Prenatal Screening and Diagnosis

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Welcome 😊

- Thank you!

- Purpose
  - Update and education on prenatal screening and diagnosis, FATC process
    - May be completely new, review, refresher

- Outline
  - Important topics
  - Break
  - Opportunity for questions
Prenatal Screening

- Every pregnancy at risk for structural defect or chromosome abN ~2-3%

- Major cause morbidity and mortality

Our role:

- Assess risk
- Accurate method
- Provide nondirective counseling
- Offer informed choices about options for screening and diagnosis
Prenatal Screening

- Individualised risk assessment
  - Maternal age
  - Gestational age
  - Previous pregnancies
  - Prior screening in current pregnancy
  - Gives a ‘risk/odds’ for outcome – not diagnostic
Beginning the Process…
Beginning the Process…

- Pre-Test Counselling
- Patient History
  - Personal
  - Pregnancy
  - Family
What Testing to Offer
What Testing to Offer

- EPR + EMST + SMST + US
- EMST + SMST + US
- SMST + US
- Refer to MMGS
Pregnancy Dating

Great dilemma!!!!
Dating—What’s the big deal?

Why is it so important?

- Prenatal Screening-Timing of testing
  - Offering tests at appropriate time for accurate results
  - Being able to offering tests in the right window, so not to miss opportunity
  - False + results due to inaccurate dates
    - patient anxiety, extra US
  - Unnecessary travel/US due to wrong dates
  - Repeat US for patients
    - not good use of resources in already overbooked FATC
    - difficult for patients to travel/miss work
Dating - What’s the big deal?

- Why is it so important? Other reasons
  - Growth concerns
    - too big, too small for ‘dates’
    - extra US
  - Preterm labour/PPROM
    - Management decisions
      - Transfer to IWK?
      - Steroids?
      - Neonatal intervention?

- Post dates

- Timing of CS bookings
Pregnancy Dating-Basics

- **LMP**
  - First day of last normal period
  - Accurate if
    - **Certain** of date
    - **Regular** menstrual cycles with normal cycle length
    - Correct for long or short cycles
Pregnancy Dating

■ IVF
  ■ Need to know
    ■ Date of embryo transfer
    ■ Day 3 or 5 embryo
    ■ Patient’s own egg
      ■ Age of patient at time of retrieval
      ■ ‘fresh’, ‘frozen’
    ■ If donor egg
      ■ Age of donor at time of retrieval
    ■ ICSI-yes, no
Pregnancy Dating

- Ultrasound dating - most accurate 1st T

  - Consider using if

    - Uncertain LMP
    - Abnormal / irregular menstrual cycles
    - < 3 cycles since last pregnancy
    - < 3 cycles since discontinuation of hormonal contraception

  - Ideal 8-10 weeks - accurate and timely

  - SOGC recommends dating US on all patients, however not good use of resources, and not feasible in the Maritimes*, so choose wisely
Pregnancy Dating

- Ultrasound dating for FATC referral…
  - Recognize limited DI resources, however…
    - Inaccurate dates->repeat US in FATC, repeat trip, anxious, unhappy patients, frustrated clinicians

- If LMP and US are discrepant…STOP…rethink the certainty of dates, as IUGR is often a feature of aneuploidy..call for guidance if unsure

- Critical to include copy of the US if done!!!!!
EPR
EPR

- Overview
- Eligibility
- Availability
Maternal Serum Testing
First Trimester
Maternal Serum Testing Requisition

Specimen to be taken on ...
(9-13^{\text{th}} \text{ weeks gestation})

\[ \frac{\text{dd mm yy}}{\text{dd mm yy}} \]

Patient Information
Prov. Health Card No. __________________ Date of Birth \[ \frac{\text{dd mm yy}}{\text{dd mm yy}} \]
Name __________________ Phone __________
Racial Origin □ Caucasian □ Black □ Other (specify)

ACCURATE PATIENT INFORMATION IS ESSENTIAL FOR VALID INTERPRETATION

Clinical Information
Patient's current weight Kg lbs

Date of last menstrual period \[ \frac{\text{dd mm yy}}{\text{dd mm yy}} \]
If an ultrasound has been performed, please give date and measurements ...
NT _____ mm CRL _____ mm EGA (by U.S.) _____ wks _____ days

Does the patient have preexisting insulin dependent diabetes? □ No □ Yes
Is this a known multiple gestation? □ No □ Yes

Physician Prenatal testing should proceed only with the informed choice of the patient
Name __________________ Signature __________________
Address Perinatal Centre 5980 University Ave.
Hfx N.S. B3H 4N1 Phone __________________

For Laboratories' Use (shipping instructions on reverse)
Originating Laboratory: __________________
Spec. Number: __________ Spec. Date: \[ \frac{\text{dd mm yy}}{\text{dd mm yy}} \]
For IWK Health Centre Lab Use
EMST

- Communication of results
- Screen +EMST-next steps
- Dating Fax
- EPR following screen+ EMST
Screen +EMST

Testing options
- FATC
  - SMST
  - NIPT
  - CVS
  - Amnio
NIPT

- Background
- Eligibility
- Cost
- Experience to date
Second Trimester
Maternal Serum Testing Requisition

Specimen to be taken on ...
(15-20 6/7 weeks gestation)

Patient Information
Prov. Health Card No. ____________________ Date of Birth
________________
Name _____________________________ Phone ________________
Racial Origin ☐ Caucasian ☐ Black ☐ Other ____________
(specify)

ACCURATE PATIENT INFORMATION IS ESSENTIAL FOR VALID INTERPRETATION

Clinical Information
Patient’s current weight ________ Kg ________ lbs

Date of last menstrual period
( / / )

If an ultrasound has been performed, please give date and measurements ...
Date ( / / )

CRL ________ mm BPD ________ mm EGA (by U.S.) ________ wks ________ days

Does the patient have preexisting insulin dependent diabetes? ☐ No ☐ Yes
Is this a known multiple gestation? ☐ ☐

Physician  Prenatal testing should proceed only with the informed choice of the patient

Name ___________ Signature __________________________
Address 544 Perinatal Centre
5980 University Ave.
Hfx N.S. B3H 4N1

For Laboratories’ Use (If shipping instructions on reverse)

Spec. Number: ________ Spec. Date: ( / / )

For IWK Health Centre Lab Use

Originating Laboratory:

Phone Direct Line Preferred

FAX __________________________

For Laboratories’ Use (If shipping instructions on reverse)
SMST

- Screen + SMST-next steps
- Dating Fax
- Testing options
  - FATC
  - Genetic sonogram
  - NIPT
  - Amnio
MST

- Communication of results
- +/- Booking appointments
Genetic Sonogram
Genetic Sonogram

- ‘FATC’ism’

- Detailed anatomy US + ☺
  - Thorough and complete anatomic review
  - Assessment of soft markers
  - Detailed review of the fetal heart structure and function

- Purpose
  - More detailed review than screening US in at risk patients
Genetic Sonogram

- Who is a candidate?
  - Determined by MFM
  - Ideal 20-21 weeks to optimize complete exam
    - +MST
    - Structural abN seen on US
    - Previous pregnancy complication

- Interpretation?
  - If complete exam, all markers assessed, no abN, and views excellent…
    - Risks reduced for particular condition (2-5 fold)
  - If incomplete exam, poor views-risks will not be adjusted
Soft Markers

- ‘Variation’ of normal
- Commonly seen in healthy babies, however, ‘associated’ with particular fetal aneuploidy
- Each marker has a particular risk (likelihood ratio)

Important Points
- All markers are not equal
- If marker identified – risk increased
- If no markers identified – risk decreased
- If marker seen on DI US
  - Isolated or multiple markers
  - Offer MST if not done
  - Will be triaged as per marker(s)/MST, risk
All markers are not equal

- Nuchal fold
- Echogenic bowel
- Hypoplastic/absent nasal bone
- Short long bones
- Echogenic focus
- CPCs
Markers

- Take home points
  - All markers not equal
  - Sometimes not ‘seen’ in FATC
  - Risk adjusted based upon US
  - Isolated vs multiple markers
Echogenic Focus
Absent Nasal Bone
Increased Nuchal Fold
Echogenic Bowel
Pyelectasis
Soft Markers

Next steps
FATC Referral Process…
Who? What? When? How?
FATC Philosophy and Aims

- ‘Excellence’ in Obstetrical Ultrasound 😊

- Provide Tertiary High Risk US services for the Maritimes
  - Routine, urgent and emergent services in ‘at risk’ women

- Prenatal screening and diagnosis

- Non-directive counselling and options in complicated pregnancies
FATC Philosophy and Aims

- Tertiary High Risk US services for Maritimes
  - Maternal disease/complications of pregnancy
  - Fetal complications
  - Pregnancies at risk

- Happy to see anyone, however,
  - aim to utilize resources most appropriately, efficiently, and effectively
  - optimize patient care
  - Improve maternal and fetal outcomes
The “Who” of FATC

- MFM Physicians
- Sonographers
- Prenatal Screening and Diagnosis coordinator
- Support Staff
The “What” in FATC

- Prenatal Screening
- Fetal Anomalies
- Obstetric Complications
- Well being assessment
- Diagnostic Testing
- Treatment and Procedures
- Support and counseling
FATC Facts

- MFM Physicians see and scan all clinic patients
- Nurse sonographers perform BPP with MFM support
- Always MFM physicians in FATC for advice/consultation
FATC Facts

- Patient population
  - 20,000 FATC visits/year
  - 5000 Anatomy US
  - 4200 BPP
  - 1800 Nuchal Translucency US
  - 1300 Transvaginal US
  - 400 Multiples
  - 150 Amniocentesis
Helpful Tips for Referrals to FATC

- Our Biggest Challenges…
"You've gotta help me! I can't read my own writing!"
Referral for ???

Don’t write like this.
Clarity…
Clarity…

DO NOT ENTER

Enter Only
Timeliness...

- Gestational age
- ‘Slow’ to get to us
- Delay in making referral
Missing Information...

- Dating!!!!!
- Previous US
- Blood Type
- Clear indication for referral
- BMI
- Results of previous testing
All leads to FATC Booking ‘Cycle of Doom’

Waiting for missing information

Doctor and Patient Anxious for appointment!

Patient info doesn’t arrive so based on ‘dates’-1 day left for EPR

Patient arrives-10+2 weeks-no EPR can be done, so repeat appointment booked

Squeeze patient in already overbooked FATC clinic

Patient frustrated and anxious, FATC challenged
Try to book at the right time

- Who needs to come and when

FATC very busy
All the time!
One option…

Stress Reduction Kit

Bang Head Here

Directions:
1. Place kit on FIRM surface.
2. Follow directions in circle of kit.
3. Repeat step 2 as necessary, or until unconscious.
4. If unconscious, cease stress reduction activity.
Is it really an Emergency ???
In order for us to go

From this…

To this…
And you to go

From this…

To this…
Referrals to FATC

Helpful Tips
Legible Writing
Clarity…

- Clear indication for referral
- Complete information
Timeliness…

- Refer as soon as you know you need an US!!!!
- Be aware of timing of tests
  - NT
  - MST
  - Anatomy
- Calculate gestational age
Timeliness: FAX or Phone

Phone 470-6461  Fax 470-7987
Emergency ???

- Urgent/emergent unit responding to multiple requests/day

- Physicians prioritize in a timely fashion
  - Indicated US
  - Urgency
  - Is it an emergency?

- Complete information

- Call if emergency or unsure!
FATC Referral Checklist

- Dating/LMP/EDC
- US done in this pregnancy – Fax Copy
- Clear medical history indicating the reasons(s) for the requested referral
- Blood Type
- BMI/weight
FATC Referral Checklist

- Patient demographics
  - Health card #, province
  - Full legal name
  - Date of birth
  - Full mailing address
  - Home phone #

- Name of referring health care provider including fax #

- Fax referral to 470-7987
Triage Process

- Referrals received
  - Fax preferrable
  - Stamped with date received
- Referrals triaged by physician
  - Indicated
  - Type of US required
  - Timing
- Dr/Patient notified
  - Fax
  - Phone
New Referral Form

- This replaces any previous forms

- Please use this for ALL referrals regardless of indication from now forward

- Complete ALL aspects for referral

- Attach any additional information
New Referral Form

- Completing the form with all information will allow:
  - Appropriate and timely triage
  - Notification for patients of appointment times
  - Minimize delayed, late referrals
  - Inability to offer US due to missing information
Referral for Consult and Ultrasound
Fetal Assessment and Treatment Centre

Phone: (902) 470-6654  Fax: (902) 470-7987

K07002307  Jun/7/2002  M
SCA,TEST Visit
ER0000145/12  HCN: 22222222
Van den Hof, TEST / TEST, Maureen
Dec/8/2012

--- Please Complete All Fields ---

Patient Name_________________________________________  DOB (dd/mm/yyyy)____________________

Address ________________________________________________  HCN ___________________________

______________________________________________________  Phone Number _______________________

Referring Physician / Care Provider ________________________

☐ Gravida  ☐ Para  ☐ Abortus

LMP (dd/mm/yyyy)_________________________  Dates certain?  ☐ Yes  ☐ No

Has an ultrasound been performed in this pregnancy?  ☐ Yes  ☐ No

If 'Yes':
Date of U/S (dd/mm/yyyy)_________________________  Gestational Age at U/S ________ weeks ________ days

** Please attach copy of ultrasound

Patient Weight_______  BMI _______  Blood Type__________  ** Please attach copy of blood type

Reason for Referral:

________________________________________________________________________

________________________________________________________________________

________________________________________________________________________

________________________________________________________________________
<table>
<thead>
<tr>
<th>Referral Received</th>
<th>Patient to be seen:</th>
<th>Patient to be seen at:</th>
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<tbody>
<tr>
<td></td>
<td>□ ASAP</td>
<td>□ within ___ Days</td>
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<tr>
<td></td>
<td></td>
<td>□ within ___ Weeks</td>
</tr>
<tr>
<td>Triage Date (dd/mm/yyyy)</td>
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<td>□ Dating / Viability</td>
<td>□ Echo</td>
<td>□ Transvaginal Ultrasound</td>
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<td>□ Anatomy</td>
<td>□ Early Pregnancy Review</td>
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<td>□ Multiples</td>
<td>□ Growth</td>
<td>□ BPP</td>
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<tr>
<td></td>
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<tr>
<td>Appointment Date (dd/mm/yyyy)</td>
<td>Time (24 hour clock)</td>
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<tr>
<td>□ Physician Notified</td>
<td>□ Patient Notified</td>
<td></td>
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<tr>
<td>Date of Notification (dd/mm/yyyy)</td>
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<tr>
<td>Method of Notification:</td>
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<tr>
<td></td>
<td>□ Other</td>
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<tr>
<td>□ FATC not indicated</td>
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</table>
So, who needs a referral???

- Busy unit
- Optimize appropriate patients for FATC
- Aim to do what is feasible and appropriate in local DI
So, who needs a referral???

- EPR
- Anatomy
Nuchal Translucency (NT/EPR)

- Indications
  - Advanced Maternal Age (>35 at delivery)
  - Previously affected baby (chromosome abN/structural abN)
  - + Family History (first degree relative)
  - Inherited Disease
  - Congenital Heart Defect in Parent/Sibling
  - Pre-existing Diabetes
  - IVF Pregnancy
  - Known Multiple Gestation
  - ‘Exposures’
  - +1st trimester MST
  - Other…
Nuchal Translucency (NT/EPR)

- Not indications
  - Past history of structural abN not detectable at EPR
  - Family history of disorder in greater than 1st degree relative
  - ART-clomid, IUI, donor insemination, past IVF
  - Previous +MST in prior pregnancy
  - Declines MST after full counselling
Anatomy

- +MST
- IVF with ICSI
- Twins
- Structural AbN
- Significant Medical disorder or exposure
- Significant obstetrical history
- Inadequate views on routine screening US
Not indication for Anatomy in FATC (as sole indication)

- Negative MST
- IVF with no ICSI
- IVF with PGD
- >40 yo as a sole indication
  - with negative complete screen-DI appropriate
Summary

- FATC aims to be accommodating and accessible
  - Routine, urgent and emergent services in high risk women

- Want to offer efficient and optimal US services

- Tips for patient referrals
  - Referral Checklist
  - New referral Form
Summary

- “If in doubt, check it out”
- Fax referrals
- Phone if you feel it’s urgent
- Indications for US
- Know timing of testing
- “Trust the triage process’
  - FATC appointment scheduled at best/most appropriate time
  - If no news after 48 hrs in urgent pt-please call
- Patient may not need to be seen in FATC