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<td>特別報告：長崎県における遺伝的カウンセリングシステム：長崎大学病院遺伝学分野の経過と現在の状況</td>
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Special Report

A Genetic Counseling System in Nagasaki Prefecture: The Course and Current Status of the Genetic Counseling Unit in Nagasaki University Hospital

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Recent progress in genetic medicine is remarkable and seems to be getting ahead of the general population. For a proper application of genetic medicine to people, genetic counseling is essential. There are few institutions that can provide sufficient genetic counseling in Japan. In response to a proposal by the Ministry of Health, Labour and Welfare to establish a genetic counseling system, Nagasaki prefecture started the genetic counseling model project in 1999 and entrusted Nagasaki University Hospital to become its core, a genetic counseling center. At the same time, Nagasaki University Hospital set up the Genetic Counseling Unit as an independent clinical division to respond the social needs. We describe here the course and current status of the trial to establish a district-adhered genetic counseling system in recent two years in Nagasaki prefecture.

Key Words: genetic counseling, Genetic Counseling Unit, Nagasaki prefecture, Nagasaki University Hospital, district-adhered genetic counseling system

Genetic medicine in Japan

The whole human genomic base-sequences will be read by 2003 in the Human Genome Project. The human genome analysis has clarified the causes of most Mendelian disorders or single-gene defects. It is focusing now on genes or loci susceptible to malignant tumors and common diseases, such as hypertension, diabetes mellitus and cardiac infarct, as well as to resistance to infections and sensitivity to drugs. Such knowledge will be used for future medicine and also for health care. Genetic testing has been applied to many inherited diseases, organ transplantation, and gene therapy. Thus, genetic information, if used appropriately, is very useful for individual health care, e.g., prevention, early diagnosis and early therapy of genetic diseases, becoming a clue for "order-made medicine" in the near future.

The humans have continuously received such genetic information from their ancestors, shuffle it to make diversity, and then transmit it to their progeny. On the other hand, a part of the information of an individual is shared with his/her family members, even in part with unrelated persons. The privacy of an individual, who is a member of the human society, sometimes has a genetic disease as a result of the gene diversity and lives in a family sharing genetic information, is an important issue, although, it has not enough been discussed in Japan because of the social climate and the paucity of genetic education.

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needs much effort to enlighten people on the knowl-
edge of such progresses, not to mislead them, and to
let them decide by themselves on the basis of accu-
rate and recent information. Education of genetics is
also absolutely essential from early age. WHO has rec-
ommended such compulsory education of genetics
with examples of human diseases from primary school
as a subject of science.

Under such a situation, the Japanese government
(Ministry of Education, Culture, Sports, Science and
Technology; Ministry of Health, Labour and Welfare;
Ministry of Economy, Trade and Industry), as well as
academic societies (The Japan Society of Human
Genetics, and seven other medical genetics-related so-
cieties) have presented "guidelines" on human genome
and gene research, or on genetic testing of inherited
diseases. However, it may take time to keep people
well informed. Many patients and/or their relatives in
Japan still accept genetic testing without enough in-
formed consent, are not given any exact genetic infor-
mation on the results, or leave without genetic coun-
seling on their future life or family planning
according to the results. One of the most essential
things concerning medical examinations and treat-
ments of genetic diseases is to assure the comprehen-
sion and free choice from several capable future ways
based on genetic counseling. It is also very important
to prepare psychological and social supports in the
daily life. However, a questionnaire survey against
families with chromosome abnormalities in Nagasaki
prefecture showed that 70% of families had not been
given genetic counseling and 80% felt its necessity.
Considering such a situation in Japan, we, all staff
members of the Genetic Counseling Unit, Nagasaki
University Hospital, agreed with the establishing of a
genetic counseling system as an urgent subject, and
thus is setting up such a system in Nagasaki prefect-
ure.

Genetic counseling model system in Japan

In 1998, the Ministry of Health, Labour and Welfare
(MHLW) of Japan received an application for estab-
lishing a genetic counseling system in the context of
social needs for genetic counseling. The purpose of
this proposal is to establish genetic counseling centers
as a core of providing genetic information in every
prefecture and to decreasing mental burden of clients
and their family members, because there are few pro-
fessional institutions for genetic counseling. The sub-
ject of the project is each prefectural government, and
its contents are to offer genetic information and
genetic counseling in cooperation with public health
centers, welfare offices, medical institutions and med-
ical associations in the prefecture. Its annual cost is
shared between MHLW and the prefectural govern-
ment by one-third and two-thirds of the total year
budget, respectively. Among three prefectures, i.e.,
Nagasaki, Saitama and Tokushima prefectures, finally
applied to and agreed with the project, it was first
being proceeded by Nagasaki prefecture. The goal in-
tended by Nagasaki prefecture is to establish a dis-
trict-adhered genetic counseling system, and a project
committee was started to discuss it periodically. The
system aimed is to establish close and functional con-
nections among the three following units, i.e., the cen-
ter (Nagasaki University Hospital), medical institutions
(practitioners) and districts (public health nurses who
front clients' personal life supports), and to make con-
sultations by clients without appointments easy. As
an internet-mediated network had already been con-
structed between the Prefectural Office and public
health centers and between the Prefectural Medical
Association office and its members (medical institu-
tions), the three units was integrated. However, be-
cause there was no network between health nurses,
the "e-mail address" of the genetic counseling center
was informed to them to be able to exchange informa-
tion. Consequently, clients can receive careful supports
individually.

We intended to recommend that primary genetic
counseling is performed by public health nurses and/or
medical practitioners, and secondary genetic coun-
seling by the center (professional counselors). As it
was unclear how many cases the practitioners and
public health nurses deal with a year, we first investi-
gated the needs of genetic counseling by analyzing questionnaires sent to all 390 public health nurses in
Nagasaki prefecture. The replying rate was 90 %, and
the results showed that about 80 % of public health
nurses had been consulted by people who had certain
 genetic diseases. In addition, 90 % or more of the
nurses felt difficulties how to respond to these consul-
tations, and many hoped to be assisted by a genetic
counseling center through a genetic information net-
work. We next directly visited 10 public health cen-
ters in the prefecture to inspect them and to train the
nurses on genetic counseling using a "guidance" re-
garding the outline of the National Genetic Counseling
Model project, basal knowledge of genetics, Q&A, and
how to introduce a client to Nagasaki University
Hospital for secondary genetic counseling.
**Genetic Counseling Unit in Nagasaki University Hospital**

The human rights based on four ethical principles, "autonomy", "nonmaleficence", "beneficence", and "justice" have been recognized to be respected also in the field of genetic medicine. Under the situation, several guidelines for genetic medicine have been presented by the government and academic societies, as mentioned above. These promoted us to establish a counseling system as a medical and social practice in Nagasaki University Hospital. We have constructed such a system in our hospital, and genetic counseling has been carrying out in an outpatient clinic of pediatric department as a part of pediatric medicine since 1986. However, it has been widely recognized that the counseling system should be involved in many other medical fields, because genetic diseases are dealt with by every field of medicine, and so-called common diseases, such as hypertension, diabetes, cardiovascular diseases, and many cancers, are also related to susceptibility genes as a basic mechanism for their onset. Therefore, Genetic Counseling Unit was set up and is working from April, 2000, as an independent division in the hospital by a collaboration with many medical doctors in different fields. The Unit is composed of Director, Vice Director, and a receptionist who receives appointments from clients. Its operative issues are being discussed in Management Committee. Staff conference consists of counseling staff, representatives from each clinical department, nurses, geneticists and a moral philosopher. Most these doctors concerned have the Japanese Board of Clinical Geneticists approved by the Japan Society of Human Genetics or by the Japan Society of Genetic Counseling. After presentation of cases by clinical geneticists/counseling staff members, principles of counseling and ethical problems are discussed in each case. When recognized to be important, the ethical problems are asked the Ethical Committees of Nagasaki University School of Medicine to be judged.

The actual flow of genetic counseling is as follows: appointment -> first counseling (pre-counseling) -> second counseling -> following-up, if necessary. When a client is introduced from a public health nurse or a medical institution (or clients sometimes contact directly the Genetic Counseling Unit), the receptionist asks the client about the outline of his/her consultation and arranges the date and time of the client's visit. The counselor(s) collects the most recent information concerning the client's problems by his/her visiting date. At the pre-counseling, after a pedigree tree is made and client's information is collected, the counselor(s) counsel the client for at least one hour, then, if necessary, the client's problem will be subjected to second counseling. By the second counseling, the counselor presents the problems (ethical problems, indication for genetic testing, and etc.) at the staff conference for discussion. According to the discussion and conclusion, the second counseling is held. The third or fourth counseling is done, if necessary, while the counseling is often closed at the first counseling, especially in the case of simple and/or less severe problems.

Genetic counseling is held on every Tuesday and Wednesday afternoon. The counseling room is soft-lighted and calm, and is flowed by soft background music, being considered for clients to be able to relax. The medical sheet (record) is made for the Unit independently from other outpatient clinics and stored in a shelf with key to protect the client's privacy. The fees of the counseling are 5,000 yen for the first counseling, and 2,500 yen for each subsequent counseling, which have been approved by the Ministry of Education, Culture, Sports, Science and Technology.

**Current status of Genetic Counseling Unit**

One hundred and twenty clients have visited for one and half year since the opening of the Unit. Most of them are habitants within Nagasaki prefecture and 15% of them came outside the prefecture. Subjects or diseases in the counseling at the time point of 97 visitors were categorized in the Table. One of the most

<table>
<thead>
<tr>
<th>Subjects and diseases</th>
<th>Number of cases</th>
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<tr>
<td>Chromosome abnormality</td>
<td>22</td>
</tr>
<tr>
<td>Congenital malformation</td>
<td>18</td>
</tr>
<tr>
<td>Neurological and muscular disease</td>
<td>19</td>
</tr>
<tr>
<td>(Spinocerebellar atrophy)</td>
<td>(5)</td>
</tr>
<tr>
<td>Psychiatric disorder</td>
<td>5</td>
</tr>
<tr>
<td>Congenital metabolic disease</td>
<td>4</td>
</tr>
<tr>
<td>Visual or auditory disorder</td>
<td>4</td>
</tr>
<tr>
<td>Hematological disorder</td>
<td>2</td>
</tr>
<tr>
<td>Familial tumor</td>
<td>1</td>
</tr>
<tr>
<td>Consanguinity</td>
<td>1</td>
</tr>
<tr>
<td>Advanced-age pregnancy</td>
<td>13</td>
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<tr>
<td>Drug usage during pregnancy</td>
<td>3</td>
</tr>
<tr>
<td>Others</td>
<td>5</td>
</tr>
<tr>
<td>Total</td>
<td>97</td>
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important problems was a pre-symptomatic diagnosis of an adult-onset, inherited, neurological degenerative disease without any available effective therapy, such as inherited spinocerebellar atrophy. Advanced aged pregnancy is counseled generally with a gynecological geneticist, and patients with chromosome abnormalities and/or congenital malformations are followed up in the outpatient genetic clinic of Pediatric Department by pediatric geneticists.

To provide genetic information, a home page of the Genetic Counseling Unit was made to link to the home page of Nagasaki University. There has been a numbers of access. On requests for genetic information by telephone, facsimile or E-mail, general information is given without referring about privacy. As a part of the genetic counseling model project, the number of visitors, current news of the Unit and some recent topics of genetic medicine are provided periodically to public health nurses.

The most important problem of our unit is that no psychological support system has been established. No clinical psychologists are assigned in Nagasaki University Hospital, therefore, some problems may arise in the near future. We have earnestly demanded to the Ministry of Health, Labour and Welfare to increase the number of staff members. In order to try to solve ethical problems that may occur in the process of counseling, genetic ethics seminars are opened, and ethical problems of clients are periodically discussed and investigated, together with professionals of philosophy, bioethics, psychiatry and education.

A goal of genetic counseling is to support clients' decision making for their lives through the process of non-directive counseling without any moral and material compulsion. Genetic information should be put on view and genetic diseases should be disclosed to the general public. Under only this situation, clients become to make self-decision easily. All staff members in Genetic Counseling Unit in Nagasaki University Hospital will make further efforts to contribute to better genetic counseling.

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