Preface
Prenatal Screening and Diagnosis

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Guest Editors

It has been an honor to serve as guest editors for this special edition of *Clinics in Laboratory Medicine*, entitled Prenatal Screening and Diagnosis. The last issue devoted to prenatal testing was in June 2003. Since that time, a lot of development has occurred in the arena of prenatal testing, particularly with the development of new screening paradigms for Down syndrome. This edition may seem to have more articles dedicated to aneuploidy screening compared with prior editions. The reason for this is the proliferation of new screening strategies in the past 5 years and the struggle among clinicians to adopt what is best for their population.

As editors, we have made an attempt to avoid overlap in the articles submitted. Some degree of overlap is, however, inevitable in the articles devoted to aneuploidy screening. Dr Bahado-Singh begins by giving an overview of first-trimester screening. Others, such as Dr Egan and colleagues, review the impact of first-trimester screening on second-trimester screening for aneuploidy, whereas Campbell and colleagues provide an update on genetic sonogram. Drs Caughey and Odibo then review the economic considerations of the various aneuploidy screening paradigms. A review of aneuploidy screening would not be complete without an article on neural tube defects screening from which aneuploidy screening was derived. Such an article is provided by Krantz and colleagues.

Among the articles on aneuploidy, several relate to first-trimester screening with free β–human chorionic gonadotropin, pregnancy-associated plasma protein A, and nuchal translucency. Drs Sonek and Nicolaides focus on the addition of new ultrasound markers in the first trimester to improve screening performance. Although many studies have reported detection rates of 90% with first-trimester screening, Dr Cuckle focuses on the important issue of maintaining quality control of nuchal translucency measurements to maintain these high detection rates when nuchal translucency assessment is pushed into widespread clinical practice. Dr Chasen's article discusses the clinical implications of first-trimester screening, including the impact of early screening on the rate of invasive prenatal diagnosis, the gestational age of prenatal diagnosis and termination, as well as the side benefit of earlier prenatal

Clin Lab Med 30 (2010) xv–xvi
doi:10.1016/j.cll.2010.04.017 **labmed.theclinics.com**
0272-2712/10/$ – see front matter

diagnosis of certain major structural abnormalities. Dr Norton's article reviews the benefits of an instant results approach to first-trimester screening in which biochemistry is performed before ultrasound and, therefore, the final risk result is available at the conclusion of the ultrasound examination. Such an approach can improve screening performance in addition to the added clinical benefit of being able to communicate with patients at the conclusion of an ultrasound examination. Dr Pergament writes about the important role of genetic counseling in first-trimester screening given the general population's understanding of risk. Dr Goetzl's article discusses the adverse implications of extreme first-trimester aneuploidy markers when aneuploidy has been ruled out.

Drs Evans and Andiole discuss multiple pregnancy, which has a lower performance of serum screening protocols as compared with singleton pregnancies. The complexity of the invasive diagnostic procedures and the risk of loss of an unaffected twin due to the sequelae of the invasive procedures.

Although screening for cystic fibrosis is now a routine part of prenatal care, Drs Cahill and Goetzinger discuss the challenging aspects of offering such screening, especially as it relates to certain racial groups and populations. Another vexing area in prenatal testing is that of screening for thrombophilias, and Rampersad and Carbone provide a comprehensive review of the subject. Another medical disorder that has received a lot of screening attention is preeclampsia. Most of the focus is now on early detection and the first trimester provides a window for early screening. Tuuli and Odibo cover this area.

The use of ultrasound and biochemical methods to screen for preterm birth is now mainstream in prenatal care. Wax and colleagues performed a detailed review of the literature and provide an evidence-based summary of the subject. This is followed by a detailed review of the role of preimplantation genetic diagnosis by Jungheim and Cooper.

There has been a lot published in recent years about screening and treating infectious diseases, such as cytomegalovirus, during pregnancy. Borgida and colleagues performed a detailed review of this subject with particular emphasis on toxoplasmosis and cytomegalovirus.

Dr Evans provides an article on noninvasive prenatal diagnosis, a technology with huge potential. Unfortunately, the large number of technical approaches that have been used is testimony to the fact that none of them has been particularly successful.

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