

Myopathology

A Practical Clinico-pathological
Approach to Skeletal Muscle
Biopsies

Balan Louis Gaspar
Rakesh Kumar Vasishta
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 Springer

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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
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Balan Louis Gaspar
Rakesh Kumar Vasishta
Bishan Dass Radotra

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In the name of God Almighty and with deep respect, an immense sense of gratitude and reverence that I thank my mentor Prof. Rakesh Kumar Vasishtha for optimal guidance that has been a great source of inspiration for me during my training period in myopathology. His words of encouragement, depth of knowledge, and simplicity acted as propelling force to enable me to bring out this work.

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I would like to thank especially Prof. Gayathri Narayanappa and Prof. Yasha T Chickabasaviah for allowing me to observe the muscle biopsy processing in the Department of Neuropathology, National Institute of Mental Health and Neurosciences (NIMHANS), Bengaluru, which helped me a lot in standardizing the neuromuscular lab in my institute.

The part played by my junior colleague Dr. Sumit Garg cannot be overemphasized, and he was with me day and night helping me out in this new journey which I thoroughly enjoyed and will remain in my memory till my last breath. I would also like to thank Dr. Charan Singh Rayat and Mr. Ishwar Negi for their valuable inputs in the technical aspects.

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I owe to my family for their patience, constant inspiration, and guidance they have shown throughout this tenure of hardship.

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Balan Louis Gaspar

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

1

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.