

# Gene Regulatory Sequences and Human Disease



Nadav Ahituv

Editor

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 Springer

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# Preface

Advanced sequencing technologies can now allow us to obtain individual genomic sequences. These genomes contain an overwhelming amount of nucleotide variation per individual, with the majority of the variants being noncoding. However, our current ability to provide a functional interpretation to these noncoding variants is extremely lagging compared to protein coding sequences. This book provides seminal examples of how these noncoding variants and epigenetic changes can be associated with human disease by altering gene regulation. While the current number of examples is very limited, the methodologies and techniques described in this book can serve as a model for researchers to associate additional noncoding variants with human disease. In addition, future development of technologies that will enable to functionally characterize noncoding gene regulatory variants in a high-throughput manner will move this field forward and expand our knowledge of gene regulation. Combined, this will lead to a better understanding of the “gene regulatory code.” Other than allowing us to obtain a better diagnosis and understanding of the genetic causes of human disease, it will be of extreme importance to numerous other biological disciplines. Biologists have long been in need of defined sequences that drive precise patterns of expression. Such sequences can be used to express recombinant proteins or to overexpress various proteins in precise locations. These sequences could also be used to target specific molecules to certain tissues for gene therapy purposes. Gene regulatory elements are also important developmental regulators, and the understanding of the factors that regulate these developmental genes can increase our knowledge of development. In evolution, regulatory sequences are thought to be a major contributor to the evolution of form. An increased understanding of the “gene regulatory code” will vastly assist these and other disciplines.

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# Contents

<b>1 Gene Regulatory Elements</b> .....	1
Nadav Ahituv	
<b>2 The Hemoglobin Regulatory Regions</b> .....	19
Betty S. Pace and Levi H. Makala	
<b>3 Regulatory Polymorphisms and Osteoporosis</b> .....	41
Huilin Jin and Stuart H. Ralston	
<b>4 Gene Regulation in Van Buchem Disease</b> .....	55
Gabriela G. Loots	
<b>5 <i>Cis</i>-Regulatory Enhancer Mutations are a Cause of Human Limb Malformations</b> .....	73
Julia E. VanderMeer and Nadav Ahituv	
<b>6 Regulatory Mutations Leading to Cleft Lip and Palate</b> .....	95
Brian C. Schutte, Walid D. Fakhouri, and Daniel Zemke	
<b>7 <i>Cis</i>-Regulatory Disruption at the <i>SOX9</i> Locus as a Cause of Pierre Robin Sequence</b> .....	123
Christopher T. Gordon, Sabina Benko, Jeanne Amiel, and Stanislas Lyonnet	
<b>8 Regulatory Mutations in Human Hereditary Deafness</b> .....	137
Jonathan E. Bird and Thomas B. Friedman	
<b>9 The Contributions of <i>RET</i> Noncoding Variation to Hirschsprung Disease</b> .....	169
Zachary E. Stine and Andrew S. McCallion	

**10 *Cis-Regulatory Variation and Cancer* ..... 195**  
Nora F. Wasserman and Marcelo A. Nobrega

**11 *Cohesin and Human Diseases* ..... 217**  
Dongbin Xu and Ian D. Krantz

**12 *Epigenetics and Human Disease* ..... 253**  
Angeliki Magklara and Stavros Lomvardas

**Index ..... 281**



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