Ethical Considerations on Pediatric Genetic Testing Results in Electronic Health Records

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Abstract

Background Advances in technology and access to expanded genetic testing have resulted in more children and adolescents receiving genetic testing for diagnostic and prognostic purposes. With increased adoption of the electronic health record (EHR), genetic testing is increasingly resulted in the EHR. However, this leads to challenges in both storage and disclosure of genetic results, particularly when parental results are combined with child genetic results.

Privacy and Ethical Considerations Accidental disclosure and erroneous documentation of genetic results can occur due to the nature of their presentation in the EHR and documentation processes by clinicians. Genetic information is both sensitive and identifying, and requires a considered approach to both timing and extent of disclosure to families and access to clinicians.

Methods This article uses an interdisciplinary approach to explore ethical issues surrounding privacy, confidentiality of genetic data, and access to genetic results by health care providers and family members, and provides suggestions in a stakeholder format for best practices on this topic for clinicians and informaticians. Suggestions are made for clinicians on documenting and accessing genetic information in the EHR, and on collaborating with genetics specialists and disclosure of genetic results to families. Additional considerations for families including ethics around results of adolescents and special scenarios for blended families and foster minors are also provided. Finally, administrators and informaticians are provided best practices on both institutional processes and EHR architecture, including security and access control, with emphasis on the minimum necessary paradigm and parent/patient engagement and control of the use and disclosure of data.

Conclusion The authors hope that these best practices energize specialty societies to craft practice guidelines on genetic information management in the EHR with interdisciplinary input that addresses all stakeholder needs.

Keywords
- ethics
- privacy
- confidentiality
- children
- genetic testing
- electronic health record

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Background and Significance

Genetic Testing in Pediatric Populations

Children with medical complexity have significant health care needs and functional impairments, and utilize health care services to greater extent than other pediatric populations.[1] These children often undergo genetic testing for diagnostic pursuit, understanding disease progression, and to guide care.[2] Within the last decade, advances in molecular technology, improved access,[3] and lower costs[4] have made genetic testing an important diagnostic and prognostic tool to improve quality of life of affected children and their families. Practice guidelines from the American College of Medical Genetics and Genomics (ACMG) consider chromosomal microarray the first line test for postnatal testing of patients with multiple anomalies not specific to a known genetic syndrome, nonsyndromic developmental delay/intellectual disability disorders, and autism spectrum disorder,[5] while next generation sequencing looks at a panel of multiple genes for a specific clinical finding such as epilepsy, myopathy, hearing loss, or cancer.[6] Newer technologies like whole exome sequencing (WES) or whole genome sequencing can help identify mutations in a gene responsible for a clinical finding or phenotype, and can contain dozens of gigabytes of information.

An abnormal genetic finding in a child often requires testing of both biological parents to identify the source of the finding, also known as familial testing, and to determine if the finding is pathogenic, benign, or uncertain and if it is inherited or a *de novo* change. Genetic testing in the pediatric population is usually done in collaboration with a genetics clinic with appropriate pre- and post-testing informed consent and genetic counseling to identify the best management approaches for the patient. When the results are received, they are included in the child's medical record, and if familial testing is done, the parental results are also often incorporated into the child's chart.

Genetic test results can have implications for a patient (and potentially their family) for years to come, and it is the responsibility of the clinician to provide patient education and to obtain informed consent regarding genetic testing.[7] Equally important is the post-testing education that assists families with interpretation of test results and explanation of implications, as well as identifying next steps. The American Society for Human Genetics (ASHG) recommends genetic testing in children should include a long-term communication plan for all results as the pediatric patient transitions from childhood to adulthood.[8] While traditionally, pediatric health care included preconception care till 21 years of age, the latter is a "soft cutoff" and the American Academy of Pediatrics (AAP) recommends arbitrary age limits on pediatric care should not be established.[9] The Patient Protection and Affordable Care Act (PPACA) enacted in 2010[10] allowed young adults to stay on their parent's health insurance and foster youth to stay on Medicaid until the age of 26, suggesting the definition be expanded to 26 years; however, states ultimately determine Medicaid eligibility.[11]

Privacy Protections and the Electronic Health Record

The Health Insurance Portability and Accountability Act of 1996 (HIPAA)[12] has set standards to protect individuals' identifiable health information, called protected health information (PHI) under the privacy rule.[13] The HIPAA security rule specifically addresses the technical, administrative, and physical safeguards required to protect the confidentiality of health information.[14] The two HIPAA rules (privacy and security) aim to encourage health care systems to develop a health information system through the establishment of standards for the electronic transmission of certain health information.[15] The Health Information Technology for Economic and Clinical Health Act updated the privacy and security laws, and outlined conditions that do not require patient authorization including for treatment purposes, public health activities, insurance payment, administrative purposes, or performance improvement.[14] In general, patient information is confidential and protected but can be released to another party with the patient's (or parent/legal guardian's) permission or if mandated by law.[16]

There has been rapid growth in electronic health record (EHR) adoption across the United States,[17] and today, the EHR can be viewed by multiple providers and patients can also opt-in to view their own records. However, while the clinician or health care organization may own the EHR, the patient owns the information within their records. Parents of children with medical complexity may want to access and share their child's health data with third parties for clinical, administrative, and research purposes. Health care organizations may not have the policies or technologies in place to easily share the information or may be reluctant to provide the health care information.[14] Security concerns with breaches of health systems compromising patient records have occurred,[18] further eroding the confidence patients and families have in the process.

Collaborating with the AAP and other stakeholders, the Office of the National Coordinator for Health IT (ONC) recently published 10 recommendations that are specific to the medical needs of children,[19] including access to maternal history with inheritable genetic conditions relevant to the newborn's health care record, and identification of children with special health care needs. An increase in the pediatric health care provider workforce may reveal gaps in the recommendations, which will lead to a need for additional changes or modifications to the ONC-developed pediatric clinical priorities certification criteria, as well as expansion of the functionality and tools used.[20]

Security procedures to protect privacy with PHI in EHRs include designating user access based on practice roles (staff support has different privileges compared with physicians and nurses), requiring users to use passwords with specific requirements, lapse periods, and time out requirements per log in; and using additional measures such as two-tier authentication, and badge or biometric access methods among others.[16] This is bolstered by mandatory training for staff and education on HIPAA rules.[16] Audit trails allow organizations to monitor access, to flag suspicious activity, to

serve as a deterrent to breaches of PHI, and to determine if the breach is from external or internal sources.[21]

Electronic Health Record, Privacy, and Genetic Results

Despite guidelines from ACMG on reporting results,[22] there remain barriers to doing so effectively in EHRs. The ASHG has recommended that the standards should be developed for permanent storage of genetic data in EHRs.[8] The size of some genetic tests alone could slow the EHR and pose difficulties in interpretation, diagnosis, and re-evaluation,[23] and existing EHRs do not have the tools or storage capacity needed to annotate, mine, and analyze genomic information, particularly over a patient's lifespan.[24] This is particularly relevant as WES testing may show large numbers of variants of unknown significance,[25–27] which over time with the growing amount of data collected, may be reclassified as "likely pathogenic" or "likely benign."

Interfaces between EHRs and outside laboratory systems (which process the genetic testing) are also variable and cumbersome; some results flow from the laboratory into the EHR via the Laboratory 2.5.1 message standard,[28] though many still send results across as digital documents downloaded from a web portal or worse yet, paper results faxed, and scanned into the record. This leads to the potential for human error (scanning into the wrong chart and incomplete scanning) and poor quality reports (fax and scan) that can make scanned data illegible, leading to errors during data transfer. Free text or direct data entry requires the clinician to enter discrete data values into drop-down lists, checkboxes or dialog boxes, or text boxes, which can also lead to errors.[29] Even with direct interfaces, there can be error reports or error queues generated, which must be reviewed by dedicated health information management (HIM) resources so that the errors do not perpetuate.[29] Additionally, if a system receives results from multiple laboratory entities, each can have their own reporting format that can cause interoperability issues when consolidating data into the EHR.[30] In 2019, Health Level 7 updated the Fast Healthcare Interoperability Resources (FHIR 4) standard that reduces technology barriers to patient information from any data source including EHRs. One of the challenges of FHIR is the bidirectional sharing of information, for example, the app can retrieve data from the EHR to the user, but few—if any—EHRs are able to receive information from the app.

Additionally, genetic data are stored/displayed in various locations in the EHR[31] from the laboratory results tab to clinician notes. Sometimes, they are displayed as a scanned document attached to an order, or stand-alone as a "media" document, which may or may not be tagged for identification. Even when genetic data are not in a scanned document, reports often contain free text fields rather than discrete data elements, making extraction and analysis for clinical and research purposes cumbersome. Often, genetic tests results can be entered as a "miscellaneous test" without notation that it is a genetic result.

An additional concern relates to the timing and disclosure of genetic results; when families can access test results via the patient portal and other treating clinicians with EHR access can look at results and disclose them to patients, prior to geneticist interpretation or genetic counseling. This is particularly challenging when results impact other family members' insurability or employment[32] as well as possibly resulting in discrimination,[33] despite the protections offered by the Genetic Information Nondiscrimination Act (GINA) of 2008[34] and PPACA.[35] The consequences of genetic data misuse can be far reaching, and protections around genetic information fail to provide adequate control to individuals over disclosures that may affect them.[36]

Ethical Considerations

Currently, primary care providers view their role as making appropriate referrals for genetic medicine,[37] but may have limitations of knowledge, skills, time, and confidence about genetic testing.[38] Such limitations about a condition or a test result may engender stigma and discrimination,[8] which in turn may lead to unjust allocation of resources[39] and potentially cause harm (see ►Table 1 for examples). An argument can be made to deter or limit provider access to such sensitive information, except to the few with the knowledge and expertise to use it in the best interests of the patient/family. Stigma from genetic etiology can be felt by families, but specifically in the context of a patient (or family's) lived experience with a particular condition[40] and is not universal.

Patients (and in the case of minors, their parents/caregivers) have a moral right to privacy, and it is the health care system's duty to uphold that right. Precautions needed to uphold these rights increase with both the sensitivity of the information as well as the degree to which it identifies the patient. Genetic information is both sensitive and identifying, and the health care provider has a responsibility to protect this information. There have been opposing views on how to

Table 1 Examples of potential harm caused by inadvertent or premature release of genetic result information

Population affected	Genetic information released or interpreted	Potential harm
Patient and family members	A single variant finding in an autosomal recessive disorder communicated as disease causing variant instead of carrier status (e.g., long-chain L-3 hydroxyacyl-CoA dehydrogenase deficiency)	Undue stress or treatment
PCP and family	Misinterpretation of a prenatal test (e.g., diagnosis of aneuploidy made on fetal DNA screening test)	May recommend pregnancy discontinuation
Adolescent and family	Disclosure of parental findings of adult onset disorders (including of parents), particularly if a life-limiting disease (e.g., Huntington's disease)	Stress and family dynamics

Abbreviation: PCP, primary care provider.

consider genetic information: one was that genetic information differed from other kinds of health information, coined "genetic exceptionalism"[41]. Recently, the view of "genomic contextualism"[42] argues that genetic/genomic tests are similar in some ways to other medical tests, but have some distinct characteristics that require a careful approach to the data with determination of the ethical issues and policy considerations.

The adolescent population is worthy of special consideration as adolescents may have the decisional capacity (by ~12 years age)[43] to be entitled to confidentiality equal that of an adult with respect to some health information (such as mental health, substance use or abuse, sex, and reproduction). The absence of such an entitlement would discourage adolescents from pursuing treatment and undermine the adolescent–physician relationship, as adolescents value engagement and confidentiality in the relationship.[44] Because of the sensitivity of and potential stigma associated with genetic information, it is reasonable to protect genetic information in the same way as information regarding these other categories.[45] Furthermore, maintaining confidentiality puts the adolescent patient in a position of control over the information and makes them central to decision-making,[46] which can help to promote the development of autonomy and strengthen the adolescent—physician relationship.

There are some circumstances in which an adolescent ought to share control over their genetic information to a greater degree; including the adolescent with intellectual disability who is unlikely to obtain the full legal authority to make their own medical decisions, and whose parents are anticipated to maintain guardianship upon the patient turning 18 years. But as long as an adolescent is anticipated to obtain the ability and the legal authority to make their own medical decisions, confidentiality should be maintained.

While a patient's right to privacy can be waived for specific purposes, a patient cannot waive *another* person's right to privacy. In the case of children, parents should be informed that disclosure of their child's genetic information may imply that their own PHI may be disclosed and a thorough informed consent is necessary for this purpose, with disclosure of when, how and to whom genetic information is disclosed.

Best Practices for Managing Genetic Information in the Electronic Health Record

Given the above background, the interdisciplinary team of authors (geneticist, informatician, ethicist, and pediatrician) provide recommendations below for professionals who are faced with managing and disclosing genetic data of children (and families) in the EHR in a stakeholder framework (families, clinicians, informaticians, and administrators). The themes coalesce around storage and display of genetic data in the EHR, and access to, and release of that data. These are not intended to serve as practice guidelines, which should be framed by the appropriate specialty societies, preferably with interdisciplinary input that addresses all stakeholder needs.[47]

Patients and Families

Patients and caregivers should be made aware of the vagaries of the EHR and genetic test reporting. Depending on the institution, if a parent's genetic test results are added to a child's record in the EHR, a child could potentially access their parent's genetic test results. This might constitute a barrier as parents would not consent to testing if they do not want their child to have access to their genetic test results. Parents should not have to cede their right to privacy with regards to genetic testing results to assist in the medical treatment of their child. Adolescents can decide whether to permit or disallow their parents as their representative or proxy (including deciding who can view their patient portal which includes diagnoses, test results, and treatment). Alternatively, the creation of separate portals for the adolescent and their parents to access different PHI can provide confidentiality to adolescents.[48]

In a scenario where a child's parents are divorced and the child's stepmother now has access to the genetic test results of the child's biological mother from the child's chart, one likely solution is to only permit disclosure of genetic information to nonbiological parents with the consent of both biological (even if one is not custodial) parents. The risk is that one parent might not consent, and this may compromise the patient's interests. In other words, when values conflict (e.g., parent's privacy vs. child's interests), parents are allowed a great deal of latitude in determining how to balance them. For foster children, an argument could be made that the parent of a child placed in the foster system (whether due to negligence or intention) may forfeit a right to privacy to the extent that that right conflicts with the health and welfare interests of the child. What this demonstrates is that the current EHR mechanisms are not conducive to all biopsychosocial frameworks. We recommend that in challenging scenarios, clinicians and administrators seek guidance from their institutional or regional ethicists or ethics board.

Genetic information should be treated similarly to information about sexually transmitted infections or mental health conditions, and have additional security protections. Release of information documents that permit sharing between different entities signed by the patients (or in the case of children, parents) need to have explicit opt-in or opt-out provisions to avoid inadvertent disclosure consequences. Opt-in implies that information cannot be shared or be available until the patient (or proxy) grants permission, while opt-out implies that the specific PHI is automatically added to the released information and the patient must specifically request that their requested data not be released. Critics of opt-out cite concerns of the violations of informed consent and confidentiality.[49] Ultimately, an institutional process must be developed that balances the pros and cons of each form of consent, taking into account the needs of the patient and the feasibility of execution.

Clinicians

Clinician education around genetics should encompass several topics including communicating genetic information and facilitating informed decision-making by patients.[50] It

is important that this education also include how to document, protect, and disclose genetic information in the EHR and how to collaborate with genetic specialists. Clinicians should be educated to avoid "copy and paste" or "forwarding" of notes with genetic information or genetic laboratories. Even if citing the original author/source, copied text may have inaccuracies[29] (especially when presenting copy number variants) that may lead to misinformation. While clinicians find this a useful shortcut and are unlikely to abandon it completely, it is helpful to highlight copy-paste content and use direct smart links from the results (though this requires discrete data input via the interface in the first place), and degradation of the links and test result amendments can affect these.

A recent analysis[51] found that patients expect transparency and access to their health care provider's communications and explanations or clarifications about potentially stressful (or serious) information, something that is relevant to genetic test results. A concern is that if caregivers were aware that others in the family might be able to access information, they would be more inclined to withhold information, thus limiting information sharing.[51] Additionally, as more patients access patient portals to review test results, there is a need to make results available in a timely fashion to them, to permit shared decision-making, and to improve quality of care.[52] However, this should be balanced against the appropriate timing to release genetic results (vs. withhold), to permit disclosure and explanation.[53] This is particularly pertinent as patients' health literacy and specifically "genetic literacy" may be barriers to interpreting the genetic information without counseling and guidance.[54] It may be appropriate for nongenetic clinicians to defer genetic result disclosure to the geneticist/counselor, who have the knowledge and skill to accompany disclosure with guidance and counseling, particularly since reclassification of results with increasing knowledge makes it imperative to maintain contact and provide updated guidance to the child/family over time.[55] Another reason to defer is to prevent premature disclosure is that it can subsequently lead to lay literature/internet searches, yielding information that may not be applicable to a particular patient and potential cause for anxiety and harm as well.

Informaticians and Administrators

The sharing of genomic information in pediatric care can be perceived as a big data problem, characterized by volume, velocity, and variety.[56,57] Since raw genetic data can range up to more than 100 GB, there are questions regarding what and how much of the data to store. There are moves to store raw data external to the EHR, for example, in the "cloud"[58] with only the variants or reported data used clinically in the native EHR (similar to radiology systems). This would allow for reporting, reanalysis opportunities, and longitudinal monitoring,[59] all which are helpful for clinical decision-making. Genetic sequencing requires faster computing speeds and the variety of data sources such as genomic and proteomic data through sequencing technologies further complicate the situation. While big data can be helpful in clinical and research applications, infrastructure must be bolstered to support these activities and regulations needed to protect the PHI generated as a result.[60]

Informaticians would need to collaborate with laboratory vendors with a view to interoperability and adoption of the FHIR 4 standard. Many laboratories do not use the format, mainly because laboratory information systems (LIS) were not designed to facilitate the use of discrete genetic data in the first place. The cost and administrative burden to implementation may also be holding back smaller laboratories from adoption. Efforts from the HL7 Genomics Working Group are leading the way by incorporating parts of Substitutable Medical Applications and Reusable Technologies (SMART) on FHIR genomics into an observation profile.[61] Other strategies include transfer of genetic data either through the LIS, or through an ancillary system like the Genomics Archive Computer/Communication System or bypassing the LIS and directly transferring information to the EHR.[59] Recently, groups have looked at providing "point-of-care" guidance to clinicians by integrating a genetic information repository with the EHR, though this only involved a few users and a few genetic variants.[62]

Challenges remain, including the breadth of genomic data variability and the need to have implementation guidelines and a resource standard to support integration efforts. However, organizations must weigh the risk-benefit of limiting access to only structured data versus any data even if "locked" in unstructured format when it comes to clinical decision-making. Another option is the Logical Observation Identifiers Names and Codes (LOINC) which has been adopted by the HL7 Clinical Genomics Working Group, and provides codes to improve structured discrete reporting of specific genetic results.[63] LOINC allows for the standardization for categorizing and coding laboratory tests and datasets, and can distinguish laboratory data from different systems to improve interoperability of test results, reduction of duplicate tests ordered, and improved confidentiality since codes are used rather than test name.[64] However, the LOINC database does not include rare genetic conditions seen in the pediatric populations, newer molecular tests may not have a code in the current LOINC system, and not all laboratories uniformly use the same codes.[65] Any changes to the LOINC code system can create challenges to EHR systems with errors in mapping to unrelated LOINC codes, either due to lack of awareness of the specific test or LOINC name, the granularity of mapping, or human error.[64,65]

The debate about opt-in versus opt-out for genetic records release comes down to a balance between security and feasibility. While it is thought that opt-out would reduce administrative burden and improve information access which can help with care delivery, the opt-in format ensures patient engagement and control of how their data are handled. Ultimately, administrators and HIM professionals should consider these aspects when constructing (or revising) their institutional policies around genetic data and the EHR, including developing forms appropriately structured for informed consent for the patient/proxy and policies to ensure HIM staff reviews and adheres to the specific policies. Ideal discussions around this

should include various stakeholders including patient advisory board members, clinicians, administrators, HIM professionals, and ethicists.

Often, role-based access control (RBAC) is determined by the organization and is not varied at the individual patient chart level; only when clinical users' roles change does access change. One solution involves discretionary access control where the patient (or proxy) specifies who can access the record, and specifies the access level of the users in terms of a sensitivity label of the data (e.g., genetic, sexual, and mental health content can be placed under higher sensitivity).[66] Finally, purpose-based access control would grant access to the information based on the "purpose" for which the provider needs access, generally limited to treatment purposes.[66] For systems, however, there are technical challenges of implementing and administering these access controls while balancing the need for clinicians to access the chart for treatment purposes and to avoid delays. Some solutions that can be done at the informational technology/architectural level include creation of variable settings within the EHR that limit or permit the release of information to specific groups of users[48]. "Disclosure filters" at the individual level can allow the individual to consent to the nature and extent of use of data for research purposes.[67]

Some strategies to limit access to genetic information in the EHR have been to make the clinician notes unavailable or flagged as confidential, which would require a specific level of access to view. This has previously been done with mental health or substance abuse treatment records. Two such methods include "break the glass" and "hard stops." "Break the glass" (derived from step you need to take to access a fire alarm) RBAC[68] is a method of EHR security to restrict, but not block, disclosure of PHI by warning clinicians that the information is private. However, in circumstances where users fear blocking EHR access would adversely impact patient care, this system can be bypassed by "breaking the glass," usually by putting in a password and indicating reason for access. Protocols usually exist for audits to confirm if access was appropriate or not. Instances of inappropriate access can result in reprimand or termination of employment.[14,32] Even more stringent are "hard stops" (inability to proceed with the task) that totally blocks access with no workaround. These are generally used in order entry systems to avoid errors, but can limits clinicians in their ability to provide treatment, enroll patients in clinical trials, and may result in unnecessary health care costs due to repeat testing.[32]

To protect patient privacy, workarounds include blocking result reporting in the EHR, but this can lead to negative consequences of miscommunication, lost records, lack of access to results, and reporting errors.[32] Other workarounds may include using pseudonyms to document the encounter, or completely bypass reporting the details of genetic testing and test result within the EHR and instead document the results in a paper format.[33] However, this runs the risk of duplication, lack of access, and the need to maintain a separate secure environment for these records,[32] as well as legal and institutional risks to the clinician.[33]

Access to genetic information should, as a default, be conservative to have the "minimum necessary" paradigm, highlighting the privacy rule to safeguard and limit PHI use to accomplish the intended purpose.[13] In regards to genetic testing, this may be only the ordering physician and the geneticist or genetic counselor who can convert the information into actionable strategies in the best interest of the patient. At the same time, access control should not be a barrier to treatment of the patient and family.

Conclusion

The ubiquity both of genetic testing and the EHR poses unique technical, legal, and ethical challenges for health care providers, systems, and patients. In pediatric health care, these issues are often further complicated due to the uniqueness of having caregivers with legal access, the complexity of adolescent rights, and the lack of clarity around laws and processes to handle the sensitive nature of genetic data in this population. An interdisciplinary, multiple stakeholder approach will be needed for systems and clinicians to be oriented to the issue, provide the necessary education, and training to clinicians and staff handling genetic data and to develop the necessary procedural and EHR architectural structure to manage genetic information, including the storage, release, and access and release thereof. Systems and clinicians should ensure that patients (and caregivers of minors) have an expectation of privacy of their genetic data, and retain the right to control access and release, and this right must be balanced with the need to deliver care with minimal barriers to access. The opinions provided in this document can serve as best practices on which professional organizations and institutions can build further guidelines, ideally with an interprofessional team involving clinicians, informaticians, ethicists, and administrators.

Clinical Relevance Statement

Methods of genetic resulting in the EHR pose challenges to disclosure while protecting the privacy of families of children and adolescents. An interdisciplinary approach involving institutional and family stakeholders is recommended to balance the needs to access the information for clinical decision-making with the rights of the family to control the access. Best practices for clinicians and informaticians can implement strategies at an institutional and EHR level that can aid in this balance with the principles of minimally necessary release and family engagement when it comes to genetic records.

Multiple Choice Questions

1. The method of electronic health record (EHR) security to restrict, but not block, disclosure of genetic test results by warning clinicians that the information is private, and that in emergencies can be bypassed is known as:

a. Hard stop
b. Break the glass

c. Opt-out health information exchange
d. Two-tier authentication

Correct Answer: The correct answer is option b. "Break the glass" role-based access control is a method of EHR security to restrict, but not block, disclosure of private health information by warning clinicians that the information is private. However, in urgent situations or in circumstances where physicians fear restricting EHR access would adversely impact patient care, this system can be bypassed.

"Hard stops" block access to PHI completely. "Opt-in or opt-out" health information exchange option is a method that patients can decide how their information is shared. "Two-tier authentication" is a password level of protection for the user.

2. A child's school has obtained parent consent and requested the child's medical record, for the purposes of qualifying for special education services. The record has diagnoses and clinicians notes, but also includes results from a recent genetic test of both the child and parents, and a note from the genetic counselor explaining the risks to the child and to mother, from whom the finding was inherited. The appropriate manner in which to release the records is which of the following?

a. Release the entire record, including the genetic results and counselor notes
b. Release the entire record, with authorization from one parent
c. Release the record except results of the genetic test and counseling notes unless authorized by both parents
d. Withhold the entire record

Correct Answer: The correct answer is option c. Results from the genetic tests of pediatric patients should be, at a minimum, behind glass. This implies that they ought to be withheld unless the person accessing the record has a legitimate medical use for them. Specifics of genetic results are unlikely to be of value for special education purposes and even less so information about the parents, which are included in the record. Hence, since releasing a child's genetic test results also releases the health information about the parents, authorization from both parents is required.

It is not necessary to withhold the entire record which might delay services for the child, given that authorization was obtained, but maintaining the "minimum necessary doctrine" is applicable for genetic test results.

3. Best practices for clinicians' documentation and disclosure of genetic results with families includes:

a. Copy and paste results from the laboratory tab into their progress note
b. Defer genetic result disclosure to a geneticist/genetic counselor
c. Implement an opt-out policy for genetic record disclosure
d. Exclude genetic information from the EHR and maintain a paper record separately

Correct Answer: The correct answer is option b. It may be appropriate for nongenetic clinicians to defer genetic result disclosure to families to the geneticist/counselor, who have the knowledge and skill to accompany disclosure with guidance and counseling as a result can lead to psychological and emotional stress for a patient and their family members. Copy and paste or forwarding of genetic results from laboratories may have (and perpetuate) inaccuracies. While it is thought that opt-out would reduce administrative burden, the opt-in format ensures patient engagement and control of how their data are handled (since the data ultimately belongs to the patient/family). Workarounds such as documenting results outside the EHR is duplicative, and may engender legal and institutional risks to the clinician plus the need to maintain separate security procedures for the paper records.

Protection of Human and Animal Subjects

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