Myopathology

A Practical Clinico-pathological Approach to Skeletal Muscle Biopsies

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This work is dedicated to my son Prahalad and the patients who have contributed to the subject.

Balan Louis Gaspar

Preface

This book covers most of the aspects of basic, essential, and recent advances and controversies in myopathology. Essential information is provided on anatomy, physiology, ultrastructure, and laboratory, and aspects in the initial chapters and the later chapters are devoted to core myopathology. Although the major content is focused on diagnostic myopathology, care is taken that relevant clinical information is not missed in any of the described individual disease entities. Special emphasis is given to rare topics such as biobanking and recent advances and controversies revolving around the rapidly progressing field of myopathology. Another special feature of this work is that it covers mostly the essential details that would be needed to establish a neuromuscular lab, and hence laboratory technical staff and research scholars would be equally benefited. The script consists of simple English (hence could easily be comprehended by the audience whose native language is not English) and is supplemented by high-quality photographs and full-color illustrations.

This book is meant for neuropathologists, histopathologists, neurologists (adult and pediatric), rheumatologists (adult and pediatric), and postgraduates of pathology, histopathology, neurology, rheumatology, internal medicine, and pediatrics. Research scholars and personnel in histotechnology in the field of neuromuscular diseases (including animal studies) would find this book an essential reading. We have taken extreme caution to make our naïve audience relish the subject of myopathology. At the same time, we have taken appropriate measures not to disappoint the experts in myopathology.

We hope our work benefits most of our readers, and we do expect a lot of feedback so as to improve our next edition.

Chandigarh, India — Balan Louis Gaspar
Chandigarh, India — Rakesh Kumar Vasishta
Chandigarh, India — Bishan Dass Radotra

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Balan Louis Gaspar

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Balan Louis Gaspar (MD Pathology, DM Histopathology) graduated from the Postgraduate Institute of Medical Education and Research (PGIMER), Chandigarh, India. He is a recipient of the prestigious "Prof. Subhash Kumari Gupta Memorial Gold Medal" in MD Pathology. He completed his 3-year DM training at the same institute from January 2014 to December 2016, thereby becoming the first candidate with a DM Histopathology certification in India. In addition, he established the state-of-the-art neuromuscular lab at the Department of Histopathology, PGIMER, Chandigarh, India. He has more than 30 publications in peer-reviewed national and international indexed journals to his credit. He is currently the Consultant-in-Chief of the Molecular Pathology, Neuromuscular Pathology, Nephropathology and Immunopathology divisions at G. Kuppuswamy Naidu Memorial Hospital, Coimbatore, India.

Rakesh Kumar Vasishta obtained his MD Pathology from the Postgraduate Institute of Medical Education and Research (PGIMER), Chandigarh, India, and completed his MRCPath and FRCPath at the Royal College of Pathologists, London, UK. Under his leadership, for the first time in India, the super-speciality DM in Histopathology course was introduced at PGIMER. He has more than 150 publications in peer-reviewed national and international indexed journals to his credit. He is currently the Professor and Head of Histopathology at PGIMER, Chandigarh, India.

Bishan Dass Radotra obtained his MD Pathology from the Govt. Medical College Rohtak, India. He completed his PhD at the Queen's University of Belfast, UK and has been a visiting scientist to the same institute. He has also been a Senior Research Fellow and Clinical Neuropathologist at the Imperial College and Hammersmith Hospital, London. He received a Commonwealth scholarship award in 1992 (UK), the Harold Miller Memorial Prize from the Irish Neurological Association in 1995, Dr. Aikat Memorial Oration at the 61st Annual conference of the IAPM, Jamnagar—December 2012 and the Dr. S. N. De Memorial Oration, West Bengal Chapter, IAPM, in September 2012. He has more than 250 publications in peer-reviewed national and international indexed journals to his credit. He is currently a Professor of Neuropathology at PGIMER, Chandigarh, India.

Introduction to Normal Skeletal Muscle: Anatomy, Physiology, Histology, and Ultrastructure

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Brief Overview

The word "muscle" is derived from the Latin word *musculus* meaning "little mouse." Skeletal muscle constitutes ~30–45% of the total body mass in an average adult and ~25% in a neonate. The skeletal muscle mass is influenced by various factors such as the genetic makeup of the individual, physical activity, nutrition, hormones, and associated comorbidities [1]. About 50–75% of the total body protein mass is made up of skeletal muscle, thereby making it the primary site for amino acid metabolism [2]. In human beings, rhabdomyogenesis starts at around 3 weeks of intrauterine life immediately following gastrulation initiated by the formation of the primitive streak [3]. The epiblasts migrate on either side of the primitive streak giving rise to paraxial mesoderm. The paraxial mesoderm develops further to form a pair of somitomeres [4]. Except for the first seven somitomeres, the rest undergo segmentation giving rise to block-shaped structures called somites. As the somites form, primitive streak simultaneously involutes. The entire sequence of events is referred to as somitogenesis [5].

Somitogenesis can be divided into four phases [6]:

1. Isochronicity of somites—somites are produced at periodic intervals.
2. Epithelialization—mesenchymal–epithelial transition (MET).
3. Specification—development of somites into different structures at different sites.
4. Differentiation—into sclerotome, dermatome, and myotome.

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