CCMG-CCGM National Office 4 Cataraqui Street, Suite 310
Kingston, Ontario K7K 1Z7
(T) 613-507-8345 (F) 1-866-303-0626
(E) info@ccmg-ccmg.org (W) www.ccmg-ccgm.org

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

Anne-Marie Laberge[1], Nolan D'Souza, Lynette Penney[2,3], Karim Jessa[4], Lauren Chad[5,6,7,8] on behalf of the CCMG Education, Ethics and Public Policy Committee

1 - Department of Pediatrics, CHU Sainte-Justine and Université de Montréal, Montreal, QC, Canada
2- Department of Pediatrics, IWK Health Centre, Halifax, NS, Canada
3 - Faculty of Medicine, Dalhousie University, Halifax, NS, Canada
4- Department of Pediatrics and Information Management Technology, The Hospital for Sick Children, Toronto, ON, Canada
5 - Division of Clinical and Metabolic Genetics, The Hospital for Sick Children, Toronto, ON, Canada
6 - Department of Pediatrics, The Hospital for Sick Children, Toronto, ON, Canada
7 - Department of Bioethics, The Hospital for Sick Children, Toronto, ON, Canada

Purpose

The aim of this document is to provide an updated statement from the Canadian College of Medical Geneticists (CCMG) regarding the current state and some future considerations on the collection, distribution, and storage of genomic information within electronic health records (EHRs), including which aspects of genomic data might warrant special attention. The original version of this document was written by the CCMG Ethics and Public Policy committee in 2010 based on data collected via an online survey of the CCMG membership at the time (Armstrong et al, 2010). It is updated here to reflect the current state of healthcare in 2024, where EHRs are almost ubiquitously used, and genomic medicine has expanded in its breadth and scope. The document was circulated to general membership for review and feedback and has been approved by the CCMG Board of Directors.

Introduction

EHRs aim to provide a useful tool for the management of complex medical information. Design features can allow for provision of care that is accessible, continuous, comprehensive, and coordinated (Scheuner et al, 2009; Ayatollahi et al, 2019). Since this statement was first written in 2010, EHRs have become nearly ubiquitous, with improved functionality and security. At the same time, the application of genomic testing in clinical care have diversified, with more complex patient choices, testing options and multidisciplinary care pathways. These factors, along with the increasing understanding of the importance of access to and integration of genomic information into care decisions make the EHR well-situated to house genomic information, with the primary goal to improve quality, safety and decision making leading to better health. As a genetics community, our questions have shifted from *whether*

genomic information should be part of the EHR to *how* it should be included, and whether there are any special points to consider in our field (McGuire et al, 2008; Hazin et al, 2013; Grebe et al, 2020). Genetic and genomic information has unique characteristics (e.g. predictive capability, general stability over a lifetime, implications for family members, etc.) but these do not justify keeping it out of the EHR based on genetic exceptionalism (McGuire et al, 2008; Evans and Burke, 2008; Grebe et al, 2020). While we recognize the difficulty in producing a national standard, this document is expected to serve as a resource for CCMG members as well as the broader genetics community, and those involved in shaping EHR design, policies and practices.

For the purposes of this statement, Electronic Health Record (EHR) will be defined as a longitudinal collection of digital information about a patient, that can be shared within and across different health care settings. EHRs currently contain a range of patient data including demographics, medical history, consultation letters, laboratory test results, and other medically related phenotypic information. Genetic and genomic information refers to genetic test results, as well as other types of genetic and genomic information such as family histories, clinical interpretation, clinical diagnoses/problem lists, raw sequence data. Some of this patient data is generated within a local organization while other information is generated externally and is incorporated into the EHR. Genetic information exists as discrete information as well as an interpretive narrative in PDF format. This has implications on downstream interoperability and ability to meet local standards.

What genetic and genomic data should be included in EHRs?

Different types of genetic and genomic information are included in EHRs, with substantial heterogeneity in how it is entered, stored, and displayed (Ronquillo et al, 2012; Shirts et al, 2015). Laboratory reports for genetic tests as well as clinical notes represent important sources of genomic data in EHRs.

Genetic tests may now be ordered by genetics and non-genetics health care professionals (HCPs) alike, across nearly all subspecialties in medicine. All clinical grade genetic and genomic tests for an individual patient, including reanalysis of previous testing, should be incorporated in an organized and easy to find fashion into a patient's EHR. These may be relevant to other HCPs in the patient's circle of care, now or in the future. While local policies and practices may differ, a lack of genetic test result visibility in the EHR may lead to duplicate clinical visits, blood draws, testing or other patient safety concerns. In this light, a pathway to include genetic test results that may be obtained in other ways (e.g. at outside centres, privately, sponsored testing, etc.) should also be considered with interoperability implications. Clinical informaticians who understand EHR design can assist with ideal incorporation of genetic information into the medical record.

While EHRs can house research genetic results that have not been clinically validated, great care should be taken to clearly mark these as research results not for clinical decision making, where the worry is they might be misunderstood to be of clinical significance. Other types of genomic results (e.g. Direct-

to-consumer genetic test results) do not have the clinical validity to be included at this time. We recognize that there will continue to be new applications of genetics and expect that new, validated clinical testing strategies that come in time will also be included within the EHR. Access to raw data, which may be desired in the future, brings with it both ethical and technical considerations and cannot be integrated into EHR at this time. Future work is needed to address this.

EHRs should also integrate other types of genetic data, such as family histories/pedigrees and phenotypic data (Ayatollahi et al, 2019). Phenotypic data should be recorded using a terminology system that enables searches and analyses (Marsolo and Spooner, 2013). As EHRs improve in functionality, the ability to search genetic-related data in EHRs will help improve and optimize patient care (Ayatollahi et al, 2019).

Who should have access to genetic and genomic test results?

- **Health care professionals:** HCPs within the circle of care of a patient should have access to all of their clinical genetic tests, with few exceptions outlined below, as they may be important to guide care. If these were performed in multiple labs and/or sites, ongoing organizational efforts underway in some jurisdictions (e.g. Ontario Lab information system (OLIS), Optilab and the Dossier Santé Québec in Quebec) to securely share current and past results should equally apply to genetic results.
- **Patients/Substitute Decision-Makers (SDM).** Genetic test results that are available in the EHR should be available to the patient and/or their SDM, in the same way that the patient can access other test results and documents from their own EHR, often through patient portals, discussed below.

How should genetic and genomic information be included in EHRs?

Given the complexity of some result types in genetics and the possibility that a clinical interpretation may differ from a laboratory interpretation, a clinical note or letter, with an accompanying clinical interpretation and/or care plan should also be easily accessed in a patient's chart. This will help ensure that results are used appropriately by all HCPs and ideally also understood and accessible for patients, discussed below (Kanungo et al, 2020; Grebe et al, 2020).

At minimum, and likely the standard at the time of this writing, genetic results should be in a digital/PDF format, ideally situated with other labs and in a section that is easy to identify. That said, the availability of clinical decision-support tools (e.g. flags for ordering certain classes of medication if pharmacogenetic testing has been performed) and current and future EHR functionality would favour the transition to genomic results as discrete data points rather than stand-alone reports. These discrete data sources should be searchable within the EHR, i.e. suitable for EHR interoperability (Marsolo and Spooner, 2013). The use of clinical integration tools (algorithms, decision aids, etc) can help increase HCPs ability to use

genetic information appropriately and effectively in their patients' care (Hazin et al, 2013). In this way, they can be fully integrated into care, queried and/or reanalyzed with ease (Grebe et al, 2020). Furthermore, current results of uncertain significance may become significant in the future. As groups move toward this, however, the special points below somewhat unique to genetics practice ought to be considered.

Special considerations for genetic and genomic information

While most genetic and genomic data are quite analogous to other types of medical data (e.g. imaging reports, biochemistry results, etc.) and belong in a patient's EHR, there are some situations - addressed below - that require thoughtful consideration (Evans and Burke, 2008; Grebe et al, 2020). Organizational ethics consultations can be sought, if needed, to ensure local practices address these unique concerns while ensuring genomic results are integrated appropriately.

1. **Predictive Testing**

In the medical 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 condition. 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 is at increased chance to develop 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. Standard of practice in predictive genetic testing calls for pretest genetic counseling, including a consenting process that identifies 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 historically developed systems to act according to patients' preferences, including the possibility of a change of mind.

There are some advantages to including predictive testing in the EHR. For a person with a positive predictive genetic test for Lynch syndrome, for example, a hereditary form of colon cancer for which prevention and treatment strategies are available, including this data would likely have more benefit 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 patient may worry about the potential risks of stigmatization or discrimination by having this information available in the EHR. For example, most HCPs would not need to have access to a patient's predictive testing result for Huntington disease. On the other hand, this information may be important if the patient presents with a possible manifestation of HD (Black et al, 2021).

In specific circumstances, for predictive test results or risk assessments with delicate implications, additional measures could be put in place to ensure an additional level of privacy (Darcy et al, 2011; Grebe et al, 2020). In the past, privacy concerns led such sensitive results to be excluded from medical records, including the EHR, often with the use of parallel charts (Eno et al, 2020). Such exclusion of genetic test results raises concerns about future access to meaningful results to ensure continuity of care. To respect patient privacy and autonomy for sensitive test results, while not impeding continuity of care, additional measures could include limited access in the EHR to these results and/or warnings about the sensitivity of the information in the report. The use of other EHR functionality like lock boxes to house sensitive data with audit capabilities is another solution.

2. **Sensitive Family Information**

Because genetic information may be shared within families, one family member's result may be relevant to others. For example, the detection of a pathogenic *BRCA1* variant in one patient may stratify breast cancer risk in family members or guide their own screening/testing strategy. While newer EHRs may facilitate the sharing of family information and/or the linking of relatives, this should only be done with the appropriate consent from the patient in whom the testing was undertaken. If information is shared, individual electronic charts should be created for the storage of each individual's data, representing a change of practice from family-based charting, which may have historically been done in Genetics clinics.

There may be specific situations where a family member's result is needed to guide testing in the patient, or where detailed family information is collected as part of a family history. In these cases, the family member's information may need be part of a patient's chart. If so, it should be put in a separate section of the EHR (e.g. "family" section). This information should not be available to the patient if they request access to their EHR, or to third parties to whom the patient grants access to their EHR. This "family" section should only be accessible to relevant HCPs, i.e. genetics professionals, or come with a warning that this information should be accessed only if clinically relevant (Kanungo et al, 2020). Practically, this may prove difficult, however, as many times a parent's test result is included as part of the patient's report or information is linked in other ways, such as maternal pregnancy information relevant to a newborn's health.

Other types of sensitive family information may be revealed through genetic and genomic testing, such as misattributed paternity or consanguinity. When such sensitive family information is uncovered, consideration for a 'sensitive notes' function, as discussed above for predictive testing, may be most appropriate (Grebe et al, 2020). Other system modifications, such as making records visible to only certain care providers (i.e. controlled access, data masking), might provide other solutions. Organisations should work with their Clinical informaticists and EHR vendors to best meet the needs of these types of results.

3. **Considerations across the lifespan**

Due to the nature of many types of problems that may be genetic in nature, testing may be performed prenatally, where issues may emerge in linking the pregnant individual with the newborn chart. Testing or screening may also be completed at birth or in childhood, when parents act as substitute decision-makers. As pediatric patients age and transition out of the pediatric health care setting, new issues regarding access to their own genetic information may emerge (Grebe et al, 2020), particularly as contemporary testing strategies like whole exome sequencing uncover secondary or other findings that may have implications for health in adulthood. Access to diagnostic genetic test results remains relevant at different stages of life, even if the diagnosis is already clear, for reproductive purposes or access to new treatments. Others may become obsolete if new evidence suggests a different interpretation or new technology allows for a more precise diagnosis. The EHR needs to be able to ensure that genetic and genomic results remain available to the patient across the lifespan, regardless of when testing was performed (Marsolo and Spooner, 2013).

Genetic testing can also be done postmortem, where similar questions have arisen as to where documentation of results ought to occur, particularly if clinical consultation and result disclosure then occurs with a family member or next of kin or scheduling/billing programs do not allow charting on deceased individuals.

Given the multiple testing options, ordering providers and the mainstreaming of genetic testing across all subspecialties and across the entire lifespan of an individual, the EHR is well suited to house an individual patients' various different encounters in genomic medicine and to develop consistent approaches across sites and jurisdictions. Some of this may require local advocacy and education in EHR design and implementation decisions, in a specialty that has historically not had the same clinical presence across all fields of medicine. Engagement and consultation with clinical informatics expertise can help facilitate optimum design. Patient portals, discussed below, are another way to ensure access to genomic results across the lifespan, with particular attention to adolescent populations with emerging autonomy where new models of care may be needed.

Considerations for other functionality afforded by EHRs

1. **Patient Access and Portals**

Many EHRs now have patient portals whereby patients can access their own health information, test results and consult notes securely, enhancing patient autonomy. This access needs to be balanced against the need for appropriate counseling and potential patient harms (Grebe et al, 2020). Use of patient-specific portals allows for the integration of support tools that can increase understanding of the implications of genetic test results to benefit the patients and families (Hazin et al, 2013). In the United States, the 21st Century Cures act has mandated immediate information sharing of results and notes (U.S. Congress, 2016). No such standards or mandates exist in Canada. For patients, genetic and genomic results on portals should be accessible in a meaningful way to support patient understanding. Release of genomic results and clinical notes to patient portals should take into account the range of

options for timing the release with the potential for a sensitive diagnosis to be received before counselling and support may be available (Grebe et al, 2020). Some centres may opt for delayed release (e.g. 7 days) in case there is communication/actionability that would be best communicated by a healthcare provider directly. Still others may opt for manual release, after results have been communicated to the patient, while others may practice immediate release. While some studies show patients prefer not to learn of consequential test results through an electronic portal (Bruno et al, 2022), there remain strong ethical arguments in favour of preserving autonomy and following local norms and practices.

2. **Consent & Communication**

With a menu of options for the return of results and current practice supporting a patient-led decision on which results to return, EHRs should ideally be leveraged to not only document a patient's consent and preferences for the return of specific genomic information, but to communicate these preferences to the lab for the generation of a report. Given genomic interpretation can change in time, consent related to future wishes to be recontacted is also ideally placed in an EHR, as well as further work toward building dynamic consent models, whereby patients can change their consent over time (Kaye et al. 2015).

3. **Decision Support**

As mentioned above, some EHRs include flags, patient specific warnings and decision-support to facilitate and improve patient safety (e.g. with pharmacogenomic input or metabolic emergency care plans) (Darcy et al, 2011; Hazin et al, 2013; Grebe et al, 2020). As well, incorporation of genomic information in EHR allows better efficiency of the health care system, by reducing duplicate testing or enabling efficiencies with electronic workflows – and should be encouraged.

4. **Research**

Not surprisingly, EHRs are also proving to be powerful tools for research in the genetics space, with the ability for cross-institutional data sharing and the development of large data sets to advance rare disease research and genotype-phenotype descriptions. At the same time, heterogenous practices in data collection and sharing and the use of different EHR platforms across centres remain significant limitations. EHRs used for larger data driven initiatives, where historically underrepresented populations may be included, some of whom may have not explicitly consented to their data being used in this way, bring a myriad of other issues to be aware of. While there might be tremendous benefit for individuals with both rare and common genetic conditions, research advanced in this way may perpetuate inequities that already exist in our healthcare system. The potential privacy of patients with rare and ultra-rare conditions should also be considered, even with fully anonymized/deidentified practices.

Conclusion

EHRs provide an opportunity to evaluate current practices and adopt new approaches for the next generation of genomic medicine. The CCMG recognizes that EHRs offer many advantages for individual patients, the health care system, and the development of care in the population. EHRs offer important benefits such as provider and patient-facing warnings and reminders that can contribute to the optimal care of the individual; cost saving changes for the system, including the prevention of duplicate or inappropriate future investigations; as well as opportunities for efficient research and development in the population-at-hand. Clinical informatics should continue to be involved in the thoughtful design of genomics information systems and how they can support clinical care and safety.

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 EHRs by the ease of access to, and rapid dissemination of, patient information. Given the broad range of care providers that may encounter genetic and genomic information, including genomic results, access to appropriate clinical interpretation remains important. The CCMG also 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 ethics committees, clinical informatics specialists, and with consideration of all family members where relevant, clinicians generating or ordering such an assessment should be able to request special protections or exclusion from the EHR.

First and foremost, the provision of genetic healthcare should continue to be patient-centered and should not be compromised because of apparent limitations of electronic technology. How EHRs will grow and adapt to house other types of genomic data remains an emerging and developing field in the years ahead. Responsible development and implementation of genetic information in EHRs should aim to benefit patients and consider clinical context and stakeholders’ views (Jacquemard et al, 2021).

Summary

- There are several unique considerations for genetic health information in the EHR, therefore genetics representation should be included at the time of EHR development.
- Genetic and genomic information (clinical summaries and test results) should be organized in a centralized, dedicated location within the EHR that is easily accessible by health care providers, patients and substitute decision makers.
- Each genetics patient should have a discreet chart, avoiding the historical practice of a family chart. EHR functionality should be used to leverage linking individual patients
- All clinical grade genomic test results for an individual patient, including reanalysis of previous testing, should be housed in the patient’s EHR, ideally in discrete format.
- Great care should be taken with research genetic results, particularly those that have not been clinically validated, which should be clearly marked as such.
- With appropriate safeguards in place, the EHR could be a powerful tool to optimize data sharing for genetics research.

- There should be specific consideration of the situation of access to sensitive results and the circumstance where a patient chooses not to receive predictive testing results and how to house those results.
- Family information (obtained with appropriate consent and important to the care of the patient) not appropriate for broad sharing among the healthcare team, such as family history information and test results of other family members, deserve special consideration within the EHR such as a "family" section.
- Immediate or delayed release of results to patients are possible and both bring with them advantages and limitations. Consideration should be given to housing of and access to results of testing initiated by parents or SDM in the prenatal or childhood setting.
- We recognize that there will continue to be new applications of genetics and expect that new, validated clinical testing strategies that come in time will also be included within the EHR.
- Clinical informatics can be valuable partners in the optimal design and application of genetic information in the EHR.

References

21st Century Cures Act. H.R. 34, 114th Congress. 2016. https://www.gpo.gov/fdsys/pkg/BILLS-114hr34enr/pdf/BILLS-114hr34enr.pdf. Last accessed June 19, 2024 at 1:34 PM

Armstrong L, Nelson T and Bruyère H. Canadian College of Medical Geneticists (CCMG) Position Statement on the Storage of Patient Genetic Information in Electronic Health Records. September 2010. Retired.

Ayatollahi H, Hosseini SF, Hemmat M. Integrating Genetic Data into Electronic Health Records: Medical Geneticists' Perspectives. Healthc Inform Res. 2019 Oct;25(4):289-296. doi: 10.4258/hir.2019.25.4.289. Epub 2019 Oct 31. PMID: 31777672; PMCID: PMC6859263.

Black KJ, Barton SK, Perlmutter JS. Presymptomatic Testing and Confidentiality in the Age of the Electronic Medical Record. J Neuropsychiatry Clin Neurosci. 2021 Winter;33(1):80-83. doi: 10.1176/appi.neuropsych.20030068. Epub 2020 Jul 16. PMID: 32669021.

Bruno B, Steele S, Carbone J, Schneider K, Posk L, Rose SL. Informed or anxious: patient preferences for release of test results of increasing sensitivity on electronic patient portals. Health Technol (Berl). 2022;12(1):59-67. doi: 10.1007/s12553-021-00628-5. Epub 2022 Jan 8. PMID: 35036280; PMCID: PMC8741580.

Darcy DC, Lewis ET, Ormond KE, Clark DJ, Trafton JA. Practical considerations to guide development of access controls and decision support for genetic information in electronic medical records. BMC Health Serv Res. 2011 Nov 2;11:294. doi: 10.1186/1472-6963-11-294. PMID: 22047175; PMCID: PMC3229457.

Eno CC, Barton SK, Dorrani N, Cederbaum SD, Deignan JL, Grody WW. Confidential genetic testing and electronic health records: A survey of current practices among Huntington disease testing centers. Mol Genet Genomic Med. 2020 Jan;8(1):e1026. doi: 10.1002/mgg3.1026. Epub 2019 Nov 7. PMID: 31701651; PMCID: PMC6978271.

Evans JP, Burke W. Genetic exceptionalism. Too much of a good thing? Genet Med. 2008 Jul;10(7):500-1. doi: 10.1097/gim.0b013e31817f280a. PMID: 18580684.

Grebe TA, Khushf G, Chen M, Bailey D, Brenman LM, Williams MS, Seaver LH; ACMG Social, Ethical and Legal Issues Committee. 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). Genet Med. 2020 Sep;22(9):1431-1436. doi: 10.1038/s41436-020-0841-2. Epub 2020 Jun 1. PMID: 32475985.

Hazin R, Brothers KB, Malin BA, Koenig BA, Sanderson SC, Rothstein MA, Williams MS, Clayton EW, Kullo IJ. Ethical, legal, and social implications of incorporating genomic information into electronic health records. Genet Med. 2013 Oct;15(10):810-6. doi: 10.1038/gim.2013.117. Epub 2013 Sep 12. PMID: 24030434; PMCID: PMC3926430.

Jacquemard T, Doherty CP, Fitzsimons MB. The anatomy of electronic patient record ethics: a framework to guide design, development, implementation, and use. BMC Med Ethics. 2021 Feb 4;22(1):9. doi: 10.1186/s12910-021-00574-x. PMID: 33541335; PMCID: PMC7859903.

Kaye, J; Whitley, EA; Lund, D; Morrison, M; Teare, H; Melham, K (2015). "Dynamic consent: a patient interface for twenty-first century research networks". *Eur. J. Hum. Genet*. **23** (2): 141–6. doi:10.1038/ejhg.2014.71. PMC 4130658

Kanungo S, Barr J, Crutchfield P, Fealko C, Soares N. Ethical Considerations on Pediatric Genetic Testing Results in Electronic Health Records. Appl Clin Inform. 2020 Oct;11(5):755-763. doi: 10.1055/s-0040-1718753. Epub 2020 Nov 11. PMID: 33176390; PMCID: PMC7657708.

Marsolo K, Spooner SA. Clinical genomics in the world of the electronic health record. Genet Med. 2013 Oct;15(10):786-91. doi: 10.1038/gim.2013.88. Epub 2013 Jul 11. PMID: 23846403; PMCID: PMC4096770.

McGuire AL, Fisher R, Cusenza P, Hudson K, Rothstein MA, McGraw D, Matteson S, Glaser J, Henley DE. Confidentiality, privacy, and security of genetic and genomic test information in electronic health records: points to consider. Genet Med. 2008 Jul;10(7):495-9. doi: 10.1097/gim.0b013e31817a8aaa. PMID: 18580687.

Ronquillo JG, Li C, Lester WT. Genetic testing behavior and reporting patterns in electronic medical records for physicians trained in a primary care specialty or subspecialty. J Am Med Inform Assoc. 2012 Jul-Aug;19(4):570-4. doi: 10.1136/amiajnl-2011-000621. Epub 2012 Apr 17. PMID: 22511017; PMCID: PMC3384119.

Scheuner MT, de Vries H, Kim B, Meili RC, Olmstead SH, Teleki S. Are electronic health records ready for genomic medicine? Genet Med. 2009 Jul;11(7):510-7. doi: 10.1097/GIM.0b013e3181a53331. PMID: 19478682.

Shirts BH, Salama JS, Aronson SJ, Chung WK, Gray SW, Hindorff LA, Jarvik GP, Plon SE, Stoffel EM, Tarczy-Hornoch PZ, Van Allen EM, Weck KE, Chute CG, Freimuth RR, Grundmeier RW, Hartzler AL, Li R, Peissig PL, Peterson JF, Rasmussen LV, Starren JB, Williams MS, Overby CL. CSER and eMERGE: current and potential state of the display of genetic information in the electronic health record. J Am Med Inform Assoc. 2015 Nov;22(6):1231-42. doi: 10.1093/jamia/ocv065. Epub 2015 Jul 3. PMID: 26142422; PMCID: PMC5009914.