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Ethical Issues in Genetic Medicine: From the Context-sensitive Approach

Introduction

It has been progressively clarified that specific genes have influence on the occurrence, treatment, and prevention of various diseases, and genes and genomes have become more involved in every field of medicine. Conventional therapies now involve genetic study of the occurrence mechanism of cancer, therapeutic effects of anticancer drugs, and susceptibility to a specific cancer by performing gene analysis of cancer cells removed from a body. It has been revealed that specific genes promote or suppress the occurrence of some types of diseases, and clinical testing of gene therapy for providing a body with necessary genes is being conducted around the world. In reproductive medicine, it is now possible for parents to determine whether to have a baby by checking the genetic qualities (genome, gene, chromosome, and biochemical features) of the parents-to-be and the child-to-be. Genes, sometimes called "a design of life," have become in such fields a target of manipulative intervention (selection, modification, addition, and removal) based on data obtained from testing and diagnosing.

Clinical medicine is required to respect the requests and intent of patients or clients, as is the case with other medicines. Medical treatment is conducted in line with the diverse emotions of patients or clients such as pain, fear, expectation, and hope. In particular, when there is a connection between genes and occurrence, treatment, and prevention of diseases, it is usually difficult to treat the disease using general principles and manuals. Therefore, an appropriate communication process is needed to solve the problem and determine a specific strategy in line with each individual case, taking full account of the values and preferences of the individuals involved.

The intent and emotion of patients or clients are regarded in most cases as a leading factor for setting the direction of the decision-making process. In such a process, taking account of why and how the intent and emotion were developed, i.e. the context surrounding each case is indispensable. We can describe the context as various multi-layered vectors that work dynamically in clinical medicine. To be more specific, the dominant concept of values in a community, the choice of the majority of people, and the system conditions can have an impact on decisions in each area of clinical medicine, and, on the contrary, individual decision can encourage and reproduce the current situation of society and in particular people's concept of values. The relation between face-to-face communication and reflective or public

communication in clinical medicine is also a problem.

In this article, I formulate these views using a context-sensitive approach to genetic medicine and organize them based on genetic counseling. Then I confirm the role and significance of context views and summarize the necessary points of the argument. Based on this work, I study the ethical issues of genetic medicine in consideration of preventing genetic diseases in reproduction (by genetic diagnosis at the premarital, preconception, preimplantation and prenatal stages). In the concluding section, I show the significance of context-sensitive approach to genetic medicine.

1. Context-sensitive Approach to Genetic Medicine: In line with Genetic Counseling

There are various types of communications in clinical medicine for solving the problems people face and determining strategies. Genetic counseling is a typical example of such communications and is defined in general as follows;

Genetic counseling is a communication process which deals with the human problems associated with the occurrence, or the risk of occurrence, of a genetic disorder in the family. This process involves an attempt by one or more appropriately trained persons to help the individual or family to

1. Comprehend the medical facts, including the diagnosis, probable course of the disorder, and the available management;
2. Appreciate the way heredity contributes to the disorder, and the risk of recurrence in specified relatives;
3. Understand the alternatives for dealing with the risk of occurrence;
4. Select a course of action which seems to them appropriate in view of their risk, their family goals, and their ethical and religious standards, to act in accordance with that decision; and
5. Make the best possible adjustment to the disorder in an affected family member and/or the risk of recurrence of that disorder.[1]

Clients undergoing genetic counseling have a variety of reasons. For example, some visit before marriage or pregnancy suspecting that their prospective children may develop a genetic disorder since a sibling has one; some visit before implantation or natality to avoid the possibility of them giving birth to babies with chromosomal abnormalities; and some visit before the occurrence of a genetic disorder to examine the possibility of them sharing the same fate as that

[1] cf. American Society of Human Genetics Ad Hoc Committee on Genetic Counseling, Genetic Counseling, *American Journal of Human Genetics,* 27:240-242,1975; "Genetic counseling, ethical issues in," S.G.Post(ed.), *Encyclopedia of Bioethics, 3rd ed.*, Macmillan, pp.948-952, 2004.

of their parents who died from a monogenic disease.

Genetic counseling is a communication process focusing on the conversation with these clients, and it has the following essential principles: "Norms of nondirectiveness: providing balanced or unbiased information, not imposing specific values or options on the clients," "Empathetic understanding of a client's anxiety, fear or suffering," and "Respect for autonomy and support of the informed decision making of the client." Medical personnel who conduct the counseling (clinical genetic specialists or genetic counselors) present necessary medical information to the clients and families such as family history of the disease, clinical history of the client, the percentage of genetic cause of the disease occurrence, possibility of being a gene carrier, risk of passing the gene to children, applicability, cost and anticipated result of the genetic test. They then determine clear strategy by taking account of economic conditions and family relationship as well as the intent of the clients and families. They listen attentively to the fears and concerns of the clients and support their autonomous decisions made based on unbiased information. It is a communication process toward decisions that are made aiming for specific problem solving and strategy making.

Now let me focus on some actual examples of the intent and emotion of the clients and raise possible issues, with which I would like to present several contexts surrounding “the autonomous choice and determination.”

> “I’m afraid that my engagement will be broken off if the test result reveals that I am a carrier of a genetic disease and my fiancée gets to hear of it.”
> ”I would like to have a healthy baby through in vitro fertilization and conducting a genetic test since people tend to shun children with disabilities.”
> ”Children with incurable diseases would be unable to live after the death of us parents, and therefore, even though we feel bad, we would choose an abortion if a genetic test detected an abnormality.”

Various responses would be made to these concerns of the clients in the counseling session. In this context, the following questions could be raised.

> *Should we affirm the “pain” and “fear” of the clients as their psychological facts? Wouldn’t it be necessary to ask how they came to formulate such an opinion in the first place and whether the emotion arose due to a biased view and lack of knowledge and information?
> *Shouldn’t we take account of the social impact of choice as well as the concept of values that support the choice in each area of clinical medicine, particularly the choice to allow specific lives to die? Could we ignore criticism that the choice would reinforce the discrimination of persons with disabilities and promote the eugenic way of thinking?
> *What information should medical personnel provide in addition to medical information? For example, information on social support systems for people with disabilities or

incurable diseases, information and claims of the persons involved, actual condition and theoretical or practical study of discrimination against disabled persons, etc.
*When medical personnel and client have the same opinion, the problem is "solved" and "smooth determination of strategy" is feasible. However, if the medical personnel lack understanding or have a biased view, would this be acceptable?

The "intent" and "emotion" the clients express should definitely be a realistic appeal even if they are based on insufficient information and biased views. They could be an earnest claim with no other choices, involuntary choice, or difficult decision. Medical personnel are not allowed to deny or demand a retraction of the choice the clients make, but should respect in principle the right to self-determination and the freedom of choice of the clients. Nonetheless, if the above questions are considered as an infringement and interference of the autonomy of the clients and turned down, various multi-layered meanings of the "emotion" and in particular of the "pain" could be overlooked.

2. Various Forces surrounding Clinical Settings of Genetic Medicine: Frame of Context-sensitive Approach

Here, with regard to evaluation, judgment, choice, and determination when facing individual intent and emotion and specific issues in clinical medicine, I organize the context as a factor that supports and contributes to such considerations.

(1) Influence of the general concept of values and system conditions in society on individual choice and determination:

The "pain" and "fear" shown with actual examples in the preceding section assume that "engagement to a woman who may bear a child with a genetic disease will be broken," that "disabled people will be shunned by people," and that "patients with incurable diseases will have to struggle to live if they have no supportive family." Are these assumptions evident "facts"? Shouldn't we ask how such thoughts came to be formulated in the first place and whether these thoughts are the only ones that people could have? It is in fact a prevailing perspective that, although in an ideal world people should not be treated unfairly with discrimination or biased views even though they have a disability or incurable disease, real life is harsh. However, we cannot derive from such a perspective the conclusion that we must embrace real life as an unavoidable reality. We should focus on the discrimination and prejudice that run deep within general public, the mechanics of exclusion and avoidance in society against people with disabilities or incurable diseases, and insufficient support in their lives (medical care, welfare,

education, employment, etc). We then need to examine the influence of these factors on the choice and the determination in each area of clinical medicine.

(2) Influence of individual choice and determination on society as a whole:

If choices and decisions made in clinical genetic medicine accompany assessment and judgment of "value and quality of life," they could have an impact on social systems and policies and people's daily practice. Of course, most of the parties involved do not have such understanding. Some may claim that, if one thinks the choices and decisions have such impact, then he/she should present objectively-verifiable factual data. However, I think the claim that assertions with no visible evidence can be ignored justifies the current situation and lacks a critical view of reality. And I think the "choices and decisions of patients or clients" that the claim supports would treat the "selection of life" only as a "private matter," and stand with no critical view on the current circumstances that are not supportive enough for people with disabilities or incurable diseases, encouraging and reproducing a negative situation. This indicates the necessity of paying attention to the context following this line of thought.

(3) Reflective communication:

The degree of "seriousness" of genetic diseases or disabilities is often a problem for preimplantation diagnosis and prenatal diagnosis to prevent the diseases and accompanying disabilities at the stage of reproduction. If a negative test result is expected to lead to terminating embryo or fetus, the diagnosis irresistibly involves assessment of life values, namely the determination of whether that life is worth living or not. Some say the persons (parents-to-be) who actually cares for the child should conduct the assessment and society including the government and academic associations as well as third parties should never even enter the argument. It is in fact a principle that the selection and decision patients and clients make should be respected and supported ultimately, but could any selection and decision be approved as "autonomous decision making of the persons involved" or as "reproductive freedom and rights"? A reflective communication process, e.g. clinical ethics committee, would be necessary for identifying a certain principle and creating an opportunity for careful consideration through the repeated exchange of information and opinions, including the seriousness of genetic diseases, quality of life and social support systems, between clients and clinical staff.

(4) Public communication:

Each decision made in clinical medicine is required to observe the rules (laws or guidelines) of the government or academic associations, or even rules of international society if the decision involves special themes such as clone technology. Therefore in the process of establishing the rules, public opinion, and the various opinions of clients and patients should never be ignored. In particular, when the decisions involve intervention in a specific quality of life, objections made by people who live with such a quality of life (patients and their families, disabled people and

their families) should have significant meaning regardless of whether the public seriously approves or disapproves of the objection, or just ignores it. How should we evaluate a social cost theory or an eugenic viewpoint which asserts that people who have the causative gene of a genetic disease should take a genetic test and should not give birth to a disabled child that will raise the social welfare cost, and that if they do decide to have the child, they should care for the child without relying on social welfare? Of course, what I mean by "eugenic" is not the improvement of genetic quality at the population level conducted by the government, but a new type of eugenics that intervenes in the genetic quality of children as an individual spontaneous choice.[2]

3. Decision Making Process as Context: Case Oriented Discussion

In a usual process, patients or clients and their families visit medical institutions with their own problems, receive necessary information, and make a decision through various communication processes and considerations. Below I present four model cases that lead to such a process for making decisions on reproduction, and conduct a context-sensitive study of the cases.

A. Premarital diagnosis

In genetic counseling, there are some cases of people who intend to marry being concerned about not only their genetic quality, but also the quality of life of their child-to-be. This issue also involves societal conditions and the public concept of values.

[Case 1] Female in her late 20s. She has several relatives who have probably-congenital mental impairments, and suspects that she possesses the causative genes. She has a fiancée and worries that she might pass on that impairments to their children. She therefore wants to undergo genetic test. If she has the causative genes, she does not want to inform anyone about it and will break the engagement without explaining why because the fiancée and his family place significance on family lineage and blood line.

"Family line," "family lineage" and "blood line" are concepts that prompt special emotions regarding "inheritance," and are deeply inherent in Japanese culture and customs. Expressions such as "shame on the family" or "person with ambiguous origin" imply the thought that "we do

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[2] cf. Shimoda, M., Designing Life and New Eugenics, in: *Challenges for Bioethics from Asia*, Eubios Ethics Institute, ed. by D.R.J. Macer, pp.312-318, Nov. 2004; Paul, D. Genetic Services, Economics, and Eugenics, *Social Context*, 1998;11(3-4):481-491; Resta, R.G., Eugenics and Nondirectiveness in Genetic Counseling, *Journal of Genetic Counseling*, 1997;6(2):255-258; Resta, R.G., Eugenic Considerations in the Theory and Practice of Genetic Counseling, *Social Context*, 1998;11(3-4):431-438.

not want to introduce strange genes into our family." "Strange genes" mean in most cases genes that may induce genetic diseases or mental, intellectual or physical impairments. These views of heredity are criticized in principle as discrimination and biased views, but when people are actually party to such cases, most of them approve those views, believing that "fundamentally everyone must hold such views." The stigma of a woman who "may bear a child with inherited intellectual impairment" could induce her strong anxiety and fear of discrimination and biased views, while she might accept it as an "inevitable fact" and make an autonomous decision to withdraw from a marriage. It is now asked whether we should regard this as a "private matter" or as "an issue that society as a whole should tackle."

B. Preconception diagnosis

It is widely approved as a reproductive freedom and right, and as happiness-pursuing rights to determine by contraception, birth control, and fertility treatment whether, when, and how to have a child. However, whether these rights include that of selecting the quality of life of the child, i.e. that of determining what type of child to have remains a controversial issue. Perhaps the answer lies in the meaning of the parent's selection of desired/undesired quality of life of the child-to-be.

[Case 2] A couple in their 30s, whose first child has an incurable nervous disorder, wish to have a second child. They came for genetic counseling to ask about the risk of the second child developing the same disease. They state that they would abandon conception if the risks are high. Although they love and care for their child with the incurable disease, they could not do the same for another child, and therefore they hope to be able to have a healthy child.

Through the care for their child with the incurable diseases, the parents might have experienced the warm/cold stares of people, the effectiveness/insufficiency of social support systems, and the value/problems of the parents' association and parties involved. They could make a decision by stepping back from the dominant concept of societal values, and we should take their decision seriously. Nevertheless the selection of quality of life and the context behind the selection must be examined independently from their intents amd circumstances. In particular, we have to carefully assess the social impact of views such as "caring for a child with an incurable disease is tough" and the "actual experience of the party involved should be respected."

Here we should pay attention to the fact that the genetic makeup of "the one who has not been born yet" is used as information for life planning of the parents-to-be, and that the meaning of the existence of the unborn baby is determined by the parents. This fact indicates the possibility of enhancing and strengthening the desire for others who are controllable, namely the desire that they should be exactly what we hope for and should not be something that will bother

or trouble us.

C. Preimplantation diagnosis and embryo selection

Preimplantation diagnosis is a method to examine the risk of a specific genetic disorder by collecting cells from in vitro-fertilized embryos and conducting various genetic tests. Usually "normal" embryos are selected according to the test result and placed back in the uterus for conception and birth, while "abnormal" embryos are frozen or disposed of. The selection patterns can be classified into two types: "negative or avoiding" and "positive or desiring." The former includes sex selection to prevent X-linked heredity diseases, the prevention of chromosome abnormality due to habitual miscarriage and late pregnancy, and the prevention of a person with a genetic disease or causative disease from passing on such problems to their children. The latter includes sex selection for non-medical purpose, donor baby for therapeutic purposes (e.g. having a sibling by selecting a high-histocompatibility embryo to conduct umbilical cord blood transplant for an elder brother/sister with leukemia), and having a child who has the same function or form as parents with deafness or achondroplasia.

[Case 3] A couple in their 30s, whose first child has an incurable disease due to a chromosome abnormality, want to have a second child. They came for genetic counseling to ask about the risk of the second child developing the same disease. If the risk is high, they want to undergo in vitro fertilization and conduct genetic test to select an embryo with no possibility of disease for conception and birth of a child.

Some approve of the selection in such cases because of the lack of other methods, compelling reasons of the parents, and the hope of having a healthy child. Namely, they emphasize individual freedom of choice or the right to pursue happiness. On the other hand, some disagree with the notions of the "selection" and the "happiness" in view of equality of life values, claiming that the selection should not be made arbitrarily and that having an incurable disease or disability should not be recognized as an unhappy state. The client of case 3, however, may not be concerned with this kind of conflict, often observed in medical research and clinical medicine that intervene in the quality of life.

The Japan Society of Obstetrics and Gynecology stated in its bulletin "Remarks on preimplantation diagnosis" (revised in 1999) that the preimplantation diagnosis should only be used for serious genetic diseases and the condition of "serious" genetic disease should be examined in individual cases because of divided views on the condition. The client would also not be concerned with the public communication that discusses whether regulation with a uniform standard in a law or guideline is appropriate or not, and whether each medical institution should create their own guidelines or not.

Nonetheless, I think it absolutely vital to provide clients with an opportunity to reflect their intent, and to provide medical personnel and, if necessary, non-medical experts and the parties involved, with an opportunity for discussing the conflict between the freedom of choice or the rights for pursing happiness and the equality of life values, and for assessing how "serious" the genetic disease is. This is because decisions and agreements made in individual clinical medicine are not final and should be updated constantly in a communication process for reflection.

D. Prenatal diagnosis and selective abortion

Many research results on this topic have been accumulated inside and outside the country particularly in connection with "disability." In this section, I would like to limit our view to the context of public communication for discussing medical tests that may result in selective abortion.

[Case 4] Female in her late 30s. She saw a poster in a hospital when she was pregnant and asked for a maternal serum marker screening. The result was positive and she was recommended by a physician to have amniocentesis. She became worried and studied related matters on the Internet. She is now thinking of having a genetic test of embryonal or fetal cells. She and her husband have decided to terminate pregnancy if any abnormality is found.

In the report on prenatal diagnosis "Remarks on maternal serum marker screening" submitted in 1999 by advisory panel to the Health and Welfare Minister, medical doctors are requested not to recommend the test but to conduct it with a sufficient explanation only if consulted by pregnant women, under the current circumstances in which there is concern about the test conducted without full understanding of its meaning as well as about insufficient specialized counseling systems. Some may say patients or clients have the right to choose whether they should take a genetic test and other various medical tests to obtain data necessary for making a selection, and that the government, academic associations, and medical institutions should not limit that right. Also, the client in case 4 may be embarrassed by the reason, for which the report suggested restricting the use of the test, that "a part of the technology might lead to the elimination of fetuses with impairments and to the denial of respect for the right to live and the life of disabled people." On the contrary, if the government or specialist groups take a clear stance of creating policy to promote the prevention of the birth of impaired children, and even if the birth prevention is not forced but promoted based on client's spontaneous choice, the social trend that "everyone should have prenatal diagnosis and it is irresponsible to give birth to a impaired child without having the diagnosis in advance" could become stronger and invisible

pressure would be placed upon people.[3]

The parents-to-be may think it questionable to claim their compelling demand would lead to denying the human rights of disabled people and it not reasonable to intervene in their right of choice for such unjustified reasons. Still, we should not overlook the inevitable involvement and influence of the choice and decision of the persons involved in the social dynamics.

Conclusion

Adult presymptomatic diagnosis has often been conducted in genetic counseling in recent years. Some problems have been pointed out in the diagnosis such as conflict between the client's privacy right and the relative's right to know, and the client's right "not to know" about incurable diseases. We can regard the problems as a context of the autonomy, adequacy and limit of an individual's choice and decision, although further discussion of this is not pursued here.

There is, in the name of "reproductive freedom and rights," a move to approve not only assisted reproductive technologies, but also preimplantation diagnosis and embryo selection, prenatal diagnosis and selective abortion, surrogate birth, and even the production of cloned humans.[4] Supporters of this movement regard the freedom and right of choice based on the client's autonomy as a "private matter" of life planning and the pursuit of happiness. As discussed in the above, however, we should not overlook the fact that individual assessment, judgment, choice, and decision could reflect, support, strengthen, perpetuate, and reproduce the social concept of values in the network (= context) of desires such as "those who may bother or trouble others should be eliminated if possible" and "any child we have should be the one that we wish for." Not only medical personnel, patients, and clients, but also the public could deliberate and reflect by recognizing the context of emotion and intent expressed in individual clinical medicine.

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[3] cf. Wertz, D., Eugenics is Alive and Well: A Survey of Genetic Professionals around the World, *Social Context*, 1998;11(3-4):493-510.

[4] cf. Robertson, J.A., *Children of Choice: Freedom and the New Reproductive Technologies,* Princeton U.P., 1994; Harris, J., Rights and Reproductive Choice, in: J. Harris and S. Holm(eds.), *The Future of Human Reproduction*, Clarendon Press, 1998.