



# Center For Advanced Fetal Care Newsletter

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## "First Things" First...

Bold. Brilliant. Defiant. Rebellious. Revolutionary. Simply put: inspirational. That is exactly how one would describe the 10th World Congress of the Fetal Medicine Foundation held in Malta June 26-30, 2011, which also marked the 20th anniversary of Professor Nicolaides' introduction of the nuchal translucency. Fast forward, 20 years later, and here we are today, with it clearly having become one of the pillars of obstetrical care and serving as the cornerstone for inverting the "pyramid of obstetrical care". As such we dedicate this issue to "first things" and to the first trimester. We bring highlights from the 10th World Congress, provide guidelines as to what may be seen at the time of the 12 week scan, present the first trimester risk calculators available to all on the website of the Fetal Medicine Foundation, in addition to our literary recommendation, news, interesting case report and upcoming congresses. We hope that this issue will further spark your interest in the first trimester and its limitless potential, praying that we may be granted the life, in order to be able to fast forward into the future, in order to experience this most exciting transition into a tomorrow with an inverted, evidence-based, revolutionary pyramid...



## Announcing the Birth of SANA Medical NGO

It is with much excitement that we announce the birth of SANA, a medical NGO dedicated to outreach work in the underserved areas of Lebanon. SANA was developed in cooperation with ISUOG and in accordance with ISUOG outreach guidelines. SANA's first outreach trip was carried out on June 7 to Knaisse, Akkar. SANA is currently in the needs assessment phase for several areas in Akkar, as well as, in the mountainous areas of North Lebanon. For details on SANA's current and future activities, please visit [www.sanango.org](http://www.sanango.org). You may also follow SANA on Facebook.



## Briefing from the 10th World Congress of the Fetal Medicine Foundation

Over 1500 delegates from 83 countries gathered at the 10th World Congress with average 12 hour days packed with live debates among the world's leaders on the most challenging issues affecting maternal and fetal health. And to be able to witness these interactions, at the highest possible human intellectual level, is a priceless unforgettable, most inspiring, privilege. The meeting started with the Eurofetus Research Group's update on the state of fetal surgery. Prof. Arzt from Austria presented the data on in utero surgery for critical aortic and pulmonary artery stenosis with upcoming plans for a randomized controlled trial (RCT) with a newly established website for a global registry of in utero cardiac surgery at [www.ifcir.org](http://www.ifcir.org). Prof. Ryan from Canada presented their experience with thoracic shunts for pleural effusions. Prof. Flake from CHOP in the USA discussed cellular stem cell therapy, as well as antenatal surgery for myelomeningocele and sacrococcygeal teratomas. Prof. Deprest from Belgium presented a Eurofetus RCT currently underway for FETO in cases of diaphragmatic hernia and details can be found at [www.totaltrial.eu](http://www.totaltrial.eu). Prof. Lewi from Belgium gave a major overview on monozygotic twins and stressed that it is an absolute must to determine chorionicity at the first evaluation in order to plan the prenatal and perinatal care. Dr. Moreira from Brazil presented the futuristic "virtual view of the placenta", a live 3D model generated from MRI acquired images, mapping the vasculature and enabling virtual in utero 3D travel, following the cords and vessels to properly plan and carry out the therapeutic procedures. Prof. Berge from the Netherlands showed unbelievable surgical outcomes in repairing craniofacial defects and stressed the importance of a multidisciplinary team approach to tackle the many challenges facing these infants. Prof. De Coppi from the UK discussed the exciting future of tissue engineering. A major highlight was Prof. Romero from the USA presenting his group's new landmark study on the use of progesterone in order to decrease the rate of prematurity by 45% in women with a midtrimester sonographically-detected short cervix. As such, the leaders recommend starting global screening by transvaginal measurement of cervical length on all women in order to identify the high risk group and commence prophylactic vaginal progesterone where one premature birth at GA < 33 weeks is prevented for every 11 women treated. First trimester cervical length is possible with care in order to delineate a measurement of the cervix alone or inclusive of the cervico-isthmic junction, a prominent area in the first trimester that adds approximately another 17mm to the cervical length. In addition, the role of cerclage and the Arabian pessary were presented by Prof. Alfirevic from the UK and Carrera from Spain. Prof. Bindas from Slovenia cautioned us on our over-eagerness to provide over-kill treatment for mild forms of cervical intraepithelial neoplasia, by either cold knife cone or LEEP, both of which put our patients at future increased risk for preterm delivery. Perhaps, one of the most intense sessions was on first trimester combined screening for preeclampsia using history, sonographic as well as biochemical markers, with a detection rate of 45% for a false positive rate of 10%. Here 75mg of Aspirin may be initiated for prevention. An update on the challenging early diagnosis of cardiac, as well as CNS abnormalities, was presented by Prof. Chaoui from Germany, and on craniofacial defects by Prof. Sepulveda from Chile. Amazing work was presented by Prof. Garatacos' team from Spain on the neurodevelopment of IUGR fetuses highlighting a unique postnatal brain modeling and development as demonstrated by sophisticated MRI techniques: connectomics and texture analysis. In the near future, AFP and PLGF will be added to first trimester biochemistry for their roles in various conditions. Prof. Tabor from Denmark discussed the effect of ART on interpreting first trimester biochemistry. Prof. Bilardo from the Netherlands provided a wonderful overview of the outcome of fetuses with an increased NT and normal karyotype. Prof. Ville from France discussed the invaluable role of ultrasound at 32 weeks for detection of fetal growth abnormalities. Prof. Jani from Belgium presented unbelievable "virtuopsy" images on first trimester fetuses using the 9.4T MRI for postmortem exams of the tiniest hearts and brains...And rightfully, it all culminated in Prof. Nicolaides' passionate call to "inverting the pyramid of obstetrical care", the evidence-based time for which has finally arrived...



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# Practical Guidelines to Assist in Performing a Fetal Anatomical Survey at 12-14 Weeks

## Introduction

With the advances in today's technology, the ability to carry out a complete anatomical survey of the fetus at the time of the nuchal translucency (NT) measurement is becoming more feasible using the transabdominal route. The components of the first trimester scan (FTS) are ever evolving. Today the role of the FTS has several components. There are the aneuploidy markers namely the NT, the nasal bone (NB), the frontomaxillary facial angle (FMFA), tricuspid regurgitation (TR), ductus venosus flow (DV) in addition to the sonographic markers such as choroid plexus cyst (CPC), echogenic bowel (EB), echogenic intracardiac focus (EIF) and pyelectasis (Dagklis 2008). The NT serves as a most powerful marker for various other structural abnormalities. In addition, an increased NT is the strongest risk factor for a fetus to have underlying congenital heart disease.

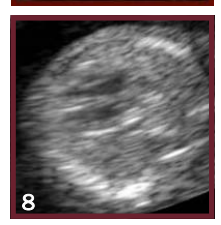
## FMF Recommendations

Though there are still no set guidelines as to the full anatomical survey at 12-14 weeks, using the NT for screening for aneuploidy is well established globally and has become incorporated into the healthcare system of all women in the UK, Denmark and most recently in the state of California. The Fetal Medicine Foundation (FMF) is the world authority on the first trimester scan and recommends that in a single mid-sagittal plane of the fetus, one can measure the NT, ascertain the presence of the NB, measure the FMFA and look at the intracerebral translucency (IT), a first trimester marker for spina bifida (Chaoui 2009), check for TR and DV flow. In addition, the intactness of the cranium can be noted ruling out acrania, the butterfly sign of the choroid plexus can be seen ruling out holoprosencephaly and the retro-nasal triangle can be noted to rule out early facial clefts (Sepulveda 2004 and 2010). The arms and legs, cord insertion and intactness of the abdominal wall can be noted. The fetal situs, basic 4 chamber view, presence of the stomach, kidneys and bladder with a 3 vessel cord may also be checked. Basic fetal biometry in addition to the crown rump length can be obtained to properly date the pregnancy. In addition, placental localization, cord insertion and umbilical artery Dopplers may be measured.



## Anatomical Components Examined at the FTS

- Mid-sagittal view for NT, IT, NB, FMFA (1)
- Sagittal view of the fetal spine (2)
- Localize the placenta and cord insertion (3)
- Visualize the butterfly sign (4)
- Check for choroid plexus cysts (4)
- Measure the BPD and HC (5)
- Examine the orbits (6)
- Visualize the upper lip (7)
- Examine the 4CV and look for EIF (8)
- Check the kidneys and for pyelectasis (9)
- Ascertain the presence of 2 legs (10)
- Ascertain the presence of 2 arms (11)
- Visualize the cord insertion (12)
- Visualize the bladder with a 3v cord (13)
- Visualize the stomach (14)
- Check for echogenic bowel (14)
- Measure flow in the DV (15)
- Visualize AoA and right atrial inflow tracts (16)

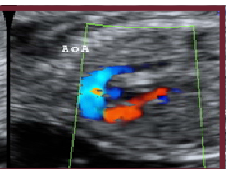
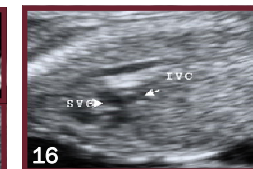
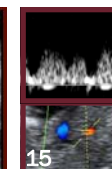
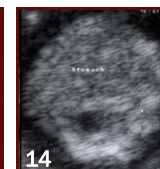
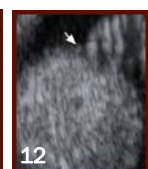


## Planes Used For Fetal Measurements/Biometry

- NT and NB (1)
- IT and FMFA (1)
- BPD and HC (5)
- Femur length (10)
- Abdominal circumference (14)
- Doppler of the DV (15)

## Practical Tips

Aim way beyond just a CRL and fetal heart tones. Just as one started with the NT, with practice, to those with a thorough knowledge of second trimester anatomy, expanding the first trimester exam to incorporate the NB, IT, FMFA, TR, DV, PI of the uterine artery, genetic markers as well as ascertaining the normalcy of all the organ systems will become a possibility. Though a first trimester scan by no means replaces the second trimester scan, it is a most powerful tool in providing early reassurance to the family against the major structural abnormalities that can be easily ruled out at 12-14 weeks. This will soon become inevitable, so the time to start practicing is NOW!



## CFAFC's Literary Recommendation: Prenatal Diagnosis 1st Trimester Screening & Fetal Therapy Special Issues

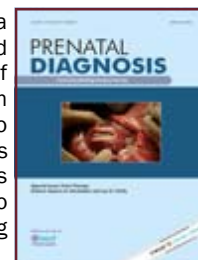
Journal of the International Society for Prenatal Diagnosis: [www.ispdhome.org](http://www.ispdhome.org)

Editor-in-Chief: Diana W. Bianchi, MD Volume 31, Issues 1 and 7, January and July 2011.

<http://onlinelibrary.wiley.com/doi/10.1002/pd.v31.1/issuetoc>

<http://onlinelibrary.wiley.com/doi/10.1002/pd.v31.7/issuetoc>

These special 2 issues of "Prenatal Diagnosis" are edited by Lyn S. Chitty, Tze Kin Lau and Kypros Nicolaides. They provide a comprehensive overview of first trimester screening and fetal therapy with several editorials in addition to original articles and thorough reviews. Professor Nicolaides provides a concise as well as a futuristic editorial with his call to "invert the pyramid of obstetrical care". All major maternal and fetal contributors to morbidity and mortality are addressed in the January issue: from aneuploidy to congenital malformations to miscarriage and stillbirth to fetal growth abnormalities to prematurity to multiplicity to maternal preeclampsia and endocrine disorders. The July issues presents lessons learnt in fetal therapy and ethical issues faced. It also reviews anomaly specific interventions and the various aspects of fetal therapy. The entire content of both issues is available for free download at the two links provided on top. CFAFC highly recommends these invaluable current references to anyone interested in an updated overview on the ever evolving role of screening in the first trimester, as well as the promising current and futuristic role of fetal therapy.



## THIS AND THAT

### 21st World Congress on Ultrasound in Obstetrics and Gynecology

18 - 22 September 2011 | Los Angeles, USA



This year, the 21st World Congress of the International Society of Ultrasound in Obstetrics and Gynecology is taking place in Los Angeles, California from September 18-22. Several exciting pre-congress workshops are being offered on September 17 and 18 on Fetal Echocardiography, Fetal MRI, Just Images in both Obstetrics and Gynecology, Doppler and Neurosonography. In addition to the rich program and live scan sessions, there will be several workshops and "Meet the Professor" sessions. For details visit [www.isuog.org/WorldCongress/2011/](http://www.isuog.org/WorldCongress/2011/)

### Fetal Medicine Foundation Online Free Risk Calculators



The fetal Medicine Foundation has made available free online calculators to determine a patient's risks for preterm delivery, preeclampsia, gestational diabetes, small or large for gestational age fetuses as well as early intrauterine demise and late still birth. Here the provider can input historical, sonographic as well as biochemical markers to calculate the risks based on assessment at 12 weeks. In order to access the calculators visit [www.fetalmedicine.com/fmf/online-education/08-pyramid-of-care/](http://www.fetalmedicine.com/fmf/online-education/08-pyramid-of-care/)

### AIUM Annual Convention



The annual convention for the American Institute of Ultrasound in Medicine is planned for March 29- April 1, 2012 in Phoenix, Arizona. Abstract submission is now open through September 28, 2011. For more information, please visit [www.aium.org](http://www.aium.org).

### Hot-Off-The-Press: Organ Targeted Prenatal Gene Therapy-How Far Are We? by Mehta, Abi-Nader, Waddington and David. Prenatal Diagnosis 2011; 31 (7): 720-734 <http://onlinelibrary.wiley.com/doi/10.1002/pd.2787/pdf>

Fetal gene therapy (FGT) aims to deliver genes to genetically defective cells, allowing correction before long term damage has occurred. Prenatal application has a number of advantages. Most important of which is that, early gestation gene delivery may result in the development of fetal immune tolerance to the transgenic protein, potentially leading to long-term gene expression and the possibility of phenotypic cure. This has been shown to be true in proof of principle studies in animal models such as the haemophilic mouse. Ultrasound-guided delivery strategies have been devised in large animals that could be used clinically to apply gene therapy to the human fetus in certain life-threatening congenital disorders. This includes for example intratracheal and intragastric injections to target the lungs and gut in cystic fibrosis and, intramuscular injection for congenital muscular dystrophies. FGT provides parents with a third choice where currently the only options are either terminating the pregnancy or continuing with an affected fetus. A number of issues will need however to be resolved before translation into clinical practice, including long-term safety and efficacy as well as regulatory and ethical questions.

### SMFM Annual Meeting



The annual meeting of the Society of Maternal Fetal Medicine is scheduled for February 6-11, 2012 in Dallas, Texas. Abstract submission is now open. For more information, visit [www.smfm.org](http://www.smfm.org). To submit your abstracts please go to <http://www.softconference.com/Subs/SMFM/2012/CFP/default.asp>.

### Annual Congress of the Lebanese Society of Obstetrics & Gynecology



Mark your calendar's for the annual meeting of the Lebanese Society of Obstetrics and Gynecology, set to take place at the Movenpick Resort on November 17-19. An exciting program is planned with distinguished international speakers discussing topics spanning all aspects of our specialty. For further details and to stay updated as to our society's activities, please visit [www.lsog.org.lb](http://www.lsog.org.lb).





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CFAFC would like to thank Drs. Abi-Nader and Yamak for their contributions to this issue.



For any interesting case reports, comments, suggestions or announcements regarding upcoming conferences, please send an e-mail to rar@cfafc.org.

## First Trimester Diagnosis of Hexalogy of Cantrell Reem S. Abu-Rustum, MD and Randa Yamak, MD

Pentalogy of Cantrell constitutes: midline anterior ventral wall defect, a cleft distal sternum, a diaphragmatic defect, a defect of the apical pericardium with communication to the peritoneum and an intracardiac defect (Allan 2008). It is invariably fatal. Recently, associated umbilical cord defects have been reported suggesting a hexalogy (Muller Brochet 2009).

A 25 yo G1P0 was referred for evaluation of a fetal anomaly at 14w2d by dates. The fetus was 12w4d by size.

A hydroptic fetus with severe kyphoscoliosis (Figure 1) was present. The fetal measurements were consistent with 12w4d and the nuchal translucency was 7.7mm. In addition, a large ventral wall defect (Figure 2) containing heart and stomach (Figure 3) was seen. The 4 chamber view was also abnormal suggestive of a single ventricle noted on color Doppler (Figure 4). The fetal cord was noted to have a single umbilical artery on color Doppler, providing evidence for a hexalogy.

The findings were discussed with the family. Given the poor prognosis, they opted to terminate and declined any further genetic testing or postmortem examination.



Figure 1



Figure 2



Figure 3



Figure 4

## Upcoming Congresses

COURSE TITLE	DATES	LOCATION	WEBSITE ADDRESS
13th World Congress of Ultrasound in Medicine and Biology	August 26-29, 2011	Vienna, Austria	www.wfumb2011.org
Patient Safety in Obstetrics 2011: Reducing Risk and Improving Outcomes	September 8-10, 2011	Las Vegas, Nevada	www.edusymp.com/meetingview.asp?productid=4365
21th World Congress of the International Society of Ultrasound in Obstetrics and Gynecology	September 18-22, 2011	Los Angeles, California	www.isuog.org/WorldCongress/2011/
9th International Scientific Meeting RCOG	September 28-30, 2011	Athens, Greece	www.erasmus.gr/en/congresses/athens/2011/rcog_2011/welcome/
Obstetrical Ultrasound in the High Risk Patient	October 14-16, 2011	Las Vegas, Nevada	https://iame.com/conferences/hr12/
Second Annual Fetal Echocardiography: Normal and Abnormal Hearts	October 28-29, 2011	Las Vegas, Nevada	www.edusymp.com/meetingview.asp?productid=4283
Ultrasound Meets Magnetic Resonance at the Louvre	October 28-29, 2011	Paris, France	www.ultrasound2011.org/index.aspx
National Conference on Ob/Gyn Ultrasound	November 4-6, 2011	Chicago, Illinois	https://iame.com/conferences/ob14/
Annual Meeting of the Lebanese Society of Obstetrics & Gynecology	November 16-19, 2011	Beirut, Lebanon	www.lsog.org.lb
14th Mid-Atlantic Ultrasound Symposium	November 18-19, 2011	Virginia Beach, Virginia	www.evms.edu/continuing-medical-education/continuing-medical-education.html
20th Annual Ob/Gyn Update for Clinical Practice	December 8-11, 2011	Fort Lauderdale, Florida	www.cmebyplaza.com/Registrants/GoHo10/About.aspx