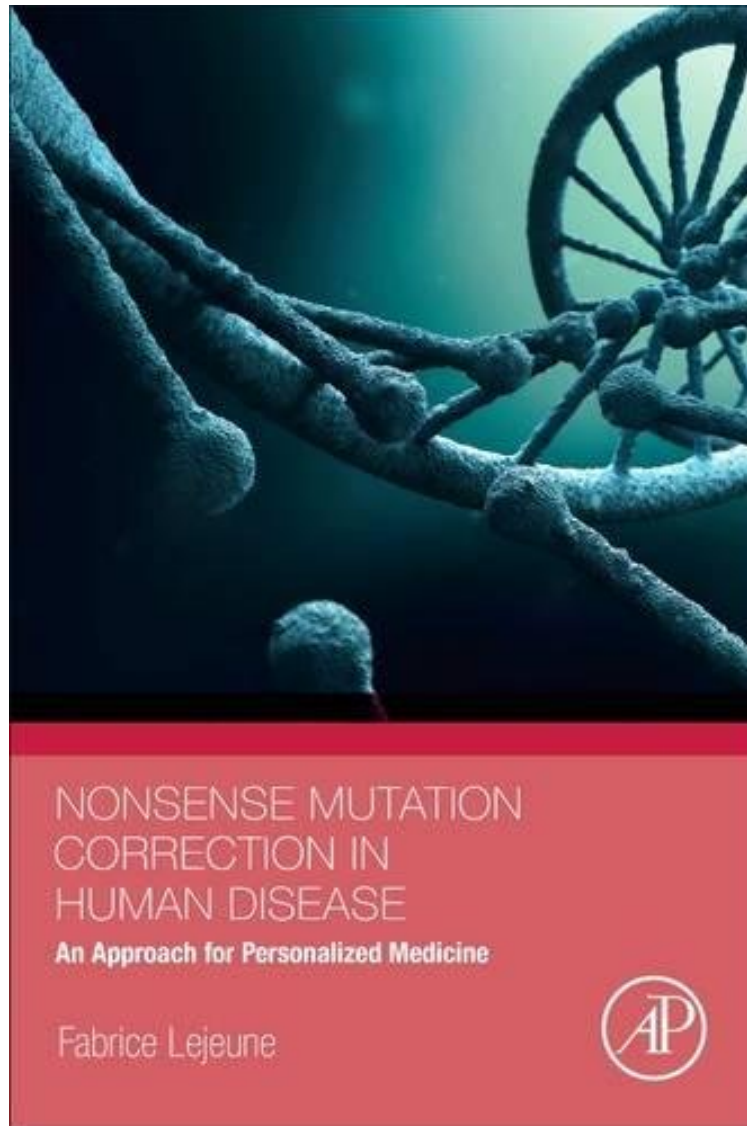


[Ebook pdf] Nonsense Mutation Correction in Human Diseases: An Approach for Targeted Medicine

# Nonsense Mutation Correction in Human Diseases: An Approach for Targeted Medicine

*Fabrice Lejeune, Hana Benhabiles, Jieshuang Jia*  
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#1464498 in Books Fabrice Lejeune Jieshuang Jia Hana Benhabiles 2016-03-28 2016-03-14Original language:EnglishPDF # 1 9.00 x .45 x 6.00l, .0 #File Name: 0128044683190 pagesNonsense Mutation Correction in Human Diseases An Approach for Targeted Medicine | File size: 65.Mb

**Fabrice Lejeune, Hana Benhabiles, Jieshuang Jia : Nonsense Mutation Correction in Human Diseases: An Approach for Targeted Medicine** before purchasing it in order to gage whether or not it would be worth my time, and all praised Nonsense Mutation Correction in Human Diseases: An Approach for Targeted Medicine:

Nonsense Mutation Correction in Human Diseases: An Approach for Targeted Medicine provides an introduction on genetic diseases, discusses the prevalence of nonsense mutations, the consequences of a nonsense mutation for the expression of the mutant gene, and the presentation of the nonsense-mediated mRNA decay (NMD). It presents the mechanism of action and rationale associated with each strategy to correct nonsense mutations with the results of clinical trials to further support this basis. In addition, the book shows how it may be possible to combine several of these strategies to ultimately improve the efficiency of correction, also suggesting the future goals and objectives to improve treatment modalities in this evolving sphere of personalized medicine. Features basic biological and clinical constructs that inform the application of genomic data to clinical decision-making. Includes theories and methods that can be used to link bio-molecular and clinical phenotypes so as to enable integrative hypothesis discovery, testing, and downstream evidence-based practice. Provides design patterns and use cases that contextualize the clinical decision-making and evidence-based practice relative to real world requirements and stakeholders.

"Not only is the content well selected and planned, but the language is clear and simple and the illustrations are magnificent. This book is unique - there are no comparable books. The authors have achieved a tremendous goal. Score: 100 - 5 Stars!" --Doody's, Nonsense Mutation Correction in Human Diseases

**About the Author**  
Dr. Fabrice Lejeune studies the mechanisms that regulate or deregulate the quality control of protein synthesis with the aim to one day utilize these mechanisms to chemically treat diseases such as cystic fibrosis, muscular dystrophy and cancer. He studied under Dr. Lynne Maquat, Director, Center for RNA Biology, and the scientist responsible for discovering the control mechanism messenger RNA carrying nonsense mutations in mammals. Dr. Lejeune continues this work at his lab at the Pasteur Institute.

Hana is a PhD student working with Dr. Fabrice Lejeune. She obtained her Masters degree in Genetics, in 2014, at the University of Lille in France. Prior coming to Lille, Hana did a biological engineering degree at Boumerdes University in Algeria. She acquired biomedical and biotechnological knowledge that she applies during her PhD. By coupling her engineering and research training, Hana is currently identifying new drugs correcting a nonsense mutation with a focus on their application in clinics to promote the expression of normally unexpressed proteins in these types of pathologies.

Jieshuang Jia is a postdoc fellow studying NMD at the institute of biology of Lille (France) since June 2015. Prior to her postdoc, she achieved her PhD at the University of Lille 2 by studying molecules with nonsense mutation correction capacity. She also got a bachelor of clinical medicine in the Second Military Medical University in July 2004 and a master of internal medicine in the Nephrology Institute of Changzheng Hospital in Shanghai, (China) with a specialty in Nephrology in June 2007. She has won several first class awards and has been the merit student for several times. She has worked in clinic for more than four years and received the advanced hospital worker status. During her clinic time from August 2007 to October 2011, she directed the students internships and taught them the diagnosis and treatment of diseases. She also performed some clinical studies on diseases such as polycystic kidney disease, chronic kidney disease with some relation with nonsense mutations until 2011. Her interest focused on new diagnostic methods and therapies of diseases. Now she is studying on NMD and would like to promote a strong interaction between nonsense mutation correction and clinic diseases.