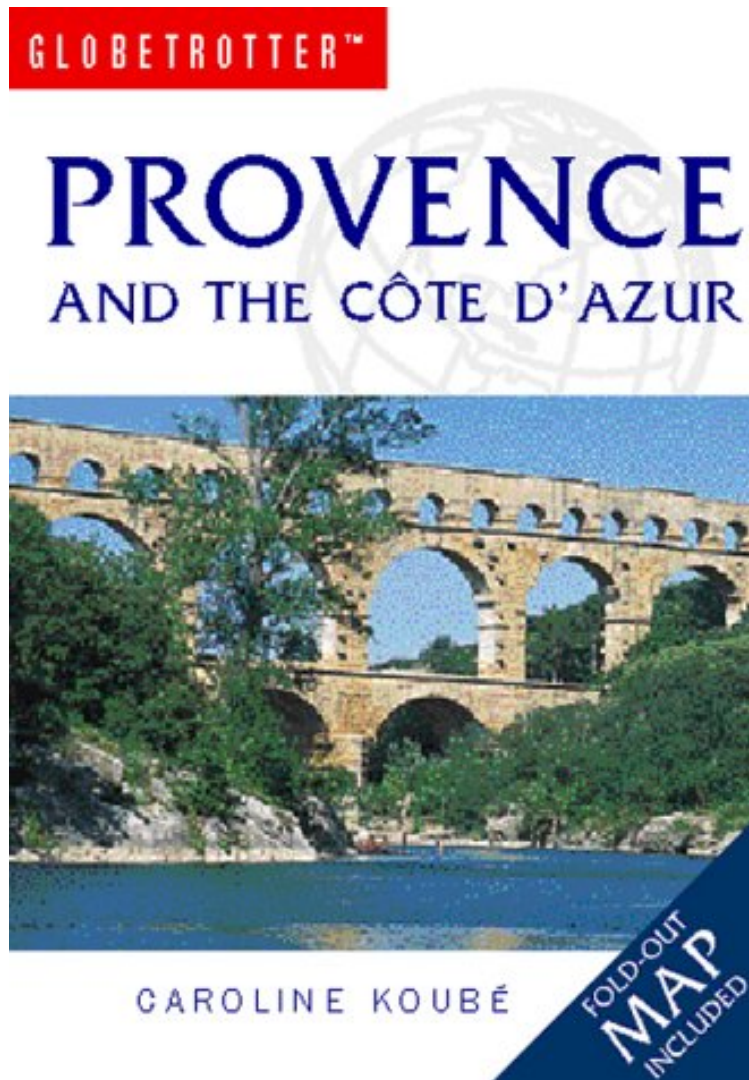


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Human Gene Mutation is a reading pleasure and will be equally helpful to the novice and the experienced. I highly recommend the book.--Professor Stylianos E. Antonarakis, Center for Medical Genetics, Johns Hopkins University School of Medicine, USA

From the Back Cover
Enormous progress has recently been made in the analysis and diagnosis of human inherited disease and in the characterization of the underlying genes and their associated pathological lesions. Human Gene Mutation brings together for the first time a short history of mutation research, a comprehensive analysis of the nature of human gene mutation (single base-pair substitutions, deletions, splice site, promoter mutations, etc.), and an assessment of the consequences of mutation both at the level of gene expression and for the complex relationship between genotype and clinical phenotype. Laboratory methods for mutation detection and analysis and current possibilities in molecular diagnostic medicine are also reviewed. The central theme of this volume is the non-randomness of human gene mutation and its implications for both the nature and prevalence of genetic disease. The cognition that certain DNA sequences are hypermutable provides vital new clues as to the basic (and endogenous) mechanisms of mutagenesis. Novel explanatory and predictive models of mutagenesis are proposed. Together this constitutes a unique approach to understanding the ultimate causes of human genome pathology. This volume in the Human Molecular Genetics series is an important companion of all human molecular geneticists and genetic counsellors as well as interested clinicians and postgraduates.