



Prenatal Screening and Diagnosis

The Who? What? When? And How? Of it...

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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

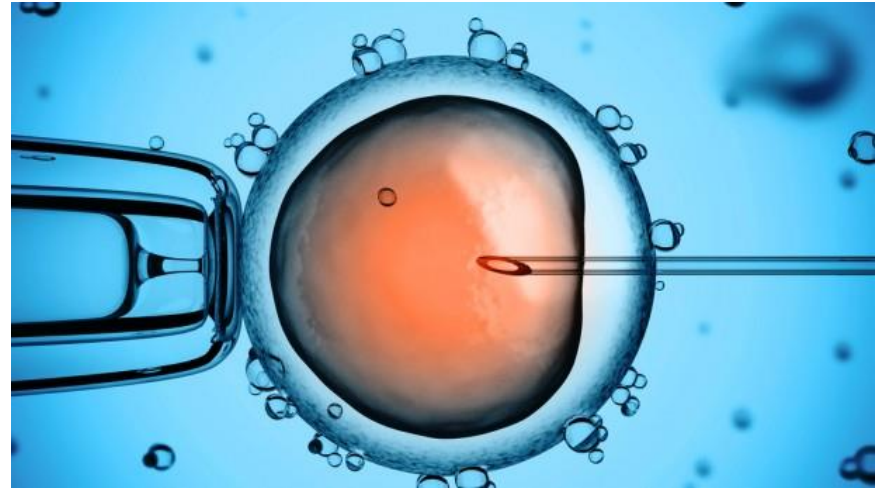
- First day of last normal period
- Accurate if
 - **Certain** of date
 - **Regular** menstrual cycles with normal cycle length
 - Correct for long or short cycles



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 ... / /
(9-13⁶⁷ weeks gestation) (dd mm yy)

Patient Information

Prov. Health Card No. _____ Date of Birth / /
(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 / /
(dd mm yy)

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

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?

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

Name 544 Signature _____

Address Perinatal Centre Phone _____
 5980 University Ave. Direct Line Preferred
 Hfx N.S. B3H 4N1 FAX _____

For Laboratories' Use (shipping instructions on reverse)

For IWKHealthCentre Lab Use

Originating Laboratory: _____

Spec. Number: _____ Spec. Date: / /
(dd mm yy)

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
-



SMST



Second Trimester Maternal Serum Testing Requisition

Specimen to be taken on ... / /
(15-20^{6/7} weeks gestation) (dd mm yy)

Patient Information

Prov. Health Card No. _____ Date of Birth / /
(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 / /
(dd mm yy)

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

CRL _____ mm BPD _____ mm EGA (byU.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 544 *Signature* _____

Address Perinatal Centre
5980 University Ave. Phone _____
Hfx N.S. B3H 4N1 **Direct Line Preferred**

FAX _____

For Laboratories' Use *(shipping instructions on reverse)*

For IWKHealthCentre Lab Use

Originating Laboratory: _____

Spec. Number: _____ Spec. Date: / /
(dd mm yy)

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

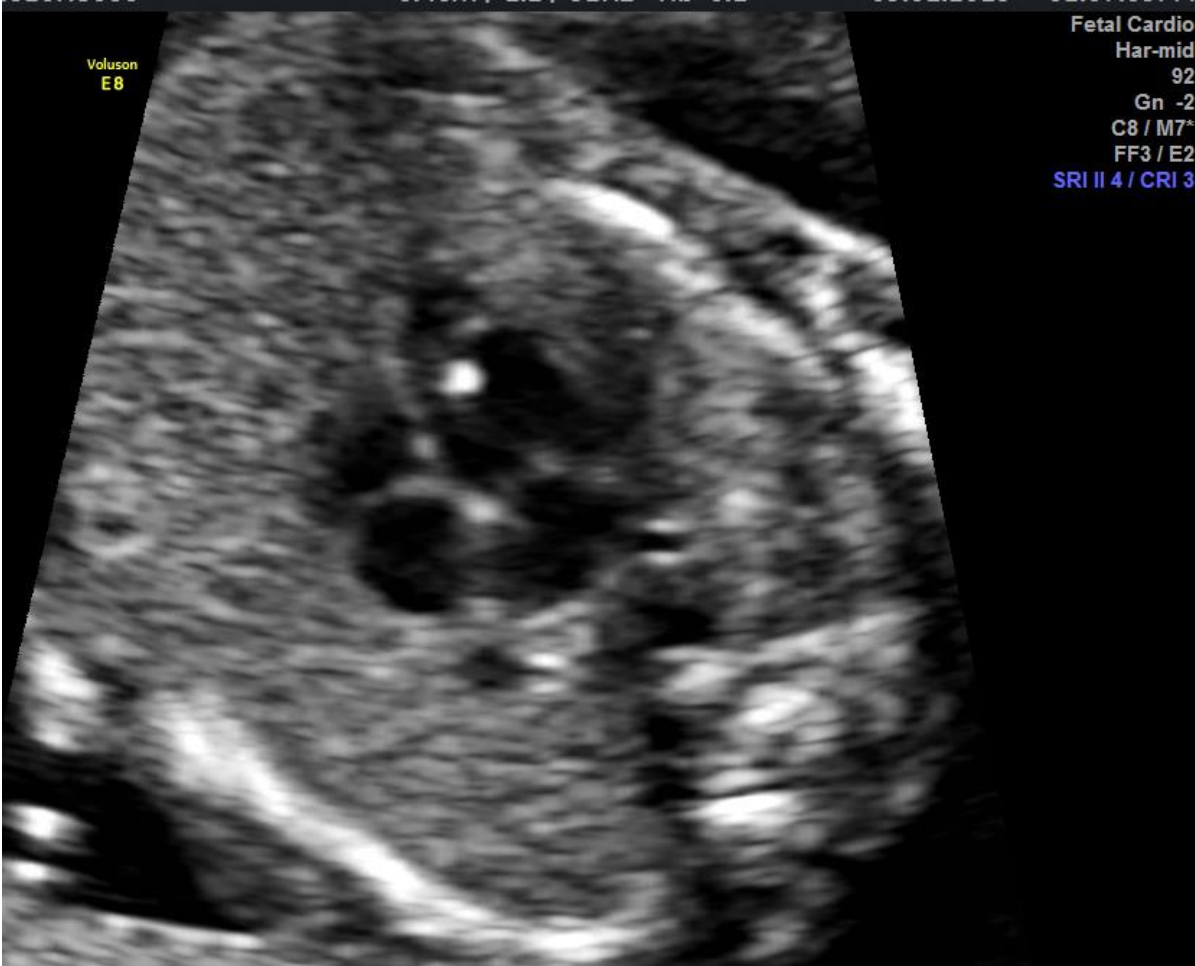
- Nuchal fold
- Echogenic bowel
- Hypoplastic/absent nasal bone
- Short long bones
- Echogenic focus
- CPCs



Markers

- Take home points
 - All markers not equal
 - Sometimes not 'seen' in FATC
 - Risk adjusted based upon US
 - Isolated vs multiple markers
-

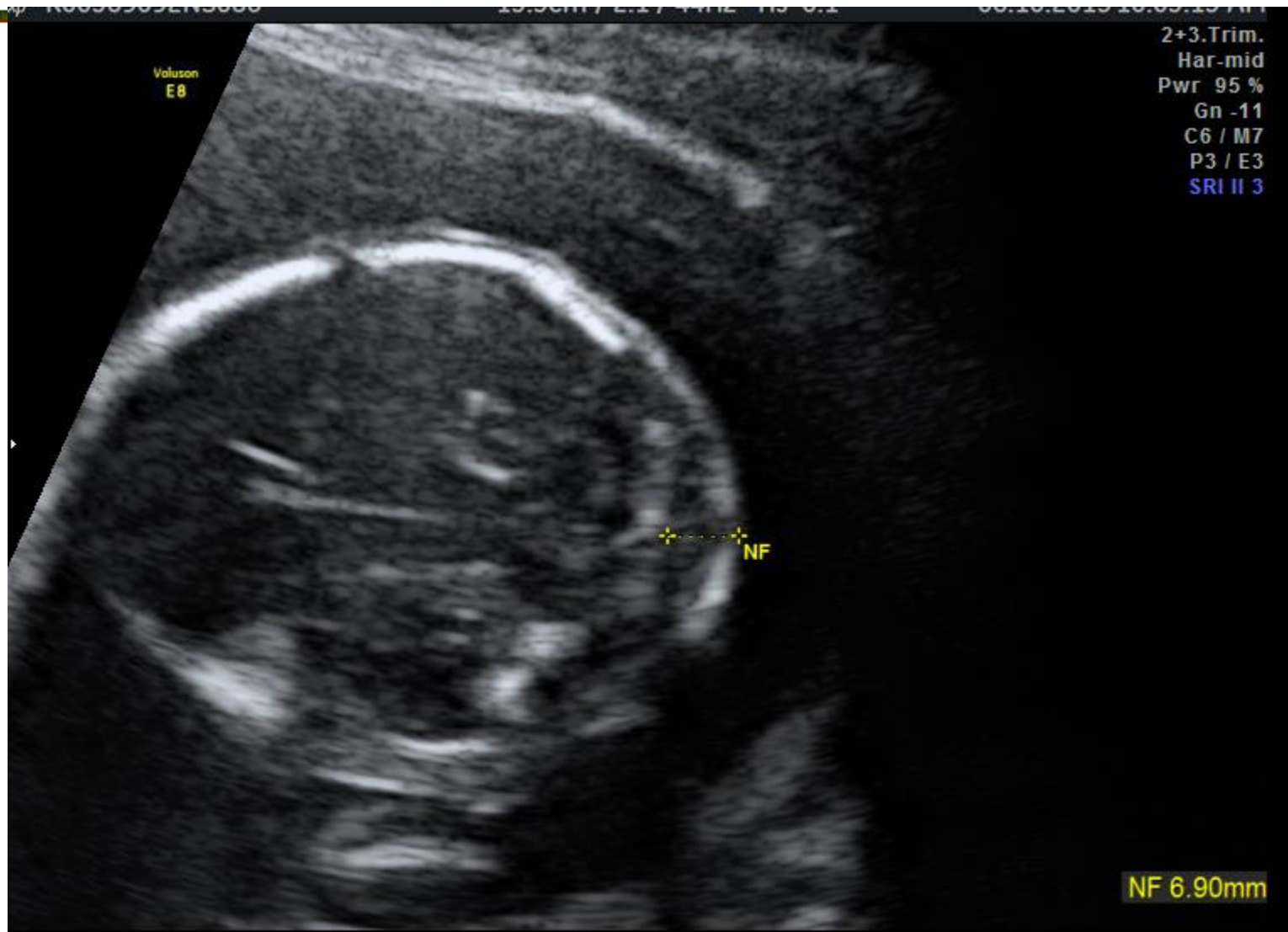
Echogenic Focus



Absent Nasal Bone



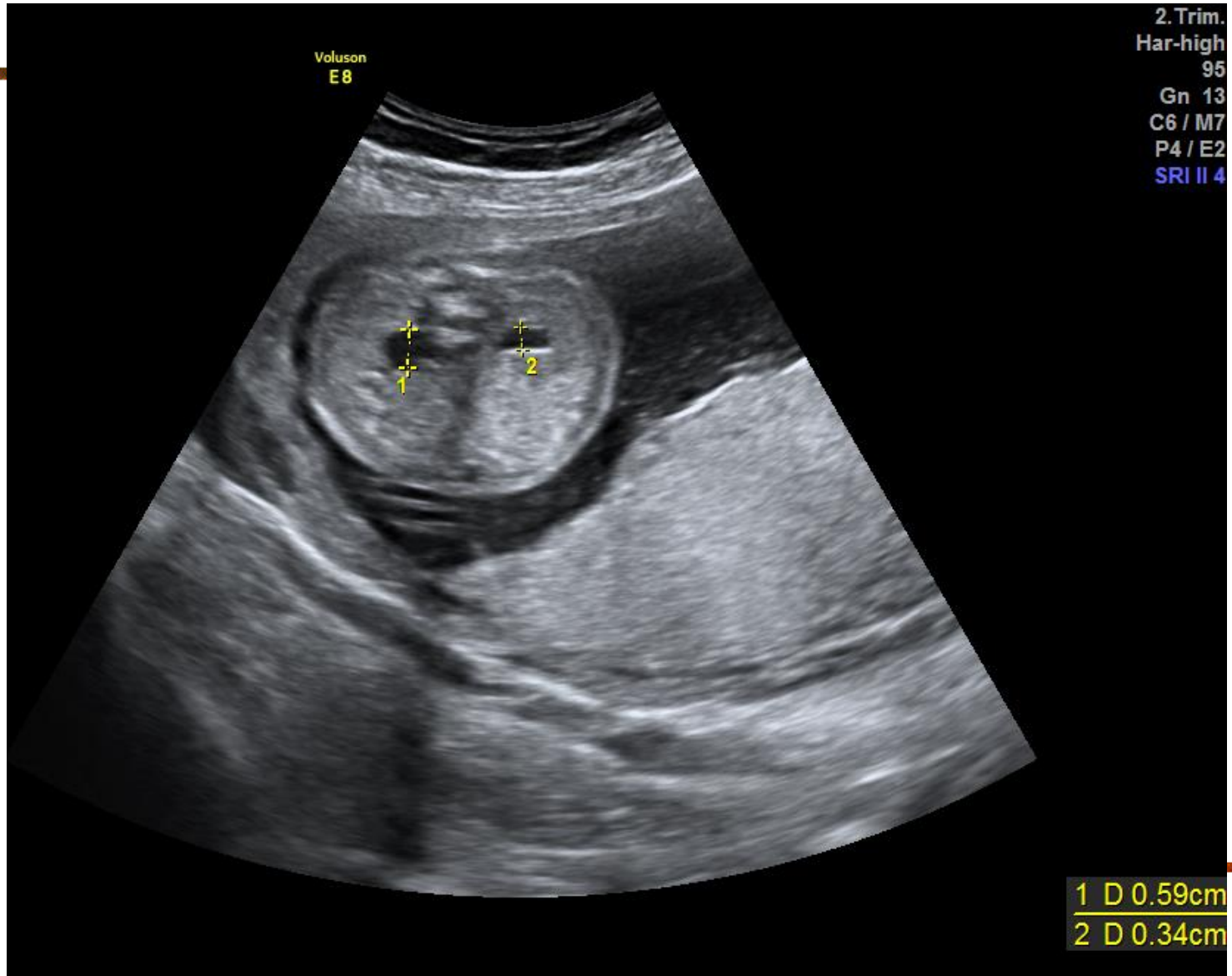
■ Increased Nuchal Fold



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
-

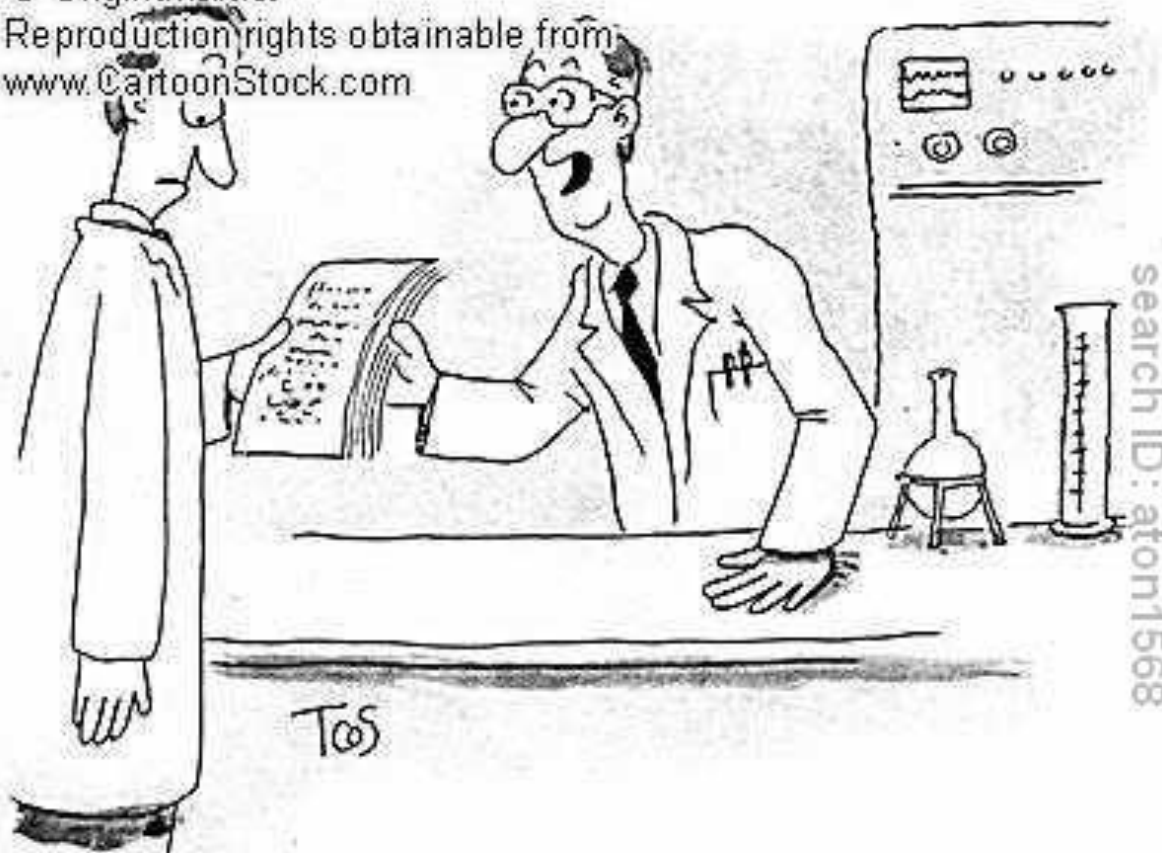
Helpful Tips for Referrals to FATC

- Our Biggest Challenges...



Cannot Decifer Writing

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search ID: aton1568

"You've gotta help me! I can't read my own writing!"

Referral for ???

23/11/90
Vene. Ad
de. to 2nd
by handypaper 2nd
but no p. 12th
no part
/

Don't write like this



Clarity...



Clarity...



Timeliness...

- Gestational age
- 'Slow' to get to us
- Delay in making referral

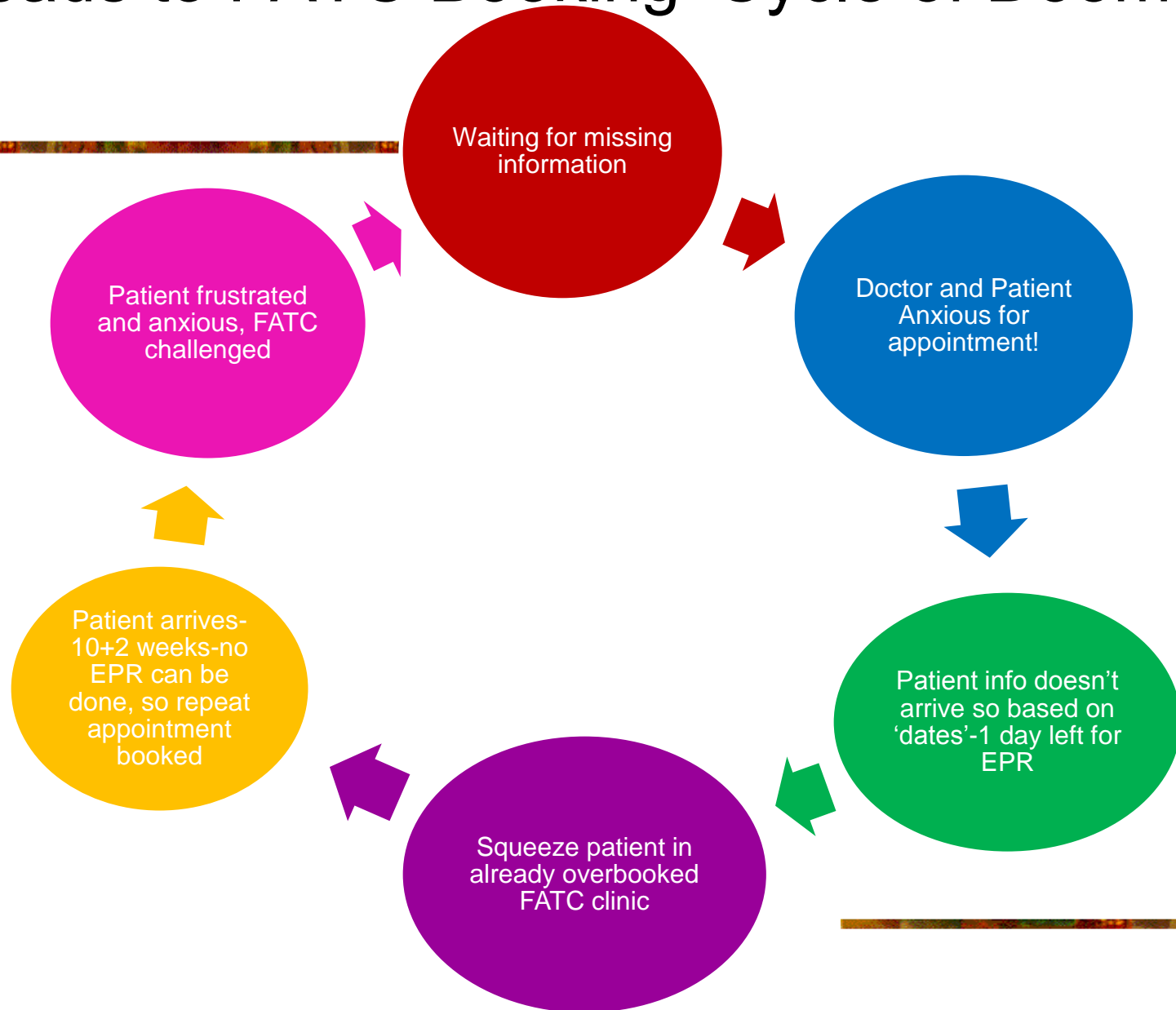


Missing Information...

- **Dating!!!!**
- Previous US
- Blood Type
- Clear indication for referral
- BMI
- Results of previous testing



All leads to FATC Booking 'Cycle of Doom'



Try to book at the right time

FATC very busy
All the time!

- Who needs to come and when



One option...

Stress Reduction Kit



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 preferable
 - 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:

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
-

