Received: 24 August 2020 | Revised: 8 July 2020 | Accepted: 17 July 2021

DOI: 10.1002/jgc4.1491

National Society of Genetic Counselors | WILEY

PRACTICE RESOURCE

Genetic counseling clinical documentation: Practice Resource of the National Society of Genetic Counselors

Katherine Hunt Brendish[1] | Devanshi Patel[2] | Kristen Yu[3] | Chelsea K. Alexander[4] | Jennifer Lemons[5] | Andrew Gunter[6] | Erin P. Carmany[7]

[1]College of Health Solutions, Arizona State University, Phoenix, AZ, USA

[2]Center for Cancer Risk Assessment, Mass General Hospital, Boston, MA, USA

[3]Neaman Center for Personalized Medicine, NorthShore University HealthSystem, Evanston, IL, USA

[4]InformedDNA, St. Petersburg, FL, USA

[5]Department of Pediatrics, The University of Texas Health Science, Houston, TX, USA

[6]Medical Affairs-HEOR, Secaucus, NJ, USA

[7]Wayne State University School of Medicine, Detroit, MI, USA

Correspondence
Katherine Hunt Brendish, College of Health Solutions, Arizona State University, Phoenix, Arizona, USA.
Email: khuntbrendish@asu.edu

Abstract

Clinical documentation is an important extension of a genetic counseling encounter. The traditional types of clinical documentation include the clinical visit note (including follow-up visit note), letter to the referring physician, letter to the patient, and result summary to the patient and referring physician. Increasing patient volumes, new genetic counseling service delivery models, transition to electronic medical records (EMR), new specialty clinics in genetics, and advances in genetic testing technologies challenge the practice of writing multiple types of clinical documents. This practice resource (PR) seeks to provide best practices for U.S.-based genetic counselors to write efficient and comprehensive clinical documentation using a hybrid clinical document designed to facilitate communication between individual providers, providers, and patients/families, and providers and payers. The content of the hybrid clinical documentation will vary by genetic specialty but may include a summary of genetic services evaluation, genetic testing options and eligibility information, genetic test results, potential risks for genetic conditions, implications for family members, and medical management recommendations. An outline of a general hybrid document along with examples of hybrid clinic notes for three types of genetic counseling specialties is included in this document.

KEYWORDS

clinical documentation, electronic medical record (EMR), genetic counseling, genetics services, practice resource, service delivery models

1 | PURPOSE

This practice resource (PR) proposes a hybrid clinical document applicable to every genetic counseling specialty (prenatal, pediatric, cancer, etc.). The practice resource addresses clinical documentation practices for multiple genetic counseling service delivery models including the following in-person, group sessions, and telegenetics (phone and videoconferencing) sessions. The intended audience is U.S.-based genetic counselors given international differences in practice patterns and documentation regulations and requirements.

2 | METHODS

This practice resource represents the opinions of a group of Certified Genetic Counselors from a wide range of disciplines (both patient facing and non-patient facing). The opinions of this author group are based on combined clinical and professional experiences as well as reviews of English-language medical articles and other governmental and institutional policies that influence clinical documentation practices. The recommendations in this practice resource are not intended to supersede the legal or institutional guidelines issued by a genetic counselors' employer or the opinions of individual genetic counselors.

Rather, this document is intended to serve as a compilation of information to consider incorporating into a clinic note, based on the relevant service delivery model; institutional requirements and policies; and a genetic counselors' specialty, professional experience, and best judgment.

3 | BACKGROUND

Clinical documentation in the United States was first developed to record and track a patient's condition and to communicate providers' recommendations and actions to the rest of the healthcare team (Kuhn et al., 2015). Clinical notes became incorporated into medical records in the late 19th century and included sections for family history, patient habits, previous illnesses, present illness, physical examination, urine and blood analyses, and progress notes and discharge diagnoses and instructions (Gillum, 2013).

The content of clinical documentation notes was further standardized by Henry Plummer in 1907 into the following categories: chief complaint, objective symptoms, subjective symptoms, and diagnosis (Gillum, 2013). Since the early 1900s, there have been additional requirements of clinical documentation including the use of documentation to: justify billing policies and practices for reimbursement, help mitigate legal liability by providing evidence for medical–legal cases, and gather information for developing measures for quality and regulatory purposes (Penoyer et al., 2014). In the late 20th century, clinical documentation notes became incorporated into one medical record with the intended use for direct patient care in the hospital and/or clinic/outpatient settings (Gillum, 2013). Most recently, the transition to an electronic medical record (EMR) has helped centralize the medical record even more but has also introduced new challenges and complexities to the practice of clinical documentation.

While there is limited guidance in the medical literature on the specific type of information to include in a clinical note, the rational for creating proper medical documentation is universally agreed upon: to ensure the best possible care for the individual and family, document the events of an inpatient or outpatient visit, and facilitate communication among healthcare providers (Doyle, 2009). Medical records today include multiple types of clinical documentation including the following: inpatient hospital and outpatient clinic notes, as well as a chart note from an initial visit along with a letter to the referring physician (Doyle, 2009). The purpose of genetic counseling clinical documentation is the same as documentation in general medical practice and similarly encompasses a chart note (structured in a similar format to notes in general medical practice) and a letter to the referring physician. What distinguishes genetic counseling documentation from other types of medical documentation practices is the use of a patient/family letter which summarizes the consultation in a manner which is understandable to the patient and serves as an education and communication tool for the patient and their family.

3.1 | Existing literature on genetic counseling clinical documentation

In order to develop the best practices for genetic counseling clinical documentation presented in this practice resource, the author group conducted a literature review in order to better understand the rationale for genetic counseling clinical documentation in the United States as well as the evolution of this practice over time. In the following paragraphs, we have briefly summarized the main types of clinical documentation traditionally utilized by genetic counselors: the clinical visit note (i.e., chart/EMR/progress note), the letter to the referring physician, and the letter to the patient/family.

There is no literature, however, addressing what content to include for each genetic counseling specialty, how to modify clinical notes depending on service delivery models, or how to write the chart note in the EMR environment while preserving the fundamental goals of clinical documentation notes.

3.1.1 | Clinical visit note (Chart/EMR/Progress note)

A chapter on medical documentation recommendations for genetic counselors was written in 2009 by Debra Lochner Doyle in *A Guide to Genetic Counseling* (Doyle, 2009). Doyle recommends that the information included in the clinical visit note, for both initial and follow-up visits, supports the scope, complexity, and time spent with the patient. Additionally, genetic counselors who bill for their services need to include information within the clinical visit note to support the Current Procedural Terminology (CPT®) billing codes.

3.1.2 | Letter to referring physician

The purpose of the letter to the referring physician is to facilitate communication between providers. The letter to the referring provider can also serve as an educational tool (similar to the clinical visit note), as it generally incorporates information about the diagnosis, prognosis, pathology, recurrence risks, and possible treatment options for conditions with which the referring physician may be unfamiliar (Doyle, 2009).

As important as the letter to the referring physician can be, there is no standard for structuring these letters to best accomplish its goals. However, preliminary research has found that referring providers' preferred content that includes diagnosis, prognosis, and management plan, rather than data items such as detailed history (Rash et al., 2018). Furthermore, letters formatted as structured templates appear to increase comprehension and may even reduce letter length (Vermeir et al., 2015). Limiting the length of the letter is also important, as physicians prefer letters that are fewer than two typewritten pages, or less than 350 words (Scott et al., 2004; Selzer et al., 2009). Since many EMR systems now allow direct access to chart notes by various

providers within a healthcare system, genetic counselors may no longer send separate letters to referring physicians. However, the author group felt that it is important to recognize the historical significance and evolution of the practice of writing letters to referring physicians.

3.1.3 | Letter to patient/Family

The patient/family letter serves as an easily understood summary of the genetic counseling session for the patient, as well as a tool to help patients understand and share information with others. Baker et al., (2002) published recommendations for writing letters to patients, noting the two primary functions of the letter: recording relevant facts and presenting information in a manner that promotes the patient's understanding.

The recommendations from Baker et al., (2002) also emphasized the importance of using clear and concise language and considering how language will impact the patient, including the importance of value-free and person-first language. Patient letters help facilitate understanding and intention to share information with others (Hallowell & Murton, 1998). A more recent study suggested that patients prefer shorter letters highlighting the basic facts; such letters were associated with positive self-reported emotional reaction compared with longer letters (Roggenbuck et al., 2015).

In addition to the patient letter describe by Baker et al., (2002), genetic counselors may utilize separate letters intended specifically to facilitate the familial communication of relevant genetic diagnoses and genetic test results. This clinical documentation practice resource does not include recommendations related to this type of correspondence. We do acknowledge, however, that this is an important topic to be considered for future research into best practices for genetic counseling clinical documentation.

3.2 | Clinical documentation practices for other medical specialties

To better understand how other medical professionals have established standards for writing effective clinical documentation, a comprehensive search of literature published in the United States between 2009 and 2018 was conducted using the keywords: best practices in medical documentation, electronic health records, and genetic specialties. The search included physicians (all medical specialties) and advanced practice providers (physician assistants and nurse practitioners, nurses, and social workers). Forty articles were identified; however, none of the articles addressed the unique component of genetic counseling documentation. There were a handful of articles that provide guidance of how to incorporate clinical notes into the EMR while preserving the intent of the note. These articles are summarized in Appendix A.

4 | RATIONALE FOR WRITING A CLINICAL DOCUMENTATION PRACTICE RESOURCE

This practice resource was conceived to help genetic counselors identify best practices for writing clinical documentation that might maximize the benefits of the practice (such as increased patient understanding and facilitation of patient care) while creating minimum acceptable documentation standards that could reduce the time burden of clinical documentation for the practicing genetic counselor. It is hoped that this might allow genetic counselors to see the maximum number of patients possible without jeopardizing patient care or genetic counselor job satisfaction.

4.1 | Clinical documentation can be a time-consuming activity

Overall, studies have shown that many healthcare providers spend a large portion of their time on clinical documentation (Baumann et al., 2018). Recent literature has shown that genetic counselors are also spending a significant amount of their time on clinical documentation (Attard et al., 2019; Heald et al., 2013; VandenBoom et al., 2018). Since clinical documentation has been found to be a time-consuming activity, it follows that genetic counselors may consider ways to decrease the time burden of clinical documentation.

4.2 | Studies have shown that there may be redundancy within the types of documentation that genetic counselors produce

Clinical documentation serves a variety of purposes; many of them outlined in this document. Additionally, studies suggest that some genetic counselors may be producing more than one type of clinical documentation for each patient with each type serving an independent function. In one study, 24.8% of cancer genetic counselors reported that they would like to reduce redundancy with documentation and pedigrees to increase workplace efficiency (Wham et al., 2010). Additionally, VandenBoom et al., (2018) found that genetic counselors were averaging up to three types of different clinical documentation for each patient. Generating multiple documents per patient could be one factor adversely affecting the efficiency of genetic counselor practice. Ideally, one clinical document that meets a minimum criterion for quality patient care and serves multiple purposes would increase genetic counselor efficiency without sacrificing patient care.

4.3 | Documentation practices lack consistency between different genetic counseling specialties

There is a small body of evidence regarding the clinical documentation practices within different genetic counseling specialties.

Overall, there seem to be differences not only between specialties but also within the same specialty. VandenBoom et al., (2018) found that pediatric and cancer genetic counselors were more likely to send clinical documentation to patients specifically using a patient-friendly format. In contrast, prenatal genetic counselors tended to write documents only intended for providers. Even within the same specialty, Heald et al., (2016) found that clinical practice can vary greatly, leading to different documentation practices. While there will always be inherent differences in genetic counseling practice both within and between specialties, some elements of genetic counseling are fundamentally the same across the profession. The definition of genetic counseling published by the NSGC identifies common elements that unite the profession (Resta et al., 2006). At the most fundamental level, genetic counselors interpret personal and family histories to establish risk, educate individuals about the various facets of genetic conditions, and provide counseling in order to promote informed choices (Resta et al., 2006). Therefore, professional best practices should exist to ensure that these fundamental elements are documented appropriately, regardless of clinical specialty and circumstance.

4.4 | There may be different documentation needs for different service delivery models

Recommendations for the clinical content to include for a traditional in-person genetic counseling session have been described by Doyle (2009). However, service delivery models for genetic counseling have evolved. For example, many genetic counselors primarily counsel patients over the telephone. Other genetic counselors use videoconferencing to conduct genetic counseling sessions. The literature review did not identify specific clinical documentation guidelines for alternative service delivery models including telegenetics and group counseling sessions. Because the number of genetic counselors utilizing alternative service delivery models will continue to increase over time, the author group felt it would be important to begin to address the clinical documentation needs for these types of genetic counseling sessions.

4.5 | Health information technology

Documentation for medical services, influenced by the advancement of EMR, and legislative initiatives, such as the Health Information Technology for Economic and Clinical Health (HITECH) Act, have impacted and shifted the ways in which medical documentation is completed, as well as the necessary components (Blumenthal, 2011). The HITECH Act was a part of the American Recovery and Reinvestment Act of 2009, which supported the transition to EMR and other technology. The U.S. government set aside additional Medicare and Medicaid payments for healthcare professionals and institutions that engaged in meaningful use, incentivized the use of EMRs, and required all public and private healthcare providers to adopt an EMR system by January 1, 2014 (Kuhn et al., 2015).

Components of this legislation are directly applicable to genetic counseling documentation. 'Meaningful use criteria' require that, after each visit, the patient receives a clinical summary of relevant information and laboratory/diagnostic tests ordered (Center for Medicaid & Medicare Services, 2014). Structured family history documentation for first-degree relatives in a patient's record is a stage 2 meaningful use objective; however, there is limited specific guidance about how to appropriately document family history/pedigree, though it is noted that it should be captured in either SNOMED CT or HL7 Pedigree standard (HL7 International, n.d.).

Additionally, many EMR systems now have patient portals in place, which allow patients to receive and interact with their own medical records. A recent literature review on the effectiveness of patient portal use in the inpatient setting reports that patient utilization of a portal resulted in improved patient–provider communication, patient safety, and adherence to medications (Dendere et al., 2019). The fact that more patients have access to their own health information through portals linked to an EMR is an important change to the process of how genetic counselors communicate with their patients and should be considered in clinical documentation practices.

5 | OTHER TOPICS TO CONSIDER FOR CLINICAL DOCUMENTATION

5.1 | Billing codes and documentation requirements

There are many different ways in which to bill/code for genetic counseling services, and genetic counseling-specific codes have been developed. Genetic counselors should be aware that institutional billing practices have implications for documentation as there may be different requirements for authorship, attestations for notes/letters, and general documentation requirements beyond those specific to genetic counseling needs. (Gustafson et al., 2011; Harrison et al., 2010). As an example, the CPT® code 96,040 specific to genetic counseling services by nonphysician providers was established by the American Medical Association (AMA) in 2007 and requires genetic counselor authorship for notes and has requirements for content (e.g., family history, medical risk assessment, genetic education, and psychosocial assessment) (Gustafson et al., 2011). Other methods of billing, such as Evaluation and Management Codes (E/M) codes, have general documentation requirements established by CMS (Center for Medicaid & Medicare Services, 1997). Additionally, different service delivery models may require different billing/coding requirements and different clinical documentation.

It is important to note that this author group is not making recommendations about billing practices. We recommend genetic counselors review and comply with their institutional policies and procedures regarding billing practices.

5.2 | Considerations for Insurance

Clinician documentation is a vital aspect of prior authorization and payment for medical genetics services, including genetic counseling and any study/assessment/testing that is coordinated or ordered by a genetic counselor. While no commercial or federal payer was identified that outlines what should be included in the documentation of a genetic counseling session, providers should familiarize themselves with the expectations of the payers with whom they work, as insurance requirements may be updated frequently. Important information to document for billing for the genetic counseling visit includes the name of the referring physician and the length of time of the consult.

There are some additional recommendations genetic counselors should keep in mind regarding insurance payments for billable services, particularly as it pertains to documentation. Genetic counselor documentation should support the medical necessity of a service by explaining what is being ordered (e.g., the name of the test and ordering code of the test used by laboratory and the name of the testing laboratory), as well as rational for why a specific test is being ordered. When a multi-gene panel test is order, consider listing the genes in the panel (only if possible) or stating the number of genes in the panel for larger panel tests which can be helpful to know whether test is negative and larger gene panels become available. Additional information that is important to include for insurance coverage includes the clinical rationale driving that assessment and plan, and the ways in which medical management will be altered based on the results of the service. Not doing so increases the chance that a payer may deny a request for coverage. If possible, consider including reference(s) or links to the relevant practice guidelines that state genetic testing is recommended and/or statement that genetic testing was recommended by additional providers.

5.3 | Privacy requirements/legislation

5.3.1 | Heath Insurance Portability and Accountability Act

The Health Insurance Portability and Accountability Act (HIPAA) of 1996 was established, in part, to create confidentiality systems to be used by healthcare facilities to keep protected health information (PHI) private. PHI is defined as any information that links a specific patient to healthcare information. Examples of PHI include, but are not limited to, a patient's name, telephone number, email address, street address, and social security number (Edemekong & Haydel, 2018).

To implement the requirement of protection for PHI, the Department of Health and Human Services (HHS) issued a Privacy Rule and a Security Rule. The Privacy Rule ensures adequate protection of PHI while allowing the flow of information needed to provide high-quality health care. The Security Rule sets national standards for protecting the confidentiality, integrity, and availability of electronic information (U.S. Department of Health & Human Services, 2017).

It is the opinion of the author group that genetic counselors should review their institutional policies and procedures regarding privacy regulations. It will also be important for the genetic counselor to review the institutional policies and procedures surrounding documentation and storage of family information in the clinical document. The institution's privacy office and the office of general counsel may also serve as resources.

A review of English-language medical articles and other governmental and institutional policies revealed no requirements for the documentation of HIPAA laws in clinical documentation.

5.3.2 | Genetic Information Non-discrimination Act

Privacy concerns surrounding genetic information are often associated with the fear of genetic discrimination. Therefore, while it is not a required element of genetic counseling clinical documentation, some genetic counseling practices and institutions will summarize the Genetic Information Non-discrimination Act (GINA) and the benefits and limitations of this law in their clinical documentation.

5.3.3 | Mandated reporting (if applicable)

While mandated reporting is not a typical component of the clinic note, the author group felt that due to the importance of this topic, it was necessary to describe the purpose and instances for which mandated reporting might need to be included in genetic counseling clinical documentation. Mandated reporting occurs when there is suspicion that a vulnerable person is or may be abused. A review of English-language medical articles and other governmental and institutional policies revealed no standard process for the documentation of mandated reporting in a clinic note or medical chart. However, the American Academy of Pediatrics has suggestions on documentation that can be helpful for later investigation of suspicious injuries, including but not limited to: standard medical and developmental history, family history (especially bleeding disorders, bone disorders, metabolic disorders, and genetic disorders), pregnancy history (wanted/unwanted, planned/unplanned, prenatal care, postnatal complications, postpartum depression, delivery in non-hospital setting, child temperament, substance abuse in family living at home), and mental health issues among family living at home (Christian et al.,l., 2015).

It is the opinion of this author group that genetic counselors should consult their department head and the Health Information Management department at their institution to determine best documentation practices for mandated reporting. It is important genetic counselors understand the necessary components of past and current medical history to document in the event of a potential lawsuit in a situation of suspected abuse or other medical–legal scenarios.

For additional information about state laws regarding mandated reporting policies, please refer to this Web site: https://www.childwelfare.gov/topics/systemwide/laws-policies/statutes/manda/

6 | JUSTIFICATION OF RECOMMENDATIONS FOR CLINICAL DOCUMENTATION

We recommend one clinic note (a hybrid note) which replaces the need to write multiple different types of clinic notes yet incorporates all of the relevant information necessary from each of the three traditional clinical documentation types. While many institutions have already shifted to a hybrid note format, there were no articles identified in the literature which summarized the transition to a hybrid note in the field of genetic counseling. The remainder of this document will outline the necessary elements to consider incorporating into a hybrid note and provide suggestions for how to format such a note to decrease time spent on clinical documentation while maintaining an effective communication tool for our patients and the healthcare providers involved in our patient care.

The primary goal of creating a hybrid clinical documentation note is to decrease the amount of time genetic counselors spend writing clinical documents by eliminating the need for multiple documentation types while maintaining or enhancing patient comprehension and utility. The proposed hybrid clinical documentation note is intended to be used as a chart note for the EMR as well as a letter to both the referring physician and the patient. Therefore, a hybrid note should include the core content outlined by Deborah Lochner Doyle in her chapter on medical documentation in *A Guide to Genetic Counseling* (2009) and be written in language that is understandable to the patient (Baker et al., 2002).

The transition to EMRs over the years has increased patient access to their medical information. With the use of patient portals becoming more common at health facilities, patients will have access to their medical information shortly after their genetic counseling visit, making it possible to eliminate the need for a genetic counselor to send a separate patient letter. If written in a manner that is understandable to the patient, the proposed hybrid note would serve as a summary of the visit, a resource for where to locate genetic test results in the EMR, and an education tool for the patient.

In addition to including the necessary clinical content for a referring physician letter and patient letter, the hybrid note includes general documentation requirements for genetic counseling billing and prior authorization. This information is not meant to supersede institutional billing practices. The intent is to educate on the content that may be necessary for some billing practices and prior authorization protocols. When the required information for billing and prior authorization is included in the hybrid note, the note can be used to justify billing codes and serve as a prior authorization request for certain types of genetic testing.

A final goal of this PR is to provide genetic counselors with a summary of the type of content to include in clinical documentation notes for different genetic counseling specialties and service delivery models. While each genetic counselor will follow his/her/their own practice and institutional guidelines for clinical documentation, the hybrid note we propose can perhaps be utilized as a template for notes across different types of genetic counseling sessions and service delivery models, thereby improving consistency in documentation throughout the various specialties.

7 | RECOMMENDATIONS FOR HYBRID CLINICAL DOCUMENTATION

7.1 | Clinical content of hybrid document

The content for the hybrid note outline (See Table 1) and example hybrid specialized notes (See Appendix B) were developed by the author group through an iterative process that incorporated best practices of genetic counseling clinical documentation identified in the literature with the professional experiences and expertise of the authors. While we reached consensus on the structure and order of the outline for these hybrid notes, we also appreciate the fact that there are many variations to clinical practices, institutional policies, and personal preferences that may result in subtle variations in the practice of clinical documentation for genetic counselors. It is important to note that these recommendations are not meant to replace institutional requirements for clinical documentation.

We recommend that the hybrid document continues to address the two primary functions of the traditional 'patient' letter as outlined by Baker et al., (2002):

1. Record relevant facts (which may include history of presenting illness/interim history, family history updates, psychosocial/social history, results of physical examination and review of systems, genetic test results, assessment of diagnosis, plan, information or counseling provided, and recommendations for medical management, treatment, and referrals which are also described as clinical note content summarized by Deborah Lochner Doyle in her chapter on medical documentation in a *Guide to Genetic Counseling* (Doyle, 2009).
2. Present information in a manner that promotes the patient's understanding using clear, concise, and non-stigmatizing language.

We recommend the initial clinic visit hybrid note focus on the most pertinent information and patient recommendations, as patients prefer shorter documentation and many have access to additional information on the Internet (Roggenbuck et al., 2015). Additional reputable materials, such as patient education sheets or brochures developed by genetic counselors for their institution, can be used to supplement the note and when provided to the patient, should be referenced in the hybrid document. Consider including links to local and national support groups as well as links to Web sites that provide information about genetic disorders to patients such as Genetics Home Reference (https://medlineplus.gov/genetics/).

TABLE 1 Example hybrid note outline

***This example includes proposed general components. The content and order of content listed below can be modified according to author preference and/or clinical indication and type of consult*. See Appendix B, for examples, of hybrid notes based on specialization.**

1. Patient Identifiers
   a. Patient Name
   b. DOB
   c. MRN
2. DOV
3. Referring Provider Name
4. If Applicable—Translator Services: If session was conducted with the assistance of an interpreter include the name and ID number of the interpreter. Note if the genetic counselor directly provided services in a language other than English.
5. Optional-Family/Friends Accompanying Patient to Visit: If appropriate and with patient permission, state whether relatives or friends attended the clinic visit with patient. For confidentiality purposes, consider using only initials to identify relatives and/or request permission to use name/initials in consult note. Only include initials for non-related individuals.
6. Indication/reason for referral and history of present illness (HPI) including relevant previous test results.
7. Summary of past histories relevant to the specialty, such as pregnancy, medical, developmental, social, and psychosocial.
   a. List relevant details with bullet points
8. Summary of family history/pedigree
   a. Reference reader to pedigree directly consider pasting directly into note or uploading to EMR
   b. Indicate whether a possible genetic condition (not associated with the current referral indication) is suspected as well
   c. Consider adding the source of the information obtained (by report or medical records)
   d. List relevant details with bullet points including ethnicity, presence/absence of consanguinity
9. Reference to findings of physical examination, if indicated
   a. Review of systems
   b. Diagnosis/differential diagnoses
10. Risk assessment
    a. Brief discussion if at risk for condition; more detailed if diagnosed (if multiple risks, consider using headings for increased readability)
    b. Risks, benefits, and limitations of all applicable options (genetic screening and diagnostic testing)
    c. Chance to test positive/negative/VUS
    d. Possible results and implications of results
    e. Additional information necessary for informed decision making
11. Genetic education (Consider using SmartTools/templates)
    a. Inheritance pattern
    b. Genes/chromosomes/description of variants
12. Natural history of condition (Consider using SmartTools/templates)
    a. Clinical features of syndrome/signs and symptoms (if applicable include penetrance, variable expressivity, anticipation, genotype–phenotype correlation, mosaicism)
    b. Prognosis (% risk for future symptoms, or associated diagnoses)
13. Discussion of recommendations for diagnostic work-up
    a. Additional evaluations necessary for establishing a diagnosis
14. Discussion of medical management guidelines
15. Recurrence Risk
    a. Risk for family members based on diagnosis or test results
    b. Summary of future reproductive options/prenatal diagnosis options
16. Psychosocial issues and concerns (the following topics can be discussed)
    a. Discussion with family members
    b. Anticipatory guidance (e.g., possibility of an uncertain finding and the need to re-contact the genetics clinic for updates on finding and/or advancement in knowledge about a genetic disorder)
    c. Support systems and coping mechanisms
    d. Mandated Reporting (if applicable)
    e. GINA and other privacy statutes-other insurance implications of genetic test results
    f. Financial/health insurance considerations (coverage of genetic testing)
17. Summary of support groups, support services, and description of educational resources and patient literature provided. Consider inserting appropriate links to patient-friendly Web sites.
18. Summary of session/recommendations: (This section could also be incorporated at the beginning of the document. Refer to Cancer Hybrid Note for an example.)
    a. ***#1. Reason for referral***-Example: Mr. Jones/Mrs. Hernandez/Mrs. Anderson was referred because of…
    b. ***#2. Risk assessment***-Example: Based on her medical history and physical exam it is possible he/she has -----condition

Example: Based on her age, her risk for a chromosomal abnormality is---

Example: Based on her personal and family history of---her risk for a possible hereditary cancer syndrome is ---

    c. ***#3. Recommendations***-Example: In order to confirm this diagnosis, the recommendations are---
    d. ***#4. Decision and Pre-authorization information (if applicable)***- Example: After considering all options, Mrs. Hernandez elected to proceed with expanded carrier screening using a 100-condition panel.

(Continues)

TABLE 1 (Continued)

Example: Mrs. Anderson elected to proceed with a multi-cancer gene panel. Based on the NCCN guidelines, Mrs. Anderson meets the clinical criteria required for coverage of an updated multi-gene panel test. Her results could affect her clinical management recommendations.

e. ***#5. Follow-up plan***- Example: Results are expected in XX weeks and a follow-up appointment has been scheduled on XX/XX/XX in order to review the results.

19. Name of Genetic Counselor
20. Telephone Number for Genetic Counselor
21. Email Address of Genetic Counselor
22. Time Spent in Direct Patient Care
23. Time Spent in Indirect Patient Care

The clinical content of the hybrid note for established patients should contain the relevant information from the initial visit along with the appropriate interim history and evaluation. Whenever possible, the document should refer the reader to the initial clinic visit hybrid documentation note for all other information.

Promoting patient understanding of written information can be difficult to achieve especially when dealing with complex medical and genetic information, numerical risk assessments, and communicating uncertainty. However, given that approximately 1/3 of the U.S. population has limited health literacy skills, using best practices in written health communication is vital (Kutner et al., 2006). The Plain Language Action and Information Network first developed the Federal Guidelines for Plain Language in the 1990s to establish best practices for effectively communicating information to the general public in writing. It includes valuable recommendations on organization for reader needs, paragraph and sentence structure, appropriate word choice to reduce medical jargon and use simpler terms, avoiding passive voice, and many others (Plain Language Action & Information Network, 2011). Additionally, the Agency for Healthcare Research and Quality more recently developed the Health Literacy Precautions Toolkit to improve patient health literacy in a variety of areas including written communication. It includes specific information regarding writing at an appropriate reading level for patients (5th- to 6th-grade levels in most cases), effectively communicating numbers in writing, and how to assess writing to improve patient understanding (Brega et al., 2015).

7.2 | Family history

We recommend that the hybrid document for the initial visit includes the family history/pedigree. A summary of only relevant family history is recommended in order to keep the note as concise as possible. No identifying information belonging to a family member should be included without appropriate consent of the family member in question. Additionally, genetic counselors should consider including the following information along with the family history summary:

1. Reference reader to the pedigree directly and consider uploading to the EMR or pasting the pedigree into the EMR.
2. Indication whether a possible genetic condition (not associated with current referral indication) is suspected as well.
3. The source of the information obtained (by report or medical records).
4. Ethnicity, especially if this information is relevant for risk assessment and/or genetic testing interpretation.
5. Presence/absence of consanguinity.

Any relevant updates to the family history should be documented in subsequent follow-up visit notes for established patients.

7.3 | Format of hybrid document

Genetic counseling clinical documentation has traditionally been formatted the same way a letter is written, with paragraphs summarizing family and medical history information. With the migration toward EMRs, we anticipate that most institutions will utilize templates or SmartTools (including SmartTexts, SmartPhrases, SmartLinks, and SmartLists) for all types of clinical documentation. Hybrid notes may be formatted with a combination of SmartTexts (templates and blocks of texts), lists, and bullet points. For example, the family history summary could list relevant details with bullet points instead of using full sentences and this format could be applied to other sections in the hybrid note such as indication/reason for referral and summary of present illness. When appropriate, use bolded or underlined text within the hybrid note to help differentiate new topics. An example of a hybrid clinic note written with a combination of full paragraphs and bullet points is provided in the appendix.

7.4 | Length of hybrid note

We recommend concise documentation, as previously recommended for patient letters (Baker et al., 2002; Vandenboom et al., 2018) and shown to be preferred by patients (Roggenbuck et al., 2015). While 1.5–2 pages have been proposed as a guideline for patient letters, the patient's situation and/or specialty-specific nuances will impact the length. For example, the length of the hybrid notes we provided in the appendix varies in length by specialty and none achieved the 1.5–2 pages preferred length. Since the proposed hybrid note includes information for both patients and providers, it may not be possible to achieve the 1.5–2 page length. However, because the literature does suggest that patients prefer more concise written information, as mentioned above, one strategy could be to consider eliminating or reducing the amount of information provided in the

genetic education section, in favor of separate educational brochures and/or links to informational Web sites.

7.5 | Service delivery models

7.5.1 | Group sessions

In addition to the general requirements of the hybrid document, it should be specifically noted if part of the session is conducted in a group setting. Pertinent information discussed in the hybrid note outline that was covered in a group session should be summarized in the clinical documentation of a group session.

7.5.2 | Telegenetics (telephone and videoconferencing)

Telegenetics consult notes follow the same general requirements of the hybrid document. Information specific to a telegenetics consult note includes a statement that telegenetics was utilized for the patient session as well as documentation of how the patient identity was confirmed (e.g., DOB). If the patient visit was intended to be an in-person visit but the service delivery model shifted due to issues such as the COVID-19 pandemic, it should be noted that the session was conducted via video due to circumstances that prevented an in-person visit from occurring. An example of language to use in this unique scenario is as follows: '*A virtual visit was used during the COVID-19 crisis in place of an in-person visit. This real-time interactive virtual clinical encounter was conducted using telephone-only technology from clinic or home office. Consent for virtual care, including informing the patient that insurance will be billed, and that in-person care is available in case of emergencies or as needed otherwise, was discussed at the time of scheduling*'.

7.6 | Result disclosure

All result disclosures to patients should be documented in the permanent patient record regardless of the manner of disclosure (in-person, by telephone, etc.). As institutional requirements and clinical circumstances may vary, this can be done in a variety of ways, including documentation embedded into the result in the EMR, a separate progress or follow-up note, or as an addendum to an initial clinic visit. As with other clinic notes, it is important to note the method of communication with the patient in the result disclosure section (i.e., in-person or telephone call).

The content of the result disclosure encounter will vary by specialty but in general should include the following: the name of the laboratory, date of report, specific test performed (if feasible, this could include a list of genes in a panel or at a minimum, stating the number of genes for larger panel tests); a summary of the genetic test result; an explanation of the clinical implication of the genetic test results (including a statement advising not to use a VUS result when making medical management decisions, if indicated); discussion about the implications of the result for family members; clinical management discussion including follow-up plan and referrals to physicians for medical management; recommendations for additional genetic testing (including testing other family members) if applicable; information about DNA banking (if relevant); and the clinic/institutional protocol for re-contacting the patient (or protocol for the patient to re-contact the clinic themselves) with updates on the classification of a result (which can occur with a VUS finding).

In an effort to improve access to genetic test results, it is recommended that when documenting the result disclosure session, a sentence be incorporated into this note that references where the genetic test results can be found within the EMR. In some situations, it may be possible to take screenshot of the report and embed it directly into the result disclosure note. It is also important to document how the patient was provided a copy of the test results (e.g., through the patient portal or sent a hard copy of the results through the mail or emailed results).

7.7 | Follow-up communication encounters (including E-communications)

Follow-up communication occurs in a variety of formats (in-person, telephone, videoconference, emails, or patient portals) and can serve a variety of different purposes (providing updated or new information, reviewing test results, etc.). For example, a common follow-up communication is to document test results and subsequent plan. In documenting follow-up communication, it is important to clarify the mode and purpose of communication as well as a summary of the information discussed, including any recommendations for future communication with the clinic to obtain updates on genetic testing options or learn new information about a genetic diagnosis/treatment options as well as the plan (e.g., follow-up visit, referrals, and additional testing). Follow-up communication can be documented in a variety of ways including the following: a separate progress or follow-up note or as an addendum to an initial clinic visit or return visit clinic note. The protocol for this type of documentation may be determined by institutional requirements.

Individuals should defer to their institutional policies with respect to allowed methods of communication (e.g., email) for this type of communication.

7.8 | Additional patient-related work

All patient-related work should be documented in the permanent patient record per institutional requirements. This includes but is not limited to: follow-up patient documentation for variant reclassification (most commonly for a VUS finding), communications with other experts, communications with genetic testing laboratories, etc. Just as with follow-up communication notes, any relevant information

surrounding patient care could be documented as a separate progress or follow-up note or as an addendum to an initial clinic visit or return visit note. Again, the protocol for this type of documentation may be determined by institutional requirements.

8 | LIMITATIONS OF THIS CLINICAL DOCUMENTATION PRACTICE RESOURCE

There are several limitations to this clinical documentation practice resource. While the recommendation for a hybrid clinical documentation note is based on the experiences of multiple genetic counselors in diverse settings as well as supporting literature, there have been no studies to validate the effectiveness of these specific recommendations. Future research evaluating the effectiveness of genetic counseling hybrid clinical documentation notes from both patient and provider perspectives is recommended.

Additionally, given the nuances of different specialties and patient needs, the authors recognize the difficulty of applying one set of recommendations to every genetic counseling encounter. Institutional legal policies, practice groups' preferences, billing policies, and medical record systems may limit applicability of these recommendations. We recommend genetic counselors review their institutions' guidelines on medical and legal accountability regarding medical documentation. Finally, we acknowledge that the landscape for medical record documentation is evolving and additional updates will likely be needed in the future.

9 | FUTURE RESEARCH CONSIDERATIONS

While this PR is intended to provide guidance on the current practice of clinical documentation in genetic counseling, we recognize that there is limited research in this area overall. Therefore, in addition to providing recommendations based on what is known currently, this document also serves as a call for more research in this area. Research is needed on the scope and differences between clinical documentation practices across the genetic counseling profession and to identify potential influential factors such as specialty, institution, and EMR use. Additionally, more research is needed on innovative approaches to genetic counseling clinical documentation (including the creation of SmartTools) that will both increase efficiency and serve the documentation needs of both patients and providers. We encourage genetic counselors work with their institution's EMR builder to write these tools in order to optimize the use of the tools to meet the needs of genetic counseling clinical documentation as described in this practice recommendation. Further research into the most effective format for genetic counseling clinical documentation is necessary as well as how and whether to include patient letters in the EMR for the purpose of sharing genetic test results with family members and how and whether to include psychosocial information in clinical documentation notes. Finally, as is true in other areas of genetic counseling, more research is needed on the impact of clinical documentation practices on patient-related outcomes.

This PR is an important starting point in addressing clinical documentation practice in genetic counseling. However, with additional research in this area, we will hopefully achieve the ultimate goal of establishing evidence-based clinical practice guidelines for clinical documentation.

10 | CONCLUSIONS

The purpose of this PR is to provide recommendations for how to respond to the challenges of traditional clinical documentation practice with the expansion and increased specialization of the profession, the integration of the EMR and patient portals into patient care, and increasing patient volumes. This PR was based on literature review and expert opinion but should not replace individual genetic counselor clinical judgment or specific institutional policies and guidelines.

DISCLAIMER

This practice resource (PR) is provided by the National Society of Genetic Counselors (NSGC) solely to serve as a helpful practice management resource and tool for genetic counselors and other healthcare providers. NSGC's PRs are not based on a systematic evidence review; instead, they are based on the recommendations and experience of the authors.

Each NSGC PR focuses on a clinical or practice-based issue, includes points for the genetic counselor or other healthcare providers to consider, and is based on review and analysis of current professional literature that the authors believe to be reliable. As such, the information provided and ideas discussed in NSGC's PRs; (i) reflect only the current scientific and clinical knowledge at the time of publication; (ii) are only current as of their publication date; and (iii) are subject to change without notice as advances emerge.

PRs do not (and are not intended to) dictate an exclusive course of management, nor guarantee a particular outcome. NSGC's PRs are never intended to displace a genetic counselor's or other healthcare provider's best medical judgment based on the clinical circumstances of a particular patient or patient population. NSGC publishes PRs for educational and informational purposes only, and neither 'approves' nor 'endorses' any specific methods, practices, or sources of information contained therein.

AUTHOR CONTRIBUTIONS

Katherine Hunt Brendish and Erin P. Carmany agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved. Katherine Hunt Brendish, Erin P. Carmany, Devanshi Patel, Kristen Yu, Chelsea K. Alexander, Jennifer Lemons, and Andrew Gunter provided substantial contributions to the conception and design of the work; drafting, and revising it critically for important intellectual content; and final approval of the version to be published.

ACKNOWLEDGEMENTS

The authors would like to acknowledge Blair Stevens, MS, CGC, for her assistance with reviewing the prenatal genetic counseling note.

COMPLIANCE WITH ETHICAL STANDARDS

CONFLICT OF INTEREST

The author group composition is in compliance with the National Society of Genetic Counselors Practice Guidelines Committee Conflict of Interest Policy. This policy requires all proposed authors to disclose conflict of interest prior to selection and imposes thresholds for conflict of interest with the potential for direct, personal financial benefit, or other real or perceived conflict of interest, through the development of the document. Katherine Hunt Brendish, Devanshi Patel, Kristen Yu, Chelsea K. Alexander, Jennifer Lemons, Andrew Gunter, and Erin P. Carmany declare that they have no conflict of interest during the development of this practice resource.

HUMAN STUDIES AND INFORMED CONSENT

No human studies were carried out by the authors for this article.

ANIMAL STUDIES

No animal studies were carried out by the authors for this article.

DATA SHARING AND DATA ACCESSIBILITY

Data sharing not applicable to this article as no datasets were generated or analyzed during the current study.

ORCID

Katherine Hunt Brendish
https://orcid.org/0000-0002-9666-1334

REFERENCES

Attard, C., Carmany, E., & Trepanier, A. (2019). Genetic counselor workflow study: The times are they a-changin'? *Journal of Genetic Counseling*, *28*, 130–140. https://doi.org/10.1002/jgc4.1041

Baker, D. L., Eash, T., Schuette, J. L., & Uhlmann, W. R. (2002). Guidelines for writing letters to patients. *Journal of Genetic Counseling*, *11*(5), 399–418. https://doi.org/10.1023/A:1016841731426

Baumann, L. A., Baker, J., & Elshaug, A. G. (2018). The impact of electronic health record systems on clinical documentation times: A systematic review. *Health Policy*, *122*(8), 827–836. https://doi.org/10.1016/j.healthpol.2018.05.014

Blumenthal, D. (2011). Wiring the health system—origins and provisions of a new federal program. *New England Journal of Medicine*, *365*, 2323–2329. https://doi.org/10.1056/NEJMsr1110507

Brega, A. G., Barnard, J., Mabachi, N. M., Weiss, B. D., DeWalt, D. A., Brach, C., Cifuentes, M., Albright, K., & West, D. R. (2015). *AHRQ Health Literacy Universal Precautions Toolkit, Second Edition*. Agency for Healthcare Research and Quality. Retrieved from https://www.ahrq.gov/sites/default/files/publications/files/healthlittoolkit2_3.pdf

Center for Medicaid and Medicare Services (1997). *Documentation guidelines for evaluation and management (E/M) services*. Retrieved from https://www.cms.gov/Outreach-and-Education/Medicare-Learning-Network-MLN/MLNEdWebGuide/Downloads/97Docguidelines.pdf

Center for Medicaid and Medicare Services (May 2014). *Eligible professional meaningful use core measures measure 12 to 13*. Retrieved from https://www.cms.gov/regulations-and-guidance/legislation/ehrincentiveprograms/downloads/13_clinical_summaries.pdf

Christian, C. W., & Committee on Child Abuse and Neglect. (2015). The evaluation of suspected child physical abuse. *Pediatrics*, *135*(5), e1337–1354. https://doi.org/10.1542/peds.2015-0356

Dendere, R., Slade, C., Burton-Jones, A., Sullivan, C., Staib, A., & Janda, M. (2019). Patient portals facilitating engagement with inpatient electronic medical Records: A systematic review. *Journal of Medical Internet Research*, *21*(4), e12779. https://doi.org/10.2196/12779

Doyle, D. L. (2009). Medical Documentation. In W. R. Uhlmann, J. L. Schuette, & B. M. Yashar (Eds.), *A Guide to Genetic Counseling* (pp. 313–329). John Wiley & Sons Inc.

Edemekong, P. F., & Haydel, M. J. (October 27, 2018). Health Insurance Portability and Accountability Act (HIPAA). Retrieved from https://www.ncbi.nlm.nih.gov/books/NBK500019/

Gillum, R. F. (2013). From Papyrus to the Electronic Tablet: A Brief History of the Clinical Medical Record with Lessons for the Digital Age. *The American Journal of Medicine*, *126*(10), 853–857. https://doi.org/10.1016/j.amjmed.2013.03.024

Gustafson, S. L., Pfeiffer, G., & Eng, C. (2011). A large health system's approach to utilization of the genetic counselor CPT® 96040 code. *Genetics in Medicine*, *13*(12), 1011.

Hallowell, N., & Murton, F. (1998). The value of written summaries of genetic consultations. *Patient Education and Counseling*, *35*(1), 27–34. https://doi.org/10.1016/S0738-3991(98)00080-9

Hansen, A. G., Martin, E., Jones, B. L., & Pomeroy, E. C. (2015). Social work assessment notes: A comprehensive outcomes - based hospice documentation system. *Health & Social Work.*, *40*(3), 191–200. https://doi.org/10.1093/hsw/hlv033

Harrison, T. A., Doyle, D. L., McGowan, C., Cohen, L., Repass, E., Pfau, R. B., & Brown, T. (2010). Billing for medical genetics and genetic counseling services: A national survey. *Journal of Genetic Counseling*, *19*(1), 38–43. https://doi.org/10.1007/s10897-009-9249-5

Heald, B., Gustafson, S., Mester, J., Arscott, P., Lynch, K., Moline, J., & Eng, C. (2013). A time study of cancer genetic counselors using a genetic counselor-only patient care model versus a traditional combined genetic counselor plus medical geneticist care model. *Journal of the National Comprehensive Cancer Network*, *11*(9), 1076–1081. https://doi.org/10.6004/jnccn.2013.0129

Heald, B., Rybicki, L., Clements, D., Marquard, J., Mester, J., Noss, R., Nardini, M., Polk, J., Psensky, B., Rigelsky, C., Schreiber, A., Shealy, A., Smith, M., & Eng, C. (2016). Assessment of clinical workload for general and specialty genetic counsellors at an academic medical center: A tool for evaluating genetic counselling practices. *NPJ Genomic Medicine*, *11*(1), 16010. https://doi.org/10.1038/npjgenmed.2016.10

HL7 International (n.d.). Retrieved from http://www.hl7.org/implement/standards/product_brief.cfm?product_id=301

Kuhn, T., Basch, P., Barr, M., & Yackel, T. (2015). Clinical documentation in the 21st Century: Executive summary of a policy position paper from the American College of Physicians. *Annals of Internal Medicine*, *162*, 301–303. https://doi.org/10.7326/M14-2128

Kutner, M., Greenberg, E., Jin, Y., & Paulsen, C. (2006). *The Health Literacy of America's Adults: Results From the 2003 National Assessment of Adult Literacy (NCES 2006–483)*. U.S. Department of Education. National Center for Education Statistics. https://nces.ed.gov/pubs2006/2006483.pdf

Leisner, B. A., & Wonch, D. E. (2006). How documentation outcomes guide the way: A patient health education electronic medical record experience in a large health care network. *Quality Management in Healthcare*, *15*(3), 171–183. https://doi.org/10.1097/00019514-200607000-00007

Meyers, S., Claire Simon, K., Bergman-Bock, S., Campanella, F., Marcus, R., Mark, A., Freedom, T., Rubin, S., Semenov, I., Lai, R., Hillman,

L., Tideman, S., Pham, A., Frigerio, R., & Maraganore, D. M. (2018). Structured clinical documentation to improve quality and support practice-based research in headache. *Headache*, *58*(8), 1211–1218. https://doi.org/10.1111/head.13348

Narayanan, J., Dobrin, S., Choi, J., Rubin, S., Pham, A., Patel, V., R., Maurer, D., Gupta, P., Link, L., Walters, S., Wang, C., Ji, Y., & Maraganore, D. M. (2017). Structured clinical documentation in the electronic medical record to improve quality and to support practice-based research in epilepsy. *Epilepsia*, *58*(1), 68–76. https://doi.org/10.1111/epi.13607

Penoyer, D. A., Cortelyou-Ward, K. H., Noblin, A. M., Bullard, T., Talbert, S., Wilson, J., Schafhauser, B., & Briscoe, J. G. (2014). Use of electronic health record documentation by healthcare workers in an acute care hospital system. *Journal of Healthcare Management*, *59*(2), 130–144.

Plain Language Action and Information Network. (2011). *Federal Plain Language Guidelines*. Retrieved from https://www.plainlanguage.gov/guidelines/

Rash, A. H., Sheldon, R., Donald, M., Eronmwon, C., & Kuriachan, V. P. (2018). Valued components of a consultant letter from referring physicians' perspective: A systematic literature synthesis. *Journal of General Internal Medicine*, *33*(6), 948–954. https://doi.org/10.1007/s11606-018-4356-3

Resta, R., Biesecker, B. B., Bennett, R., Blum, S., Hahn, S. E., Strecker, M. N., & Williams, J. L. (2006). A new definition of genetic counseling: National Society of Genetic Counselors' Task Force Report. *Journal of Genetic Counseling*, *15*(2), 77–83. https://doi.org/10.1007/s10897-005-9014-3

Roggenbuck, J., Temme, R., Pond, D., Baker, J., Jarvis, K., Liu, M., Dugan, S., & Mendelsohn, N. J. (2015). The long and short of genetic counseling summary letters: A case-control study. *Journal of Genetic Counseling*, *24*, 645–653. https://doi.org/10.1007/s10897-014-9792-6

Scott, I., Mitchell, C., & Logan, E. (2004). Audit of consultant physicians' reply letters for referrals to clinics in a tertiary teaching hospital. *Internal Medicine Journal*, *34*, 31–37. https://doi.org/10.1111/j.1444-0903.2004.00424.x

Selzer, R., Foley, F., Ellen, S., & McGartland, M. (2009). What do GPs want from a psychiatrist's letter? *Australasian Psychiatry*, *17*, 385–388. https://doi.org/10.1080/10398560902971771

Simon, K. C., Hentati, A., Rubin, S., Franada, T., Maurer, D., Hillman, L., Tideman, S., Szela, M., Meyers, S., Frigerio, R., & Maraganore, D. M. (2018). Successful utilization of the EMR in a multiple sclerosis clinic to support quality improvement and research initiatives at the point of care. *Multiple Sclerosis Journal - Experimental, Translational, and Clinical*, *4*(4), 2055217318813736. https://doi.org/10.1177/2055217318813736

U.S. Department of Health & Human Services. (June 16, 2017). HIPAA for Professionals. Retrieved from https://www.hhs.gov/hipaa/for-professionals/index.html

VandenBoom, E., Trepanier, A. M., & Carmany, E. P. (2018). Assessment of current genetic counselor practices in post-visit written communications to patients. *Journal of Genetic Counseling*, *27*, 681–688. https://doi.org/10.1007/s10897-017-0163-y

Vermeir, P., Vandijck, D., Degroote, S., Peleman, R., Verhaeghe, R., Mortier, E., Hallaert, G., Van Daele, S., Buylaert, W., & Vogelaers, D. (2015). Communication in healthcare: A narrative review of the literature and practical recommendations. *International Journal of Clinical Practice*, *69*(11), 1257–1267. https://doi.org/10.1111/ijcp.12686

Wham, D., Vu, T., Chan-Smutko, G., Kobelka, C., Urbauer, D., & Heald, B. (2010). Assessment of clinical practices among cancer genetic counselors. *Familial Cancer*, *9*, 459–468. https://doi.org/10.1007/s10689-010-9326-9

How to cite this article: Hunt Brendish, K., Patel, D., Yu, K., Alexander, C. K., Lemons, J., Gunter, A., & Carmany, E. P. (2021). Genetic counseling clinical documentation: Practice resource of the national society of genetic counselors. *Journal of Genetic Counseling*, 30, 1336–1353. https://doi.org/10.1002/jgc4.1491

APPENDIX A

Article	Medical provider or healthcare professional performing clinical documentation	Clinical documentation challenge	Key recommendations applicable to clinical documentation practices for genetic counselors
Meyers, et al.; Structured Clinical Documentation to Improve Quality and Support Practice-Based Research in Headache; *Headache*; 2018	Physicians	The Department of Neurology at NorthShore University sought to create an in-house toolkit of customized templates (based on best practices) within the EMR to establish consistency among providers' documentation.	Templates were developed that included custom navigators (sidebar index of processes to choose from), electronic forms, best practice advisories, order sets (based on national and international guidelines for migraine diagnosis), and optional free-text fields.
Narayanan et al.; Structured clinical documentation in the electronic medical record to improve quality and to support practice-based research in epilepsy; *Epilepsia*; 2017	Physicians	How to utilize the EMR to capture structured clinical data and provide clinical decision support to promote quality improvement and practice-based research in epilepsy.	A structured clinical documentation support (SCDS) tool was built into the EMR that supports best practices in epilepsy. The SCDS tools write notes, capture data fields (including score tests; brain imaging; blood laboratory and electroencephalography results; and treatment outcomes). These tools are shared with other sites and used to conduct trails using the EMR.
Simon, et al.; Successful utilization of the EMR in a multiple sclerosis clinic to support quality improvement and research initiatives at the point of care; *Multiple Sclerosis Journal-Experimental, Translational and Clinical*; 2018	Physicians, Medical Assistants, and Nurses	Three multiple sclerosis specialists practicing at four outpatient centers associated with NorthShore University HealthSystem identified an opportunity to standardize care to best practices and use the EMR to facilitate quality improvement and practice-based research.	A structured clinical documentation support (SCDS) toolkit was developed (specific for multiple sclerosis) to document initial, interval, and annual follow-up encounters. The content of the toolkit included a sidebar index of processes to choose from (known as a navigator), electronic forms, and summary flowsheets.
Hansen et al.; Social Work Assessment Notes: A Comprehensive Outcomes-Based Hospice Documentation System; *Health & Social Work*; 2015	Social workers	The authors could not identify a social worker specific patient assessment tool in the literature that could be used for patients in the hospice setting.	Hansen et al. created a social work-specific psychosocial assessment tool called the *Social Work Assessment Notes*. This tool is intended to serve as a comprehensive hospice social work documentation system. The tool includes assessment, planning, and outcomes measurement for patients' and caregivers' needs related to end-of-life, a method to facilitate collaborative care plan development and to measure patient- and family-centered outcomes.

Article	Medical provider or healthcare professional performing clinical documentation	Clinical documentation challenge	Key recommendations applicable to clinical documentation practices for genetic counselors
Leisner & Wonch; How documentation outcomes guide the way: a patient health education electronic medical record experience in a large health care network; *Quality Management Health Care*; 2006	Various providers within a VA system, including nursing	Leisner & Wonch used focus groups, pilot testing, usage monitoring, local action planning, and feedback surveys to create a usable EMR-based method of documentation of patient health education.	Implementation of a new EMR documentation tool requires perseverance, collaboration, and continuous monitoring and development.
Penoyer et al.; Use of Electronic Health Record Documentation by Healthcare Workers in an Acute Care Hospital System; *Journal of Healthcare Management*; 2014	Physicians, nurse practitioners, physician assistants, nurses, managers, medical nurse assistants, social workers, and ancillary diagnostic personnel	Penoyer et al. sought to determine: What type of information in the EHR is used by clinicians, how this information is used for patient care and the amount of time clinicians review and document information in the HER. These data were collected in an effort to improve effectiveness and efficiency of documentation.	Recommendations: Administrators/leaders and EHR designers should add fields for narrative descriptions in documentation; create shared documentation between disciplines to reduce redundancy; and include the clinical practitioners in discussions about changes to the documentation system that will affect them.
Kuhn et al.; Clinical Documentation in the 21st Century: Executive Summary of a Policy Position Paper from the American College of Physicians; *Annals of Internal Medicine*; 2015	Physicians	Kuhn et al. wrote a position paper that reviews current and emerging purposes of clinical documentation, forces that may influence or distract from these purposes and challenges and opportunities afforded by EHRs.	The purpose of clinical documentation is to support patient care and improve clinical outcomes through enhanced communication; physicians, medical societies, and other should define professional standards for clinical documentation practices through their organizations.

APPENDIX B

Example of General Genetics Hybrid Note (This is intended to represent a Genetic Counseling only consult.)

Patient Name: Kayla Brown
DOB: 1/1/2017
MRN: 123,456
DOV: 8/18/2019
Referring Provider: David Jones, MD
Primary Care Provider: John Doe, MD
Indication/reason for referral:

- Two years old with a history of developmental delay and failure to thrive

SUMMARY OF PAST MEDICAL HISTORY/PREGNANCY, DEVELOPMENTAL AND SOCIAL HISTORY; RELEVANT PREVIOUS TEST RESULTS

- Mother's age at birth: 26 years old; father's age at birth: 32 years.
- G2P2001
- Pregnancy exposures: N/A.
- Pregnancy complications: First-trimester bleeding.
- Prenatal tests/labs/ultrasounds and results: Normal first-trimester screen. No additional prenatal testing.
- Delivery: 38 weeks 1 day; vaginal.
- Hospital/City/State of birth: Your City, USA.
- Birth parameters: weight/%tile: 10%; length/%tile: 12%
- Neonatal course: History of high bilirubin in the neonatal period, treated with phototherapy and discharged after 2 days. Patient was noted to have lost 10% of weight at the newborn follow-up and had difficulty feeding.
- List diagnosis history: Developmental delay, failure to thrive.
- List surgical history: None.
- List medications: None.

Developmental history

- Developmentally appropriate for age: No.
- Major milestones: Rolled 6 months, sat alone 9 months, crawled 15 months, cruising at 20 months, unable to walk unassisted. Not currently toilet training. Not fully transitioned to solid foods.
- Name/Type of school and classes or stay-at-home: Daycare 3 days a week, home with family members 2 days a week.
- Therapies (ST/OT/PT): Patient has been receiving PT/OT since age 9 months.

Social history

- Patient lives at home with both parents and a 6-year-old sister.

Relevant previous test results

- Myopia identified on eye exam [DATE])
- Negative Newborn screen [STATE], normal ALT/AST, negative lead testing [value/date].No previous genetic testing results.

SUMMARY OF RELEVANT FAMILY HISTORY/PEDIGREE

Maternal:

- No known family history of developmental delay, chromosomal or genetic condition; no personal or family history of multiple miscarriages.

Paternal:

- No known family history of developmental delay, chromosomal or genetic conditions; no family history of multiple miscarriages.

Ethnicity/Consanguinity:

- Maternal: African American
- Paternal: African American
- No consanguinity reported in maternal or paternal history

Please see the pedigree located under the 'family history tab' for additional details.

The information provided is based on the patient's recollection of the family history and in the absence of complete medical records. If the family history changes or if more information is obtained, the patient was asked to contact the genetics clinic as this may alter the recommendations or impression of the family history.

Physical exam

Per Kayla Brown's physical examination with Dr. Jones, it was found that her weight, height, and head circumference were below the 5% for her age. The remainder of the physical examination was unremarkable.

Review of systems

Per Kayla Brown's review of systems with Dr. Jones,

- EYES—**Reports:** Wears glasses for myopia
- List other positives by pulling from below paragraph and detailing here

Except as stated in the history of present illness, review of systems for GENERAL, EARS, NOSE/MOUTH/THROAT, HEMATOLOGY/LYMPHATIC, RESPIRATORY, CARDIOVASCULAR, GASTROINTESTINAL, MUSCULOSKELETAL, RENAL/URINARY, NEUROLOGICAL, ENDOCRINE, INTEGUMENT, ALLERGY/IMMUNOLOGY are negative.

Differential diagnoses

Based on Kayla's history of unexplained developmental delay, failure to thrive, and unremarkable family history, there is a high suspicion for a chromosome abnormality for the patient.

Risk assessment

The chance for any child with developmental delay to have a chromosome abnormality that can be identified by chromosome microarray (CMA) analysis is ~10%

Genetic education (Consider use of SmartTools/templates for this section)

Chromosomes are the structures that carry our genes and can be found in every cell of the body. Genes found inside our chromosomes contain the instructions (or blueprints) for our overall health. If a chromosome or section of a chromosome is missing, or duplicated, individuals could be affected with various health problems depending on which part of the chromosome is involved.

Chromosome microarray testing (CMA) is a genetic test which is able to determine whether someone was born with any chromosomes that are missing (either completely or partially) or duplicated (either completely or partially). While CMA testing is able to detect most chromosomal deletions and duplications, it cannot diagnose all genetic diseases.

Natural history of condition

N/A until results are available.

Discussion of diagnostic work-up/options for genetic testing

- As CMA is recommended as a first-tier evaluation for individuals with neurodevelopmental disability and congenital anomalies (Genetics in Medicine, 2013), testing is recommended for Kayla
- Additional testing may be recommended based on the results from the CMA analysis

Recurrence risk

Will be reviewed with family after results are available from CMA

Summary of session/recommendations

1. Kayla Brown was referred for a genetics visit by Dr. Jones due to her history of developmental delay and failure to thrive.
2. We reviewed the chance for any child with developmental delay to have a chromosome abnormality that would be identified by chromosomal microarray (CMA) is ~10%
3. After discussion of the benefits and limitations of genetic testing, Kayla's parents consented to have Kayla's blood drawn and sent to [Laboratory] for SNP-microarray testing with CPT codes (81,229). Based on Kayla's history of unexplained developmental delay and failure to thrive, there is a high suspicion for a chromosome abnormality for the patient. As CMA is recommended as a first-tier evaluation for individuals with neurodevelopmental disability and congenital anomalies (Genetics in Medicine, 2013), testing is recommended for Kayla as it may be able to provide an accurate diagnosis for the patient, ensure that she is seen for appropriate follow-up, and allow for accurate genetic counseling and risk assessment to be provided for her in the future as well as her family.
4. Recommendations/follow-up plan:
   a. The patient had her blood drawn for a SNP-microarray today, and results are expected in approximately 2–3 weeks. Specific recommendations for follow-up will be made at that time.

b. Continue to work with primary care physician on feeding and nutrition
c. Follow-up genetic visit is scheduled in 6 months
d. Follow-up genetic counseling once results are available.

Name of Genetic Counselor, MS, CGC
T: XXX-XXX-XXXX (direct & confidential)
Email:
Time Spent in Direct Patient Care:
Time Spent in Indirect Patient Care:

Example of Prenatal Hybrid Note

Patient Name: Maria Hernandez
DOB: 1/1/1980
MRN: 123456
DOV: 8/18/2019
Referring Provider: David Jones, MD

Indication/reason for referral:
Patient and partner both carriers for cystic fibrosis referred to discuss prenatal testing options.

- Summary of pregnancy/obstetrical and medical history and previous test results (if applicable):
- 25 yo female
- G1 P0 at 16 weeks' gestation
- No reported complications, medical conditions, or pregnancy exposures
- Patient found to be a carrier of the deltaF508 pathogenic variant in the CFTR gene. Expanded carrier screening panel for 100 other conditions was negative. A copy of the genetic test results can be found in EMR under 'laboratory results tab'
- Partner subsequently found to be a carrier of the deltaF508 pathogenic variant through sequencing of the CFTR gene.

Summary of relevant family history/pedigree

Maternal:
- No known family history of cystic fibrosis or related symptoms
- No additional family history relevant for the current pregnancy

Paternal:
- No known family history of cystic fibrosis or related symptoms
- Partner's uncle with schizophrenia

No consanguinity reported in maternal or paternal history

Ethnicity:
- Maternal: Hispanic, Mexican
- Paternal: Hispanic, Mexican

Please see the pedigree located in 'family history tab' for additional details.

The information provided is based on the patient's recollection of the family history and in the absence of complete medical records. If the family history changes or if more information is obtained, the patient was asked to contact the genetic clinic as this may alter the recommendations or impression of the family history.

Risk assessment for current pregnancy (combined with genetic education and natural history): (Consider use of SmartTools/templates for parts of this section and providing patient with education sheets or brochures and document that these education sheets or brochures were provided to the patient. In addition, consider including links to Web sites that provide information about genetic disorders to patients such as Genetics Home Reference (http://medlineplus.gov/genetics/))

Cystic fibrosis: Cystic fibrosis (CF) is one of the most common inherited genetic conditions in the general population. It is inherited as an autosomal recessive genetic condition. With this type of inheritance, a person affected with CF inherits two altered copies (pathogenic variants) of the gene that causes CF called CFTR, one from each parent. Parents of a child with CF are carriers. They have one altered copy and one functional copy of the CF gene and do not have any symptoms of cystic fibrosis. Since both partners are carriers of the deltaF508 pathogenic variant, *there is a 25% chance with each pregnancy that the child will have cystic fibrosis, a 50% chance that the child will be a carrier like the parents and a 25% chance that the child will have two normal CF genes and neither be affected nor a carrier.* Please see https://medlineplus.gov/genetics/condition/cystic-fibrosis/ for information additional information on cystic fibrosis.

Schizophrenia: Schizophrenia, like other psychiatric conditions, typically has multifactorial inheritance. That is, there are multiple genes that are inherited that interact with each other and unidentifiable environmental factors to produce the condition. Because they are partly caused by genes, these types of conditions may recur within a family. *The risk for the partner to develop schizophrenia may be approximately 3% due to having a second-degree relative. Overall, the risk to the pregnancy is likely not significantly increased over the general population risk of 1%.*

Options for genetic testing and/or screening with associated risks: We discussed the option of fetal CFTR testing for the deltaF508 pathogenic variant by amniocentesis. In addition, routine chromosome analysis would also be performed. We discussed the risks, benefits, and limitations of this testing including the 1/300 risk for miscarriage associated with the procedure.

We discussed the option of not having fetal testing performed for cystic fibrosis. In this case, all babies born in the United States undergo newborn screening for cystic fibrosis; therefore, if the baby has cystic fibrosis, it would likely be detected within a few weeks after birth.

As in any pregnancy, we recommended a level II ultrasound examination. Some pregnancies affected with cystic fibrosis can have echogenic bowel (intestines that look brighter than typical) seen on ultrasound. However, not all babies with cystic fibrosis have an echogenic bowel; therefore, a normal ultrasound examination would not rule out cystic fibrosis or guarantee a healthy baby.

Risk for family members: We recommended that Mrs. Hernandez and her partner inform their family members of their carrier status so these individuals can have carrier screening, should they so choose.

Psychological issues/concerns: We discussed the possible psychological implications of genetic testing, issues of family dynamics, and concerns regarding confidentiality, insurance coverage, and genetic discrimination.

Mandated reporting can be inserted here if applicable.

Decision about genetic testing: After considering the possible options, Mrs. Hernandez decided to not pursue an amniocentesis for cystic fibrosis genetic testing at this time.

Summary of support groups/educational resources/patient literature provided: Mr. and Mrs. Hernandez were offered patient brochures about cystic fibrosis but they declined receiving these resources until after the birth of their child.

Summary of session/recommendations:

1. Mrs. Hernandez was referred by Dr. Jones to discuss the risks for cystic fibrosis in the current pregnancy since she and her partner are both carriers for the deltaF508 pathogenic variant in the CFTR gene.
2. The chance for cystic fibrosis for this and any future pregnancy this couple has together is 25% with a 50% chance to have a child who is a carrier but unaffected.
3. Mrs. Hernandez and her partner chose not to have an amniocentesis for cystic fibrosis at this time.
4. Recommendations/follow-up plan:
   a. Level II ultrasound examination between 18 and 20 weeks' gestation
   b. Prenatal care per obstetrician recommendations
   c. Mrs. Hernandez should notify the pediatrician of the chances for cystic fibrosis in the baby to ensure close follow-up of newborn screening results and possible additional testing/evaluation for cystic fibrosis after birth.
   d. Mrs. Hernandez and her partner were encouraged to inform family members of their cystic fibrosis carrier status. We are happy to see any family members for genetic counseling and consideration of testing.
   e. Follow-up genetic counseling if there are any additional concerns.

Name of Genetic Counselor, MS, CGC
T: XXX-XXX-XXXX (direct & confidential)
Email:
Time Spent in Direct Patient Care: 45 min
Time Spent in Indirect Patient Care: 60 min

Example of Cancer Hybrid Note

Patient Name: Jane Anderson
DOB: 1/1/1963
MRN: 123,456
DOV: 8/18/2019
Referring Provider: David Jones, MD

Indication/reason for referral:

- Personal history of contralateral breast cancer
- Family history of breast and ovarian cancer
- Updated genetic testing

Summary of session/recommendations

1. Mrs. Anderson was referred by Dr. Jones for her personal history of bilateral breast cancer and her family history of breast and ovarian cancer.
2. Based on her personal history of bilateral breast cancer and family history of breast and ovarian cancer, her risk to test positive for a BRCA 1 or BRCA 2 pathogenic variant is XX% based on the Tyrer-Cuzick model (optional to use risk assessment calculations).
3. After discussing the risks, benefits, and limitations of additional genetic testing, Mrs. Anderson elected to proceed with (name of genetic test and number of genes in panel) with associated CPT code (XXXX) to be sent to (Laboratory). Based on the NCCN guidelines, Mrs. Anderson meets the clinical criteria required for coverage of an updated multi-gene panel test. Her results could affect her clinical management recommendations.
4. A follow-up appointment to receive the results has been scheduled for 9/1/2019. Medical management recommendations will be made based on the result of her genetic analysis.
5. Mrs. Andersons's family history was suggestive of a possible hereditary neurological syndrome, and therefore, we recommended she consider scheduling an appointment in the neurogenetic clinic for additional evaluation.

Summary of personal history of cancer and previous test results (if relevant)

- 62 yo female
- Stage II, IDC, age 38 (2001)
- Negative BRCA1/2 testing, age 38. Testing performed at Myriad Genetics Laboratory (2001) See 'laboratory results tab' for a copy of test results
- Contralateral DCIS, age 50

Summary of relevant family history/pedigree

Maternal

- Mother died at age 61 from ovarian cancer diagnosed at age 60. She was also diagnosed with breast cancer diagnosed at age 50.
- Aunt died at age 42 from breast cancer diagnosed at age 40. Through this relative:
- First cousin, age 55: ovarian cancer, age 53.
- Grandmother died at age 72: Died from complications of diabetes. Breast cancer, age 60.

Paternal

- There was no additional paternal family history specific to a hereditary cancer syndrome.

The family history was significant for her brother complaining of weakness and difficulty lifting the foot while walking as well as numbness with associated pain in his hands and feet. These findings are suggestive of a possible hereditary neurological syndrome, and therefore, we recommended Mrs. Anderson consider meeting with a genetic specialist in the neurogenetic clinic for further assessment. Please see the pedigree (located under the 'family history tab' for additional details.

No consanguinity reported in either maternal or paternal family history

Ethnicity

- Maternal: Italian
- Paternal: Irish, Native American

The information provided is based on the patient's recollection of the family history and in the absence of complete medical records. If the family history changes or if more information is obtained, the patient was asked to contact me as this may alter the recommendations or impression of the family history.

Risk assessment for a hereditary cancer syndrome: Mrs. Anderson's personal and family history is suggestive of a hereditary predisposition given multiple family members with related cancers (i.e., breast and ovarian cancer), cancer in multiple generations of the family, early-onset cancers (i.e., before the age of 45), and finally patient's diagnosis of more than one cancer.

Genetic education: (Consider use of SmartTools/templates for this section and providing patient with education sheets or brochures. Be sure to reference the fact that patient education sheets or brochures were provided to the patient in the document. In addition, consider including links to Web sites that provide information about genetic disorders to patients.)

Mrs. Anderson's family history of breast and ovarian cancer is most suggestive of Hereditary Breast and Ovarian Cancer HBOC) syndrome caused by changes (called pathogenic variants) in the *BRCA1* and *BRCA2* genes. Please see https://www.cancer.net/cancer-types/hereditary-breast-and-ovarian-cancer for information on HBOC including genetics, inheritance, cancer risks, management, and prevention.

Recommendations for genetic testing: Per NCCN guidelines, Mrs. Anderson qualifies for a more comprehensive multi-gene panel analysis due to the fact that she was age 38 at the time of her first breast cancer, she was diagnosed with bilateral breast cancer and has a family history of breast and other cancers.

Implications of genetic testing and brief overview of medical management: (Consider use of SmartTools/templates for this section)

Prior to deciding whether to undergo genetic testing, we discussed the possible results. If the test is positive for a pathogenic or likely pathogenic variant, in a hereditary breast cancer gene (meaning the variant found in a gene is related to an increased risk for breast cancer), there may be an increased risk to develop additional cancers in the future. In this situation, the necessary follow-up care will be recommended to help lower the risk for future cancers and/or to detect cancer at an earlier stage.

If the result is negative, then there is no identifiable hereditary cancer pathogenic variant, but because the personal and family history is significant, it is possible there is a pathogenic variant that was not identified through this test. Therefore, we recommend contacting us periodically to learn about any updated genetic tests that might be relevant.

The third possible result is a variant of uncertain significance or a VUS. A VUS is an alteration found in a gene but the laboratory does not have enough information to determine whether the alteration is a benign (harmless) genetic variant or if it is associated with an increased risk of cancer. As more information becomes available, some VUS are eventually reclassified, and research shows that if a VUS is reclassified, the odds are high that it will turn out to be benign. We recommend that you contact us periodically for an update on the status of the VUS.

Risk for family members: If the result is positive, there is a 50% chance to pass this pathogenic variant to each offspring. In addition, siblings have a 50% chance to have inherited the same pathogenic variant. Other family members may also be at risk of having inherited the familial pathogenic variant.

If the result is negative, there is nothing identified to pass to offspring. If the result is a VUS, we do not recommend testing at-risk family members unless or until the VUS is reclassified as a positive result.

Psychological issues/concerns: Finally, we discussed the possible psychological implications of genetic testing, issues of family dynamics, and concerns regarding confidentiality, insurance coverage, and genetic discrimination.

Mandated reporting can be inserted here if applicable.

Decision about genetic testing: After discussing the risks, benefits and limitations of undergoing genetic testing including the potential implications of test results on clinical management and the fact that the clinical use and understanding of some of these genes included on the multi-gene panel analysis is limited, she decided to pursue an updated panel test for hereditary breast and ovarian cancer syndromes.

Name of Genetic Counselor, MS, CGC
T: XXX-XXX-XXXX (direct & confidential)
Email:
Time Spent in Direct Patient Care: 45 min
Time Spent in Indirect Patient Care: 60 min