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Titolo	Agammaglobulinemia / / edited by Alessandro Plebani, Vassilios Lougaris
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ISBN	3-319-22714-9
Edizione	[1st ed. 2015.]
Descrizione fisica	1 online resource (126 p.)
Collana	Rare Diseases of the Immune System, , 2282-6505 ; ; 4
Disciplina	616.0798
Soggetti	Immunology Rheumatology Hematology Pediatrics Cytology Cell Biology
Lingua di pubblicazione	Inglese
Formato	Materiale a stampa
Livello bibliografico	Monografia
Note generali	Description based upon print version of record.
Nota di bibliografia	Includes bibliographical references at the end of each chapters and index.
Nota di contenuto	Early B Cell Biology -- Agammaglobulinemia: Basic Pathogenesis and Clinical Spectrum (Including X-Linked and Autosomal Recessive Forms) -- Pulmonary Complications in Agammaglobulinemia -- Immunoglobulin Replacement Therapy: Past, Present, Future -- Mutational Spectrum of BTK: A Comprehensive Description -- Novel Therapeutic Options for X-Linked Agammaglobulinemia -- BTK in Non B Cells.
Sommario/riassunto	This book provides an updated overview of agammaglobulinemia, a rare form of primary immunodeficiency which is considered the prototype of the congenital humoral defects, and which is characterized by the absence of peripheral B cells and very low serum immunoglobulin levels. The book opens by discussing the highly orchestrated early B cell development in the bone marrow and the genes involved based on both human and animal models. The pathogenesis and clinical presentation of X-linked agammaglobulinemia, caused by mutations in the BTK (Bruton’s

tyrosine kinase) gene, are then presented in detail, followed by descriptions of the clinical manifestations and molecular basis of the less frequent autosomal recessive and autosomal dominant forms of agammaglobulinemia. Patients' management in terms of respiratory complications, gammaglobulin replacement therapy and the potential value of novel experimental therapeutic strategies are discussed. The book's closing chapters offer a comprehensive and updated description of mutations in the BTK gene, and the expression and function of BTK in cells other than B cells.