First Trimester Fetal Ultrasound Scan as A Screening and Diagnostic Tool

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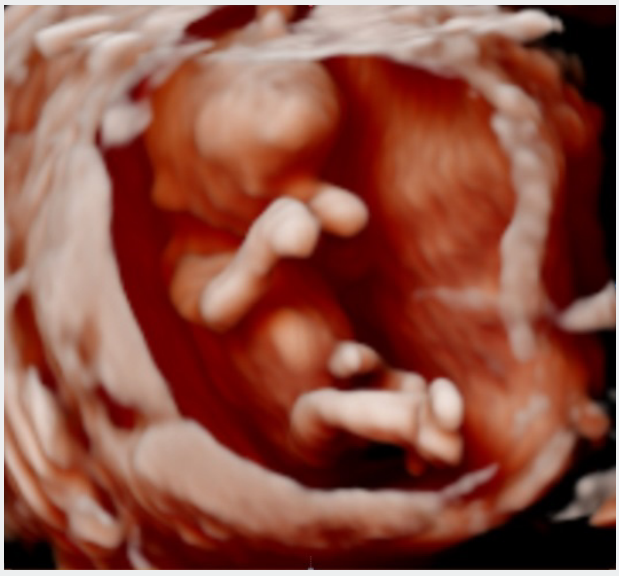

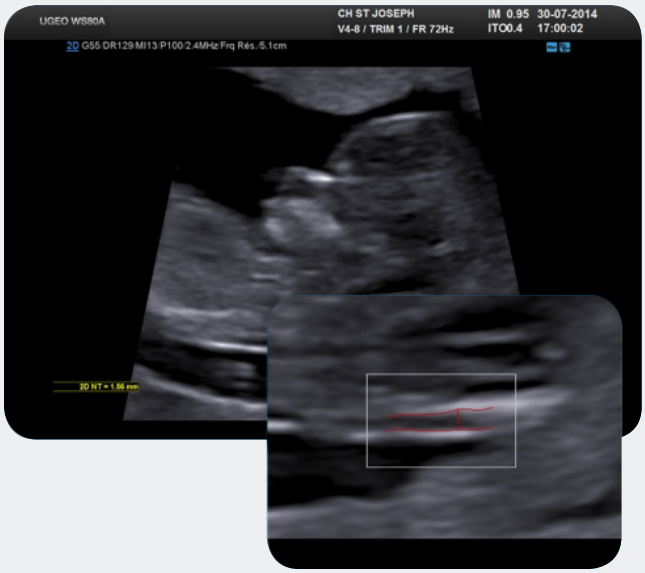

" Due to the recent enhancement of the first trimester US quality, the differentiation between routine screening ultrasound and diagnostic ultrasound becomes pointless. "

Introduction

First trimester ultrasound (US) has drastically changed during the past twenty years. The information provided from the first trimester scan has considerably expanded. When we discuss about first trimester US we are surprised to see how this scan has evolved from a "basic" scan with the function confirming pregnancy, its viability, location, and establishing gestational age to a more "sophisticated" scan with additional biometrics and morphological data as well as evaluation of various maternal and fetal risks. We will exclude the workup for maternal risks this time in order to focus on the fetal assessment.

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1) Fetal biometrics

It is mandatory to perform some measurements during the first trimester US. In most patients, biparietal diameter (BPD) and crown-rump length (CRL) (Figure 1a and b) allow us to estimate gestational age with some degree of accuracy up to 15 weeks gestational age. This information will not be amended thereafter and will mainly be used in management of intra uterine growth retardation (IUGR), in planning congenital anomaly test, and in management of post-term complications. US scan between 11 + 0 and 13+ 6 weeks shall be continuously compared to the one performed between 7 and 10 weeks. Therefore, first trimester US screening should be possible to detect some abnormalities such as fetal triploidy.[1]

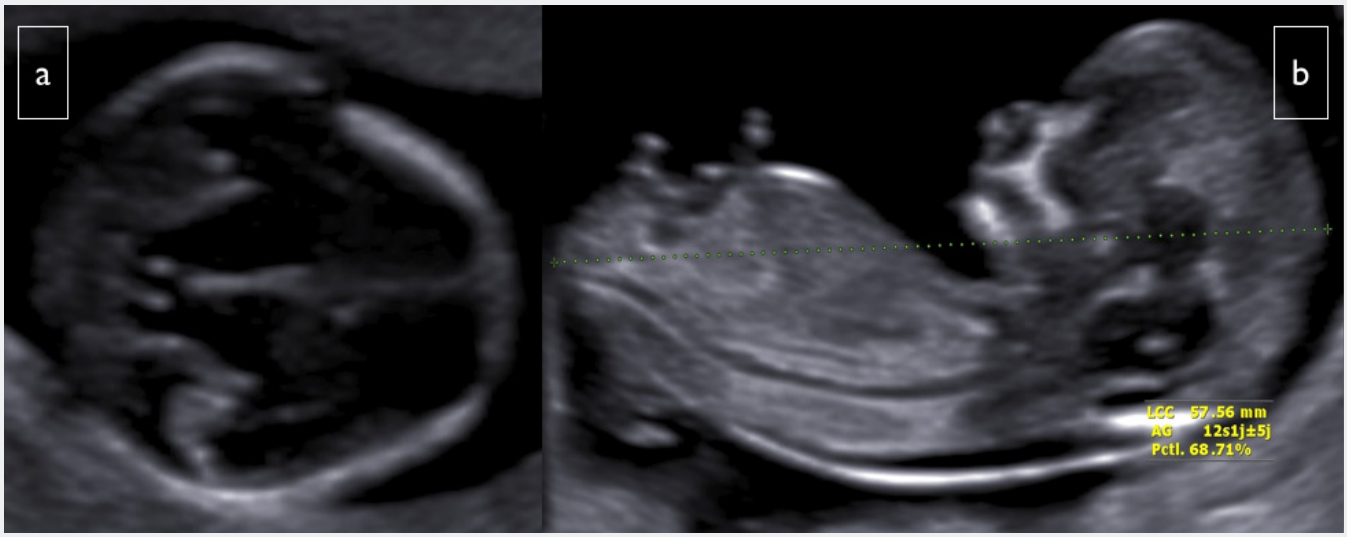

Figure 1a and b. Estimating the gestational age during the beginning of the pregnancy by using (a) BPD and (b) CRL measurements.

2) Nuchal translucency

In the early 1990s, the concept of nuchal translucency (NT) measurement during the first trimester of pregnancy was introduced, and we know that this application is not only a marker of aneuploidies but also an indicator of a wide variety of syndromes and malformations.[2] K. Nicolaides and his group has proposed a new model for aneuploidy screening as he established the rules for the proper usage of this method, in particular with the need for a learning curve, as well as the need for auditing both qualitatively (through the use of quality scores imaging) and quantitatively (median, delta-NT, etc). The Fetal Medicine Foundation has standardized this clinical protocol and established the certification policy to improve quality of care and reduce screening variability.[3,4] NT measurement can be acquired manually (Figure 2a) as well as semi-automatically (Figure 2b) to improve the reproducibility of the measurements. Moreover, advanced measurement of NT, 5D NT developed by Samsung, recognizes the correct mid-sagittal plane and provides improved Herman score.

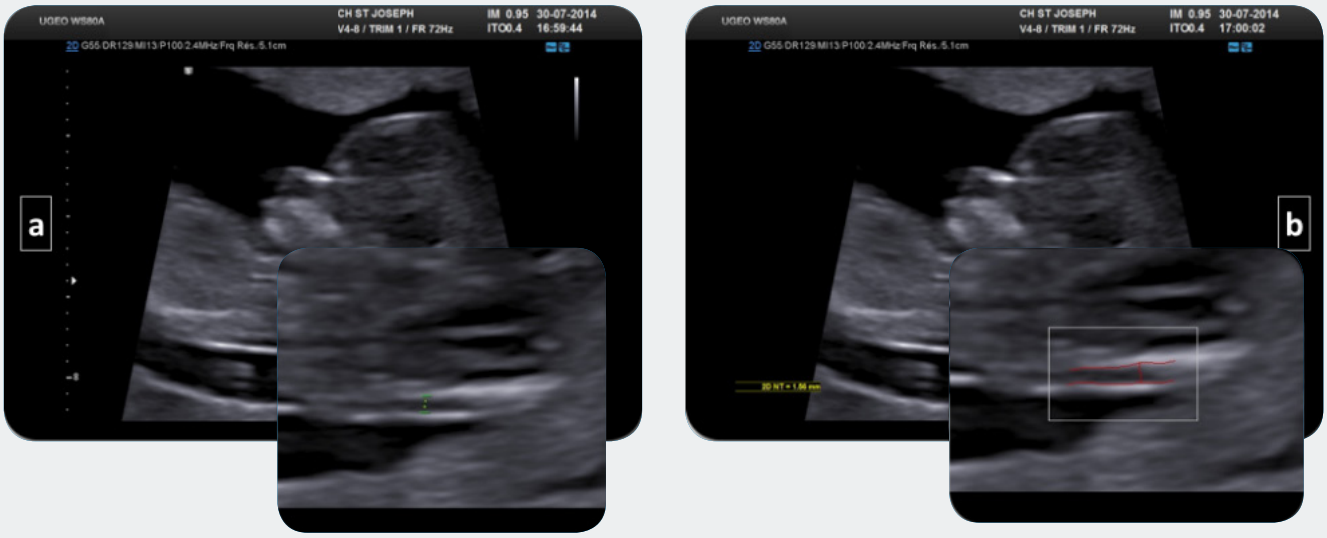

Figure 2a and b. Nuchal translucency measurement can be achieved manually (a) or semi-automatically (b)

3) The morphological assessment

The reported detection rate of defects in the first trimester of pregnancy vary considerably in the literature depending on populations, methods, and periods considered.[5] Morphological analysis during the first trimester of pregnancy has evolved along with the development of ultrasound technology. The International Society of Ultrasound in Obstetrics and Gynecology(ISUOG) Practice Guidelines, published in 2013, suggests anatomical assessment of fetus between 11+0 and 13+6 weeks of gestation (Figure 3).[6]

We can identify various anomalies from a first trimester US scan: 1) the visible anomalies - anencephaly, alobar holoprosencephaly, gastroschisis, omphalocele, body stalk syndrome, megacystis, and some lethal osteochondrodysplasias, 2) the invisible anomalies - brain, digestive, urological, and skeletal diseases, and 3) certain anomalies requiring analysis - cardiac, facial and limbs conditions, spina bifida.

Due to the recent enhancement of the first trimester US quality, the differentiation between "routine screening ultrasound" and "diagnostic ultrasound" becomes pointless. Advancement in 3D/4D and automated measurement technologies provide more capabilities for accurate morphological assessment.

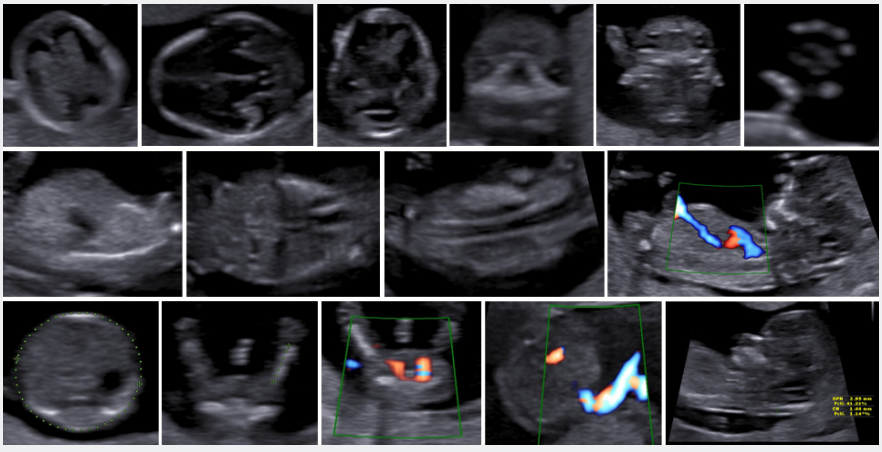

Figure 3. Anatomical details from the first trimester scan

4) The diagnosis of chorionicity and amnionicity

Diagnosis of chorionicity (bichorial vs. monochorial) and amnionicity for monochorionic pregnancies (mono vs. biamniotic) is the cornerstone of scanning twin pregnancies. In the setting of multiple pregnancies, this information is essential for an appropriate management of a discordant malformation, growth retardation, intrauterine fetal death, or fetal sampling. The accuracy of this diagnosis can be reached up to 100% during the first trimester and then decreases throughout pregnancy.[7] The visualization of the "Lambda" sign (Figure 4a) confirms bichorionicity. In contrast, the absence of visualization of the Lambda sign or identification of a "T" connection (Figure 4b) of the inter amniotic membrane which is perpendicular to the chorionic plate allows us to assess monochorionicity.

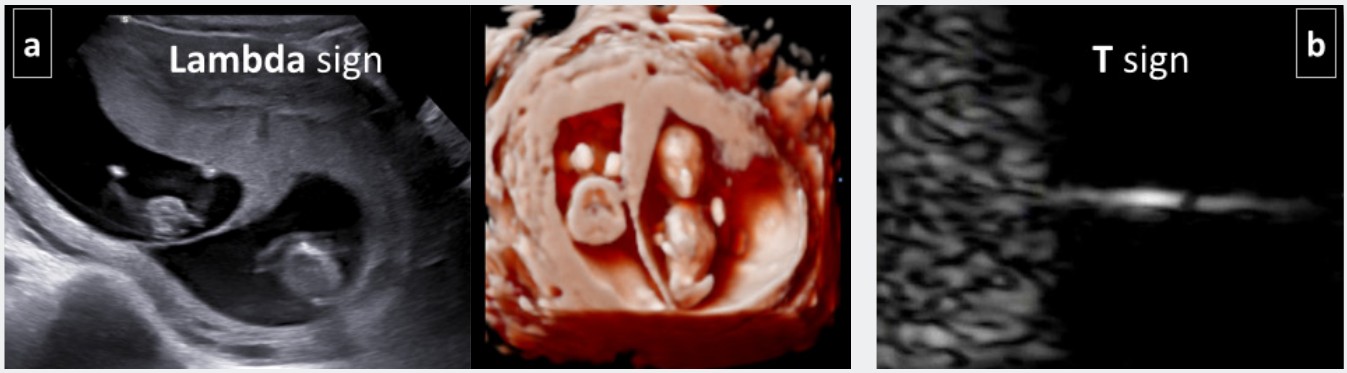

Figure 4. Diagnostic of chorionicity and amnionicity during the first trimester by assessing the "Lambda" (a) and "T" signs (b)

5) The assessment of fetal risk

First trimester US also has the ability to characterize several fetal risk such as aneuploidies, congenital heart defects, and spina bifida.

A. Aneuploidies

Ninety percent of fetuses with major aneuploidies can be detected by the combined exam of maternal age, NT measurement and maternal serum markers (PAPP-A and free beta hCG).[8] This screening can be improved by an earlier use of serum markers between 9 and 10 weeks and US scan during 12 weeks for additional markers such as fetal nasal bone, blood flow in the ductus venosus and through the tricuspid valve (Figures 5a, b, c, and d).[9] In addition, Non Invasive Prenatal Testing (NIPT) enables us to go one step further to screen common fetal chromosome anomalies.

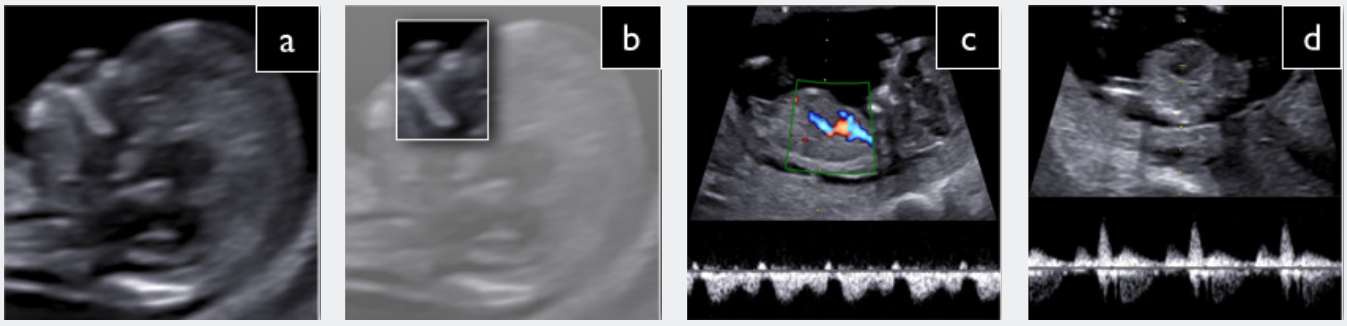

Figure 5a,b,c and d. Evaluation of aneuploidies by ultrasound markers such as nuchal translucency (NT), nasal bone (NB), Ductus venosus (DV), and tricuspid flow (TF)

B. Congenital heart defects

Early fetal echocardiography can be performed for populations at high risk of congenital heart defects (CHD). This examination is usually scanned by experts who are familiar with the prenatal diagnosis of CHD. Fetuses can be classified as high risk for CHD, not only based on personal, family histories and toxic exposure, but also enlarged NT and abnormal blood flows in the ductus venosus and through the tricuspid valve.[9] A "detailed" fetal echocardiography is usually reserved for high-risk fetuses while a "basic" echocardiography is not yet currently recommended for the general population.

However, the basic ultrasound could mainly be supported by the careful use of Color Doppler energy modes in order to identify the presence of atrioventricular flows on the four-chambers (Figure 6a) and the three vessels and trachea views (Figure 6b). Obtaining such views can reassure parents about the absence of severe cardiac disease such as single ventricle, ventricular hypoplasia, complete atrioventricular septal defect (AVSD), aortic or pulmonary atresia and some types of arterial malpositions.

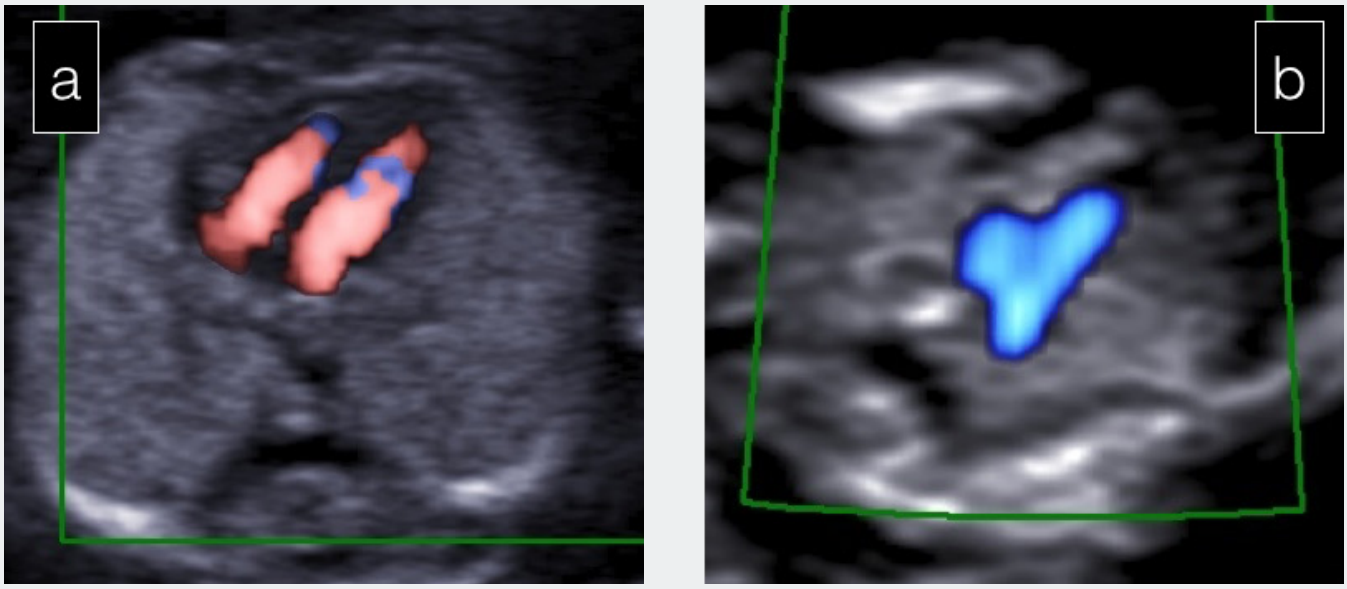

Figure 6a and b. Early fetal echocardiography using Color Doppler energy mode by visualizing the four chamber (a) and three vessel tracheal (b) views.

C. Spina bifida

Historically, doctors and sonographers have been screening neural tube defects, mainly open spina bifida, during the second trimester US scan. We have seen in recent years that the advent of early sonographic markers obtained between 11 and 14 weeks (intra cranial translucency, the use of BPD and BPD / ATD ratio) allow us to consider some fetuses as being at high risk of open spina bifida (Figure 7).[10-12]

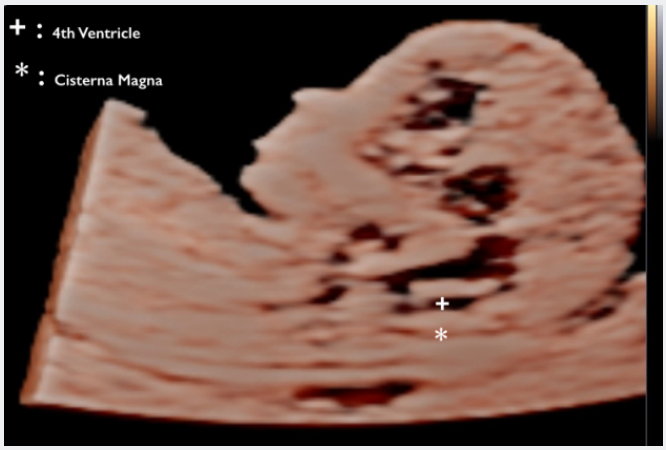

Figure 7. Cranial intra translucency (IT) in the setting of screening US of open spina bifida

Conlcusion

We have been witnessing the significant transformation of first trimester US during the last two decades. The first trimester of pregnancy has become the critical period for maternal-fetal risk assessment, personalized counseling, and follow-up. We expect further development and maturity in the use of first trimester US scan as the crucial screening and diagnostic tool.

Supported Systems

- WS80A with Elite
- WS80A

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