

Genetic Testing: Policy Issues for the New Millennium

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The advent of molecular genetic testing in clinical laboratories has led to the introduction of a bewildering array of tests for many hundreds of genetic disorders. With the completion of the human genome project, the number is expected to increase significantly since one of the first applications of knowledge about the molecular basis of a particular disorder is the ability to develop a clinical diagnostic test.

Computational advances and genomic technologies that enable rapid parallel testing of many genetic markers and mutations will further stimulate the growth of this sector. This progress is expected to lead to major changes in the infrastructure, performance and delivery of genetic services and, as a consequence, is challenging the limits of current services and the related ethical and regulatory frameworks.

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Evaluations of the provision of genetic services in various European countries in 1997 by the Concerted Action on Genetic Services in Europe (CAGSE) [1] and in 1998 at an International Symposium sponsored by the European Commission, pointed to the need for guidelines and internationally agreed principles [2]. In the United States the 1997 report by the Task Force on Genetic Testing [3] and results of a survey done under the auspices of US-CDC to assess quality assurance in molecular genetic testing laboratories [4] underscored similar concerns.

It is against this background that the OECD was invited to 'consider whether the approaches of member countries for dealing with new genetic tests are appropriate and mutually compatible'. The United Kingdom and Austria heeded this call and with the support of the European Commission, hosted the OECD workshop 'Genetic Testing: policy Issues for the new millennium', held in Vienna 23–25 February 2000.

Among the many issues that surround genetic testing and were set forth by the OECD Steering Committee, which was charged with the organisation of the workshop, four broad questions were retained and discussed extensively: (1) What are the likely consequences of a rapid expansion of genetic testing? (2) What policies are needed to safeguard quality and equitable access? (3) What are current incentives and barriers to diffusion of novel genet-

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ic tests? (4) What are the major ethical, legal and social issues?

The twelve papers in this special issue are representative of the presentations and expert opinion offered at the workshop in response to these questions.

The papers by Patterson et al. and by Kristoffersson review the current challenges in safeguarding quality of genetic tests and services. While many OECD countries have established adequate oversight or legislative authority for diagnostic laboratory practices, this oversight is not in fact being implemented for genetic testing. Errors are therefore more common than is generally thought [4, 5]. To remedy this situation, most OECD countries have developed a number of external quality assessment schemes and professional guidelines in the last five years and dedicated regional networks, such as the European Molecular Genetics Quality Network. There are, nonetheless, significant differences among OECD approaches to proficiency testing and QA (for a review of national approaches see OECD web site [6]), which, in the background of increasing trends for tests and services to cross national boundaries point to the need for international standards.

The quality of the counselling process is highly relevant to the performance of a genetic test, particularly in the case of molecular genetic testing. This because, as the papers by Frontali and Jacopini, and Blancquaert validly argue, while the new knowledge from genetic testing gives patients hope, it can also create problems and fears if it is not applied appropriately and effectively. The major concern for patients is what the information gained from testing will mean for them. Yet, owing to the growing number of tests available (for both rare and common disorders), and the development of easier and cheaper molecular techniques, the demand for genetic testing is likely to exceed the supply of counsellors and genetic services. In addition, as Modell and Darlison underscore in their paper, as our ability to test grows, the information technology needs of genetic services also increase. Health workers will require continuing updating in genetics and ready access to information on genetic mutations and related materials designed both for them and for their patients. To meet these challenges, the three papers advocate an enhancement of current counselling services, genetic training and public information, including the development of compatible electronic information systems in genetics.

Genetic tests for hereditary cancer susceptibility are a striking example of how the interpretation of test results is not simple and how counselling is a most essential ele-

ment of testing, particularly in light of the limited therapeutic options. The papers by Grann and Frank review the factors that may influence the decision to test in the context of breast cancer susceptibility. Among the questions they ask are: What is the efficacy of genetic testing and preventive treatment for women who test positive for BRCA1 and BRCA2? Who should be tested?

As health care systems embrace population-based medicine and genetic epidemiology, genetic testing is, however, expanding beyond its most traditional use and definition. Genomic knowledge is being applied to develop tools for patient segmentation based on genetic profiles and for the elucidation of individual disease risk profiles to optimize prevention, diagnosis, and drug use. This approach, as Middleton et al. forecast, holds also the promise of improved and rational drug R&D approaches. It does, however, entail the development of new sophisticated informatic tools, and the assembly of population-based databanks for genetic association studies, ultimately to establish what has been called 'cradle-to-grave' individual electronic medical records [7].

The policy implications, as reviewed in the papers by Blatt, Knoppers and Zweig, are staggering. Whether samples are identified, coded or anonymous, banking of DNA and of genetic information raises serious issues pertaining to access, informed consent, privacy and confidentiality of genetic information, civil liberties, patenting and proprietary rights. A key concern regarding stored genetic information is whether unauthorised third parties could gain access to or view genetic profiles or results of simple DNA tests. A related concern is whether samples or data are used for purposes different from those for which they were originally collected. This makes it all the more important to link the performance of genetic tests to stringent data protection requirements. There are strong economic pressures for expanding the use of genetic tests. Yet, genetic tests have a cost and health budgets cannot expand indefinitely [8]. This raises crucial issues related to access to genetic tests. Should access be limited? If so, how and under what conditions? More generally, what should be the goal of regulating this area? These advances if not adequately regulated, will also and inevitably find their way into the courts of law.

The critical issue, as Guillod's paper highlights, is ultimately how and to what extent to regulate genetic testing. The future of genetic testing, health-care sector databases and of post-genomics will depend critically on good regulations. Quality assurance, privacy and encryption policies need to be trustworthy if they are to generate confidence in the public. One of the greatest challenges of regu-

lating genetic testing is, however, to find the appropriate mix of normative instruments to achieve convincing and effective but flexible regulation. Middleton et al., indeed warn that regulations should be carefully tailored to the various applications of genetic testing as not to stifle promising post-genomic advances, which are transforming pharmaceutical industry R&D, medical practice and public health.

The present situation is, however, rather chaotic: in addition to the laws passed in many countries, there are, in virtually every country, a long list of professional, ethical and other guidelines, recommendations and policy papers [6].

Thus, the overwhelming conclusion from the workshop, adequately reflected in this selection of papers and further documented in the recent OECD policy report on genetic testing [9] is that international action is urgently needed to address five areas: (1) development of internationally recognised and mutually compatible best-practice policies for analytical and clinical validation of genetic tests, including quality assurance and accreditation of genetic services; (2) exploration of strategies to enhance current counselling services, genetic training and public information (including the development of compatible electronic information systems in genetics), particularly as a means to provide the individual with accurate infor-

mation and to protect his/her autonomy; (3) examination of possible impacts of restrictive licensing practices; (4) guidance on how existing privacy, security and cryptography guidelines can apply in the context of genetic testing, particularly to ensure adequate security of genetic databases, and (5) exploration of broader implications of developments in pharmaco-genetics and high-throughput parallel processing.

This workshop on genetic testing is part of a broader programme of work within the OECD on issues regarding biotechnology, genomics, and human health. Since the early 1990s, the OECD Biotechnology Unit has been reviewing and analysing the safety and socio-economic implications of developments in biotechnology for health, such as live viral vaccines, gene therapy and gene delivery systems, transgenic technology, new molecular diagnostics and xenotransplantation, under the guidance and close co-operation with expert delegates to its Working Group on Human Health Biotechnologies.

In addressing these areas and in recognition of the global importance of these issues, the OECD will continue to strengthen co-operation with other international organisations particularly the WHO, as highlighted in a Framework for Co-operation signed by the Directors of the two organisations on 16 December 1999.

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