A Pictorial Essay/Literature Review

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The Ever Evolving 11–14-Week Scan

About the Authors

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ABSTRACT

Advancements in ultrasound equipment have led to improved resolution of smaller structures. In turn, this has allowed for the evolution of the 11–14-week scan. At its inception, the first-trimester scan was used primarily to indicate the risk for aneuploidy in a fetus. However, as time passed, the ability to detect structural abnormalities complemented the risk for chromosomal abnormalities. This literature review will discuss the evolution of the 11–14-week scan up to current-day recommendations for population screening.

Keywords: 11–14-week scan; screening; aneuploidy

Introduction

Before the 1990s, amniocentesis was the gold standard method for detecting a chromosomal abnormality. Maternal age was used as a determining criterion for amniocentesis as it was an established risk factor for Trisomy 21 (Down syndrome), the most common chromosomal abnormality in infants. By age 35 the estimated incidence of Trisomy 21 is 1 in 350 live births. This increases to 1 in 200 live births by age 40.[1] Amniocentesis is associated with a risk for miscarriage, so there was the impetus to develop a non-invasive screening method for Trisomy 21. Research showed that an increased nuchal thickness correlated with an increase in aneuploidy, and its measurement could be performed on an ultrasound image of the fetal profile.[2] The 11–14-week scan (initially called the 11–13+6 week scan)[2] was developed in the 1990s to aid in the screening for aneuploidy, precisely Down syndrome. In the following 10 years, the scan evolved to encompass screening for other trisomies (13, 18) and Turner's syndrome (X0). The current first-trimester screen incorporates biochemical markers, nasal bone status, and the nuchal translucency (NT) measurement to ascertain the risk for aneuploidy. The addition of a standardized

checklist of fetal anatomy would permit the detection of congenital structural abnormalities, making the first-trimester screening more complete.

Evolution of the First Trimester Scan

The 11–13+6 week scan was introduced as a screening tool for Trisomy 21 (Down syndrome) in the 1990s. The scan included assessing the maternal environment, crown-rump length (CRL) measurement, fetal heart rate (FHR), chorionicity if multiples, and measurement of the NT.

In 2000, Nicolaides et al. determined the efficacy of the 11–13+6 week scan combined with maternal age to provide predictive screening for Trisomy 21 with a 75% detection rate.[2] They further demonstrated that with the addition of serum biochemical markers (free-beta-human chorionic gonadotropin [βHCG] and pregnancy-associated plasma protein-A [PAPP-A]), the detection rate for chromosomal abnormalities increased to 90%.[2] A large NT measurement (>3.5 mm measured when CRL is between 45 mm and 84 mm)[2] was also shown to be associated with other chromosomal abnormalities, syndromes, heart defects, defects of the great vessels, and fetal skeletal dysplasia.

Nicolaides et al. also showed that standardization of the method of measurement of the NT was essential for its' reproducibility and developed the Fetal Medicine Foundation (FMF London, UK) NT measurement criteria and the 11–13 weeks training/certification program, which has been widely adopted (https://fetalmedicine.org/education/the-11-13-weeks-scan). Figure 1 is an example of an NT measurement scan, showing the FMF criteria. The image was obtained ML on a fetus in a neutral position. The image is magnified to include only the chest and head. The nasal bone, skin line anterior to the nasal bone, nose tip, mandible tip, palate, intracranial translucency, and diencephalon are all demonstrated. The NT is measured at the thickest point with the calipers placed perpendicular to the NT, crosshairs "on" the borders of the NT.

For centers offering NT for aneuploidy screening using the FMF risk algorithm, the NT scans should only be performed by sonographers with FMF UK certification who comply with ongoing yearly audits to ensure the requirements were adhered to.[2]

As of 2003, evidence supported a correlation between fetal nasal bone ossification at 11–13+6 weeks

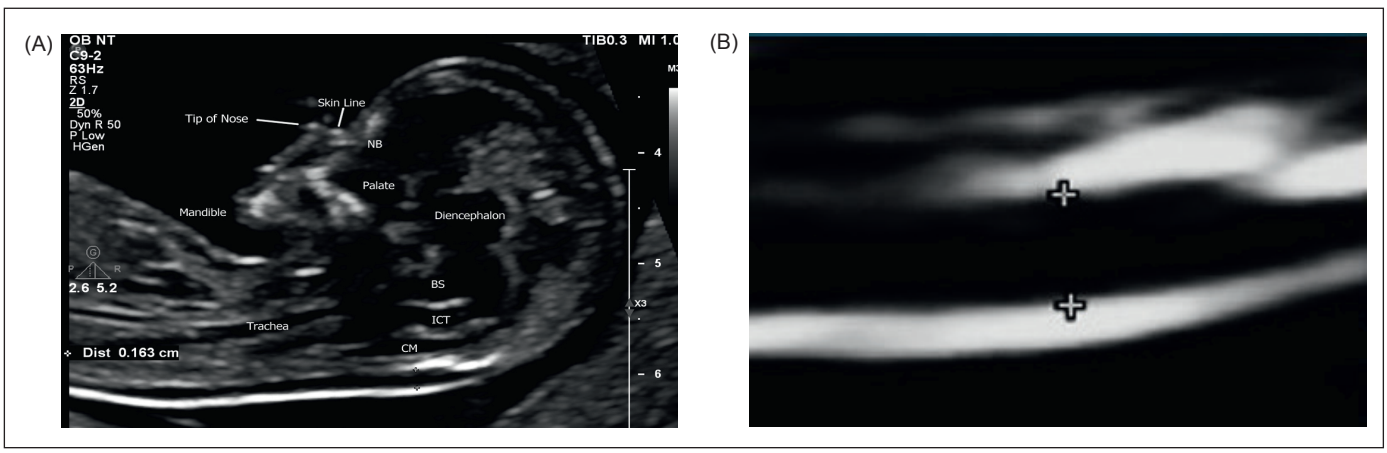


Figure 1. (A) The NT measurement should be taken on a ML sagittal image with the nasal bone seen (should not see zygoma) magnified so only head and thorax are in image. Head should be in neutral position (flexion or extension will alter measurement). Amnion should be visualized. Demonstration of the brain stem (BS), intracranial translucency (ICT), cisterna magna (CM), nasal bone, upper palate, tip of the nose, ML spine are required for an accurate measurement. (B) Calipers placed perpendicular to the nuchal translucency with the cross hairs "on" borders of NT.

and fetal trisomy. Cicero and others showed that the nasal bone was absent or hypoplastic in 60–73% of fetuses with Trisomy 21, 53–57% Trisomy 18, 32–45% Trisomy 13 and 9% of Turners syndrome at the 11–13+6 week scan.[3,4] Increased sensitivity for demonstration of the nasal bone was achieved if the assessment was performed after 12 weeks gestation.[3,4] As with NT, the FMF UK has developed a training and certification program for nasal bone assessment to ensure standardization of technique and compliance with ongoing quality assurance, allowing sonographers to become certified in nasal bone imaging in conjunction with NT licensing. https://fetalmedicine.org/education/the-11-13-weeks-scan. The MFM recommended protocol for the 11–13+6 week scan is displayed in Table 1.

From 2005-2007 evidence supported the additional evaluation of fetal anatomy at the time of the 11–13+6 week NT scan to detect major structural anomalies.[5] Improvements in ultrasound imaging provide better resolution of fine structures. By expanding the fetal anatomical survey during the first trimester, it is possible to detect approximately 50% of major abnormalities.[5]

In 2013 a systematic review of the literature for 11–14 week scans found the overall detection rate of fetal structural anomalies was 51% when a basic anatomy scan was included. This improved to 62% with the addition of endovaginal ultrasound.[6] Detection rates were improved to 67% when the patient had a known increased risk for abnormality (e.g., large NT).[6] The authors identified those factors that had a proven influence on screening performance. Changing the time frame to 12–14 weeks improved the detection rate even further. The inclusion of a standardized protocol, use of endovaginal scanning when visualization is suboptimal, sonographer training, and quality of equipment were all identified as factors that would improve detection rates with these scans.[6]

ISUOG (International Society of Ultrasound in Obstetrics and Gynecology) published the first practice guidelines the same year for the first-trimester anatomy scan.[7,8] The basic anatomy recommended on first-trimester ultrasound scan to detect not only aneuploidy risk but also structural anomalies is outlined in Table 2. Figure 2 demonstrates this anatomy.

Additional complementary papers detailed which anomalies should always be detectable, anomalies that are detectable approximately 50% of the time, and those that are not likely to be demonstrated in the first-trimester scan if anatomy is assessed. Syngelaki et al. provided a breakdown of anomalies and their relative probability of detection on a first-trimester anatomy scan (Table 3, Figure 3).[7]

Table 1. Current Protocol for 1st Trimester Screening Ultrasound

Protocol for the Nuchal Scan (First Trimester Screening Ultrasound)
Biometry: Crown Rump Length, Biparietal Diameter
Fetuses/Chorionicity if >1
Fetal Heart Rate
Placenta, Cervix, Adnexa
Amniotic Fluid Volume (qualitative assessment)
Nuchal Translucency
Nasal Bone

Table 2. Basic 1st Trimester Anatomy Protocol Recommended by ISUOG.

Basic 1T Anatomy Protocol (present / absent)
Cranium, Choroids, Profile
Midline Falx
Nuchal Translucency
Chest/Heart (situs)
Stomach
Cord Insertion (abdomen)
Bladder
Extremities • 3 segments (+hands/feet)
Placenta

Ultrasound Obstet Gynecol 2013; 41: 102-113.

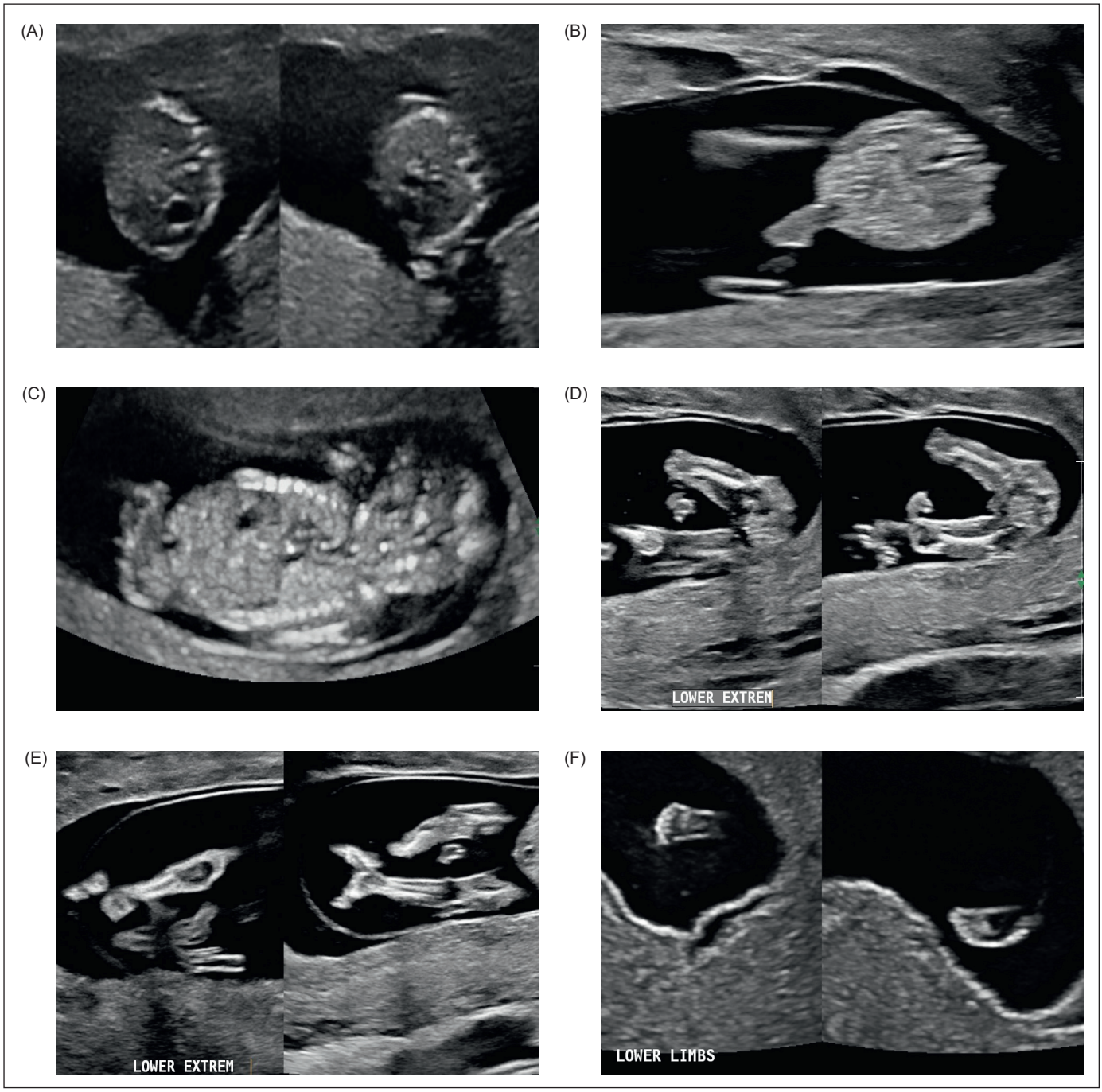


Figure 2. Anatomy recommended for First Trimester Basic Anatomy Scan. (A) Stomach in left quadrant, (B) Intact abdominal wall with cord insert, (C) Bladder in fetal pelvis (coronal image bladder/stomach/heart/lungs/diaphragm), (D,E,F) Major bones of lower extremity.

ISUOG updated its recommendations in 2019, but the only pertinent change was a recommendation that basic fetal anatomy should be reviewed whenever obstetric ultrasound is done at 11–14 weeks, while women with increased risk of fetal structural and genetic abnormalities can be offered enhanced screening if performed by ultrasound providers with appropriate imaging expertise.[9,10] The list of basic anatomy to be reviewed was unchanged from the 2013 guidelines.

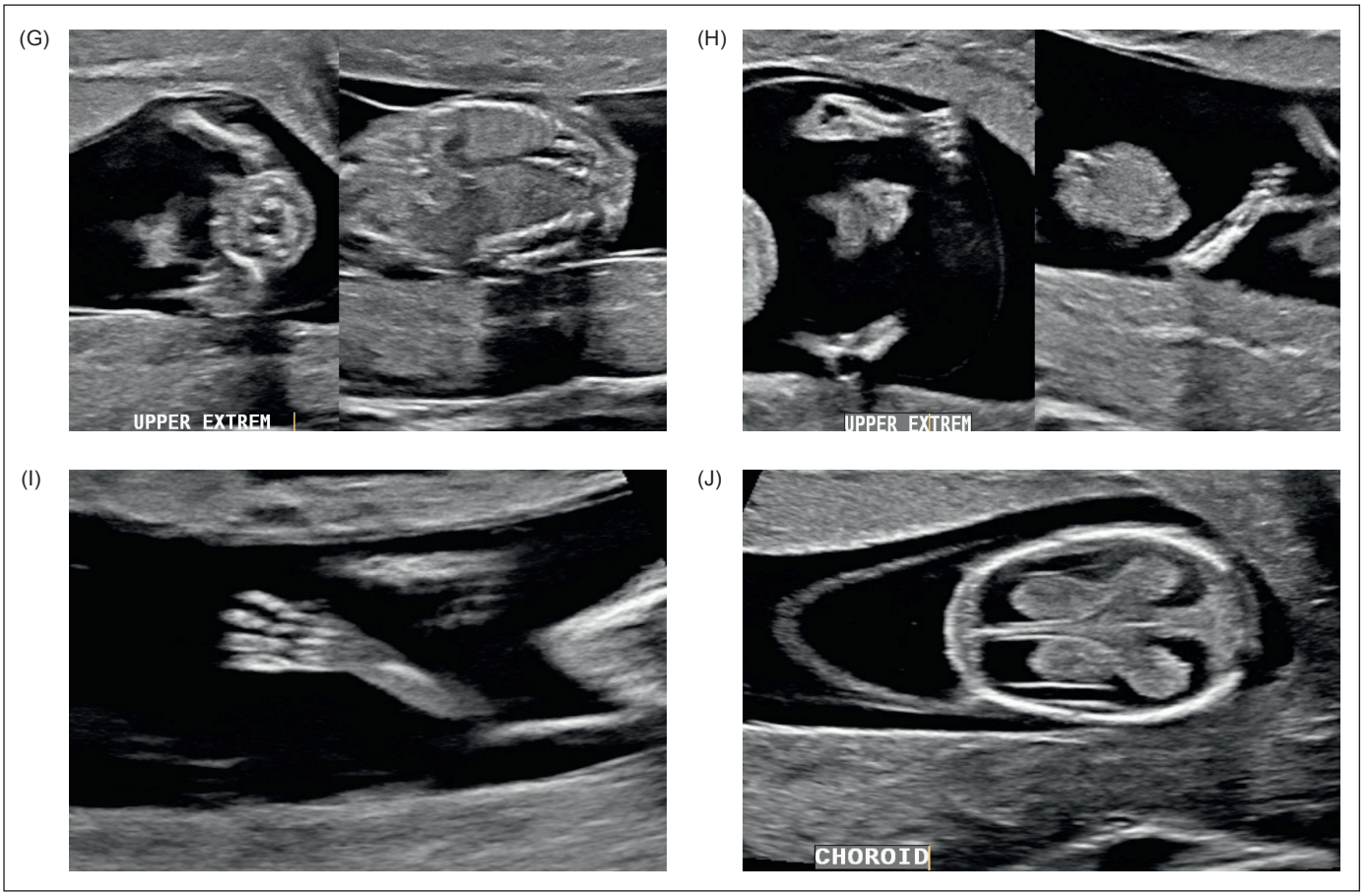
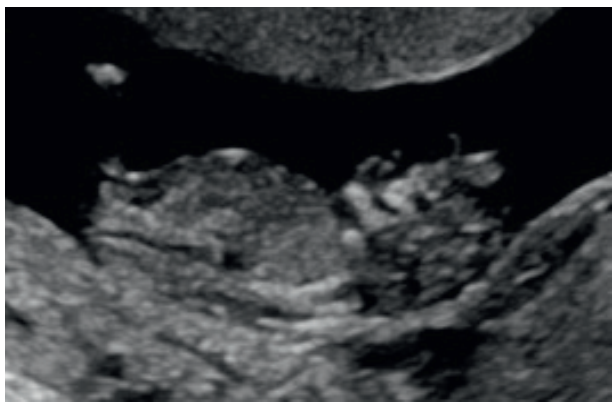


Figure 2. (G,H,I) Major bones of upper extremity, (J) Cranial ossification, choroid plexus.

Table 3. Breakdown of Anomalies Diagnosable, Potentially Diagnosable, and Likely Not Diagnosable In the First Trimester If Anatomy Scan Performed.[7]

Always Diagnosable (100%)	Potentially Diagnosable (~50%)	Likely NOT Detectable (<10%)
Anencephaly	Open Spina Bifida	Ventriculomegaly
Alobar Holoprosencephaly	Lower urinary obstruction	Agenesis of corpus callosum
Encephalocele	Fetal akinesia sequency	Isolated cleft lip
Pentalogy of Cantrell	Lethal skeletal dysplasia	CPAM
Ectopia Cordis	Dandy Walker malformation	VSD
Abdominal wall defects	Major heart defects	Unilateral renal agenesis, multicystic kidney, hydronephrosis, duplex kidney
Megacystis	Diaphragmatic hernia	Hypospadias
Phocomelia	Polydactyly	Talipes
Body Stalk Anomaly		

(A)



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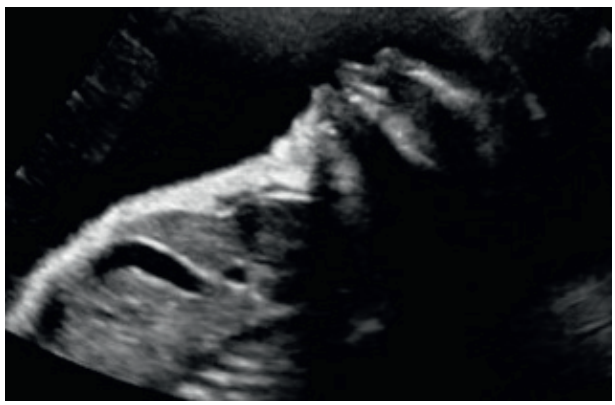

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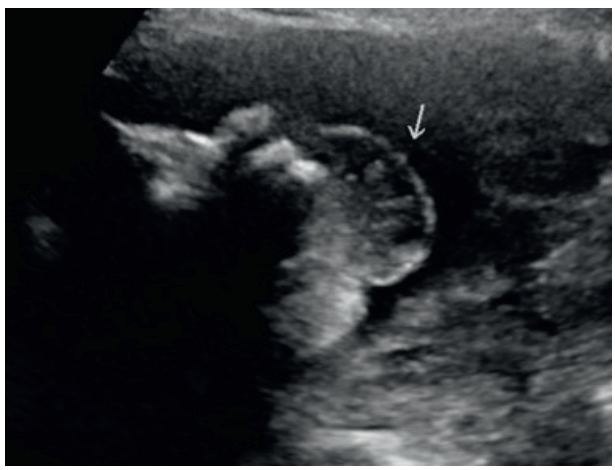

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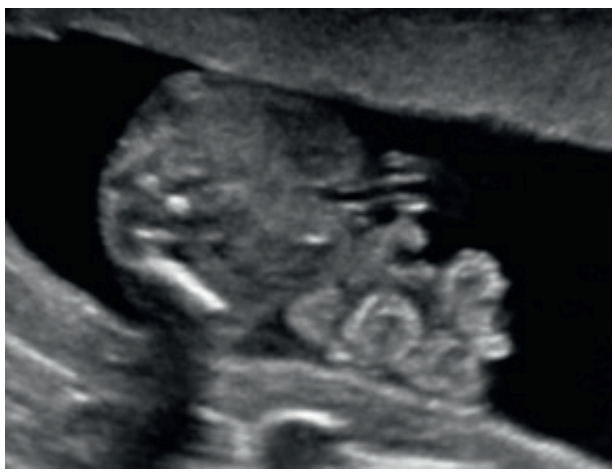

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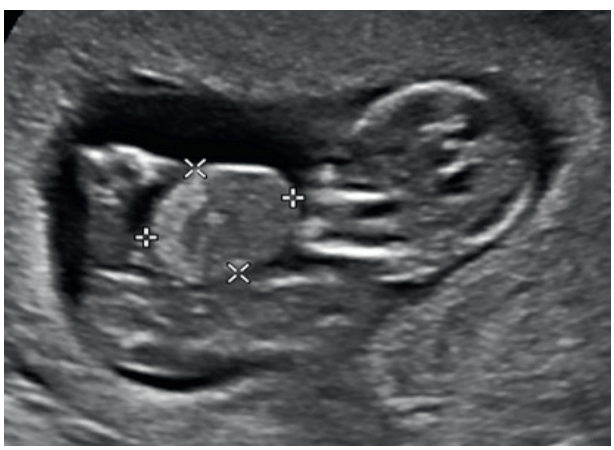

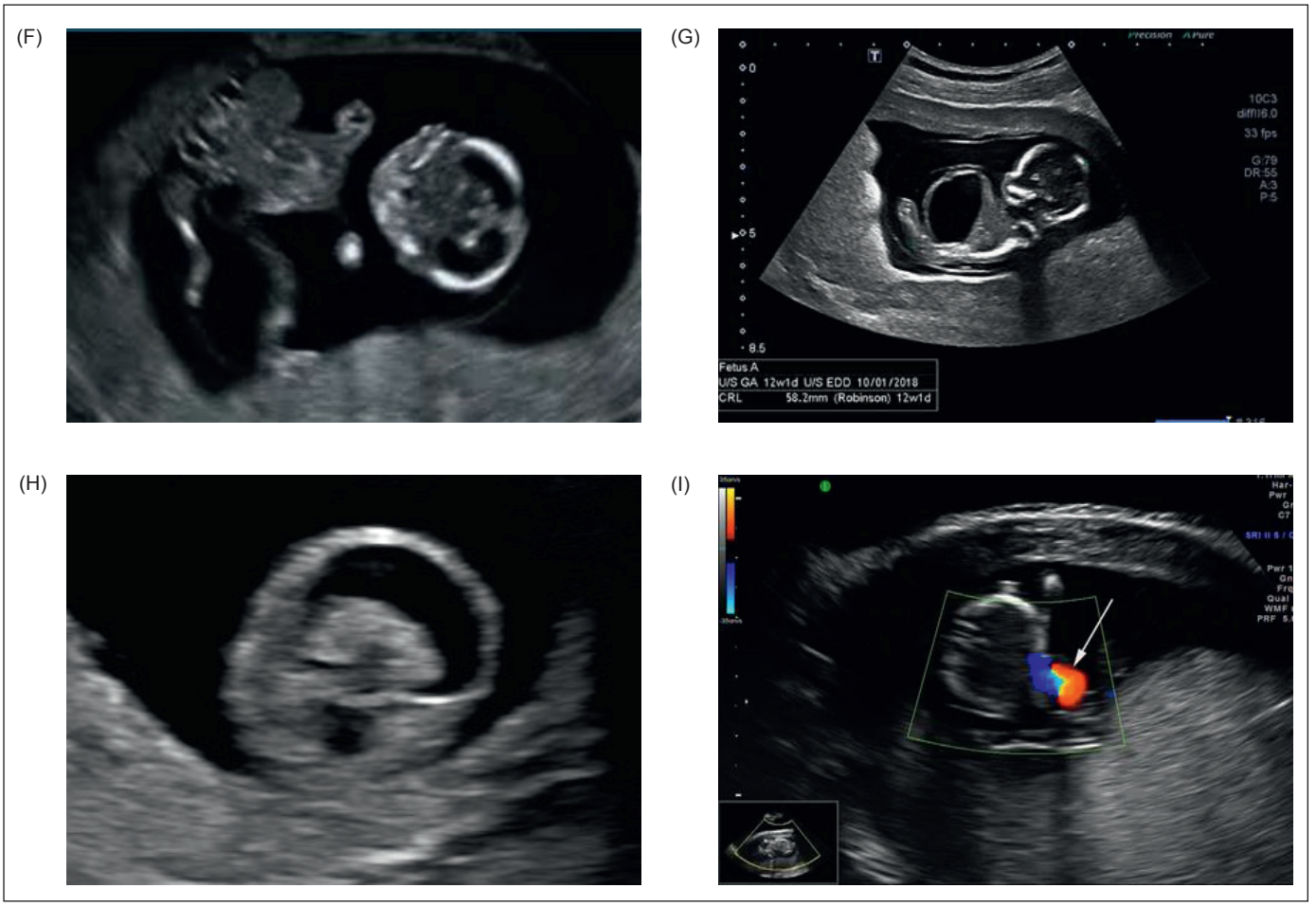

Figure 3. 9 Major anomalies are always detectable if an anatomy scan is performed in first trimester. (A) Acrania, (B) Anencephaly, (C) Encephalocele, (D) Gastroschisis (E) Omphalocele. (F) Body Stalk Anomaly, (G) Megacystis, (H) Holoprocencephaly, (I) Ectopia Cordis.

The most recent ISUOG practice guideline, published January 2023, sets a new recommended 2-tiered approach to the 11–14-week scan. The first tier utilizes the previously recommended anatomy scan as the minimum required testing assessment.[11–15] These minimum requirements are listed in Table 4. Image 4 illustrates the additional images needed in combination with those in Figure 2 to complete the minimum requirements for the 11–14 week scan. Abnormal findings from the first-tier scan or high-risk maternal risk factors would reflex the patient to the second-tier scan. This additional scan is a comprehensive and detailed assessment of the fetus that should be performed by technically qualified personnel at MFM or tertiary care centers. Recommended second-tier anatomy is outlined in Table 5.[15] Additional recommendations from the practice guideline include an assessment of uterine artery Doppler as a marker for increased preeclampsia risk and biochemical testing recommendations detailing appropriate usage[15] (outside the scope of this review).

Current Standard in Canada

Current standards in Canada recommend the first-trimester ultrasound be used for: dating, an indication-based early anatomic review, the NT measurement for aneuploidy, multivariable preeclampsia (PE) risk assessment, and use of open neural tube defect (oNTD) markers for screening where expertise and resources exist.[10] These standards do not include which anatomy should

Table 4. ISUOG New Guideline 2023 Minimum Scan Criteria for 11-14 Week Scan, Optimal Visualization After 13 Weeks.[15]

Minimum Requirements for Scan at 11-14 Weeks Gestation	
Anatomic Region	**Minimal Requirement for Scan**
General	Confirm singleton pregnancy
Head and Brain	Axial View of head to demonstrate: Calcification of cranium, contour/shape of cranium (no bony defects), 2 brain halves separated by interhemispheric falx, choroid plexuses almost filling lateral ventricles in their posterior two-thirds (butterfly sign)
Neck	Sagittal view of head and neck – confirm whether nuchal translucency thickness <95th percentile
Heart	Axial view of heart at level of four chamber view – heart inside chest with regular rhythm
Abdomen	Axial view – stomach visible, intact abdominal wall. Axial or sagittal view – bladder visible and not dilated
Extremities	Visualize four limbs, each with three segments
Placenta	Ascertain normal appearance without cystic structures
Biometry	Sagittal view – crown-rump length and nuchal translucency thickness Axial view – biparietal diameter.

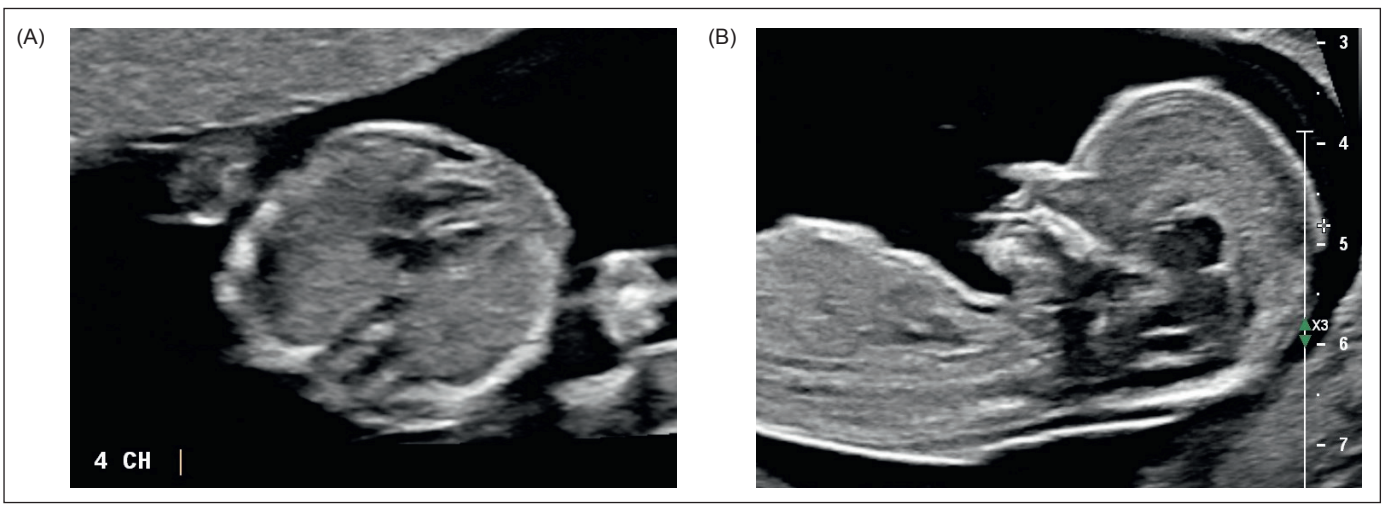

Figure 4. Additional images recommended by ISUOG 2023 guideline for minimal anatomy First Trimester Anatomy Scan. (A) Axial 4-Chamber Heart, (B) Profile (head/neck).

be included in the anatomic review. Equity of care across Canada is poor as many 11–14 week ultrasound exams are still performed using the protocol developed and implemented in 2003 (see Table 1). Other first-trimester providers have adopted the inclusion of some or all anatomy dictated by department protocols; there is no consistency across local, provincial, or national levels. As a result, all patients are not getting the same standard of care.

Discussion

Performing a first-trimester anatomy scan has determined clinical utility for multiple purposes. Patients with abnormal findings can be referred to a tertiary care/MFM site for a timely comprehensive scan. This maximizes the options parents have for pregnancy management; earlier access to genetic testing/counselling, additional time to consider termination of pregnancy (TOP), and earlier access to TOP.[13] An earlier anatomy scan can reduce maternal anxiety, especially in high-risk patients. Another advantage is with patients with increased BMI. Obesity is a risk factor for fetal anomalies and is known for decreasing the completion rate for routine ultrasound exams in the second trimester. Before the uterus leaves the maternal pelvis the fetus can more easily be scanned beneath

Table 5. ISUOG New Guideline 2023, Anatomical Structures That Can Potentially Be Visualized on Detailed Fetal Scan at 11-14 Weeks Gestation (in sagittal, axial and coronal view as needed).[15]

Detailed Fetal Scan Criteria	
Anatomic Region	**Minimal Requirement for Scan**
General	Confirm singleton pregnancy Overview of fetus, uterus and placenta
Head and Brain	Calcification of cranium, contour/shape of cranium (no bony defects), 2 brain halves separated by interhemispheric falx, choroid plexuses almost filling lateral ventricles in their posterior two-thirds (butterfly sign), thalami, brainstem, cerebral peduncles with aqueduct of Sylvius, intracranial translucency (fourth ventricle), cisterna magna
Face and Neck	Forehead, bilateral orbits, nasal bone, maxilla, retronasal triangle, upper lip, mandible, nuchal translucency thickness, no jugular cysts in neck
Thoracic	Shape of thoracis wall, lung fields, diaphragmatic continuity
Heart	Heart activity present with regular rhythm, establish situs, position – intrathoracic heart position with cardiac axis left (30-60 degrees), size – one third of thoracic space, 4 chamber view with 2 distinct ventricles on grayscale and color doppler in diastole, left ventricular outflow tract view on grayscale and color doppler, 3 vessel and trachea view on grayscale and color doppler, absence of tricuspid regurgitation/antegrade ductus venosus A-wave on pulsed-wave Doppler
Abdomen	Stomach – normal position in left upper abdomen, Bladder – normally filled in pelvis (longitudinal diameter <7mm), abdominal wall – intact umbilical cord insertion, two umbilical arteries bordering bladder, kidneys – bilateral presence.
Spine	Regular shape and continuity of spine
Extremities	Upper and lower limbs, each with three segments and free movement
Placenta	Size and texture normal without cystic appearance, location in relation to cervix and to previous uterine c-section scar, cord insertion into placenta.
Amniotic fluid and membranes	Amniotic fluid volume, amniotic membrane and chorion dissociated physiologically

a maternal pannus. The first-trimester anatomy scan objectives can usually be achieved with the ability to perform transvaginal ultrasound in combination with transabdominal scanning. When a first-trimester anatomy scan has been performed, the interpreting physician can consider the first and second-trimester anatomy scans to evaluate all imaging criteria. It is often easier to see the fetal profile and extremities in the first trimester than during the second trimester's routine exam. Finally, the entire fetus can be imaged in one view. A midline sagittal image of the fetus can assess as many as 12 different criteria in one image (CRL, profile, NT, Nasal bone, Maxilla, Mandible, Mid-brain, Bladder, Cord insertion, Extremities, Fetal sex, and Ductus venosus) (Figure 5).[13]

Non-invasive screening based on cell-free DNA (NIPS) has emerged as the best screening test for common trisomies (21, 13, 18). NIPS detects chromosomal abnormalities; it does not evaluate for congenital structural anomalies. Hence, whenever NIPS is done, it should be complemented by a first-trimester anatomy scan. While NIPS can be done before or after the first-trimester scan, there are certain advantages of offering it after the first-trimester scan. This is because NIPS may not be indicated if a large NT or fetal anomalies are detected due to the significant association with atypical chromosome abnormalities not detectable by NIPS. Either of these scenarios should elicit immediate referral for the comprehensive scan at tertiary care/MFM, allowing the patient to be provided the appropriate genetic counselling and testing options. In addition, structural anomalies (2–3%)[13] are far more common than chromosomal anomalies (1:700 overall incidence).[13] Performing the scan first ensures triage of patients

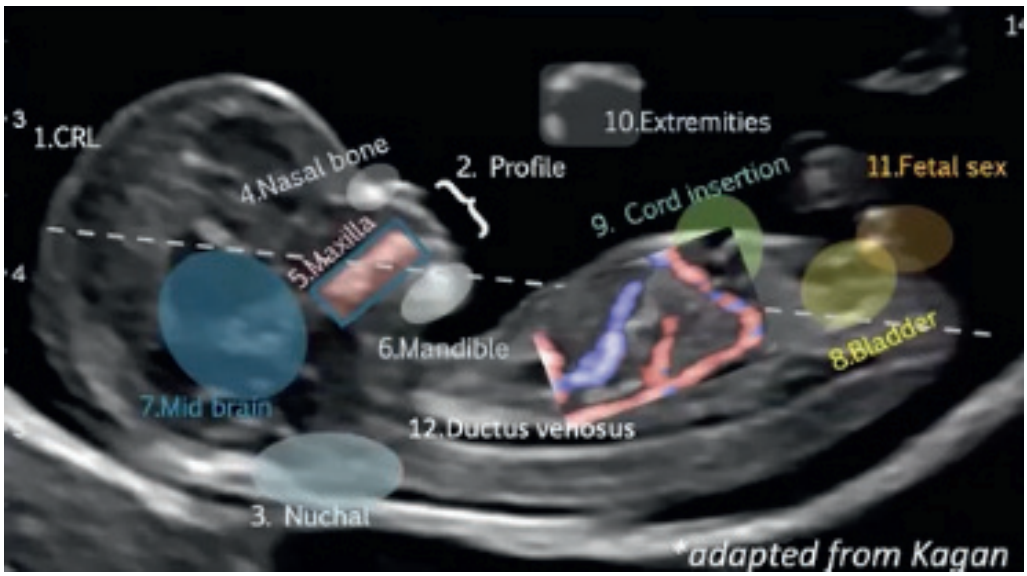

Figure 5. Demonstrates the fetal anatomy demonstrated on a good midline sagittal image of the entire fetus. This anatomy includes (1) CRL, (2) profile, (3) Nuchal translucency, (4) Nasal bone, (5) Maxilla, (6) Mandible, (7) Mid brain, (8) Bladder, (9) Cord insertion, (10) Extremities, (11) Fetal sex, (12) Ductus venosus. Hoopmann M, Kagan O. The Fetal Profil. Ultraschall in Med 2017; 38: 611–618.

to the appropriate subsequent testing (i.e., invasive testing amniocentesis or CVS versus NIPS). This approach also has the potential to be cost-effective, expedite referral to MFM/tertiary care, and decrease the incidence of undue parental expectation from negative NIPS result.

Conclusion

Including the basic minimal anatomy scan in first-trimester prenatal screening protocols will improve the detection of anomalies. This will offer parents more informed choices earlier in pregnancy. In addition, it will provide better utilization of NIPS-based cell-free DNA testing. In addition, ultrasound imaging will be better optimized for obese patients with the benefit of improved visualization of imaging criteria while the uterus is best positioned for imaging, and with the use of transvaginal sonography. Finally, it allows for the evaluation of imaging criteria from first and second-trimester anatomy scans, improving the completion rate of anatomy criteria and the subsequent management of the pregnancy.

Acknowledgement

Images were graciously provided by MIC Medical Imaging Edmonton AB through Cathy Ridsdale and the University of Calgary Fetal Medicine Department through Dr. Jo-Ann Johnson.

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