

## Special Report

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

Tadashi MATSUMOTO<sup>1,5)</sup>, Tatsuro KONDOH<sup>2,5)</sup>, Norio NIKAWA<sup>3)</sup>, Noriko MAEDA<sup>1)</sup>, Tadayuki ISHIMARU<sup>4,5)</sup>

1) Department of Nursing, Nagasaki University School of Medicine

2) Department of Pediatrics, Nagasaki University School of Medicine

3) Department of Human Genetics, Nagasaki University School of Medicine

4) Department of Gynecology and Obstetrics, Nagasaki University School of Medicine; 5) Genetic Counseling Unit, Nagasaki University Hospital, Nagasaki Japan

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.

ACTA MEDICA NAGASAKIENSIA 46 : 7-10, 2001

**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 hyperten-

sion, 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.

Gene analysis has exclusively developed in recent years, while some research institutions or commercial laboratories where genetic tests are done take precedence of research or benefit over the personal privacy and variety of ethical problems, so that there have been big social problems. Recently advanced cell-engineering technologies including the handling of embryonic stem (ES) cells, and several reproductive technologies for infertility evoked also the ethical/legal issues. Such rapid progresses in genetic technologies and their derived problems may cause a negative mood against the whole scientific genetic-research in Japan. Consequently, the dark image against inherited diseases has not been erased completely. Thus, it

**Address Correspondence:** Tadashi Matsumoto, M.D.  
Department of Nursing, Nagasaki University School of Medicine, Sakamoto 1-7-1, Nagasaki 852-8523, Japan  
TEL: +81-95-849-7924 FAX: +81-95-849-7924  
E-mail: [pediatr@net.nagasaki-u.ac.jp](mailto:pediatr@net.nagasaki-u.ac.jp)

needs much effort to enlighten people on the knowledge of such progresses, not to mislead them, and to let them decide by themselves on the basis of accurate and recent information. Education of genetics is also absolutely essential from early age. WHO has recommended 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 societies) 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 informed consent, are not given any exact genetic information on the results, or leave without genetic counseling on their future life or family planning according to the results. One of the most essential things concerning medical examinations and treatments of genetic diseases is to assure the comprehension 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<sup>1)</sup>. 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 prefecture.

### **Genetic counseling model system in Japan**

In 1998, the Ministry of Health, Labour and Welfare (MHLW) of Japan received an application for establishing 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 professional institutions for genetic counseling. The subject 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 medical associations in the prefecture. Its annual cost is shared between MHLW and the prefectural government 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 intended by Nagasaki prefecture is to establish a district-adhered genetic counseling system, and a project committee was started to discuss it periodically. The system aimed is to establish close and functional connections among the three following units, i.e., the center (Nagasaki University Hospital), medical institutions (practitioners) and districts (public health nurses who front clients' personal life supports), and to make consultations by clients without appointments easy. As an internet-mediated network had already been constructed between the Prefectural Office and public health centers and between the Prefectural Medical Association office and its members (medical institutions), the three units was integrated. However, because 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 information. 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 counseling by the center (professional counselors). As it was unclear how many cases the practitioners and public health nurses deal with a year, we first investigated the needs of genetic counseling by analyzing questionnaires sent to all 390 public health nurses in Nagasaki prefecture<sup>2)</sup>. 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 consultations, and many hoped to be assisted by a genetic counseling center through a genetic information network. We next directly visited 10 public health centers in the prefecture to inspect them and to train the nurses on genetic counseling using a "guidance" regarding 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<sup>3)</sup>, "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.** Subject for Genetic Counseling

Subjects and diseases	Number of cases
Chromosome abnormality	22
Congenital malformation	18
Neurological and muscular disease (Spinocerebellar atrophy)	19 (5)
Psychiatric disorder	5
Congenital metabolic disease	4
Visual or auditory disorder	4
Hematological disorder	2
Familial tumor	1
Consanguinity	1
Advanced-age pregnancy	13
Drug usage during pregnancy	3
Others	5
Total	97

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.

#### References

- 1) Yamaguchi K, Kondoh T, Matsumoto T, et al: A questionnaire survey concerning genetic counseling. Part I: Against families with chromosome abnormalities. *Nihon Shonika Gakkaizassi* (submitted, Jpn)
- 2) Yamaguchi K, Kondoh T, Matsumoto T, et al: A questionnaire survey concerning genetic counseling. Part II: Against public health nurses. *Nihon Shonika Gakkaizassi* (submitted, Jpn)
- 3) Wertz DC, Fletcher c, Berg K, Boulyjenkow V: Guidelines on ethical issues in medical genetics and the provision of genetics services. World Health Organization 1995