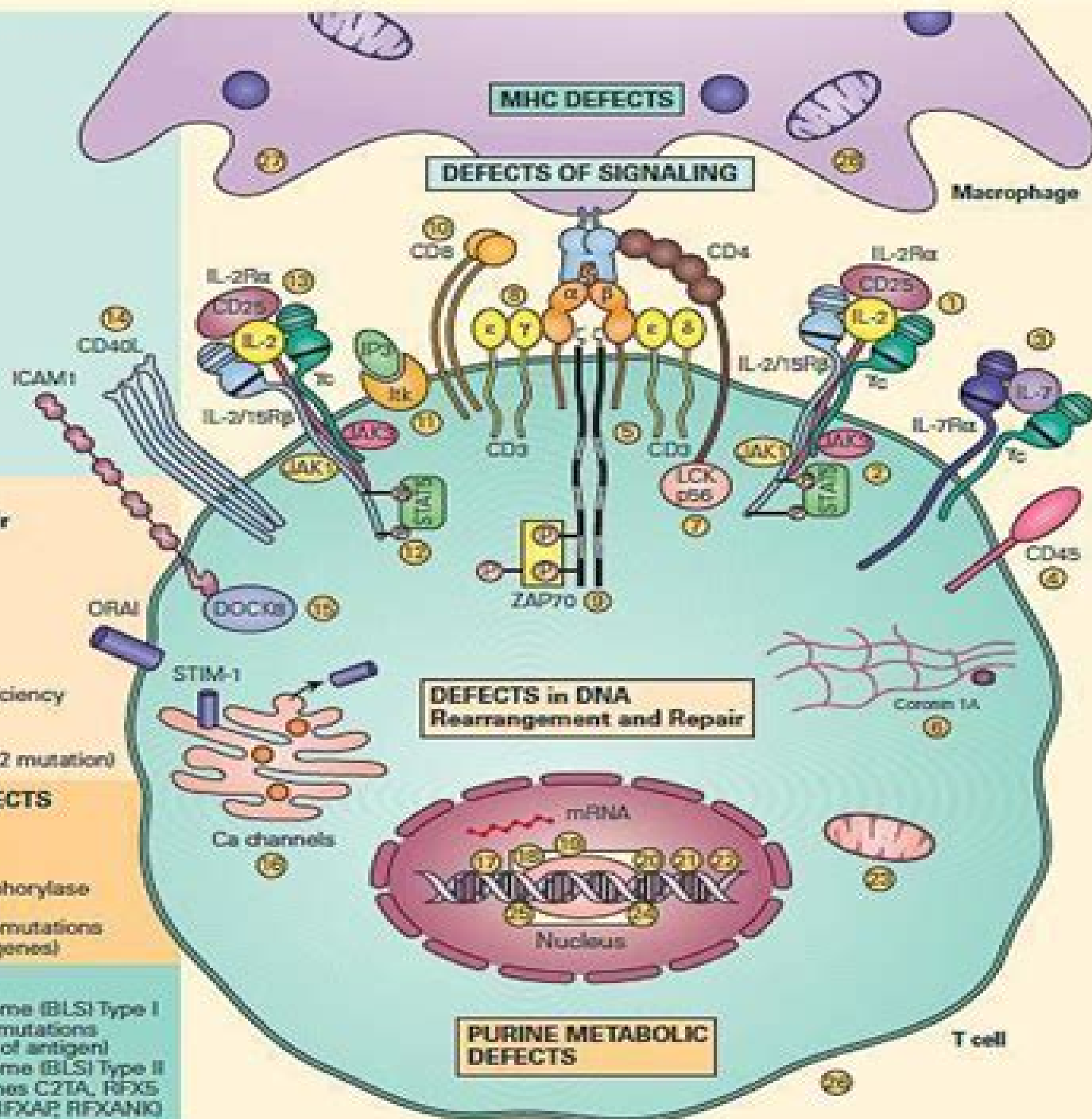
PURINE METABOLIC DEFECTS

T-B-NK-SCID

- 24 Adenosine deaminase (ADA) deficiency
- 25 Purine nucleoside phosphorylase (PNP) deficiency
- 26 Omenn syndrome (hypomutations in several SCID-related genes)

MHC DEFECTS

- 27 Bare lymphocyte syndrome (BLS) Type I (TAP1, TAP2, or Tapasin mutations affecting MHC-I loading of antigen)
- 28 Bare lymphocyte syndrome (BLS) Type II (deficiencies in chaperones C2TA, RFX5 or transcription factors RFXAP, RFXANK)



Biological Basis Of Immunodeficiency

I. Edward Alcamo



Biological Basis Of Immunodeficiency:

Biological Basis of Immunodeficiency Erwin W. Gelfand, Hans-Michael Dosch, 1980 *Genetic Basis of Primary Immunodeficiency*, 1994 *Biological Basis of Immunodeficiency*, 1980 AIDS: The Biological Basis Benjamin S. Weeks, I. Edward Alcamo, 2009-02-02 **AIDS, the biological basis** I. Edward Alcamo, 1993 **Biological Basis of Immunodeficiency** Erwin W. Gelfand, Hans-Michael Dosch, 1980 *Principles and Practice of Medical Genetics* Alan E. H. Emery, David L. Rimo, 1990 **Biomedical Index to PHS-supported Research**, 1987 **Biomedical Index to PHS-supported Research: pt. A. Subject access A-H**, 1994 *Primary Immunodeficiency Diseases* Hans D. Ochs, MD, Dr. med, C. I. Edvard Smith, PhD, Jennifer M. Puck, MD, 2013-11-01 Primary immunodeficiency diseases first recognized 60 years ago are inherited disorders that affect human adaptive and innate immunity In most cases affected individuals experience recurrent infections but they may also suffer from autoimmune diseases and malignancies This third edition of *Primary Immunodeficiency Diseases* provides readers with the historic and scientific background clinical presentations immunologic characteristics and the molecular genetic underpinnings of this rapidly enlarging class of diseases With up to date diagnostic tools and therapeutic options from prophylactic anti infective measures to hematopoietic stem cell transplantation and gene therapy this volume will remain an authoritative resource on this increasingly important area

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In a fast-paced digital era where connections and knowledge intertwine, the enigmatic realm of language reveals its inherent magic. Its capacity to stir emotions, ignite contemplation, and catalyze profound transformations is nothing lacking extraordinary. Within the captivating pages of **Biological Basis Of Immunodeficiency** a literary masterpiece penned with a renowned author, readers embark on a transformative journey, unlocking the secrets and untapped potential embedded within each word. In this evaluation, we shall explore the book's core themes, assess its distinct writing style, and delve into its lasting effect on the hearts and minds of people who partake in its reading experience.

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