ACMG
American College of Medical Genetics and Genomics

February 1, 2013

Recent Activities of The American College of Medical Genetics and Genomics

The American College of Medical Genetic and Genomics (ACMG) is the professional home to nearly 1,600 board certified clinical and laboratory genetics professionals and is the only nationally recognized medical organization dedicated to improving health through the practice of medical genetics and genomics. The College's mission includes the following major goals: 1) to define and promote excellence in the practice of medical genetics and genomics and to facilitate the integration of new research discoveries into medical practice; 2) to provide medical genetics and genomics education to fellow professionals, other healthcare providers, and the public; 3) to improve access to medical genetics and genomics services and to promote their integration into all of medicine; and 4) to serve as advocates for providers of medical genetics and genomics services and their patients. This report summarizes key activities of the ACMG between September 2012 and January 2013.

ACMG Announces New President-Elect and Directors

ACMG recently held its biennial election, with the following individuals beginning their six-year terms at the end of the 2013 Annual Clinical Genetics Meeting in March. Gerald Feldman, MD, PhD, FACMG is the President-Elect. The other newly elected Directors are Joel Charrow, MD, FACMG (Biochemical Genetics); Kristin Monaghan, PhD, FACMG (Molecular Genetics); Lorraine Potocki, MD, FACMG; Robert Steiner, MD, FACMG and Maren T. Scheuner, MD, MPH, FACMG (Clinical Genetics).

ACMG Releases New Position Statement on the Public Disclosure of Clinically Relevant Genome Variants

In November the ACMG Board of Directors released a new Position Statement on the *Public Disclosure of Clinically Relevant Genome Variants* to address problems arising from gene patent monopolies that have allowed for the development of proprietary databases of the clinical meaning of the variants in particular genes that become trade secrets.

In a related press release, ACMG's executive director, Michael S. Watson, PhD, FACMG, explained, "The next phase of the human genome project, which is to annotate the human genome sequence with the clinical and biological meaning of the sequences and variants, will require capturing information from a very large number of people from diverse populations across the US and internationally. Information that informs us about the meaning of genome sequences should be in the public domain where it can be used for the benefit of all."

The complete Position Statement, which is appended to this report, states, "The American College of Medical Genetics and Genomics (ACMG) believes that gene testing and the clinical data on which genetic data are interpreted must remain widely accessible and affordable, and that the development and improvement of safe and effective genetic tests should not be hindered. Monopolistic practices that limit a given genetic test to a single laboratory are inconsistent with ACMG's goals of broadly accessible and affordable genetic tests.

ACMG Petitions Supreme Court on Gene Patent Case: Schedule of Follow-up Events Now Available

While the legal proceedings in the Myriad Genetics gene patent case have been a long and winding road—albeit a closely followed one in the human genetics and genomics communities—ACMG (and other plaintiffs) successfully petitioned the United States Supreme Court for a *writ of certiorari*. What this means is that the plaintiffs asked the Supreme Court to review (and, ultimately, to reverse) the decision of the US Court of Appeals for the Federal Circuit, which most recently upheld Myriad's patents on the BRCA1 and BRCA2 gene sequences while striking down patents on the methods to compare those genes. The Supreme Court has agreed to take on the case, and a timetable has been established for the receipt of various court documents that need to be produced by both sides. It is anticipated that oral arguments will be heard in mid-April, with a final decision by June 2013.

New ACMG Video Encourages Medical Students to Consider a Residency/Combined Residency in Medical Genetics

As part ACMG's ongoing commitment to attract more college and medical students to pursue careers in medical genetics and genomics, the College has produced a video aimed at encouraging medical students to pursue a residency in medical genetics. The video, *Medical Genetics is the Future of Medicine*, is available on TheACMGChannel on YouTube at www.youtube.com/theacmgchannel and has been distributed to medical schools and Medical Genetics Training Programs around the country. The video includes interviews with current and recently graduated medical genetics trainees and has real-life footage of patient care, team meetings, and other activities in several settings. For much of the video, medical genetics trainees 'talk' directly to the medical student viewers. Individuals may request a copy of the five-and-one-half minute video on flash drive by emailing cpowell@acmg.net.

Other outreach efforts to grow the medical geneticist pipeline include: 1) the ACMG Summer Genetics Scholars Program, whereby medical students can apply for a paid summer genetics position under the mentorship of a board-certified geneticist; 2) a growing number of ACMG Medical Student Interest Groups: 3) free student sessions at the ACMG Annual Meeting; and 4) free student memberships, with discounts for other trainee categories.

The ACMG Foundation for Genetic and Genomic Medicine provided funding for the Medical Genetics Residency Video.

ACMG Takes to Social Media for 9th Annual Family Health History Month

In 2004, then U.S. Surgeon General, Vice Admiral Richard Carmona, MD, MPH, FACS, declared Thanksgiving as National Family Health History Day—an event that came about through strategic educational efforts by NHGRI and the major human/medical genetics organizations. In the ensuing years, this public health initiative has grown such that November is now celebrated as National Family Health History Month. This past fall, ACMG invited the public to celebrate Family Health History Month by sharing a Family Health History Fact or Resource each week on social media. ACMG reminded fans and followers that talking with their health care provider about their family health history can help them and their loved ones stay healthy. ACMG also shared resources such as the Surgeon General's My Family Health Portrait, and encouraged the public to print their family health history to share with family or healthcare workers and to save this information to update over time.

ACMG Grants and Contracts

Newborn Screening Translational Research Network (NBSTRN) Coordinating Center

The Newborn Screening Translational Research Network (NBSTRN), now in its fifth year, is funded by a contract from the *Eunice Kennedy Shriver* National Institute of Child Health and Human Development (NIH) ACMG. The mission of the NBSTRN is to improve the health outcomes of newborns with genetic or congenital disorders by means of an infrastructure that allows investigators access to robust resources for newborn screening research. Information about all NBSTRN programs can be found at www.nbstrn.org. Several updates follow:

Virtual Repository of Dried Blood Spots (VRDBS)

Dried blood spots (DBS), collected within the first days of life from infants who have rare genetic diseases, are difficult to acquire for investigators working to improve newborn screening; yet, DBS are at the core of translational research. The NBSTRN's VRDBS provides an efficient way for investigators to find and share DBS. The Repository consolidates specimen information and offers tools for navigating state policies for requesting access to DBS.

NBSTRN has partnered with 5AM Solutions, Inc. to develop the Virtual Repository. The VRDBS is a secure web□based tool that presents information from participating states and provides a centralized, de□identified view of DBS, allowing researchers to locate and request specimens. For public health departments, the virtual repository

provides the ability to review and manage specimen requests, track shipments, and obtain feedback on specimen storage condition.

Currently, 4 are states participating in the VRDBS (CA, IA, MI, and NY), representing over 2 million specimens. Public training webinars for the VRDBS are held monthly and are open to all interested parties.

Development of the Longitudinal Pediatric Data Resource (LPDR)

The Center for Biomedical Informatics (CBMi) at the Children's Hospital of Philadelphia (CHOP) and the NBSTRN have partnered to present a solution for collecting, managing, and delivering long-term care data for individuals identified with conditions through newborn screening. Effective capture of long-term clinical data for these individuals represents an exciting opportunity to establish a longitudinal data collective, which will produce a transformative resource for biomedical research. Delivery of a mechanism to collect, manage, and disseminate long-term clinical data in the context of newborn screening information and biomaterials is a key outcome for the remaining years of the initial NBSTRN project period.

Dr. Peter White and his team at CHOP are spearheading this work, through a subcontract administered by the NBSTRN. The LPDR and data capture tool are being developed using Research Electronic Data Capture (REDCap). Since its introduction into the academic research community in 2007, the REDCap data management system has quickly emerged as an academic standard for investigator-driven research data management, with over 175 institutions and nearly 5,000 research studies now using the tool.

Additional NBSTRN Resources

- **State Profiles:** In cooperation with the Association for Public Health Laboratories (APHL) and the National Newborn Screening and Genetics Resource Center (NNSGRC), the NBSTRN has collected specific information about each of the states' newborn screening program and has compiled it in one, easy to access, resource on its website. The State Profiles include: 1) newborn screening program contacts; 2) residual dried blood spot retention, storage and use data; 3) state-specific IRB protocols and 4) other information pertinent to newborn screening related research. Specific information for those states participating in the VRDBS includes: 1) years of data included in the VRDBS; 2) frequency of upload of data; 3) state-specific IRB and MTA information; 4) fees for specimen recovery, processing and shipping; and 5) additional review or steps needed outside of the VRDBS.

- **Investigator FAQs:** Innovative translational must be conducted in a manner that respects and protects the rights of children and their families and includes IRB oversight. The FAQs Resource is intended to provide potential newborn screening researchers and IRB members with guidance regarding some of the ethical and regulatory issues that may arise when designing and conducting translational research to improve newborn screening.

National Coordinating Center for Regional Genetics and Newborn Screening Service Collaboratives (NCC)

Since 2004, the ACMG has received funding through a cooperative agreement with the Maternal and Child Health Bureau, Health Resources and Services Administration (DHHS) to serve as the National Coordinating Center for the seven Health Resources and Services Administration (HRSA) Genetics Collaboratives. The mission of the seven Regional Genetics and Newborn Screening Service Collaboratives (RCs) and their National Coordinating Center (NCC) is to strengthen and support the genetics and newborn screening capacity of the states and to improve the availability, accessibility, and quality of genetic services and resources for individuals with or at risk for genetic conditions, and their families.

NCC Invited to Assist with Implementation of the Affordable Care Act (ACA)

Maintaining its current focus on quality improvement and program evaluation, the NCC responded to a request from HRSA with a number of proposed activities to be carried out in the next 3, 6 and 12 months, nationally, regionally and locally, to ensure that the expertise of genetic service providers and the needs and perspectives of individuals and families living with heritable conditions that require on-going health services and public health support will be integrated into implementation of the Patient Protection and Affordable Care Act (ACA). The NCC and the RCs have a unique focus on genetic services and resources, with the RCs strategically positioned as information "nodes" that are also connected with the service delivery sites and the state public health infrastructure across the US. The NCC, on the other hand, brings to this process the ability to leverage established relationships with national agencies and organizations. The following goals, adapted from principles delineated in the ACA, will guide the work and facilitative role of the NCC/RC system and its partners in this important endeavor:

- Essential health benefit (EHB) packages should address medical/health needs of individuals with heritable conditions.
- Changes in both public and private insurance programs should provide adequate reimbursement for needed health services for individuals with or, at risk for, heritable conditions.
- Patient care navigators, who are part of the health insurance exchanges, should have an understanding of the unique needs of the genetics population in order to help select the best plan(s) for them.
- Medical homes and care coordination, that comprise a core part of the health delivery system reform, must meet the needs of individuals with heritable disorders, including consideration of the roles of specialty centers and genetic specialists.
- The healthcare workforce, especially primary care providers, should be educated about genetics and heritable disorders.
- Accountable care organizations (ACOs) should have quality indicators that target the medical/health needs of individuals with heritable disorders and address those needs through quality improvement activities within their systems of care.

Genetics in Medicine Updates

The following Laboratory Guideline was published in *Genetics in Medicine* between September 2012 and January 2013:

> Lyon E, Foster JG, Palomaki GE, Pratt VM, Reynolds K, Sábato MF, Scott SA and Vitazka P; A working group of the Molecular Genetics Subcommittee on behalf of the American College of Medical Genetics and Genomics (ACMG) Laboratory Quality Assurance Committee. **Laboratory testing of *CYP2D6* alleles in relation to tamoxifen therapy.** *Genet Med* 14(12):990-1000 (December 2012)

2013 ACMG Annual Meeting Updates

The 2013 ACMG Annual Clinical Genetics Meeting will be held March 19-23, 2013 in Phoenix, AZ, in conjunction with the 44th Annual March of Dimes Clinical Genetics Conference on Skeletal Dysplasias and Connective Tissue Disorders. Up-to-the-minute information can be found on the ACMG Meeting website at www.acmgmeeting.net. Several highlights follow:

March of Dimes Session to Honor the late Dr. David L. Rimoin

The 44th Annual March of Dimes Clinical Genetics Conference will be presented in honor of ACMG's Founding President and Genetics Pioneer, the late David L. Rimoin, MD, PhD, FACMG, whose life-long interest in understanding the genetics and treatment of skeletal dysplasias and connective tissue disorders earned him international accolades. Brendan Lee, MD, PhD, of Baylor College of Medicine, will present the March of Dimes Pruzansky Lecture, held during this session.

Inaugural R. Rodney Howell Symposium in Public Genetics and Genomics to Examine Fifty Years of NBS

When R. Rodney Howell, MD, FACMG retired from his many Washington duties, including his roles as the first Chair of the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children (DHSS), Special Assistant to the Director of NICHD, and ACMG's long-time liaison the NACHGR, the ACMG Foundation for Genetic and Genomic Medicine created a symposium on public health genetics and genomics, to occur at the ACMG Annual Clinical Genetics Meeting, in honor of Dr. Howell's lifetime contributions to children's health. The inaugural symposium, *Fifty Years of Newborn Screening: Impact on Public Health Genetics and Genomics,* will trace the history of newborn screening (NBS) from its early social and scientific challenges to being heralded as one of the most successful children's public health program of the last decade. The symposium will end with future perspectives, emphasizing on the rich collaborative research opportunities for the genetics and genomics communities. Speakers include V. Reid Sutton, MD, PhD, FACMG (Baylor College of Medicine/Texas Children's Hospital), Michele Caggana, ScD, FACMG (New York State Department of Health/Wadsworth Center), Susan Berry, MD, FACMG (University of Minnesota) and Leslie Biesecker, MD, FACMG (NHGRI/NIH).

Two Short Courses to Precede Annual Meeting

As the growth of genomic technologies continues to accelerate the pace at which medical genetics permeates all of healthcare, the Short Courses held prior to the ACMG Annual Meeting have become popular forums for medical genetics professionals to maintain cutting-edge skills and expand their depth of knowledge in specialized areas, all

while learning from the leaders in the field. Darrel Waggoner, MD, FACMG (University of Chicago) will direct the short course, *Next Generation Sequencing From a Clinical Perspective: What are You Getting and What Does it Mean?* The second short course, *Arrays, NIPT and Expanded Carrier Screening: Advances and Controversies in Prenatal Genetics*, will be directed by Anthony R. Gregg, MD, FACMG (University of Florida) and Brian Shaffer, MD (Oregon Health and Science University).

The Highlights Plenary Looks to the Future

The popular Highlights Plenary, *Progress in Gene and Cell Therapy Clinical Trials for Genetic Diseases* will feature two talks focusing on the work of colleagues involved in advanced clinical trials for genetic diseases—Cell Therapy Strategies for Adrenoleukodystrophy and AAV Strategies for Hemophilia B. This cutting-edge session represents a collaboration between the American Society of Gene and Cell Therapy (ASGCT) and the ACMG.

Second Annual Open Forum Scheduled

Based on the popularity of the 2012 Open Forum, *Reporting of Incidental Findings in Whole Genome/Exome Clinical Testing*, a similarly structured session will occur at the 2013 Annual Meeting. The topic will be *Whole Genome Analysis Consent and Prenatal Findings: An Open Forum on Return of Genetics Results*. The session will include an interactive discussion with attendees able to provide feedback via an Audience Response System.

Opportunities for Trainees

The ACMG Annual Meeting will once again feature many learning and networking opportunities for Residents, Post-Doctoral and Clinical Fellows, and Students. These include: 1) an exclusive welcome reception; 2) a workshop on manuscript publication; 3) a trainee/mentor luncheon; and 4) an informational session on careers in medical genetics.

Annual Meeting Mobile App Introduced

In response to the overwhelming number of requests, the ACMG Meetings Team is introducing a new mobile app to assist attendees in better navigating their meeting experience, including options to schedule education sessions, committee meetings, special events and exhibit hall visits, as well as to carry out social networking.

2013 ACMG Genetics and Genomics Review Course Now Offers Webinar Option

ACMG has scheduled its next Genetics and Genomics Review Course for June 20-23, 2013. The Course will be held at USF Health's Center for Advanced Medical Learning and Simulation (CAMLS), a state-of-the-art, three-story facility for health professional education and training located in downtown Tampa, FL. The Course will be presented live in Tampa and simultaneously broadcast via webinar for those unable to attend in person. The webinar will be archived for later purchase to allow professionals to take advantage of this learning opportunity for up to two years. Hotel information, online registration and the program schedule will be available on the ACMG Website in February.

Further information about all ACMG activities and a full listing of our press releases and clinical genetics laboratory and practice guidelines can be found on our website at www.acmg.net. The ACMG website now houses an Online Learning Center, as well. ACMG uses Facebook, LinkedIn, YouTube, and Twitter to augment its educational and advocacy missions, provide news and resources related to medical genetics, and improve communication with and among its members and stakeholders.

Submitted by Michael S. Watson, PhD, FACMG
ACMG Liaison to the National Advisory Council for the National Human Genome Research Institute, NIH

ACMG
American College of Medical Genetics and Genomics

Position Statement on the Public Disclosure of Clinically Relevant Genome Variants

Genetic and genomic tests are playing an increasing role in medical practice, enabling prevention, diagnosis, and management of both rare and common disorders and the analysis of genetic changes associated with cancer. Testing is done in commercial, academic, and hospital laboratories throughout the world. The clinical interpretation of rare variants requires access to data on clinical annotation, but some laboratories have maintained private databases that are not publicly available. In some cases, these are deliberately withheld from public access for business reasons; in other cases, laboratories that have focused on rare disorders have not taken the steps to make data publicly available. The American College of Medical Genetics and Genomics (ACMG) believes that gene testing and the clinical data on which genetic data are interpreted must remain widely accessible and affordable, and that the development and improvement of safe and effective genetic tests should not be hindered. Monopolistic practices that limit a given genetic test to a single laboratory are inconsistent with ACMG's goals of broadly accessible and affordable genetic tests.

These practices:

- Limit the accessibility of competitively priced genetic testing services and hinder test-specific development of national programs for quality assurance.
- Limit the number of knowledgeable individuals who can assist physicians, laboratory geneticists and counselors in the diagnosis, management and care of at-risk patients.
- Place the laboratory that possesses the data in the position of practicing medicine, since only they have the ability to use data to inform medical decision-making.
- Inhibit the training of the next generation of medical and laboratory geneticists, physicians, and scientists.

Therefore, it is the ACMG's position that:

- Clinical data underlying genome annotation that informs the clinical interpretation of molecular variants are fundamental to the practice of genetic and genomic medicine.
- Withholding/restricting the use of such information impedes its integration into medicine.
- Interpretations of genomic variants should be informed by the best clinical information available to clinicians and scientists.
- Payers, regulators and providers should work to bring the clinical data into publicly available resources.

Approved by the ACMG Board of Directors on November 6, 2012

7220 Wisconsin Avenue, Suite 300, Bethesda, MD 20814 Phone 301-718-9603 Fax 301-718-9604 **www.acmg.net**