Despite the continued controversy around the utility of electronic fetal monitoring (EFM) in obstetrics, the fact remains that it is used in the overwhelming majority of births in the United States. While the technology has been unable to accurately predict long-term neurologic outcome, it can identify fetuses at risk of hypoxia and acidemia. In addition, allegations of misinterpretation and inappropriate management of fetal heart rate (FHR) tracings continue to be the basis for many malpractice claims. In 2004, The Joint Commission issued a Sentinel Event Alert aimed at preventing infant injury during delivery. Specific recommendations included developing clear guidelines for fetal monitoring and interpretations, and educating all members of the labor and delivery team to use standard terminology and communication around abnormal FHR tracings. Both National Institute of Child Health and Human Development (NICHD) and the American College of Obstetrics and Gynecology have also emphasized the importance of standard terminology.

In 2014, the Perinatal Quality Foundation created a Fetal Monitoring Credentialing (FMC) exam to aid in efforts to improve standardization. To facilitate collaboration among the team members, there is a version tailored to L&D nurses in addition to the exam for obstetric physicians and midwives. The FMC exams measure knowledge and judgment related to FHR tracings. The former is done in a traditional fashion using multiple-choice questions; the later is assessed in a unique fashion using script concordance theory (SCT). This type of question is particularly challenging, because it is unfamiliar to most clinicians and there is no single correct answer.

Script Concordance Theory is based on the premise that stored knowledge and experiences, called scripts, are mobilized and organized to help sort through a differential diagnosis and then establish a management plan when presented with a specific clinical situation. SCT questions are written with the intent of modeling the uncertainty of common clinical scenarios and the varied approaches that are typically encountered in daily practice. A brief clinical vignette is given and accompanied by a proposed management plan. This is then followed by an additional piece of information such as a change in the patient’s clinical status or in the FHR tracing. Based on this new piece of information, examinees are asked to state whether or not they would alter the original management plan. The possible responses use a five-point Likert scale with options ranging from the new information strongly supports the original management plan to it strongly invalidates it.

An example of a script concordance question is shown on the next page.
Case Context: A 40-year-old G1P0 patient is admitted with a cervical exam of 7/0/80%/vertex. You review her FHR tracing shown in Panel A.

Your management plan is …
Expectant management while initiating conservative measures …
and then you learn the following additional information:
One hour later, a repeat pelvic examination reveals the cervix to be complete/+2/100%/vertex.

You review her FHR tracing shown in Panel B.

How does this additional information affect your thinking about the management plan?
- Strongly invalidates: (selection of this answer would give FULL credit)
- Could invalidate: (selection of this answer would give PARTIAL credit)
- No impact
- Could support
- Strongly supports

SCT questions are scored based on the responses provided by an expert panel. For each question, there are typically two and sometimes three answers that have been selected by the experts. Credit is given in proportion to the number of experts that gave the same response. It is very intentional that there is not just “one” correct answer, and this approach is very well suited to interpretation and management of EFM, as there is very often more than one legitimate approach to a given clinical situation. One provider may choose to immediately proceed to delivery while another may choose to use various conservative interventions, while still another may choose to simply observe for a while longer. All of these choices are the result of judgment based on knowledge of the relationship of the FHR tracing to neonatal outcome informed by clinical experience.

Since its introduction in mid 2014, the FMC exam has been increasingly incorporated into the quality and safety programs of a number of organizations. To date 42 institutions are utilizing the exam with a total of 1340 individuals having completed the exam and another ~ 1250 scheduled to take it. Physicians make up 51%, CNM’s 9%, and nurses 40% of participants. Of those taking the exam, 90.3% have passed and been credentialed. The FMC exam also can be used to meet one of the American Board of Obstetric and Gynecology (ABOG) annual Part IV Maintenance of Certification requirements (see page 3). More information on FMC can be found on the Perinatal Quality Foundation web site (www.perinatalquality.org).

In addition to FMC, CLEAR is now also being accepted by ABOG for MOC Category IV: Improvement in Medical Practice.

To navigate to FMC and CLEAR on the ABOG site, follow these steps in the screenshots below:

1. Log in @ ABOG.COM
2. Click on “Improvement in Medical Practice (IMP) Activities” under Part IV of your MOC page.
3. Click on Upcoming Simulations and QI Activities.
4. Find CLEAR and FMC among the list of upcoming Simulations and QI Activities.
UPDATE ON THE PERINATAL QUALITY FOUNDATION NTQR PROGRAM

In January 2014, Mary D’Alton and other members of the Perinatal Quality Foundation (PQF) Board of Directors published a history of the development of the Nuchal Translucency Quality Review Program (NTQR). As of February 2013, NTQR had 6600 credentialed participants of which 66.5% were sonographers, and in July 2013, approximately 12% of the epidemiologic reports demonstrated measurements that were statistically “out of range” compared to a standard reference curve.

The last few years have brought rapid change in prenatal testing. Insights and updates from recent NTQR statistics are noted below:

1. Despite the commercial availability of cell free DNA screening for common aneuploidies since 2011, the number of NT/CRL data points submitted and analyzed by NTQR has not changed dramatically; Figure 1 shows the number of data sets submitted per month through February 2016.
2. NTQR regularly inactivates participants who fail to complete the required quality maintenance activities to which they are assigned as well as participants who do not pay their annual participation fees. Nevertheless, the numbers of new registrants and inactivated participants have remained relatively balanced since 2013. Currently there are 7044 credentialed participants, of which 74% are sonographers.
3. The number of participants with data insufficient for analysis has increased slightly from 28% in 2013 to 33% in 2016.
4. In April 2016, 86% of sonographers and 93% of physicians with data sufficient for analysis were “in-range”, this is an increase from 79% and 85% in 2012.
5. Some participants have inquired about the impact of not submitting large NT values to the laboratories and to NTQR. While this may be common practice, we have no evidence that it is impacting epidemiologic reports, as the percentage of epidemiologic reports demonstrating NT/CRL measurements “out of range LOW” has continued to decrease to approximately 7.6%. NTQR encourages participants to submit their own data under “PERFORMANCE IMPROVEMENT” within their accounts or by submission directly to ntqrsupport@ntqr.org if there is concern.

The PQF Board of Directors continues to monitor changes occurring in prenatal testing, and is committed to our mission to improve the quality of obstetrical medical care and to facilitate the transition of new technology into clinical practice. The NTQR has served this mission since 2007, and our newer programs including CLEAR and GEM continue to aid in the transition of new technologies to best clinical practice.

REFERENCE:

INCREASED NUCHAL TRANSLUCENCY BEYOND THE KARYOTYPE:
NOONAN SYNDROME
By Renée Chard, MSc, CGC

The Increased Nuchal Translucency: Beyond the Karyotype series debuted in the previous newsletter with a review of the value of chromosomal microarray in the setting of increased nuchal translucency (NT). However, fetuses with increased NT and normal karyotype and microarray remain at risk for structural defects and single gene disorders, most commonly Noonan syndrome. It is estimated that 1-3% of euploid fetuses with an NT measurement above 3.5 mm and over 10% with cystic hygroma have Noonan syndrome.

Noonan syndrome is estimated to occur in 1 in 1000 to 1 in 2500 births; however, this may be an underestimate because associated features are extremely variable and mild cases may be undiagnosed. Clinical findings of Noonan syndrome include characteristic facial features that tend to become less pronounced with age such as a deeply grooved philtrum, hypertelorism, ptosis, low-set and posteriorly rotated ears, and micrognathia. Short, webbed neck with a low posterior hairline, short stature, pectus deformities, and scoliosis are also common features. Most affected individuals have a heart defect, often pulmonary valve stenosis. In addition, there is an increased risk of hypertrophic cardiomyopathy, bleeding disorders, and developmental delay.

Noonan syndrome is an autosomal dominant condition, meaning that only one genetic mutation is required to cause the condition. Several genes have been implicated, and most cases result from a mutation in PTPN11 (50%), SOS1 (10-15%) and RAF1 (5-10%). A small number of cases are due to mutations in KRAS, BRAF, or NRAS genes; but in 20% of cases the causative mutation is unidentified. The associated genes are important for production of proteins involved in cell division and differentiation. Over half of Noonan syndrome cases result from sporadic mutations; but in some cases, careful examination reveals that one parent is mildly affected and the mutation inherited.

Prenatal ultrasound may be useful for the detection of some features associated with Noonan syndrome. Baaker et al suggest that the diagnosis should be considered if second trimester ultrasound reveals increased nuchal fold or cystic hygroma together with at least one of the following: hydrops, pleural effusion, congenital heart disease, polyhydramnios, or characteristic facial findings. In another study, Croonen et al evaluated 75 fetuses with a normal karyotype and ultrasound findings associated with Noonan syndrome and found that 13 (17.3%) were found to have a mutation in PTPN11, KRAS, SOS1, or RAF1. Of those found to have a Noonan syndrome mutation, all 13 had an increased NT ranging from 4.2 - 14 mm (mean 8.0). Based on their results, Croonen et al recommended that testing of the PTPN11, RAF1, and KRAS genes be offered when a fetus with an increased NT and normal karyotype is found to have at least one of the following: polyhydramnios, hydrops, renal anomalies, distended jugular lymphatic sacs, hydrothorax, CHD, cystic hygroma, or ascites.

Advances in DNA testing with high throughput sequencing continue to expand testing options while reducing cost and turn-around-time for results. Laboratories can not only test for many mutations in a gene at the same time but also test for many mutations in multiple genes at once giving rise to multi-gene panels. Multi-gene panels allow for efficient testing of genetic conditions for which there are several causative genes or in a situation where clinical findings may be explained by a number of conditions with overlapping features. One must bear in mind; however, that DNA testing at this level can result in the discovery of variants of unknown significance (VUS). At the 2012 meeting of the Society of Maternal-Fetal Medicine, Meck et al reported that of 212 tests performed for Noonan syndrome and related conditions using a multi-gene sequencing panel, 8 (4%) had a VUS. In the cases with a VUS in a Noonan syndrome gene, not only is it important for parental testing to determine if the VUS was inherited or de novo, but physical examination of the parents may also be indicated to rule-out mild features of Noonan syndrome. For this reason, it is important that testing be offered in the context of comprehensive pretest counseling. A genetic counselor can be found by clicking on the ‘Find A Genetic Counselor’ link on the website of the National Society of Genetic Counselors (nsgc.org).
Stay tuned for the next installment of our “Increased Nuchal Translucency: Beyond the Karyotype” series for more information on fetal cardiac defects associated with increased nuchal translucency.

References

GEM FOR PRENATAL GENETIC TESTING: IMPROVING COUNSELING AND FACILITATING PATIENTS’ DECISION-MAKING

By Erica Sturm, MS, CGC

The Perinatal Quality Foundation (PQF), the GEM Task Force, and founding supporters are eagerly anticipating the forthcoming Genetic Education Modules (GEM). By the end of the summer, practices will finally have access to these web-based clinical tools. Evidence continues to build demonstrating the need for GEM and the ways in which it will be most useful.

Laura Colicchia and colleagues recently published “Patient-Health Care Provider Conversations About Prenatal Genetic Screening” in the June issue of Obstetrics and Gynecology. In their analysis of 210 first prenatal visits, they found that 10% of patients were not offered testing at their first visit and that providers spent an average of 1.5 minutes discussing prenatal screening during this visit.

The article summarized ACOG’s recommendation that counseling about prenatal genetic screening include discussion of: the screening process, the conditions screened as well as those not captured, the differences between prenatal screening and diagnostic testing, the implications of a positive result, encouragement to explore personal values and beliefs, and the elective nature of prenatal testing. Colicchia et al report that few providers included all recommended counseling points, that detection and false-positive rates were rarely included, and that inclusion of more counseling points correlated with longer discussion times. Investigators concluded that “the lack of in-depth discussions by many health care providers raises concerns that health care providers may not be obtaining truly informed consent.”

In addition to ACOG’s recommended discussion topics, we expect that many factors may affect a patient’s decision to pursue or decline prenatal testing. For example, ability to access educational materials, ability to limit distractions and focus on counseling, and inclusion of a partner or other support person(s) may impact a patient’s decision-making. GEM provides detailed but concise information about
all prenatal genetic testing options, including declining testing altogether. The Patient Module is accessible to patients outside of the office and allows patients to review short sections of easily understandable information when, where and with whom they choose. Personalizable tools reinforce pertinent details and guide patients as they explore personal factors influencing their decisions. Similarly, the Provider Module equips prenatal care providers with tools to counsel patients and answer questions about testing options. Through these modules and other resources, GEM will bridge the gap between patients and providers and facilitate patient education and decision-making. This is expected to enable providers to comply with recommended counseling points, empower patients to make informed decisions, and increase satisfaction on all sides.

Reference


UPCOMING LAND-BASED COURSES:

NTQR First Trimester Assessment Courses (contact IAME.com to register):

<table>
<thead>
<tr>
<th>Course</th>
<th>Date</th>
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<tbody>
<tr>
<td>Obstetric Ultrasound in the High Risk Patient</td>
<td>September 9-11, 2016</td>
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<tr>
<td>Encore at the Wynn</td>
<td>Las Vegas, NV</td>
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<td>Intercontinental Chicago</td>
<td>Chicago, ILL</td>
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<td>CLEAR Courses:</td>
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CLEAR related courses or lectures are planned in Salt Lake City, Utah, at ACOG Region II in New York, at the Society of OB/GYN Hospitalists meeting in Colorado, and in Oklahoma City. For contact information please e-mail support@perinatalquality.org.