



Preface

Genetic screening and counseling



Anthony R. Gregg, MD Joe Leigh Simpson, MD
Guest Editors

The human genome has now been reliably and almost completely sequenced. In view of the huge investment required to reach this milestone, there are tremendous pressures to apply this information. Putting this knowledge to work in the field of medicine has the potential to immediately quell critics and show a direct benefit to mankind. With the unraveling of the human genome sequence, new challenges in genetic counseling have emerged for health care providers. For example, the burden to offer patients testing for genetic diseases can no longer be dismissed, interpretation of tests has become more complex, previously unrecognized ethical issues have emerged, and of course the cost of providing health care services has not diminished.

New demands placed on the obstetrician-gynecologist to provide genetic counseling caused us to devote the first two articles of this issue of *Obstetrics and Gynecology Clinics of North America* to genetic counseling. We hoped to offer some proof that genetic counseling is more than a casual conversation with a patient or couple. We start our discussion of genetic screening for specified diseases with a discussion of cost efficacy. Screening programs already in place for Tay-Sachs disease in the United States and β -thalassemia in Sardinia have proven effective; however, they target a very specific ethnic group. Canavan disease screening is relevant because it is straightforward and like Tay-Sachs and β -thalassemia targets a specific ethnic group. New standards for carrier screening of cystic fibrosis have recently been established through a combined effort of the American College of Obstetricians and Gynecologists, American College of Medical Genetics, and National Institutes of Health. Although directed at specific ethnic groups, these guidelines guarantee the obstetrician-gynecologist will need to have a minimal understanding of the clinical and genetic aspects of cystic

fibrosis. Finally, we included discussions of breast cancer because of its immediate association with women's health care and tri-nucleotide repeat disorders (e.g., fragile X syndrome) because of their importance in the etiology of mental retardation and a spectrum of additional disorders affecting the nervous system.

We hope this issue of the *Obstetrics and Gynecology Clinics* will confer on all obstetrician-gynecologists the exciting promise that the "new genetics" offers for genetic screening and medicine in general.

Anthony R. Gregg, MD

Joe Leigh Simpson, MD

*Departments of Obstetrics and Gynecology
and Molecular and Human Genetics*

*Baylor College of Medicine, 6550 Fannin Suite 901 A
Houston, TX 77030, USA*