Case 41 Annie Ploidy
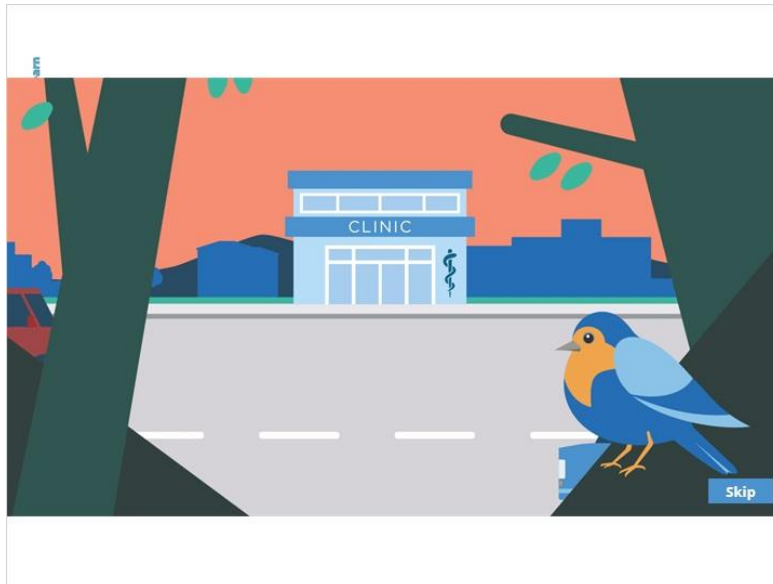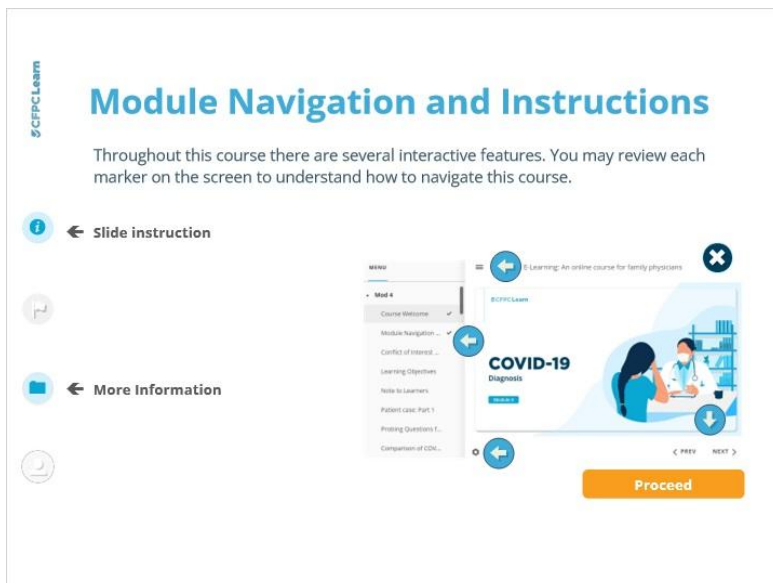
1. Conflicts

1.1 Intro Video

1.2 Module Navigation and Instructions

1.3 Learning Objectives

Learning Objectives

At the end of this module participants should be able to:

1. Identify key factors/findings in family medicine to help establish common clinical diagnoses.
2. Evaluate best evidence/guidance and practice tools to determine preferred treatment options for patients.
3. Synthesize patient preference, evidence and experience to formulate comprehensive plans for patient cases.

Approximately 15 minutes to complete each patient case.

Proceed

2. Annie Ploidy

2.1 Meet Annie Ploidy

Annie Ploidy, 36

Annie is a new patient to you. She has had a positive home pregnancy test.
Her last menstrual period was about seven weeks ago. She is 36 years old, G3P1, with a three-year-old child at home.
She is otherwise healthy and has started on prenatal vitamins about 6 months ago.

Conversation I   Conversation II

Moving Forward

Annie 1 (Slide Layer)

Annie 2 (Slide Layer)

2.2 Choose all that apply:

2.3 Review

First trimester (Slide Layer)

Ultrasound at 11-14 weeks (Slide Layer)

Second trimester ultrasound (Slide Layer)

First trimester serum markers (Slide Layer)

Different Provinces (Slide Layer)

Age (Slide Layer)

Review

First trimester U/S
First trimester U/S, II
Second trimester U/S
First trimester markers
Provincial variability
Screening Test
Age

- Age alone is considered poor minimum standard for referral for invasive fetal diagnostic testing. (SOGC 2017)

  - Ultrasound at 11-14 weeks and/or multiple marker serum screening advised depending on province as an initial test, depending on province.

Moving Forward

Screening Test (Slide Layer)

Review

Screening Test	Sensitivity* for Trisomy 21
First trimester serum markers alone	70
Nuchal translucency (NT) alone	71
First trimester serum and NT	87
First and second trimester serum	85-87
First and second trimester serum and NT	92-95
Second trimester serum alone (QUAD)	74-83

- *when specificity set at 95% (=false positive rate set at 5%). Specificity can be "set" by moving the threshold for a positive test in a test that has a continuous value.

2.4 Conversation

2.5 Which statements about cell-free DNA (cfDNA) testing or non-invasive prenatal test (NIPT) are most accurate?

(Multiple Response, 10 points, 1 attempt permitted)

2.6 Review

cfDNA (Slide Layer)

cost effective (Slide Layer)

Ontario cost analysis (Slide Layer)

Ontario cost analysis - II (Slide Layer)

sens,spec (Slide Layer)

Positive predictive value (Slide Layer)

Review

- cfDNA "is not a substitute for invasive for invasive testing" (SOGC 2017)
- Positive predictive value (PPV) decreases in lower prevalence populations (but same sensitivity and specificity).

	Sensitivity	Specificity	Age 25 PPV	Age 40 PPV
T21	99.3	99.8	33	87
T18	97.4	99.8	13	68
T13	91.6	99.6	9	57

Adapted from SOGC 2017, data from Obstet Gynecol 2014;126:e31-7.

Moving Forward

False Positive (Slide Layer)

Review

- Cell-free DNA
- Cost Effectiveness
- Cost analysis, I
- Cost analysis, II
- Sensitivity, specificity
- Positive predictive value
- False positives

False positives are from placental mosaicism where the placenta (the source of the fetal DNA detected in the maternal circulation) may have a chromosomal abnormality but the fetus doesn't.

Moving Forward

2.7 Recommendation

(Multiple Response, 10 points, 1 attempt permitted)

"Too bad that more accurate test can't just replace the amniocentesis. I was also reading that the miscarriage rate is high!"

Miscarriage rate with chorionic villous sampling is 3% and amniocentesis is 1 %.

☐ True

☑ False

Submit

2.8 Review

Review

Answer: False

Miscarriage Rates

Early invasive testing

Click on the buttons to view additional information

Moving Forward

Miscarriage Rate (Slide Layer)

Amniocentesis (Slide Layer)

2.9 Create a Plan

Let's Make a Plan

Using what you have learned write a plan for Annie. It can include any/all of the following: lifestyle intervention(s), prescription intervention(s), lab(s)/test(s) required, follow up appointment time frame, and referral required.

Fill in your plan for the patient here

See how your plan compares

2.10 Plan Answers

Annie Ploidy

This is the proposed plan for Annie. How does your plan compare?

Serum Testing

Ultrasound

Select details on the left to see suggested recommendations

What's in a name?

Serum Testing (Slide Layer)

Ultrasound (Slide Layer)

Name (Slide Layer)

2.11 References