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EDITED BY

Ashok Kumbamu,
Mayo Clinic, United States

REVIEWED BY

Cheng Hung Tsai,
Cheng Shiu University, Taiwan
Patrick J. Dillon,
Kent State University at Stark, United States
Bryan Abendschein,
Western Michigan University, United States

*CORRESPONDENCE

Jennifer D. Cotter
✉ cotter@clermson.edu

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Effective patient-provider communication in healthcare genetics: a concept analysis

Jennifer D. Cotter^{1*}, Karyn O. Jones², Stephanie C. Davis³,
Anna Baker⁴, Anne Koci⁵ and Luigi Boccuto¹

¹Healthcare Genetics and Genomics Program, School of Nursing, College of Behavioral, Social and Health Sciences, Clemson University, Clemson, SC, United States, ²Department of Communication, College of Behavioral, Social and Health Sciences, Clemson University, Clemson, SC, United States, ³School of Nursing, College of Behavioral, Social and Health Sciences, Clemson University, Clemson, SC, United States, ⁴Department of Psychology, College of Behavioral, Social and Health Sciences, Clemson University, Clemson, SC, United States, ⁵College of Nursing, Texas Woman's University, Denton, TX, United States

Effective communication of genetic testing results between healthcare providers and patients remains essential, but the concept generally lacks a precise definition. Since clinical genetic testing has increased dramatically in recent years, it is crucial to clarify the concept of effective communication as it pertains to patient-provider communication in healthcare genetics. The Walker and Avant methodology of concept analysis was used to operationalize the effective communication definition, along with the identification of associated characteristics and illustrative examples of the concept's application. This refined definition informs communication in healthcare genetics practice and supports the development of future research methodologies for assessing the effectiveness of communication approaches in clinical genetics.

KEYWORDS

concept analysis, effective communication, physician-patient communication, health communication, genetic testing

1 Introduction

Rapid technological advancements in genetics have driven not just a greater understanding of genetic diseases but also a necessity for incorporating genetic testing into healthcare practice (Durmaz et al., 2015). Personalized medicine, using one's genetic profile to inform health decisions such as in family planning, medication and treatment selection, determination of appropriate clinical follow-up frequency, and cascade testing, is now more accessible than ever due to increased availability and decreased cost (Phillips et al., 2018). Since genetic information is inherently complex, this increased access and utilization presents significant new challenges to healthcare providers and their patients (Medendorp et al., 2021), from determination of specific test applications and utility (Lubin et al., 2008) to interpretation of results and their implications (Scheuner et al., 2012; Recchia et al., 2020). Effective communication of genetic information is essential to overcome challenges such as these and provide high-quality healthcare services. However, while *effective communication* is used frequently in the lexicon of healthcare genetics, a clear, universal definition is rarely described. Further, reliable measurement and the constituents of *effective communication* in genetics are vague and inconsistent.

The phrase *effective communication*, frequently used alongside *genomic literacy*, is defined as "the capacity to obtain, process, understand, and use genomic information for health-related decision-making" (Hurle et al., 2013). While the two concepts are closely related, they are not

interchangeable. Additionally, in the broader field of health communication research, representation of the related concept of *effective health communication* is limited, and research has been primarily focused on message persuasiveness as a measure of effectiveness (Kreps et al., 2003; Fishbein and Cappella, 2006; Rimer and Kreuter, 2006; Keller and Lehmann, 2008). Kreps, Bonaguro, and Query briefly discussed the concept of effectiveness in their summary of the history and development of the field of health communication, noting that communication research is intended to “direct the knowledge gained toward helping participants in the modern health care system use communication strategically to accomplish their health goals” (Kreps et al., 2003). Fishbein and Cappella used data from research on smokers’ intentions to continue or quit smoking to discuss the critical role of communication theory in efforts to change health behaviors, and Rimer and Kreuter advanced persuasion theory as the driving force behind tailored health communication (Fishbein and Cappella, 2006; Rimer and Kreuter, 2006). Finally, Keller and Lehmann performed a meta-analysis to identify factors influencing the effectiveness of health communication, using individuals’ intent to comply as a measure of effectiveness and found several communication approaches and individual characteristics that contribute to health communication effectiveness (Keller and Lehmann, 2008).

While genetic communication can be regarded as a subset of health communication as the two fields share many commonalities, nuances exist that are specific to healthcare genetics. Genetic information is inherently complex, associated recommendations may not be clearly defined or standardized, and results may have implications beyond the individual, including impacts on family members and future generations. Therefore, a precise definition of effective communication as applied to clinical genetics is needed to ensure consistent assessment of the effectiveness of communication strategies within the field, which can ultimately lead to deficits in patient care (Mazzola et al., 2019). As such, clarification of *effective communication* can benefit genetic healthcare practice and future healthcare genetics research. The purpose of this concept analysis is to refine a definition and identify qualities of *effective communication* as applied to clinical genetic information using the Walker and Avant methodology (Walker and Avant, 2018).

2 Methods

A concept analysis is a method by which a refined theoretical definition and description of a concept can be distilled through literature review and rigorous linguistic evaluation. This iterative theory-development process is used when a concept is poorly understood, not clearly defined, or easily confused with another concept, which hinders its use within the field of interest (Walker and Avant, 2018). Concept analyses have been applied across many disciplines, including philosophy, political science, linguistics, information technology, nursing, and medicine. Though several specific approaches to conducting a concept analysis exist, all involve clarifying the concept itself and elucidating its defining features. The concept analysis method used here was adapted from Walker and Avant and consists of the following steps: select a concept, determine the aims and purpose of the analysis, identify instances of use of the concept, determine defining attributes and characteristics, identify a model case, identify borderline, related, contrary, and illegitimate

cases, identify antecedents and consequences, and define empirical referents, if they exist, and determine potential implications of the concept (Walker and Avant, 2018).

This concept analysis aims to derive a definition for and describe the components of the concept of *effective communication* to enable operationalization in healthcare genetics research and practice. According to Walker and Avant, a literature search is employed to evaluate the concept’s uses, attributes, antecedents, and consequences (Walker and Avant, 2018). This literature search is not intended to represent a systematic literature review.

The search strategy we adopted implied the utilization of resources including PubMed, Academic Search Complete, Web of Science, Google, and Google Scholar using the search terms “effective,” “communication,” “genetic,” “genetics,” “effective communication concept,” and “effective genetic communication concept analysis.” These search terms were used both individually and in combination with one another. The search terms were intended to be general to return the broadest selection of results and to determine that a concept analysis had not already been performed on the selected concept. The search was limited to peer-reviewed scholarly literature published within the past 15 years, spanning January 2008 to November 2021, with the earliest censor date chosen based on the presumed influx of interest, development and utilization of clinical genetic testing with the introduction of the Genetic Information Nondiscrimination Act (GINA) of 2008, which protects Americans from employment and health insurance discrimination (P.L. 110–233, 122 Stat. 881). Returned search results were limited to include only those relevant to medical usage of *effective communication* in genetics, resulting in a final compilation of articles and dictionaries. Definitions and historical use of “effective” and “communication” were obtained online from the Merriam-Webster Dictionary. Self-awareness of potential bias due to the authors’ personal beliefs and interpretations was practiced to maintain scientific rigor.

2.1 Purpose and aims

The purpose of this concept analysis is to arrive at a useable definition and elucidate operationalizable characteristics of the use of *effective communication* in healthcare genetics. Identified sample cases are described to illustrate the definition. Descriptions of and relationships between antecedents and consequences, attributes and characteristics, and empirical referents are also presented. This analysis aims to improve research methodology and medical practice by generating improved clarity regarding *effective communication*.

3 Results

3.1 Instances of use

3.1.1 Dictionary definition

With the purpose and aims identified, the next step in the analysis process is to identify all uses of the concept. For this purpose, a dictionary definition is ideal, as it encompasses a range of both modern and historical usages of a concept. *Effective communication* is, by nature, a phrase, the meaning of which incorporates the individual meanings of two terms. As there is no clear definition for the

combination of effective and communication yet, the dictionary definition will be considered as the summation of the definitions of the individual component terms. To that end, each term must first be considered independently.

The Merriam-Webster Dictionary definition of effective is “producing a decided, decisive, or desired effect;” “impressive or striking;” “operative;” “actual;” or “ready for service or action” ([Effective Definition and Meaning – Merriam-Webster, 2021](#)). Further, effective can be used as a noun to mean “one that is effective” ([Effective Definition and Meaning – Merriam-Webster, 2021](#)). For this concept analysis, only the foremost usage and definition will be considered, as it is most relevant to this analysis. Effective was first used to mean “producing a desired result” in the 14th century. It originates from the Middle English word *effectif*, derived from the Late Latin term *effectivus*, which means “producing a result,” and the earlier Latin origin of *effectus*, meaning “involving an end product.” *Effectus* is the past participle of *efficere*, which means “to make, bring about, produce, carry out” ([Effective Definition and Meaning – Merriam-Webster, 2021](#)). As this dictionary definition suggests, the meaning of effective has remained relatively constant despite the many derivations that have occurred: to bring about a desired result.

Merriam-Webster Dictionary defines communication as “a process by which information is exchanged between individuals through a common system of symbols, signs, or behavior;” “personal rapport;” “information transmitted or conveyed;” or “a verbal or written message” ([Communication Definition and Meaning – Merriam-Webster, 2021](#)). It was first used in the 14th century to mean transmission of information ([Communication Definition and Meaning – Merriam-Webster, 2021](#)). It is derived from the Latin term *communicatio*, which means “a sharing or imparting” ([Durham Peters, 2008](#)). The meaning of communicate, therefore, has consistently been based on the transference of information.

To fully appreciate the meaning of *effective communication*, the two individual definitions must be combined. Recall that the definition of effective was “to bring about a desired result,” while the definition of communication was “the transference of information.” Taken together, the complete dictionary, or theoretical, definition for *effective communication* is the transference of information in such a way that the desired result is obtained.

3.1.2 Instances of use in literature

While *effective communication* is rarely defined, and even more rarely explicitly defined, in published literature, the intended meaning of *effective communication* as it applies to the context of healthcare genetics practice is, in some cases, implied within the text. These inferred definitions are often presented indirectly in the form of descriptions of what is not *effective communication*, rather than descriptions of the constituents of *effective communication* itself. For instance, Joseph and colleagues reported that *effective communication* of clinical genetic test results is hindered by excessive or irrelevant informational content, complex or jargonistic language, lack of patient engagement, and provision of unclear recommendations ([Joseph et al., 2017](#)). It can then be inferred, given that these components contribute to ineffective communication, that *effective communication* would comprise the contrary. Alternatively, an incomplete inferred definition for *effective communication* is sometimes presented in the form of one or a few components of the concept serving as a focal point in the literature. In one such example, Farrell and colleagues reported that

an important component of *effective communication* is awareness of and respect for personal values and beliefs ([Farrell et al., 2020](#)).

3.2 Defining attributes

The next step in the concept analysis process is to identify defining attributes. These attributes are characteristics or traits that comprise the concept. They define and distinguish the concept from similar or related concepts ([Walker and Avant, 2018](#)). For this portion of the analysis, key characteristics identified through a review of literature sources are compiled.

Several characteristics were identified during this analysis. *Effective communication* within the field of healthcare genetics is (a) dissemination of information ([Communication Definition and Meaning – Merriam-Webster, 2021](#)) in a manner that is (b) situation and audience conscious ([Lubin et al., 2008, 2009; Scheuner et al., 2012, 2013; Farrell et al., 2020; Recchia et al., 2020](#)), displayed by being (c) appropriately paced ([Joseph et al., 2019](#)), and (d) avoidant of jargon ([Lubin et al., 2008, 2009; Scheuner et al., 2012, 2013; Joseph et al., 2019; Recchia et al., 2020](#)). Further, *effective communication* (e) includes only actionable and/or relevant information ([Lubin et al., 2008, 2009; Scheuner et al., 2012, 2013; Joseph et al., 2017; Recchia et al., 2020](#)), (f) provides recommendations ([Lubin et al., 2008, 2009; Scheuner et al., 2012, 2013; Joseph et al., 2017; Recchia et al., 2020](#)), (g) sufficiently fosters understanding ([Michie et al., 2005; Lubin et al., 2008, 2009; Scheuner et al., 2012, 2013; Pollard et al., 2020](#)), which is (h) confirmed by questioning ([Michie et al., 2005; Joseph et al., 2017](#)).

3.3 Model case

Identification of a model case serves to exemplify the definition of the concept. The model case should exhibit all defining attributes and represent a definitive, exemplary embodiment of the concept ([Walker and Avant, 2018](#)). Since, based on this definition, no model case could be identified within the field of healthcare genetics literature for *effective communication*, an invented, hypothetical model case will be utilized instead.

Olivia is a 45-year-old pre-menopausal female who recently underwent surgery to remove a cancerous lesion from her left breast. Dr. Smith, her physician, ordered prognostic testing on the excised tissue. Olivia has returned to the clinic today to obtain her results from Dr. Smith.

Dr. Smith begins by greeting Olivia and asking how she is recovering from surgery. Olivia explains that she is still quite sore but healing well. She also reports that she is anxious for her test results. Dr. Smith acknowledges her anxiety and comments that it is typical and expected to feel that way, given the situation. He then shares that her imaging tests suggest that all the tumor tissue was removed and that he has good news regarding her prognosis. He explains that, based on the genetic assessment of her tumor tissue, her cancer has a very low risk of spreading, only about 3–5% over the next nine years. He speaks at a conversational pace and pauses periodically to allow Olivia to ask questions. Dr. Smith informs her that her genetic testing results suggest that chemotherapy is unnecessary and that she can instead be treated with an oral medication that can help prevent her cancer from returning by interrupting the action of the hormone that

allows the cancer cells to grow. Notably, Dr. Smith finishes by asking Olivia if everything he said made sense and if anything was unclear and needed to be repeated. Once Olivia has had the chance to ask any questions and obtain responses to her satisfaction, and she expresses consent to the proposed treatment plan, Dr. Smith writes her prescription, explains how to take it, and requests that she follow up with him in 6 months.

This example is a model case because it embodies all the attributes of *effective communication*. Dr. Smith provided information in a manner that was conscious of Olivia's fear and anxiety, demonstrated by acknowledgment and reassurance. He spoke at a pace that was appropriate and avoided using jargon or highly technical terms. He provided relevant, actionable information and recommendations by detailing the metastatic risk and treatment implications of her test results, as well as the follow-up and treatment plan he prescribed. Lastly, he ensured that understanding was generated and verified this by asking Olivia if any information needed further clarification.

3.4 Borderline case

The description of a borderline case serves to highlight an example of a case that exemplifies most, but not all, of the defining attributes of a concept (Walker and Avant, 2018). For comparison, refer to the above model case in which Dr. Smith delivers Olivia's breast cancer prognostic genetic testing results with a single change—Dr. Smith does not inquire about Olivia's understanding of the information with which she was just presented.

While this example closely resembles the model case described earlier, the lack of assessment of understanding inhibits the ability to correct any misunderstandings or unclear information. Even though all other characteristics are represented, the criteria for classification as *effective communication* still need to be met. This case is, therefore, used as an example of a borderline case.

3.5 Near-contrary case

The purpose of including a contrary case is to illustrate a situation where none of the concept's attributes are demonstrated (Walker and Avant, 2018). As dissemination of information is a defining characteristic of *effective communication*, a contrary case is a complete lack of the provision of information. Therefore, in the context of *effective communication*, a near-contrary case will instead be used to consider Olivia and Dr. Smith's encounter. However, alterations to the narrative will be more significant than for the borderline case examined above.

Dr. Smith enters the room and sits on the stool near the examination table. He looks at Olivia's chart momentarily, then quickly states that Olivia's tumor came back as "ER-positive, HER2-negative, and node-negative" and that she has "nothing to worry about." He writes a prescription for Olivia and hands it to her before leaving the room.

In this example, while Dr. Smith provided information to Olivia regarding her test results, he did not do so in a manner that reflected an understanding of her anxiety, situation, or genomic literacy. He spoke quickly and acted rushed instead of appropriately pacing his speech and behaviors. His usage of genomic terminology to describe

Olivia's test results is a distinct usage of jargon instead of lay terminology. He provides no relevant or actionable results or recommendations, indicating that she "has nothing to worry about" with no further information. Dr. Smith did not foster understanding or evaluate if understanding was achieved at any point in the example. As this example does not reflect almost all the defining attributes of *effective communication*, it represents a near-contrary case of the concept.

3.6 Antecedents and consequences

Antecedents are required instances that must exist before a concept can take place (Walker and Avant, 2018). Several antecedents for *effective communication* were identified. First, proper communication skills are necessary to enable basic communication (Medendorp et al., 2021). Further, to communicate healthcare genetic information effectively, the provider must have a strong understanding of genetics and confidence in their understanding (Scott and Trotter, 2013; Wilkes et al., 2017; Mazzola et al., 2019). The provider must understand the purpose and risks of a genetic test and the associated results' interpretations and implications (Lubin et al., 2008, 2009; Scheuner et al., 2012, 2013; Recchia et al., 2020). Lastly, the provider must recognize the contextual impact of the communication (Michie et al., 2005; Wilkes et al., 2017), dedicate sufficient time to the communication (Farrell et al., 2020), and demonstrate empathy and respect (Joseph et al., 2019).

Consequences are the occurrences that are the result of the concept (Walker and Avant, 2018). Multiple beneficial consequences, but no negative consequences, were identified for *effective communication*. *Effective communication* results in improved patient understanding (Michie et al., 2005; Lubin et al., 2008, 2009; Scheuner et al., 2012, 2013; Scott and Trotter, 2013; Wilkes et al., 2017; Mazzola et al., 2019; Pollard et al., 2020; Recchia et al., 2020; Medendorp et al., 2021), more informed decision making (Michie et al., 2005; Lubin et al., 2008, 2009; Scheuner et al., 2012, 2013; Wilkes et al., 2017; Recchia et al., 2020), and improved confidence in the credibility of the provider (Wilkes et al., 2017; Mazzola et al., 2019). Further, *effective communication* yields improved collaboration between patient and provider (Lubin et al., 2008, 2009; Scheuner et al., 2012, 2013; Recchia et al., 2020), as well as increased satisfaction (Lubin et al., 2008, 2009; Scheuner et al., 2012, 2013; Pollard et al., 2020; Recchia et al., 2020) and improved outcomes (Lubin et al., 2008, 2009; Scheuner et al., 2012, 2013; Pollard et al., 2020; Recchia et al., 2020).

3.7 Empirical referents

Empirical referents are occurrences that prove the existence of the concept in the real world. They serve to provide a measure by which the concept defining attribute occurrence can be determined through and are thereby directly linked to them (Walker and Avant, 2018). Because of this, each defining attribute is restated, followed by a description of the associated empirical referent.

Prior work in the broader field of health communication led to the development of an instrument for the measurement of healthcare provider communication in clinical practice settings. This instrument, the Global Consultation Rating Scale or GCRS, enables the assessment

of components of *effective communication* in healthcare across 12 domains (Burt et al., 2014). Notably, several of the domains assessed by the GCRS are aligned with the identified defining attributes of *effective communication* in the context of genetics healthcare, and as such, these domains are discussed as empirical referents to which each is applicable.

Measurement of the situation and audience consciousness can be obtained by determining whether the patient felt respected and spoken to appropriately by the provider. Within the GCRS, the corresponding assessment domain is entitled “Building the Relationship,” and encompasses demonstrated acknowledgement of and consideration for patients’ perspectives (Burt et al., 2014). Appropriate pacing can be measured by whether the provider presented the information too quickly, too slowly, or at the ideal rate to the patient. The associated domains within the GCRS are “Providing Structure” and “Providing the Correct Amount/Type of Info for the Individual Patient,” which together comprise the appropriateness of the amount and flow of information (Burt et al., 2014). Determination of whether the provider avoids jargon would reflect whether the provider used highly technical terminology and corresponds to the “Aiding Accurate Recall and Understanding” domain of the GCRS, which includes the provision of information in a comprehensible manner (Burt et al., 2014). The provision of actionable and/or relevant results can be measured by whether superfluous information was communicated and whether all pertinent information was conveyed. Within the GCRS, the applicable domain is “Achieving a Shared Understanding,” which comprises the association of information provided with the patient’s specific circumstances (Burt et al., 2014). Likewise, the provision of recommendations would be determined by whether recommendations were provided and clarified, as well as whether patient feedback on their preferences was elicited, in alignment with the GCRS domain “Planning,” which includes shared decision making (Burt et al., 2014). In order to evaluate whether the provider sufficiently fostered patient understanding, the patient’s pre-communication and post-communication knowledge would need to be assessed. Lastly, in determining if the communication was supported by questioning, the measure would be whether or not the provider asked if the patient understood or if clarification was needed, as well as whether the patient intends to engage in the provider’s recommendations. The relevant domain within the GCRS for both sufficient fostering of understanding and confirming understanding by questioning is “Closure,” and is composed of confirmation of shared understanding achievement (Burt et al., 2014).

4 Discussion

Effective communication is necessary in healthcare genetics, significantly impacting patient satisfaction and care. Of particular relevance are the implications for informed consent. In healthcare, informed consent hinges on the assumption that a patient not only has an adequate decision-making capacity and is able to consent voluntarily but also possesses sufficient comprehension of relevant information to make an informed decision. In genetics, more than many other fields of healthcare, the provision of information in a manner that supports these assumptions prior to obtaining consent

for testing is crucial since the consequences of genetic test results may impact not only the patient’s life but also the lives of their family members as well as future generations. Due to the advancing applications of genetics in healthcare and the aforementioned necessity, *effective communication* was chosen as the focus for this concept analysis. This analysis provided not only a theoretical definition of *effective communication* based on uses but also an operational definition based on defining attributes, antecedents, consequences, and empirical referents. The theoretical definition of *effective communication* that emerged is the transference of information in such a way that the desired result is obtained. The operational definition that was developed through this concept analysis is that *effective communication* within the field of healthcare genetics is the dissemination of information in a manner that is situation and audience-conscious, appropriately paced, avoids jargon, includes only actionable and/or relevant results, provides recommendations, sufficiently fosters understanding, and is evaluated by questioning. Lastly, a model case, a borderline case, and a near-contrary case have been presented to demonstrate the concept further.

The Walker and Avant method of concept analysis was used to develop the concept of *effective communication* (Walker and Avant, 2018). The strengths of this method are that it is rigorous and thorough. Potential limitations of this method are that the literature search method may have resulted in some relevant literature being overlooked or excluded from the analysis and that the focus of this analysis was specific to the application of *effective communication* in healthcare genetics and, therefore, may have been limited by not including the broader field of communication. Additionally, this concept analysis assumes that the communicated information is true and accurate and that the provider’s intentions are not to deceive the patient. Subterfuge and intentional obfuscation are potentially important related concepts beyond the scope of this analysis; however, they could serve as confounders to the description of *effective communication* as the concept is described in this analysis. Lastly, this concept analysis was focused on *effective communication* in healthcare genetics within the context of a single clinical encounter, wherein only the provider and patient are participating. Extension of the patient-provider interaction over multiple encounters or the inclusion of additional participants, such as a patient’s family members or additional clinical staff members, could necessitate expansion or modification of the concept of *effective communication*, as it is defined in this analysis.

This analysis has potential future implications in healthcare genetics’ clinical, industrial, educational, and research settings. A clear, conceptual definition may inform future both genomic communication practices and research study design by providing a critical foundation on which novel strategies may be developed and approaches to measurement of the effectiveness of such strategies may be measured. Specifically, the defining attributes and empirical referents described could be applied to assessing the effectiveness of an approach to communication within healthcare genetics, contributing to its improvement.

Author contributions

JC: Conceptualization, Data curation, Formal analysis, Investigation, Writing – original draft, Writing – review & editing. KJ: Writing – review & editing. SD: Writing – review & editing. AB:

Writing – review & editing. AK: Writing – review & editing. LB: Writing – review & editing.

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