For Referring Providers

First Trimester Screening using nuchal translucency (NT) and biochemistry

The evolution of prenatal screening for Down syndrome continues with the introduction of nuchal translucency (NT) measurement – along with PAPP-A, hCG, and dimeric inhibin-A – in the first trimester. This combination of markers provides early consistent and reliable screening for Down syndrome and trisomy 18. This test is available for women who would rather not wait for conventional second trimester screening.

At a fixed screen positive rate, detection rates for First Trimester Screening are comparable to those seen in the second trimester:

Prenatal Detection of Down syndrome at 5% Screen Positive Rate

	NT + Biochemistry First Trimester Screening*					Quad Test
GA (wks)	10	11	12	13	**10-13**	Second Trimester Screening
Detection	81%	83%	87%	90%	**86%**	**81%**

*Actual positive rate will depend on age distribution of screened population
Table adapted from Wald et at, SURUSS Report, study funded by NHS, UK

Screening Requirements

- CRL between 45 and 84 mm
- NT measured by certified* sonographers at DHMC
- Serum sample drawn between 11w and 13w 6d gestation
- Completed First Trimester Screening requisition

*Fetal Medicine Foundation, Society for Maternal-Fetal Medicine, Nuchal Translucency Quality Review

An accurate measurement of Down syndrome risk

First Trimester Screening takes advantage of the association between fetal nuchal translucency (NT) and Down syndrome risk. The observation that newborns with Down syndrome had excessive skin on the back of the neck led to prenatal ultrasound examination of the width of the translucent, fluid-filled space beneath the fetal skin on the back of the neck. Multiple studies have confirmed that increased NT is associated with increased risk for Down syndrome in the fetus. In addition, several first trimester maternal serum markers are associated with Down syndrome risk. By combining fetal NT measurement with maternal biochemical measurements, an accurate Down syndrome risk can be determined.

Advantages

Patients who wish to complete the screening process during a time when the pregnancy is not yet obvious may find First Trimester Screening to be preferable to second trimester screening. While the first and second trimester detection rates for Down syndrome are comparable at a given screen positive rate, in practice positive rates for First Trimester Screening are typically higher, reflecting the higher maternal ages of women who tend to utilize this service. Patients may also utilize their first trimester results to help them decide for earlier diagnostic testing such as chorionic villi sampling (CVS). CVS is typically performed between 10 - 13 weeks. Amniocentesis is available starting at 15 weeks.

Precise, expert ultrasonographers

Because the accuracy of the screening depends on the integrity of the ultrasound of fetal nuchal translucency measurements, sonographers with documented expertise in first trimester measurements of this type must conduct the NT measurements. Specialized training as well as ongoing image review are required as part of certification.

Reimbursement

Most insurers should cover First Trimester Screening to the extent that they cover prenatal ultrasound and serum screening procedures. Patients are encouraged to check with their insurance carriers if in doubt.

If you would like us to provide First Trimester Screening to your patients:

- Fetal CRL must be between 45 and 84 mm as determined by DHMC sonographers. This corresponds to approximately 11 weeks - 13 weeks and 6 days gestation.
  - ☐ If the patient is obese, we would suggest scheduling the patient closer to 13 weeks.
- If NT cannot be measured accurately, the patient may need to be rescheduled.
- Blood for the serum test is drawn following the NT measurement.
- Patients referred because of increased risk, e.g., advanced maternal age or family history, will be offered genetic counseling by DHMC counselors.
- Results are available in 3-5 days.

  Screen positive results will be called to your office and your patient by a Dartmouth-Hitchcock genetic counselor.

  Screen negative results will be faxed to your office by FBR. You will need to inform your patient of the results.
- Patients with screen negative results can have AFP Only analysis in the second trimester to rule out open fetal defects. Second trimester multiple marker screening is CONTRAINDICATED in patients who have undergone First Trimester Screening.

Refer a patient:

If you would like to refer a patient, please call the Physician Connection Line at 866-DHMC DOC (866-346-2362)
Download referral forms and lab requisition: www.dhmc.org/goto/referralforms

When making a referral, fax a completed referral and lab requisition form to
Lebanon: (603) 653-3545 | Manchester: (603) 623-7216 | Nashua: (603) 577-3497

FBR

Prepared in collaboration with the Foundation for Blood Research, Scarborough, Maine

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