

# Molecular and Translational Medicine

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Paula E. North • Tara Sander  
Editors

# Vascular Tumors and Developmental Malformations

Pathogenic Mechanisms and Molecular  
Diagnosis

 Humana Press

*Editors*

Paula E. North  
Department of Pathology, Division of  
Pediatric Pathology  
Medical College of Wisconsin  
Milwaukee, WI, USA

Tara Sander  
Department of Pathology, Division of  
Pediatric Pathology  
Medical College of Wisconsin  
Milwaukee, WI, USA

ISSN 2197-7852

ISSN 2197-7860 (electronic)

Molecular and Translational Medicine

ISBN 978-1-4939-3239-9

ISBN 978-1-4939-3240-5 (eBook)

DOI 10.1007/978-1-4939-3240-5

Library of Congress Control Number: 2015956616

Springer New York Heidelberg Dordrecht London  
© Springer Science+Business Media New York 2016

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# Preface

*Vascular anomalies* encompass a dizzying array of distinct clinicopathologic entities that can be grouped, roughly, into two general categories: vascular tumors and developmental vascular malformations. The vascular tumors are intrinsically proliferative lesions, at least in some phase of their inception, and include both benign and malignant neoplasms as well as reactive proliferations. The vascular malformations are more static congenital errors in development of the vasculature that may evolve over time under environmental and/or genetic influences. Many of these anomalies, even those officially “benign,” can be absolutely devastating for patients over the course of a childhood or a lifetime, also placing great burden on families and frustrating healthcare providers who struggle to provide relief. Current mainstream treatment options are relatively limited, often ineffective, and may be accompanied by significant clinical complications. Surgery and/or interventional radiological approaches are sometimes good solutions, but effective medical intervention would be a much better option for many patients. Design of rational targets for such medical intervention requires understanding of mechanisms of disease. Fortunately, discovery in that regard is escalating.

In this book we bring together a complimentary group of authors representing clinical practice, surgical pathology, molecular diagnostics, and basic science to present different aspects of the puzzle of vascular anomalies. We believe the timing is right, as pieces of this puzzle are now rapidly beginning to fall together. Interest in vascular anomalies is increasing among clinicians and scientists alike, spurred in part by new basic science discoveries as well as by recent beneficial use of “old” drugs like propranolol and rapamycin for new purposes in this field. More and more, basic scientists focused upon vasculogenesis and angiogenesis are attracted by meaningful translational applications in the field of vascular anomalies in addition to other medical fields such as cancer and wound healing. Multidisciplinary centers for the diagnosis, treatment, and study of vascular anomalies have sprung up worldwide. The marvelous new tools we now have for next-generation sequencing and highly sensitive PCR have opened up previously unavailable opportunity for rapid discovery of germline and/or somatic mutations that underlie the development of many vascular anomalies and for development of useful molecular clinical diagnostic tests.

We would like to thank our families and colleagues, of course, for patience and assistance as we and our partnering authors have tried to bring together key elements from an enormous amount of collective information in a rapidly evolving field, still plentiful of collegial controversy to be certain. On that note, we would also like to acknowledge the critical role of the International Society for the Study of Vascular Anomalies (ISSVA) and its global membership in bringing vascular anomalies into the light of international, multidisciplinary scrutiny and consensus for the past three decades. By bringing clinicians, radiologists, pathologists, and basic scientists together to talk and present science regularly, ISSVA has done a great service. Most of all, we would like to thank our patients and their families – they are the real inspiration and driving force behind this most difficult and perplexing of medical fields. Together, we are making progress.

Milwaukee, WI, USA

Paula North  
Tara Sander

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# Contributors

**David Bick** Pediatrics Department, Medical College of Wisconsin, Milwaukee, WI, USA

**Chang Zoon Chun** Department of Medicine, Division of Nephrology, Hypertension, & Renal Transplantation, University of Florida, Gainesville, FL, USA

**Beth A. Drolet** Dermatology Department, Division of Pediatric Dermatology, Medical College of Wisconsin, Milwaukee, WI, USA

**Kelly J. Duffy** Department of Radiology, Medical College of Wisconsin, Milwaukee, WI, USA

**Kristin E. Holland** Pediatric Dermatology–Administrative Research Offices, Medical College of Wisconsin, Milwaukee, WI, USA

**Michael E. Kelly** Pediatrics Department, Division of Pediatric Hematology, Oncology, Bone Marrow Transplantation, Children’s Hospital of Wisconsin, Milwaukee, WI, USA

**Paula E. North** Department of Pathology and Laboratory Medicine, Children’s Hospital of Wisconsin, Milwaukee, WI, USA

Department of Pathology, Division of Pediatric Pathology, Medical College of Wisconsin, Milwaukee, WI, USA

**Ramani Ramchandran** Departments of Pediatrics, and Obstetrics and Gynecology, Neonatology, Medical College of Wisconsin, Inc., Milwaukee, WI, USA

**Tara Sander** Department of Pathology and Laboratory Medicine, Children’s Hospital of Wisconsin, Milwaukee, WI, USA

Department of Pathology, Division of Pediatric Pathology, Medical College of Wisconsin, Milwaukee, WI, USA

**Rashmi Sood** Department of Pathology, Medical College of Wisconsin, Inc., Milwaukee, WI, USA

**Sara Szabo** Department of Pathology and Laboratory Medicine, Children's Hospital of Wisconsin, Milwaukee, WI, USA

Department of Pathology, Division of Pediatric Pathology, Medical College of Wisconsin, Milwaukee, WI, USA

**Monte S. Willis** Pathology & Laboratory Medicine; McAllister Heart Institute, University of North Carolina/UNC Hospitals, Chapel Hill, NC, USA