Role of Ultrasound and Second Trimester Serum Screening in Predicting Adverse Pregnancy and Perinatal Outcome

Thesis

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وأنزل الله عليك
الكتاب والحكمة
وعلَّمَكَ مَا لَمْ تَكُن
تَعْلَمُ وكَانَ فَضْلُ
اللَّهِ عَلَيْكَ عَظِيماً
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This work is dedicated to . . .

My Father, *to him I will never find adequate words to express my gratitude.*

Also to My **Husband** for dealing patiently and tactfully during this work

Also to my **daughters**

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List of Abbreviations

AC : Abdominal circumferance
AFP : Alpha feto proteins
BHCG : Beta human chorionic gonadotrophines
CF : Cystic fibrosis
CHD : Congenital heart defects
CNS : Central nervous system
CP : Cerebral palsy
CRL : Crown rumbe length
DM : Diabetes melittus
DPRS : Double positive results
DS : Dawn syndrome
D-wave : Diastolic wave
EFW : Estemated fetal weight
FEB : Fetal echogenic bowel
FGR : Fetal groth restriction
FHR : Fetal heart rate
FN : False negative
FP : False positive
G1,g2 : Gravida 1,gravida 2
HCG : Total human chorionic gonado trophines
IUD : Intrauterine death
IUFD : Intrauterine fetal death
LSD : Less significant difference
MOM : multiple of median
MSS : Maternal serum screening

List of Abbreviations *(Cont...)*

N,No : Number
NBL : Nasal bone length
NINDS : Nationalinstitutofneurologicaldisordesand stroke
NT : Nuchal translucency
NTD : Neural tube defects
ONTDS : Open neural tube defects
PAPP : Pregnancy associated plasma protiens
SD : Standard deviation
SGA : Small for gestational age
SLE : Systemic lupus erythamitosis
S-wave : Systolic wave
T21 : Complete trisomy
TN : True negative
TP : True positive
TRANS-DS : Translocation leading to dawn syndrome
UE3 : Unconjugated estriol
US : Ultrasound
X2 : Chi –squared ratio

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Introduction

In the last 3 decades, perinatal medicine has made tremendous advances in scientific knowledge and in the successful application of this knowledge toward understanding the fetal aspects of pregnancy. Evaluation of the health of the fetus and screening for birth defects has become an important part of prenatal care ***(Rappaport, 2008).***

Congenital anomalies constitute an important cause of infant death with the exception of vitamin supplementation for the prevention of neural tube defects ***(MRC Vitamin Study Research Group, 1991)*** there has been no significant progress in the prevention of congenital malformations. Instead, emphasis has been placed on prenatal detection. Prenatal screening for Down syndrome and neural tube defect has been the standard of care in obstetrics for some time**.** The frequency of these disorders is known to be sufficiently high to justify population screening ***(ACOG, 1996)***.

Rowley noted that the aim of genetic screening should be to maximize the options that are available rather than to reduce the prevalence of genetic diseases ***(Rowley et al., 1984).***

Prenatal diagnosis allows time for a thorough counseling and allows the family to make an informed decision regarding continuation or termination of pregnancy, instigation of medical therapies, preparation for postnatal surgery, and more recently,

the possibility of invasive intrauterine interventions such as valvuloplasties ***(Tworetzky et al., 2004).***

Pregnant women are able to use sophisticated screening information to make scientifically and ethically rational decisions about invasive testing for detection of trisomies and other congenital anomalies. These empiric data compliment the arguments of normative ethics to create evidence-based ethical standards for informed consent regarding invasive testing ***(Chasen et al., 2004).***

We must help pregnant woman to make a decision about invasive testing if needed. This equally important autonomy-based component has been defended on the basis of ethical analysis and argument. The data reported demonstrate that pregnant women exercise their autonomy in a scientifically and ethically rational manner in decisions about invasive testing when that exercise is provided a rigorous evidence base. The decision of undergoing invasive testing must be fully supported by both ethics and science. This is a major contribution to the emerging field of evidence-based obstetric ethics ***(Chasen et al., 2004).***

Replication of the results requires not only high-quality screening techniques and interpretation but also, a high-quality informed consent process and adequate time for an evidence-based informed consent process must be created. Obstetricians who are unable or unwilling to do so should refer the patients to centers prepared to meet the standards of evidence-based obstetric ethics ***(Kypros et al., 2004 b).***

During the last 30 years, extensive research has aimed at developing a noninvasive method for prenatal diagnosis of chromosomal and other abnormalities through the isolation and examination of fetal cells that are found in the maternal circulation. However, on the basis of currently available data, there is no realistic prospect that, in the future, noninvasive diagnosis will replace the need for invasive testing ***(Joe, 2012).***

Invasive prenatal diagnosis requires either amniocentesis from 16 weeks of gestation or chorionic villous sampling from 11 weeks of gestation. Randomized studies have demonstrated that the procedure-related risk of miscarriage is the same (approximately 1%). Consequently, invasive testing is carried out only in pregnancies that are considered to be at high risk for chromosomal abnormalities ***(Bianchi et al., 2002).***

The traditional method of screening is maternal age, with which invasive testing in 5% of the population identifies approximately 30% of the fetuses with trisomy 21. There is now extensive evidence that ultrasound examination at 11 to 13 weeks of gestation, combined with maternal serum biochemical testing, can identify > 95% of the fetuses with major chromosomal abnormalities So the most effective method of screening for chromosomal defects is by first-trimester fetal NT and maternal serum biochemistry ***(Kyriaki et al., 2005).***

Increased fetal nuchal translucency thickness is associated with trisomy 13, trisomy 18, trisomy 21, Turner syndrome, other sex chromosome abnormalities, as well as