

Stem Cell Biology and Regenerative Medicine

Series Editor

Kursad Turksen, Ph.D.

kursadturksen@gmail.com

More information about this series at <http://www.springer.com/series/7896>

Martin K. Childers

Editor

Regenerative Medicine for Degenerative Muscle Diseases

 Humana Press

Editor

Martin K. Childers
Department of Rehabilitation Medicine
University of Washington
Seattle, WA, USA

Institute for Stem Cell and Regenerative
Medicine, School of Medicine
University of Washington
Seattle, WA, USA

ISSN 2196-8985 ISSN 2196-8993 (electronic)
Stem Cell Biology and Regenerative Medicine
ISBN 978-1-4939-3227-6 ISBN 978-1-4939-3228-3 (eBook)
DOI 10.1007/978-1-4939-3228-3

Library of Congress Control Number: 2015956772

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

This work is subject to copyright. All rights are reserved by the Publisher, whether the whole or part of the material is concerned, specifically the rights of translation, reprinting, reuse of illustrations, recitation, broadcasting, reproduction on microfilms or in any other physical way, and transmission or information storage and retrieval, electronic adaptation, computer software, or by similar or dissimilar methodology now known or hereafter developed.

The use of general descriptive names, registered names, trademarks, service marks, etc. in this publication does not imply, even in the absence of a specific statement, that such names are exempt from the relevant protective laws and regulations and therefore free for general use.

The publisher, the authors and the editors are safe to assume that the advice and information in this book are believed to be true and accurate at the date of publication. Neither the publisher nor the authors or the editors give a warranty, express or implied, with respect to the material contained herein or for any errors or omissions that may have been made.

Printed on acid-free paper

Humana Press is a brand of Springer
Springer Science+Business Media LLC New York is part of Springer Science+Business Media
(www.springer.com)

Preface

Regenerative medicine—an emerging multidisciplinary field composed of clinicians, bioengineers, pharmacologists, surgeons, and others—seeks to develop new ways of repairing and replacing cells, tissues, and organs. A great example of regenerative medicine technology was pioneered by Anthony Atala, MD, who pioneered the world’s first urinary bladder made from the patient’s own cells and grown in a laboratory before being implanted into the patient. This process of regenerative medicine differs radically from conventional medicine by not only treating symptoms but by recreating damaged tissues and organs in the human body. Unthinkable only a few years ago, clinicians and scientists are rapidly exploiting nature’s secrets to replace damaged tissue or to stimulate the body’s own natural repair mechanisms to heal previously irreparable tissues or organs. For degenerative muscle diseases, this is a tough order, since skeletal muscle comprises the largest tissue type in the human body. Not only is skeletal muscle the largest “organ” in the body, but most degenerative diseases, such as the muscular dystrophies, are caused by rare inherited genetic mutations. So in this case, regenerative technologies must overcome two very difficult problems. The first problem is to create enough “medicine” to treat the entire volume of skeletal muscle in a human body, and the second is to replace defective genes. Both problems may seem insurmountable at first glance, but the history of medical progress, particularly in the field of genetics and gene therapy, tells us otherwise.

In this book, the first three chapters are dedicated to an overarching view of how genetics and regenerative technologies can synergistically work together to target skeletal muscle genetic diseases. The next five chapters explore how an understanding of cellular biology (particularly stem cell biology and cellular reprogramming) proffers alternative methods to gene replacement. Chapters 8 and 9 examine new ways to look at our conventional approaches (rehabilitation medicine and nutrition) when combined with regenerative technologies. And the final chapters introduce the concepts of “micro-RNAs” and how fish and dog models can be used to enhance our understanding of these devastating and often fatal diseases of skeletal muscle.

Altogether, these new concepts, technologies, and approaches should give patients, families, and doctors cutting-edge information about incredible regenerative advances already under way in laboratories and clinics worldwide.

Seattle, WA, USA

Martin K. Childers

Contents

1	Regenerative Medicine Approaches to Degenerative Muscle Diseases	1
	Martin K. Childers and Zejing Wang	
2	An Overview of rAAV Vector Product Development for Gene Therapy	21
	Richard O. Snyder	
3	Gene Discovery in Congenital Myopathy	39
	Laura L. Smith, Vandana A. Gupta, and Alan H. Beggs	
4	Stem Cell Transplantation for Degenerative Muscle Diseases	85
	Berkcan Akpinar, Elizabeth C. Stahl, Aiping Lu, and Johnny Huard	
5	Spinal Cord Cellular Therapeutics Delivery: Device Design Considerations	109
	Khalid Medani, Jonathan Riley, Jason Lamanna, and Nicholas Boulis	
6	Patient-Derived Induced Pluripotent Stem Cells Provide a Regenerative Medicine Platform for Duchenne Muscular Dystrophy Heart Failure	129
	Xuan Guan, David Mack, and Martin K. Childers	
7	Overview of Chemistry, Manufacturing, and Controls (CMC) for Pluripotent Stem Cell-Based Therapies	157
	Amy Lynnette Deusen and Michael Earl McGary	
8	Regenerative Rehabilitation: Synergizing Regenerative Medicine Therapies with Rehabilitation for Improved Muscle Regeneration in Muscle Pathologies	205
	Kristen Stearns-Reider and Fabrisia Ambrosio	

9 Practical Nutrition Guidelines for Individuals with Duchenne Muscular Dystrophy..... 225
Zoe E. Davidson, Greg Rodden, Davi A.G. Mázala,
Cynthia Moore, Carol Papillon, Angela J. Hasemann,
Helen Truby, and Robert W. Grange

10 Identifying Therapies for Muscle Disease Using Zebrafish 281
Elizabeth U. Parker and Lisa Maves

11 miRNAs in Muscle Diseases 295
Diem-Hang Nguyen-Tran and Hannele Ruohola-Baker

12 Canine-Inherited Dystrophinopathies and Centronuclear Myopathies 309
Joe N. Kornegay and Martin K. Childers

Index..... 331

Contributors

Berkcan Akpınar Department of Orthopaedic Surgery, University of Pittsburgh School of Medicine, Pittsburgh, PA, USA

Fabrisia Ambrosio Department of Physical Medicine and Rehabilitation, University of Pittsburgh, Pittsburgh, PA, USA

McGowan Institute for Regenerative Medicine, University of Pittsburgh, Pittsburgh, PA, USA

Alan H. Beggs Division of Genetics and Genomics, The Manton Center for Orphan Disease Research, Boston Children's Hospital, Harvard Medical School, Boston, MA, USA

Nicholas Boulis Department of Neurosurgery, Emory University, Atlanta, GA, USA

Martin K. Childers Department of Rehabilitation Medicine, University of Washington, Seattle, WA, USA

Institute for Stem Cell and Regenerative Medicine, School of Medicine, University of Washington, Seattle, WA, USA

Zoe E. Davidson Nutrition and Dietetics, Monash University, Notting Hill, VIC, Australia

Amy Lynnette Van Deusen Regenerative Medicine Strategy Group, LLC, Los Angeles, CA, USA

Robert W. Grange Human Nutrition, Foods, and Exercise, Virginia Tech, Blacksburg, VA, USA

Xuan Guan Institute for Stem Cell and Regenerative Medicine, School of Medicine University of Washington, Seattle, WA, USA

Vandana A. Gupta Division of Genetics and Genomics, The Manton Center for Orphan Disease Research, Boston Children's Hospital, Harvard Medical School, Boston, MA, USA

Angela J. Hasemann School of Clinical Sciences, University of Virginia Children's Hospital, Charlottesville, VA, USA

Johnny Huard Department of Orthopaedic Surgery, University of Pittsburgh School of Medicine, Pittsburgh, PA, USA

Joe N. Kornegay Department of Veterinary Integrative Biosciences (Mail Stop 4458), College of Veterinary Medicine, Texas A&M University, College Station, TX, USA

Jason Lamanna Department of Neurosurgery, Emory University, Atlanta, GA, USA

Department of Biomedical Engineering, Georgia Institute of Technology, Emory University, Atlanta, GA, USA

Aiping Lu Department of Orthopaedic Surgery, University of Pittsburgh School of Medicine, Pittsburgh, PA, USA

David Mack Department of Rehabilitation Medicine, University of Washington, Seattle, WA, USA

Institute for Stem Cell and Regenerative Medicine, School of Medicine, University of Washington, Seattle, WA, USA

Lisa Maves Center for Developmental Biology and Regenerative Medicine, Seattle Children's Research Institute, Seattle, WA, USA

Department of Pediatrics, University of Washington, Seattle, WA, USA

Davi A.G. Mázala Department of Kinesiology, School of Public Health, University of Maryland College Park, College Park, MD, USA

Michael Earl McGary Regenerative Medicine Strategy Group, LLC, Los Angeles, CA, USA

Khalid Medani Department of Neurosurgery, Emory University, Atlanta, GA, USA

Cynthia Moore Nutrition Counselling Center, University of Virginia, Charlottesville, VA, USA

Diem-Hang Nguyen-Tran Departments of Biochemistry, Biology, Bioengineering, Genome Sciences, Institute for Stem Cell and Regenerative Medicine, University of Washington, School of Medicine, Seattle, WA, USA

Department of Neurosurgery, McKnight Brain Institute, University of Florida, Gainesville, FL, USA

Carol Papillon Human Nutrition, Foods and Exercise, Virginia Polytechnic Institute and State University, Blacksburg, VA, USA

Elizabeth U. Parker Center for Developmental Biology and Regenerative Medicine, Seattle Children's Research Institute, Seattle, WA, USA

Jonathan Riley Department of Neurosurgery, Emory University, Atlanta, GA, USA

Greg Rodden Human Nutrition, Foods, and Exercise, Virginia Tech, Blacksburg, VA, USA

Hannele Ruohola-Baker Departments of Biochemistry, Biology, Bioengineering, Genome Sciences, Institute for Stem Cell and Regenerative Medicine, University of Washington, School of Medicine, Seattle, WA, USA

Laura L. Smith Division of Genetics and Genomics, The Manton Center for Orphan Disease Research, Boston Children's Hospital, Harvard Medical School, Boston, MA, USA

Richard O. Snyder Department of Molecular Genetics and Microbiology, University of Florida, College of Medicine, Gainesville, FL, USA

Atlantic Gene Therapies, INSERM UMR 1089, Université de Nantes, CHU de Nantes, Nantes, France

Center of Excellence for Regenerative Health Biotechnology, University of Florida, Alachua, FL, USA

Elizabeth C. Stahl Department of Orthopaedic Surgery, University of Pittsburgh School of Medicine, Pittsburgh, PA, USA

Kristen Stearns-Reider Department of Integrative Biology and Physiology, University of California Los Angeles, Los Angeles, CA, USA

Helen Truby School of Clinical Sciences, Monash University, Melbourne, VIC, Australia

Zejing Wang Medicine/Clinical Research, University of Washington/Fred Hutchinson Cancer Research Center, Seattle, WA, USA

About the Editor

Martin K. (Casey) Childers, D.O., Ph.D. is a professor in the Department of Rehabilitation Medicine and Investigator at the Institute for Stem Cell & Regenerative Medicine, University of Washington. He is a graduate of Seattle Pacific University (B.A., Music Performance), Western University (D.O., Medicine, Osteopathic), and The University of Missouri (Ph.D., Physiology & Pharmacology; residency, Rehabilitation Medicine). The Childers' laboratory works in two areas of investigation. In a series of preclinical studies, they address the hurdles required for systemic gene replacement delivery for patients with X-Linked Myotubular Myopathy (XLMTM). In other studies, they use a “disease-in-a-dish” approach with induced pluripotent stem (iPS) cells to study heart disease in patients with Duchenne muscular dystrophy (DMD). Dr. Childers' clinical medicine practice at the University of Washington Medical Center is dedicated to serve patients with neuromuscular diseases.