ACMG
American College of Medical Genetics and Genomics

August 30, 2012

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 May and August 2012.

ACMG Grants and Contracts

Newborn Screening Translational Research Network (NBSTRN) Coordinating Center

- Part of a program authorized and appropriated under the Newborn Screening Saves Lives Act; operates under a contract from NICHD/NIH.
- Includes a Standing Committee and working groups in the areas of Bioethics and Legal Issues, Newborn Screening Laboratories and Programs, Clinical Centers, and IT and Informatics.
- Opens a multistate virtual repository for dried blood spots held by State Newborn Screening Programs in September; the only large biospecimen repository of the general population in the United States. Over two million bloods spots will be available to investigators.
- Provides the tools for longitudinal point-of-care data capture related to diagnosis, evaluation and management with deposition into a centralized data warehouse.
- Currently involves grantees representing 25 institutions in 12 states that are studying more than 50 conditions that are identified by newborn screening programs or other conditions that are candidates for newborn screening. Capacity to integrate exome sequencing research with the clinical database is in development.

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

- Part of a program funded by the Maternal and Child Health Bureau of the Health Resources and Services Administration (HRSA).
- Facilitates interactions among the seven regional collaboratives, public health and primary care providers, and specialists (including medical geneticists).
- Develops clinical decision support tools for primary care providers preparing to meet with a patient/family that has screened positively in newborn screening.
- Coordinates regional activities in telemedicine, patient transition from pediatric to adult care, roles of primary care medical homes in collaborative management of patients, relationships with other HRSA and federal resources.

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ACMG Receives Funding to Continue as the National Coordination Center for the HRSA Regional Genetics Collaboratives: Focus to be on Quality Improvement and Evaluation

Since 2004, ACMG has served as the National Coordinating Center (NCC) for the seven Health Resources and Services Administration (HRSA) Genetics Collaboratives (RCs) through a cooperative agreement with HRSA. ACMG staff recently responded to a funding announcement to continue to serve as the NCC for another three-year period. In June, we learned that funding was renewed through another cooperative agreement with HRSA. Genetic Alliance will partner with ACMG to coordinate the National Genetics Education and Consumer Network (NGECN), a key component to the NCC. The central goal of the NCC has been to bring quality genetic and newborn screening services to local communities and to build bridges between public health, primary care/Medical Home, geneticists and other specialists, and families and consumers. With continued funding the NCC will also focus on the following goals:

1. Development of a comprehensive quantitative and qualitative evaluation program assessing project outcomes and measuring the health impact of the NCC/RC system as a whole;
2. Engaging RC project/workgroup leads in NCC workgroups to ensure consensus on national definitions (e.g., Medical Home, Telegenetics), develop evaluation measures, and identify data sources that inform the evaluation workgroup and permit system-wide evaluation and quality improvement;
3. Development of electronic medical record (EMR) specifications for clinical genetics patient care with attention to the integration of clinical decision support tools such as the Newborn Screening ACT Sheets;
4. Supporting further development of the existing collaborative work on data sets for follow-up of newborn screening and development of data dictionaries and common ontology for use with EMRs and registries;
5. Providing assistance to the RCs to facilitate communication strategies, partnerships, and collaborations on a local level through tri-annual publication of the *NCC Collaborator* newsletter, monthly inter-RC calls, quarterly educational webinars on topics of interest to NCC and RC collaborators, joining efforts with other national centers and federally funded-projects related to the work of the RCs, and exhibiting NCC and RC products at national meetings to increase awareness; and
6. Development of the NGECN to assist and engage consumers and consumer groups, bridge between consumers and services provided by HRSA-funded programs and centers to ensure access to genetics information, services, and resources, enhance and improve state-level linkages with a broad range of constituencies to promote genetics education, disseminate existing resources and services information, and retain an advisory body of stakeholders that ensures that projects meet these requirements.

NBSTRN Announces Two Unique Webinar Series

The Newborn Screening Translational Research Network (NBSTRN) is now offering two informative webinars. The first is the *Virtual Repository of Dried Blood Spots* (VRDBS) monthly training webinars series. These one-hour webinars are intended to provide a general overview of the NBSTRN, goals of the VRDBS, and a quick review on using the system. Each month's presentation is catered to either states or investigators. Attendees in previous webinars range from academic institutions to state newborn screening programs and federal agencies. This has proven to be a quick and efficient way to demonstrate the functionality and capabilities of the VRDBS. Please join us at any of the scheduled VRDBS Training Webinars or contact NBSTRN for a private demonstration.

The second webinar series, *Providing Newborn Screening Specimens for Research: Legal Issues Faced by State Health Departments*, is a partnership with the Network for Public Health Law, the National Newborn Screening and Genetics Resource Center, and the Association for Public Health Laboratories. This series is intended for states that store and provide residual dried blood spots (DBS) for secondary uses or are considering storage and secondary uses. These webinars cover legal and related ethical issues concerning secondary uses of DBS and associated data. The webinars cater to public health attorneys, newborn screening programs and laboratories, institutional review boards, HIPAA privacy boards, privacy officers, researchers, and others who have interest in legal and ethical issues related to secondary uses of residual DBS. Information on previous as well as upcoming sessions can be found at NBSTRN.org.

New ACT Sheets Posted to ACMG Website

Twelve new ACT Sheets, encompassing Transition to Adult Care, Newborn Screening and Carrier Screening, recently approved by the ACMG Board of Directors, now can be found on the ACMG website. Included are urea cycle disorders, congenital hypothyroidism, congenital adrenal hyperplasia, galactosemia, homocystinuria and sickle cell disease (transition from pediatric to adult care) and sickle cell carrier screening. The ACT Sheets are point-of-

care education tools that offer critical clinical management information for a variety of conditions not always familiar to practitioners.

ACMG Foundation Update

ACMG Foundation for Genetic and Genomic Medicine Announces *The David L. Rimoin Lifetime Achievement Award* and Endowment Campaign

Following the recent and untimely passing of David L. Rimoin, MD, PhD, FACMG, medical genetics pioneer and Founding President of both ACMG and the ACMG Foundation, the Board of Directors of the ACMG Foundation for Genetic and Genomic Medicine chose to honor Dr. Rimoin in perpetuity by renaming its most prestigious award *The David L. Rimoin Lifetime Achievement Award*. A fundraising campaign is currently being launched to endow this honor to include a significant monetary award, thereby elevating it to the most prestigious award in medical genetics and genomics.

Dr. Rimoin's distinguished career and his enormous legacy truly represent the breadth of accomplishments associated with recipients of this award. These include excellence, unparalleled vision, and national leadership in medical genetics scholarship and research, patient care, teaching, mentoring trainees, and voluntary service that has advanced the field. In 2010, Dr. Rimoin was honored as the second recipient of the ACMG Foundation's Lifetime Achievement Award, following Dr. Arno Motulsky, who received the inaugural award in 2009. Drs. Charles Epstein and R. Rodney Howell were the 2011 and 2012 honorees, respectively.

The David L. Rimoin Lifetime Achievement Award will be given as deemed appropriate, to individuals who exemplify the lifetime achievements and qualities embodied by Dr. Rimoin. The ACMG Foundation is presently working with Dr. Rimoin's family to develop the criteria for the Award.

For more information about the ACMG Foundation's Awards Programs, visit its website at www.acmgfoundation.org.

***Genetics in Medicine* Updates**

Nature Publishing Group took over publication of ACMG's monthly journal, *Genetics in Medicine*, in January 2012. The journal continues to thrive with faster turnaround times from submission to publication and record numbers of submissions coming from an international cadre of medical geneticists. The 2012 impact factor for *Genetics in Medicine* is 4.762, which places us in the top 25 Genetics and Heredity journals, only three of which are clinical.

The following Policy Statement was published in *Genetics in Medicine* between May and August 2012:

> ACMG Board of Directors. **Points to consider in the clinical application of genomic sequencing.** *Genet Med* 14(8):759-761 (August 2012)

2013 ACMG Annual Meeting

The 2013 ACMG Annual Clinical Genetics Meeting will be held March 19-23, 2013 in Phoenix, Arizona, in conjunction with the 44th Annual March of Dimes Clinical Genetics Conference on Skeletal Dysplasias and Connective Tissue Disorders. The meeting website has just opened, with information to be added weekly to include preliminary program and session information—with descriptions of the two pre-conference short courses—on-line abstract submission, registration, hotel reservations and more. To learn about this rich genetics education opportunity, please visit www.acmgmeeting.net. Abstract submission opens in October!

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
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