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# Confidential genetic testing and electronic health records: A survey of current practices among Huntington disease testing centers

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## Abstract

**Background:** Clinical care teams providing presymptomatic genetic testing often employ advanced confidentiality practices for documentation and result storage. However, patient requests for increased confidentiality may be in conflict with the legal obligations of medical providers to document patient care activities in the electronic health record (EHR). Huntington disease presents a representative case study for investigating the ways centers currently balance the requirements of EHRs with the privacy demands of patients seeking presymptomatic genetic testing.

**Methods:** We surveyed 23 HD centers (53% response rate) regarding their use of the EHR for presymptomatic HD testing.

**Results:** Our survey revealed that clinical care teams and laboratories have each developed their own practices, which are cumbersome and often include EHR avoidance. We found that a majority of HD care teams record appointments in the EHR (91%), often using vague notes. Approximately half of the care teams (52%) keep presymptomatic results out of the EHR.

**Conclusion:** As genetic knowledge grows, linking more genes to late-onset conditions, institutions will benefit from having professional recommendations to guide development of policies for EHR documentation of presymptomatic genetic results. Policies must be sensitive to the ethical differences and patient demands for presymptomatic genetic testing compared to those undergoing confirmatory genetic testing.

## KEYWORDS

confidentiality, electronic health record, Huntington disease, presymptomatic testing, privacy

## 1 | BACKGROUND

Clinical laboratories and clinicians follow strict policies for resulting and storage of data from clinical testing. Usually, the ordered test is clearly identified and resulted in a way that the care teams can readily access them. The goal of

clear, accessible records has become even more widespread with the advent of the electronic health record (EHR). While genetic information in the EHR has become mainstream, a few genetic tests still defy these practices for the sake of confidentiality and to address clinician and patient concerns about stigmatization and potential discrimination, such as

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in insurability and employment. While there are a handful of these conditions, including frontotemporal dementia [OMIM: 600274] and autosomal dominant Alzheimer disease [OMIM: 607822, 104300], the most established is the protocol (HDSA, 2016) for presymptomatic genetic testing for the presence of the expanded trinucleotide repeat associated with Huntington disease (HD) [OMIM: 143100]. HD is an autosomal dominant neurodegenerative disorder associated with an expansion of a polymorphic CAG trinucleotide repeat in exon 1 of *HTT* (Gusella et al., 1983; HDCRG, 1993). While the disease is typically late-onset, most commonly between the ages of 30–50, individuals of any age can have HD (Roos, 2010). HD is invariably fatal following a several-decade progressive decline in motor control, cognitive faculties and behavioral disturbances. Direct testing in symptomatic patients and presymptomatic testing in at-risk family members has been available since 1993. Whereas HD testing is widely available and fairly routine (Losekoot et al., 2013), uptake of testing remains low among healthy people at risk for HD, with fewer than 10% of at-risk adults in the United States getting tested. This number is only slightly higher in countries with a socialized system of health care; in the UK approximately 17% of people at-risk for HD are tested (Baig et al., 2016).

Due to the nearly 100% penetrance of this adult-onset condition, the decision for an individual to undergo testing, especially prior to the onset of symptoms, is difficult, as the results may not just affect their life and family but could also produce serious psychosocial reactions and lead to future financial planning, educational, and employment complications (Divino et al., 2013). The Genetic Information Nondiscrimination Act of 2008 (GINA, 2009) and the Affordable Care Act (ACA, 2010) protect individuals from health insurance discrimination; however, other insurance products such as disability, life, and long-term care are not protected by these laws. Thus, a positive presymptomatic HD test result may render an otherwise healthy individual uninsurable. Stakeholders such as the Huntington's Disease Society of America (HDSA), the American Society of Human Genetics (ASHG), and the American College of Medical Genetics and Genomics (ACMG) (ACMG/ASHG, 1998), have stressed extensive pre-test counseling and confidential testing to “protect the well-being of individuals who choose to be tested” (HDSA, 2016). However, maintaining strict confidentiality of test results is difficult, especially in the age of the EHR, and may actually compromise patient care by restricting access to essential diagnostic information.

The field of genetics has wrestled with the possibility of discrimination since the advent of testing for sickle cell disease (Fulda & Lykens, 2006). Since HD was one of the first genes for which presymptomatic individuals could choose to be tested, protocols were established to maintain utmost confidentiality. Results are generally not withheld from the EHR

except for certain genetic results which are deemed confidential, which is in line with “genetic exceptionalism” (Murray, 1997), namely the practice of treating genetics results differently based on a paternalistic view of patient care. Even though there was a precedent for constraint in the early days of HIV testing (Malmberg, Phan, Harmon, & Nauert, 2012; Williams, 2011), resulting of HIV status in a secure EHR is generally promoted over paper records, so as to help the individual with their personal care and as a mechanism for public health reporting (Malmberg et al., 2012).

If strict confidentiality is to be maintained, special precautions must be in place from the moment the patient enters the clinic, from scheduling, through test ordering, resulting, to post-test disclosure and genetic counseling; however, confidentiality of the laboratory test report is a moot point if the information ends up in a clinician's note. This survey of US clinical care teams who routinely order HD presymptomatic testing of individuals addresses the use of the EHR throughout the process as a means to reveal the day-to-day issues with confidentiality in light of EHR use. The ACMG and ASHG statements regarding HD testing only speak to the technical standards and guidelines of the test itself (ACMG/ASHG, 1998) and do not offer recommendations for how the care team handles the test order or results. The HDSA testing protocol (HDSA, 2016) addresses standards for educating and supporting individuals being tested; however, it does not offer standards for managing the legal obligations and challenges of maintaining confidentiality in the EHR. Lastly, the reporting of a single type of test differently because of confidentiality concerns may need to be examined, and a more practical protocol may need to be established.

## 2 | METHODS

### 2.1 | Ethical compliance

Neither certification of exemption from UCLA IRB review nor UCLA IRB approval of the proposal and survey questions were required since the activities did not involve “human subjects” research as defined by federal regulations for human subject protections.

### 2.2 | Survey procedures

We contacted 43 United States Centers of Excellence listed on the HDSA website (Huntington's Disease Society of America, 2019) to query their policies regarding availability of genetic results for presymptomatic individuals. We emailed the contact person listed on the site, and those who did not respond were contacted one additional time. Twenty-three clinical care teams from 17 states offering HD genetic testing agreed either to a phone interview or to fill out a written questionnaire regarding their current practices. The single

member representing the common practice at their institution included genetic counselors, social workers or clinicians. For the phone interviews, the survey was sent prior to the conversation, filled out based on their statements, and responses were confirmed during the phone call. During the phone interviews, representatives were generally more open to sharing their concerns and giving examples of disparities in this testing population (see Discussion). Two centers rejected the survey because they do not currently perform presymptomatic genetic testing. Eleven never responded to correspondence, and two responded but did not want to participate. In five cases, the contact person responded to the email, indicating they were no longer involved with the listed center and offered to forward the request to the correct person; however, no response from that center was ever received.

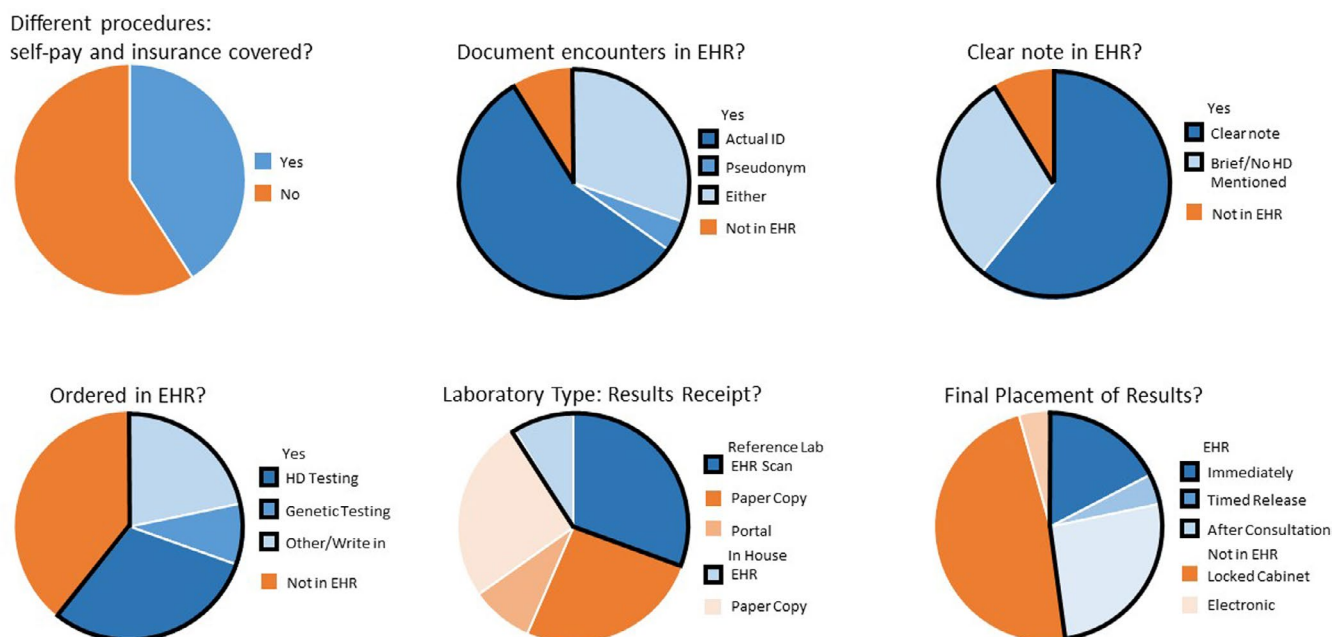
Questionnaires focused on how the HD gene analysis is ordered, resulted, and stored. The questionnaire was multiple choice, and answers were compiled and counted. Each member representing their institution was instructed to answer based on how their institution handles presymptomatic individuals (Figure S1). All answers were kept confidential.

### 3 | RESULTS

All responding centers perform presymptomatic testing for expanded HD alleles. A minority of institutions (39%) have different procedures for individuals choosing to use self-pay versus insurance, including sending the test to a different laboratory and documentation in the EHR (Figure 1). Two institutions do not allow for a self-pay option, and one does

not bill insurance for their services/testing. Approximately half of the patients (52%) choose to use insurance when possible, and multiple institutions noted that the percentage of patients choosing insurance to cover the test has been increasing in the past few years. Some institutions noted that the majority of the individuals choose insurance coverage for the initial encounter with an ordering physician, even if the individual chooses self-pay for the laboratory test. A majority of institutions (56%) document encounters in the EHR using the individual's true identity, and 30% of centers allow for the individual's choice of true identity or a pseudonym. A minority of institutions (9%) have avoided EHR use completely. However, one of the centers avoiding the EHR is unique as it is a research center rather than a clinical center, thus allowing it this freedom. Of the institutions documenting the individuals in the EHR, a majority of clinicians (67%) write a clear, brief note in the EHR, discussing the testing ordered and family history, and of the remaining 33%, the clinician note does not state the testing in question with some referring to a paper note ("shadow chart") accessible offline.

A majority of institutions (61%) order testing through the EHR. Of those, 57% order the test under "HD testing". The remaining (43%) teams order tests that are not specific to HD, such as "Genetic Analysis," "Special Procedure" or "Molecular Pathology". In the latter cases, the care team wrote in "HD testing". A minority of institutions (39%) order by paper requisition or through a laboratory portal not linked to the individual's EHR. A majority (65%) uses a reference laboratory for testing, some noting this is unique only to their self-pay patients and not their patients using insurance coverage. The remaining centers (35%) conduct the testing



**FIGURE 1** Results from a clinical care team survey on the process of ordering, resulting, and storage of presymptomatic genetic testing for Huntington disease. Blue represents tasks completed on the EHR and orange represents tasks completed off the EHR

in-house. Interestingly, whether or not the care team ordered the test through the EHR is not correlated with the type of laboratory (in-house or reference) running the test ( $p = .91$ ).

Of those using a reference laboratory, 47% receive the results via the EHR, and 53% receive results either via fax or through a laboratory portal. Of those care teams utilizing an in-house laboratory, most (75%) received a paper copy, and 25% of the teams noted the results came through the EHR. No matter the type of laboratory (reference or in-house), the test results are accessible to all practitioners in the EHR at 48% of total institutions, with 54% of these releasing results in the EHR after the care team has the results appointment with the individual. A slim majority of institutions (52%) store the results outside of the EHR, either in a locked cabinet (91%) or in electronic form (9%). Two institutions upload the results into the EHR depending on discussions with the patient.

We asked the institutions for their impression as to the confidentiality concerns of patients seeking presymptomatic testing. Answers ranged from adamant about patient confidentiality to being content with having results in the EHR (Figure S1). A minority of institutions (30%) noted the latter; however, three centers do not have an option to avoid the EHR. Those institutions with the option of anonymity noted that many to all their patients are concerned enough to have a discussion. Multiple institutions chose all three options for this question as, not surprisingly, their patients have different opinions as to confidentiality of their results.

## 4 | CONCLUSION

Strict maintenance of confidentiality in the age of universal EHR use is difficult. Institutional adherence to the laws of medical privacy is typically in conflict with the patient's understanding of and wishes for privacy. In the world of EHRs, particularly nearly universal medical record access when multiple unrelated institutions use the same EHR platform, routine clinical care may also infer an ability for providers of all types to have unfettered access to genetic testing results. Many patients will view this as a breach of their confidentiality when in reality this access is fully within the bounds of HIPAA and other privacy policies. Our survey results suggest that HD centers have a desire to protect the confidentiality of patients; however, this is becoming increasingly difficult with the modernization of medical records through EHRs and legal requirements. Still, centers often create workarounds to avoid notice in the EHR, which in some cases places an unnecessary burden on the laboratory and clinic at a time when efficiency in the EHR is preferred, necessary, and sometimes mandated by the hospital/institution to receive reimbursements from the US government (ARRA, 2009). These workarounds also increase the chances for identification errors, inability to retrieve results when needed, loss of records, and

miscommunication. Since testing protocols (HDSA, 2016) do not address the laws and policies regarding maintenance of records, each individual center must establish methods within the bounds of their larger medical institution. While this survey addresses HD as an example, establishing best practices for testing and reporting would be helpful for other cases of presymptomatic genetic testing of individuals harboring detrimental variants or expanded alleles in highly penetrant, late-onset diseases.

### 4.1 | Pre-test appointments

Starting with the act of booking the initial neurologic or genetics appointment, individuals must decide whether or not to use insurance to pay for the appointment(s). In many cases, for insurance to cover the appointment(s), a note and (suspected) diagnosis must be documented in the EHR. Centers that document appointments in the EHR ranged from stating “see paper note” to stating “family history of neurologic disorder” or “family history of HD”, although most stated they keep the note as brief as possible. One clinician who does not mention HD in his note in order to conform to longstanding practices at that center felt that not being explicit as to the testing did not represent appropriate medical care. A possible solution could be an access-limiting note, which would deem such notes as private and appear to maintain the argument of “genetic exceptionalism” (Green & Botkin, 2003; Murray, 1997). Access-restricted notes were utilized at one center; however, recently their institution has blocked this feature, and all notes are available to the EHR in their entirety. In order to achieve true informed consent for HD testing, centers must make an effort to explain to the patient, prior to scheduling their initial testing appointment(s), the practices and limitations of the documentation and its visibility in the EHR. This is a difficult task, particularly as these practices and views of “genetic exceptionalism” change over time (Evans & Burke, 2008; Murray, 2019; Rothstein, 2007).

### 4.2 | Testing order name

Clinical laboratory test ordering needs to be clear and concise to avoid mistakes between the care team and laboratory interface. In many cases in the medical field, the test names are overly complex and full of abbreviations leading to confusion (Passiment et al., 2013); however, this survey shows the opposite may have the same effect. One institution uses the term “Genetic Analysis,” which is a specific “unwritten code” for HD testing at that laboratory. This unclear orderable may lead to inaccurate orders, which must then be clarified. For example, clinicians not familiar with the “code” may use this entry to order a multitude of genetic tests, for example, Lynch syndrome. The laboratory then may contact the



clinician by email to confirm the test, stating they received an order for “Genetic Analysis” and specifically stating HD testing in the email, thus breaking the confidentiality that was originally intended by the discreet name of the test order.

A minority (39%) of care teams avoid the EHR for ordering the test, opting for paper requisition forms or ordering through a separate laboratory portal. Thus, they are able to avoid the confidentiality breach that may occur by ordering the test through the patient's personal EHR. However, some clinicians and informaticists (Jamoom, Patel, Furukawa, & King, 2014) argue that ordering through the EHR allows for efficient use of resources: lower costs, fewer errors, and quicker turn-around time.

### 4.3 | Storage of results

When the test is resulted, most institutions do not place results in the EHR, instead handing out a paper copy. While this maintains confidentiality, avoiding the EHR places a burden on the individual and the care team to keep results easily accessible. In general, genetic testing reports must be retained by the laboratory for a minimum of 10 years (Schwartz, 1999). However, individuals may undergo presymptomatic HD testing many years prior to presenting symptoms of the disease. In addition, a recent survey found the age of presymptomatic testing has decreased, thus resulting in a longer period of time between testing and symptoms (Holman et al., 2018). EHR is more permanent, allowing for easier maintenance of results.

A majority of care teams choose to keep results in a locked cabinet, which is counter to recommendations that storage of genetic results be in the EHR (ARRA, 2009; Botkin et al., 2015; Shoenbill, Fost, Tachinardi, & Mendonca, 2014). While one can argue that the storage cabinet is safer, it places excessive burden on the staff member charged with security. Beyond those concerns, as defined by CLIA and HIPAA, the patient has a right to access their test reports upon request, and laboratories subject to CLIA must be able to provide copies of the requested completed test (CMS et al., 2014; Schwartz, 1999). Individuals returning decades post-testing for a copy of their results will result in an overwhelming amount of work to retrieve the correct files, which in the interim may have been relocated to remote sites or stored on media that is no longer readable.

### 4.4 | Care team accessibility

Accessibility of testing results to the care team is a substantial concern. While the EHR is intended to provide an accurate medical history, there is the worry of intended and unintended views of protected health information (PHI). State laws on the subject of who can receive/access results vary across the US, with a majority allowing for only

medical professionals who “need to know” having access to results. In this matter, one could argue that placement of any type of test where the results were deemed to need a higher level of protection should not be in a fully accessible EHR.

Maintaining confidentiality of laboratory testing is not new to the medical records field. Beyond genetics, adolescent care, mental health, and substance abuse treatment teams have been the most vocal about confidentiality of the EHR, and thus have built-in privacy functions in the software platform used. These teams have created “safeguards” to avoid intended and unintended views of patient PHI, such as, “soft barriers” or “hard stops”. “Break-the-glass” is intended to warn physicians that they are embarking on information deemed more private. Those who do “break-the-glass” are then tracked and if the access to that information was deemed inappropriate, may lead to reprimand or termination based on the breach severity. However, this feature is generally seen as a pseudo-protection since it does not block access for any clinician, nurse, medical student or laboratorian using the interface. Thus, it is only a perceived protection.

The “hard stops” refer to a block in the information, such that only those designated by the program can have access to such data. One principal reason the EHR was developed was to make data sharing easier with other clinicians; thus, every informatically placed block leads to a decrease in the availability of information to another care team who may be called upon to treat the patient without knowing the historical context (Shenoy & Appel, 2017). Some commented that it is important for any future care team to know HD results in case an individual becomes symptomatic and needs proper care. Thus, keeping genetic test results out of the EHR may have the potential to cause delays in care, misdiagnoses, unnecessary repeat testing, or increase costs of care in the future.

Knowledge of rare, highly penetrant, late-onset genetic diseases may be relatively low in general medical practice. One study found that few providers felt knowledgeable on the subject of genetics, interpreting genetic test results and able to care for patients with genetic testing (Hauser, Obeng, Fei, Ramos, & Horowitz, 2018). For example, a positive presymptomatic test result for HD is not the same as a clinical diagnosis of HD.

EHR confidentiality extends beyond the health of just the individual. Some states/insurance carriers will cover in vitro fertilization (IVF)/preimplantation genetic diagnostic testing (PGD) (Drazba, Kelley, & Hersherberger, 2014); however, to cover such testing, the genetic variant must be on record. The same would apply for entry into any clinical trials of HD prevention or therapy. In one case cited in the survey results, the couple decided that having the positive HD genetic test recorded in the EHR was a much greater risk than paying

out-of-pocket for IVF with PGD testing, a process known to cost tens to hundreds of thousands of dollars (Goldman et al., 2018). Thus, if the EHR included this information in a more confidential matter, families may be more comfortable with recording this information in their EHR and not have to make these difficult decisions.

Many institutions surveyed mentioned the results are immediately placed in the EHR but withheld completely or for a period of time from the patient portal. No specific laws or regulations have been established to address the degree of information suppressed from a patient-accessible portal. HIPAA privacy rule deems that patients have a right to their laboratory results, and these results should not be withheld *based on the sensitive nature or potential for causing distress to the individual* (CMS et al., 2014). Thus, it may behoove professional bodies to decide if all genetic results be available in a patient portal to allow patients to access their data which either they or their insurance paid for.

## 4.5 | Limited professional guidance

We found a wide range of institutional protocols ranging from full disclosure (clinician note, test ordered and results) to absence in the EHR. In an era of reliance on the EHR, the reporting of a single test differently from all other laboratory results because of confidentiality concerns may need to be examined and a more practical and uniform protocol may need to be established by professional bodies. Survey responses revealed a number of other concerns, such as cumbersome storage and retrieval of test results, lost results, vague or misleading clinical notes, additional intralaboratory burdens, and, now that targeted therapies (Dickey & La Spada, 2018) for HD are emerging, loss of eligibility for tested patients to enter clinical trials due to delays in obtaining test results outside of EHRs. The recommended HDSA protocol for testing suggests a level of confidentiality that was effective when all documentation was maintained on paper. However, with many hospitals and laboratories predominantly using EHRs for all appointments, notes, communications with patients, and testing, we found a large spectrum of practices are occurring in the field.

Beyond presymptomatic HD testing, institutions also need to consider which other genetic conditions deserve this highest level of confidentiality, thus begging the question of how to make the determination and who should decide. Testing for asymptomatic carriers of *BRCA* variants has been done for roughly the same amount of time; however, those results are not concealed from the EHR (though some patients still prefer to pay out of pocket and use a pseudonym). Of course, hereditary breast/ovarian cancer is a quite different disease with lower penetrance and far more

options for prevention and treatment. Beyond genetics, HIV status, which early in the epidemic was kept strictly confidential, is now encouraged to be documented in the EHR under the argument of effective care for the person (Green & Botkin, 2003; Herwehe et al., 2012; Safran et al., 1995). Recommended criteria to guide clinical teams and laboratories when establishing a protocol for confidentiality of predictive genetic testing do not exist. Therefore, professional recommendations for the appropriate use of EHRs to document, order, and report genetic testing for late-onset, highly penetrant conditions in presymptomatic individuals are needed to help standardize and optimize current clinical and laboratory practices.

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## CONFLICTS OF INTEREST

None of the authors have any conflicts of interest to disclose.

## AUTHOR CONTRIBUTIONS

CCE designed, conducted, and organized survey data and wrote the initial draft of the manuscript. SKB and ND participated in concept development and survey design. SDC, JLD, and WWG advised throughout survey development and edited the manuscript. All authors reviewed the final manuscript.

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## SUPPORTING INFORMATION

Additional supporting information may be found online in the Supporting Information section.

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