
Preface

The first edition of *Molecular Diagnosis of Genetic Diseases* was published in 1996 and was organized to present a series of key methodologies in the context of the diagnosis of particular genetic conditions. It constituted a handbook covering the analysis of the most common inherited conditions.

This second edition revisits and updates some of these areas and is organized around the diagnosis of a range of specific inherited conditions. It includes both commoner and rarer genetic conditions and though being firmly aimed at the service diagnostic laboratory, is also relevant to related research areas and to clinicians and science students with an interest in diagnostics.

Since 1996, technologies have moved on, but the revolution promised through the use of hybridization arrays or other parallel processing techniques has yet to emerge as a reality for most diagnostic centers. This perhaps reflects the need in clinical areas for well-characterized and robust technologies.

In 2001, the working draft of the human genome sequence was announced. Mining this resource has resulted in a growing number of gene targets associated with clinical conditions. No simple literature source can hope to detail the thousands of potential diagnostic genetic tests. However, we can hope that the future will bring a simplification of the problem faced by diagnostics; perhaps one or two combinations of analytical chemistry and generic instrumentation will be sufficient for most demands. Until then genetic diagnostics remains based to a large extent on individual scientific skills in the design of assays, their execution, and their interpretation—hence this book.

A problem for the diagnostic sector as a whole is to meet the demands posed by the huge diversity of applications made possible through the human genome project while meeting the need for timely, accurate, and reliable services, including tests.

These challenges are being faced by scientists, regulators, and planners in all health systems and the balance in service provision between specialization and diversity, centralization and dispersal, public and private sector approaches is yet to be decided.

This large number of possible tests poses another challenge—to public health planners, the insurers, and the public funding agencies for health care. Is every genetic test relevant and a best use of resources? How can we prioritize and approve genetic tests for introduction into service? To date these decisions have been made on an ad hoc basis, but service providers and planners increasingly

need to reach a consensus to develop coherent, evidence-based, and defensible policy in these areas.

Taking inspiration from the huge success of the international genome sequencing efforts, one solution increasingly being discussed for genetic service provision, especially for specialized or “orphan” areas is risk-sharing through collaborative national and even international networks of diagnostic centers.

It is already clear that many genetic referrals cross international frontiers. A level playing field needs to be created which service providers and consumers have mutual confidence in the quality and ethical standards in force and where reimbursement is regularized and straightforward. Under these conditions an international network of genetic diagnostic centers could permit comprehensive access to diagnostics. It could help free patients facing barriers to access to services at national frontiers or simply owing to their misfortune to be at risk of a particularly rare genetic condition.

A successful international collaboration in genetic health care is a prize worth working for.

Rob Elles, PhD
Roger Mountford, BSc



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Elles, R. (Ed.)

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