

# Implementation of First Trimester Genetic Screening at Whittier Clinic

Thanks to the "Steering Committee for First Trimester Genetic Screening"  
Nancy Newman, Lourdes Pira, Michelle Karsten, Alicia Erickson, Anne Zachman

ACOG recommends that all pregnant patients be offered the full range of options of genetic screening. (ACOG Practice Bulletin, Number 77, January 2007)

**Background:** At HCMC pregnant patients can receive first trimester genetic screening through OBTU/Genetic Counseling. It is offered in a "Stepwise Sequential" manner. This incorporates an Integrated first trimester screen (nuchal translucency, PAPP-A, B-hcg level) and a follow up second trimester quadruple screen and ultrasound. The first trimester screen is accompanied by an appointment with the genetic counselor as well. Follow up is arranged and managed through the Genetic Counselor (Marie Runyon).

The first trimester testing provides a higher Down syndrome detection rate at an earlier gestational age than the second trimester testing alone. The quad screen in the second trimester allows for screening for trisomy 18 and neural tube defects. There are various advantages to each approach (Please see ACOG bulletin for details). Other than termination of pregnancy there is no treatment available prior to delivery for any of these genetic conditions.

## Plan:

1. All pregnant women who present for OB intake appointments at less than 12 + 0 weeks GA will be offered first trimester and second trimester approaches to genetic screening. Patients will be given information about these options during their OB intake appointment by the RN. Patients with more questions after the intake appointment will be offered appointments with providers at Whittier or Marie Runyon, depending on patient preference. Patients presenting for OB intakes after 12+0 weeks GA will be offered 2<sup>nd</sup> trimester screening only.

2. When there is uncertainty about dating of the pregnancy, a dating ultrasound should not be ordered as part of the OB intake appointment if the patient plans to meet with Marie Runyon to discuss first trimester screening as a dating ultrasound will be done as part of first trimester screening. If a patient declines first trimester genetic screening, then a dating ultrasound should be ordered in the usual way.

## **Outstanding Issues:**

1. Uniform, patient friendly education tools need to be available that outline options for genetic screening. Video presentations in English and Spanish may be ideal. Patients can be easily swayed by the way these choices are presented by providers; a teaching video offers an ethical and accessible way of presenting this complex information.
2. The patients need to go to HCMC for multiple appointments when they choose first trimester genetic screening, we hope that appointments can be scheduled so as to minimize the numbers of those visits. We need to monitor of the numbers of appointments and challenges of getting these appointments scheduled, as they impact clerical people at Whittier as well as patients.
3. Add orders for OBTU ultrasound and Genetic Counseling referral to nurse OB intake order set.
4. Ongoing review of how this new option for genetic screening positively or negatively impacts on our patients.