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BOOK REVIEW

Impact of Genomics on Health Care

Edited by G Poste, J Bell, K Davies, P Goodfellow, N Hastie
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With an introduction by Peter Goodfellow, this issue of expert reviews is a significant contribution to the understanding of the achievements, limitations and future possibilities of genome research in relation to health care. In 12 chapters, a series of outstanding authors review different aspects of what the impact of this new knowledge and improved technology will be in the coming years. In addition, the contributions of T Marteau on communicating genetic risk information, S Thomas on the implications for ethics and education, and R Fears, D Weatherall and G Poste on medical education and training place the review in a much broader context than pure technological considerations. The last chapter by K Lindpaintner is an original review of the issues and problems surrounding individualised treatment in common complex diseases.
The fact that the authors, all established scientists, are drawn both from academia and industry, gives the reviews a dimension which could probably not have been achieved if they all had a common background.

In the first chapter a critical review is given of available and forthcoming technologies. As expected, the emphasis is on high throughput, high resolution systems such as DNA chips and microarrays as well as mass spectroscopy. These reviews are comprehensive and give the reader a clear picture of the state of the art. The second chapter discusses the applications of the different technologies in diagnosis, presymptomatic testing and prenatal diagnosis in all its aspects. The increasing need for high throughput methods is considered to be one of the major challenges facing DNA diagnostic laboratories. The need for quality assessment and of high quality laboratory services is stressed, but the subject is not fully developed and viewed mainly from a UK perspective. C Matthew suggests that 'integration of chemical and laboratory services within one department or centre' is a 'very effective model for the provision of genetic services', but he suggests this may be wishful thinking, given the reality of different situations in many countries. The last sentence 'ensuring that the new information is applied appropriately for patient care' is unquestionably the greatest challenge for geneticists in the next decade, but the reasons why this is so are actually developed in the chapter on communicating genetic information. In carefully chosen terms, T Marteau argues that predicting responses to genetic information is not as straightforward as one might hope. The perception of risks and the adherence to prescribed medication, to give just two examples, are complex and are dramatically influenced by the expertise of the counselling team and the quality of the information given.

The chapters on ethics and education and on medical education and training summarise in a very clear and concise way what the ethical and educational challenges or consequences of genomics are. Both chapters arrive at the obvious conclusion that education of the public, starting with the school core curriculum, and of the medical and paramedical profession, in particular at primary health care level, will be necessary. In addition, 'New concordats might be developed to include patients, research charities and industry, and public awareness of the benefits of clinical research must be promoted'. Indeed, public perception of what is happening in genetic research, and the declining resources allocated to clinical research and training, could lead to a paradox in which a better informed public will have to interact with a decreasing number of expert professionals, who would become completely disconnected from the necessary research and development.

In addition to issues pertaining strictly to the human genome, two chapters discuss topics of related interest: the pathogen virulence genes, and the infectious disease susceptibility. The first of these chapters illustrates the contribution of bacterial genome research in the development of new drugs and vaccines in an attempt to regain a certain control over antibiotic-resistant bacteria. The second gives an overview of our still limited understanding of the genes that contribute to susceptibility and resistance to infectious diseases. The hope here is also that as more molecules and pathways are unravelled, new targets for pharmacological intervention will become available. The presence of a chapter on xenotransplantation, in the absence of a chapter on gene therapy or other genome-based therapeutic strategies is somewhat surprising. Nevertheless, P Morris gives a clear overview of where we stand, in particular in the removal of hyperacute rejection triggered by α-1,3-galactose antigen. His conclusion that 'there should be no illusions about the obstacles to be overcome' is a sobering note in the context of the technological achievements of genome research.

The pharmacogenetic polymorphisms within the cytochrome P450 family are the best characterised. Other polymorphisms are being evaluated and 'the ability to routinely use genetically-based methods to predict individual response to drug treatment is now becoming a realistic goal'. C Wolf and G Smith illustrate what can already be done by using the CYP2D6 polymorphisms. They also state that 'an increasing number of pharmaceutical companies are genotyping their clinical trial populations'. While this statement is intended

to draw our attention to the increasing importance of this information for drug registration, it brings us also to the complex ethical issues of informed consent, DNA storage and access to databases, treated in further chapters.

The collection of appropriate samples with correct clinical information is also a major issue in drug discovery. K Lindpaintner gives us a remarkable review of the power of genomic information to develop new drugs. At the same time he gives a very clear overview of the challenges and pitfalls of this research, in particular how difficult it would be to identify genes that have only a limited contribution to the development of a common complex disease. The author proposes short and long term strategies to tackle the problem of identifying modest signals in a multitude of sources of noise, including the use of inbred populations, the screening of SNPs and their variants for drug targets and the use of pharmacogenetics to distinguish effects on metabolism and drug activity. In his ethical considerations, the author attempts to demystify genetic information and to bring it back to the level of all medical information. In his view 'not so much the privacy of medical information, but the way it is used' is important. It is clear that for the author and for the pharmaceutical industry in general, the protective attitude of the geneticist over genetic information is considered to be an obstacle to progress in drug discovery. 'The creation of the knowledge base required to carry out profiling for genetic risk and the acceptance of these new approaches among the general public and the patients' are the two major 'hurdles to be overcome if we want to realize the new potential of the pharmaceutical identity in our lifetime.' Industry has identified the problems very clearly but whether they will find the correct approach to solving these remains to be seen.

In conclusion, this edition of the expert reviews should be digestible for geneticists and non-geneticists alike. The information contained in this booklet should greatly facilitate serious discussions and exchanges between the different parties involved: the public, the professionals and industry. Hopefully this will bring a consensus closer on how and when this technology and this knowledge will be applied to the benefit of our society.

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