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Canadian College of Medical Geneticists (CCMG) Position Statement on the Storage of Patient Genetic Information in Electronic Health Records

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Purpose

The aim of this document is to provide a statement of the current opinion of the CCMG membership regarding the use of electronic systems for the distribution and storage of genetic information. The specific focus is on whether genetic data should be stored within electronic health records, and if so, whether there are some types of data that warrant exemption. The document does not review important aspects of electronic health record systems, such as database security and back-up, as these concepts apply to all health information. This document is expected to serve as a resource for Canadian College of Medical Geneticists (CCMG) members, and those who are involved in transitioning genetic information from paper records to electronic records, as they produce their own individual policies and practices.

Document development

The Ethics and Public Policy committee drafted this document based on data collected via an online survey of the CCMG membership. The document was circulated to the membership for review and modification. This document has been approved by the CCMG Board of Directors.

Historical Context

Geneticists and Genetic Counsellors have traditionally viewed some aspects of their practices as distinct from conventional screening or diagnostic consultations/tests in medicine, and have evolved particular practices in serving their patients. Many Genetics departments have traditionally opted out of the transcription and medical records systems in their institutions, maintaining a within department system and custodianship; the charting has, in many of the departments, been family- rather than individual-based. This practice of blanket genetic exceptionalism¹, designed especially to protect the sensitive aspects of the practice, is no longer feasible as non-Geneticist health care professionals and the general public become more informed about genetics, the applications of and access to genetic testing diversify, care of individuals becomes more complex and contributed to by great numbers of providers, and institutions move toward more integrated electronic health records. It is important to consider how patient genetic information will be best stored in the electronic age.

Electronic Health Record

For the purposes of this guideline, Electronic Health Record (EHR) will be defined as a longitudinal collection of electronic health information about an individual patient². An EHR will be considered to be a collection of digital information that can be shared within and across different health care settings through the use of network-connections, and that contains a range of patient data including demographics, medical history, consult letters, laboratory test results, and other medically related information. An EHR may be generated and maintained provincially, by a medical institution, a clinic, or a physician's office.

EHRs aim to provide a useful tool for management of complex medical information³. Design features can allow for provision of care that is accessible, continuous, comprehensive, and coordinated⁴. Perceived advantages include introduction of patient specific warnings and reminders to facilitate and improve safety of current and future care for the individual; better efficiency of the health care system, by, for example, reducing duplication of testing and promoting evidence based medicine; and by facilitating compilation of data about populations for the purposes of research and development. However, this accessibility of the data may make patients' privacy and confidentiality more vulnerable. Data bits stored in a well indexed, readily accessible system are much more likely to be seen (often even inadvertently), or sought by more people. A consultation or lab report may become less of a communication to a specific requesting physician, and more of a posting for as yet unknown readers. Thus, implementation of EHRs will require considerations to ensure a patient's data are not misinterpreted and misused.

Should genetic data be posted on EHR?

Most genetic data are quite analogous to other medical data and would be considered to have a favourable benefit to risk ratio for posting on the electronic health record systems. There are some situations in which the ratio is more likely unfavourable. The two more common scenarios are "predictive" testing, and sensitive family information.

In the Genetics community, the term "predictive" is used to refer to an assessment of the future chance of disease in someone who *at present has no known features of the disease process*. Predictive genetic assessment differs from conventional screening or diagnostic consultations/tests as the aim is not to diagnose a current pathology, but rather to establish whether a patient will or will not be affected

by a disease in the future. This prediction can be complicated by concepts such as expressivity and penetrance; which, where relevant, should be clarified explicitly in any health record. Predictive genetic data might take the form of a consult note detailing a presymptomatic risk assessment, a pedigree, or a genetic test result. Standard of practice in predictive genetic testing calls for pretest genetic counseling, informed consent and identifying a patient's preferences regarding how the results will be communicated and with whom they will be shared. Occasionally a patient will defer receipt of, or decide not to receive, their reported result. Genetics departments have developed systems to act according to patients' preferences, including the possibility of a change of mind.

There are some predictive data pieces for which posting would be an advantage, and others for which it would likely not. For a person with a positive predictive genetic test for Lynch syndrome, a hereditary form of colon cancer for which prevention and treatment strategies are available, posting the data would likely do more good than harm. The various care providers can contribute to ongoing surveillance and appropriately modify diagnostic possibilities when assessing presenting complaints. The risk-benefit analysis might be the converse for late-onset conditions that do not have treatment options to prevent the development of symptoms. During the asymptomatic period, the wide availability of the information might have an unfavourable risk-to-benefit ratio for the patient and patient's family. For example, consider the 20 year old woman with a genotype predictive of onset of an autosomal dominant disease at an approximate age of 50 years. If the patient has received adequate counselling around recurrence-risk and family planning options, and chooses no intervention, there maybe no need for the health care professionals involved in the care of her pregnancies and deliveries to be privy to this information. However, the patient may worry about the potential risks of stigmatization or discrimination by having this information widely available in an EHR⁵.

Because genes are shared within families, there are times when a relative's information is collected to inform a patient's consultation. Conversely, there are times when a patient's data risk stratifies or establishes a diagnosis in another family member. For example, detection of a BRCA gene mutation in a given woman establishes the presence of a mutation within the family, thereby significantly stratifying breast cancer risk in her relatives. As a further example, a patient with a maternal grandparent affected with Huntington disease may request predictive genetic testing. If the patient's mother is asymptomatic and has chosen not to pursue predictive testing, a positive test result in the patient also identifies the mother as positive, taking away the mother's choice to not know her disease risk. Individuals requesting testing may have many reasons to seek the result for themselves, but delay or decline to inform family members. The posting of such information pieces in an EHR might pose more risk of violation of privacy through inadvertent disclosure, etc, than benefits to the relatives.

Therefore, when providing patient- and family- centred care there will be some genetic tests or some patient/family contexts that warrant special exemption from regular posting. First and foremost, care must be patient-centric and should not require compromise because of external pressures or apparent limitations of electronic technology. It is the responsibility of anyone involved in a patient's care to aim for creative solutions where they need to be found, in order to be confident that patient-centric care is maintained. System modifications, such as making records visible to only certain care providers (i.e. controlled access, data masking), might avoid potential discrimination. When not possible, an opt-out option (whether on the part of the patient or on the part of the clinic or laboratory posting predictive genetic information) will ensure patient- and family-centred care.

Conclusion

New electronic health record systems provide an opportunity and incentive to evaluate current practices and the potential for adoption of new approaches and technologies. Electronic health record systems offer important benefits such as warnings and reminders that can contribute to the optimal care of the individual; cost saving chances for the system, including the prevention of duplicate or inappropriate future investigations; as well as opportunities for efficient research and development in the population. There are certain types of genetic data that warrant special sensitive treatment. While privacy and confidentiality issues are relevant in any data system, their relevance and the potential risk of harm to patients might be magnified in today's electronic health record systems by the ease of access to, and rapid dissemination of, patient information.

Statement

The CCMG recognizes that electronic health records offer many advantages for individual patients, the health care system, and the development of care in the population. Given that genetic data are frequently subject to misinterpretation, even by health care professionals, and that the electronic health record systems facilitate ready viewing of data by a broad range care providers, postings (letters, reports, etc) should include appropriate interpretation to aid in understanding the implications of this information to the patient and to her or his care. The CCMG recognizes that some genetic information, particularly that which falls under the category of "predictive" and/ or contains data on other family members, is unique and may require special considerations and protections within any record system. In consultation with individual patients on an as needed basis, and with consideration of other family members where relevant, clinicians generating or ordering such an assessment should be able to request special protections or exclusion from the electronic health record.

References:

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