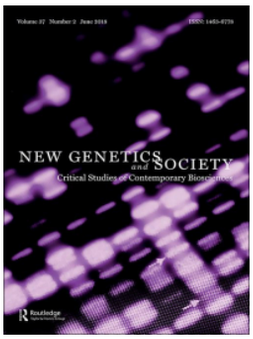

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# "Doing genetic literacy": a discourse-oriented approach to literacy in genetic contexts

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This paper proposes a discourse-oriented approach to genetic literacy. The increased availability of genetic testing requires a certain level of genetic literacy among the public. This is important to understand the benefits and the risks of genetic testing. In this paper, we suggest that genetic literacy is not just a set of knowledge and skills that people *have*, but something that they *do*. It is discursively accomplished, and socially and culturally constitutive. We illustrate our approach using a genetic counseling consultation for Sudden Arrhythmic Death Syndrome (SADS). We examine how in this consultation, through careful orientation to the clients' understanding of genetic information, extended explanation-giving and education on genetics, the geneticist supports the clients in their decision about genetic testing. We discuss our findings in relation to clinical and non-clinical genetic contexts and highlight the importance of genetic literacy and professional support in diverse situations where genetic testing is considered.

**Keywords:** Genetic literacy; genetic counseling; discourse

## Introduction

Rapid scientific and technological advances in genetic and genomic medicine have led to the emergence of new genetic knowledge. This knowledge is guiding medical professionals in gaining a more comprehensive understanding of rare genetic diseases, as well as providing new explanations for some common diseases (e.g. cancer, diabetes). As such, genetic and genomic medicine is increasingly having an impact on people's everyday lives. In the past, genetic tests were typically administered to individuals who were considered at an increased risk of developing a specific genetic condition. However, decreasing costs as well as a wider availability of these tests mean that the general public are also able to

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utilize them to manage their personal health and well-being. For example, in some countries, pregnant women now have an option of a non-invasive prenatal test (NIPT) that taps into fetal DNA and identifies the risks for a number of genetic conditions, including Down's syndrome. The test is a maternal blood test, and therefore, safer than the tests used in the past (Minear *et al.* 2015), such as a chorionic villus sampling (CVS) or amniocentesis.[1] One can hypothesize that by the time current primary school children reach adulthood, many health-related decisions that they will be making will include at least some considerations of genetics.

Moreover, genetic knowledge is extending itself beyond the traditional clinical domains. Through genetic tests that are marketed direct-to-consumer (hereafter, DTC), the general public are now able to find out about their ancestry, various health risks, and even physical traits such as baldness, athletic ability, and ear wax type. These types of tests that provide individuals with genetic information about their health, ancestry, and other biological traits are known as personal genomic testing (Savard *et al.* 2019). Since in many cases, personal genomic testing is not done for medical reasons, the majority of these tests are not covered by public health systems and are only available through private or commercial providers. Following the wide publicity and the FDA approval of some of the genetic tests offered by the US-based DTC company 23andMe, a growing number of companies have used the Internet, print advertising, and social media to market personal genomic tests. In most cases, however, these marketing activities are very loosely regulated (Hall *et al.* 2017). It also means that genetic testing is provided without specialist attention, that is, it is not accompanied by genetic counseling by genetic professionals as is the case in clinical contexts.

Considering the increased flow of information and commercial offerings of genetic testing, we believe it is crucial to examine how the general public understands genetic testing and its implications. While genetic tests undoubtedly provide useful information for the management of one's health, this information may also be "risky". Namely, finding out about one's genetic make-up may carry serious implications, for example, for one's familial relations, reproductive choices, or employment opportunities. Tapping into genetic make-up on a mass scale is also not without controversies, for example, in relation to issues of genetic privacy, discrimination, and social engineering. Understanding the benefits and the risks of genetic testing ensures that any decisions that individuals make with regards to genetic information and testing are *informed decisions*, that is, that they are based on reliable information and are not coerced by effective marketing strategies (Luo, Zayts, and Shipman 2020).

General understanding of genetic information and the ability or skills to apply that understanding to decisions regarding one's personal health and broader genetic issues is known as *genetic literacy* (Kampourakis 2017). Considering the increasing exposure of the general public to genetic information and the

importance that such information plays in individuals' health-related and social decisions, in this paper, we seek to re-conceptualize genetic literacy from a discourse-oriented perspective. We situate our understanding in a broad historical context of relevant strands of literacy research, as well as existing sociological literature on genetic knowledge and "lay expertise" (e.g. Kerr, Cunningham-Burley, and Amos 1998; McGowan, Fishman, and Lambrix 2010). Building on this research, we argue that taking a discourse-oriented approach offers a more holistic and comprehensive understanding of genetic literacy, compared to its earlier conceptualization in terms of abstract knowledge and skills. Specifically, a discourse-oriented view of genetic literacy provides valuable insights into the actual processes of how people, as members of social groups, "*do* literacy" *in situ* as they engage in social interactions where such literacy is vital. Ultimately, discourse analytic investigations of genetic literacy contribute to our understanding of what could be done to address the issue of the general public making informed decisions in relation to the new genetics and personal health.

To exemplify our understanding of genetic literacy, we draw on empirical data of naturally occurring interactions in the context of clinical genetic counseling. Our approach provides a unique perspective of how people make decisions about genetic testing in the *actual* interactions between genetic professionals and patients. While previous literacy and sociological research provides strong grounds for our investigation, they have not addressed the moment-by-moment negotiation of genetic literacy at the sufficient level of communicative detail, that is, how it is actually accomplished and what challenges may arise. To address this gap, in this paper, we examine real-life empirical data that illustrate the role of genetic literacy in facilitating decision-making about genetic testing in a genetic counseling encounter. In line with the broader context of genetic literacy extending beyond the clinical domain that we set up above, we also discuss how a discourse-based understanding of literacy can be applied to both clinical and non-clinical contexts, and how our conceptualization of genetic literacy could provide detailed insights into how it is actually accomplished in an interactional process.

## Conceptualizing genetic literacy: from a static to a more dynamic approach

Genetic literacy is generally understood as a form of health literacy, which, in turn, is based on general literacy. Early research on general literacy primarily comes from educational research. That research approached literacy from a static viewpoint, defining it as one's cognitive ability to read and write (Ivanič 1998; Pahl and Rowsell 2005). Crudely, it maintained that readers and writers engage in mental processes such as decoding, retrieving information, comprehending, and inferencing (Gee 2015). Those studies also believed that literacy could be parsed into "a discrete set of skills" (Hamilton 2010, 7) that, in turn, can be taught and acquired.

Applying the static view of literacy to health contexts, the U.S. Department of Health and Human Services, in its *Health People 2030 Framework*, defines health literacy as "the degree to which individuals have the ability to find, understand, and use information and services to inform health-related decisions and actions for themselves and others". Most available research on genetic literacy comes from the medical education and public health literature. The definitions in this strand of research echo the definition in the Framework quoted above, focusing on the cognitive aspects of genetic literacy, that is, the knowledge and skills that individuals need to *have* in order to understand genetic information and make informed decisions about their genetic health (e.g. Bowling *et al.* 2008; Pearson and Liu-Thompkins 2012). This knowledge typically includes understanding of key genetic concepts (e.g. gene expression and transmission) and the relation between one's genetic make-up and health. What transpires across most of this research is that genetic literacy is viewed as a static mental capacity that an individual either has or does not have. In line with this understanding, studies in medical education and public health have primarily focused on developing genetic literacy assessment instruments (e.g. Erby *et al.* 2008; Fitzgerald-Butt *et al.* 2015; Hooker *et al.* 2014). These studies have demonstrated a generally low level of genetic literacy among the general public (Krakow *et al.* 2018; Little, Koehly, and Gunter 2022). Condit (2010, 2), for example, observes that public understanding of genetics "tends to be […] general and has many inconsistencies in detail". He further notes that the general public understands the basics of heredity rather than the scientific basis of genes, and this understanding is based on people's social experiences.

While these studies offer some insights into the genetic literacy of the general public, their focus on assessing individuals' technical competence in genetics prevents a comprehensive view of the sociocultural complexities involved in public understanding of genetic science. This research reflects a "deficit model" of public understanding of genetics (Kerr, Cunningham-Burley, and Amos 1998; Wynne 2005) that has dominated scientific, medical and bioethical literature (e.g. Goltz and Acosta 2015; Kampourakis 2017). The deficit model foregrounds the public's ignorance and lack of knowledge of and interest in genetic science as barriers to lay participation in policy discussions and decision-making about the new genetics and health. This "deficit" understanding legitimizes policy efforts to educate the general public on scientific matters and to promote their genetic literacy. Under the influence of this deficit model, the general public are often reduced to a homogeneous group of "ignorant" subjects who are unable to understand and engage with genetic information.

The "deficit" view has been challenged by social scientists who favor a constructivist approach to examining the complexities of public understanding of genetic science (e.g. Bates 2005; Santos 2006). A unique aspect of the constructivist model is that members of the general public, rather than being viewed as ignorant, are conceptualized as "lay experts" (Irwin and Wynne 1996; Kerr,

Cunningham-Burley, and Amos 1998; Sarangi 2001) with a wide array of knowledge and situated understandings about genetics and health. These lay experts are able to draw on their own epistemology to contribute significant social and cultural meanings to genetic science (McGowan, Fishman, and Lambrix 2010). As Kerr, Cunningham-Burley, and Amos (1998) aptly note, the public is not simply in need of facts about genes or chromosomes to participate in social discussions about new genetics; rather their already *sophisticated level of knowledge* and diverse views could be further enhanced by greater availability of information and opinions about genetics. Focusing solely on technical knowledge deficiencies obscures the representation of lay people's knowledge about genetics and health. Noteworthy is that individuals' actual encounters with genetic science and technologies (e.g. in a consultation with a genetic specialist, or while deciding to buy a DTC genetic test online) are always complex, socially embedded activities. This study extends the previous sociological insights on new genetics and lay expertise by providing *contextualized, micro-analytic* evidence to support the dynamic view of genetic literacy as people's situated experiences.

The dynamic view of genetic literacy we adopt in this paper also builds on the social and cultural approach to literacy, generally known as the New Literacy Studies (hereafter, NLS) (e.g. Barton and Hamilton 2012; Cook-Gumperz 2006; Heath 1983; Pahl and Rowsell 2005; Street 1995). A common critique made by NLS scholars is that the traditional view of literacy as a neutral set of cognitive skills that resides in people's heads neglects the complex, socially situated nature of reading and writing (Pahl and Rowsell 2005). NLS argue that literacy is something people *do* in the material world and the society, it is not just inside their heads (Gee 2015). As Ivanič (1998, 65) maintains, literacy is "not […] a set of transferable cognitive skills, but a constellation of practices which differ from one social setting to another". In NLS, context thus becomes very important (Bloome and Green 2015). In short, literacy only becomes meaningful when considered within specific social practices of a particular group, and it loses its meaning when abstracted from "historically and culturally situated social practices" (Gee 2012a, 374).

While the focus of earlier NLS is on print-based literacy (Ivanič 1998), a general consensus among literacy theorists nowadays is that literacy is inclusive of meaning-making practices that integrate various semiotic resources and technologies (e.g. speech, writing, still and moving images, sounds, and music). More recent literacy research considered the multimodal and digital nature of reading and writing (Kalantzis *et al.* 2016; Mills 2016). Along with these developments, the term *discourse* has increasingly been used to describe literacy practices that involve various modes of meaning-making. Gee (2012b, 3) uses the term "Discourses" (with a capital "D") in place of literacy, defining it as socially recognized ways of using language and other semiotic resources. For Gee, Discourse concerns "ways of behaving, interacting, valuing, thinking, believing and speaking […] and often reading and writing" to identify as a member of a social group (Mills 2016).

In discourse-oriented research nowadays, the terms discourse and literacy may be used interchangeably to refer to socially recognized ways of meaning-making through the use of linguistic and other semiotic resources. In other words, literacy is approached as a discursive and interactional construct (Rex *et al.* 2010). People are thought to be constructing "ways of knowing, being, and *doing* [our emphasis] literacy" through language-in-use (Bloome and Green 2015, 23).

Drawing on these latter studies of literacy as a discursive and social practice, as well as earlier sociological research reviewed above, in this paper, we propose a discourse-oriented approach to genetic literacy. This approach understands genetic literacy as a dynamic construct – it is something that people *do*, through language-in-use, rather than simply something that they *have*, as a static mental capacity. Our use of the term "doing genetic literacy" foregrounds the action and epistemological orientation of discourse by viewing it as "a practical, social activity that does things and brings about effects; it constructs knowledge and constitutes factual versions of reality" (Arribas-Ayllon, Sarangi, and Clarke 2011, 56). In other words, discourse is not only seen as a medium of communication, but more broadly, as a means of knowledge production and taking social actions. As such, our analysis focuses on how participants of genetic counseling (i.e. geneticists and clients) engage in communication and interactions around genetic health issues, and how they *discursively* construct situated knowledge for managing personal and familial health.

We maintain that genetic literacy, as an interactional phenomenon, should be viewed as a dynamic, intersubjective accomplishment that involves not just patients/clients/the general public but also people "on the other side" of the equation, such as other interlocutors in face-to-face encounters (e.g. genetic professionals), or other parties in computer-mediated communication (e.g. message producers of DTC advertising). Genetic literacy could also be negotiated, confirmed, or contested. It is context-dependent, and socially and culturally constitutive. Approximating Gee's broader description of literacy to genetic literacy, we maintain that it reflects "relationships between individuals […] and physical, social, and cultural environments in and through which individuals think, feel, act and interact with others" (Gee 2012a, 372). We illustrate this approach to genetic literacy in the analysis that follows.

## Data and method

The data we draw on in this paper come from a large-scale research project examining genetic counseling for Sudden Arrhythmic Death Syndrome[2] (SADS) in Hong Kong. The project was approved by the Institutional Review Board of the University of Hong Kong/Hospital Authority Hong Kong West Cluster (IRB project reference number UV 15–111). The project, which was launched in 2018 at a public hospital in Hong Kong, involved collecting and analyzing a corpus of 42 audio – and video-recorded cardiac genetic counseling consultations

between medical professionals (e.g. geneticists, cardiologists) and clients. These consultations included first-time consultations when a genetic cause of a patient's cardiac condition is suspected, and post-test consultations where cascade testing[3] of other family members are discussed. All participants gave their informed consent prior to their inclusion in the project.

All recorded consultations were transcribed verbatim, using simplified transcription conventions traditionally used in conversation analysis (Jefferson 2004, see the Appendix). As such, the transcription of the consultations involved noting down the verbal, paralinguistic (e.g. pause, intonation), and non-verbal (e.g. nod, smile) details of interaction. This transcription method allowed us to examine how microlevel features of talk contribute to participants' meaning-making and their doing of genetic literacy. The recorded consultations were conducted either in Cantonese with Hong Kong Chinese participants, or, in the cases of clients originating from outside of Hong Kong, in English. The latter was used as a lingua franca (a common language among speakers of different languages). The consultations conducted in Cantonese were translated into English and verified by two native Cantonese-speaking research assistants. All personal identifiers were removed in the transcription process to protect the anonymity of the participants. The examples included in this paper are English translations of a single consultation originally conducted in Cantonese. The choice to include one consultation was deliberate, as we aimed to demonstrate how clients' genetic literacy has evolved over the course of the consultation, that is, how it was dynamically negotiated by the participants of the consultation.

The analysis of the data draws on theme-oriented discourse analysis (TODA, Roberts and Sarangi 2005), which has been widely applied to empirical research on communication in genetic counseling and other healthcare settings (e.g. Thomassen Hammerstad, Sarangi, and Bjørnevoll 2020; Zayts-Spence, Fung, and Chung 2021). As a qualitative discourse approach, TODA focuses on how meanings are made and negotiated through contextualized language use. As such, it serves as a valuable toolkit for investigating how participants of genetic counseling encounters discursively construct and negotiate their literacy with regards to genetic testing, and how it impacts on their subsequent decision-making. At the analytical level, TODA involves mapping the interactional data along the main/focal themes and analyzing the specific linguistic/rhetorical strategies that participants draw on in the interaction (e.g. different types of explanations, comprehension checks, simile). Through an inductive process of repeated readings of the transcripts, both authors first independently identified and then agreed on the recurrent focal themes. After that, we conducted a micro-analytic mapping along the main linguistic/rhetorical strategies that participants employ in the interactions. While mapping along the main themes resembles thematic analysis (Braun and Clarke 2006), it is the micro-analytic focus on interactional features that distinguishes TODA from the latter approach.

As we demonstrate in the analysis that follows, genetic literacy is prominent in these consultations, as medical professionals orient to clients' construction of their understanding of genetic information and facilitate their decision-making about genetic testing. The three focal themes related to genetic literacy that were identified as a result of the broad thematic mapping of the data include: explanation-giving about the genetic condition, client education on genetics, and decision-making about genetic testing.

## Doing genetic literacy in genetic counseling consultation

To illustrate how participants *do* genetic literacy *in situ*, we present an extended case analysis (Hutchby and Wooffitt 1998) of one genetic counseling encounter. By delving into the minutiae of communication in a carefully selected case, this method has the potential of revealing how a specific phenomenon of interest (in our case, genetic literacy) unfolds in the details of talk-in-interaction. This choice also closely aligns with our aim to explore how clients' genetic literacy "evolves' over the course of a consultation. The case we examine is a post-test consultation where the genetic result is delivered, and cascade testing is discussed. The participants are a clinical geneticist (G), a mother (M), and a father (F) of a child (not present in the consultation) who experienced recurrent syncope[4] when exercising. Based on an electrocardiogram, cardiologists diagnosed the child with catecholaminergic polymorphic ventricular tachycardia (CPVT), which is a form of arrhythmia. Since CPVT is one of the syndromes covered by the umbrella term SADS, and because there may be a genetic cause for that condition, the child was also referred for arrhythmia gene panel testing.[5] The positive genetic result confirmed the diagnosis, and the child became the first person diagnosed in the family (known as the proband). In what follows, we examine in detail how the participants construct and negotiate genetic literacy, focusing on three focal themes in the interaction: explanation-giving about the genetic condition, client education on genetics, and decision-making about testing. These focal themes and the communicative strategies that the participants employ to do genetic literacy are indicative of the interactional features observed in the overall corpus of our study.

### *Theme 1: explanation-giving about the genetic condition*

In cascade testing, explanation-giving about a genetic condition typically takes place when medical professionals reveal a proband's genetic test result.

#### Example 1

1 G: CPVT (.) it's just four letters. So normally we'd – if we talk to:: our relatives, sometimes we might use this word. If you hear this, it is indeed an abbreviation (.) of the name of this very special.h hereditary heart disease, alright? As for its causes, actually it's because of his heart cells – in the cell, one of the areas is responsible for the movement of calcium. You have heard of calcium, right?

2 M: Yeah, yeah.
3 G: You obtain calcium as you drink milk ((imitates the action of drinking))
4 M: Yeah, yeah.
5 G: Indeed, keeping calcium in the heart cell is very important. If the level of calcium is imbalanced, the movement of the cell, meaning its contraction =
6 M: Mhm.
7 G: =and its motion will be affected. Besides, the heart rhythm, or the transfer of impulses will also be affected, alright? And, therefore, the gene that we tested is a gene that controls the movement of calcium (.) molecules, (.) alright? So when there is a change in the gene, this indeed means it's a situation where one gets excited, especially when one exercises vigorously, or when one gets thrilled,=
8 M: Mhm.
9 G: =in this situation, it's possible that the heart may (.) beat abnormally, or that's what we call arrhythmia.
10 M: Mhm.
11 G: When the condition continues, the heart (.) malfunctions, not sending blood to the brain. He will faint, that's it. Do you understand? (.) Alright. And that can be regarded as a relatively critical condition as you could see last time when your son was sent to the hospital, his situation was very serious. Actually, last time, there is a need for first aid.
12 M: Yeah, yeah.

In this example, explanation-giving about the condition includes both physiological and hereditary explanations, that, as previous research attests, serve to educate the clients and prepare the grounds for subsequent advice-giving and decision-making (Zayts and Sarangi 2013). In the context of SADS, a potentially lethal condition, clients' understanding about the physiology and heredity of the condition is vital, as precautionary measures could be taken. Throughout this example, the geneticist orients to low genetic literacy of the clients, assuming that their knowledge about the genetic condition is minimal. This is evidenced by how he simplifies the information about the condition. Rather than using the full technical name of the syndrome (catecholaminergic polymorphic ventricular tachycardia), he uses an abbreviation, CPVT (turn 1).

While abbreviations as part of medical jargon may make medical discourse more obscure to lay people, in this case, the abbreviation serves to simplify the explanation. The geneticist suggests that this shorter form can be used in discussing the condition with other family members, thus also orienting to their low genetic literacy. The geneticist then proceeds to explain the hereditary causes of the condition. In a similar manner, the explanation is non-technical, it is a life explanation that draws on familiar habitual actions, such as drinking milk (turn 3), exercising, or "getting thrilled" (turn 7). While some medical jargon is used (e.g. "cells," "hereditary heart disease," "heart rhythm," "molecules," "arrythmia"), the geneticist explains and defines the main concepts. For example, "the movement of the cells, meaning its contraction" (turn 5), "the heart rhythm, or the transfer of impulses" (turn 7), "the heart may beat abnormally, or that's what we call

arrhythmia" (turn 9). Moreover, in explaining what a critical condition is, the geneticist draws on the example of what happened to the child, namely that he was sent to the hospital and first aid was performed. This is what in previous research on genetic counseling was described as a localization (as opposed to externalization) strategy (Sarangi *et al.* 2003), that is, a more contextualized explanation that takes the client's specific circumstances into account. It facilitates the explanation and makes it more personalized and accessible.

During the explanation-giving, the geneticist draws on comprehension checks, such as explicit checking of understanding ("do you understand" in turn 11), tag questions with a tag "alright" (e.g. "hereditary heart disease, alright?" in turn 1). While tag questions are typically used to seek confirmation from the addressee, the geneticist proceeds with the explanation without giving the clients an opportunity to respond or ask questions about the provided information. In fact, the clients' contributions to this interaction are confined to minimal acknowledgements as they respond with interjections "yeah" or "mhm". These minimal acknowledgements are ambiguous and could convey a range of meanings, from signaling understanding to confusion and lack of understanding (Kang and Zayts 2013). These could also be used to fill in the interactional turns when clients are expected to say something, without indexing any degree of understanding. While the geneticist is actively positioning the clients as having a low genetic literacy and needing detailed explanations about the condition delivered in lay terms, the clients are also actively co-constructing their lack of knowledge by taking on a rather passive interactional role confined to minimal acknowledgements. The low genetic literacy of the clients is thus co-constructed by all participants of this interaction.

### *Theme 2: client education on genetics*

In explaining a hereditary condition, at least some level of genetic literacy on the part of the clients is crucial to ensure their understanding of provided information and informed decision-making. The medical professional in the consultation, therefore, also educates the clients on basic genetic concepts.

### Example 2

| | | |
|---|---|---|
| 1 | G: | Have you heard of – what does gene mean? |
| 2 | F: | No::. [I've heard of the term but I don't know what it] means. ((smiles)) |
| 3 | M: | [I've heard of the term (.) but I don't quite get it]. |
| 4 | G: | You don't quite understand. |
| 5 | M: | [Yeah.] |
| 6 | G: | [Right.] Actually, in recent years, probably – well, the last eight to ten years or so I believe more and more people have heard the word from television or other media. Genes, DNA, genetic codes, and so on. We should have heard them but since they aren't of immediate concern to you, then of course you wouldn't pay special attention to them, right? |

7 M: Yeah, yeah.
8 G: So perhaps let me explain to you a bit about what genes – what DNA refers to. In fact, in each nucleus in our bodies, the central part of the cell is called nucleus, which looks like a seed=
9 M: Mhm
10 G: =[which means]=
11 M: [Nucleus]
12 G: =nucleus. In the center, it contains something called the genetic material, the genetic code, and in English DNA is used to refer to this substance.=
13 M: ((nods))
14 G: =As for its functions, actually it's like a set of instructions, meaning right after birth, it determines one's (.) height and weight, one's skin color, whether one's eyes are brown or blue, whether one is a sporty or a quiet person. These things may be affected by the genes.
15 M: These are what the genes may affect.
16 G: They may be influenced by the genes. Of course, there are other postnatal factors, yet such prenatal factors exist. For instance, you may probably know someone – in some families – you may find that some of your friends and their families are all very fat, alright? Or in some families all members are very tall. In fact, to a certain extent this is due to the influence of the genetic code. It is one of the factors. Similarly, as I just told you, one of the things that controls heart rhythm, it is also subject to the influence of the gene, and so when there are changes, mutations or abnormalities in the gene, it will be like what we see in little ((boy's name)). There will be a – meaning a heart rhythm abnormality, alright?
17 M: ((nods))
18 G: Of course, I've mentioned many different body features, yet if we go back to (.) little ((boy's name)), the gene responsible for controlling his heart rhythm – actually, all genes come in pairs, meaning all genes have two alleles=
19 M: Yeah.
20 G: =Alright? But why is that the case? In fact, for every person, half of the genes come from (.) the father, =
21 M: Ah.
22 G: =half come from the mother.
23 M: Ah, that's how it works.
24 G: Do you get it?
25 M: Got it.
26 G: Then what it means is that if one of the, say one of the genes in which one of the alleles comes from my dad, and the other one comes from my mum, so I have got a pair. Do you understand? (.) As I mentioned, this genetic material is – actually, once we realized that in little ((boy's name)) body there's a pathogenic change in one gene, we started to think about.h would it be the case that this gene is actually inherited from his father or the mother? Then, this is what we mean by a congenital illness.
27 M: ((nods))

In this example, both the mother and the father become more participatory in the interaction. This could be attributed to the geneticist addressing a direct question to them at the start of the interaction, "Have you heard of what does gene mean?" (turn 1) and checking comprehension as the interaction progresses ("Do you get it?", turn 24; "Do you understand?", turn 26). Similar to Example 1, there are also indirect comprehension checks, such as the use of tags "alright" and

"right" (turns 6, 16). Both parents admit to having heard of the term but not knowing what it means (turns 2 and 3). Such admissions about one's low genetic literacy could be face-threatening; therefore, both participants mitigate it. Note the father's smile as a face-saving strategy (turn 2), and the mother's use of the adverb "quite" in "don't quite get it" to mitigate the degree of her lack of knowledge (turn 3). The geneticist also mitigates the clients' low genetic literacy by normalizing this lack of knowledge: since this information was not "of concern" to the clients, it is normal that they did not pay attention to it (turn 6). Understanding of this basic information on genetics is, nevertheless, important. Therefore, the geneticist proceeds with educating the clients on it. In terms of the specific discursive strategies employed to facilitate the explanation, there are definitions of concepts (e.g. "the central part of the cell is called nucleus," turn 8; "this is what we mean by a congenital illness," turn 26), synonymic expressions, including in another language (English) (e.g. "the genetic material, the genetic code, and in English DNA," turn 12), and paraphrases (e.g. "all genes come in pairs, meaning all genes have two alleles," turn 18).

The geneticist also draws on the rhetorical device of simile to compare the concepts that he is explaining to more familiar objects (e.g. "nucleus, which looks like a seed," turn 8; "its functions, actually it's like a set of instructions," turn 14). The simile is a kind of life explanations as described in Example 1. Also forming part of life explanations are references to familiar people in the clients' everyday lives, "some of your friends, and their families … " (turn 16). In this extract, the geneticist shifts between generalizations and localizations (Sarangi *et al.* 2003), from "more and more people," "some families" to "you," also "little ((boy's name))". More general information is aimed at reassuring the clients' that their situation is not unique and other people's lives are also influenced by their genes. The localization of information helps to focus on the clients' specific circumstances, making sure that important information is taken on board, such as information about mutations in the child's genes and the resulting arrhythmia.

From turn 18, the geneticist moves to a hereditary explanation that focuses on how the genes are passed from parents to their children. This could be a very sensitive piece of information, as particularly in Chinese culture, mothers are typically blamed for the health issues that their children may have (Zayts and Pilnick 2014). The importance of this explanation is both to address potential guilt and responsibility that the parents may experience on receiving their child's genetic result; in the context of cascade counseling, it is also to explain why other family members, either on the mother's or the father's side, also need to consider testing for CPVT. The latter is important because while other family members may not yet have experienced any critical episodes, they may, nevertheless, be at risk.

In this example, we can see how the clients' genetic literacy is co-constructed interactionally. In other words, the genetic literacy of the clients evolves and develops in the course of the encounter. As the geneticist uses a range of interactional strategies to make the general information on genetics more accessible, the clients'

also actively display their understanding of the provided information. This is evidenced both through minimal acknowledgements (e.g. "mhm," turn 9; nodding in turns 13, 17 and 27), and other explicit cues that suggest their understanding, such as turn co-construction (turns 10 and 11 where the mother completes the turn for the geneticist) and statements signaling understanding ("Ah, that's how it works," turn 23; "got it," turn 25).

In the next example, we examine how an enhanced level of genetic literacy facilitates clients' decision-making about testing.

### *Theme 3: decision-making about testing*

Previous research has shown that genetic counseling in Hong Kong tends to be directive, that is, medical professionals often suggest to clients that testing should be done in their best interests; and clients, in turn, explicitly express their preferences of being supported in decision-making (Zayts and Schnurr 2013, 2014). In the context of SADS, a potentially lethal condition, there are, however, clinical indications for testing that, at least partly, justify the directiveness of the medical professionals (see Clarke 1997). Whether directive or not, decision-making needs to be informed (Zayts and Schnurr 2011).

Example 3 focuses on decision-making about cascade testing. Prior to this extract, the geneticist explained to the family the benefits of cascade testing, in particular, that it can be used to protect other family members at risk from critical incidents and improve their quality of life. The mother acknowledged her understanding of this information and participated in discussions about cascade testing with the geneticist, which attested to her enhanced level of genetic literacy.

### Example 3

| | | |
|---|---|---|
| 1 | G: | So I think, at this stage, because now your son has it, so at this stage, it should be daddy and mummy take the test first, because he doesn't have any siblings,= |
| 2 | M: | Mhm. |
| 3 | G: | =right? So, you need to consider this, right? So, eh, if you want to do it today, I will arrange it for you, right? And then we will ask you to come back later, by phone, because we need to get the report and have all the arrangements done before asking you to come, so we'll discuss with you again later. So today we won't give you a date for the next consultation. |
| 4 | M: | Okay. |
| 5 | G: | If the report returns, we will call you. That's it. |
| 6 | M: | Are we going to have the blood collected in your room? |
| 7 | G: | Outside, [yes]. |
| 8 | M: | [I] see::. Then let's [get it] done, as we are now here already. We came from Tsuen Wan. |
| 9 | F: | [Outside]. |
| 10 | G: | Okay. Are there any other questions? Yes, I know you live far away, right? |
| 11 | M: | Yes. |
| 12 | G: | Okay, alright. So::, okay. Would you please wait outside, and in a minute my colleague will take the blood for you. |

While the suggestion to take the test is framed with modals "should" (turn 1) and "need to" (turn 3), the geneticist mitigates the directiveness of the suggestion by providing an explanation of why the mother and the father should take the test ("because he doesn't have any siblings," turn 1). This mitigation, however, does not bring into question the actual need for testing: in case there were any siblings, they would have taken the test first. The suggestions are framed as questions; however, they are tag questions that are typically used to elicit agreements. The suggestion to have it done "today," and the acknowledgement that the clients live far from the hospital, and, therefore, it would be more convenient for them to take the test on the day of the consultation, also direct the clients toward taking the test. While the geneticist's directive stance may conflict with the professional tenet of nondirectiveness endorsed by genetic counseling communities, a nondirective approach has also been questioned as potentially requiring a complete suspension of expert judgment, which may not always be in the best interests of the clients (Pilnick and Zayts 2012). As mentioned previously, the clinical indication for testing in the context of SADS also contributes to justifying the directive stance of the geneticist. The mother does not object to taking the test. The practical consideration appears to be the decisive factor in the clients' decision-making ("Then let's get it done, as we are now here already," turn 8). While it can be argued that the clients' consent to take the test may not be truly informed, the mother's active engagement in discussing the possibility of cascade testing for an extended family member (i.e. her sister's young daughter) with the geneticist before making the decision (not presented in the extracts due to space consideration) suggests that the choice is likely an informed one. This observation can also be supported by the mother's display of her enhanced genetic literacy in Example 2. The geneticist then checks if the clients have any further questions (turn 10). The formulation of the question ("any other questions," cf. *some* other questions) suggests that the geneticist does not expect further questions (Heritage and Robinson 2011). The consultation ends with the geneticist asking the clients to wait outside for the blood test.

With regards to genetic literacy, toward the end of the consultation, the geneticist appears to assume that the clients have reached an adequate level of understanding to make a decision about testing. The mother actively constructs her literacy here by asking a practical question about where the blood test will take place (turn 6), suggesting that she has understood the information and has made the decision to take the test (turn 8). The interaction in this example also suggests that after the explanations about the condition and the general educational information on genetics, the clients were able to arrive at a decision. In other words, in the course of this interaction, their genetic literacy has evolved from a generally low level of knowledge to being able to reach an informed decision. One observation that can be drawn from this interaction is that decision-making and genetic literacy can be mutually reinforcing, co-constitutive, and interdependent.

Specifically, with clients who display a higher level of genetic literacy, there is a tendency for genetic professionals to prompt them toward making a decision to take the test on the basis that they already understand relevant information; with "less literate" clients, however, more educational and interactional work needs to be done to enable clients to make a decision.

To conclude the analysis, we note that we view the "*doing"* of genetic literacy as a resource for engaging clients in exploring the benefits and risks of genetic testing (Miyoshi and Watanabe 2023). This is also to enable clients to participate in personal and social decisions about genetics and health (Kaphingst 2017). We do not suggest that an enhanced level of genetic literacy always leads to clients' consent to genetic testing; rather, we believe that the outcomes of "doing" genetic literacy may also include an informed decision to opt out of testing. The main aim of "doing" genetic literacy is thus to enhance clients' understanding of the risks and benefits of testing and its outcomes.

## Discussion and conclusion

In this paper, we outlined a discourse-oriented approach to genetic literacy. Our approach builds on previous research on general literacy in education, as well as health and genetic literacies in medical education and public health. It also draws on previous sociological work on genetic knowledge and lay expertise. We have discussed the trajectory of literacy research from viewing it as a static set of cognitive skills and knowledge to a more dynamic approach that considers the social and cultural sides of literacy. Furthermore, drawing on an example from the clinical context of genetic counseling for SADS, we have illustrated the dynamic, intersubjective side of genetic literacy. Specifically, we have examined how it is co-constructed and co-accomplished by the participants of the interaction (i.e. the geneticist and the parents of a child diagnosed with CPVT, one of the SADS syndromes). Our emphasis on the intersubjective nature of genetic literacy echoes social scientists' call for greater involvement of the lay public in decision-making about the new genetics and health (Kerr, Cunningham-Burley, and Amos 1998). While using the example of one consultation can be a potential limitation, a close examination of three extracts from the same interaction allowed us to demonstrate how the clients' genetic literacy has "evolved" over this consultation – from being low to a level where the clients were able to make an informed decision to take a genetic test. By investigating how genetic literacy is "done" in talk-in-interaction, this study has extended previous sociological work on genetic knowledge and lay expertise by offering *contextualized, micro-analytic evidence* on how clients discursively accomplish a situated understanding of a specific genetic condition, and how it informs their decision-making about testing.

At the start of the consultation, the clients produced multiple interactional cues of their low genetic literacy (ranging from ambiguous minimal acknowledgements to explicit acknowledgements of not knowing even basic genetic information).

The geneticist actively addressed the clients' low genetic literacy by providing elaborate explanations, educating them on the basic genetic concepts, and supporting the clients in their decision-making about testing. Generally speaking, when genetic testing is considered, clients' adequate genetic literacy is important to enable their engagement with provided genetic information and informed decision-making. In the context of counseling for SADS, where the genetic condition could potentially be fatal, literacy can literally be life-saving. In this particular example, parents' understanding of the fact that one of them could have the same condition and be facing the same risks as their child, could also have a "ripple positive effect" on the rest of the at-risk family members either on the father's or the mother's side. Genetic testing is, however, not uncontroversial. It involves risks that we briefly touched upon in the Introduction (e.g. the risk of disclosing one's genetic status), therefore, an involvement of a counselor could be crucial to weighting in all the risks and the benefits. As noted earlier, in some clinical contexts where there are clinical indications for genetic testing, risks associated with not taking the test would typically outweigh any other considerations.

Of note is a wide range of linguistic strategies that the geneticist employed to ensure that the complex information provided in the consultation is understood by the clients. These included using different types of explanations (physiological, hereditary, life explanations), providing definitions, drawing on synonyms, similes, generalizing or localizing information, among other strategies. Our analysis has demonstrated how the geneticist "picked up" on the interactional cues that the clients produced that signaled their understanding. The geneticist also drew on several comprehension check strategies, the most effective ones were direct questions to clients, such as "do you understand?". We noted that tag questions with an "ok" or an "alright" tag may not be as effective in checking comprehension. The discourse-oriented, micro-analytic examination of the counseling process in this paper has provided evidence that prior to receiving the explanations, the clients may not necessarily have an adequate level of genetic literacy to make an informed decision. Compared to the cognitive, skills-based approach to genetic literacy, a discourse-oriented approach has the benefits of showing how genetic professionals actively address the literacy needs of clients in the moment-by-moment unfolding of an interaction, to enhance their genetic literacy and involvement in decisions about genetic health. By showing how genetic literacy evolves and is interactionally accomplished in clinical encounters, our paper challenges the normative assumption underpinning clinical framings of informed decision-making, where individuals need to be pre-equipped with a certain level of genetic literacy to understand information and thereby make an informed decision (e.g. van den Berg *et al.* 2006). As such, our research aligns with existing sociological work that understands informed decision-making as a socially situated process shaped by social interactions and relationships (Bruch and Feinberg 2017).

Clinical contexts could be considered "prototypical" settings where clients need a certain degree of genetic literacy to understand the information provided

in a consultation and to make an informed decision regarding the next step. This next step may be about consenting to or opting out of testing as an individual or as part of cascade testing for a family member, disclosure of one's genetic result to others, or managing one's condition. While in this paper, we used an example of counseling for SADS in a clinical setting, our discourse-oriented approach to genetic literacy is applicable to other contexts where genetic testing takes place. In other words, the "doing" of genetic literacy may well extend beyond clinical settings to encompass more day-to-day encounters with genetics. An example of such encounters is in DTC genetic testing, which has been examined in Luo and Zayts-Spence's (2023) recent study on the discursive production of genetic knowledge among consumers of DTC tests in an online health forum. In contrast to the traditional clinical setting where clients' genetic literacy is largely mediated by professionals, these consumers interact in a more open and collaborative environment where they actively draw on their lay expertise accrued from their lived experiences to make sense of DTC tests, and at times, to question the validity of these tests. Taking a dynamic view of literacy, these consumers can be viewed as lay experts who actively participate in co-constructing and negotiating the meanings of DTC tests, which may contribute significantly to the uptake and application of the emergent technology by the society.

To conclude, we concur with Hamilton's (2010) view of literacy as an emancipating tool: an increased exposure to genetic information requires that the general public actively develop their genetic literacy to "*do* things" in a variety of social contexts related to managing personal and familial health. While this argument may reflect a neoliberal ideology of self-governance where the responsibility for managing one's genetic health is transferred from the state/medical institutions to individuals (Arribas-Ayllon, Sarangi, and Clarke 2011; Resnik 2014), such "responsibilization" can also *empower* individuals (Zinn 2020) to become active participants in social issues around genetics and health. Since individuals with perceived "low" genetic literacy (e.g. children) and those who may not have had sufficient experience with genetic information can be excluded from becoming active participants in this regard (Hui, Zayts-Spence, and Chung 2022), more targeted efforts could be made to create more opportunities for engaging these groups in public discussions about genetics and health. Research as ours is able to highlight best professional practices in clinical genetic contexts that could be extended to other social contexts where genetic information plays a role.

## Notes

1. CVS is a procedure in which a needle is inserted into the placenta under ultrasound guidance and a small amount of chorionic villi (placental tissue) is aspirated for chromosomal study. Amniocentesis involves inserting a needle into the mother's uterus to withdraw 20 ml of amniotic fluid. Both procedures carry a risk of miscarriage: 0.5% for amniocentesis and 1% for CVS (Tabor and Alfirevic 2009).

2. Sudden Arrhythmic Death Syndrome (SADS) is an umbrella term encompassing conditions that lead to sudden unexpected death without an observable cardiac structural defect in childhood and adulthood (Vavolizza et al. 2015). Genetic diagnosis can yield up to 80% success rate of detection and death prevention.
3. Cascade testing is the process of informing family members of a genetic condition discovered within the family, followed by family members getting tested for the condition. It can help improve health and quality of life for these family members by identifying and managing the condition early.
4. Syncope is a medical term referring to temporary loss of consciousness (i.e. fainting) caused by a fall in blood pressure.
5. Arrhythmia gene panel testing is used to identify genes associated with the disease in symptomatic individuals.

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## Appendix

### *Transcription conventions*

| Symbol | Definition |
|---|---|
| (.) | A just noticeable pause |
| = | No pause between the end of a turn and the beginning of the next turn |
| [ | The beginning of overlapping speech |
| ] | The end of overlapping speech |
| , | A continuing intonation |
| . | A stopping fall in tone |
| ? | A rising intonation |
| - | A sharp cut-off of a prior word or sound |
| :: | Prolongation of the immediately prior sound |
| .h | Inhalation |
| ((word)) | Transcriber's comments additional to transcription |