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**The Interface of Genomic Information with the Electronic Health Record:
an ACMG Points to Consider Statement**

Bethesda, MD – June 1, 2020 | Advances in genetic and genomic testing technology have not only introduced the utilization of clinical genomic information into virtually every area of medical care, this testing has become an essential tool to achieve the goal of precision medicine. As genomic data become more complex, so too must the electronic health record (EHR) evolve to provide optimal care for patients, maximizing benefits while minimizing harm. Issues of patient autonomy, access, genetic literacy, privacy and protection, and transferability of data, as well as the appropriate genomic data set, are key in facilitating the incorporation of genomic information into patient care.

In an effort to provide practical guidance and important considerations regarding how genomic information can be incorporated into electronic health records, the American College of Medical Genetics and Genomics (ACMG) has released, "[The interface of genomic information with the electronic health record: a points to consider statement of the American College of Medical Genetics and Genomics \(ACMG\).](#)"

"The electronic health record serves as a powerful interactive tool in improving the healthcare of patients and populations," said Terri Grebe, MD, FACMG, chair of the ACMG Social, Ethical, and Legal Issues Committee. "As an integral component of medical treatment, genomic data in the EHR must therefore be continuously and easily accessible to both patients and providers, while simultaneously receiving appropriate privacy protection, to achieve the goal of personalized medicine. This ACMG document provides guidelines on the storage and access of genomic information, improvements in EHR systems, and ethical issues surrounding the sharing of genomic data."

This new ACMG points to consider document addresses types of genomic information in the EHR, mechanisms of placement, data entry, usage, patient/provider access, results disclosure, portability, and privacy. It highlights patient, family, and societal benefits, discusses areas of concern, identifying where further modifications are needed, and makes recommendations for further optimization. It also highlights unique characteristics of genomic information that require additional attention, as they relate to universal bioethical principles.

A few of the specific points to consider include:

- Genetic data in the medical record should be readily and continuously accessible to the patient, including test results, secondary findings, AND the clinician's interpretation.

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- Caution should be exercised in assessing the quality and medical actionability of outside results from other institutions and laboratories uploaded to the EHR by the patient, particularly direct-to-consumer testing companies. These results would be best stored in a separate section of the EHR or flagged in such a way as to clarify the origin of the report.
- Further optimization of the interoperability of EHR networks is encouraged to allow separate institutions who provide care to the same patient to be able to view the patient's genetic data, furthering coordinated care and minimizing the risk of duplicate testing, with the attendant waste of resources. The use of standards such as the Health Level 7 (HL7) genomics model, and Fast Healthcare Interoperability Resources (FHIR) including the emerging FHIR genomics standards by EHR vendors is encouraged.
- In the future development/revision of EHRs, the ability to easily retrieve genomic information will be vital to enable targeted testing for family members, facilitating cost reduction, earlier diagnosis and treatment.
- Informed consent should be adapted to reflect these points to consider, explicit on right of access, mechanism of access, delayed release of certain results, and potential usage of personal genomic information by the ordering institution as well as outside agencies such as public health programs and genomic databases.

The statement concludes that further research is needed to determine the optimal approaches for patient access to and use of genomic information in the EHR, as well as protecting patient privacy and avoiding harm. While direct patient access to the EHR is appropriate and will facilitate patients' involvement in their own health care, it is not a substitute for face-to-face interaction, which remains the ideal method of communication of potentially life-altering personal health information. These points to consider should be viewed as guidance for the ordering provider, clinical geneticist, laboratory geneticist and genetic counselor, and for institutions and vendors. They are intended to assist providers, institutions and vendors to develop policies and procedures that optimize the use of the EHR in the delivery of healthcare to maximize patient benefit, minimize harm, improve population health and decrease healthcare costs.

About the American College of Medical Genetics and Genomics (ACMG) and ACMG Foundation

Founded in 1991, the American College of Medical Genetics and Genomics (ACMG) is the only nationally recognized medical professional organization solely dedicated to improving health through the practice of medical genetics and genomics, and the only medical specialty society in the US that represents the full spectrum of medical genetics disciplines in a single organization. The ACMG is the largest membership organization specifically for medical geneticists, providing education, resources and a voice for more than 2,400 clinical and laboratory geneticists, genetic counselors and other healthcare professionals, nearly 80% of whom are board certified in the medical genetics specialties. ACMG's mission is to improve health through the clinical and laboratory practice of medical genetics as well as through advocacy, education and clinical research, and to guide the safe and effective integration of

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genetics and genomics into all of medicine and healthcare, resulting in improved personal and public health. Four overarching strategies guide ACMG's work: 1) to reinforce and expand ACMG's position as the leader and prominent authority in the field of medical genetics and genomics, including clinical research, while educating the medical community on the significant role that genetics and genomics will continue to play in understanding, preventing, treating and curing disease; 2) to secure and expand the professional workforce for medical genetics and genomics; 3) to advocate for the specialty; and 4) to provide best-in-class education to members and nonmembers. *Genetics in Medicine*, published monthly, is the official ACMG journal. ACMG's website (www.acmg.net) offers resources including policy statements, practice guidelines, educational programs and a 'Find a Genetic Service' tool. The educational and public health programs of the ACMG are dependent upon charitable gifts from corporations, foundations and individuals through the ACMG Foundation for Genetic and Genomic Medicine.

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