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Book Review

Book Review: Small Supernumerary Marker Chromosomes. Basics

Vladimir A. Trifonov [1, 2, ‡, *]

1. Research Department for Limnology, University of Innsbruck, Mondsee str. 95310 Mondsee, Austria; E-Mail: Vladimir.Trifonov@uibk.ac.at
2. Institute of Molecular and Cellular Biology SB RAS, Lavrentiev ave 8/2 630090 Novosibirsk, Russia

‡ Current Affiliation: Institute of Animal Physiology and Genetics CAS, Rumburská 89 27721 Liběchov, Czech Republic

* **Correspondence:** Vladimir A. Trifonov; E-Mail: Vladimir.Trifonov@uibk.ac.at

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Abstract

This review provides a critical assessment of the content and structure of the recently published book by Dr. Thomas Liehr, 'Small Supernumerary Marker Chromosomes: Basics.'

Keywords

Small supernumerary marker chromosomes; clinical diagnostics; chromosome aberrations

Typically, the number of chromosomes in a karyotype is regarded as a stable characteristic of a species in biology. Nevertheless, there are exceptions when the number of chromosomes changes due to chromosomal rearrangements or whole-genome duplications. These exceptions have garnered particular interest recently, especially with the advancement of the genomics era, as many structural variants remain largely unexplored but may harbor crucial genomic innovations. One noteworthy observation is the occurrence of supernumerary or extra chromosomes, in addition to the standard chromosome set, which may be associated with specific genetic syndromes or remain

neutral for carriers. When these structurally abnormal chromosomes are present in humans as an extra copy alongside the normal chromosome set, they are referred to as small supernumerary marker chromosomes (sSMCs). The first description of such elements in humans dates back to 1961 [1]. In the case of some other species, when genetically neutral chromosomes accumulate in the population, they are referred to as additional or B-chromosomes. The first discovery of B-chromosomes was over a century ago [2]. The origin, genetic activity, and evolution of such elements are not yet sufficiently studied, making them an interesting subject for both clinical geneticists and geneticists studying eukaryotic genomes.

Thomas Liehr's book, 'Small Supernumerary Marker Chromosomes: Basics' [3], delves into a variety of essential questions concerning the biological and clinical characteristics of sSMCs. Over several decades, the author's team has analyzed over 2500 corresponding clinical cases, employing a broad array of modern cytogenetic, molecular cytogenetic, and genomic approaches. Their extensive collection has enabled them to generalize existing data at a new level. Furthermore, detailed descriptions of all cases are available not only in specialized scientific literature but also on the website [4]. The book, partially based on this online resource, is very well-structured and easy to read. I would recommend it to both (i) individuals without a biological and medical background and (ii) specialists in human genetics. For clinical specialists, I would also recommend the earlier book by the same author, 'Small Supernumerary Marker Chromosomes (sSMC): A Guide for Human Geneticists and Clinicians' [5], which also includes family reports. Furthermore, families with a familial sSMC and/or those with someone affected by an sSMC-related syndrome may benefit from the current book. It provides insight into how sSMCs can form, their potential to cause clinical problems, their behavior over generations, and the current state of therapy options.

In the first section of the book, comprising seven chapters, the author provides general genetic definitions and terms necessary for understanding information in the subsequent sections. Additionally, the author offers a comprehensive definition of sSMC, explains their usual detection methods, and discusses the clinical syndromes with which they may be associated. Notably, chapters on how sSMCs can serve as markers of uniparental disomy (UPD), the importance of considering possible mosaicism for the presence of sSMC, and the characteristics of sSMC in tumor cells stand out.

The second section is devoted to the classification of sSMC described to date. While various classifications could be proposed, they would all consider the region(s), the mechanism of origin, as well as the structure and stability of these elements. This section might be too complex for beginners in genetics, but it holds crucial information for specialists dealing with unusual cases that require clinical decisions. Additionally, it is emphasized here that a gain of copy numbers is only deleterious if triplication-sensitive genes are involved.

The third section is equally crucial for clinical geneticists as it outlines the necessary steps for diagnosis and provides a list of questions to be addressed during the investigation. This section offers essential recommendations for the timely diagnosis of sSMC. The last chapter in this section also includes information for patients and their relatives, creating an opportunity to seek alternative information and obtain additional consultations.

The fourth section is particularly significant for professionals interested in advancing knowledge in the diagnosis and research of sSMC. It outlines potential directions in which these studies may develop shortly and how this knowledge can potentially be applied in other areas of biology.

I sincerely believe that reading this book can be highly beneficial for clinical cytogeneticists, as nearly all of them will encounter sSMCs in their practice sooner or later. Additionally, for ordinary biologists, the book can unveil a series of unusual phenomena in human genetics that are not widely discussed in genetic literature. For instance, few have heard about the self-correction of the embryo's genome or subgenomic clustering of parental chromosomes in the nucleus, and topics like chromothripsis or uniparental disomy are not common knowledge. These issues may spark the interest of geneticists from entirely different disciplines, encouraging them to investigate the presence of these phenomena in their respective subjects.

The overall style and narrative of the book are maintained at a moderate and comfortable pace. Very complex genetic terms are explained in simple language, facilitating an understanding of important genetic processes. I thoroughly enjoyed reading the book and cannot help but recommend it to my colleagues and interested laymen! I especially appreciate that the book is available in five languages-English, German, French, Portuguese, and Russian-with further languages. This will help spread knowledge about small supernumerary marker chromosomes across the world.

Author Contributions

The author did all the research work for this study.

Competing Interests

The author has declared that no competing interests exist.

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