"Welcome to the Summer 2010 edition of the NT Examiner. Since its inception in 2005 the NTQR program has grown and matured. We now have a full time Executive Director Jean Lea Spitz, MPH, RDMS and have elected Dr. Alfred Abuhamad, Mason C. Andrews Professor and Chairman of the Ob-Gyn Department at Eastern Virginia Medical School as the new Chairman of the Maternal Fetal Medicine Foundation (MFMF) who also serves as the head of the Nuchal Translucency Oversight Committee (NTOC). The NTQR has exceeded 5,000 registrants. The program is generating individual epidemiologic reports on a regular basis. NTQR has active committees; including an Education committee that is responsible for updating and improving the web based and land courses, a Technology committee that has developed the website and web based calculators, a Benefits committee to assure that all participants receive optimum benefit from their participation, a Quality Assessment committee that designs and monitors the performance improvement program, and a Research and Innovation committee. Congratulations to all. The MFMF and the NTQR program look forward to an expanding role in clinical management.

Steven L. Warsof, MD

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Participant Survey
Thank you for your Input!

In late 2009 the NTQR sent an electronic survey to 5,053 participants. A number of e-mails were returned so the total population receiving a survey request was 4,867 persons. 1,380 participants returned the survey giving an overall response rate of 28.4%.

The survey respondents were reasonably representative of the NTQR participants. A typical respondent has performed nuchal translucency exams for less than 3 years and performs less than 20 NT measurements per month. More than 75% have performed NT exams for less than 5 years and perform less than 50 NT measurements per year. Interestingly, 58.2% work in a practice that is accredited by the American Institute of Ultrasound in Medicine (AIUM).

Respondents appreciate the availability of the the online didactic course, the image review criteria and scoring, the quarterly epidemiologic reports, and not having to submit images yearly. Participants requested more continuing medical education (CME) and patient education resources. Participants also requested web calculators, and the opportunity to voluntarily submit images for review. The NTQR is actively working on all of these requests.

Respondents included questions in the comment section of the survey. NTQR will be answering each of these participant questions in this and upcoming issues of the newsletter. Additional input and questions are welcome at any time.

How many NT measurements do you perform or supervise?

<table>
<thead>
<tr>
<th>How many NT measurements do you perform or supervise?</th>
<th>Response Percent</th>
<th>Response Count</th>
</tr>
</thead>
<tbody>
<tr>
<td>Less than 5 per month</td>
<td>14.2%</td>
<td>194</td>
</tr>
<tr>
<td>5-20 per month</td>
<td>45.6%</td>
<td>622</td>
</tr>
<tr>
<td>20-50 per month</td>
<td>27.7%</td>
<td>378</td>
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<tr>
<td>50-100 per month</td>
<td>9.9%</td>
<td>135</td>
</tr>
<tr>
<td>more than 100 per month</td>
<td>3.1%</td>
<td>42</td>
</tr>
</tbody>
</table>

ANNOUNCEMENT: The NTQR now provides a web calculator for the exclusive use of NTQR-credentialed providers. The calculator uses a model, based on a combination of CRL, maternal age, and NT thickness without serum analytes to estimate Trisomy 21 and Trisomy 18
Genetic Syndromes Associated with Increased NT Measurements: Skeletal Dysplasias

By Renee Chard, MS, CGC
Maine Medical Center
Genetic Counselor and Member
Nuchal Translucency Oversight Committee

Nuchal translucency (NT) measurements greater than the 95th centile have been associated, not only with an increased chance for aneuploidy, but also with a significantly increased risk for structural defects, especially congenital heart defects and diaphragmatic hernia, and with genetic syndromes. In this issue, The NT Examiner will review the reported association of increased NT measurements with skeletal dysplasias in a continuing series of articles reviewing the genetic syndromes most frequently reported in association with increased NT measurements.

While the true prevalence of skeletal dysplasias remains uncertain, it is estimated to be 2-3/10,000 births. Well over 200 conditions are included under the category of skeletal dysplasia and further classification continues to evolve with clarification of etiologies. Skeletal dysplasias that have been reported in association with increased nuchal translucency measurements including achondroplasia, osteogenesis imperfecta, thanatophoric dysplasia, diastrophic dysplasia, and campomelic dysplasia just to name a few. The mechanism for the increased nuchal translucency in such cases is unconfirmed, but theories include (1) narrow thoracic cage causing mediastinal compression, a common finding in many skeletal dysplasias, (2) changes in the dermis secondary to collagen defects or (3) reduced fetal movement.

Given the number of skeletal dysplasias it can be difficult to make a specific diagnosis prenatally, but there are evaluations that can be done to uncover clues to a diagnosis. Ultrasonography is a very important tool in gathering more information about fetal anatomy. Sonographic findings that are associated with skeletal dysplasias and might be identified in the first trimester include short and/or bowed femora, small thorax, hypomineralization of the skull and vertebral anomalies. However, first trimester findings are limited and often, features of skeletal dysplasias do not present until the second trimester, especially in non-lethal forms.

It is recommended that fetuses with an increased nuchal translucency in the first trimester have a high-resolution ultrasound examination in the second trimester. Second trimester sonographic findings suggestive of skeletal dysplasia are numerous. In addition to those seen in the first trimester include, frontal bossing, depressed nasal bridge, micrognathia, oral cleft, stippled epiphysis, polydactyly, trident hand and polyhydramnios.

Genetic counseling plays an important role in the evaluation of a fetus with skeletal dysplasia. In addition to careful ultrasound examination, detailed review of pregnancy, medical and family histories is essential and may provide useful diagnostic clues. Moreover, a genetic counselor can provide patient education, support and anticipatory guidance.

Not only are skeletal dysplasias clinically heterogeneous, they are also genetically heterogeneous. The underlying gene has been identified for several conditions. For example, mutations in the FGFR3 gene cause achondroplasia, hypochondroplasia and thanatophoric dysplasia. Type II collagen mutations are responsible for several skeletal dysplasias including spondyloepiphyseal dysplasia, Kniest dysplasia and achondrogenesis. The majority of cases of osteogenesis imperfecta types I, II, III and IV are due to mutations in COL1A1 or COL1A2.
Unfortunately, even the most thorough prenatal evaluation rarely yields a definitive diagnosis, especially in the absence of a family history of skeletal dysplasia. Postnatal testing including genetic evaluation, radiology and histopathology may be required to finalize a diagnosis providing patients with prognostic information as well as recurrence risks and the availability of prenatal diagnosis in future pregnancies. In a family with a known mutation, molecular testing of at risk relatives is highly accurate both pre- and postnatally. In this setting a referral to a medical geneticist would be indicated.

Cedars-Sinai Medical Center is home to the most experienced experts in the area of skeletal dysplasias and offers a consultation service for diagnostic dilemmas. Their team includes genetic counselors, physicians, and scientists. Dr. Deborah Krakow, MD is an attending physician and MFM consultant for the center. More information is available at their website, [http://www.csmc.edu/Patients/Programs-and-Services/Skeletal-Dysplasia](http://www.csmc.edu/Patients/Programs-and-Services/Skeletal-Dysplasia).

REFERENCES


YOU ASKED, NTQR ANSWERS

Why doesn't the NTQR require images every year?

The NTQR program requires images as part of the initial credentialing process. In this way participants demonstrate that they are capable within their practice to produce NT or NB images that meet standard criteria. The NTQR also requires images when participants are selected on the basis of epidemiologic monitoring for required quality improvement. NTQR does not require images every year.

The NTQR recognizes that displaying "best" images each year does not assure that "all" images meet the standard criteria. Large epidemiologic studies have shown that image review is subjective and that quality monitoring by epidemiologic analysis maximizes performance. In addition epidemiologic analysis can be automated by data download from laboratories and individual monitoring can be accomplished more efficiently.

How does NTQR know that participants are doing a good job?

For monitoring purposes, statistical analysis of NT / CRL data provides much more information than looking at 3-5 selected images. The epidemiologic analysis of data submitted for each participant provides information on the day-to-day practice of NT measurements.

If the analysis indicates that the NT median MoM is high, the participant may be making systematic errors that lead to larger measurements, such as not visualizing a separate amnion. If the NT median MoM is low, the participant may be making systematic errors that are leading to smaller measurements, such as not measuring the widest portion of the NT space. The NT median MoM is not the average of NT measurements. It is NTs adjusted by gestational age.

If the standard deviation (SD) is low the calipers on the equipment may be sticking or there may be a tendency to report similar measurements. If the standard deviation is high there may be more than one person using the same number or there may be differences in equipment or practice that are influencing NT measurements.

If the distribution of the measurements is atypical there may be errors in CRL measurements. As can be seen the epidemiologic analysis reveals a great deal about the NT measurements. The NTQR provides feedback to each participant on a quarterly basis. Participants are encouraged to improve their NT measurements by studying this feedback and their own practice patterns.

How can we improve the accuracy of the reports?

First by reviewing what data is needed for the reports and how it is obtained; then by looking at what the participants, laboratories, and the NTQR may do to improve accuracy.
The NTQR epidemiologic reports are based on data sets that include the following:

- Sonographer NTQR ID#
- Supervising physician NTQR ID#
- Number of fetuses
- NT for each fetus
- CRL for each fetus
- Date of ultrasound exam

The NTQR does not receive patient identification or follow-up data. Data sets for each NT examination may be submitted in the following ways:

- Participating laboratories send data directly to NTQR.
- Participants may submit information for individual examinations by logging into their account on http://www.ntqr.org and filling in the information requested in "Data Submission" under "Performance Improvement" on the upper menu.
- Participants may collect their NT/CRL data and submit it regularly in Microsoft Excel or ASCII format. You may submit data through the "Data Submission" link in your NTQR account on the website or by sending an e-mail attachment to NTQRSupport@ntqr.org.

To improve the accuracy of the reports, NTQR encourages participants to do the following:

- Put the NTQR ID# of the sonographer who performs the measurement and the supervising physician who reads on every requisition submitted with patient serum to a laboratory.
- If you change from another credentialing organization to NTQR it is helpful to fax your NTQR certificate to the laboratory and you must place your NTQR ID# on each requisition.
- If your NTQR epidemiologic reports do not reflect your NT examination numbers, then contact NTQR through phone or E-mail so we can work with you and the laboratory to obtain more data. When you contact NTQR we will ask for your NTQR ID# and the laboratories that are receiving samples with your NT / CRL data.
- If a patient has an NT measured but no "biochemistry" is being done, submit that data individually by logging into your NTQR account and going to "Data Submission" under "Performance Improvement".

It is worth noting that at the laboratory level:

- Data may be sorted by the sonographer NT ID# initially since not all samples are received with a supervising physician NT ID#. Data with an NTQR ID# in the sonographer slot will be sent to NTQR. NTQR may not receive supervising physician data when the sonographer ID# is from another organization.
- When a participant has both an NTQR ID# and a FMF ID# the laboratory may only send data from requisitions that contain the NTQR ID#.

Before sending reports, NTQR performs the following data tasks:

- Duplicate data sets are removed.
- Data sets that are incomplete or that list invalid numbers for NT or CRL are eliminated.
- Data sets with invalid NTQR ID#s are eliminated.
- If the data has a physician's ID# in the sonographer field and there is not a supervising physician number indicated, then the physician ID# that is in the sonographer field is placed in the supervising physician field as well.
- If there is a sonographer ID# number in the supervising physician field, that sonographer's number is removed and the ID for "Unknown Sonologist" is inserted.

Can multiple people in the same practice use the same NTQR ID#?

NO, the NTQR ID# is given to individual participants. It should be used to identify NT measurements that are performed or supervised by that individual.
Please provide more images of "good" NTs and "bad" NTs.

The NTQR, as part of our performance improvement module, now offers a voluntary self-assessment image quiz to all participants. Participants will be able to review images and compare their assessment of the image to our review team’s score. The voluntary quiz will soon be available under the menu heading, "Performance Improvement" in your NTQR account.

Samples from this image quiz are provided below. These two images below do not show the amnion. In each image there is at least one other NT image or measurement criteria that is incorrect.

![Image 1](image1.png)  ![Image 2](image2.png)

**NTQR series of false negative screening results**

Dr. Ron Wapner, Dr. Howard Cuckle, and other researchers associated with the NTQR are interested in collecting information about false negative screening results. If you have a patient who has a low-risk screen and then delivers a child with Down Syndrome, NTQR would be interested in seeing the images and the analyte report for the pregnancy. You may send de-identified reports and images to the NTQR and specify that it is a false-negative screen. As the series that we have collected grows we will be reporting on the results.

![NTQR Quality Maintenance Process](image3.png)

**NTQR Quality Maintenance Process**

The Nuchal Translucency Quality Review Program has initiated a quality maintenance process that includes voluntary performance improvement activities and required quality maintenance. NTQR quality monitoring is based on epidemiologic analysis of individual participants’ NT case data. Epidemiologic analysis commences when an individual submits at least 30 NT measurements to the NTQR program.

NT review is based on a comparison of the practitioners’ measurements to those of a standard referent curve. For review purposes, NT measurements are converted to multiples of the gestational age specific referent median (MOMs). The ideal practitioner’s median NT should be 1.0 MOM with an expected 90th percentile range from 0.9 to 1.1 MoMs. Practitioners with NT median MOMs or standard deviation values that are statistically outside the expected range may be required to complete quality maintenance activities.

In evaluating a practitioners’ performance both long-term cumulative results as well as those from the most recent interval are evaluated.

**Participants with Less than 30 NT data sets submitted annually:**

30 NT measurements are required to project an individual’s measurement trend. If there are fewer than 30 NT measurements submitted a report will be generated but results of statistical testing will be stated as invalid due to low numbers.

Participants with fewer than 30 NT measurements will be required to submit a minimum of three images per year to demonstrate continued...
competency to produce NT images meeting the standard criteria for measurement. There will be no additional cost to NTQR participants for
digital submission of images. The images may be submitted through the "Image Refresher" link (soon to be released) that will be found under
"Performance Improvement" upon login to the NTQR website.

If the submitted images do not pass the image review process, additional images will be required until the standard criteria for NT image
acquisition and measurement have been fulfilled.

Report Generation and Distribution:

If more than the requisite 30 NT measurements are submitted, a report will be generated, and the results of statistical testing will be stated.

The results of the epidemiologic monitoring will be reported directly to:
   1. The individual participant,
   2. The individual's supervising physician if indicated by a recorded NTQR number on the requisition slips and
   3. The designated practice administrator upon request.

Participating laboratories have access to reports of participants who submit NT data to their laboratory.

The NTQR epidemiologic monitoring reports provide participants quarterly feedback on their NT measurements.

A number of participants who are the farthest outside the range will be required to undergo quality maintenance. Those selected will be notified
and must complete the activities specified under Required Quality Maintenance.

The NTQR Quality Assessment (QA) committee will determine on a regular basis the participants who are required to complete quality
maintenance. Selection is based on NT Median MOM and standard deviations.

All participants outside the range are encouraged to engage in Preliminary Performance Improvement.

Preliminary Performance Improvement:

When a provider is first notified that analysis of their NT measurements is outside the expected range, they should seek out credentialed
colleagues whose values fall within the expected range to observe NT image acquisition and measurement and offer constructive critiques to
the participant.

It will be the responsibility of the NT credentialed medical director or their designee to monitor progress of these individuals.

NTQR recommends targeted performance improvement activities for individuals whose NT values are reported to be outside the expected
range. All performance improvement activities are available at no cost to NTQR participants. The tasks chosen may include any combination of
the following:

1. Review of the individual’s NT measurements.
2. Review of the technical lectures within the NTQR online course. The lectures may be found under “Education” upon login to the
   NTQR website.
3. Review of specific suggestions related to measuring high or low that are provided.
4. Voluntary submission and review of NT images through the "Image Refresher" link (soon to be released) that will be found under
   "Performance Improvement" upon login to the NTQR website.
5. Monitoring NT measurement analysis by review of quarterly NTQR epidemiologic reports.
6. Completion of the image review self-test available on-line.

Required Quality Maintenance:

If a provider is notified of required quality maintenance, the provider will be required to complete the following:

1. Update practice and supervision information.
2. Review specific suggestions related to measuring high or low that are provided.
4. Complete the image review self-test available on-line.
5. Submit for review five new images from five separate fetuses.
If the submitted images do not pass the image review process, additional images will be required until the standard criteria for NT image acquisition and measurement have been fulfilled.

Sonographers will be required to document supervision by an NT credentialed physician. Statements signed by NT credentialed physician(s) verifying supervision of NT measurement scans performed by the sonographer are mandatory.

Required quality maintenance must be completed within 3 months of the date of notification. If remediation is not completed within 3 months, the relevant laboratories will be notified that the NTQR credential number is invalid until remediation is completed.

Required quality maintenance will not occur for a given provider more frequently than once per year.

**Laboratory Notification of Required Quality Maintenance:**

Participating laboratories have ongoing access to quality monitoring reports of participants who submit data to their laboratory. A laboratory may request regular notification of participants selected for required quality maintenance. All laboratories receiving NT data from participants will be notified if quality maintenance activities required of individuals are unsuccessful. The credential status of such individuals will be reported to laboratories on a regular basis.

**Appeal Process:**

A participant who is required to participate in quality maintenance activities may write a letter of appeal to NTQR. Appeals are reviewed by a designated committee and if sustained may substitute for required quality maintenance.

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**Join NTQR and Get Credentialed**

The Nuchal Translucency Quality Review Program (NTQR) is a United States based effort seeking to establish a NT quality control system and help formalize set standards. NTQR offers a unique opportunity to learn about the proper techniques and theories involved in obtaining accurate and reproducible NT measurements from the 11-14 week ultrasound scan and first trimester risk assessment for Down Syndrome, while also offering a method to evaluate and track provider proficiency though ongoing NT quality monitoring reports.

**Two ways to join NTQR and get credentialed!**

1. **On Line**
   1. Go to [http://www.ntqr.org](http://www.ntqr.org)
   2. Register
   3. On your computer, watch the same lectures given at NTQR's land-based courses. (This doesn't have to be done in one sitting)
   4. Take the same on-line test as land-based course participants
   5. Submit 5 NT images for quality review
   6. Get credentialed

2. **Plan to attend one of these upcoming NTQR land-based courses:**
   1. Register and attend a 2010-2011 planned Land-Based Courses (see below)
   2. Take the on line exam
   3. Submit 5 NT images for quality review
   4. Get credentialed

**National Conference on OB-GYN Ultrasound**
Chicago, Illinois
November 5-7, 2010
[Registration Information](#)

**19th Annual OB / GYN Ultrasound Update for Clinical Practice**
Lago Mar Resort & Club
Ft. Lauderdale, Florida
December 2-5, 2010
[Registration Information](#)

**Obstetric Ultrasound in the High Risk Patient**
Las Vegas, Nevada

**Society of Maternal Fetal Medicine**
**31st Annual Meeting**
The Pregnancy Meeting
NTQR Program Fast Facts

Results of Epi Reports 4/11/2010

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Program Statistics 3/31/2010

- 5,492 providers of NT measurements have registered with the Nuchal Translucency Quality Review Program
- 3,928 providers have been credentialed through NTQR
- Over 28,000 NT images have been reviewed by NTQR’s Expert Reviewers
- Over 800,000 data sets have been provided by participants or by our partner laboratories. These data sets were analyzed to produce individual epidemiologic reports. Over 3,100 personalized reports were sent to participants in April 2010.
- To see a list of our partner laboratories, go to http://www.ntqr.org
Answers to Image Questions in article, YOU ASKED, NTQR ANSWERS

- Left Image: The amnion is not seen and the fetal neck is hyperflexed.
- Right Image: The amnion is not seen and the nuchal space is not measured at the widest point.

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You may also send e-mail to NTQR Support.