Patient's Guide for Informed Consent

Forwarding Your Diagnostic Sample and Information to Other Facilities

If you have consented to the preservation of your sample at NCNP, and with this sample if you wish to receive other facilities' diagnostic services which are not available at NCNP, please submit the request form "Request for Forwarding Diagnostic Sample" to us. We will forward your sample in accordance with your request. Similarly, if you wish to obtain your diagnosis result once again or if you have changed a hospital, please submit the request form "Request for Diagnostic Information" to us. Both forms are required to confirm the consent by you or your authorized representative. Please contact your attending physician or us at the Sample Reception Desk of the MGC if such a need has arisen.

Preservation / Research Use

The cause of many neuromuscular disorders is still unknown and no effective cure is available. The promotion of researches using your sample and clinical information (including clinical history and findings) is indispensable for the fastest elucidation of disease causation and development of new medicines. NCNP as well as many domestic and foreign research institutes are conducting researches on disease causation and treatments. Therefore, an individual's sample, including your sample and clinical information, is considered a valuable research resource for the discovery of remedy for neuromuscular disorders. We would like to ask for your understanding and cooperation on this. Of course, even when you do not consent to the research use, there is no disadvantage for you at all. Also, you may cancel the research use at any time. Similarly, consenting to the research use does not affect the volume of sample collected from you nor require additional collection.

1. Purpose of Our Research

Even when you consent to the research use, we utilize your sample and clinical information including clinical history and findings only for the purpose of "the elucidation of pathomechanism and the development of therapy for neuromuscular disorders". These researches sometimes require genetic analysis to identify a disease causation. However, the use of obtained information is strictly limited to the above mentioned research purpose.

2. Protection of Your Personal Information

Upon conducting an analysis, as a principle we give each sample a number, and your identifiable personal information is removed as much as possible. However, it is necessary to verify the correct patient is matched with the correct sample number so we can send out an analysis result. For this purpose, we record your identification into a comparison list prior to anonymization. The personnel responsible for the administration of this comparison list is called the Privacy Officer, and the Director-General of the Translational Medical Center is responsible for this role. The names of Subofficers responsible for conducting the actual operations are listed at the end of this handbook. When we outsource your diagnostic analysis to an external facility, we will never provide your personal information.

Additionally, in case we use your sample for researches based on your consent, your anonymized sample and information goes through a second anonymization process where the initially anonymized sample and information is replaced by another number. This anonymization allows us to promote clinical researches through comparing clinical information to a research result. In case of a joint research with other facilities, we only provide such second-anonymized samples and information. When various types of data such as analysis results and

clinical information are presented for an academic and educational purpose, such information is completely anonymized in any reference materials.

If you wish, we shall contact you individually to disclose research achievements in the future, however we are not able to specify when it would happen.

3. Confirmation of Diagnosis in the Future

Even when a definite diagnosis is not available for you at this moment, with your sample the diagnosis might become available in the future. In this case, we will report the diagnosis result to your attending physician. However, we might withhold the report in these circumstances : 1) when your contact address is not available anymore due to reasons such as too much time has passed since the submission of the first sample, or 2) when disclosure of diagnosis result is likely to harm you or a third party's life, body, property and other rights and interests. Also, with regard to the disclosure of genetic information to a minor, we will discuss with the patient and/or their authorized representative, as well as refer to the advice by our Ethics Committee, in order to decide whether or not to disclose, and ways to disclose such information. Upon informing you about a new diagnosis, if the new diagnosis is based on a genetic analysis, we will contact your attending physician so you are provided with genetic counselling before and after the diagnosis. Our genetic counselling specialist can also help you.

4. Deposition of Genetic Sequencing Data in Public Databases

Various large-scale genetic analyses performed at NCNP are in most cases supported by various public research funding such as AMED funding (Japan Agency for Medical Research and Development). Hence, in principle, any genetic data obtained through sequencing is to be deposited in a public database. The type of data which does not reveal an individual's identity, namely variant frequency information as to how frequent the detected genetic change is observed in the general population or in a group of patients with a same disorder, is deposited into open databases, such as the database specified by AMED and others so that the data is utilized by an unspecified number of researchers. On the other hand, an individual's detailed genetic sequencing data is, together with disease information, deposited into a closed database specified by AMED and others. Any personal identifiers such as name, address, contact number and hospital ID are completely removed prior to such data registration. However, please understand that, even when you decide to withdraw your consent, removing your data from such databases might not be possible anymore since the research has been proceeded through the shared information.

5. Range of Use of Your Sample and Clinical Information

Your sample and clinical information is used only for the purpose of "the elucidation of pathomechanism and the development of therapy for neuromuscular disorders" at NCNP and other domestic research institutes. If you consent to the use of your resource by foreign research institutes, please check the appropriate box. This enable us to utilize your resources for international joint research projects.

6. Offering Your Resource to Public Biobanks

Public biobanks are the public organization with the purpose of storing research resources and making them available to laboratories. In Japan, well-known biobanks include those operated by RIKEN, the Japan Health Science Foundation, and the National Institute of Biomedical Innovation. Your resource offered to these biobanks will be widely utilized for scientific researches. When your resource is offered to biobanks, we will remove your identifiable information from the resource in order that it can never be traced back to you, so we can ensure your privacy is certainly protected. However, in this case none of research result can be fed back to you.

In our consent form you can find a section to confirm whether you wish to offer your resource to such public biobanks for research use. Please let us know your wish.

7. Offering Your Resource to Commercial Enterprises

Most of very new medicines are developed by commercial enterprises, namely by pharmaceutical companies. The research & development activities conducted by such commercial enterprises including pharmaceutical companies are also considered as helping the elucidation of pathogenesis of neuromuscular disorders and the development of new treatments. If you consent to offering your sample to such commercial enterprises, please check the appropriate box.

We provide your sample to commercial enterprises but only for researches which scientific and ethical validity has been acknowledged by an ethical review within NCNP

8. Incidental findings

When such a large-scale analysis is performed, theoretically the result can reveal your vulnerability as well as predisposition to various disorders. In some circumstances, your health-threatening finding could be unexpectedly identified, which is called an "incidental finding".

If you wish to be informed about such a finding, please check the appropriate box. However, even when a large analysis is performed, the subsequent analysis to interpret the raw sequencing data is limited to targeted regions/targeted genes aiming to elucidate disease causation and pathogenesis, therefore, it is not certain that all kinds of health-harming information can be obtained. Also, information such as a patient's vulnerability to various disorders is usually not detected, as our analysis does not intend to serve such a purpose.

Amendment to Your Consent

You may make changes to what you have agreed in the "Consent Form for Sample Analysis, Preservation and Research Use" at any time. Please submit to us the "Request Form to Amend Consent" to us. This form would be provided to you together with the copy of Consent form. If you have any question about this form, please contact our Sample Reception Desk at the MGC.

List of Analysis Conductors (also the Administrators of Samples and Bioresources) and Privacy Officers

Analysis Conductors and Administrators of Samples and Bioresources：

Ichizo Nishino (Director of the Department of Genomic Medicine Development of the MGC, Director of the Department of Neuromuscular Research, Clinical fellow at the Genetic Diagnosis Unit of the National Center Hospital)

Narihiro Minami (Researcher of the Department of Genomic Medicine Development and the Department of Neuromuscular Research, Medical Technician at the Genetic Diagnosis Unit)

Yuichi Goto (Director-General of the MGC, Director of the Department of Mental Retardation and Birth Defect Research, Head Physician at the Genetic Diagnosis Unit of the National Center Hospital)

Privacy Officer : Keiji Wada (Director-General of the Translational Medical Center)

Subofficer : Toshiaki Hirata (Head of the General Affairs Section)

Advisors :

Ichizo Nishino (Director of the Department of Genomic Medicine Development of the MGC, Director of the Department of Neuromuscular Research, Clinical fellow at the Genetic Diagnosis Unit of the National Center Hospital)

Narihiro Minami (Researcher of the Department of Genomic Medicine Development and the Department of Neuromuscular Research, Medical Technician at the Genetic Diagnosis Unit)

Yuichi Goto (Director-General of the MGC, Director of the Department of Mental Retardation and Birth Defect Research, Head Physician at the Genetic Diagnosis Unit of the National Center Hospital)

The above list of personnel is subject to change without notice due to organization and personnel change.

Contact:
National Center of Neurology and Psychiatry
MGC Sample Reception Desk
Address: 4-1-1 Ogawahigashi-Cho, Kodaira-Shi, Tokyo, JAPAN
Telephone: +81-(0)42-341-2711 (Operator), +81-(0)42-346-1770 (Direct)

Approved by Ethics Committee on 24 February, 2017

Modified in conformity with the revised ethical policy, on 30 May, 2017