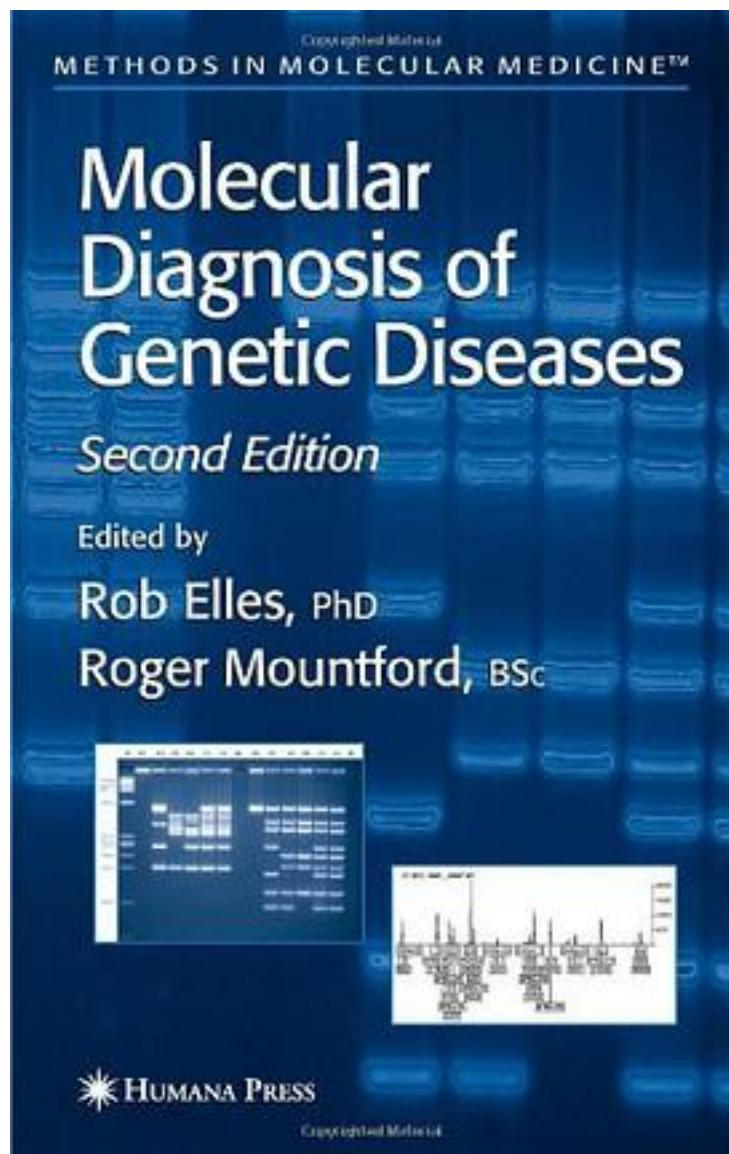


Molecular Diagnosis of Genetic Diseases



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This completely revised and updated second edition to integrates the many new technologies and insights now available for the diagnosis of genetic diseases. The authors use such methodologies as PCR optimization dosage analysis, mutation scanning, and quantitative fluorescent PCR for aneuploidy analysis, Neurofibromatosis type 1, and Duchenne muscular dystrophy. These largely generic methodologies may be adapted to most genetic conditions for which a molecular diagnosis is relevant, no matter how frequent or rare their incidence. Molecular Diagnosis of Genetic Diseases, Second Edition offers diagnostic molecular geneticists a unique opportunity to sharpen their scientific skills in the design of assays, their execution, and their interpretation.

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