The Practice of Genetic Counseling for Predictive Genetic Testing in Japan

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1. Backgrounds

After the publication of the Guidelines for Genetic Testing in 2003 by genetic medicine-related societies in Japan, medical institutions are set to provide genetic counseling following originally developed guidelines. These guidelines and principles state that “Upon an examinee’s request, follow-up genetic counseling should be planned and performed to assist autonomous decision making (1).” There have been, however, a few reports on counseling practice itself, including how autonomous decision making is confirmed on site or whether counseling practice should extend beyond its confirmation(2)(3). Some practitioners suggest that genetic counseling is incompatible with the fetters of guidelines because different clients and families have different circumstances even with the same disease (4). Thus, genetic counseling practice can vary greatly depending on the particular situation of each case, and there is little understanding of the actual challenges faced by practitioners in the clinical setting and the practical meaning of these challenges.

In our previous report, "Genetic Testing and Counseling in Japan (5)," we show that clinical geneticists or doctors of other specialty departments related to genetic disease play a major role in current genetic counseling in Japan. The present article describes the practice of genetic counseling for predictive genetic diagnosis, which raises a range of ethical issues in Japan (6)(7), highlighting practitioners' narratives based on their experiences. Describing these episodes provides insight into the current situation of genetic medicine in Japan and practical skills and knowledge accumulated by forerunners with illustrative cases.

2. Study methods

2.1. Method selection

This article describes how genetic counseling practice is performed, remarkable findings and issues and how they feature in the clinical setting based on how genetic counseling providers describe what they did and thought during the practice and discussed at conference meetings. As the experience of each counseling provider (doctor) is significant only in his or her context, we describe each experience subjectively from a phenomenological perspective (8).

The oral reproduction of previous practice does not represent the practice itself but the process of redefining own practice through answering questions. In this article we also review the redefinition process.

2.2. Study participants

Medical doctors with experience in genetic counseling were requested to participate in interview. Two doctors agreed and became study subjects (Doctors A and B).

2.3. Ethical considerations

Study participants gave their signed informed consent after having been given oral and written explanations on the purpose of the study, requested duties, and that subjects who do not submit consent will not suffer any disadvantage; subjects can withdraw from the study at any time; subject's privacy is fully protected; study results may be published; and subjects appear anonymously in the publication. The subjects also agreed to being given an opportunity to read the manuscript before permitting publication.

2.4. Investigation procedure scheme (9)

An interview was performed by a lead interviewer (Q1), an assistant interviewer, and two or three record-keepers (Q2, Q3, and Q4). A private room in either participant's workplace or the investigators' laboratory
was used. Participants were independently interviewed once, for about two hours. The interviews were recorded by an IC recorder after obtaining the subject's permission. The interview started by asking "please recall the most impressive cases you experienced in genetic counseling for predictive genetic diagnosis and describe the counseling you gave," and then the subjects talked freely regarding their case experiences (semi-structured interview). The interviews were transcribed verbatim, indicating any silences and hesitations.

2.5. Analysis and interpretation methods (9)

The transcripts were used to analyze and interpret the focus of attention, the belief of the practitioners regarding what genetic counseling should be given, and how these were actually performed in their narratives. Then we highlighted the counseling sessions provided to a key client and associated the experience of the practitioner and identified the theme of the practice, as well as recounting their experience of the practice. In the descriptions, personal practice experience and the oral presentation of the experience of the practitioners were featured, abstracting the themes of the stories from the transcripts. Original page numbers were indicated at the end of each transcript for reference to note when it was told during the interview.

3. Description of counseling practice

This chapter introduces the genetic counseling practice of each study participant by theme. For easy understanding of counseling flows and issues of note, a series of counseling provided for one client is featured. The clients were the main topic of each study participant. They were designated as Client H and J in sequence to avoid identification by third persons and to distinguish from practitioners for anonymity.

3.1. Practice by Doctor A

Doctor A has performed genetic medicine and genetic counseling for more than 10 years. Doctor A says "we" or "in our team" to refer to working together with Psychotherapist C in counseling. Other psychotherapists, interns in genetic medicine, and medical specialists may have attended the counseling. Doctor A privately discusses issues proposed by clients with Psychotherapist C. In public, the current status and problems are discussed in weekly conferences with the others involved. The story recounted by Doctor A therefore represents the experience of multiple practitioners.

Clients come alone or in a pair (with a spouse, etc.) to counseling. Each counseling session has a different theme, such as spouse or psychological analysis. The following describes the counseling practice conducted for Client H, Doctor A considered a notable case. Over a period of 1 year, Client H received ten or more counseling sessions.

1) Suggesting the meaning of predictive genetic testing

Client H, a married woman in her 30s, visited the outpatient clinic seeking genetic testing. First, we consider the introduction of Client H recounted by Doctor A.

A...was the disease positive...she...her mother had Huntington's disease with a confirmed genetic diagnosis. She didn't bring the results of genetic diagnosis with her at the initial visit. The woman visited us many times. She came alone at first for counseling, and I asked about the disease, family history, and so
In the introduction of Client H, it is noteworthy that her mother has Huntington's disease and she already had a confirmed diagnosis by genetic testing. As Doctor A said "she didn't bring the results of genetic diagnosis with her at the initial visit," the genetic counseling (which covers predictive genetic testing of Huntington's disease) began with whether her parents had a history of Huntington's disease. In fact, Doctor A officially started counseling for predictive genetic testing after asking Client H to reconfirm her mother's test results.

One of the things that need to be checked at the early stage of counseling is the client's will. After saying "I asked the client's will," Doctor A referred to her family history as below:

A ...Her parents live together and dad usually takes care of mom. As their daughter, she understands that dad has many difficulties everyday because she knows what mom is like, but at the same time, she also knows that mom lives a happy life. So...she has been hoping to have genetic tests. Mom has Huntington's disease and mom's sisters as well...she was afraid that all women in her family may acquire the disease...I told her that the disease does not always develop just because you are a woman. She understood that, but meanwhile, she was sure she would have it and told me she wanted to know early because then she could prepare for it...

While talking about the life of her mother with Huntington's disease and the care-giver, her father, and her idea that she would surely develop it, Doctor A said it was the client's will to know whether she would develop Huntington's disease in the future. This shows that Doctor A recognizes her will as inseparable from her parents' life and the familial disease, and closely related to surrounding situations and relevant knowledge. That is to say, Doctor A talked about the client's will as a variable that depends on the definition of the situation.

The client's will that was given at the first visit was frequently reconfirmed by the practice of repeated and well-considered inquiries in subsequent counseling sessions.

At the early stage of counseling, Doctor A showed the client the guidelines of predictive genetic testing, which were developed in Japan and the hospital where Doctor A practiced, and obtained her consent to use them as standards in counseling. In talking to her, Doctor A emphasized "dialogue rather than a simple transmission of information" when showing the guidelines. With the attitude to open guidelines to clients, Doctor A seems to explain the guidelines via dialogue, rather than just telling policies written on them. This dialogue proceeds in a mood of discussion. As described below, the guidelines here may also imply a hurdle of predictive genetic testing. If the implication is true, the guidelines would become the main pillar of counseling practice.

Doctor A added that the client was informed that predictive genetic diagnosis may be "serious effects" and "high hurdles" for her with time-consuming involving a few months of counseling. This information is presented early in counseling so that the clients understand that predictive genetic testing is not easy to perform as many problems may occur unless the clients review and understand the significance. Disclosing information and obtaining clients' consent seem to be the first steps in counseling.

2) Preparing with the partner

Doctor A and colleagues once performed counseling in the absence of the client's partner because the client was not married yet and they "wanted to avoid damaging the relationship." It was left up to the client
to tell their partner. Although, the partner was told the test results, he was confused and had difficulty understanding the situation. The experience suggests that even if all the words and phrases said during counseling are conveyed to the partner, it is no more than a transmission of information and the partner cannot understand problems easily without experiencing the process together. Counseling was subsequently provided for the partner. Whether a test result is positive or negative, the client needs others who understand and can provide support through challenges that may emerge later. These third persons must also share various burdens with the client. From this experience, Doctor A proposes that one of the roles of counseling is identifying the client's supporters and preparing for consequences after revealing the test results to the partner. Counseling can be used to nurture attendants who just came with the client into becoming partners who can share problems with client. The next section introduces a case in which a partner attended counseling.

(1) Looking for supporters

Reflecting on the experience above and considering the “serious effects” of predictive genetic testing, Doctor A felt that partners should attend with clients in the case of predictive genetic testing. “We should find someone who can support the client, a key person, so the client does not have to receive a positive test result alone,” said Doctor A.

Client H’s husband was always busy, but he attended counseling with Client H from the next session upon request. He knew the name of the disease suffered by his wife’s mother before marriage. It seemed that Client H had consistently told him that she wished to undergo genetic testing. Her husband first attended the fourth or fifth session. His first impression was strong, stable, and assertive. He said “I cannot say ‘No’ if she (Client H) wants to do genetic testing.” Doctor A felt that Client H allows her husband to do what he likes too. But, he showed a change at the first counseling.

At that time I told them what Huntington's disease is and gave a detailed explanation in case symptoms appear after genetic testing...and if a test is positive, what kind of support he can provide to her. He said he could support her, very firmly, like "yes, I will do it"...but at the end of the first counseling, he became serious and his facial expression was completely changed...

Doctor A explained to the husband about Huntington's disease and asked what he was going to do to deal with the possible consequences. For example, what if his wife is genetically diagnosed as having the disease, or what if she actually suffers it after the diagnosis. Doctor A repeatedly used the word "if" in between asking him each question. By using "if," Doctor A seemed to be showing that undergoing predictive genetic testing is not assumed and there is a possibility of not undergoing the testing. Despite that, in the case that the client decides to have a predictive diagnosis, Doctor A provided him an opportunity to be more realistic for situations occurring after the testing by asking him to design specific measures. It is likely that Doctor A induced him to think of future possibilities as his own problems, while showing other choices by using the expression for questioning. It is not just the alternative of undergoing or not-undergoing genetic tests, —Doctor A embodied what should be done in the words of questions within an inch of a decision and implemented in the session. With such detailed, concrete talking, the husband became serious and his facial expression completely changed.

(2) Confirming and/or creating the partner’s will

In the counseling process, clients are asked to design the rest of their lives in the case of a positive result,
and partners to think how they would support the client and describe what they think of the result, both in writing. This writing provides clients and partners with a chance to organize their own ideas, and Doctor A and colleagues felt that it was a beneficial experience for both clients and partners.

Client H and her husband were asked to express their ideas in writing as well. Doctor A and colleagues were very surprised by what the husband had written.

A ...We were moved to tears reading what he had written...it said that he owes everything to her and he is grateful for everything that his wife has done. Although he knew her family carries the disease, he could not make sense of the fact. But when he had counseling, he realized why his wife had insisted on knowing. He also said that were he to know that his wife would develop the disease; he would work hard until the time comes, and reduce the amount of work after onset, so that he could stay with his wife throughout his life.

Doctor A, after reading what he had written, said that his impression was now totally different from his first impression, noting that he "must have been that kind of person all along." As Doctor A stated that "we were moved to tears and changed our opinion of him." when talking about his change, the husband's change has altered the view of counseling staff on him and Client H. Of course, Client H herself was "very happy to see the husband changed like that." Following "a substantial" discussion, Doctor A finally judged that her family's support system was mature enough even if Client H has genetic mutation. In the ethical committee, the ethics chairman read what he had written out loud "with tears in his eyes." The writing was subjected to the review of committee members as well as the family support system and the understanding of Client H, and the conduct of genetic testing was approved.

Her husband visited the hospital with her on the two different days when blood samples for genetic testing were taken and the day of disclosing the result. Her results, which were tested twice, were both positive.

(3) Getting involved in the relationship with the partner

The changes that occurred in counseling were not limited to her husband or the medical provider's view. Client H looked "lonely" at first, but her attitude and thinking also changed through the counseling.

A I think the problem was not a couple issue but an individual issue for Client H. She seemed to want to know for herself, but didn't want to trouble her husband. When we read what her husband had written to her, everyone in our office was amazed. I asked the wife, "Have you read it before?" She said “No.” So I asked the husband, "May I show it to her?" He said “OK.” After she read what he had written in our office, she looked really impressed. This seemed to improve their relationship, so genetic counseling helped to make them a better couple.

According to Doctor A, "she wanted to know for herself, but didn't want to trouble her husband," she had made up her mind to accept any results not as "a couple" but as "herself" alone, until she read his letter. But, she came to understand her husband's feelings after reading his letter in counseling. Through this process, their married relationship seemed to have changed, at least in the eyes of Doctor A.

In genetic counseling, clients and partners answer questions from counselors and face serious consequences together. This often helps them to reaffirm their underlying wills and feelings toward each other. We can see the practice of communication, reconstruction, and reaffirmation of mutual feelings in counseling here, rather than just revealing inner feelings that are usually concealed. This is why it is difficult
to communicate the outcome of counseling discussions to someone who was not actually there. Doctor A also said in the latter half of the interview that counseling served as a place of mutual communication between the wife and husband.

A ...And when he heard them both talking, he felt more at ease, he said.  
Q1 Hmm...so it was a place of mutual communication for the couple?  
A Yes, that's right, a place of mutual communication. I felt their feelings connected to each other while we talked.

Genetic counseling investigates diverse family circumstances. Husbands are exposed to wives' ideas that were previously unspoken and vice versa in this practice of investigation. This opportunity can also help to dispel preconceived ideas of both sides. Doctor A felt more at ease when he heard "them both talking..." Such a dialogue between the wife and husband via a counselor produced a place of "mutual communication for the couple," which Doctor A perceived as a heart-to-heart connection. The husband's seriousness shown in the first counseling may have been an initial sign of such interaction. If this were the case, the practice of investigating family circumstances may provide clients and their families with a secure environment in which to share and consider ideas and worries between clients and family members. Client H and her husband have changed, and our "own" feelings have changed too—I mean, including the counselors'—connected through this place of intercommunication. This may be the reason that Doctor A found the case of this couple so remarkable. However, Doctor A said that some clients and partners have separate counseling. Revealing one's own ideas directly to a partner does not always result in a good exchange, or worse, may hurt the feelings of the partner. Some clients feel able to say things to clinical psychotherapists that they could not say in front of other people. Doctor A and clinical psychotherapists have different responsibilities in this regard.

3) Repeating questions and listening with patience

According to Doctor A and colleagues, questions were repeated "over and over" and "with patience" during the counseling for Client H. The decision to undergo predictive genetic testing was the final result reached through a thousand of such careful inquiries. The next section describes how the practice progressed.

(1) Shaking the client's feelings

When Client H visited for counseling, she was seeking genetic testing. But, as we have seen, the "clients' will" can change, or can even be determined as a result of counseling.

A I tested her repeatedly to see whether she might change her mind. I asked her what if she gave up the idea of genetic testing, explained the advantages of giving up the testing and living without knowing the result. But, she stuck with doing it...She had looked for somewhere offering to examine the test at any cost since she knew the disease is inheritable...Her desire for the testing was still strong and unchanged, consistent throughout.

Doctor A tried to understand the client's feelings by asking the same the questions "over and over" and "with patience." The practice aims not only to reveal her feelings but also to "test if her feelings sway." In the case of Client H whose "desire" for genetic testing was strong, Doctor A tested her to see whether she would change her mind by having her consider options such as "giving up the idea of genetic testing" and
"the advantages of giving up the testing and living without knowing the result." Despite repeated upsetting opinions, "her "desire" for the testing was still strong and "consistent throughout."

It is possible to say that her feelings remained the same from the start, as well as that she, fully considered various possibilities each time, and reached a conclusion at last. As the relationship with her husband and preparation for accepting the test results changed through counseling, her understanding of genetic testing also changed. Her desire for genetic testing may have been reconstructed during the course of counseling.

(2) Confirming the source of feelings

Why does Doctor A place such much importance on taking time to listen to clients so patiently? Doctor A ascribed it to personal experience; one client wanted to "clear up doubts" from the parents of the marriage partner; another client requested genetic testing with recommendations by the parents of both families. For this reason, Doctor A allows plenty of time to ask about the background and ascertain the client's feelings.

A ...Analysis of the source of the desire for performing the tests is fundamental. Generally speaking, most of those who visit us want genetic testing to show the husband's parents that they are clear. But, undergoing genetic testing for this reason would then be a problem if the result were positive. So I tell them that some people have a better outcome if they do not undergo the testing. Clients think it over when we tell them that they may be unable to say they are clear because of a positive test result. You know, they are obsessed with taking genetic testing, demanding "I need it! I need it!" So, I settle down to listen...

Doctor A talks about the practice of "questioning and listening to clients with patience" here to analyze the underlying circumstances behind the desire for genetic testing. It is not a matter of simply asking the clients questions, the suggestions are made to help the clients consider possibilities that they may not have imagined, such as "some people have better results without knowing the fact" or "you may be unable to say you are clear because of a positive test result..." By these suggestions, Doctor A leads clients to "think it over." According to Doctor A, some clients are "medically not recommended" for genetic testing. For example, persons with mental instability or depression are referred to attending psychiatrists to avoid any crises. Doctor A judges whether such clients can go for genetic tests before starting "serious, high-hurdle" discussion associated with the genetic diagnosis. This approach to counseling is based on Doctor A's attitude as shown below:

A After all, we never know what will happen, depending on the result. Any harm to clients caused by test results must be avoided...One stance is that we act defensively to protect them and do not allow them to have genetic tests in an irresponsible way...So there should be complete understanding between us, and we should seek to understand their feelings, circumstances, and personal emotions by interview.

According to Doctor A, "Any harm to the clients caused by the test results must be avoided" and "we make a complete defense to protect them" depending on the results of predictive genetic testing. These words imply that the results of predictive genetic testing may cause the person harm. A counselor or the place of counseling per se, must tell the results, but as well, must protect the client from the consequences. In this context, it can be understood why Doctor A does not allow clients to have genetic testing "in an irresponsible way." Doctor A considers that even though the decision is up to the client's "voluntary will," not only the clients but also we, counselors, must be responsible for the decision. To make a "complete" defense to protect clients of genetic testing is the responsibility of counselors because the client's decision
is identical with the will of all counseling participants who reconstructed the "client's will." This is reflected in
the need to understand the client's feelings, circumstances, and personal emotions by interview. To put it
differently, the will for genetic testing and post-test support systems are determined or created by the client
and counselors together, rather than by either one of them individually, and the responsibilities are thus not
alternative.

These matters are clear from what Doctor A said about another couple who asserted "self-decision." "They
insisted on self-decision sometimes" for genetic testing, Doctor A said, and added "maybe our counseling
was a little unemotional." The story shows that Doctor A takes care in counseling so that the clients do not
make such an "unemotional" self-decision, or not decide for oneself and shoulder all the problems and
responsibilities of genetic testing.

(3) Tapping into family issues

Doctor A, as a counseling provider aiming at better mutual understanding, gave the following reply when
asked about how to become involved in or maintain personal relations with clients:

A ...Within the range they can disclose to us, like the partner problem I have just told you about, we kind of
penetrate into the inside of families and try to resolve psychological problems together. So, in my opinion,
there must be a robust relationship with mutual trust. Without sufficient rapport to disclose such problems,
counselors cannot proceed easily. We don't want the clients to feel any regrets later.

Doctor A and co-workers penetrate into "the inside of families" and try to resolve "psychological problems"
in counseling practice. This practice itself can shed light on problems, but it is limited to "within the range
they can disclose to us" and by the level of "rapport" developed, however deep they penetrate.

Doctor A said that the counselor needs to judge when to back off, and if the person seems to want to keep it
on a less personal level, the counselor should support the attitude. Whatever the personal relationships are,
Doctor A constantly considers how best to protect the clients from harm.

4) Prepare for the future

(1) "I expected the result"

This type of counseling is, in a way, a kind of preparation. This section describes some aspects of the
preparation. As confirmed previously, Client H was positive for genetic testing. After revealing the result,
Doctor A still maintains a relationship with the couple.

A ...The couple came together to hear the result. As we expected, the husband looked shocked. But, he
came with her to follow-ups. His mood was a little better by then. In contrast, the wife was rather
unemotional and said she wanted to keep in good health and had started hiking...The husband said,
although depressed still, that he would commit to his business until the disease appears because he knew
what he wanted and expected the result...He said he thought he must do it, too.

Client H and the husband, who had used counseling as a place of mutual communication, came together to
hear the result and to seek follow-up support. With regard to the test result, it sounds as if the husband was
more shocked, but he was "a little better" in counseling after two weeks and they began to turn their eyes to
their future life.
As the husband said he "expected the result," the prospective view has probably been developed through the counseling provided up until that point. Counseling for predictive genetic testing was an opportunity to confront the clients and lead them to put future possibilities into words. In parallel, it was a "preliminary" preparation for life afterwards.

(2) Introducing other specialties

In addition, Doctor A had been performing the activities below before and after genetic testing.

A  The client was referred to a neurologist before testing...We explained to the client that your neurologist would follow up if the test was positive. The neurologist met the client in our office to join the preliminary discussion...and witnessed the disclosure of the positive result, too.

In the case that she suffers from the disease, Doctor A arranged meetings among the client, a possible attending neurologist, and other related persons "before testing" and "at the time of disclosure" of the positive result. The "perfect system" described by Doctor A includes making preliminary connections among related persons. Reduction of fears for the uncertain future of Client H and the husband may be a reason for the arrangement. Probably, Doctor A also took into consideration the need for the neurologist to know the couple and the content of the counseling. Furthermore, Doctor A introduced the couple to some patient associations where neurologists were involved. Doctor A makes the most of every opportunity to link clients to prospective primary physicians.

(3) Building a personal relationship

Genetic counseling for predictive genetic testing involves the decision-making process for genetic testing and the process of imagining and preparing for life after knowing the result. The practice of preparation still continues after testing, because whether a test is positive or negative, the client may still feel distressed. Some clients with negative results may continue to have a sense of guilt for being the one to escape the genetic disease, especially when other family members are diagnosed as having the disease.

Client H has been receiving counseling once every two months with a prospect of disease onset.

A ...Including life after that, I talked about many things with her...unrelated to the disease she chatted with clinical psychotherapists or with me. For instance, she asked about the progress of medicine or current medical systems for specified diseases. With respect to specified diseases, we introduced her to a case worker for consultation. I tried to take care of these things, too.

In the follow-up counseling after genetic testing, the client and counselors continued to have a personal relationship, not only by having a positive discussion for future life, but also by talking about other things "independent of the disease." Occasionally, the preparation extended to arranging discussions with a case worker or other necessary medical providers. For Doctor A, building a personal relationship, which forms the foundation of the practice of counseling, is an essential part of the preparation.

3.2. Practice of Doctor B

Doctor B has over 20 years experience in clinical medicine and has been involved in genetic counseling for neurological diseases for over ten years. As the counseling is provided with a clinical psychotherapist
Psychotherapist E) and a nurse (Nurse F), Doctor B often used the word "we" in the interview. Nurse F, who is a contact person or liaison of counseling, answers the phone and makes appointments for prospective clients. Nurse F asks first-time callers the disease name, condition, family history, and relation with clients and patients. The set of information is transmitted to the doctor or clinical psychotherapist in charge and discussed in a conference prior to counseling.

In a counseling room, Doctor B sits in front of the clients across a table, with Nurse F beside Doctor B, and Psychotherapist E besides the clients. Psychotherapist E sits next to the clients to listen to and ask questions of the doctors and nurses from the client side. This alignment is "not intentional" but "spontaneous."

This section describes genetic counseling practice focusing on the narrative of clients who were impressive for Doctor B considered particularly noteworthy.

### 1) Concern whether the client can accept the test results

#### (1) Initial impressions

Client J, a man in his 30s, is a client who Doctor B considered particularly noteworthy. He was referred to the hospital from a patient association and received counseling about 10 times during a period of more than two years. Client J requested predictive genetic testing for Huntington's disease. His father was diagnosed as having Huntington's disease a few years ago (Client J had undergone no genetic testing at the time of the initial consultation). He said he had felt "bound by Huntington's disease" since he knew of his father's disease.

He came to the first counseling alone. Three medical providers, Doctor B, Psychotherapist E, and Nurse F, attended his counseling. He talked as if "he were giving vent to every thought and feeling that he had ever repressed." He said he had received consultation in many clinics already and failed to have genetic testing. According to Doctor B, Client J was very emotional and wanted others to understand his unhappiness. Doctor B also felt that the client only hoped "to confirm a negative test result" and "was not mentally prepared for the possibility of having a positive result." His counseling lasted two to three hours. Everyone requires this much time at the first counseling, according to Doctor B.

Asked what impressed him the most in Client J's counseling, Doctor B said "I felt great concern whether he could accept the test results and lead a normal social life" and "strongly felt" concern at the initial counseling. The concern for Client J came up in a weekly conference and the following issue was checked:

B This man showed a serious desire for genetic testing and his voluntary will was also obvious, but whether he could directly accept the test results was the first issue of concern...There was another problem of who would support this man...

Before starting counseling, Doctor B paid attention to check the client's desire based on voluntary will, mental capacity for the test results, and the existence of supporters. In particular, whether Client J "could accept the test results was the first issue of concern," and "everyone," including the psychotherapist and his father's personal physician, was afraid of his response to a "possible positive result." Supporters of Client J also told him about their concern.

#### (2) Identifying supporters and finding out the surrounding circumstances

Genetic counseling should identify the existence of a spouse, brothers and sisters, or parents who would...
support the client.

B ...I think it best that supporters address genetic testing with the same level of understanding as the client. For example, the spouse, if any, or brothers, parents, or someone else should sit with the client, listen to us, and preferably imagine a range of possible situations. It is difficult without such support even if the client is tough and courageous.

Supporters in genetic counseling offer more than just support and should "be provided counseling with the client," understand the meaning of the tests, and "be able to predict various situations." According to Doctor B "it is difficult...even if the client is tough and courageous," Doctor B expects the genetic counseling to address the whole environment including the supporters, because genetic testing may affect not only the clients but also the children, brothers, sisters, and those who spend the rest of their life with the clients.

Client J was a bachelor. His brothers did not know about his disease. His father lost the ability to make autonomous judgment because of illness. Therefore, Doctor B felt the need to talk to his mother about genetic testing for his father. Accordingly, his mother was asked to visit for an individual interview or counseling with Client J, and had discussions with Doctor B over the phone several times. The mother, who should be a primary supporter, said that she had been "very worried" about genetic testing on Client J. Not only "whether he can accept the test results," but also the whole environment around Client J, including the to-be supporter, mother, indicated the difficulty of providing genetic tests. Doctor B and colleagues thus decided on a policy of "not providing genetic tests easily" for Client J. This policy consideration applies to all clients who desire predictive genetic testing, Doctor B said.

Both the mother and client desired genetic testing of his father. It was performed prior to that of Client J because they expected a small possibility of incorrect clinical diagnosis. But the result was positive. The result was directly communicated to both the mother and client "who had been receiving counseling together."

2) Seeing whether they change their mind

(1) Explaining every aspect

Doctor B "explains every aspect of the disease, genes, and the significance of the test" to the applicants of predictive genetic diagnosis. This is the practice of providing clients with information for making a decision and to correct wrong understanding. Moreover, the clients' behavior when listening to the explanation can be useful in assessing the strength of desire for genetic testing.

B ...This man consistently requested the test each time. He had a strong desire to find out after not knowing for so long. We explained some guidelines and the level of difficulty in our office, but he was consistent in wanting to complete the testing to make it end, saying he could meet any requests for the purpose.

The repeated words, "all the time" and "consistent," show that these explanations such as "level of difficulty" were used to question the client's will and "desire," expecting a change in his will. But, Client J never changed his will despite the upsetting explanations. Nonetheless, it's not that the words of Doctor B and colleagues did not reach Client J.

B For instance, during the course of counseling, he consulted a psychiatrist or sought neurological examination in intervals between counseling sessions. He looked relieved when I told him that he was
normal. So, he was not insensitive to what physicians or others say, I think. But, his desire for predictive genetic testing did not change at any time.

Although Doctor B thought that Client J appeared stable despite the upsetting counseling explanation, he did show relief when being told the results of other medical examinations. Nonetheless, Doctor B said that his will did not change "at any time." This may be because he was determined to show his steadiness to Doctor B and co-workers at that time.

In genetic counseling, as described above, "every aspect" of genetic testing is provided, assuming that clients may change their mind as they begin to understand the disease and the significance of undergoing genetic testing. Following such changing process with clients is an important part of counseling practice.

(2) Helping the clients imagine post-diagnosis life

The narrative of Doctor B included a client who underwent predictive genetic testing.

B ...We explain many things to clients using examples...this one came down to this particular status because of the test, that one ended with that, and another one dropped out of social life due to mild mental problems even though the test was negative, and such like. When he heard these stories, he thought for a moment or said he would think it over, but he never showed a desire to postpone the decision or take an interval to think about it...His attitude "I really understand it, but still I want to do it" did not change, from beginning to end.

All example cases presented to clients usually end with problematic "conclusions" such as "this particular status" or "mental problems." Apparently, these cases are prepared as a tool to induce consideration for the significance of genetic testing at a later interval, as estimated from the narrative that Client J did not say a single word about "postponing the decision" or "taking an interval to think about it." Although Client J never said "I will think about it again," he must have thought a lot over the two years and several months, the period required until his genetic testing was performed. The case presentations were also intended to help the clients imagine post-diagnosis life.

B ...As is often the case, few people are good at predicting what will happen to their own life or family life when they are diagnosed positive. Primarily, counseling provides time to imagine clear scenarios of expected events when the test result is positive. But, this man just wanted to make it clear by undergoing genetic tests anyway. The bottom line was that he wanted a negative result...Conversely, he had neither prediction for future events nor definite plans for coping with a positive diagnosis, even after all these years...

The message, repeated by Doctor B through the narrative, is that you should have a good prediction ability, or imagination for your own and family lives that follow when you are diagnosed positive. Doctor B prompted Client J to think about the meaning of genetic testing as a matter of "himself", as that of his family life involved in the disease, or as a real, substantive matter of his own, rather than just as knowledge. In addition to a concern that "Client J may be unable to accept the disease," Doctor B could not see his substantive post-diagnosis plans in relation to his mother and younger brother or different life conditions, which may have been a factor of anxiety for Doctor B. This also explains why Doctor B introduced an example of a client who planned his future life with the spouse in the story of Client J. The example client needed a definite diagnosis because there were clear future plans and clear necessity of genetic testing.
Client J, however, could not imagine his future life until the end. He seemed to think that everything starts after knowing the result.

(3) Adjusting the client to the counselor’s pace/build up a relationship of mutual trust

The practice of explaining heredity or genetic disease and helping the client to imagine post-test life inevitably implies "a compromise with counselors," according to Doctor B. Some clients who strongly desire genetic testing saying "I will take full responsibility" change their mind through continued counseling. The possibility of change has a great importance for Doctor B and co-workers, as described previously.

B ...These persons come to a sort of compromise with us over long-term discussion...I do not know why, but I can feel it somehow. The most remarkable case was one client who was tested positive. You know, at first the client was so...like trying to lock others out, erected a high mental barrier to hide feelings, and expressed only what the client wanted. But, the client gradually opened their mind and showed signs of going at our pace...in this case.

It should be noted that Doctor B repeatedly said "our pace" here. As Doctor B presented a client who "erected a high mental barrier" gradually "opened their mind" through counseling over "long-term" discussion, it represents an agreement to accept the counselors' requirements that the client should consider the meaning of the tests by suppressing their own desire for immediate execution of testing. Doctor B asks clients to imagine post-test family life. It is therefore possible to say that genetic counseling is, in a way, the practice of adjusting the clients to the counselors' pace.

On the other hand, although details are provided later, Doctor B and co-workers also adjusted their attitude in response to the clients' ideas. These changes occur on both sides as counseling sessions go on. For Doctor B and co-workers, this leads to familiarity with the clients and to a real sense of a relationship of mutual trust between Doctor B and the clients. "Adjusting to the counselors' pace" also means changes in the personal relationship with the clients.

(4) Judgment is based on the participation of many different people

Whether clients can clearly imagine post-diagnosis events or not is an important measure of the appropriateness of the genetic testing.

B We decide that the person had better not undergo the testing when the client looks unprepared for the subsequent events and changes occurring both in oneself and the family after hearing either a positive or a negative result. We explain this by telling the client "we would like to continue your counseling a little while because we think you are not mentally prepared yet."

According to Doctor B, they tell the clients that "we think you are not mentally prepared yet" to hear the test results. Communicating the counselors' judgment can help clients to realize their situation more fully. So, how is the judgment made?

B ...Clients can take time to see other doctors in the genetic medicine department, for instance with a psychiatrist. Nurse F and Psychotherapist E always take part in counseling. Sometimes trainees who study to become qualified as genetic counselors come in and observe counseling. Anyway, many people observe clients and make a judgment...Whether clients are mentally prepared or not is the only issue. Other matters
and requirements (in guidelines) do not hamper the process.

Through observing and listening to clients, Doctor B and associate doctors, nurses, clinical psychotherapists, residents, or others judge the level of "mental preparation" of clients. According to Doctor B, this "counseling time" is a chance "to involve a diverse range of people and to analyze and observe the person from multiple aspects." Observation does not literally mean just looking at clients. He mentions "multiple aspects," because clients show a wide variety of expressions towards others in different situations. The level of "mental preparation" can be judged only by using the eyes of a diverse range of people to provide clues. Predictive genetic testing is related to gaining knowledge of something that one has not experienced yet. It is difficult to assess the clients' ability to imagine future events, so it is useful to judge from many different people's viewpoints.

Meanwhile, however hard we exercise our ingenuity for a long time, "no one knows what really is in the clients' mind." In this sense, "whether it is OK or not depends on the length of time and level of effort that everyone considers sufficient." Satisfaction of all counseling participants constitutes one aspect of preparation for genetic tests. In the case of Client J, there has been "considerable" discussion in counseling and conferences, and conformity to guidelines was confirmed. The results of these discussions may become evidence for allowing Client J to take genetic tests, even if his mind appears unprepared.

(5) Caring for clients beyond the borders of medicine

Most medical tests are performed to treat patients by detecting a disease or checking the health condition. Predictive genetic testing can detect diseases before individuals experience it. Huntington's disease can be detected by genetic testing, but currently has no established prevention or treatment and genetic testing for the disease itself can cause new problems.

That is all the more reason for genetic counseling to cross the borders of medicine as stated by Psychotherapist E. Doctor B and co-workers have even provided counseling to a client who requested predictive genetic testing because his father and older brother had symptomatic Huntington's disease. As the client and his brother had different birth fathers, the client was medically supposed to have little or no risk. According to Psychotherapist E, "these kinds of persons must take more time and receive more careful counseling." "These persons are all the more unprepared" because "they think they are OK" and request the tests "to remove any small lingering doubt." So Psychotherapist E emphasizes that "much time should be devoted to them." In counseling, clients are provided with medical knowledge, which is included in the practice of discussing future life with clients and related persons for preparation.

In spite of unresolved concerns, the moment of decision will come. Expenses for counseling itself or transportation increase the burden each time. A physician who participated in counseling once pointed out that there was the question of only repeating counseling without performing the tests. The time left to make a decision is limited, while counseling providers hope for a better situation for clients in practice. As a certain judgment is made in counseling, the next schedule can be planned. Such judgments are made each time only if a number of staff participate in counseling.

3) Informing people who have the same risk

(1) Confirm again and again

Client J has a younger brother who was not told the disease name of his father or Client J's approach to genetic testing. Doctor B was concerned that nothing would be told to the brother who has the same risk as
Client J. "I was concerned that just one of them was running ahead out of control and how the mother or client felt about the fact that the brother would not know anything, whether the test were positive or negative...and how they would deal with the fact."

Only Client J knew the disease name because he was there when his father's diagnosis was informed. His brother was too young to be there at that time. His younger brother had spent his life in ignorance of the disease "not unintentionally but consequently." Concerning treatment for the brother, Doctor B said:

B...I suggested to him many times that he should tell his brother. I think that if brothers have the same risk to some extent and one brother is informed, the other brothers should be informed about the disease. I thought it would be a problem later if one of them did not know. I still think so...so I suggested that the other brother should be informed, but both mother and the client said they would not tell him.

Doctor B suggested many times to Client J and his mother that they should tell the brother about the disease. This was repeated often in the interview. If his brother alone is kept from knowing the disease, and besides, only Client J knows the test results, the knowledge gap would be wider. Whether Client J is positive or negative his brother's condition is unknown. In either case, genetic testing for Client J would result in the situation where Client J and his mother would become nervous about his brother's disease, even though the brother does not realize it at all. This may complicate discussion on the disease or request for being a supporter in the future. The words, "I thought it would be a problem later.... I still think so," reveal that Doctor B has been concerned about this brother since then.

(2) Challenging your thinking

There was a clear reason for not telling his brother the disease name. Client J has been in distress for many years since he knew about his father's disease. He did not want to have his brother go through the same experience. The mother had told Doctor B that she was suffering from the knowledge gap between the brothers regarding the father's disease. She also said "it may be too late now, but I feel nervous about keeping it secret from him" when she heard about the genetic test result of her husband. "I tried many times to persuade him to tell his brother, but I couldn't."

This experience prompted Doctor B to challenge his own thinking.

B My basic thinking has not changed, but what I think has changed is, because there is a difference in age or residence...and the personal relationship with the parents is not always equivalent, it's case-by-case, really...so I thought the family decision is the best and right decision. But in principle, particularly when considering special medicine such as predictive genetic testing, I still believe that those who have the same risk should know it, though. Of course, I could never compel them to agree with me because each family has different circumstances between brothers, sisters, parents and children. It's beyond our control, or maybe not a subject in which medical providers should force correction...Of course we say straight out that you can think differently, or please consider whether you would really still be OK in keeping it secret from your brother as you were before the testing when you did not know whether you were negative or positive, but in the end, the judgment of which is right or wrong is not our responsibility.

Over and over again, Doctor B said "those who have the same risk should know it," which is the "principle" that "basically does not change."
The repeated word "still" may indicate that Doctor B is conscious of the unchanging basic idea even if
something is different from the "first" idea. Doctor B implied other possible ways of thinking by adding "though" after the sentence of own idea. The idea "what I think has changed," as represented by the words "case-by-case," is that "the family decision is the best and right decision" even if the principle is presented as a way of "thinking" because the family situation is different by each brother and parent-child relationship. However, as Doctor B repeated "My basic thinking has not changed" followed by saying "but what has changed." Doctor B did not completely change his thinking nor did he want to change his thinking. Doctor B just questions whether his own thinking needs to be modified in accordance with the words and situations of the clients each time.

(3) Considering keeping the information secret

The family circumstances and family judgment led to the secret from the brother with the same risk. Doctor B was slowly inclined to take the circumstances of this family into account despite his basic thinking.

B We asked the mother and client, on many occasions, whether it was really OK not to inform the brother as matters progressed and the client was ultimately ready to undergo the test. But, I was not sure whether it was really wrong. His brother may get married and have children ordinarily without knowing anything at all. Someone may think, even if Huntington's disease develops 20 or 30 years later from now, he may be happy in his way because he lived a happy married life with children for a given period of time. Otherwise he may be bothered for 10 years like the client if he knows it early, and as a result, he may take genetic testing. But, whether negative or positive, I have a feeling that it is difficult to judge which is better when these 10 years are compared with another way of spending 10 years.

From the beginning to end Doctor B talked about the brother being unaware of the disease in this part of the interview. In short, he wondered whether telling the disease to the brother would really make him happy when considering only his own life. He believed that the brother should know the fact, otherwise an information gap or relation with those "with the same risk," for instance, may cause a problem about disclosure of the disease in relation to others around him. Doctor B started to suppose a situation of "complete secrecy" after thinking about the information gap and different family relationships with the surrounding people. This is a case in which a counselor inclined to take the circumstances of the family into account despite his basic thinking.

4) Resistance to genetic testing/notification of results

(1) Explaining the option of declining to be told the results

Client J was approved to undergo predictive genetic testing after review by the ethical committee on the condition that "the course must be reported to the ethical committee." Doctor B spoke to Client J as follows in the counseling immediately before the test.

B I told him that results would automatically arrive in four weeks, so I would make the next appointment on such and such a date to tell him the results. I also told him another option that he could come back during the four weeks and decide to decline to hear the results.

Blood sampling was performed for the test at the ninth counseling. Again this time, Doctor B told him that he could he could "come back during the four weeks and decide to decline to hear the results."
genetic testing employs the practice of stepping on the brake just ahead of the testing. By making blood sampling and notification of test results separately, clients have another opportunity to think things over. Doctor B also said that after blood sampling clients have to encounter a variety of options such as who hears the test results with the clients and who shares the information with the clients. These situations had already been imagined in the course of counseling. Clients put it in practice in counseling, and at the same time, the clients' condition is checked in the counseling as well.

The client was asked to come along with his mother to the 10th counseling when the test results would be opened. This counseling required two hours or more. Doctor B confirmed again whether he would like to be told the results in front of them.

B ...I said, "I can tell you the results now, but are you really ready for it?" As soon as I confirmed his will, I told him the results. I think there was not much of an interval at that time. We had already had full discussion.

Soon after starting counseling, Doctor B told Client J the test results. Despite this, two hours or more was required because counseling for those who request predictive genetic testing always pays attention to post-test situations. For Doctor B and co-workers who were concerned "whether he could accept the test results" or "that he was not mentally prepared for life after knowing the test results," it may have been the last important opportunity to observe Client J before telling him the results.

(2) Hesitation in genetic testing
With regard to genetic testing for Client J, Doctor B said "I was sort of relieved to see a negative result," but some disquiet remained.

B ...There was little feeling of achievement. So it is still difficult to say whether it was really right...You know, there remains the problem of the brother. I do not quite know if that man really became happy with the negative result.

At the end of the interview, Doctor B said "I wonder whether he was glad to undergo the test, thinking 'I should have taken the test even if the result was positive...I made the right decision and I will not regret it.'"

B Many things cannot be judged only from our side...We regard predictive genetic testing in one way, but others may regard it in a different way...I mean, there are different views from various angles. Of course, I guess test results can benefit some people in designing their lives, and some can accept bad results and live a positive life...

Genetic testing is also used to confirm clinical diagnosis. Doctor B expressed resistance to this usage as "I often question the real need to do this." The reluctance seems to be caused by the nature of genetic testing, which diagnoses not only the person but also the future of the genetic relatives, as compared to clinical diagnosis which diagnoses only the person who experience disease. "Some people prefer not to know and stop within an inch of conclusive risk disclosure to children. I understand the idea in a way, you know," Doctor B added.

A few years have passed since Client J was told the test results, but Doctor B is still concerned about him.
4. Summary

The article describes practical counseling based on the narratives of two doctors who experienced genetic counseling for predictive genetic testing. The counseling experience of clients who requested predictive genetic testing for Huntington's disease was the main topic of both doctors, which may be because the validity of predictive genetic testing of the disease has been questioned due to a lack of established prevention or treatment. Predictive genetic testing is indicated for late-onset dominant genetic disease with high penetrance. It should be noted that such a disease has been reviewed in the present narratives of the counseling for predictive genetic testing.

The two doctors introduced in this article had been committed to clients with different backgrounds for a long time. As their personal relationships are reflected in the experience they describe, a summary of the experience is not presented here. We would like to end this article with some characteristics that were revealed through transcription.

The first to note is their methods of checking the "voluntary will" of the clients requesting genetic testing. The clients' resolve to undergo the tests was confirmed not only by being asked but also by being upset with a variety of explanations. In other words, the doctors attempted to reconstruct the "will" over the course of subsequent counseling with clients who visited and wanted to undergo genetic testing. The will can also be created by clarifying the meaning of predictive genetic testing with partners and imagining the future—driven by the participation of the doctors, as counseling providers. If wills are formed as described, the "will" for genetic testing does not belong only to the clients but is shared by all counseling participants. Specifically, not only counseling clients but all participants share the responsibilities of the decision, genetic testing, and test results. This practice of sharing responsibilities also allows participants to keep personal relationships independent of their goal, the implementation of tests, because predictive genetic testing does concern beyond the results; it is the practice of collaborative preparation for the future. The same applies to the case where they decided not to undergo genetic tests. It may well be said that counseling functions as such tools.

In the process of creating wills or building relationships, clients and families were upset by explanations and questions from doctors and co-workers, gravitated to their ideas, and transformed their wills. Concurrently, they also drew doctors toward their own ideas and challenged the thoughts of the counterparts. The ideas of counseling providers were created in discussion with each client and in a counseling team by being questioned and reconstructed through encounters with the clients. This may be because counseling for predictive genetic testing is deeply involved in the life and future of each client and family and extends beyond medical practice despite concerning medical technology or knowledge. This is also the reason why doctors cannot be conclusive in genetic counseling. The present review suggests that genetic counseling practice for predictive genetic testing is supported by knowledge that has been explored and accumulated by counseling practitioners, who faced the situation together with each client and family in counseling. The narratives in this article reveal some of this accumulated knowledge.
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