
Contents

<i>Preface</i>	<i>v</i>
<i>Contributors</i>	<i>ix</i>

PART I MUTATIONS

1 DNA Diagnostics and Exon Skipping	3
<i>Umasuthan Srirangalingam and Shern L. Chew</i>	
2 Bioinformatics and Mutations Leading to Exon Skipping	17
<i>F.O. Desmet and C. Bérout</i>	
3 Minigenes to Confirm Exon Skipping Mutations	37
<i>Lourdes R. Desviat, Belén Pérez, and Magdalena Ugarte</i>	
4 Analysis and Interpretation of RNA Splicing Alterations in Genes Involved in Genetic Disorders	49
<i>Maaïke P.G. Vreeswijk and Heleen M. van der Klift</i>	
5 Exon Skipping Mutations in Neurofibromatosis	65
<i>Emanuele Buratti and Diana Baralle</i>	

PART II TOOLS TO INDUCE EXON SKIPPING

6 Overview on Applications of Antisense-Mediated Exon Skipping	79
<i>Willeke M.C. van Roon-Mom and Annemieke Aartsma-Rus</i>	
7 Overview on DMD Exon Skipping	97
<i>Annemieke Aartsma-Rus</i>	
8 Overview on AON Design	117
<i>Annemieke Aartsma-Rus</i>	
9 Optimizing RNA/ENA Chimeric Antisense Oligonucleotides Using In Vitro Splicing	131
<i>Yasuhiro Takeshima, Mariko Yagi, and Masafumi Matsuo</i>	
10 Optimizing Antisense Oligonucleotides Using Phosphorodiamidate Morpholino Oligomers	143
<i>Linda J. Popplewell, Alberto Malerba, and George Dickson</i>	
11 Optimizing Splice-Switching Oligomer Sequences Using 2'-O-Methyl Phosphorothioate Chemistry	169
<i>Carl Adkin, Sue Fletcher, and Steve D. Wilton</i>	
12 Exon Skipping Quantification by Real-Time PCR	189
<i>Alessandra Ferlini and Paola Rimessi</i>	
13 Antisense-Mediated Exon Skipping to Shift Alternative Splicing to Treat Cancer	201
<i>Jing Wan</i>	

14	Antisense-Mediated Exon Skipping to Generate Soluble Receptors	209
	<i>A. Seda Yilmaz-Elis and J. Sjeff Verbeek</i>	
15	Antisense-Mediated Exon Skipping to Reframe Transcripts	221
	<i>Sandrina Turczynski, Matthias Titeux, Nathalie Pironon, and Alain Hovnanian</i>	
16	U1 snRNA as an Effective Vector for Stable Expression of Antisense Molecules and for the Inhibition of the Splicing Reaction	239
	<i>Julie Martone, Fernanda Gabriella De Angelis, and Irene Bozzoni</i>	
17	Engineering U7snRNA Gene to Reframe Transcripts	259
	<i>Aurélie Goyenville</i>	
18	Dynamic Fluorescent and Luminescent Reporters for Cell-Based Splicing Screens	273
	<i>Claude C. Warzecha, Ruben Hovhannisyan, and Russ P. Carstens</i>	
19	Antisense-Mediated Exon-Skipping to Induce Gene Knockdown	289
	<i>Petra Disterer and Bernard Khoo</i>	
20	Antisense-Mediated Exon Inclusion	307
	<i>Yimin Hua and Adrian R. Krainer</i>	
21	Antisense Genes to Induce Exon Inclusion	325
	<i>Rachel Nlend Nlend and Daniel Schümperli</i>	
22	Using Mini-genes to Identify Factors That Modulate Alternative Splicing	349
	<i>Robert Morse, Adrian G. Todd, and Philip J. Young</i>	

PART III DELIVERY

23	Overview of Alternative Oligonucleotide Chemistries for Exon Skipping.	365
	<i>Amer F. Saleh, Andrey A. Arzumanov, and Michael J. Gait</i>	
24	Identification of Peptides for Tissue-Specific Delivery	379
	<i>Hans Heemskerk</i>	
25	Systemic Delivery of Antisense Oligomer in Animal Models and Its Implications for Treating DMD.	393
	<i>Qi Long Lu and Bo Wu</i>	
26	Cell-Penetrating Peptides Enhance Systemic Delivery of Antisense Morpholino Oligomers	407
	<i>Hong M. Moulton</i>	
27	Optimizing Tissue-Specific Antisense Oligonucleotide–Peptide Conjugates.	415
	<i>Corinne A. Betts, Suzan M. Hammond, Hai-fang Yin, and Matthew J.A. Wood</i>	
	<i>Index</i>	437



<http://www.springer.com/978-1-61779-766-8>

Exon Skipping

Methods and Protocols

Aartsma-Rus, A. (Ed.)

2012, XI, 440 p. 70 illus., 8 illus. in color., Hardcover

ISBN: 978-1-61779-766-8

A product of Humana Press