

Margaretha van Mourik

H. Skirton, C. Patch: Genetics for Healthcare Professionals: A lifestyle approach

Bios Scientific Publishers, 2002 (ISBN 1-85996-043X) softcover, £22.99

Received: 28 April 2003 / Published online: 26 July 2003

© Springer-Verlag 2003

We live in the era of the „new genetics“. New discoveries rapidly explored by the media fuel hopes, expectations and confusion for an increasing section of the population. The application of new technologies holds the potential for benefit, but also for possible harm. The quagmire of the World Wide Web may have an adverse or converse influence. Therefore, many people look towards health professionals to elucidate them. Health professionals in turn may be unsure on how to explain the possibilities and consequences opened up by these new technologies and how to promote accessibility to genetic services. There is, therefore, an increasing need for educational material that not only presents genetic information but also shows the relevance to practice for health professions. This book helps to fill that gap. It looks at many aspects of genetic conditions but remains concise and pragmatic, thereby preventing the newcomer to this field from drowning in information and concepts. It is also a useful book to dip into for medical, scientific and genetic counselling colleagues who may be interested in exploring counselling models, a holistic family-orientated approach or to refresh their memory on risk calculations.

The authors are both highly experienced genetic nurse counsellors, who derived their knowledge and expertise not merely from academic studies but from day to day, year in year out, clinical work with families. Their educational interest and expertise helped to develop the logical and original layout of the book.

The first chapter is an introduction to clinical genetics, the types of genetic testing and psychosocial, ethical and insurance implications. In this chapter six families are introduced. They are used throughout the book to demonstrate the clinical application of the particular topics under discussion and enable exploration of clinical situations.

The following chapters explore the importance of the family history and genetic counselling issues. The authors have tackled the „basic concepts in genetic science“ head on. They succeeded in making the information accessible by breaking it up in clear sections with appropriate illustrations. This chapter is an excellent and very necessary preparation for the „flesh on the bone“ of this book, where the authors have organised chapters relating to particular life stages from preconception to adulthood.

The pre-conception chapter explores factors that may affect the fetus, such as maternal age and the role of folic acid in lowering the risk of neural tube defects. It also looks into genetic conditions affecting the mother. It explodes some traditional superstitions attached to cousin marriages and emphasises the importance of the preparation of couples for prenatal diagnosis where there is a family history of a genetic condition.

Midwives would find the checklist of genetic conditions useful when booking a woman for antenatal care, a time fraught with the sheer volume of information that needs to be discussed. Maternal genetic conditions may alter the management of the pregnancy and particular emphasis was given the importance of jointly managed care between the obstetric team experienced in the care of women with high risk pregnancies, and physicians experienced in the care of patients with a particular condition. This is a good chapter and it is, therefore, regrettable that the importance of professionals allied to medicine, such as dieticians and physiotherapists, were not mentioned in the context of shared care. Nevertheless antenatal screening and prenatal testing were clearly set out and I liked the explanation of exclusion testing in the case study.

The boxed „key practice points“ in this book are extremely clear and useful. For instance, colleagues in cytogenetics would be pleased to note that a practical point highlighted the fact never to place a skin biopsy from a stillborn child in formalin but in tissue culture medium, and why a speedy delivery is important.

Other health professionals in turn would welcome the clear definition of the categories and patterns of birth defects and the guidance on breaking bad news. Neonatal

M. van Mourik (✉)
The West of Scotland Regional Genetic Service,
Yorkhill Hospitals, Glasgow, G3 8SJ, Great Britain
Tel.: +44-141-2010365, Fax: +44-141-3574277,
e-mail: mvanmourik@yorkhill.scot.nhs.uk

screening was covered and included elucidating key practice points. As well as an exploration of dysmorphology, the reader is asked to consider how the health service should address the impact on the social and educational development in children and adolescents.

The last chapter of the life-stages approach looks at the adult onset conditions, which are divided into four groups: familial cancers, neuro-muscular disorders, psychiatric disorders and genetic haemochromatosis.

The section on cancer genetic is clear and concise, with guidelines for referral to the genetic service, screening protocols, and the psychological implications of a risk status. I was delighted to see the inclusion of haemochromatosis, because, in spite of the high carrier rate in the northern European population, few health professionals are aware of this treatable adult onset disorder. It also gave the authors the opportunity to touch on the arguments of cascade versus population screening.

The concluding chapter embraces the development of genetic services and the genetic counselling profession.

This book has great strengths. It is well referenced and eleven pages of website addresses include descriptions of the sites and whether they are useful for the professional and/or the family. Because of the key practice points, the case-study approach, exploration of ethical dilemmas, a

good glossary and review questions, it would be very useful as the main text in midwifery and nursing courses, and a concise introduction to clinical genetics for medical and genetic counselling students. It would also benefit a wide variety of nurses and other health professionals outside of the genetic community.

If there are weaknesses, they are minor. The pre-conception chapter would have gained if the topic of pre-implantation genetic diagnosis had been included. The authors touched on the difficult issues posed by the genetic influence on psychiatric conditions – a subject not well understood. The inclusion of a case study, exploring the difficulties experienced during a genetic counselling session when there is no clear pattern of inheritance, would have been useful.

My final very small personal niggle is the American spelling used throughout. As this book has a strong British/European flavour, it seemed at times intrusively inappropriate. I also noticed that a few useful UK website addresses were left out in favour of some transatlantic ones.

Apart from this, it is an excellent text, which clearly illustrates the human side of clinical genetics with the „family“ firmly identified as the pivotal point of the medical genetic service.

Copyright of Human Genetics is the property of Kluwer Academic Publishing / Academic and its content may not be copied or emailed to multiple sites or posted to a listserv without the copyright holder's express written permission. However, users may print, download, or email articles for individual use.