May 11, 2016

Summary of Recent Activities of The American College of Medical Genetics and Genomics

The American College of Medical Genetics and Genomics (ACMG) is the only nationally recognized medical organization dedicated to improving health through the practice of medical genetics and genomics. ACMG represents over 2000 members, nearly 80% of which are board certified clinical and laboratory geneticists and genetic counselors. ACMG's mission, as redefined in the 2015 Strategic Plan, is to "develop and sustain genetic and genomic initiatives in clinical and laboratory practice, education and advocacy." Three guiding pillars underpin ACMG's activities: 1) Clinical and Laboratory Practice: Establish the paradigm of genomic medicine by issuing statements and evidence-based or expert clinical and laboratory practice guidelines and through descriptions of best practices for the delivery of genomic medicine. 2) Education: Provide education and tools for medical geneticists, other health professionals and the public and grow the genetics workforce. 3) Advocacy: Work with policymakers and payers to support the responsible application of genomics in medical practice. This report highlights key activities of the ACMG between February and mid-May 2016.

ACMG Celebrates 25 Years of Translating Genetic Discoveries into Better Patient Care During Its Most Successful Annual Meeting to Date

• Held in Tampa, FL, March 8-12, 2016, this year's Annual Clinical Genetics Meeting featured many special events to celebrate 25 years of ACMG. The Program included more targeted student/trainee sessions, and new in 2016: Posters remain available online as ePosters at http://epostersonline.com/acmg2016/. We also set new records for attendance, exhibitors, exhibit hall space and press/media coverage. A major example follows:

- *Frontline Genomics* magazine released an "ACMG 2016 Issue" for the 25th Anniversary, featuring an exclusive "Clinical Genetics Now and Then" interview with Executive Director, Dr. Michael Watson, and a 25-year timeline of milestones for the College and ACMG's Foundation for Genetic and Genomic Medicine.

Advocacy, Policy and Practice Activities

• ACMG continued its activities related to educating members and policymakers about its position on the *regulation and oversight of Laboratory Developed Tests (LDTs).*

- On March 1st ACMG sponsored a webinar with the Association for Molecular Pathology, "Impending Federal Legislation on Laboratory Developed Tests", in which the implications of proposed legislative actions and policies on clinical and laboratory practice were discussed.

- Following the late April approval by the House Appropriations Committee of H.R. 5054, the FY 2017 Agriculture, Rural Development, Food and Drug Administration and Related Agencies Appropriations Act, which includes report language requiring that *"…FDA suspend further efforts to finalize the LDT guidance and continue working with Congress to pass legislation that addresses a new pathway for regulation of LDT's in a transparent manner,"* ACMG joined a coalition of medical society and industry stakeholders in bipartisan meetings with the offices of key Senators in the appropriations process to discuss the inclusion of this language in the Senate's Appropriations legislation for FY 2017. ACMG's goal continues to be educating policymakers about our concerns with the FDA draft Guidance and the current FDA-centric draft legislation in the House Energy and Commerce Committee, as well as our recommendations for regulating LDT's through CLIA modernization.

• ACMG Executive Director, Dr. Michael Watson, joined a broad representation of stakeholders at the first *Building the Medical Information Commons* Advisory Committee Meeting. The meeting provided Advisory Committee members and invited guests with a platform to discuss current issues concerning data-sharing policies and practices while simultaneously providing the project team with guidance and feedback. This project seeks to develop an ethical and policy framework for building a medical information commons – a networked environment in which diverse sources of health, medical and genomic data on large populations become broadly available for research and clinical use.

• *wRVU Benchmarking data now available for Clinical Geneticists*: Until two months ago, reliable benchmarking data for the practice of clinical genetics did not exist. This was largely due to the fact that the University Healthcare Consortium's (UHC) Faculty Practice Solutions Center, which collects this data, did not have enough clinical geneticists enrolled in their program. ACMG Medical Director, Dr. David Flannery, made a request to our members, asking that they change their practice category from the specialty of the academic department in which they hold their primary appointment, to Clinical Genetics. With over 250 Clinical Geneticists now enrolled in the wRVU benchmarking process, UHC is generating robust data showing the 25th, 50th, 75th and 90th percentile of wRVU productivity specifically for Clinical Geneticists. ACMG members who are at institutions that participate in the UHC can obtain these objective and externally validated data for use in discussions with department chairs and other administrators.

• *ACMG's Secondary Findings Working Group,* co-chaired by Drs. David Miller and Christa Martin has developed an ongoing process for maintaining the list of mandatory genes that are recommended for testing when patients opt to learn about secondary findings following WES/WGS. Using a standardized, evidence-based vetting process genes will be continually added to (and perhaps removed from) the list originally dubbed "the ACMG 56".

• In Press: Two revised cancer cytogenetics laboratory guidelines:

  • Mikhail FM, Heerema NA, Rao KW, Burnside RD, Cherry AM and Cooley LD; on behalf of the American College of Medical Genetics and Genomics (ACMG) Laboratory Quality Assurance Committee. **Section E6.1–6.4 of the ACMG technical standards and guidelines: chromosome studies of neoplastic blood and bone marrow–acquired chromosomal abnormalities.** *Genet Med* 16(6):635-642 (June 2016)

  • Cooley LD, Morton CC, Sanger WG, Saxe DF and Mikhail FM; on behalf of the American College of Medical Genetics and Genomics (ACMG) Laboratory Quality Assurance Committee. Section **E6.5–6.8 of the ACMG technical standards and guidelines: chromosome studies of lymph node and solid tumor–acquired chromosomal abnormalities.** *Genet Med* 16(6):643-648 (June 2016)

•In Preparation:

  • *ACMG's Revised Position Statement on Non-Invasive Prenatal Screening (NIPS),* which is expected to be published early this summer. The Statement discusses the application of this modality for all pregnant women when screening for Down, Edwards and Patau syndromes. It also clarifies the current state of the science related to using NIPS to screen for sex chromosome aneuploidy and selected conditions involving CNVs. The need for pre- and post test patient education and counseling are emphasized.

  • *Points to Consider in Genomic Screening of Asymptomatic Individuals* will discuss the use of NGS in screening the healthy adult population for genetic predisposition to common conditions as well as late onset rare disorders with a heritable component.

  • Laboratory guideline addressing the *selection of genes in a gene panel:* This guideline will build upon work done by ClinGen that resulted in the recent publication of proposed levels of evidence relating a gene with a disease/phenotype. These proposed evidence levels will provide a framework for recommendations on the evidence needed to support inclusion of a gene on a panel. The guideline will also include standard reporting of exon coverage as well as other quality information (limitations and or confirmation) in the clinical report.

Grant and Contract Updates

• In March the National Coordinating Center for the seven HRSA-funded Regional Genetic Service Collaboratives (NCC) released a final brief and set of recommendations for *Regional Genetic Service Delivery Models* that would reach underserved populations. A working group, developed the document over an eight-month period, and reports primary findings collected using multiple qualitative and quantitative approaches. This final document has been

submitted to HRSA for review.

• The NCC is launching a new state NBS program lab and clinical data collection initiative; the kick-off meeting was held May 11, 2016.

• *Curating the Clinical Genome*, the 2016 ClinGen/DECIPHER public meeting will be held on the Wellcome Genome Campus, Hixon, Cambridge, UK, June 22-24, 2016.

Meetings and Education Updates

• Plans for the *2017 Annual Clinical Genetics Meeting* are under way. The meeting will be held March 21-25, 2017 in Phoenix, AZ. Visit www.acmgmeeting.net for ongoing updates, including forthcoming registration and housing information.

• ACMG continues to sponsor live *Case Conferences* on a monthly basis. There are two ongoing series, and all of the conferences are delivered via webinar and then archived in the ACMG Genetics Academy.

• Genomics Case Conferences occur on the third Wednesday of each month at 2:00 PM ET, with a break during June-August. The May 19th Conference, *Which Error Do You Prefer to Make? The Challenge of Secondary Findings*, will be presented by NHGRI.

• Adult Genomics Case Conferences occur on a quarterly basis and are a terrific learning resource for both medical genetics professionals and primary care practitioners. The next session will be on Tuesday August 9 at 2:00 PM ET.

ACMG Foundation for Genetic and Genomic Medicine Updates

• During the 2016 ACMG Annual Meeting, the ACMG Foundation honored nine individuals for excellence in scientific achievement through a range of *awards*, totaling $350,000. Five fellowship awards were presented to supplement medical genetics residency training with translational research opportunities, particularly in medical biochemical genetics, with one of these awards given for the first time in 2016. Four additional awards recognized scholarly work that either resulted in publication by a trainee, or a presentation at the Annual Meeting. The ACMG Foundation graciously acknowledges the generous corporate contributions and individual donations that have made its robust giving program possible. An overview of the 2016 ACMG Foundation awards is available at www.acmgfoundation.org.

• The *Summer Genetics Scholars Program* (SGSP) has selected twenty-two institutions in nineteen states to participate in this year's program. Pairing a rising second-year student with a medical geneticist mentor (at the student's home institution or at another of the participating schools), the six-to-eight week hands' on experience is intended to give future physicians a "real world" introduction to Medical Genetics and Genomics, and its opportunities and rewards as a career.

•The ACMG Foundation recently received *a $1.65 million commitment from Shire to fund 10 one-to-two year training awards for medical geneticists over the next 3 years* through the ACMG Foundation/Shire Laboratory Geneticist Fellowship and Clinical Genetics Residency Awards Program. This is the single largest corporate gift to the ACMG Foundation in its history, and it comes at a critical time in terms of addressing the shortage of medical geneticists in the US. The partnership between Shire and the ACMG Foundation will also help foster a generation of geneticists worldwide who will play crucial roles in the diagnosis and care of patients with rare and common diseases. ACMG and its Foundation have formed a Task Force that is currently developing a strategy for a cohesive and synergist program to connect all of its training awards.

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.

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