

1. Record Nr.	UNINA9910877191103321
Autore	Federici Augusto B
Titolo	Textbook of Von Willebrand Disease : Basic and Clinical Aspects
Pubbl/distr/stampa	Newark : , : John Wiley & Sons, Incorporated, , 2024 ©2024
ISBN	1-119-41952-2 1-119-41947-6 1-119-41949-2
Edizione	[2nd ed.]
Descrizione fisica	1 online resource (306 pages)
Altri autori (Persone)	BerntorpErik E LillicrapDavid MontgomeryRobert R
Disciplina	616.1/57
Soggetti	Von Willebrand Diseases
Lingua di pubblicazione	Inglese
Formato	Materiale a stampa
Livello bibliografico	Monografia
Nota di contenuto	Cover -- Title Page -- Copyright Page -- Contents -- List of Contributors -- Foreword -- Preface -- Chapter 1 Historical perspective on von Willebrand disease -- Introduction -- The scientist of the disease -- First description of the disease: the Åland family -- Other early clinical reports -- The search for a new factor-the bleeding time factor -- The end of the beginning -- Recent scientific visits to Åland Islands -- References -- Chapter 2 Biosynthesis and organization of von Willebrand factor -- Introduction -- Terminology -- Molecular biology of VWF -- The VWF gene -- VWF domain structure -- VWF promoter -- Cell biology of VWF -- VWF processing and dimerization in the endoplasmic reticulum -- VWF processing in the Golgi -- VWF multimerization -- Regulated storage of VWF in Weibel-Palade bodies and -granules -- Weibel-Palade body biogenesis -- Regulated release of VWF -- Postsecretion modification of VWF -- References -- Chapter 3 Von Willebrand factor structure and function -- Introduction -- VWF function -- Structure of VWF -- VWF domain structure -- Functional biochemistry of VWF -- Interaction of VWF with FVIII -- Two sites of interaction between VWF and platelets -- Proteolysis of VWF -- Carbohydrate modifications of VWF -- Clearance

of VWF through autoantibodies and other mechanisms -- Increased clearance as a cause of type 1C VWD -- Critical disulfide structure of VWF -- Shear and its effect on VWF -- Structure and function of the VWF propeptide -- Summary -- Acknowledgement -- References -- Chapter 4 Regulation of von Willebrand factor expression -- Introduction -- VWF gene structure and chromosomal location -- VWF expressing cells -- VWF promoter structure -- Transcriptional activators -- Transcriptional suppressors -- NFY, a dual-purpose transcription factor -- Influence of regulatory sequence variants on VWF expression.

Regulatory variants in the proximal regulatory region: the VWF promoter -- Regulatory variants in the distal regulatory region: VWF enhancers -- Epigenetic regulation of VWF expression -- Posttranscriptional regulation of VWF expression -- Splicing events and differential VWF expression in endothelial cells -- MicroRNAs regulate VWF biosynthesis, maturation, and secretion -- Physiological, pathological, and environmental factors affecting VWF expression -- Hemodynamic shear stress regulates VWF expression -- VWF is an acute-phase reactant -- Hormones raise blood VWF levels -- Conclusion -- References -- Chapter 5 Modulation of VWF by ADAMTS13 -- Introduction -- VWF structure, synthesis, and function -- ADAMTS13 structure -- ADAMTS family proteases -- Prodomain -- Metalloprotease (MP) domain -- Disintegrin-like (Dis) domain -- Thrombospondin type 1 repeats (TSP) -- Cysteine-rich (Cys-rich) domain -- Spacer domain -- CUB domains -- ADAMTS13 conformation/latency -- Global latency -- Local latency -- ADAMTS13 function -- Physiology and pathophysiology of the VWF-ADAMTS13 axis -- Summary -- References -- Chapter 6 Assessment of VWF clearance -- Introduction -- Macrophage-mediated VWF clearance -- Low-density lipoprotein receptor-related protein 1 (LRP1) -- Scavenger receptor class A member I (SR-A1) -- Macrophage galactose lectin (MGL) -- Siglec-5 -- Endothelial cell contribution to VWF clearance -- C-type lectin domain family 4 member M (CLEC4M) -- Stabilin-2 -- Scavenger Receptor Class A Member 5 (SCARA5) -- Conclusion -- References -- Chapter 7 Classification of VWD -- Classification of VWD -- von Willebrand factor -- Historical classification -- Type 1 VWD -- Type 1C VWD -- Type 1 versus low VWF -- Type 2 VWD -- Type 3 VWD -- Clinical classification of VWD -- Genetic classification of VWD -- Conclusion -- Acknowledgement -- Conflict of Interest.

References -- Chapter 8 The epidemiology of von Willebrand disease -- Introduction -- Historical studies on the prevalence of VWD -- Prevalence of bleeding patients in the general population -- Bleeding score: A new diagnostic tool to assess clinically relevant VWD -- The problem of diagnosing mild VWD -- Prevalence of intermediate VWD -- Prevalence of severe VWD -- Prevalence of a mutant VWF gene -- Conclusions -- References -- Chapter 9 Clinical aspects of von Willebrand disease: bleeding history -- Introduction -- Bleeding history in VWD -- Bleeding symptoms in VWD -- Cutaneous symptoms -- Epistaxis -- Gynecologic bleeding -- Oral cavity bleeding -- Postoperative bleeding -- Gastrointestinal bleeding -- Other bleeding symptoms -- Specific situations -- Women-the value of gynecologic assessment -- Bleeding assessment tools -- Conclusion -- References -- Chapter 10 Laboratory diagnosis of von Willebrand disease: the phenotype -- Screening diagnostic tests -- Bleeding time -- PFA-100/200 and other global shear stress test systems -- Activated partial thromboplastin time -- Platelet adhesion -- Platelet count -- Extended diagnostic tests -- Assay of the FVIII/VWF complex -- Ristocetin-induced agglutination in platelet-rich plasma (RIPA) -- Botrocetin-

induced aggregation in platelet-rich plasma (BIPA) -- Binding studies with washed platelets -- VWF propeptide (VWFpp or VWF:Ag II) -- Qualitative changes in VWF -- VWF multimers -- VWF fragments -- FVIII-binding capacity of VWF (VWF:FVIIIB) -- VWF antibodies/inhibitors -- Diagnosis in neonates and young children -- Diagnosis in pregnancy -- Desmopressin trials as an aid to the diagnosis and functional characterization of VWD -- An algorithmic approach to diagnosis of VWD -- Future perspectives -- References -- Chapter 11 Molecular diagnosis of von Willebrand disease: the genotype.
Introduction and role of genetic testing in VWD -- Techniques used in molecular analysis of VWD -- Direct Sanger sequencing -- Next generation sequencing -- Copy number variant analysis -- Assessment of variant pathogenicity -- Molecular spectrum of VWD -- Type 1 -- Type 2 -- Type 3 -- Challenges and future of VWD genotyping -- Acknowledgments -- References -- Chapter 12 Clinical, laboratory, and molecular markers of type 1 von Willebrand disease and low von Willebrand factor -- Introduction -- The epidemiology of type 1 von Willebrand disease -- Clinical features of type 1 von Willebrand disease -- The laboratory diagnosis of type 1 von Willebrand disease -- The genetics of type 1 von Willebrand disease -- The role of ABO blood group and type 1 von Willebrand disease -- von Willebrand factor gene mutations and type 1 von Willebrand disease -- Recurrent type I von Willebrand disease candidate mutations -- Non-coding sequence variants in type 1 von Willebrand disease -- Type 1 von Willebrand disease and accelerated clearance of von Willebrand factor -- Future priorities in type 1 von Willebrand disease characterization -- References -- Chapter 13 Clinical and molecular markers of type 1C VWD -- Introduction -- Epidemiology -- Clinical features -- Laboratory diagnosis and studies on increased clearance in type 1 VWD -- Genetics and the specific mutations of type 1C VWD -- The role of glycans in increased VWF clearance -- Summary -- References -- Chapter 14 Clinical and molecular markers of VWD2A -- Introduction -- Pathophysiology -- Clinical manifestations -- Laboratory diagnosis -- Molecular markers -- Treatment -- References -- Chapter 15 Clinical and molecular markers of VWD2B -- Introduction -- Pathophysiology -- Clinical manifestations -- Laboratory diagnosis -- Molecular markers -- Treatment -- Conclusions -- References -- Chapter 16 Clinical and molecular markers of type 2M VWD -- Introduction -- Clinical features of type 2M VWD -- Laboratory classification of type 2M VWD -- Problems with ristocetin in VWF assays -- Genetic variants found in type 2M VWD -- Treatment of type 2M VWD -- Summary -- Acknowledgements -- Conflicts of Interest -- References -- Chapter 17 Clinical and molecular markers of VWD2N -- The VWF-FVIII interaction -- Laboratory diagnosis -- Molecular analysis -- Proposed algorithm for the diagnosis of type 2N -- Clinical symptoms -- Therapeutic options -- Conclusions -- References -- Chapter 18 Clinical, laboratory, and molecular markers of type 3 von Willebrand disease -- General definition, history, and epidemiology -- General definitions -- History -- Epidemiology -- Clinical markers of VWD3 -- Laboratory markers of VWD3 -- Molecular markers of VWD3 -- Inheritance pattern -- Large gene deletions -- Nonsense mutations -- Small deletions and insertions -- Splice site mutation8 -- Missense mutation -- Additional VWF molecular markers found in 3WINTERS-IPS patients -- Prenatal and molecular diagnosis of VWD3 -- Treatment and prevention of bleeding in VWD3 -- Treatment of patients with alloantibodies to VWF -- Secondary long-term prophylaxis -- Future perspectives -- Acknowledgments -- References -- Chapter 19 Pediatric aspects of von Willebrand disease -- Introduction --

Diagnosis of VWD in childhood -- Clinical presentation and screening tests -- Diagnosis of type 1 VWD versus low VWF levels as a risk factor for bleeding -- VWD in neonates -- Acquired VWS in childhood -- VWD in adolescents -- Treatment strategies in children -- General principles -- DDAVP -- Replacement therapy -- Conclusions -- References -- Chapter 20 Women with von Willebrand Disease -- Introduction -- Gynecology -- Approach to screening and diagnosis of VWD in women with HMB.

Gynecological problems in women with VWD.

Sommario/riassunto

"Von Willebrand Disease: Basic and Clinical Aspects Second Edition describes the important and complex role of von Willebrand factor in hemostasis and thrombosis. In addition to the current understanding of its molecular biology, this book gives particular focus to the association between genetic variants of von Willebrand factor and different von Willebrand disease phenotypes. It also reviews the important area of the obstetric and gynecological manifestations of von Willebrand disease, as well as the treatment of acute bleeding, GI bleeding, and how to prepare VWB patient for surgery. Many advances in agents are included in this updated edition as well as the wide topics such as VWF beyond Hemostasis, in Angiogenesis, and VWF/ADAMTS13 as risk factors of thrombosis. This valuable book is written by an international team of editors and contributors and is a valuable resource for hematologists in practice and in training, and specialists in thrombosis and hemostasis"--
