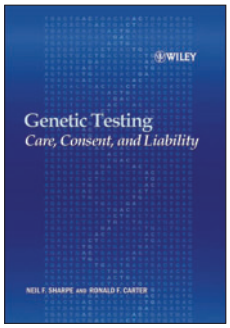
When the past isn't prologue–it's the whole book



Genetic Testing: Care, Consent and Liability

Edited by Neil F Sharpe & Ronald F Carter

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Reviewed by Kathy Hudson

Contrary to media hype, the relevance of genetics to medicine did not begin with the Human Genome Project. The fields of reproductive and pediatric medicine have long relied heavily on genetic testing. Prenatal diagnosis has given prospective parents some, albeit limited, options: parents could be relieved to know that their child would not have a specific condition; they could prepare for the birth of an affected child; or they could terminate the pregnancy. In short, patients and health care professionals have been grappling with what to 'do' with genetic test results for a very long time. But today, genetics has expanded well beyond the reproductive context.

And there is another set of issues that separates the early days of genetic testing from today's reality, let alone the promise of the future. Historically, genetic tests could diagnose or predict disease, but there were few effective interventions to reduce that risk or slow disease progression. Without clear relevance to clinical management, patient-focused decision-making and nondirectiveness—the ethical constraint on medical professionals to use genetic tests only to inform, not direct, patient care—remained the norm. This therapeutic gap is narrowing, though much more slowly than many would have hoped.

There are too few medical geneticists and genetic counselors to meet the projected need for clinical genetic testing services, and primary health care providers remain largely ill equipped to offer knowledgeable advice about the need for and meaning of genetic tests. The time certainly is right—even overdue—for a comprehensive review of genetic testing for health care professionals, a review that addresses current and emerging challenges.

Genetic Testing: Care, Consent and Liability, edited by Neil Sharpe and Ronald Carter, could have been such a review, but it isn't. Things are moving fast in genetic testing, and the authors pay far too much attention to what is now the distant past. For example, the chapters on counseling, communication, psychological aspects and informed consent are written almost exclusively in the context of reproductive genetic testing and fail to clearly distinguish between the very different moral and medical circumstances presented by different types of tests and different contexts for testing. Prenatal testing for Tay-Sachs disease presents very different challenges and options for the clinician and patient than, for instance, testing for variants that inform drug dosing and treatment choice. And contemporary concepts such as pharmacogenetics and personalized medicine are hardly mentioned in this book.

The book is divided into 15 chapters, each with a lengthy section by Sharpe and Carter followed by one or more sections on specific topics by notable experts. While books with multiple authors frequently suffer from some redundancy and disjointedness, this book is striking in that much of the repetition and internal inconsistencies are introduced by the editors themselves. For example, the first three chapters include several discussions, and several definitions, of genetic counseling and nondirectiveness, each presented as if the topic is being introduced for the first time. Nondirectiveness makes sense in the reproductive context, but as genetic tests proliferate and interventions emerge, does it really make sense to hold fast to old notions and treat genetics as distinct from all other areas of medicine?

The inconsistencies in the editing are particularly prominent in the chapter on 'referral and diagnosis'. For example, on pages 108–109, there is a section on 'need to know/obligations of the clinician', with bullet points outlining what clinicians must do with respect to diagnosis. Then, on pages 110–111, there is a section on 'watch out for/competency and the obligation to refer'. These two sections say essentially the same thing, although different cases are cited and the nuances may be slightly different. It is not helpful to the reader to break things up this way.

In addition, reflecting the book's dated focus, this chapter deals almost exclusively with legal cases alleging wrongful life and wrongful birth—a situation arising uniquely in reproductive genetics. This section incorrectly and nonsensically presents the case *Breyne v. Potter* as involving a woman who was "told that genetic testing showed that her baby would be born with Down syndrome." The woman proceeded to have an abortion. The following day, the physician telephoned the woman, admitting that he had been mistaken and said that the baby "actually had trisomy 21." In actuality, the doctor incorrectly reported that the fetus had Down syndrome but then correctly reported that the fetus had 47 XXX. This error might be overlooked were it not for numerous other errors.

While there is little to like in this volume, there are some good bits. The contributed material on reproductive testing in chapter 8 is quite good, including a summary of the relevant clinical practice statements and ethical guidelines of the various health professional societies. Similarly, the chapter "Test Samples and Laboratory Protocols" includes a useful list of physician and laboratory obligations, though it sometimes veers into the impractical, such as suggesting that physicians must know the validity of tests and reliability of labs—information that is often difficult if not impossible to come by.

Each chapter includes sections on 'need to know' and 'what to watch out for' that highlight court cases in which physicians have been found negligent. But the authors could have better aided readers in avoiding such a fate by providing clear directions to professional guidelines that often serve as an indicator of the standard of care. Indeed, the reader would be better informed by reviewing those guidelines rather than laboring through this volume.

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