*Perspectives in Genetic Counseling*
**Volume 34, Number 2**
**Summer 2012**

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# President's Beat

As the NSGC's President, I've fielded a number of media inquiries this year about the implications of genetic testing for consumers. Reporters often ask about whole genome sequencing in the same breath as *BRCA* genes, noninvasive prenatal diagnosis, and direct-to-consumer testing, struggling to make sense of the differences. I find myself comfortable in this role, calling on the genetic counseling tools of analogy and example to make understandable, relevant distinctions between deterministic and susceptibility tests, genetics and genomics, the "old" and the "new," the "before" and the "after." I can do this because, even though the genomic era is upon us, I have spent a long career counseling people about genetic disorders, and my core skills still apply. I can readily cite real life examples of human reactions to uncertainty, the uniquely familial nature of genetic disease, and marked differences in decision-making styles from one person to another, all issues that come into play whether we are talking about single or multiple genes. I've counseled women about the chance of unintentionally revealed non-paternity – an incidental finding if ever there was one. I've struggled over the ethics of carrier testing for asymptomatic minors. Questions about genomic medicine feel familiar because they *are* familiar, albeit on a more massive scale than we've experienced before. This is not to downplay the practical challenges that genetics professionals will all face as the predicted avalanche of genomic information rolls across our healthcare system over the next few years. Still, of all the potential players in the health care arena, who but genetic counselors and our clinical genetics colleagues have the depth of training and experience to roll up our sleeves and begin the work?

At recent clinical and laboratory genetics meetings, the buzz about whole genome and exome sequencing has been palpable. There is tremendous excitement about the powerful technologies on our doorstep. At the same time, there is worry and a sense of urgency about the need to develop practical models to handle the implications of genomic testing. Which findings should be made available to patients? How will informed consent be handled? How should variants of uncertain significance be explained? The questions now being posed by laboratory and clinical geneticists are downright… "genetic counselor-ish." More than ever before, genetic counselors are participating in, and leading, these important discussions. I look at this age as one of great transformation that will be cited by future historians, for better or worse, as a turning point in human biological

awareness. It is also an age of great opportunity for the genetic counseling field, and one that compels us to collectively seize the day.

In a recent issue of *Journal of Genetic Counseling*[1], former NSGC president **Wendy Uhlmann** and Richard Sharp propose an interesting solution for developing practice models as we move into the genomic age. They point out that we no longer have the luxury of methodically building and documenting an evidence base before establishing best practices for genetic testing, because genomic medicine is already here and guidance is needed now. They suggest that input on global issues related to genetic testing can be more quickly synthesized through the creation of Genetic Testing Integration Panels (GTIPs), local expert groups that bring together the experiences of clinical and laboratory geneticists and genetic counselors, as well as professionals from pertinent related disciplines such as bioethics and health communication. By meeting regularly and sharing experiences, successes, and dilemmas encountered while navigating the rapidly changing landscape of genetic testing, GTIPs can help to identify concerns, needs, and strategies. Ultimately, the collective experiences of many local GTIPs could be combined through a central web-based forum, fostering the ongoing evolution of best practices and standards. This is guideline development on the fly. Ready or not, we're being thrown into the deep end of the gene pool, and it's sink or swim. There's no longer any time to carefully weigh, evaluate, and tweak before putting a tentative toe in the water; a growing number of genetic counselors have already jumped in with both feet, and their pioneering experiences will serve to blaze the trail for the rest of us.

The development of a centralized experience repository, such as the one proposed by Uhlmann and Sharp, could ultimately prove to be an efficient model for molding practice. It will take time, resources, and the collaboration of major genomic institutes, local genetics centers, and individual genetic counselors and geneticists. In the meantime, each of us can already contribute to the collective conversation through discussions with colleagues, presentations at regional and national conferences, blogs, articles, case reports, focus groups, and research studies. We need to call on practicing genetic counselors as well as graduate students in training to observe, document, and imagine creative solutions, because the genomic era is already upon us. In a way, all of our professional training and experience has been leading up to this moment, as we find ourselves face to face with DNA in its original container (i.e., individuals and families) on the new frontier of personalized medicine. It's here, and for a while at least, it's not going to be pretty. It already feels overwhelming and uncomfortable, particularly for a profession that prides itself on being thoughtful, careful, and methodical. We can still be all of these things, but we need to move quickly before other more impulsive, but less sure-footed professionals fill the void. Genetic counselors and clinical geneticists are uniquely positioned to tackle the human aspects of the genomic era. Informed consent? Incidental findings? Pre-test counseling? Bring it: let's go out and own this thing.

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[1]Uhlmann, WR and Sharp, RR. Genetic Testing Integration Panels (GTIPs): A novel approach for considering integration of direct-to-consumer and other new genetic tests into patient care. *J Genet Counsel.*, DOI 10.1007/s10897-011-9468-4. 2012.

Brenda Finucane, MS, CGC
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# Feature Article

## Health Information Technology 101

*By Megan Doerr, MS, CGC and Heather Fecteau, MS, CGC*

Health information technology (HIT) is the area of information technology that focuses on the design, development, creation, use, and maintenance of information systems for the healthcare industry. Automated and interoperable healthcare information systems are expected to lower costs, improve efficiency, and reduce error, while also providing better consumer care and services.

***Why is Health IT important to genetic counselors?***

- It may change the way you document the health care you provide
- It could improve the number and quality of your referrals
- It may reduce your per-patient workload

**It may change the way you document: Use of the electronic health record (EHR)**

As part of the recently enacted U.S. stimulus bill, $19.2 billion was allocated to promote the adoption and meaningful use of HIT. This release of funding is known as the Health Information Technology for Economic and Clinical Health or "HITECH" Act, part of the American Recovery and Reinvestment Act of 2009 (ARRA). One of the primary outcome goals of the HITECH Act is to make an EHR available for every American by 2014, a challenge originally issued by former U.S. President George Bush in 2004. Since few U.S. doctors or hospitals — perhaps 17% and 10%, respectively — have even basic EHRs, EHR adoption is sure to change the way many, if not most, genetic counselors schedule patients, document encounters, record test results, and help coordinate follow-up for their patients.

EHRs are often confused with electronic medical records (EMRs) and personal health records (PHRs). An EHR is an individual's official, digital health record; it differs from an EMR in that it meets nationally recognized interoperability standards, allowing providers, insurers, and others to securely share health information about a patient. EHRs are distinct from PHRs because they are not managed, controlled, and shared by the patient, but rather by a health care provider(s) or organization(s). EHRs often have patient portals for electronic provider-patient/patient-provider communication, clinical decision support, and computerized physician order entry. These definitions are further described in Table 1.

**Table 1. Health Information terminology**

| | |
|---|---|
| Electronic Health Record (EHR) | Individual's official, digital health record; shared among multiple facilities and agencies |
| Electronic Medical Record (EMR) | An individual's health record within a healthcare provider's facility |
| Personal Health Record (PHR) | An individual's self-maintained health record |
| Regional Health Information Organization (RHIO) | Group that oversees communications among the other elements and unifies them geographically. |
| Health Risk Assessment (HRA) | Tool to evaluate an individual's likelihood of developing a disease (e.g., Gail Model) |
| Clinical Decision Support (CDS) | An interactive electronic tool which is designed to assist physicians and other health professionals with decision making tasks |
| Health Information Exchange (HIE) | The mobilization of healthcare information digitally across organizations within a region or community |
| Computerized Physician Order Entry (CPOE) | A computerized system that allows a physician's orders for services such as medications, laboratory tests, and other tests to be entered electronically |
| Interoperability | The ability of different information technology systems and software applications to communicate, to exchange data accurately, effectively, and consistently, and to use the information that has been exchanged |

**It could improve the number and quality of your referrals: Meaningful use**

A second outcome goal of the HITECH Act is the concept of "meaningful use" of HIT, with provision for incentive payments to hospitals and providers administered through the Center for Medicare and Medicaid Services (CMS). Meaningful Use Stage 1 (MU1) criteria were established and implemented in 2010. Meaningful Use Stage 2 (MU2) criteria were proposed earlier this year – originally intended with the intention for implementation 2013 – a date that has just recently been pushed back to 2014.

What is most important to genetic counselors about the Meaningful Use (MU) criteria is that health systems and providers will be focusing time and money on meeting MU criteria in order to qualify for incentive payments. There are several MU criteria that are of particular pertinence to genetic services. MU1 contains provisions to maintain problem lists of current and active diagnoses based on International Classification of Diseases (ICD)-9 or Systematized Nomenclature of Medicine (SNOMED) codes, and to generate lists of patients by condition to use for quality improvement, reduction of health disparities, and outreach. These two provisions point to EHRs that are capable of queries

that can, with advanced approval by an Institutional Review Board (IRB), return patient registries for diagnoses of interest to genetics providers.

For example, an EHR query of the ICD-9 code 237.3 would identify all paraganglioma patients within the EHR, allowing genetic counselors to reach out and coordinate appropriate genetics follow-up for these patients. MU2 contains a requirement that family health history be stored as structured (i.e., searchable) data, including adoption of the HL7 Pedigree Standard. This requirement has the potential to significantly improve the way EHRs store family history data, allowing the creation of patient registries of asymptomatic patients for appropriate offers of genetic counseling based on family history alone.

**It may reduce your per-patient workload: Automated record sharing**

Meaningful Use criteria also stipulate that EHRs must be interoperable, allowing for automated record sharing between providers and insurance companies, and similar third parties. This automated record sharing may reduce the need for separate letters of medical necessity (LMN) for genetic testing, as insurers could be given access to genetic counselors' encounter documents. Additionally, MU requirements for clinical summaries are improving EHRs' abilities to created succinct after-visit summaries, which genetic counselors could use in place of authoring separate patient letters. It is vitally important that genetic counselors critically appraise these features to determine the appropriateness of their use within their practice of genetic counseling.

Other elements of HIT that are already reducing per-patient workloads include pedigree drawing programs and internet decision aids. These tools are addressed in a second article in this issue of *Perspectives in Genetic Counseling* by **Kristin Baker Niendorf**, **MS**, **CGC** and **Emily Gabitzsch**, **MS**.

In conclusion, there is growing need for further innovation and collaboration in the development of HIT to address the demands for personalized medicine. As facilities are upgrading to EHRs, genetic counselors have the opportunity to play an active role in HIT development so that HIT solutions reflect the unique needs of our profession. We challenge genetic counselors to become knowledgeable about HIT and involve themselves in the implementation and meaningful use of HIT within the practice of genetic counseling.

# For Your Practice

## Information Technology Tools for Genetic Counseling Practice: What is available now and where do we go from here?

*By Kristin Baker Niendorf, MS, CGC and Emily Gabitzsch, MS*

Genetic counseling is a field of "new": new discoveries, new genes, and new treatments. As we grapple with so much novel information, we also need to prepare for technology that will assist us in caring for our patients and improving our practices. While the art of pedigree analysis, dysmorphology assessment, and genetic counseling can never be wholly mechanized, automation of some elements of genetics services could be a boon to genetic counselor productivity. Technology is currently available to automate some of these elements, including family history collection/pedigree generation and risk assessment.

### Past genetic counseling documentation methods

Family history collection has always been a component of medical care; however, in the past, family histories have been taken in various ways and used (or not used) as determined by the individual provider. Apart from the obvious benefit of identifying those at increased risk for hereditary conditions, a family history also provides important documentation that improves screening behaviors[1,2,3] and influences risk perception[4].

Usually, family history collection does not involve a nuanced and comprehensive assessment from a genetics professional, but relies on the more basic documentation found in primary care. As a result, evidence abounds of limited identification of at-risk families in medical practice and physician referral barriers to genetic counseling. The gap between specialties further reflects an unmet need.[6-10] To help with this, we have prepared a collection of information technology tools currently available to healthcare professionals who collect a family history, those who compute a genetic risk assessment, and those who do both (Tables 1 and 2).

### Current genetic counseling tools

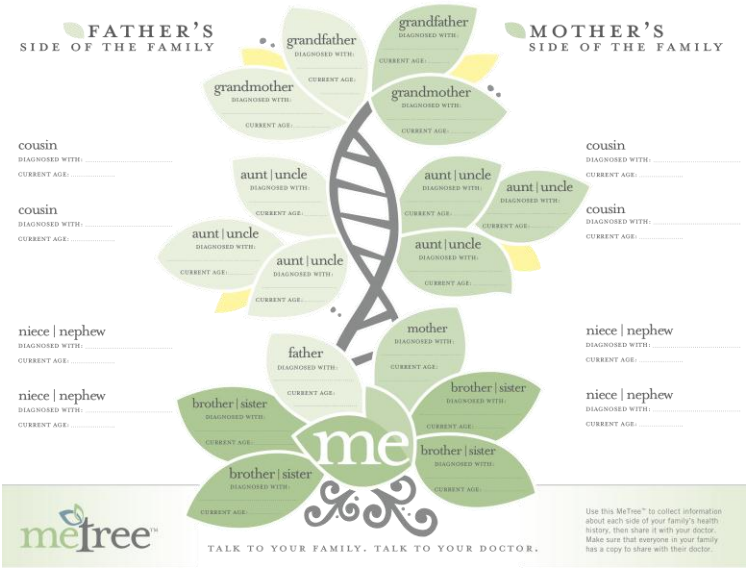
Depending on the intended user, family history collection tools vary in their breadth of coverage (number and degree of relatives), distinct details (consanguinity, assisted reproductive technology, adoptees), and items available for inclusion (cancer family history, cardiovascular disease, other).[5] Previous inquiries regarding use of online methods to collect a family history suggest that many patients find online family history tools desirable but, ideally, other options (e.g., paper format) should exist for those individuals not interested in online mechanisms.[11] Regardless of the source of the data, electronic pedigree generation tools may offer compatibility with the patient's electronic

health record (EHR), and provide systematic strategies to collect and update the family health history (FHH).[12]

In addition to documenting family history, risk assessment tools have been developed to aid healthcare professionals in personalizing genetic risk for individuals, determining the appropriateness of genetic testing, and offering tailored preventive healthcare messages to implement improved screening behaviors and lifestyle changes. Two examples of these are shown below. These differ in their intended areas of focus (cancer, heart disease, diabetes mellitus) and the data upon which they are built. The convergence of these tools (patient-entered pedigree and clinical algorithms to determine risk) holds great promise to 1) increase the amount and quality of family history, and 2) provide essential point-of-care clinical decision support and useful risk assessments. Current availability ranges from private tools within a healthcare institution, platforms that can be customized and adopted into specific healthcare systems, and those that are publicly available and/or aimed at the general population.


*"My Family Health Portrait," a pedigree generation tool for public use from the U.S. Surgeon General (https://familyhistory.hhs.gov/fhh-web/home.action)*

We outline several tools currently available in Table 1 and Table 2. Both will be made available on the National Society of Genetic Counselor's website. These tables are representative but certainly not a comprehensive list of tools currently available. Areas of interest for each tool include: online/electronic collection of family history data, pedigree creation, risk assessments, risk assessment recommendations, interaction with mutation probability software (e.g., CAGENE for hereditary cancer), production of risk assessment letter, interaction with EHR, production of genetic counseling summary note within or without the EHR, and research versus clinical focus. By reviewing these comparative tables, genetic counselors should be able to make informed choices about the benefits or disadvantages of a tool that may be used within their clinical or research environment. This comparison should also be of benefit to those programs that intend to, or are in the midst of, producing their own tools.




*"MeTree," a pedigree construction and risk assessment tool from University of North Carolina-Greensboro/Duke University Medical Center/Cone Health and an accessory to "The Genomedical Connection" risk assessment for 46 diseases (http://www.genomedical.com/whats_the_connection/your_connection.cfm)*

**Future directions**

As with any new technology, several versions will be proposed at the beginning. In the future, it will become clearer which version, if any, stands up to the test of time. Additionally, new innovations will be developed to address future needs. As genetic counselors, we are at the forefront of knowing what tools would benefit our patients and ourselves in our clinical practice.

The next step in this process is further systematic assessment of current tools. What utility do these tools bring to genetic counselors and other providers? To patients? What tools and features are desired by genetic counselors, and would best assist genetics care? Benchmarks for future success include validation of an increase in the following: identification of at-risk families, appropriate referrals for genetic counseling, improvement in efficiency of genetic counseling services and patient/provider adherence to medical management recommendations.

We raise a challenge to all genetic counselors to consider the use of technology within your practice. We are in the position to be in the driver's seat; let us be the guides.

**Table 1. Comparison of information technology genetics tools**

| Tool Name | Pedigree Generation | Risk Assessment | Available for Public Use |
|---|---|---|---|
| *My Family Health Portrait* | ✓ | | ✓ |
| *Our Family Health* | ✓ | | |
| *Health Heritage* | ✓ | ✓ | |
| *Hughes Risk Apps* | ✓ | ✓ | |
| *MeTree* | ✓ | ✓ | |
| *My Breast Health* | ✓ | ✓ | |
| *My FHH* | ✓ | ✓ | |
| *My Generations* | ✓ | ✓ | ✓ |
| *GenTri* | ✓ | ✓ | |
| *CancerGeneConnect* | ✓ | ✓ | |
| *Family Health Link* | | ✓ | ✓ |
| *B-RST* | | ✓ | |
| *Score Against Colon Cancer* | | ✓ | ✓ |

## Table 2. Clinical Genetics Information Technology Tools

Definitions: EMR = Electronic Medical Records, GC = Genetic Counseling, IT = Information Technology, NCCN = National Comprehensive Cancer Network, SNOMED = Systematized Nomenclature of Medicine, IBIS = International Breast Intervention Study, CCF = Cleveland Clinic Foundation, FDR = First Degree Relative, SDR = Second Degree Relative, MRI = Magnetic Resonance Imaging

| Tool | Creator | URL/Ref | Description | Components | Notes |
|---|---|---|---|---|---|
| **Jameslink/ Family Health link** | Ohio State University | URL (1) | Web-based, patient-entered risk assessment tool | - Publicly available online<br>- Family History data (first, second, some third-degree relatives); no ethnicity; Ashkenazi Jewish ancestry<br>- Cancer, CHD risk assessment<br>- No EMR compatibility | - High, moderate, or average risk summary<br>- No pedigree generated<br>- Patient letter recommendations include: GC referral, general medical management, health behavior promotion |
| **Hughes Risk Apps v. 2. 7-1** | Kevin Hughes | URL (2) | Identifies individuals at increased risk for breast cancer and hereditary breast cancer<br>Medical and family history data input via tablet computer (web access can be requested via IT support at cost) | - Two levels of questionnaires (standard, high risk)<br>- Generates risk estimates via BRCAPRO, Myriad, Gail, Claus<br>- Can export to Progeny, CaGene<br>- Generates clinic note (through IT contract, templates can be modified as institution-specific)<br>- Cut-off of 10% likelihood *BRCA* mutation (BRCAPRO) for increased risk (GC referral) | - Pedigree generated<br>- Requires tablet computer for patients<br>- Desktop format for providers<br>- Program is free to download individual copies, but requires IT support to implement "at cost" (versus retail cost) to institutions<br>- Trademarked<br>- Every update requires IT support<br>- System incompatible with other scheduling programs (may require building / purchasing interface) |

| Tool | Creator | URL/Ref | Description | Components | Notes |
|---|---|---|---|---|---|
| **My Generations** | Northshore Hospital-Evanston | URL | Web-based, patient-entered family health history and risk assessment tool | - North Shore Hospital site for storing cancer family history<br><br>- Risk assessment for many cancer syndromes based on BRCAPRO, Gail, and Hampel algorithms<br><br>- Includes ethnicity, Ashkenazi Jewish ancestry<br><br>- Free to all users; public information stored in secure database | - Free of charge<br><br>- Pedigree generated<br><br>- Personalized risk assessment includes both general cancer risk and risk for hereditary cancer, and instructions for patient to follow-up |
| **Health Heritage/ GenE EMR** | University of Virginia<br><br>Current Funding: National Cancer Institute<br><br>Past Funding: Robert Wood Johnson Foundation | URL (3) | Decision support tool for cancer risk assessment based on personal and family history risk factors | - Patient portal<br><br>- Pertinent health information from record pre-populated into tool for application of decision support risk algorithms.<br><br>- Uses Surgeon General's tool for family history collection, internal risk assessment algorithm<br><br>- Assessment of 7 cancers; planned expansion to other diseases<br><br>- Personalized risk report includes evidence-based risk reduction recommendations, drawing primarily from NCCN<br><br>- Risk report displayed for the patient and shared in Epic with primary care provider | - Pedigree generated<br><br>- Risk levels: significantly increased, moderately increased, general population<br><br>- In pilot, a number of participants took >1 hour to complete<br><br>- Pilot found the tool identified 60% of specific conditions vs. 24% from chart review<br><br>- Available for research collaborations, (especially Epic users)<br><br>- Stand-alone Health Heritage tool also available for settings without an EMR<br><br>- Capability for sharing family medical data electronically, reducing need for self-reported family health history entry |
| **My Family Health Portrait** | Health and Human Services, HHS | URL (4) | Surgeon General's web-based, patient-entered family health history collection tool | - Publicly available online<br><br>- Customizable for use by centers<br><br>- Includes ethnicity, twins, adoptees | - Family history collection displayed in tabular format and pedigree<br><br>- No risk assessment |

| Tool | Creator | URL/Ref | Description | Components | Notes |
|---|---|---|---|---|---|
| **Our Family Health** | Intermountain | URL (10) | Web-based, patient-entered family health history collection tool | - Secure patient portal; internal health system<br><br>- No EMR compatibility (but planned)<br><br>- Handles twins, adoptees, divorced / separate partners, first-, second-, third-degree relatives<br><br>- Print-out of family history available in tabular form | - No risk assessment (but planned)<br><br>- Features "common condition" box, and additional search box of over 500 conditions (SNOMED)<br><br>- Quickstart guide gives users option to start with blank pedigree, input integer number of relatives for a skeleton pedigree, or upload GEDCOM data |
| **MyBreast Health** | Amigenics | Contact Erica Ramos: Eramos77 @gmail.com | Amigenics tool for collecting personal and family history for use by genetic counseling in High Breast Cancer Risk Clinics. | - Patient portal family history via secure website<br><br>- Pedigree generation via Progeny integration<br><br>- Risk assessment: Gail, Claus, Penn II, Myriad, BRCAPRO, Boadicea, IBIS<br><br>- GC summary note created | - Is available on a fee-basis by contacting Erica Ramos |
| **Breast/ Ovarian Cancer Genetics Referral Screening Tool (B-RST)** | Cecelia Bellcross | URL (7, 8) | Simple, validated online screening tool which identifies individuals at 5-10% or greater risk for *BRCA* mutations to prompt genetic counseling referral | - 8 question online tool, free of charge<br><br>- Patient and provider portal<br><br>- Generates printable report with result, interpretation, links, family history information<br><br>- Includes definitions, resources and references pages | - Screens positive / negative (low & moderate)<br><br>- Intended to quickly screen for patients appropriate for cancer genetic counseling referral<br><br>- Tablet application in development |
| **Score Against Colon Cancer** | Cleveland Clinic | URL | Web-based, patient-entered risk assessment for colon cancer | - Risk assessment, including behavioral and lifestyle recommendations provided to patient | - No pedigree generated<br><br>- Risk levels of high, medium, low |

| Tool | Creator | URL/Ref | Description | Components | Notes |
|---|---|---|---|---|---|
| **MyFHH/ CDST** | Cleveland Clinic | Internal provider / patient access only | Web-based, patient-entered family history collection and risk assessment tool | - Currently used for Cleveland Clinic patients only<br>- Primary-care focused<br>- EMR compatible | - Pedigree generated<br>- Risk assessment to provider |
| **CancerGene-Connect** | University of Texas Southwestern | URL | Online patient driven cancer genetic risk assessment program<br>Virtual Genetic Counselor Environment | - Online family/medical history collection<br>- Risk Models (Claus, Gail, BRCAPRO, MMRPRO, PanPro, MelaPRO)<br>- GC documentation<br>- Provides online fact sheets on Cancer syndromes<br>- Data Collection and Reporting function | - Pedigree generated<br>- Currently only available at UT Southwestern, (but plans to offer system to other cancer genetic sites)<br>- English and Spanish versions<br>- Genetic Navigation system |
| **GenTri/ Progeny** | North Memorial Healthcare/ Humphrey Cancer Center/ Progeny | Contact Joy Larsen Haidle: Joy.larsen.haidle@northmemorial.com | Tool for collecting personal, medical and family history information; risk assessment and oncology patient care | - Cancer risk assessment<br>- Data collection by web questionnaire, Tablet, digital pen<br>- Uses General Oncology questionnaire<br>- Family history information<br>- Risk assessment letter<br>- EMR integration | - Pedigree drawn automatically in Progeny<br>- Useful for triaging clinic population into cancer genetics clinic<br>- Once in Progeny, data can be transferred into specific risk assessment models |

| Tool | Creator | URL/Ref | Description | Components | Notes |
|---|---|---|---|---|---|
| **MeTree** | University of North Carolina - Greensboro/ Duke University Medical Center/ Cone Health | URL (9) | Family history intake & decision support program for select cancers & thrombophilia | - Patients enter personal & family history of 46 conditions on touch screen computer prior to primary care visit<br><br>- Output includes pedigree & decision support for 4 pilot diseases: breast, ovarian & colon cancer, & thrombophilia<br><br>- Patient & physician receive decision support printouts to discuss during visit<br><br>- Research participation | - Pilot disease decision support based on published professional guidelines/expert opinion<br><br>- Decision support is action based (e.g., refer to genetic counseling) rather than a risk level<br><br>- Collects data on FDRs & SDRs<br><br>- Runs Gail model for breast cancer chemoprevention recommendations<br><br>- Runs BRCAPRO (lifetime breast cancer risk) for breast MRI recommendations<br><br>- May add cardiovascular disease, diabetes modules<br><br>- May become available outside research context |

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# Licensure / Billing & Reimbursement

## Coding Corner

### New NSGC "Ask the Expert" Discussion Forum for Billing, Coding, and Licensure Questions!

*By Pia Summerour, MS, CGC and Kaylene Ready, MS, CGC*

*The Coding Corner is supported by the Coding Subcommittee of the National Society of Genetic Counselors' (NSGC) Access and Service Delivery Committee and aims to assist NSGC members with the application and understanding of governmental regulations and guidelines regarding terminology and Current Procedural Terminology (CPT) / International Classification of Diseases (ICD) coding in genetic services as well as keep the membership educated regarding billing and reimbursement issues.*

We are happy to report that a new discussion forum for questions about billing, coding, and licensure has been created on the NSGC website. This new discussion forum will provide the easiest and most efficient way to get answers for all your billing, coding and licensure questions! Since these topics affect, or have the potential to affect, many genetic counselors, we encourage all interested genetic counselors to join the forum. The Billing, Coding, and Licensure "Ask the Expert" Discussion Forum will be moderated by the Coding Subcommittee Co-Chairs, **Pia Summerour** and **Kaylene Ready**.

Answers to questions about billing, coding, and licensure are complex, can vary by region, state, and licensure status, and must be mindful of other legal issues, such as antitrust laws. Therefore, unlike other discussion forums, submitted questions will not immediately appear on the website and answers will only be posted by the moderators and other topic experts after review. By structuring the discussion forum in this way, our goal is to provide the most accurate and up-to-date information. No more confusion or misinformation!

Here's an overview of the process. First, you submit a question to the Billing, Coding, and Licensure "Ask the Expert" Discussion Forum. Then, your question will be reviewed by the moderators and a panel of experts to generate the most informative answer. Depending on the complexity of the question, turn-around time for an answer may vary.

Once an answer is generated, the question and its answer will only be posted to the discussion forum, and a notification will be sent to all discussion forum members. If a question is initially posted on a different forum, such as the General Discussions Forum, the question will be answered on <u>both</u> the General Discussions and the Billing, Coding, and Licensure Discussion Forums. However, to maximize efficiency, eventually these types of questions will <u>only</u> be answered on the Billing, Coding, and Licensure

Discussion Forum. Therefore, we strongly encourage all interested genetic counselors to join the Billing, Coding and Licensure "Ask the Expert" Discussion Forum so that you have access to important information!

To join the Billing, Coding and Licensure "Ask the Expert" Discussion Forum, just follow these simple steps:

1. Log in as a member on www.nsgc.org
2. Click on "Member Center"
3. Under "What's New", Click on "Discussion Forums"
4. Under "General NSGC Discussion Forum," click on the "Billing, Coding and Licensure" link
5. Click in the box next to "Click to subscribe to this forum" to receive emails when questions and answers are posted

We hope to hear from you soon!

*The Coding Corner is your resource for questions about coding. If you have questions you wish to be considered for this section, please send them to* ***Pia Summerour*** *(pia.banerji@utsouthwestern.edu) or* ***Kaylene Ready*** *(kaylene@counsyl.com)*

# SIG Speak

## From the Pediatric and Clinical Genetics Special Interest Group

### Whole Exome Sequencing in the Medical Genetics Clinic

*By Carolyn Dinsmore Applegate, MGC, CGC*

About two weeks into January, I turned to my clinical Director and said, "Do you realize that what we are saying to patients is completely different than what we were telling patients before Christmas?" We sighed, shook our heads, and reminded each other that this is part of what makes genetics such an interesting and challenging field.

What changed in our clinic as of January 1st? The availability of clinical whole exome sequencing (WES) through three, U.S.-based, genetic testing laboratories and the launch of the Baylor-Hopkins Center for Mendelian Genomics (BHCMG), a large, National Institutes of Health (NIH)-funded grant project aimed at identifying the genetic cause of unknown Mendelian inherited disorders by WES. Initially, WES appeared to be a natural extension of our clinical and research practices but, as we began consenting patients for clinical WES and enrolling patients into the BHCMG, we realized this technology raised a multitude of practical, clinical, technical, and ethical issues that we had only partially anticipated. The most pressing issues that our clinic faced were to whom this testing should be offered, at which point in time, and which patients to enroll in research versus clinical WES.

The initial patients were easy to identify: those who very likely had a genetic cause for their clinical history, but lacked a molecular diagnosis and were desperate for an answer for the purposes of prenatal diagnosis or peace of mind. These were patients that we could name off the tops of our heads. Concurrently, we began consenting families and individuals for the NIH Mendelian study. These were families with genetic conditions without a molecular diagnosis, but whom we suspected had a mutation that would hopefully be identified by WES. These families were also motivated, but the sense of urgency was not present.

After arranging clinical WES for the initial patients and obtaining consent from a number of families for research WES, the decision to offer clinical versus research WES became much more complicated. When deciding whether to offer a patient clinical exome sequencing versus research exome sequencing, we as clinicians and research consenters have an inherent bias in that we know how patients will be prioritized.

Families with more than one affected individual and/or consanguineous families will be among the first sequenced, whereas isolated probands are unlikely to be sequenced in the near future. Therefore, if the goal is to quickly establish a molecular diagnosis, clinical WES sequencing appears to be the appropriate choice for many of our families. The fact that clinical WES will provide a result within a specified time frame, even if negative, is

important to some families, as the open-ended nature of research can cause more stress and anxiety than a definitive answer.

A family's motivation for pursing WES can also affect the decision to perform clinical versus research WES. If the family is motivated to find the molecular cause of the genetic condition in their particular family, clinical WES tends to be more appropriate. If the family is comfortable with genetic ambiguity, less concerned with molecular diagnosis, and motivated to contribute to the field of genetics as a whole, research WES may be more appropriate. Many families express both of these factors as motivation to pursue WES, but establishing the primary motivation can help determine the type of testing that would be most useful.

Cost and insurance coverage are also factors when deciding between clinical and research WES. Clinical WES requires payment by insurance or the patient, and grant funding covers research testing. The ability to obtain samples from other family members can also play a role in the decision to pursue clinical versus research WES. Most clinical laboratories rely on trio testing (proband and two parents) to narrow down the list of candidate variants. Thus, if the parent(s) or other family members that are necessary to perform segregation analysis are unavailable, research testing may be the more practical option.

Discussion of all of the aspects of clinical versus research testing can be very time consuming, but thus far at our institution we have been successful in developing a testing strategy that is agreeable to both the patients and providers. Even though many aspects of these counseling/consenting sessions are novel, we continue to utilize the same genetic counseling process with which we are all familiar.

Even if it is clear that clinical WES should be performed, there is still the issue of optimal timing to offer and perform this testing. Some questions that arise include:

- What is the probability of identifying a causative mutation?
- How likely are we to make an incidental diagnosis?
- How likely are we to find variants of unknown significance?
- What role do incidental findings play in determining the timing of testing?
- When should WES be performed as the first line genetic test?
- What are the technical and analytical limitations of the current clinical platforms and laboratory tests?
- What are the limitations in our ability to understand and interpret the results?

In a general medical genetics clinic, a large proportion of patients appears to have a genetic cause underlying the medical condition(s) and/or intellectual disability/ies they have; however, we are unable to either establish a syndromic diagnosis based on clinical features or identify a causative mutation by molecular testing. Many established patients have already had the standard biochemical and cytogenetic work-up, in addition to sequencing analyses of candidate genes. These patients are appropriate candidates for WES.

The appropriateness of clinical WES becomes less obvious with the remainder of patients. One can argue for or against clinical WES based on cost-effectiveness. For instance, if one gene or panel has a high likelihood of identifying a mutation and is less than the cost of WES, then traditional single-gene testing for that candidate gene or panel is most cost-effective. On the other hand, if we have more than one candidate gene in mind, with the additive cost being equal to or greater than the cost of WES, then clinical WES would be the more cost-effective approach.

Cost-effectiveness as a determinant of when to send site-specific genetic testing versus clinical WES appears to be a simple decision-making tool until you consider that: 1) WES does not cover all exons and genes equally, and 2) clinical WES may lead to incidental diagnoses or a large number of variants of unknown significance. For example, the emotional cost of possibly diagnosing an adult-onset condition in a one-week-old baby with multiple congenital anomalies may outweigh the cost-effectiveness of WES.

Another hypothetical scenario is one in which a parent comes to the clinic for a very specific purpose: their child has an intellectual disability and/or dysmorphic features, but they also have a family history of an adult-onset condition for which the parent has explicitly chosen to forego pre-symptomatic testing. WES could potentially identify not only the status of the child, but also of the parent and other family members who have not yet been tested. Ethical dilemmas are far from new to the field of genetics; however, this technology may lead to a greater number of ethical issues or, at the very least, to unprecedented situations.

The cost-effectiveness of clinical WES is also difficult to quantify due to technical limitations and differences in laboratory policies. The coverage of specific genes and exons varies between platforms and between laboratories. Thus, in some cases WES may not identify a mutation in a particular gene or exon due to the fact that there is scanty coverage of that region. Some labs will provide a list of the genes/exons that are not covered to a certain threshold, and other labs will provide information about specific genes upon request. In either case, there is no way to know before testing occurs whether specific genes will be sufficiently sequenced.

Laboratory policies on data storage and re-analysis are also a factor when determining the value of clinical WES. Some laboratories destroy the data once analyzed, some laboratories will provide the patient/provider with the variant call files, and some laboratories are maintaining the data so that re-analysis can be performed in the future. At this time, it is not clear what the follow-up will be for patients with negative WES results. Will we order reanalysis of previously derived data or collect new samples to be tested on newer platforms? These issues are not quantifiable with respect to cost, but play a role in determining the value of clinical WES for the sake of a cost-benefit analysis.

As is commonplace with genetic testing, there is the issue of insurance coverage. Thus far, insurance companies have been surprisingly agreeable to covering clinical WES. It's unclear, however, whether this will continue. Unfortunately, some insurance companies

have set forth policies stating that chromosome microarrays are experimental; one could foresee these companies taking a similar stance on WES. One could also argue that the cost-effectiveness of WES versus the traditional model of gene-by-gene testing will encourage insurance companies to cover WES.

It will be interesting to see whether insurance companies will cover a second WES on the same individual as platforms naturally improve over time. From the perspective of a genetic counselor who has spent many hours getting insurance pre-authorization for testing one gene – only to have to go through the process again when the first genetic test result was negative – the thought of having to get insurance pre-authorization for only one test per patient is quite enticing; a greater proportion of our time could be spent on providing direct patient care, and less time would be spent providing indirect patient care such as obtaining insurance authorizations.

At present, there are no published guidelines regarding utilization of clinical whole exome sequencing in the medical genetics clinic. Also, laboratory guidelines about reporting medically actionable incidental findings are currently non-existent. However, the practical, clinical, and ethical issues that arise with this technology are not entirely new to the field of genetics or genetic counseling. As we continue to work through these very real ethical, practical, and technological dilemmas, we need to let our past experiences guide us and remember our primary role as patient advocates who provide comprehensive knowledge to promote autonomy.

*If you would like additional information, the Pediatric and Clinical Genetics SIG is actively creating resources and references about whole exome sequencing for the National Society of Genetic Counselors' membership.*

# NSGC News

## NSGC Committee Updates

Have you wondered what type of activity is taking place within the National Society of Genetic Counselors' five management committees? Below, you will find an update on many exciting activities that were underway during the first quarter of 2012. Committee updates will periodically be presented in *Perspectives in Genetic Counseling* to keep you informed and help you determine where you might want to get involved!

### Access and Service Delivery Committee

**Shanna Gustafson**, Chair
**Stephanie Cohen**, Vice Chair

- Continues to educate members, legislators and key stakeholders regarding state licensure for genetic counselors.
    - Received three applications for state licensure grant awards.
    - Additional states are being supported at various levels of the process.
    - New Hampshire and Nebraska recently held Senate hearings.
    - Rhode Island is developing a conscience clause with American Civil Liberties Union (ACLU).
- Developing a webinar on personalized medicine to educate the NSGC membership on its use and benefits.
- Continues to educate members regarding Current Procedural Terminology (CPT) coding issues to help promote NSGC coding consults for members' employers/institutions.
    - An article on International Classification of Diseases (ICD)-10 delay was published in the Spring 2012 *Perspectives in Genetic Counseling* issue.
    - Watch for a new discussion forum on Billing/Reimbursement.
- Service Delivery Models Task Force continues to work on two publications about service delivery models.
- Payer Task Force is developing and expanding relationships with third party payers. A reference sheet on commonly asked questions for payers and a list of tools to enhance progress have been developed.

### Practice Guidelines Committee

**Margo Grady**, Chair
**Adam Buchanan**, Vice Chair

- Updated the practice guideline development and approval process. Proposals for several new guidelines are under review.

- Guidelines in the final stage of review and/or publication include:
  - Cystic fibrosis
  - Fragile X syndrome
  - Prenatal screening
  - Prenatal microarray
  - Hereditary breast and ovarian cancer

**Communications Committee**

**Kimberly Barr**, Chair
**Sara Riordan**, Vice Chair

- Monitoring internal and external communication vehicles to ensure NSGC is consistently incorporating our brand messages and member value proposition.
- Started initial discussions for the Physician Reference Link project. The committee is formalizing a program to post reference links on hospital/physician websites directing users to NSGC. The primary intent of this project is to create reference links on hospital or healthcare providers' websites.
- Beginning an assessment of current publications to make recommendations regarding the content to be developed, updated or enhanced.
- Currently working to provide feedback on phase two of the marketing tool kit.
  - Collaborating with the Special Interest Groups (SIGs) to develop specialty-specific content in line with the brand messages.
  - Continuing to provide recommendations for content specific to physicians and healthcare providers.

**Education Committee**

**Leigha Senter**, Chair
**Kelly Jackson**, Vice Chair

- The Annual Education Conference (AEC) Subcommittee has selected the Preconference Symposia, Plenary presentations, and Educational Breakout Sessions for the 2012 AEC. The Abstract Workgroup is now selecting concurrent papers and posters following the close of the Call for Abstracts on May 14, 2012.
- The Webinar Subcommittee scheduled six webinars at 12:00pm Central Standard Time to be held in 2012:
  - January 25 - Navigating the New NIH Genetic Testing Registry
  - March 28 - Fetal Diagnosis and Intervention: Past, Present and Future
  - May 16 - Advanced Degrees in Genetic Counseling: What are the Options?
  - July 25 - TBD
  - September 26 - From Seeking to Finding: Improving Your *PubMed* Searches

    - November 28 - TBD
- The Online Course Subcommittee is finalizing the 2012 course, focused on "Why choose just one gene? Large panel tests for genetic conditions." Additional information was made available in late May 2012.
- The Outreach Education subcommittee is reviewing proposals from the Prenatal and Assisted Reproductive Technology SIG.
- To keep up with the demand for reviewing both Category 1 and Category 2 Continuing Education Unit (CEU) applications, the CEU Subcommittee has added two review teams in 2012, for a total of 17 review teams.
- The *Journal of Genetic Counseling* CEU program opened February 15, 2012.

**Membership Committee**

**Bronson Riley**, Chair
**Kami Schneider**, Vice Chair

- The Awards Subcommittee administered the 2012 Student Rotation Scholarship program, awarding one summer rotation with Lineagen Inc. Launched the new cycle of the NSGC Mentorship Program and is preparing for the next session launch in June 2012.
- Reviewed the 2012 Nominations process and provided recommendations for process improvements and three additional at-large members to be added to the Nominating Committee and Board of Directors.
- The Professional Status Survey (PSS) Subcommittee closed the 2012 PSS survey in February. The survey results are now available for the membership.

**Public Policy Committee**

**Flavia Facio**, Chair
**Jill Stopfer**, Vice Chair

- Submitted a presentation entitled, "The new landscape of genetic testing: How to approach testing minors for adult onset conditions in the era of large scale genomic testing." This was selected for an Educational Breakout Session at the 2012 AEC.
- Completed the NSGC Position Statement and White Paper on Non-Invasive Prenatal Testing.
- Completed the NSGC Position Statement and White Paper on Newborn Screening.
- Completed the NSGC Position Statement and White Paper on Blood Spot Storage.
- Convened Task Forces to work on revising:
    - Stem Cell/Fetal Tissue Research Position Statement
    - Testing Minors for Adult Onset Conditions Position Statement

- Revised a new liaison education and reporting process to ensure liaisons have the appropriate level of support and information to effectively represent the NSGC with various external organizations.

# ABGC Update

## The ABGC Launches Two New Initiatives

*By the ABGC Board of Directors*

The American Board of Genetic Counseling (ABGC) is excited to announce the launch of two new initiatives: the online **Practice Examination** and an **Online Recertification System**. These programs were developed to expand the services provided by the Board in response to feedback from our diplomates (both current and future). More details are below.

### Online Practice Examination

The Practice Examination (PE) is available for purchase now. The PE is a 100-question online examination developed by the ABGC to mirror the content and difficulty of the actual ABGC Certified Genetic Counselor® Examination.

The PE can be a useful tool to assess areas of strength and weakness in your exam preparation, and to give you practice answering questions. Upon completion of the PE, the candidate will receive several performance reports, including an overall score report that includes 1) the number of questions answered correctly in each area of the Detailed Content Outline; 2) a feedback report that lists each question and whether it was answered correctly (green checkmark) or incorrectly (red X); and 3) a mastery report showing the percentage of questions answered correctly in each area of the Detailed Content Outline.

Access to the PE and performance reports is available for sixty days from the date of purchase. Individuals can move in and out of the PE as many times as they wish during this period, provided that they have not yet marked "Finish and Display results" or "Finish and Exit." **Once these buttons have been selected, the individual may access the reports only.** The PE also has a timing feature that allows the examinee to monitor the amount of time it takes to complete the exam. This helps examinees prepare for the actual exam by ensuring that they are moving through the questions at an adequate pace.

The current cost of the PE for genetic counselors, including graduates and students, is $55. A group discount is available for five or more copies of the PE purchased at one time, with each copy currently available for $50.

**Online Recertification System – now live!**

The Online Recertification System (ORS) allows diplomates to enter and track their continuing education activities, and complete the recertification process entirely online.

To access the ORS, click on the "Member Login" button on the ABGC website or click on this link: ABGC Member Login. You will need to use your Member ID # as the login and your current password. If you do not have your ABGC Member ID and password, contact the Executive Office at info@abgc.net or 913.895.4617.

Once you have logged in, click "Recertify" in the upper right corner of the page to begin entering continuing education units (CEUs) and/or Professional Activity Credits (PACs). The "My CEUs/PACs Summary" page will appear. You may enter as many CEUs/PACs as needed at any time prior to your certification expiration date. The system will save and store the information for you, and the summary of CEUs/PACs will be automatically updated. This summary can be found on the "CEUs/PACs Summary Table" at the bottom of the screen.

Although you may enter CEU/PAC activity at any time, you may ***only*** recertify in the year that your current certification is set to expire. Once you meet the required minimum number of CEUs (25 CEUs for Certified Genetic Counselors (CGCs) with 10 year certification; 12.5 CEUs for CGCs with 5 year certification), the "Recertify" button will be activated and you can click it. If you owe Certification Maintenance Fees (CMF) for previous years or the current year, you will be prompted to pay the outstanding CMF fees online before you can recertify.

After the system processes your recertification information, you will receive a confirmation online and via email that the recertification process is complete. A ***random sample*** of recertification applications will be selected for ***audit*** to confirm the validity of all CEUs/PAC information submitted. Your confirmation will clearly indicate whether or not you have been selected for audit. Those audited will be required to submit documentation of attendance and/or participation in an activity to support their claimed CEU/PAC credits. **Therefore, it is critical for a diplomate to retain all documentation even though s/he may be recertifying online.** For information on the appropriate documentation for each CEU/PAC credit, go to www.abgc.net.

After your recertification is complete, your electronic account will automatically be updated with your new certification expiration date. Within thirty days of recertifying, the ABGC will mail you a new certificate reflecting the new expiration date. All new certificates will be issued for a five-year period ending on December 31 of the final year of certification.

# Global Genetics

*By Rawan Awwad, MS, CGC*

When I decided to move back home to the Palestinian Territories in 2008, I had been studying and working in the United States on a non-immigrant visa for years. I was excited by the potential of giving back to a community that needed my services, but overwhelmed with the fear that my career might plummet if I could not create a job there. I was, after all, moving back to a place where genetic counseling services did not yet exist. If things were not going to work out for me in the Middle East, obtaining another U.S. work visa was not going to be a guaranteed or straightforward process. Living close to my family, however, was enough of a priority at that time that I was determined to take a risk and make it work.

Several months before making the move, I got in touch with genetics professionals in both the Palestinian Territories and Israel. As is commonly known, the politics of the Palestinian-Israeli coexistence in that historical region of the Middle East is extremely complicated. For the purpose of this article, it is important to point out that the governments, health care systems, insurance plans, freedom of movement, and quality of life for both people are not equal and are, in fact, non-comparable. In the occupied Palestinian Territories west of Jordan, commonly known as the "West Bank," there is one board certified clinical geneticist, no genetic counselors, and limited genetic laboratory services. There are several Palestinian health care professionals in the region who are working to create more services, but with limited resources and outside support, there are struggles of sustainability. It is in the West Bank where my hometown, Ramallah, is located.

In Israel, there are several advanced genetics centers, multiple geneticists, multiple genetic counselors, a genetic counseling program within the Hebrew University of Jerusalem, and a wide array of molecular, cytogenetic, and biochemical testing services. As a Palestinian born in East Jerusalem, I was able to access (live, travel, and work in) both regions. Working inside the West Bank meant serving Palestinians who needed my services, but in clinics with extremely limited resources. Working inside Israel meant having access to a large variety of advanced services, but serving only those Palestinians who were physically able to cross the border to Jerusalem and reach Israeli medical

centers. As mentioned above, freedom of movement is not a privilege of every Palestinian living in that region. Palestinians in the West Bank and Gaza Strip are occupied and have no citizenship rights.

Contacting professionals in the region before my arrival was not entirely successful. In-person communication was more hopeful, but I still had a long way to go. I ran into issues of staff limitations, funding, licensure, or simply no need to hire a genetic counselor when the work was covered by a physician/geneticist.

After several months of actively seeking to create a position, I decided to volunteer at the Human Genetics Center at Hadassah University Medical Center in Jerusalem, one of the largest clinical genetics centers in the region. My training in the U.S. and my bilingual skills in Arabic and English were useful. I was needed to serve the Arabic speaking population at the hospital. I worked with dedicated pediatric and prenatal clinical geneticists and genetic counselors who served both Israelis and Palestinians. I am particularly appreciative of Dr. Annick Rothschild, a pediatric geneticist who saw a great need for my services with the Palestinian population. She was keen to have me work with her on pediatric cases, which opened up several doors of opportunity for me in the region. I am also grateful to Professor Vardiella Meiner, the Medical Director of the genetics center, who supported my work and my incorporation into the department.

Having the respect and support of these geneticists, I decided to try to extend services into the West Bank. I wrote letters of support for entry permits for Palestinians with chronic and/or undiagnosed conditions to enter Jerusalem for genetic assessment and follow-up. For patients who were denied permits, depending on the indication, I travelled to their home towns and we met in public places or inside their homes for genetic counseling. When appropriate, I coordinated patients' blood draws and transferred their samples back to the hospital for further evaluation by the geneticists. As I worked with these patients, I identified areas of ethical and social concern for Palestinians and presented these cases at professional conferences locally and in Europe.

Billing and reimbursement for services were not easy. In general, because Palestinian insurance plans did not cross over to the Israeli health care system, the only billing option available was out-of-pocket with discounted prices. This was not feasible for many families. There were a few non-governmental organizations willing to cover the costs of certain procedures for chronic conditions, but the coverage was not always guaranteed. For Palestinians from East Jerusalem, who were the majority of patients able to reach the center, they had the same insurance plans as Israelis; those plans covered most visit and genetic testing costs.

Over time, I was successful at getting compensation for my time. I worked part-time on a grant-funded research project with Professor Azaria Rein, the head of Pediatric Cardiology at Hadassah, to create a database of congenital heart defects of unknown etiologies among Palestinian families. After certification by the Israel Board of Medical Genetics in 2009, I was offered a salaried position as a prenatal genetic counselor through Hadassah to work at a satellite clinic (Maccabi Medical Center) in Jerusalem. I saw

Palestinian patients independently and Professor Meiner signed off on my reports. The types of indications I counseled people for were similar to what I saw people for in the U.S., except that the cultural and psychosocial issues were very different. In addition, the high level of consanguinity among Palestinians provided exposure to unique cases.

Among the several rewarding experiences for me in the Palestinian Territories was participating in the early stages of the creation of the first Palestinian genetic counseling center. I participated in designing a training workshop on genetic counseling, alongside two other Palestinian professionals. They are Dr. Samir Khatib, PhD and Dr. Wafa Dakkak, MD, the founders of The Genetics and Metabolic Diseases Center, in Al-Quds University in East Jerusalem. The workshop aimed to raise awareness among doctors, nurses and social workers in the Palestinian Society, and to increase referrals. The workshop received a lot of positive response, and the center is still receiving patients today.

In 2010, the National Society of Genetic Counselors recognized my efforts by awarding me the International Leadership Award. It was a tremendous honor for me. I am now back in the United States and live with my husband in Michigan, where he has been based for years. I work as a pediatric genetic counselor at Children's Hospital of Michigan, Detroit, serving both the Arabic and English speaking patient populations. I am still active at extending services to the Middle East, and currently work on a project with Wayne State University's Pediatric Research Center, under the direction of Professor William Lyman, PhD, to start newborn screening in the Palestinian Territories.

Through my work in the Middle East, I learned that:

- **Being persistent and willing to volunteer can be more effective in grabbing the attention of a potential employer than prior experience and degrees/certificates earned.** They are excellent ways to prove how valuable our services can be to a clinical practice.
- **Knowing the language of the population of interest is very helpful; however, knowing the culture is just as important.** There is more to how people communicate than the words they say.
- **Bureaucracy in the work place is everywhere, and is usually a hindrance to effective progress of any newly introduced system.** Invested health care professionals are always interested to hear new ideas, but whether the implementation of these ideas will be supported is a totally different thing.
- **The Palestinian population is rich and interesting from a genetic perspective.** There is a high level of consanguinity with a high incidence of rare conditions, which are often confined to very small geographic regions within the country. Sometimes carrier screening is tailored based on the region of origin. In addition, the occasional detection of more than one recessive mutation for the same condition within members of highly consanguineous families brought about theories of "heterozygote advantage."
- **Psychosocial and ethical issues related to genetic testing among Palestinians are different from what I experienced in the U.S.** For example, there is no

defined age of majority; rather, autonomy in decision-making may be associated with certain life events, such as marriage.

Political struggle and tension interferes with every aspect of life for the Palestinians. These hardships influenced the effectiveness of extending health care services into the West Bank. Nonetheless, it was extremely rewarding to help several families there, and the hope is to be able to extend services on a much larger scale in the future.

*If you are or know of a genetic counselor with an interesting international story, please contact **Janice Berliner**, column editor, at berlinej@mskcc.org to discuss submission of an article.*

# Student Forum

## Through the Eyes of a Genetic Counseling Student: My experience at the 2011 National MPS Society Family Conference

*By Julie Jesiolowski, MS, University of North Carolina at Greensboro, Class of 2012*

My experiences in graduate school have taught me that families are resilient; despite hardships and turmoil, many people can handle the cards dealt to them by the unpredictable wheel of life. Families dealing with mucopolysaccharidoses (MPS) could be the "poster children" for such resilience. After a child is diagnosed with an MPS, uncertainty and fear can be incapacitating for the entire family. Although some forms of MPS have treatments available, others types are still in the beginning stages of research and are not well understood. Thus, some families are faced with the devastating news that their child may never gain independent skills or reach adulthood. And yet even after experiencing this overwhelming diagnosis, these families develop strong bonds with one another and cherish each day with their children.

When I was selected to intern with the National MPS Society in the summer of 2011, I did not realize how impactful this experience would be. As part of the internship, I attended the annual family conference sponsored by the society in St. Louis, Missouri. This event offers informative breakout sessions and lectures by experts on the disease and provides invaluable networking opportunities as families bond with one another, celebrate successes, share challenges, and remember those who have lost their battles.

I got off the plane in St. Louis barely able to pronounce "mucopolysaccharidoses," but eager to learn as much as possible. While waiting for the hotel shuttle to pick me up, I met my first family with a child who has an MPS. Twelve-year-old Emily* looked at me from her wheelchair with bright blue eyes. I greeted her and her family, and while she could not say hello back to me, I hoped she could understand. Her family was incredibly sweet and open with me. I knew at that moment that this conference was going to be very special.

As I was preparing for the welcome dinner, I was unsure what to expect but excited to meet each of the families. Before going to the dining area, I decided to visit the childcare room, dubbed "Camp Courage." Upon entering the hotel ballroom, I found a space designated exclusively for children with an MPS and their siblings to play and express themselves. My initial anxiety quickly subsided as I met the adorable children that lit up the room with their smiles. Within minutes, I was surrounded by a semicircle of children and holding two in my lap. Puzzles were dumped, toys were thrown, and loud shrieks of joy were all around me. These kids knew they could be themselves, and their energy was contagious. I played dolls with one hand and built a puzzle with another. I have done some multitasking in graduate school, but this was an entirely new level!

*Emily checking me out between break-out sessions*

*Enjoying dinner with Ariana*

Throughout the weekend, I was welcomed by each of the families with open arms. They shared intimate details about daily living with an MPS and the effect it has on their relationships and lives. I laughed while looking at scrapbooks of silly photos and listened as wonderful memories were shared. I cried with them during the memorial ceremony as we remembered those who had lost their fights. I could sense the anxiety in their voices during the clinical trial lectures; I too was desperately wishing that there would be a promising trial for each and every one of these children.

Many parents shared concerns with me about the lack of knowledge among health care professionals about forms of MPS; often, these families know more about the disease than their own primary care providers. They spoke of the pain of watching dreams for their child's future shatter. Parents who experienced a "roller coaster ride" to diagnosis expressed frustration at not having an explanation for their child's physical and cognitive decline during the years leading up to the diagnosis. Some individuals were misdiagnosed with other health conditions, and had been prescribed medications and therapies that provided no relief. Other families shared how they personally diagnosed their child after months of searching on the Internet, finally stumbling across pictures of other children with an MPS who looked similar to their own child. We could all learn a lesson from these parents, who act as advocates for their children and stand up for what they believe in. The road to diagnosis was not always easy, but these families never stopped looking for the answer.

Siblings of children with an MPS are also incredible; they are often willing to provide a helping hand. They talked with me about how proud they were of their family members, and shared their impressive fundraising efforts for various forms of MPS. They taught me

the importance of cherishing each day and how valuable strong relationships are. I was inspired by their strength and humbled by their courage. These families are truly warriors, fighting with honor and bearing an assault that often feels hopeless. I noticed a common theme among them: they each have an undeniable appreciation of each day, and have learned to take things as they come. They will not allow the love and strong bonds within their families to be broken.

While an MPS diagnosis invades many facets of families' lives, it does not define them. Their children's openness with themselves and others is enviable. If they are hungry, they express themselves loudly until satisfied. Bodily functions are not hidden; there is no shame. If they are unhappy with a current activity, they move onto the next one without regard. If they wish to speak with someone, they let it be known. A trip to the water park and time at the pool proved that these families like to splash water and eat French fries as much as anyone else. In the shallow end of the pool, our "noodles" became imaginary horses, and the children nominated me an honorary princess. I will not forget Justin, always welcoming me with a smile and a "Hi!" at breakfast each morning. Though their lives may be much different than anticipated, this syndrome does not impede these families from excelling.

As a second-year genetic counseling graduate student, the National MPS Society Family conference gave me a weekend I will never forget. This experience has positively changed my journey toward becoming a genetic counselor. I have learned so much from these families, and I hope to be able to one day make a difference in their lives. I will always remember their advice for all health care professionals: "Truly listen to your patients." I thank each of them for their compassion and openness. Also, I want to extend my thanks to the National MPS Society for providing this life-changing experience.
For more information about the National MPS Society, please visit http://www.mpssociety.org/ or e-mail info@mpssociety.org.

[*]*All names have been changed.*

# Genetic Counselor Publications

## Feature Article

*By Christine Colón, MS*

**Leitsalu L**, **Hercher L**, Metspalu A. Giving and withholding of information following genomic screening: Challenges identified in a study of primary care physicians in Estonia. *J Genet Counsel*. 2011. Epub.

*Liis Leitsalu, MS*

Groundbreaking technological advances in genetics and genomics offer more screening options to providers and patients, making genetic information more relevant to patient care than ever. The variety of options available yields an immense amount of complicated data – a fact that carries both positive and negative consequences. Often, it is a genetic counselor who sifts through this information to prioritize and interpret it for the patient, in order for them to make informed decisions about their health. However, physicians can also have this role, yet may not share the same level of expertise as genetic counselors within the subject of genetics and delivery of genetic test results. In this type of situation, it is unclear how prepared physicians are to explain such information to their patients.

A recent study from the Estonian Genome Center of University of Tartu (EGCUT) has begun to investigate this issue. A small research team, including **Liis Leitsalu**, **MS**, designed and distributed a survey to examine the attitudes of primary care physicians in Estonia regarding learning about and discussing genetic information with their patients. Questions focused on physicians' comfort levels discussing inheritance patterns and the genetics of complex disease, taking a family history, speaking to families diagnosed with a genetic disease, recommending testing, and informing patients of potential risks and ethical and social consequences.

Overall, the participating group of primary care providers demonstrated eagerness to apply genomic information in practice, as well as willingness to improve their knowledge base in genetics and genomics. Results of the study, however, also highlighted the need

for policies regarding return of genomic information to patients and/or research participants while safeguarding autonomy and the right not to know.

Born and raised in Estonia, Liis moved to Oslo, Norway in 2000. After completing high school in 2004, she began attending the University of Edinburgh in Scotland, where she received her Bachelor of Science (BSc) with honors. She came to the United States in 2008 and began her genetic counseling training at the Joan H. Marks Graduate Program in Human Genetics at Sarah Lawrence College, Bronxville, New York. Since her graduation in 2010, she has moved back to her home country after having been away for 10 years. Liis explains: "I had become increasingly homesick. As an additional bonus, by returning to Estonia I was the first genetic counselor in the country with a Masters of Science in Human Genetics."

Initially, Liis became involved with the EGCUT during her genetic counseling training. "At the time, when students were picking topics for their thesis projects, I thought of doing something that would involve my international background," she said. "One of the first things that came in my mind was the genome center. I contacted the director and he answered. Apparently he had been looking for someone with a genetic counseling background for a while!" After establishing a working relationship with the EGCUT, Liis completed her thesis, "Counseling for Complex Diseases – Incorporating Predictive Genetic Testing into the Health care System," in collaboration with the University of Tartu.

Upon returning home, Liis received an invitation to work in a research position at the EGCUT. Currently, she acts as the head of the Translational Genomics Working Group. She describes her responsibilities as, "leading the activities to broaden the use of genomic information generated by the genome center in health care, and raising awareness of the potential role of biobanks in public health care." In addition, she recently began her studies to earn a Ph.D. in Gene Technology at the University of Tartu. Eventually, Liis envisions herself working on the development of personalized medicine and how to responsibly incorporate genomic information into medical care.

According to Liis, the fact that Estonia is not as large as other nations has its advantages. "Having a small population size of 1.34 million[2] makes it logistically more feasible to implement innovative projects on a nationwide scale," she said. It also lends itself to a unified system of health care that is regulated nationally, provides genomic information from a single center, and offers health care education through only one university. This allows for more cutting-edge projects in the fields of health care and public health, such as creating a national database that can be used to further integrate genomic information into clinical care. It is a future goal of the EGCUT to create such a database, and allow all physicians in Estonia access to the patient information it contains. Projects of this nature may benefit those outside Estonia as well. "A redesigned health care system in Estonia can be a model for systems approach to medicine for other countries," Liis explained.

[2] POPULATION BY SEX AND AGE GROUP, 1 JANUARY

Successfully completing research and developing manuscripts acceptable for publication can be a long and arduous process, as many seasoned professionals can verify. For those with limited experience in this area, the ordeal can seem even more daunting, if not impossible. Liis offers advice to those thinking about publishing: “Find a good advisor/mentor that you enjoy working with and who would be interested in your topic and take the time to work with you. For my first publication, my supervisor, **Laura Hercher**, **MS**, **CGC**, played a crucial role. Do not get discouraged if the first journal does not accept the publication or asks for several revisions. Publishing is a long process, and it might take a while, but keep persevering!”

# AEC Update

## NSGC 31st Annual Education Conference

*By Claire N. Singletary, MS, CGC, 2012 AEC Chair and*
*Quinn Stein, MS, CGC, 2012 AEC Vice-Chair*


**Something for everyone!**

Program brochure with all of the dates and deadlines for the Annual Education Conference (AEC), which will be held **October 24-27**, **2012**, have been mailed to the NSGC membership. We look forward to expanding the spectrum of our educational offerings at the Hynes Convention Center in Boston, Massachusetts. We hope that the membership will enjoy the variety of sessions in our new convention center venue. The AEC will feature a wide variety of content in the hopes of addressing the educational and professional needs of our diverse group. This year we are proud to offer an app to help you stay informed about the AEC. Search your iTunes or Google Play store for the "2012 NSGC AEC Mobile App" to download a version; you can also follow us on Facebook and Twitter to stay connected. As an added technological advantage, AEC attendees will enjoy free wireless internet in the convention center.

**Plan ahead: Stay for the entire AEC and stay over to see Boston**

The 2012 AEC will begin on Wednesday with the "Welcome to the AEC" orientation, followed at 3:30 p.m. by the opening Plenary Session with the Janus Series and Best Abstract Awards. Concluding this kickoff will be the Welcome Reception in the Exhibitor Suite on Wednesday evening. There will be three full days of outstanding educational opportunities within the Plenary and Educational Breakout Sessions on Thursday and Friday, followed by a shorter day on Saturday that concludes by 4:00 p.m. We hope many of you will make plans to stay over Saturday evening so that you do not miss educational opportunities at the end of the conference, and are then able to enjoy Boston on Sunday.

The 2012 AEC will offer 2.48 CEUs (24.8 Contact Hours). An additional 0.50 CEUs (5 Contact Hours) will be available for those individuals who choose to register for a Pre-Conference Symposium.

**Pre-Conference Symposia**

Based on the continued positive feedback on Pre-Conference Symposia offerings, we will again have six high-level, in-depth symposia on the opening day (Wednesday, October 24). Each session will last five hours, allowing for a deeper review and discussion of a particular topic, such as The Impact of NextGen Sequencing of Cell-Free DNA on Prenatal Genetic Counseling, Epigenetics, Disorders of Sex Development, and Surgical Options for Hereditary Cancer Syndromes. The attendance at each symposium will be smaller than at the Educational Breakout Sessions, which will allow for a more interactive experience. Pre-Conference Symposia requires separate registration from the AEC and will have limited space available. Sign up early!

**Outreach in Boston**

In an effort to reach out to the community of our host city, the NSGC annually conducts an Outreach Event during the AEC. This year's event is being coordinated by **Katherine Lafferty**. Katherine and her Outreach Committee are already hard at work providing outreach to college students in the Boston area. Interested students will be invited to join us for an afternoon during the AEC to attend educational sessions and to hear a panel discussion of genetic counselors from a variety of job experiences and work settings. If you are interested in helping with this endeavor, please contact Katherine at katherine.lafferty@gmail.com

**Prepare for Boston**

The Boston area has much to offer in the Back Bay neighborhood surrounding the convention center, such as a nearby food court and shopping center, historical sites along the Freedom Trail, museums, and much more. Visit http://www.cityofboston.gov/visitors/thingstodo.asp or http://advantageboston.com/Hynes/Advantages.aspx for more information. Two nearby hotels, the Boston Marriott Copley Place and the Sheraton Boston Hotel, will have room blocks available for NSGC AEC attendees. Please book your rooms at these hotels after you have registered for the conference so that all confirmed attendees are able to enjoy rooms within the hotel room block.

**Many thanks**

We would like to thank our Conference Subcommittee members – **Mary Jarvis Ahrens**, **Julie Culver**, **Katie Dunn**, **Patricia Devers**, **Lori Erby**, **Lauren Hache**, **Katherine Lafferty, Brandie Leach**, **Anne Madeo**, **Catherine Vendola**, **Meredith Weaver**, and **Emily Windsor** – we owe them all a huge debt of gratitude. This conference is the result of the tireless efforts of this outstanding Subcommittee. Without each and every one of these individuals, this conference would not be possible. We would also like to thank

**Leigha Senter Jamieson** and **Kelly Jackson**, Education Committee Chair and Vice-Chair, and **Janet Williams**, NSGC Board of Directors liaison, for their constant guidance during the planning for the 31st AEC.

Join us as we embark upon a new path for expanded genetic counselor education by utilizing a conference center location rather than a hotel at the NSGC's 31st AEC. We look forward to seeing you in Boston.

2012 AEC Chair & Vice Chair
**Claire N. Singletary** (Claire.n.singletary@uth.tmc.edu)
**Quinn Stein** quinn.stein@sanfordhealth.org.

# Resources / Book Review

*Reviewed by Rob Finch, MS, CGC*

**Eating Pomegranates: A Memoir of Mothers, Daughters, and the *BRCA* Gene**

**Author:** Sarah Gabriel
**Publisher:** Scribner (2009)
**Pages:** 272
**Retail price:** $25.00
**ISBN-10:** 1439148198
**ISBN-13:** 978-1439148198

Eating Pomegranates: A Memoir of Mothers, Daughters and the *BRCA* Gene is Sarah Gabriel's story about struggling with memories of losing her 42-year-old mother to ovarian cancer when she was only 17 years old. Gabriel had genetic testing that showed the M18T alteration in the *BRCA1* gene. At the time, this was classified as a variant of uncertain significance, but to Gabriel the significance was 'certain,' since her grandmother also succumbed to ovarian cancer and a cousin was diagnosed with breast cancer at age 42.[1]

Feeling as if she were living on borrowed time, Gabriel, at the age of 42, felt relieved that a mammogram earlier in the year came back without any suspicion. One evening, however, while lying on the couch, flagrantly mocking the brochure on how to perform a breast self-exam, she noticed a lump. Still not taking things seriously – after all, she had her ovaries removed the previous year, which was supposed to offer her some protection against breast cancer – she debated with her husband about whether she should even bother mentioning her findings during her appointment the next day at the Cancer Genetics Clinic of the Royal Marsden Hospital in London, England. At the conclusion of that appointment, as Gabriel was shaking hands and agreeing to follow-up in a year's time, she blurted out, "Oh, by the way, it's probably nothing, but I think I may have found something…"

The rest of the book takes us through Gabriel's experience with her diagnosis of Grade III multifocal estrogen receptor-negative breast cancer; her struggles in deciding what type of surgery to choose; her nearly unbearable course of chemotherapeutic treatment with FEC (5-fluorouracil, epirubicin, and cylophosphamide); and having to pick up the pieces after the conclusion of her journey through breast cancer treatment. All this, while trying to make informed decisions on the best way to share her trials and tribulations with her young daughters, something she felt resentful that she had not been afforded by her own mother.

While at times very ominous in her descriptions, such as describing her situation as having a "wretched gene that would very likely kill her unless she amputated large

chunks of herself, that threatened to leave her children in the same parlous condition she was left in herself," it does provide a very colorful view of what it might feel like to be a *BRCA* mutation carrier, particularly to those of us who disclose genetic test results and don't live with the consequences of that disclosure every day.

I fully admit to not being an avid reader… but my experience with this book required patience and tenacity. I began my tour through this memoir with a false start. I struggled to get through the many vignettes, some of which were only tangentially relevant to Gabriel's experiences, some of which she admitted to fabricating. I began reading this book on an airplane but, feeling frustrated, I accidentally (or maybe subconsciously?) left it in the seat pocket at the end of the flight. It wasn't until I got a call from the airline that I even realized I had left the book behind.

Faced with the task of providing an honest review of this memoir, I tried again to begin my journey through the pages of Gabriel's life, this time with the anticipatory guidance that the first attempt afforded me. Once able to look past the side stories (in my opinion, equally as irrelevant as if I paused to give you a detailed history of the specific airline in the previous paragraph) I was actually able to enjoy the memoir and respect it for what it truly gave me: a very personal, very raw account of what one woman with a *BRCA* mutation encountered during her journey from being a previvor to becoming a survivor.

While the scientific literature addressing *BRCA1* and *BRCA2* mutations is endless, there have been relatively few books on the subject geared towards the lay individual. Because of this, many patients going through genetic testing and/or diagnosis of hereditary breast cancer will undoubtedly place this book on their reading lists. Genetic counselors and other healthcare providers in the field may find this book to be on the pessimistic side. However, I believe that it is an important one to read, not only to be familiar with what patients are reading, but also to get the perspective of one patient and her psychosocial journey through her experience.

[1]The M18T variant in *BRCA1* has since been reclassified to "Suspected Deleterious."