

Obstetrical Ultrasound Examination And Biochemical Markers As Contemporary Tool Assessment For Fetal Anomalies In Albania

Rustem Celami

Obstetrician and Gynecologist at University Hospital of Obstetrics and Gynecology "Koço Gliozheni" Bajram Curri Boulevard, Elbow, Tirana, Albania, Tel: + 355 66 60 70810. Email: rustemcelami@gmail.com

ABSTRACT

Obstetrical ultrasound screening has taken a principal role nowadays in obstetrics and gynecology practice. It has captured in a way even medical professionals' thoughts besides general population attention of having done such examination. With the revolutionary progress of technology the ultrasound machine itself offers tremendously stunning images from two dimensional, three dimensional and now has become routine even four dimensional ultrasound examination, bringing clinical practice to a level of excellence patient care delivery.

The obstetrical clinical practice in Albania, pregnant women are suggested routinely the combination of obstetrical ultrasound examination with biochemical markers. A nuchal translucency (NT) more than 3 mm is associated with an increased risk of structural fetal abnormalities, fetal heart abnormalities, subsequent pregnancy loss, and other. Double, triple and quadruple test are accessibility and commonly biochemical markers done as complementary tests to ultrasound screen in early detection fetal anomalies in our clinical practice.

In conclusion, nowadays medical practice in obstetrics and gynecology has adopted the classic and modern aspects in order to offer a superior care to patients. The application of up to date technology in medicine such as obstetrical ultrasound examination and laboratory tests combined with permanent professionals' continuing education offer a better not to say a gold standard assessment tool of probably diagnosis. Patients need to be counseled about the positive and negative findings that ultrasound and laboratory tests may reveal so they are prepared for unexpected pregnancy knowledge and the possibility of further testing options being offered. Consequently, a well organized national health care with a universal medical services proper training and employ of assets sustain our mother and child care service to western standards.

Keywords: Obstetrical ultrasound screening, biochemical markers, structural fetal abnormalities

Introduction

Obstetrical ultrasound examination principally is influenced by two essential factors, first the component of human resources such as a proficient examiner which takes time and expenses in addition to everlasting education and practice, second component is the machine itself with all the supplementary elements that consist of. Many aspects of antenatal care attendance means have changed in the past few generations. Today, the availability of proper preconception and prenatal care such as early screening, assessment and diagnostic tests, parents are given a chance to prepare themselves for the possibility that their child might be born disabled and to educate themselves about the options. Although the primary aim of first trimester screening is to identify pregnancies at risk of aneuploidy, first trimester findings may give insight into other adverse pregnancy outcomes.

The mainstay of antenatal non invasive screening of the first and second trimester are serum biochemical markers commonalty namely; double, triple and quadruple test combined with ultrasound markers explicitly; nuchal translucency (NT), absent nose bone, dilated brain ventricles, spina bifida and other. Structural fetal abnormalities have significant short and long term impact in health care services and socially in broad spectrum. The practical approach to interpreting abnormal first trimester findings and assess what further testing may be indicated should include a multidisciplinary manner. Abnormalities discussed in this article will include: I. Nuchal translucency, II. Abnormal fetal anatomy detected at first trimester screening and III. Abnormal maternal serum analytes in addition to a harmonized consultation of multidisciplinary experts by respective medical disciplines



Discussion

Nuchal Translucency

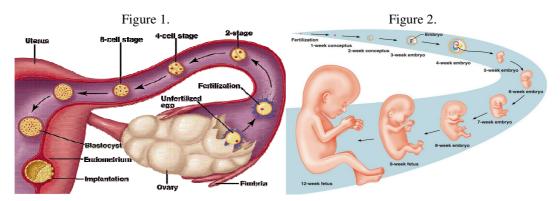
As in many other fields in medicine we do refer our data to the national and international guidelines; evidence based medicine is today an additional polar star which guides us in addition to our theoretical and practical knowledge and experience in making proper assessment, diagnosing and management. So far there are accepted as normal range of nuchal translucency (NT) less than 3 mm, of course taken into strict consideration adequately assessed favorable fetal lie and gestational age CRL 45 – 84 mm [1, 2, 3, 5]. Thickened NT has been associated with a numerous of syndromes, nevertheless most of these have only been reported very few in the literature [2].

In our clinical practice do offer relatively the same standards as in western clinics in regards to obstetrical ultrasound examination such as the measurements of NT and laboratory tests. The question is; how we keep away our professional integrity from marketing of medicine? In a way rather than giving to the patients proper professionally care we do overstress them during a natural process that has been taken place since mankind presence on earth. Consequently, unless a specific medical family history is present that tips to an amplified likelihood of explicit disorder, it is unlikely to be clinically reasonable to counsel patients regarding all these rare possibilities. On the other hand, having these means of assessment and diagnostic, we should be cautious and keep in mind that human assets dominate the decision subsequent to the ultrasound and laboratory findings and deliver the information to the patients in a wised professional manner.

Abnormal fetal anatomy detected at first trimester screening

Before the health technology era in clinical practice which encompasses a broad range of <u>healthcare</u> products used to assess, diagnose, monitor or even treat diseases or medical conditions affecting <u>humans</u>, we did not have many options to conclude to a fetal abnormal anatomy diagnosis other than professional medical knowledge applied into clinical practice.

Development begins at fertilization followed by cleavage of embryo and its implantation in uterus which takes time from day one to first week, pursued by the differentiation of the primary germ layers and culminates in primary organogenesis when tissues and major organ rudiments of embryonic body are formed by eight week. In Albania we do see in our clinical practice most of pregnant women around eight to nine weeks after the first day of last menstrual periods, yet the latest literature do mention the importance of early antenatal visit [4, 5, 6, 7]. Early stages of embryo development shown in figure 1 and stages from fertilization to twelve weeks described in figure 2.



Some of structural fetal abnormalities are visible during first trimester of pregnancy from eleven to thirteen weeks and six days. We do offer obstetrical ultrasound examination during the first and second trimester in our obstetrical clinical practice in Albania such; nuchal translucency measurement and later on the detailed anatomical obstetrical ultrasound examination up to twenty four weeks of gestation. In the mean time we assess other obstetrical ultrasound soft markers; face (visualization of nose, nasal bone, orbits, lips), skull and brain (skull ossification, lateral ventricles, cerebellum, cistern magna, cavum septi pellucidi), anencephaly, encephalocele, explore for occurrence of cystic hygroma, spine (search for presence of neural tube in longitudinal and transverse view, heart (rhythm of fetal heart, four chamber view, and assessment of great vessels), genitourinary (presence, size and shape of kidneys; existence, size and shape of urinary bladder;



presence of genital organs), gastrointestinal (stomach and bowel, abdominal wall and umbilical vessels as well), extremities (evaluate extremities, and digits). When visualization of fetal structures is not optimal or a structural abnormality is suspected during transabdominal scan, transvaginal scan is always performed. Some of abnormal fetal anatomy diagnosed at our medical center are; an increased nuchal translucency - Figure 3, cystic hygroma at 12-13 weeks of pregnancy - figure 4, Dandy Walking malformation - figure 5.

Figure 3. Figure 4. Figure 5.

Abnormal maternal serum analytes

Serum biochemical markers are requested routinely in a way at our clinical setting, the first trimester biochemical screening in combination with obstetrical ultrasound findings are suggested two markers or as frequently called double test (beta-human chorionic gonadotropin β -hCG and pregnancy associated plasma protein A – PAPP-A) both measured from eleven to thirteen weeks and six days accordingly crown-rump length up to 84 mm. Second trimester serum biochemical screen consist of triple test (AFP, hCG, and unconjugated estriol-UE₃) and measurement of an additional hormone levels to triple test known as inhibin A which is released by placenta is called quadruple test. These two last tests are done from $15^{th} - 22^{nd}$ weeks, however in order to obtain most accurate values we suggest to be done between 16^{th} and 18^{th} weeks of gestation.

Countless factors affect levels of maternal serum analytes which ought to be taken into consideration when calculating risks. These include; previous screening results, gravidity and parity, smoking, alcohol and other substance abuse, previous or present harmful exposure, assisted reproduction, pregnancy complications, maternal age, weight and diabetic status. Screening test suggested in first and second trimester of pregnancy identify pregnant women mainly as high risk or low risk, where in the high risk group is proceeded to diagnostic procedures, and in low risk group keep on regular assessment. So in the other words, positive results in these screening test only places the pregnant women to a high risk group and does not in any way imply decisively that fetus is affected with abnormalities.

Conclusions and Recommendations

Maternal and child health is one of the most important indicators of any country national health standards. Continuous professional medical education of human resources together with application of up to date assets and technology in clinical practice will bring short and long term impact to a better health service delivered to population. Proficient medical knowledge in cooperation with appropriate interpretation of obstetrical ultrasound examination findings and laboratory test results will aid pregnant women and their loved ones to make an informed decision throughout pregnancy.

A well-organized national health care service with a universal medical system, proper training and utilization of assets withstand our mother and child care service to western standards

Finally, suggested recommendations are:

- 1. Albanian professional license of obstetrical ultrasound practitioners must express the proficiency level of exam interpretation.
- 2. The privileged professionals exercising this medical practice must be licensed and accredited by a recognized western university clinic/center of fetal medicine.
- 3. Multi-disciplinary consultation in accordance with patients and/or her family should make the final informed decision of any fetal malformation over 12 weeks of gestation.



Acknowledgments:

I would like to thank for the encouragement and support my mentor Prof. Dr. Krenar Preza (Mother Teresa University Hospital Center|Faculty of Medicine, Medical University of Tirana, Tirana, Albania), Dr. Zef Delija, Dr. Genc Kabili, Dr. Kastriot Dallaku, Dr. Islam Mamica, Dr. Rubena Moisiu University of Obstetrics and Gynecology Koco Gliozheni|Faculty of Medicine, University of Medicine Tirana, Tirana, Albania), and Emanuela Celami BSc, MSc in Biology (Faculty of Natyral Sciences, University of Tirana, Tirana, Albania).

References

American Academy of Pediatrics, American College of Obstetricians and Gynaecologists, author.Guidelines for perinatal care. 5th ed. Elk Grove Village, (IL): American Academy of Pediatrics, American College of Obstetricians and Gynaecologists; 2002.

Athena P. Souka, Constantin S. von Kaisenberg, Jonathan A. Hyett, Jiri D. Sonek, MD, Kypros H. Nicolaides. Increased nuchal translucency with normal karyotype. American Journal of Obstetrics & Gynecology Volume 192, Issue 4, Pages 1005-1021, April 2005.

Bower S, Chitty L, Bewley S, et al. 1995. First trimester nuchal translucency screening of the general population: data from three centres (Abstract). Presented at the 27th British Congress of Obstetrics and Gynecology, Royal College of Obstetrics and Gynecology, Dublin, Eire.

Kypros H. Nicolaides, Victoria Heath and Simona Cicero. Prenatal Diagnosis. Prenat Diagn 2002; 22: 308–315. Published online in Wiley InterScience (www.interscience.wiley.com). DOI: 10.1002/pd.308.

National Institute for Health and Clinical Excellence, author. Antenatal care: Routine care for healthy pregnant women. 2003.

Low Pamela, Paterson Janis, Wouldes Trecia, Carter Sarnia, Williams Maynard, Percival Teuila. Factors affecting antenatal care attendance by mothers of Pacific infants living in New Zealand. The New Zealand Medical Journal. 2005;118:1216. [PubMed].

Salvador J, Borrell A, Lladonosa A. Increasing detection rates of birth defects by prenatal ultrasound leading to apparent increasing prevalence. Lessons learned from the population-based registry of birth defects of Barcelona. Prenat Diagn. 2005 Nov;25(11):991-6.