Nuchal Translucency Screening for Down Syndrome

Last Review Date: December 10, 2021	Number: MG.MM.RA.11eC

Definitions

Nuchal translucency (NT) measurement — Accumulated fluid behind the fetal neck, referred to as the nuchal area, is measured in a standardized way utilizing ultrasonography.

During the first trimester of pregnancy (0–14 weeks), a small amount of fluid collects in the fetal nuchal area, which typically drains away via the maturing lymph system. This fluid (edema) is visible on an ultrasound scan as the translucent space referred to as nuchal translucency. A larger than normal NT space is thought to occur in approximately 75% of fetuses with Down syndrome (DS).

Utilizing a combination technique, referred to as combined screening, DS detection increases to 82–87%. The combination refers to the following:

1. 1st trimester NT measurement.
2. Serum markers of pregnancy associated plasma protein-a (PAPP-A) and free or total beta-human chorionic gonadotrophin (hCG).
3. Maternal age.

Guideline

First trimester combined screening is appropriate (regardless of age) as follows:

1. Women who present at 10.0–13.6 weeks are eligible for combined screening (NT, serum hCG, PAPP-A) in singleton and twin gestations.
2. Women who present at > 13 weeks, with any of the following risk factors, are eligible for combined screening utilizing a 24-hour-turnaround fingerstick test:
    a. Advanced maternal age.
    b. History of a prior pregnancy with a trisomy.
    c. Ultrasound suggestive of trisomies.
    d. Parental balanced Robertsonian translocation with increased risk of fetal trisomy 13 or 21.

Note: For accurate gestational dating, the fetal crown-rump length should be between 45-84mm.

Women found to be at increased risk of having a fetus with Down syndrome or trisomy 18 with first-trimester screening should be offered genetic counseling and the option of non-invasive prenatal testing for fetal aneuploidy, chorionic villous sampling or mid-trimester amniocentesis.

Repeat testing during the course of the pregnancy is not recommended.

Integrated testing utilizing NT measurement (fully integrated screening, stepwise sequential screening or contingent sequential screening) is appropriate (regardless of age) for women who present at 10.5–14 weeks.

Limitations/Exclusions

1. NT testing requires that a highly skilled ultrasonographer perform the service so that optimal test result accuracy can be assured; therefore, coverage is limited to an ultrasound-credentialed setting whereby ongoing quality monitoring can be demonstrated.
2. The following screening approaches are not regarded as medically necessary:
    a. First trimester serum analyte testing (beta-hCG, PAPP-A or invasive trophoblastic antigen [ITA]) alone without NT measurement.
    b. First trimester NT measurement alone (without first trimester serum analyte testing) in the absence of fetal cystic hygroma in singleton pregnancies.

Revision History

Dec. 11, 2020	Added note for crown-rump length pertaining to accurate gestational dating.
Dec. 13, 2019	Amended time-frame when screening can be performed (10 0/7 to 13 6/7 weeks rather than 10 5/7 to 14 0/7) and added coverage for twin gestations.

Applicable Procedure Codes

76813	Ultrasound, pregnant uterus, real time with image documentation, first trimester fetal nuchal translucency measurement, transabdominal or transvaginal approach; single or first gestation
76814	Ultrasound, pregnant uterus, real time with image documentation, first trimester fetal nuchal translucency measurement, transabdominal or transvaginal approach; each additional gestation (List separately in addition to code for primary procedure)
81508	Fetal congenital abnormalities, biochemical assays of two proteins (PAPP-A, hCG [any form]), utilizing maternal serum, algorithm reported as a risk score
81509	Fetal congenital abnormalities, biochemical assays of three proteins (PAPP-A, hCG [any form], DIA), utilizing maternal serum, algorithm reported as a risk score
81510	Fetal congenital abnormalities, biochemical assays of three analytes (AFP, uE3, hCG [any form]), utilizing maternal serum, algorithm reported as a risk score
81511	Fetal congenital abnormalities, biochemical assays of four analytes (AFP, uE3, hCG [any form], DIA) utilizing maternal serum, algorithm reported as a risk score (may include additional results from previous biochemical testing)
81512	Fetal congenital abnormalities, biochemical assays of five analytes (AFP, uE3, total hCG, hyperglycosylated hCG, DIA) utilizing maternal serum, algorithm reported as a risk score
82105	Alpha-fetoprotein (AFP); serum
82677	Estriol
82397	Chemiluminescent assay

84163	Pregnancy-associated plasma protein-A (PAPP-A)
84702	Gonadotropin, chorionic (hCG); quantitative
84704	Gonadotropin, chorionic (hCG); free beta chain
86336	Inhibin A

Applicable ICD-10 Diagnosis Codes

O09.511	Supervision of elderly primigravida, first trimester
O09.512	Supervision of elderly primigravida, second trimester
O09.521	Supervision of elderly multigravida, first trimester
O09.522	Supervision of elderly multigravida, second trimester
Q90.0	Down syndrome Trisomy 21, nonmosaicism (meiotic nondisjunction
Q90.1	Down syndrome Trisomy 21, mosaicism (mitotic nondisjunction)
Q90.2	Down syndrome Trisomy 21, translocation
Q90.9	Down syndrome, unspecified
Q91.0	Trisomy 18, nonmosaicism (meiotic nondisjunction)
Q91.1	Trisomy 18, mosaicism (mitotic nondisjunction)
Q91.2	Trisomy 18, translocation
Q91.3	Trisomy 18, unspecified
Q91.4	Trisomy 13, nonmosaicism (meiotic nondisjunction)
Q91.5	Trisomy 13, mosaicism (mitotic nondisjunction)
Q91.6	Trisomy 13, translocation
Q91.7	Trisomy 13, unspecified
Q92.0	Whole chromosome trisomy, nonmosaicism (meiotic nondisjunction
Q92.1	Whole chromosome trisomy, mosaicism (mitotic nondisjunction)
Q92.2	Partial trisomy
Z36	Encounter for antenatal screening of mother
Z36.0	Encounter for antenatal screening for chromosomal anomalies
Z36.1	Encounter for antenatal screening for raised alphafetoprotein level
Z36.2	Encounter for other antenatal screening follow-up
Z36.3	Encounter for antenatal screening for malformations
Z36.4	Encounter for antenatal screening for fetal growth retardation
Z36.5	Encounter for antenatal screening for isoimmunization
Z36.8	Encounter for other antenatal screening
Z36.81	Encounter for antenatal screening for hydrops fetalis
Z36.82	Encounter for antenatal screening for nuchal translucency

Z36.83	Encounter for fetal screening for congenital cardiac abnormalities
Z36.84	Encounter for antenatal screening for fetal lung maturity
Z36.85	Encounter for antenatal screening for Streptococcus B
Z36.86	Encounter for antenatal screening for cervical length
Z36.87	Encounter for antenatal screening for uncertain dates
Z36.88	Encounter for antenatal screening for fetal macrosomia
Z36.89	Encounter for other specified antenatal screening
Z36.8A	Encounter for antenatal screening for other genetic defects
Z36.9	Encounter for antenatal screening, unspecified

References

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Screening for fetal chromosomal abnormalities. ACOG Practice Bulletin No. 77. American College of Obstetricians and Gynecologists. Obstet Gynecol 2007;109:217–27. [Reaffirmed 2013].

Screening for fetal aneuploidy. Practice Bulletin No. 163. American College of Obstetricians and Gynecologists. Obstet Gynecol 2016; 127:e123–37.

Spencer K, Nicolaides KH. Screening for trisomy 21 in twins using first trimester ultrasound and maternal serum biochemistry in a one-stop clinic: a review of three years experience. BJOG 2003; 110:276–80.

Specialty matched clinical peer review.