IN THE NAME OF GOD

National guidelines for the prevention of chromosomal abnormalities in Down syndrome

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Nuchal Translucency and Trisomy 21

- Fetuses with trisomy 21 as well as other aneuploidies often have excess fluid in the subcutaneous tissue behind the fetal neck.
- Sonographically, this appears as an echolucent fluid collection between the soft tissue over the cervical spine and anechogenic line representing the skin edge.
- This fluid space is called the **nuchal translucency** (NT)

- The lucency is thought to represent mesenchymal edema and is often associated with distended jugular lymphatics The prevailing theory suggests an alteration in lymphangiogenesis and delayed lymphatic development.
- Other possible causes include cardiac failure and abnormal extracellular matrix, but these do not explain the localized and transient nature of the NT.
- Most likely, the cause is a complex interaction of factors.
- NT normally increases with advancing gestational age, and therefore the measurement is compared to **crown-rump length**


NB
NT

Standardization of Nuchal Translucency Measurement Technique

- The fetal CRL must be between 45 and 84 mm
- The accuracy of the NT measurement and the CRL is critical because the NT measurement is converted into multiples of the median (MoM) based on the CRL.
- An adequate sonographic image to measure the CRL requires the fetus to occupy a majority of the image space and be in a neutral position.

CRL

- The longest straight line between the fetal crown and rump is measured at least three times and the average of three good measurements is used.

- In measuring CRL, pay attention to the tallest area by attaching the head to the farthest part of the soft tissue

Criteria for an Accurate Crown-Rump Length

ALARA: Thermal Index BONE < 0.7[40]

MAGNIFICATION

The fetus fills the majority of the image space available

MIDSAGITTAL VIEW

Fetal spine midsagittal

Profile, spine, and rump are visible

NEUTRAL POSITION

The spine is in line with the head

Fluid is visible between the fetal chin and chest

MEASUREMENT

Angle of insonation perpendicular to fetus

Fetus horizontal on image

Calipers are placed on the outer border of the skin at crown and rump


CRL
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Fetal Nuchal Translucency (NT) Measurement Technique

ALARA: Thermal Index BONE < 0.7[40]

CLEAR NT MARGINS
Thin NT line
Angle of insonation perpendicular to NT space
Fetus horizontal on image
TIPS for optimal imaging:

1. Optimize your focal zone
2. Reduce your dynamic range
3. Reduce the gain
4. Review harmonics. Possible edge enhancement optimized with harmonics off
5. Avoid post freeze zoom
6. Narrow your sector

FETUS IN MIDSAGITTAL PLANE
Fetal spine midsagittal in thoracic and cervical region
Tip of nose in profile
Third and fourth ventricles in brain demonstrated

FETUS OCCUPIES MAJORITY OF IMAGE
Head, neck, and upper thorax fill image
Fetus occupies more than 50% of image space; a second fetus of the same size would not fit in the image space

HEAD IN A NEUTRAL POSITION
Head in line with the spine, angle of neck and chest is less than 90 degrees
Pocket of fluid should be visible between chin and neck

AMNION SEEN SEPARATELY FROM NT LINE
Fetus is seen away from uterine wall and separate from the amnion

MEASUREMENT
Calipers (cursors) must be + (plus/positive)
Crossbar of caliper is on the NT line at the inner border adjacent to the lucency
Measurement is perpendicular to long axis of fetus
Measurement is taken at the WIDEST part of the lucency (If nuchal cord, measure above and below the cord and average)

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Thin NT line

Angle of insonation perpendicular to NT space

Fetus horizontal on image

TIPS for optimal imaging:

1. Optimize your focal zone
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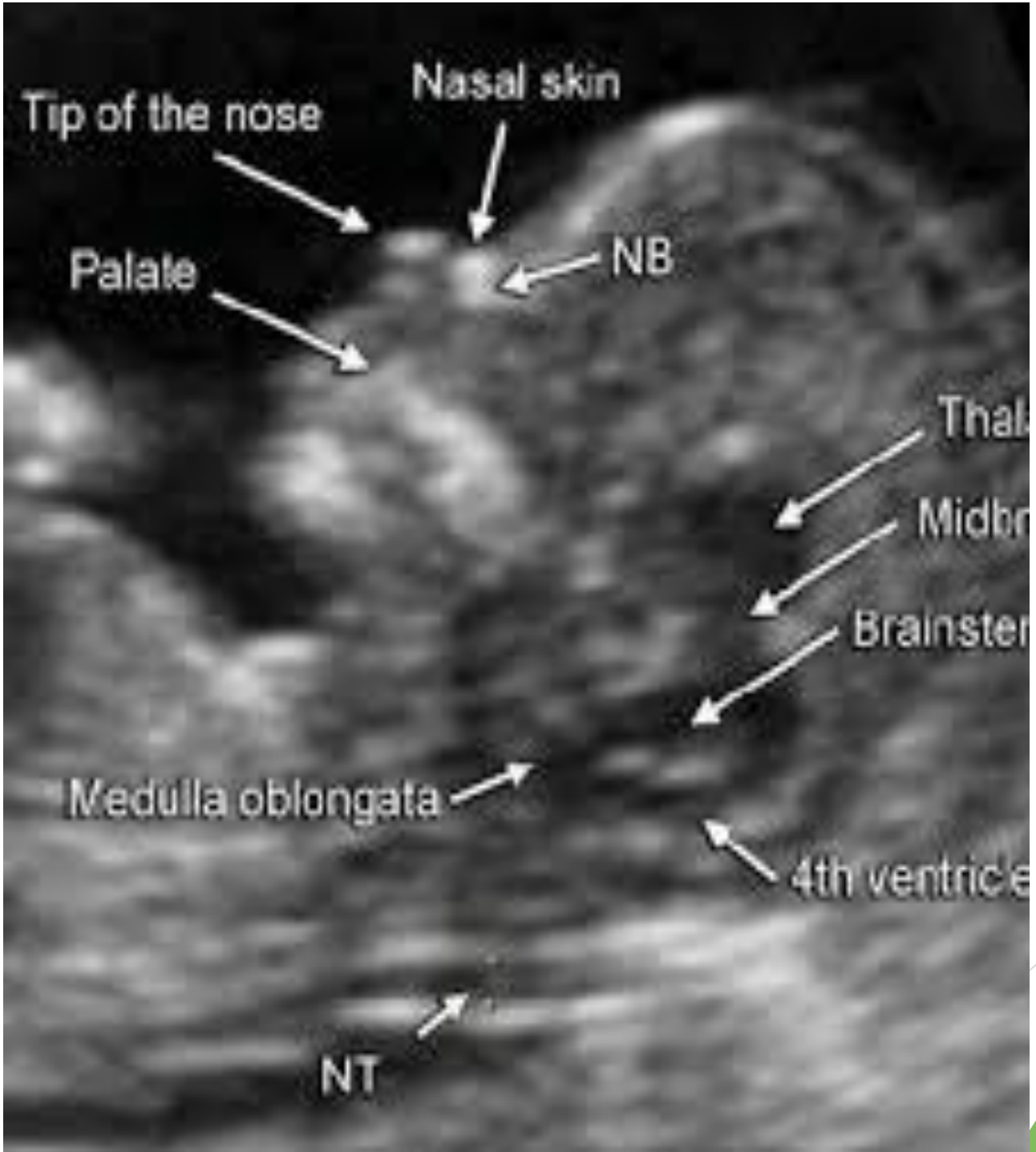
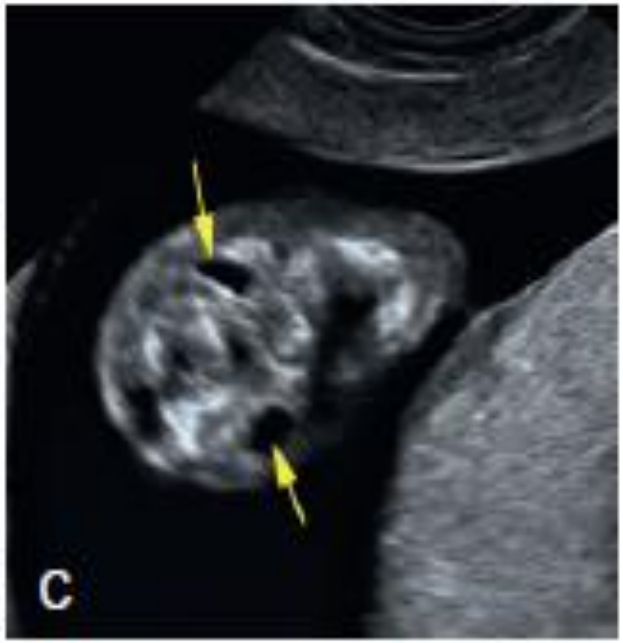
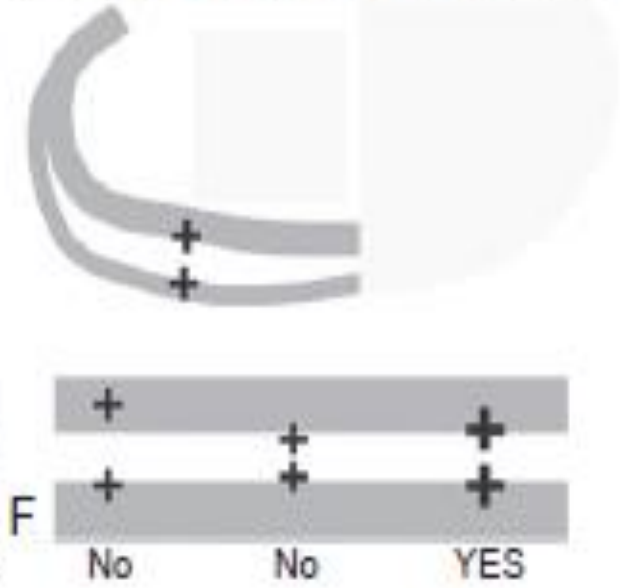

Tip of the nose
Nasal skin
NB
Palate
Thal
Midbr
Brainster
Medulla oblongata
4th ventricle
NT



Caliper alignment for NT measurement



Nasal Bone

many fetuses with trisomy 21 have a small or absent nasal bone.

The fetal nasal bone can be seen by ultrasound starting at approximately 11weeks' gestation.

The first-trimester nasal bone evaluation is technically challenging, and competency in assessing nasal bone reportedly takes an average of 80 scan

. Initial first-trimester screening studies report that the nasal bone is absent in 73% of fetuses with trisomy 21 and 0.5% of euploid

Criteria for Nasal Bone (NB) Evaluation

- ALARA: Thermal Index BONE < 0.7[40]
- Margins of fetal anatomy clear without ambiguity in nasal anatomy
- Fetus in midsagittal plane
  - Fetal spine midsagittal in thoracic and cervical region
  - Tip of nose clearly seen in profile and the skin edge over the nasal bridge is identified
  - Care must be taken to demonstrate the skin edge separately from the NB so that an equal (=) sign is apparent
  - Third and fourth ventricle are identified in the brain
- Fetus occupies majority of image
  - Head, neck, and upper thorax fill image
  - Fetus occupies more than 50% of width and length of image
- Angle of insonation 45 degrees to fetal profile, perpendicular to NB
- Echogenicity of NB is comparable to other bony structures and similar or brighter than the overlying skin.

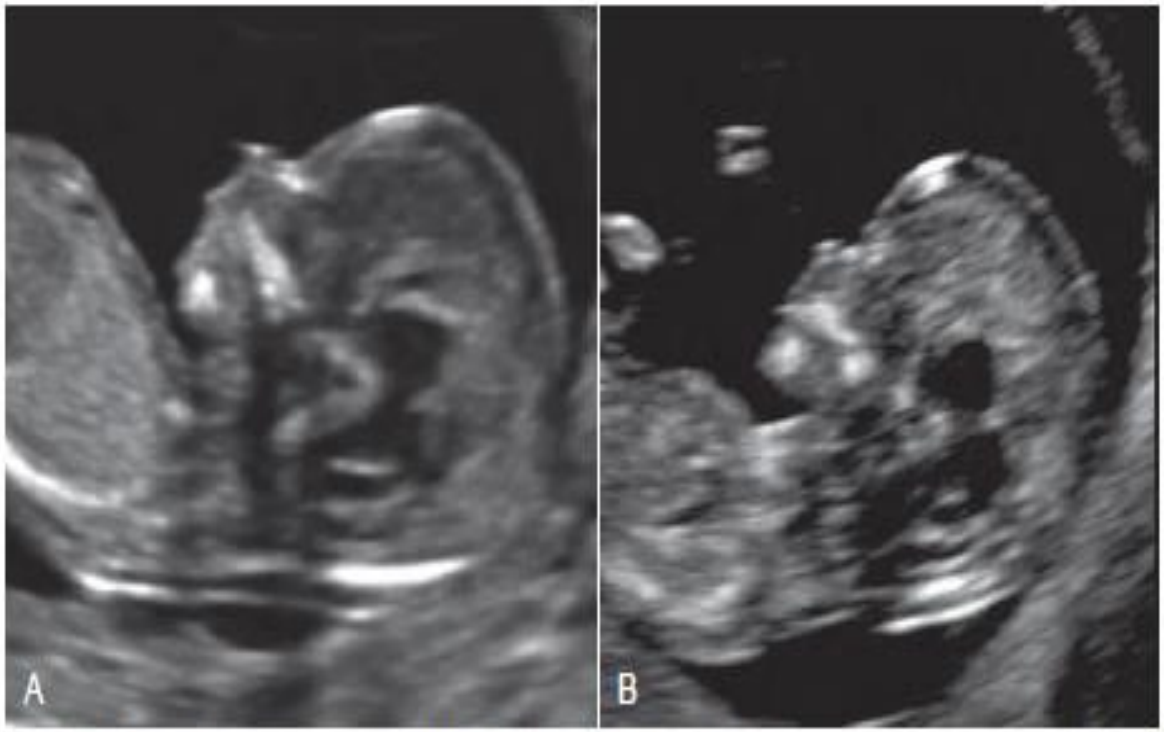


FIG. 31.4 First-Trimester Nasal Bone Assessment. **(A)** Midsagittal profile of a fetus shows the correct method of assessing the nasal bone. Note that the nasal bone is more echogenic than the overlying skin and they form an equal (=) sign. **(B)** Midsagittal profile of a fetus with trisomy 21 shows the echogenic overlying skin but **absent nasal bone.** See also Video 31.3.

Other Markers for Aneuploidy

Reversed Flow in Ductus Venosus

The ductus venosus directs well-oxygenated blood from the umbilical vein to the coronary and cerebral circulation.

Abnormalblood flow demonstrated as a reversed *a* wave in the ductus venosus is seen in 80% of fetuses with trisomy 21 and in 5% of euploid fetuses

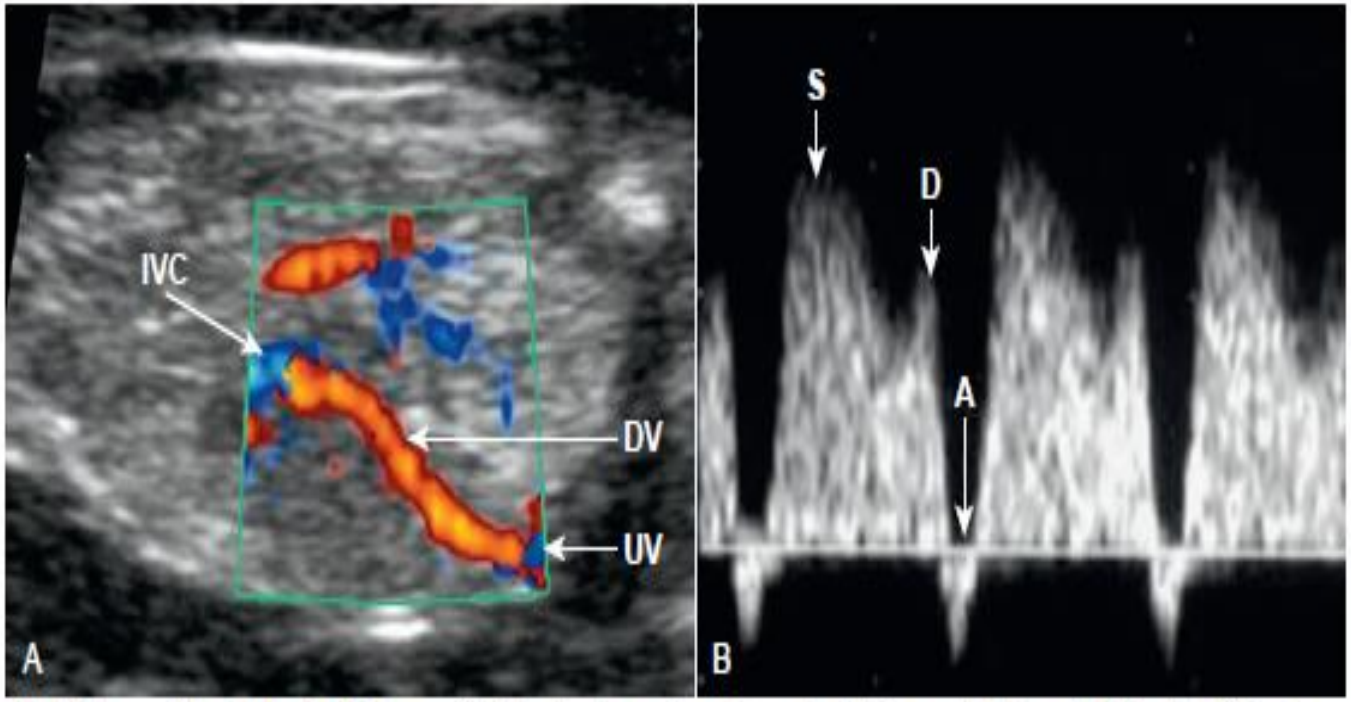


FIG. 31.5 Reversed Flow in Ductus Venosus. (A) Color Doppler anatomy of vessels at oblique sagittal view of fetal trunk. *DV,* Ductus venosus; *IVC,* inferior vena cava; *UV,* umbilical vein. **(B)** Abnormal ductus venosus sonogram shows a **reversed *a* wave.** Absent or reversed a-wave flow can occur in cardiac failure, with or without cardiac defects, and in chromosomally abnormal fetuses. *D,* Diastole; *S,* systole.

Tricuspid Regurgitation

Tricuspid regurgitation has also been proposed as a method of risk assessment.

Tricuspid regurgitation was identified in (74%) of trisomy 21 fetuses and in (7%) of euploid fetuses.

No relationship between tricuspid regurgitation and the levels of maternal serum free ß-hCG and PAPP-A was identified.

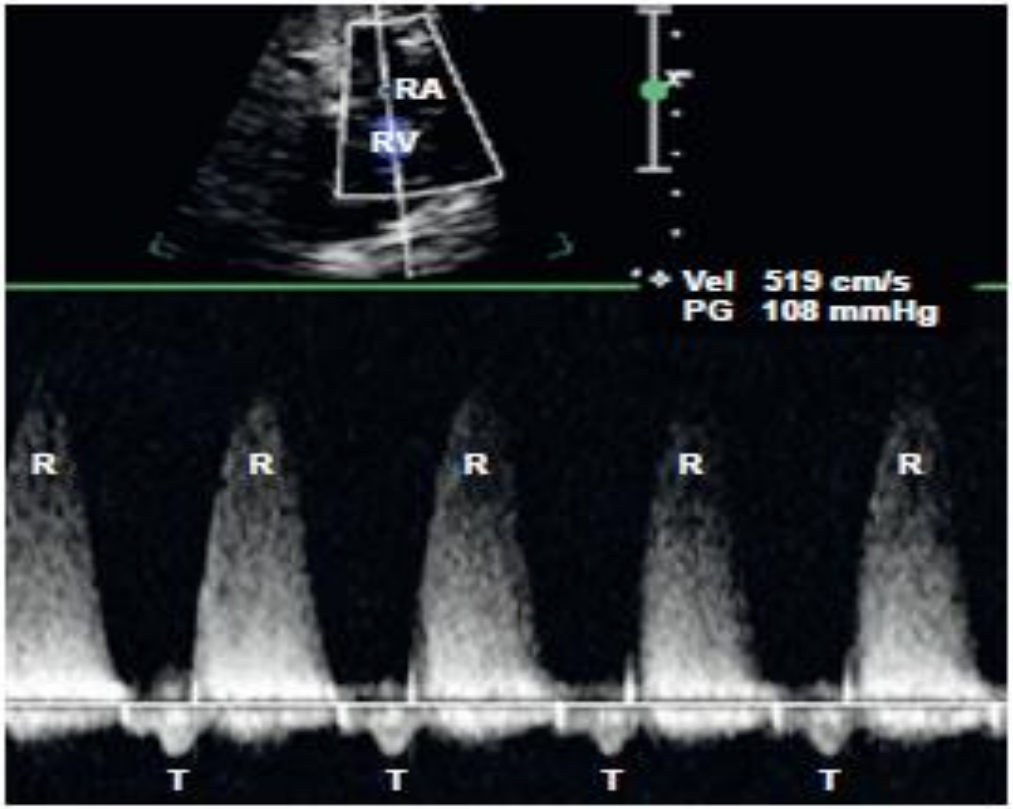


FIG. 37.17 Tricuspid Insufficiency. Spectral Doppler sample volume is placed proximal to the tricuspid valve in the right atrium *(RA)*. The regurgitant flow *(R)* can be seen above the baseline. This implies that the valve has not closed completely during systole, and therefore blood flow is retrograde into the right atrium. *RV*, Right ventricle; *T*, tricuspid valve.


Thanks For Your Attention

Screening for mothers who have access to standard NT ultrasound

- In this case, the method of choice will be the Combined Test. Therefore, NT ultrasound and biochemical tests including Free β hCG and PAPP-A are requested by the physician at 11 to 13 weeks and six days of gestation.

- NT ultrasound takes precedence over biochemical tests

- 1) If NT≥3.5 mm or NT to CRL size ratio is more than 99 (99% percentile) Biochemical tests have not been performed and the mother should be instructed by an ultrasound specialist to see the referring physician at the earliest opportunity to take the necessary measures.
- These should be reported by the sonographer to the county of the mother's place of residence. (These mothers should be referred by a gynecologist for a genetic diagnosis.)

- 2) - In cases where NT <3.5 and the size ratio of NT to CRL is more than 95 (95% percentile) and less than 99%;
- The mother is considered a positive screening and should be referred by the sonographer to the referring physician at the earliest opportunity to take the necessary measures. These cases should be reported by the sonographer to the county of the mother's place of residence. Be referred for NIPT.)

- 3) - If NT <3.5 mm and NT to CRL size ratio is less than 95% (95% percentile), At the initial request of the physician, the mother should be instructed by an ultrasonographer to perform biochemical screening tests for timely referral to a medical diagnostic laboratory.

- Tests are best done the same day (or at most the next day). It should be noted that sampling of biochemical tests should be done up to 13 weeks and 6 days of pregnancy, and these tests will be worthless after this time.

TABLE G-1 Distribution of Nuchal Translucency Measurements by Crown-Rump Length

Crown-Rump Length (mm)	Gestational Age (wk)	*N*	Mean (mm)	Median (mm)	Standard Deviation (mm)	PERCENTILE CUTPOINT (mm)			PERCENTILE CUTPOINT (MoM)			Percentile Equivalent at 3.5 mm
						90th	95th	99th	90th	95th	99th	
45.0	11.3	404	1.17	1.1	0.31	1.5	1.7	2.0	1.31	1.42	1.82	99.8
46.0	11.4	620	1.22	1.2	0.38	1.6	1.8	2.9	1.37	1.58	2.45	99.6
47.0	11.5	667	1.21	1.2	0.33	1.6	1.8	2.2	1.30	1.44	2.02	99.8
48.0	11.6	796	1.26	1.2	0.39	1.7	1.9	2.9	1.35	1.55	2.32	99.6
49.0	11.7	853	1.25	1.2	0.33	1.6	1.8	2.5	1.28	1.45	1.90	99.8
50.0	11.7	1048	1.29	1.2	0.36	1.7	1.8	2.6	1.34	1.49	2.08	99.7
51.0	11.8	1256	1.31	1.2	0.38	1.7	1.9	2.8	1.34	1.53	2.17	99.8
52.0	11.9	1401	1.31	1.3	0.30	1.7	1.8	2.3	1.29	1.40	1.71	99.9
53.0	12.0	1783	1.35	1.3	0.39	1.7	1.9	2.6	1.32	1.44	1.98	99.7
54.0	12.0	1919	1.37	1.3	0.34	1.8	2.0	2.4	1.33	1.47	1.83	99.8
55.0	12.1	2089	1.41	1.4	0.42	1.8	2.0	2.6	1.32	1.46	2.02	99.6
56.0	12.2	2247	1.42	1.4	0.36	1.8	2.0	2.6	1.33	1.45	1.89	99.7
57.0	12.3	2439	1.44	1.4	0.35	1.8	2.0	2.5	1.33	1.46	1.85	99.9
58.0	12.3	2695	1.48	1.4	0.39	1.9	2.1	2.6	1.35	1.48	1.92	99.6
59.0	12.4	2831	1.51	1.5	0.42	1.9	2.1	2.8	1.35	1.49	1.93	99.6
60.0	12.5	3011	1.54	1.5	0.39	2.0	2.2	2.7	1.38	1.50	1.98	99.7
61.0	12.6	3050	1.55	1.5	0.37	2.0	2.2	2.6	1.35	1.50	1.79	99.8
62.0	12.6	3185	1.57	1.5	0.37	2.0	2.2	2.8	1.36	1.48	1.91	99.8
63.0	12.7	3156	1.60	1.5	0.38	2.1	2.3	2.7	1.35	1.48	1.85	99.8
64.0	12.8	3217	1.62	1.6	0.41	2.1	2.3	2.8	1.38	1.51	1.81	99.8
65.0	12.8	3125	1.64	1.6	0.39	2.1	2.3	2.8	1.35	1.47	1.83	99.7
66.0	12.9	3171	1.67	1.6	0.42	2.2	2.4	2.9	1.37	1.50	1.91	99.6
67.0	13.0	3083	1.69	1.6	0.42	2.2	2.4	2.8	1.37	1.51	1.81	99.7
68.0	13.1	2872	1.70	1.7	0.40	2.2	2.4	2.8	1.35	1.50	1.76	99.9
69.0	13.1	2779	1.72	1.7	0.41	2.2	2.4	2.9	1.35	1.46	1.80	99.8
70.0	13.2	2662	1.74	1.7	0.40	2.2	2.4	2.9	1.35	1.46	1.78	99.8
71.0	13.3	2483	1.79	1.7	0.41	2.3	2.5	3.0	1.37	1.48	1.77	99.7
72.0	13.4	2337	1.78	1.7	0.43	2.3	2.5	3.0	1.34	1.48	1.79	99.7
73.0	13.4	2150	1.79	1.8	0.43	2.3	2.5	2.9	1.34	1.45	1.74	99.7
74.0	13.5	2015	1.82	1.8	0.44	2.3	2.5	3.0	1.34	1.45	1.83	99.7
75.0	13.6	1920	1.80	1.8	0.43	2.3	2.5	3.1	1.31	1.49	1.75	99.7
76.0	13.7	1799	1.83	1.8	0.44	2.4	2.6	3.1	1.30	1.42	1.75	99.6
77.0	13.8	1542	1.85	1.8	0.45	2.4	2.6	3.3	1.30	1.43	1.75	99.4
78.0	13.8	1477	1.81	1.8	0.44	2.4	2.5	3.1	1.29	1.42	1.73	99.5
79.0	13.9	1215	1.85	1.8	0.47	2.4	2.5	3.0	1.27	1.38	1.65	99.7
80.0	14.0	778	1.84	1.8	0.43	2.4	2.7	3.1	1.27	1.39	1.69	99.8
81.0	14.1	625	1.86	1.8	0.45	2.4	2.7	3.2	1.24	1.39	1.67	99.6
82.0	14.2	592	1.86	1.8	0.45	2.4	2.6	3.3	1.23	1.33	1.66	99.4
83.0	14.2	611	1.85	1.8	0.44	2.4	2.6	2.8	1.18	1.27	1.47	99.8
84.0	14.2	383	1.86	1.8	0.45	2.5	2.6	3.1	1.23	1.35	1.84	99.7

MoM, multiples of the median.

From Jelliffe-Pawlowski LL, Norton ME, Shaw GM, et al: Risk of critical congenital heart defects by nuchal translucency norms. Am J Obstet Gynecol 212(4):518.e1-518.e10, 2015.

Terms of cooperation of sonographer in the national program of Down Syndrome screening

- A sonographer volunteering to work with the program (which is the referral site for NT sonography in the program) including a radiologist or perinatologist must have a certificate from FMF or ISR
- It is mandatory for the sonographer to have an archive of photos for ultrasound up to 5 years after the sonography

- The number of scans is effective in quality and accuracy and the minimum number of acceptable scans is 30 per year.

- The ultrasound report (in addition to the items mentioned in the first section) should specify the NT size ratio for the CRL and the corresponding percentile

The original photo along with the report must be provided to the mother


از حسن توجه شما سپاسگزارم