The Human Side of Service Engineering, Vol. 182, 2025, 195–202
https://doi.org/10.54941/ahfe1006419

The Process of Explaining Secondary Findings in Cancer Genome Panel Testing, Putting into Consideration Patient Benefit

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ABSTRACT

This study aims to derive measures for handling of "secondary findings" (reports of genetic mutations not primarily intended) which have been pointed out as an issue in cancer genome medicine and to seek improvement from the perspective of patient benefit. Specifically, semi-structured interviews and participant observation were conducted on five medical professionals at two hospitals in Japan from November to December 2023. The gene panel test used in cancer genome medicine aims to select treatment for cancer patients who have either completed or are not undergoing standard treatment, i.e. radiation, surgery, or drug therapy. Still, hereditary pathological gene mutations can be detected at a certain rate, as called secondary findings (SF). Hence, in SF there is a possibility that the mutation is inherited between parents and children and the results can extend the risk of developing diseases such as cancer in blood relatives. The benefits of this test for patients are uncertain as the possibility of SF can be burdensome. For medical staff, explaining SF to patients takes tremendous work, as well as interpreting the test results and dealing with patients. In analyzing the ethnographic data, we focused on patient benefits and used the "delayed benefit" model proposed by service management scholars. The results of the field research suggest that appropriate support from genetic healthcare professionals can bring about "perspective benefit" and "emotional benefit" with potentially positive change and satisfaction in patients. If genetic counselors initiate face-to-face interviews early on and continue the dialogue with patients until the results are disclosed, patients can actively participate, reflect, and communicate with their relatives. This approach reduced the overall burden on cancer genome healthcare professionals with improved patient benefit.

Keywords: Genetic counselling, Medical service, Benefit delay, Service management

INTRODUCTION

This study explores ways to improve on the issues in cancer genome medicine, gene panel testing explanation process for SF and alleviate the burden on healthcare professionals from the perspective of service management by focusing on patient benefit. The field research was conducted via semi-structured interviews and participant observation on five cancer genome

medicine professionals selected from two hospitals. The research was conducted with the cooperation of an oncologist, a genetic specialist, two genetic counselors and a hereditary tumor specialist.

The evolution of the next-generation sequencing (NGS) technology, which can comprehensively detect gene mutations that cause cancer, and the emergence of cancer drugs that target gene mutations have accelerated the adoption of precision medicine in cancer treatment in Japan. NGS-based panel testing meets the need to find as many gene mutations as possible that may lead to treatment. Still, the comprehensiveness of the testing also increases the likelihood of finding SF. According to a previous study by an oncologist, SF was detected in 14.3% of cases, compared to a treatment success rate of 3.6% (Inagaki et al., 2021). In Japan, since June 2019, panel testing for patients without standard treatment or who have completed standard treatment can be reimbursed by Japan's National Health Insurance system. For the majority of patients, the original purpose of finding a prospective treatment was not found, while partly informed of the possibility of SF. In the limited time remaining for the patient, one can easily imagine the burden of sharing this result with relatives. Some patients refuse to undergo confirmatory testing even after being informed of the possibility of SF. According to the medical professionals in one research, the most common reason given for refusing to check is that it is "not useful for my treatment" (Shimada et al., 2022). Shimada et al. (2025), certified genetic counselors and researchers in medical ethics and medical genetics, suggest this is due to "patients' lack of understanding of SF". First of all, it presents the difficulty of explaining SF to patients and gaining their understanding of SF. Secondly, it indicates the headstrong belief on the part of medical professionals that it is good to make use of SF in genetic medicine. There is a possibility of discrepancy from the perspective of patient benefit. European guidelines state that patients' "autonomy and desire to know or ignore" should be respected (Cozzi, 2021:95).

In this study, the data obtained from the field research was analyzed using a service management researcher's "Benefit Delay" model (Fujimura, 2018). The model proposed by Kazuhiro Fujimura, a researcher in service management made it possible to visualize the benefit for patients even when the achievement of the main objective was uncertain such as in this case study subjects. Furthermore, we derived measures to improve the explanation process of SF from this perspective.

METHOD

The two facilities that were the subject of the field research for this study were O University Hospital, a general hospital that advocates 35 medical specialties, and P Cancer Center, which specializes in cancer treatment. An overview of the two facilities is given in Table 1.

Table 1: Overview of the facilities surveyed.

Name of Facility	O University Hospital	P Cancer Center
Number of Beds	930	410
Number of Departments	35	26
Number of outpatients per day	2,400	550
Number of inpatients per day	800	350

The interviewees were five people in total: specialist in genetic medicine B, oncologist C, genetic counselor D, genetic counselor E, all of whom belong to O University Hospital, and specialist in hereditary tumors G, who belongs to P Cancer Center. We conducted semi-structured interviews with all of them. The average length of the interviews was about 60 minutes. We asked Genetic Counselor D to role-play a patient explanation, which took about 30 minutes. At O University Hospital, we conducted participant observation (30 minutes) of the SF discussion that was held every week.

Table 2: Informants.

Age/Gender	Organization	Position – Pseudonym	Duration
50s / Male	O Univ. Hosp.	Genetic Medicine Specialist B	17 yrs
30s / Female	O Univ. Hosp.	Oncologist C	3 yrs
20s / Female	O Univ. Hosp.	Genetic Counselor D	2 yrs
60s / Female	O Univ. Hosp.	Genetic Counselor E	13 yrs
50s / Male	P Cancer Ctr.	Hereditary Tumors Specialist G	23 yrs

A verbatim transcript was created from the audio data of all interviews and participant observations. Codes were added to the resulting text data, and a cross-sectional and comprehensive examination was conducted to derive common themes. MAXQDA 24 (ver. 24.1.0, VERBI Software, Berlin, Germany) was used for data coding, comparison, and examination. The following five codes, which were frequently used, were considered important themes and were re-contextualized.

RESULTS AND DISCUSSION

The following is a description of the five themes derived from the field research results analysis.

SF Explanation and Disclosure Process

The star mark indicates the point of contact between the genetic counselor and the patient. At O University Hospital, the oncologist explains SF to the patient at the initial examination. Unless there are special circumstances or reasons, a genetic physician or counselor is not required to attend. The topic of conversation is to find the treatment which is the main goal. On the other hand, at the P Cancer Center, the hereditary tumor coordinator is present at the initial examination explanation, and the coordinator takes over from the doctor in charge to explain SF. This increases the point of contact with the patient, and as a result, the probability of visiting the genetic outpatient

clinic is high. The differences in the starting point and the contents of the SF explanation potentially have an impact on the rate of access to genetic medicine afterwards.

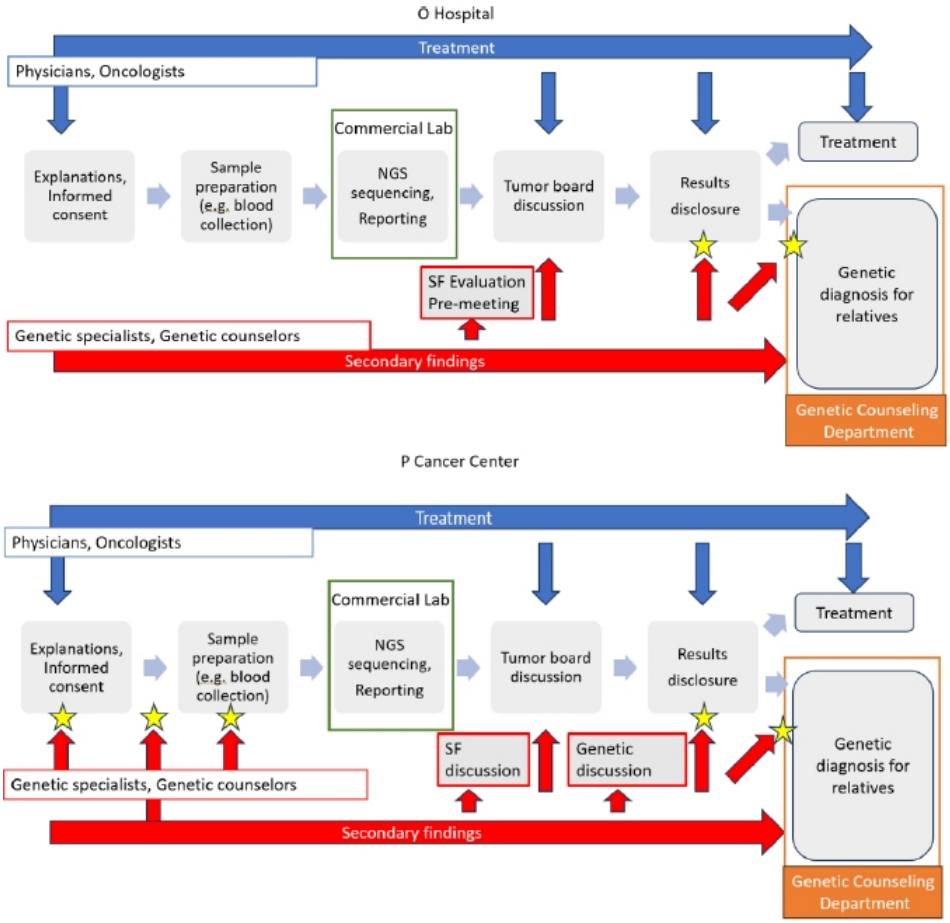

Figure 1: The differences in the workflow at the two facilities.

The Advantages and Disadvantages of SF

In order to delve deeper into this argument, I would like to take this opportunity to summarize how the medical professionals interviewed here think about the advantages and disadvantages of SF. All of the interviewees expressed the view that the advantage of SF is that they can lead to the prevention of the next onset of the disease in the patient's relatives. This suggests that there is a firm common understanding among medical professionals that SF should be used in genetic medicine, and that this understanding may take precedence over the wishes and desires of the patient.

> *"What I think about genetic medicine and hereditary tumor treatment is that when something of this importance is found, we can manage everyone's risk so there's no need to feel sorry for the parents or the children. Even so, there are still patients who worry about it."*
>
> (Interview with Hereditary Tumors s-Specialist G, December 5, 2023)

Regarding the disadvantages, Genetic Medicine Specialist B, Oncologist C, and Hereditary Tumors Specialist G mentioned that those patients who hold negative views of SF and heredity have social and psychological burden, fear, and are vigilant against social inequality.

Sharing With Family and Blood Relatives

For the patient themselves, communicating the risk of a hereditary disease to their blood relatives is often painful. The patient's feelings will vary depending on the family structure and relationship with blood relatives. For patients who do not want to tell their family about their SF, Genetic counselor D sometimes questions whether informing the family will lead to family surveillance (taking measures such as regular check-ups for undiagnosed cancer) is actually beneficial. In such cases, she first asks about the patient's own situation and feelings.

> *"Please also think about who you want to tell. Do you want to keep it to yourself and not tell anyone, or do you feel like you have to tell your family?"*
>
> (Genetic Counselor D, Patient Explanation Role Play, December 13, 2023)

Only then, measures are taken to encourage the sharing of information with family members who may become guardians of children, such as the spouse, and to arrange for the family to have ongoing communication with the Department of Genetic Medicine, with the aim of balancing the patient emotions and the family benefits.

Negative Reactions and Positive Changes

In multiple interviews at both facilities, it was mentioned that the older the patient, the more likely they were to negatively react to the idea of genetic testing.

Genetic counselor D indicated that if a patient shows a negative reaction to SF, multiple counseling sessions can help the patient feel emotionally stable, encourage their participation, and deepen their understanding. It is also useful to pay attention to what the patient says and explain things that might be of interest to them based on their situation.

> *Even if you can't think about it at the time, you might think about it later, or what you think about it then might be different from what you think about it later on. It could go either way. It could change for the better, or it could change for the worse. It also depends on whether the illness is curable or not. Even if it's cancer, it could be something that can be cured or treated, or it could be something that can't (and it would be different).*
>
> (Interview with a genetic medicine specialist B, December 13, 2023)

As the number of counseling sessions increases, the relationship and trust improve. It also becomes possible to divide complex explanations into

multiple sessions and check communication progress between the patient and family.

Point of Contact With the Patient

In the previous section, we mentioned the frequency and number of interviews as factors that bring about positive change in patients, but it is speculated that the number of interviews that can actually be held is limited. It is considered that increasing the frequency will increase the time burden on medical staff, but we have seen comments from genetic counselors themselves that indicate otherwise, they would like to meet up more with patients. It is expected of counselors to convey the doctors' message "get a medical checkup" in a way that takes into consideration patient's emotion and patient's family relationships and convey it in a way that the patient can understand.

> *"I always say that genetic counseling is like a dot. It's a point of contact with the patient. For doctors and nurses, when a patient is admitted to hospital, the dots connect to form a line, and then a plane is created, but for us (genetic counselors), we are just dots. In some cases, after just one session and it's over, it's really scary. If the patient stops listening to us because of something we say or do, then we won't be able to protect the relatives either."*
>
> (Interview with Genetic Counselor E, November 17, 2023)

It became clear that fulfilling this role poses difficulty as contacts with patients are limited. On the other hand, it was suggested that increasing the number of contacts would actually reduce the stress on medical staff when working with patients who experiencel anxiety during the consultation.

Based on the results of the above research, we describe the results of our analysis of how SF in cancer genomic medicine benefit patients using the Benefit Delay model. We will then describe the findings obtained regarding measures to improve the process of explaining SF by healthcare professionals. Finally, we will describe the theoretical and practical implications of the results of this study and the implications for future research.

The field research results were plotted on Fujimura's model "The effects of three types of benefits on customer satisfaction and customer participation" (Fujimura 2018: 213) as a frame for analyzing the field research data (Figure 2). As a result, it was concluded that even if the results do not lead to treatment for the cancer patient themselves, SF brings Perspective Benefit and Emotional Benefit for the patient. The equivalent of Perspective Benefits in SF is the possibility of managing the risk of hereditary tumors in blood relatives. The aim is not just to manage one's own health and treatment but also to manage the risk of hereditary diseases in one's relatives, such as children, siblings, and parents, through genetic medicine and avoid the onset of disease or lead to early detection and treatment. Perspective Benefits encourage customer participation and also affect the cultivation of goals and happiness even after service delivery has ended.

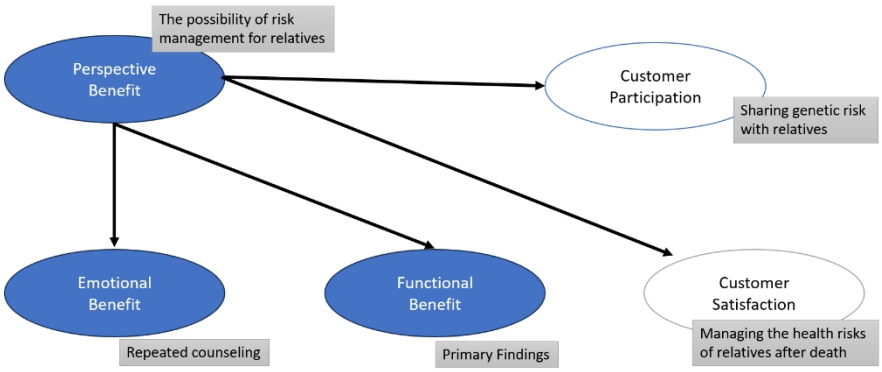

Figure 2: The author created this based on "The effects of three types of benefits on customer satisfaction and customer participation" (Fujimura 2018: 213).

Emotional Benefits are the benefits gained from good relationships between medical staff and patients, repeated communication, and active listening. Genetic counselors are assigned to specific patients, and whenever possible, the same person provides genetic counseling and related explanations. This person also participates in the medical examination of the patient's SF (including doubts), and while maintaining a high level of expertise, they communicate in a way that is empathetic to the patient's psychology and work to build a relationship of trust. Emotional Benefits foster patient trust. Genetic medical care for the patient's relatives may not begin while the patient is still alive. In order to realize genetic medical care and its benefits, it is essential to gain the trust of patients and their relatives in the Department of Genetic Medicine and the Genetic Outpatient Clinic for the future.

CONCLUSION

In this study, we clarified the actual situation in cancer genomic medicine regarding the benefits to patients and the handling of SF through field research of healthcare professionals and attempted to analyze them using the "Benefit Delay" concept in service management. "Benefit Delay" concept defines "Functional Benefit", "Perspective Benefit" and "Emotional Benefit", which were used in the explanation of medical services are expected to be applicable not only in cancer genome medicine, but also in other areas such as genetic medicine, including hereditary diseases and reproductive medicine, and pre-symptomatic screening services that go beyond the genome. In these growing fields, where it is difficult to perceive "Functional Benefit", the Benefit Delay framework used in the design and verification of service delivery processes, is expected to bring the perspective of patient behavior to the service providers (healthcare professionals) and contribute to the efficient improvement of service quality.

In terms of practical implications, several genetic healthcare professionals indicated that the participation of genetic counselors from the beginning

of the SF disclosure process and the increase of contact with patients will facilitate the execution of work related to SF. It is expected that the initiation of communication with patients regarding SF by specialized genetic healthcare staff during the initial examination explanation will encourage patient participation in actual clinical practice and lead to improvements in patient benefits.

Limitations of This Study and Implications for Future Research

In this study, the field research subject was limited to healthcare professionals, hence we could indirectly know how patient benefits were actually perceived. In addition, further research is needed to demonstrate whether the results of the field research at the two facilities can be used as a general model.

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