

in

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Genetic Applications in practice

bridging the GAP from bench to bedside

A Newsletter for Health Professionals Courtesy of NCHPEG

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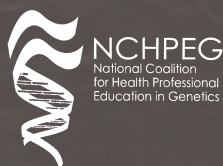
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Doctor, what do you mean my genetic test came back positive?

Contributed by: **Ira M. Lubin, PhD**

Centers for Disease Control and Prevention
Atlanta

Consider receiving a call from your cousin who is recovering from a deep vein thrombosis. He informs you that the doctor performed a genetic test and found a mutation that may or may not be associated with increased risk for thrombosis in other family members. What does that really mean? Should you be concerned? Should you be tested? Treated? Knowing more about genetic testing will help shed some light on this and other clinical conundrums.

Typically, a doctor orders a test for one of two reasons: to make a diagnosis or to screen for conditions for which the

patient is at increased risk. Colonoscopy for men and women age 50 and over is an example of a routine screening test. A culture for strep throat is an example of a diagnostic test ordered for symptomatic patients. In both cases, the doctor needs additional information about the patient (e.g., age, symptoms) to order the correct test and to interpret the result correctly. The same applies to genetic tests.

All genetic tests either are available in a variety of flavors, but all either directly or indirectly assess the base sequence that defines the structure of DNA. An

important point to appreciate is that our current technology does not make it practical to determine one's entire DNA sequence and identify all possible changes contributing to disease. For many common conditions, multiple genetic and environmental factors work in concert to cause disease. Therefore, it is important to recognize the limitations of knowledge when interpreting genetic test results. Nonetheless, such DNA sequence information can be helpful in disease management and prevention, in establishing parentage, and in determining identity (i.e., forensic analysis) and ancestry. Examples of

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The Family History Newsletter: Expanding Its Scope

During the last three years, NCHPEG has published nine on-line issues of the Genetic Family History In Practice newsletter, and they have been downloaded almost 60,000 times. In those issues, we have heard from nurses, physicians, genetic counselors, speech language pathologists, educators, and persons with genetic conditions – each with a unique personal or professional perspective on the implications of family history information.

Because family history information is central to the application of genetics, we will continue to emphasize the topic, but we have decided to expand the scope of our newsletter. Genetic testing; genetic discoveries; ethical, legal, and social issues; the economics of genetics in healthcare; and NCHPEG announcements are now in the repertoire.

The new name, Genetics Applications in Practice (GAP), new tagline, "bridging the GAP from bench to bedside," and new design reflect our expanding purview. In addition, we have included Web-friendly hotlinks directly to pages of interest. What has not changed is our commitment to collaborate with healthcare providers across disciplines and to provide interesting, useful information about genetics in practice in relatively brief, conversational articles.

Dr. Ira Lubin, a geneticist in the Laboratory Practice Evaluation and Genomics Branch at the Centers for Disease Control and Prevention, is the guest editor for this inaugural issue of GAP, and as such was instrumental in outlining a cohesive theme and in identifying expert authors. Dr. Lubin's area of interest is the improvement of laboratory practices associated with genetic testing. The Centers for Disease Control also has agreed to sponsor this issue, which will allow us to print a limited number of hard copies for distribution at professional meetings.

The staff at NCHPEG, our contributing authors, and Dr. Lubin hope that our new-and-improved newsletter will be a pleasure to read, and ultimately, that its content will inform and improve your practice. Please don't hesitate to contact the NCHPEG office at (410) 583-0600 with any questions, or to submit ideas for future articles.

Sincerely,
Erin K. Harvey, ScM, CGC

genetic testing and medical applications include:

Carrier testing

An asymptomatic adult with a mutation in one copy of the CF gene will be at higher risk of having a child affected by cystic fibrosis, depending on the carrier status of his or her partner.

Diagnostic testing

A child who inherits two CF mutations (one from each parent) will be diagnosed with cystic fibrosis. Diagnostic testing for several conditions also may be performed on fertilized ova prior to in-vitro fertilization or during pregnancy.

Newborn screening

Infants in each state in the U.S. are screened for a number of genetic conditions whose early diagnosis and treatment can usually prevent life-long disability and premature death. The number of conditions screened for varies by state - see <http://www.genetests.org>.

Pharmacogenetic testing

Pharmacogenetic testing (e.g., the cytochrome P450 gene family) is used to determine how an individual is likely to respond to a range of drugs from painkillers to chemotherapy agents. The results can help physicians prevent adverse drug reactions by helping them select the best medication and tailor the most effective dose for a specific patient.

Susceptibility / Predictive testing

Susceptibility testing helps determine the likelihood of developing a disease or complication if a specific genetic

alteration is present. Susceptibility testing is not a guarantee that disease will develop, but it is a valuable tool in risk assessment and in preventive management (e.g., about 3% of BRCA1 mutation carriers will develop breast cancer by the age of 30, but by age 70, about 85% of women with a BRCA1 mutation will have developed breast cancer— see www.genetests.org).

The best example of predictive testing is found in Huntington disease. Looking at the number of repeated DNA bases in the HD gene can diagnose individuals who are still asymptomatic and can predict the age range when symptoms are most likely to develop.

The family medical history is perhaps the oldest and most common genetic "test." Without laboratory analysis, it can provide clues about heritable diseases that may be traveling through generations in a family. As with the other types of testing described, knowledge about one individual in a family can also have implications for other family members who share genes and so may be at increased risk.

So, what type of test did your cousin, recovering from a deep vein thrombosis, have? It is most likely that he had a susceptibility test to look for the presence of a mutation known to increase the chance of thromboses. To interpret that test accurately, however, the physician must communicate additional information to the reporting laboratory, and the laboratory must effectively communicate the implications of the test results back to the physician. Miscommunication between

the lab and the provider can result in misinformation, or confusing information, being reported to the patient and his or her family. You need additional information to make your cousin's genetic test results useful to you, including: how closely related you are to your cousin, whether or not you've had any symptoms indicative of thromboses, your age, and any medications you may be taking or lifestyle habits that could exacerbate a genetic susceptibility. After considering those variables -- particularly if you do have any symptoms suggestive of thrombosis, or if you are pregnant or considering the use of oral contraceptives -- then you and your provider should discuss testing you for the same mutation found in your cousin.

The challenges to providers include: communicating effectively with the laboratory to order the proper test; incorporating genetic test results into other clinical findings and family history information, determining the relevance of the results for the patient as well as other family members, and creating a management plan, which may include further testing or clinical evaluation, counseling, treatment, and/or referral. Articles in this issue highlight some of these challenges and take-home messages in the responsible delivery of genetic services in everyday care.

When is a mutation associated with disease?

Case 1

Contributed by:

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Salt Lake City

DNA-based testing seems simple enough: test for the presence or absence of disease-associated mutations and we're in business. In practice, unfortunately, that is not always the case. It is sometimes challenging to differentiate mutations that are associated with disease from those that are not. In addition, current testing strategies can interrogate only selected regions of a DNA sequence, and many tests look only for a handful of known mutations, missing mutations in untested regions, or mutations that are in the sequence but not picked up by the test. It is particularly challenging to interpret sequence variations that are detected but have no data associated with their clinical significance. Experts estimate that single nucleotide polymorphisms (single-base sequence variations) occur approximately every 100 to 300 bases along the 3-billion-base human genome. The vast majority of these are thought to be benign, but it can be difficult to predict the clinical significance of mutations that have a major effect on protein structure. This case study illustrates how family information is useful in deciphering whether or not a previously uncharacterized mutation causes or contributes to disease.

We tested a boy with symptoms of hereditary hemorrhagic telangiectasia (HHT), an autosomal dominant disease characterized by nosebleeds, telangiectasias, and arterio-venous malformations. The original clinical information stated that there was no family history of HHT.

In sequencing the genes responsible for this disease, we found a missense variant – a change that replaces one amino acid for another – and a small deletion (in theory, a deletion is more likely to have a negative effect on the resulting protein structure and therefore function). Because neither of these mutations had been reported previously in scientific journals or databases, they were reported as "variants of uncertain clinical significance," which means we couldn't predict their role in the disease. Because we believed the deletion was more likely to be the deleterious mutation, we tested the boy's parents for the same deletion. Neither parent carried it. Therefore, we initially reported the child's deletion as a new mutation in the family and the likely cause of HHT, and we assumed that the missense variant was benign. In this scenario, any future children of this boy's parents would have a low risk of being affected, because the chance of a second new mutation in the same gene is extremely low.

In a later conversation with the clinic office, however, we learned that the father (who did have the missense variant seen in his son) and some of his relatives had some minor suggestive of HHT. The implications of that new family history information were that the missense mutation could be causing the disease, and the parents could have up to a 50% chance with each future pregnancy to have another affected child. The only way to rule that out was to evaluate more closely the "symptoms" in the father. It turned out that his symptoms were not consistent with a diagnosis of HHT, so we were able to confirm our initial conclusion that a new mutation (occurring post-fertilization and therefore not inherited from the parents) was responsible for the son's diagnosis of HHT.

In this case, the genetic test results, combined with testing of additional family members and a careful clinical evaluation of the father, were essential to correctly identify the cause of disease and to provide accurate risks to other family members.

Lessons Learned:

- Characterizing variants of unknown clinical significance as benign or deleterious requires additional genetic testing and clinical information about family members.
- Accurate clinical information helps in the interpretation of test results and in the evaluation of other family members.
- The occurrence of a new mutation in the affected child (not inherited from the parents, but occurring for the first time in that child) indicates that the parents' chance of having another affected child is very low. The affected child, however, has a 50% risk of passing the HHT-causing mutation on to each of his future offspring.
- The laboratory and the clinic must work together to interpret genetic test results correctly.

Communicating with the lab: Is the staff on board?

Case 2

Contributed by:

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For many of us, a trip to our doctor's office includes interactions with multiple staff, several of whom are responsible for aspects of our care. Similarly, when a genetic test is approved, others may be responsible for actually placing the order, interacting with the laboratory, and reviewing the results. As with any team, success depends on how well staff members work together and on their having a common understanding of the process that will result in optimal care. Using molecular genetic testing for cystic fibrosis as an example, this case study illustrates the importance of effective communication both within the clinical setting and between the clinical and laboratory settings.

The lab received a peripheral blood sample from an obstetrician's office with a request for cystic fibrosis carrier screening. The requisition provided no family history or medical history information other than that the blood was drawn from a 21-year-old Caucasian female.

When the lab results showed two copies of the delF508 mutation, consistent with a diagnosis of cystic fibrosis, lab personnel immediately called the obstetrical practice with the results. According to the nurse who answered the phone, the laboratory had made a mistake because prenatal patients in the practice are referred only for carrier testing. So, the test was reordered, the sample was reprocessed, and the entire analysis was repeated. The result was the same.

When the lab reported a positive diagnosis a second time to the obstetrician's office, a second nurse indicated that the results must have been in error because the blood was from an unaffected prenatal patient, so at most, only one mutation should have been detected. Twice rebuffed, the laboratory began a series of rigorous experiments to determine if a benign polymorphism or base-pair change had interfered with the validity of their results. Lab staff used two different sets of amplification and detection reagents, but two mutations were identified each time.

After numerous calls, the laboratory director finally spoke with the referring physician, only to learn that the patient had already been tested and was well aware of her diagnosis of cystic fibrosis, but had undergone routine (but in her case, unnecessary) carrier screening at the same time as her partner while at the clinic.

The lab director also learned that because the husband had tested negative, the couple was told (erroneously) that they had no risk of having a child with cystic fibrosis. Because neither the referring physician nor the patient understood that there was still a residual risk for the partner to carry a mutation not detected by the lab test, the couple was referred to a genetic counselor. The counselor corrected the misinformation the couple had been given and ordered further mutation analysis that showed a greatly reduced risk that the husband was a carrier.

This case highlights the critical need for the referring physician to provide pertinent clinical and family history data on the requisition for the genetic test. Had the patient's mutation status been discussed among the clinic staff, re-ordering the test would not have been necessary, nor would the laboratory have wasted valuable time and resources to test their assays.

Lessons Learned:

- The laboratory must receive pertinent clinical and family history information with each test requisition.
- It is essential that clinical and laboratory staff have easy access to each other before and after genetic testing to ensure that the tests ordered are appropriate and to ensure that the results are interpreted accurately and communicated effectively to the patient.

You've got the right result, but does the patient really 'get it'?

Case 3

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The utility of a genetic test is ultimately determined by how well a patient understands and uses the results. The following case study illustrates how patients' perceptions can negate a test's utility.

A 25-year-old pregnant woman was referred to our reproductive genetics clinic for genetic counseling. She had undergone cystic fibrosis (CF) carrier screening by her obstetrician and was subsequently identified as a carrier of the most common CF-causing mutation. Her partner also was found to be a carrier. We discussed these findings with her, and the 25 percent chance that this child (and each subsequent child with the same partner) may be affected with CF. However, she seemed remarkably calm throughout the session, considering she had just been told that her baby may be at risk for a potentially serious genetic disorder.

The genetic counselor paused and remarked on her degree of serenity. To which the patient responded, "This is no big deal. I had the exact same thing happen to me in my last pregnancy too."

The purpose of the counseling session was to discuss the test results and to help the patient understand their implications for her, her family, and her unborn child so she could make informed decisions about potential prenatal/newborn management plans or possible termination of the pregnancy. The patient's response indicated that she believed that her normal first

pregnancy was evidence that her current pregnancy was not at risk for CF. Ideally, she should have understood prior to testing that, independent of her first pregnancy, she could still be at substantial risk for having an affected child.

We cannot know for sure whether she received any pre- or post-test counseling following her previous positive test. However, it is apparent that she did not fully understand the implications. This leads to the first lesson of this story: be cognizant of what the patient thinks she or he understands about the test and use that knowledge as an entree into communicating the test's strengths and limitations.

The test for CF has characteristics different from other tests commonly ordered during the prenatal period, such as that for maternal alpha-fetoprotein (AFP). In CF testing, increased risk can be associated with outcomes from previous pregnancies, but it also depends on the carrier status of the partner. Knowing a patient's CF carrier status obviates the need for her to be re-tested with each pregnancy, but her partner's status needs to be known to make an accurate risk assessment. If both partners are carriers, the risks are the same with each conception. AFP tests on the other hand, are often repeated, regardless of the partner, because the results can have different implications for each pregnancy.

That leads to the second point of this case: in performing genetic carrier testing, we are able to comment on risk for future pregnancies and the implications for other family members (i.e., this patient's siblings also may carry a CF mutation). So, the future implications of a patient's genetic test results also should be discussed.

The major theme here is that patient counseling and education are critical to informed consent and to informed decision making, and therefore critical to patient care. Proper counseling, however, requires the provider to understand the genetics of the condition at hand and to make a time commitment to helping the patient understand several complex issues, including: possible phenotypes for a given genetic condition; the risks and limitations of the genetic test for the condition; and the implications of the results for the patient as well as for current and future family members. Because time for counseling and education is generally poorly reimbursed, each provider must consider how best to provide pre- and post-test counseling and education to his or her patients, whether that is in-house or through referrals to genetics professionals.

Lessons Learned:

- Informed decision making involves an understanding of the disorder, of the molecular test, and of the individual and family implications of a positive result.
- The patient's preconceptions about genetic testing are central to an informed discussion about the benefits and limitations of genetic tests and results. Keep that in mind when discussing the testing process.
- Appropriate counseling is important before and after the test is performed. If you wish to provide this in the confines of your practice, evaluate available resources carefully to determine whether they are adequate for the task. Otherwise, consider opportunities for referrals.
- As population-based genetic testing becomes part of routine care, new strategies may be required to ensure the avoidance of unnecessary repeat testing.

Family history in the broader context of genetic testing: From single-gene disorders to complex disease

Contributed by:

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Geneticists have long recognized that thorough family medical histories can aid in diagnosis and identify other family members at increased risk for disease. Until recently, the majority of referrals to genetics services were for diagnosis of rare single-gene disorders. Now, however, genetic risk assessment is being applied increasingly to common chronic conditions such as heart disease, stroke, diabetes, and certain cancers, which are caused by a combination of genetic and environmental factors.

This paradigm shift in the application of genetics in medicine is most apparent in the emphasis placed on family history by the Surgeon General's Family History Initiative. For many chronic conditions, family history is a strong indicator of risk for family members and provides the opportunity for disease prevention and early detection. For most common diseases, there does not appear to be an easily identifiable cause or a single gene of major affect, but for some, there are multiple genetic contributors that we can test. For example, statistics tell us that approximately 1 in 1000 persons in the US will have a venous thrombosis. Two independent mutations, mutations in the Factor V Leiden and Factor II genes are known to increase significantly the risk for thrombosis, and tests are available for both mutations. Although we require more evidence on the utility of these tests, test results may lead to the identification of others in the family who are at increased risk and who should be monitored more closely.

Another example is colorectal cancer. A positive family history, especially when combined with some telltale clinical signs (e.g., history of multiple polyps), is

a solid indication for mutation analysis in known cancer predisposition genes. Testing can aid in diagnosis and identify other family members who may benefit from prophylactic preventive measures. Likewise, approximately 5%-10% of individuals with breast cancer will have associated mutations in their BRCA1 or BRCA2 genes and are likely to have a family history. A carefully taken family history can reveal those who may benefit from testing, and if disease-associated mutations are detected, interventions may be offered to multiple family members to decrease the chance of disease onset.

Despite the clear value of family history in identifying those at increased risk, a recent review of the collection and use of family history in adult primary care found several substantive barriers to its successful application, including lack of time during the patient visit, reimbursement policies, and clinicians' limited knowledge and skills in interpreting family histories and in counseling patients about their risk (Rich et al. *Journal of Genetics in Medicine* 2004;19:273).

Several efforts are underway to address these challenges. For example, the Surgeon General's Family History Initiative and other federal and state activities have begun to raise awareness among the public about the importance of collecting family history information and sharing it with health professionals. NCHPEG also emphasizes the importance of family history data in all of its genetics education programs.

If these and other public health efforts to increase the awareness of the value of family history are successful, there may be an increasingly consumer-driven demand for providers to interpret and act on family health histories. Indeed, knowledge of increased risk for chronic diseases due to family history can directly influence the clinical

management and prevention of a disease. Prevention strategies include: 1) lifestyle changes such as diet, exercise, and smoking cessation; 2) screening at earlier ages, more frequently, and with more intensive methods than might be used for individuals at average risk; 3) instituting the use of chemoprevention agents such as aspirin; and 4) referral to a genetics specialist for a thorough risk assessment. Screening and prevention guidelines are available for many chronic disorders, and data are accumulating about the effectiveness of these and other strategies in high-risk individuals.

As more genetic variants are identified, we will be able to refine our capabilities in diagnostic and predictive testing. However, as is the case with BRCA1/2 testing, most predictive genetic tests will not be appropriate for the population at large. Subgroups of the population who are at increased risk – often due to family history – will be the recipients of these tests.

A number of studies have found that although primary care providers acknowledge the increasing role of genetic services in primary care (i.e., family history taking, risk assessment, informed decision making, genetic testing, and test interpretation), their level of confidence in delivering those services is low. Educational efforts to raise health professionals' knowledge and skills will help, but equally important are computerized decision-support systems that will allow faster and more accurate methods to assess disease risk, offer genetic tests, and implement prevention guidelines (see Emery et al. *British Journal of Medicine* 2000; 321:28)

For additional information about the US Surgeon General's Family History Initiative, see <http://www.hhs.gov/familyhistory/>.

Tying It All Together:

Challenges and opportunities for genetic testing in primary care

Contributed by:

Ira M. Lubin, PhD

Centers for Disease Control and Prevention
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Genetics is an evolving field that is an important component of diagnosis, risk assessment, and disease management. These applications to medicine are not new, but the integration of DNA-based genetic testing into practice presents benefits and challenges.

The first question to ask of any medical test or intervention is whether its use benefits the patient. For DNA-based genetic tests, the benefit derives from our understanding of the correlation between known sequence variations and disease. Ideally, well-defined, population-based studies provide those correlations, but there are few such studies compared to the number of genetic tests available or in development. A pilot program called EGAPP, developed under the auspices of the Centers for Disease Control and Prevention, is attempting to narrow the gap between discovery and research (see <http://www.cdc.gov/genomics/gTesting.htm>)

Another challenge lies in the delivery of genetics services in primary care settings. Each of the case studies in this issue addresses the importance of understanding the process of genetic testing, and of open communication within and between the laboratory and clinical settings. Ineffective communication can result in compromised patient care and can add unnecessary time and costs to clinical services.

A meeting held in 2003, hosted by Mt. Sinai School of Medicine and the Centers for Disease Control and Prevention, provided a national forum to begin discussion of these service-delivery issues. Attendees concluded that despite a growing number of excellent guidelines, communication suffers from lack of a common understanding of terminology and basic concepts (see <http://www.phppo.cdc.gov/dls/genetics/comm052003.aspx>).

A workshop held in November 2005, co-hosted by the Wadsworth Center, New York Department of Health, the Association for Molecular Pathology Clinical Practice Committee, and the Centers for Disease Control and Prevention, followed up on the Mt. Sinai meeting to chart a course for improving communication and basic knowledge of genetic testing. CDC plans to convene a series of workgroups to address barriers to communication and to consider common terminology for ordering, reporting, and interpreting genetic tests, all in the hope of more effective use of genetic tests in primary care. In considering these issues, it will be critical to consider the role of electronic information systems and the electronic medical record.

Accurate collection and reporting of information take on added significance given that some genetic tests are moving to non-specialty, high-volume laboratory and clinical settings, as is the case with DNA-based testing for cystic fibrosis. These evolving practices are cost effective and save time for the

patient and the healthcare system, but the challenge resides in ensuring that appropriate resources are available, accessible, and used to make sense of the test result for the provider and the patient.

A helpful starting point is to review the relevant processes in our own settings and to identifying credible genetics resources. Several excellent web-based resources provide authoritative information about genetic testing. GeneTests provides information about the use of genetic testing in diagnosis, management, and genetic counseling (see <http://www.genetests.org>), and the American College of Medical Genetics has developed a number of policy statements that outline the proper use of genetic tests in laboratory and clinical practice (see <http://www.acmg.net>). A number of other professional organizations such as the American Society of Clinical Oncology (<http://www.asco.org>) have addressed various issues pertinent to their specialty. Ultimately, our efforts to increase knowledge, to bring genetic technologies from the bench to the bedside, to reduce costs, and to ensure the accurate interpretation of results will help us serve our patients and their families more effectively.

Race, genetics, and healthcare: What we know and what it means for your practice

On April 20, 2006, NCHPEG hosted its first live television broadcast of a CME program in genetics. The 90-minute program originated from Dallas, reaching approximately 700 hospitals nationwide through the VHA satellite network, run by the Voluntary Hospitals of America.

The broadcast, supported by the Robert Wood Johnson and Josiah Macy, Jr. Foundations, is intended to teach primary-care providers (PCPs), and anyone who visits the NCHPEG website, about race, genetics, and health care, in particular:

- whether race has validity as a biological entity,
- the differences between biological and cultural conceptions of race,
- the relationship between genetics and race in particular with respect to health care, and
- the meaning and utility of race in the clinical setting and in biomedical research.

A live studio panel responded to simulated, pre-recorded interactions between patients and providers who addressed the following clinical issues:

Case 1: Metabolic syndrome: Understanding risk on an individual level

Case 2: Who is at high risk? Screening for prostate cancer in a 40-year-old African American

Case 3: G6PD deficiency and the history of human biological variation

Case 4: BiDil – pharmaceuticals targeted to specific populations

Case 5: Tay-Sachs disease and hemochromatosis

Case 6: Race, genetics, and health disparities

The panelists were:

Gary Gibbons, MD

Morehouse College of Medicine;

Howard Levy, MD, PhD

Johns Hopkins School of Medicine; and

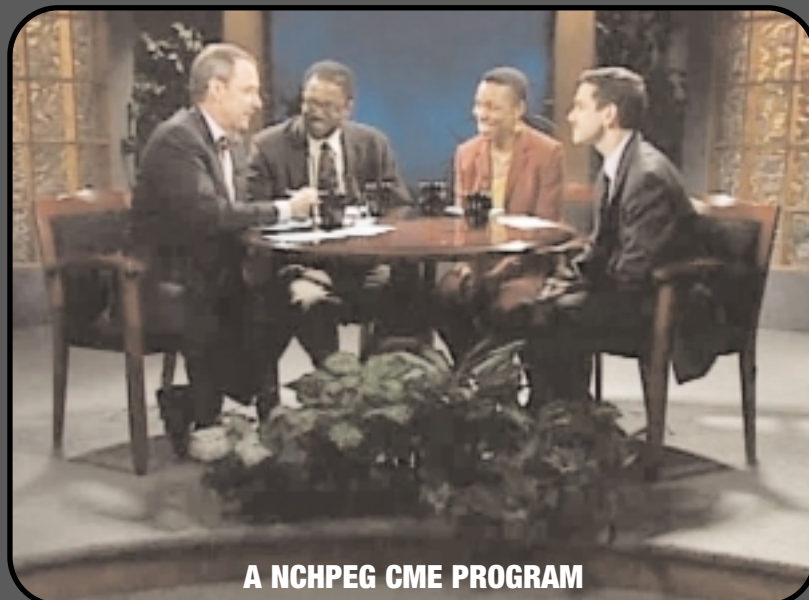
Charmaine Royal, PhD

National Human Genome Program,

Howard University.

Francis Collins, chairman of NCHPEG's board of directors and director of the National Human Genome Research Institute, provided introductory comments and, in Case 3, an overview of the history and nature of human genetic variation.

The American College of Medical Genetics, a NCHPEG member organization, provided CME credits for the broadcast and will provide credits for those who view the program on-line at www.nchpeg.org.



A NCHPEG CME PROGRAM

NCHPEG's Newest Staff Member



NCHPEG is pleased to welcome Eleanor K. Reed as its newest staff member. Kate, as she is known, joined the staff on March 13 to serve as project director for our new CDC-funded project to develop a resource network for single-gene disorders (the Genetic Alliance is the lead agency).

Kate will spend 40 percent of her time doing genetic counseling in pediatrics and cancer genetics at the University of Maryland, Baltimore, where she will be working with Mimi Blitzer, a NCHPEG board member. Kate holds an MPH in public health genetics from the University of Washington and recently graduated from the genetic counseling program at Johns Hopkins and the National Human Genome Research Institute.

Cynthia Prows wins 2006 Scotti Award



Attendees at NCHPEG's 9th annual meeting saw Cynthia A. Prows, MSN, RN, receive the third annual Michael J.

Scotti, Jr. Award for contributions to genetics education for health professionals. Ms. Prows, a clinical nurse specialist and educator at the Cincinnati Children's Hospital, was honored for "her substantive and enduring leadership in genetics education for nurses, and for her many contributions to NCHPEG's programs and activities."

Ms. Prows has been active in developing, implementing, and evaluating genetics education programs since 1992, when she created a genetics program for nurses at Cincinnati Children's Hospital. In 1996, she received a National Human Genome Research Institute (NHGRI)/Ethical Legal Social Issues (ELSI) grant, later supplemented by the Health Resources and Services Administration, to determine the effectiveness of genetics education programs targeting nursing faculty within the United States. A competitive grant renewal extended the program audience to advanced practice nurses. From that program of education research, seven onsite genetics summer institutes, and seven 18-week web-based genetics institutes (WBGI) provided genetics instruction to 314 nursing faculty and advanced practice nurses from 230 different schools of nursing and

healthcare organizations within 45 different states, Puerto Rico, Japan, South Korea, Nigeria, and the Virgin Islands.

Follow up of nursing faculty participants demonstrated that the institutes provided the necessary instruction and resources to enable them to increase genetics content in their curricula. The WBGI's success can also be measured by its sustainability without grant funding.

In 2003, Ms. Prows received an NHGRI/ELSI grant to develop web-based, independent self-paced modules for nurses, particularly those in medically underserved communities. Four modules are freely accessible at <http://gepn.cchmc.org> and two additional modules were pilot tested in spring 2006.

NCHPEG initiated the Michael J. Scotti, Jr. award in 2004 to honor the contributions Michael J. Scotti, Jr., MD, a former vice president at the American Medical Association and a founding member of NCHPEG's Board of Directors. Previous winners include Dr. Scotti and Jean Jenkins, PhD, RN, formerly at the National Cancer Institute and now on staff at the National Human Genome Research Institute.

Case studies in genetics for physician assistants: NCHPEG's 2007 targeted audience for genetics education

Physician assistant (PA) students, faculty, and professionals constitute the target audience for NCHPEG's 2007 online, genetics education program. The content on the site will be case driven, using clinical examples relevant to PA practice. In addition to family history and basic genetics tutorials and quizzes, case studies will focus on clinical problem solving for patients who present with developmental delay and mental retardation, cancer, thrombophilia, and high cholesterol. A "teaching tools" section of the site will house PowerPoint slides, articles, and online references to help faculty incorporate genetics content into existing curricula.

The PA genetics education site is a collaborative effort between NCHPEG, the American Academy of Physician Assistants, and PA faculty at the University of Utah. PA practitioners, faculty, and students from across the country have also agreed to serve as advisors and pilot testers. The site is expected to go live in the summer of 2008, and will be available at the NCHPEG website (www.nchpeg.org) free of charge.

The rationale for the site is provided by several studies that suggest that the incorporation of genetics into PA programs has lagged behind the application of genetics in clinical practice. There continues to be concern among educators that providers are not adequately prepared for the expanded role that genetic information demands. Recognizing this shortfall, the Accreditation Review Commission on Education for Physician

Assistants (ARC-PA) has created a new accreditation standard, effective September 2006, that mandates teaching "the genetic and molecular mechanisms of health and disease" in all PA programs.

Typically, PAs have close relationships with their patients and know their medical histories and attitudes towards health care. Coupled with this critical foundation, genetic skills that are increasingly important for PAs include the following:

- **identification of individuals - by eliciting an informative genetic family history - who may benefit from genetic services;**
- **recognition of physical findings and other red flags indicating a genetic susceptibility or a genetic diagnosis;**
- **provision of basic genetic information and counseling to facilitate informed decision-making;**
- **knowledge of available genetics references and access to genetics colleagues; and**
- **collaboration with genetics specialists in the management of patients with complex and rare disorders that have a genetic basis.**

Ultimately, the 2007 targeted genetics education website for PAs aims to help bridge the gap between genetics knowledge and its application to every-day practice. NCHPEG has developed similar online resources for family physicians, dentists and dental hygienists, and speech-language pathologists and audiologists. Funding for NCHPEG's annual genetics education programs is provided by the Health Resources and Services Administration, the National Human Genome Research Institute, and the Office of Rare Diseases. For more information, contact the NCHPEG office at 410-583-0600.



10TH ANNUAL MEETING
NCHPEG/GROW

PHARMACOGENOMICS

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