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BOOK REVIEW

Sharad Gogate: Guidelines and Standards for Maternal Serum Screening for Down's Syndrome, Neural Tube Defects, and Other Obstetric Problems

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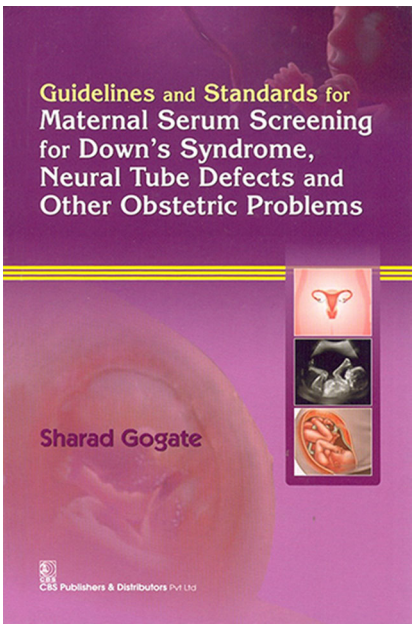
About the Reviewer

Dr. Pooja Vaziraani had her postgraduate training in Obstetrics & Gynaecology at the L.T.M.M. College and Hospital, Mumbai, which she completed in 2005. She proceeded to Mediscan Fetal Care Center in Chennai, India, to pursue a 2-year Clinical Fellowship Programme in Fetal Medicine in 2008. Since 2010, she is working as a Consultant in Fetal Medicine in the high-risk Obstetrics unit at Dr. L.H. Hiranandani Hospital, Mumbai, Grace Hospital and Mediscan Fetal Medicine Center, Mumbai. She has been a faculty to various National conferences and workshops. She has contributed chapters in textbooks of Obstetrics as well as Fetal Medicine. Her special interests in the field of Fetal Medicine is screening for aneuploidy, multifetal gestation and fetal Cardiology.

Maternal serum screening for birth defects began in the early 1970s with the discovery of serum alpha-fetoprotein in the second trimester. It was found to be increased in fetuses with only open neural tube defects [1]. The book explains the advent of the serum screening for open neural tube defects. Indeed, the use of multiples of median (MoMs) to report results was partly adopted because of vast interlaboratory differences [1]. However, now in today's era of excellent USG machines and experts in the field of imaging, we have now shifted our focus from



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serum screening to USG detection and diagnosis of the open neural tube defect. Apart from the well-known second-trimester signs of ONTD, i.e., lemon-shaped skull and obliterated posterior fossa, we have methods or markers for the detection of ONTD in the 11–13 + 6 weeks scan. Thanks to Prof R. Choui for the introduction and correlation of the fourth ventricle in the 11–13 + 6 weeks scan as the intracranial translucency [2]. The chapter on the anatomy in the 11–13 + 6 weeks describes the sign in detail with good pictorial illustration. There are other signs as well which could not have been included, but readers can refer to the Journal of Ultrasound in Obstetrics for the same.

In the early 1980s, it was discovered that lower levels of AFP could also be indicative of the presence of fetal chromosomal abnormalities [3]. Over the past few years, extensive research on refining serum screening for detection of aneuploidies especially for Down syndrome has lead to a standardization of the 11–13 + 6 weeks combined first-trimester screening for T21, T18, and T13 [4]. The combined first-trimester screening gives us 90 % detection rate with 5 % FPR if maternal age, NT, and serum-free beta HCG and PAPPA are taken into account. The main part of the screening is, however, the measurement of Nuchal translucency. The chapter on 11–13 + 6 weeks scan describes the criteria in detail [4]. The chapter's heading has a printing error of 10–13 + 6 weeks instead of 11–13 + 6 weeks scan.

The other USG markers in the first trimester, i.e., imaging the nasal bone, ductus venosus flow, and the tricuspid regurgitation are important, and they should be included in the screening protocol if the screen is in the intermediate category [4–6]. The book mentions the high-risk cutoff as 1/100 and the low risk as 1/1000. The intermediate group thus will lie between 1/100 and 1/1000. This is, however, followed in the UK. We in India still follow the risk of 1/250 as the cutoff, and thus, for us the value between 1/250 and 1/1000 will be the intermediate group. We are hoping that in few years we too shall have our adjusted cutoff values that will suit our population. As far as the second-trimester screening for aneuploidy is concerned, we have not shifted from the triple marker test to the quadruple screening. We adopt the second-trimester screening only in those cases which have missed the FTS. It is indeed sad to say that in current practice, people are unaware about the implications of the screening test unless it turns out to be alarming. It has to be understood that screening is a program and not mere a blood test and has the diagnostic testing as the end point.

The revolution in screening, i.e., the noninvasive prenatal testing for T21 has changed the outlook to screening programs. It has got certain indications which can be looked up to in Journal of USG in Obstetrics and Gynaecol. In 1997, cell-free fetal DNA was discovered in maternal plasma. Though designated as fetal, the cell-free fetal DNA is placental in origin and results from apoptosis of trophoblast cells. The chapter on NIPT (noninvasive test for aneuploidy, i.e., T21, T18, and T13) gives extensive information about the different technical methods used in NIPT interpretation and risk calculation. In singleton pregnancies, cell-free DNA (cfDNA) testing in maternal blood can detect about 99 % of cases of trisomy 21, about 97 % of trisomy 18, and 92 % of trisomy 13, with respective false-positive rates of <0.1, <0.2, and <0.2 %, respectively. 1cfDNA testing in twins is feasible, but the reporting rate of results is lower in singletons due to a lower fetal fraction [7, 8].

Screening for preeclampsia which is not mentioned in detail is the need of the hour. Its importance cannot be overemphasized as there is a 40 % reduction in severe early preeclampsia by a simple method of prevention, i.e., low dose ecosprin if started <16 weeks. The book gives importance to pre- and posttest counselling which is the most crucial part of the screening program [9].

To summarize the review, I would say that it has simple but valuable information on the screening for aneuploidy and neural tube defects. It is a good handbook for day to day office work. Interested readers should, however, read journal articles on the latest screening protocols, NIPT, and more information on the new screening protocols for preeclampsia.

Compliance with Ethical Standards

Conflict of interest There is no conflict of interest.

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