

ゲノム支援とは
About the Grant-in-Aid for Scientific Research on Innovative Areas
“Genome Science”

Background

“Genome Science” is a part of the “Support Programs for Three Fields in Life Sciences (cancer, genome and brain sciences)” established by the Ministry of Education, Culture, Sports, Science and Technology (MEXT) under the Grant-in-Aid for Scientific Research on Innovative Areas. “Genome Science” is a simplified form of the official title of the program: “Promotion of genome sciences through support for large-scale genome information production and advanced bioinformatics analysis”. This new program is established for continuing to provide support for three areas of life sciences, following the discontinuation of coverage by the MEXT’s program under the Grant-in-Aid for Research on Specific Areas. This is based on a conclusion made by a working group of the Council for Science and Technology: A new mechanism needs to be established to continue the functions previously provided by the General Management Team and Support Team in the earlier framework for supporting research on the three areas.

Objectives of “Genome Science”

The advancement of genome sciences, such as the emergence of next-generation DNA sequencers, is leading to revolutionary progress in life sciences and exploration of new frontiers. Such progress requires large-scale systems for both wet and dry analyses. Genome Science will develop next-generation genome analysis systems, while ensuring the most efficient use of hardware, technologies and human resources prepared under the previous programs, and support research projects that need such next-generation analysis systems to address relevant themes. We expect to receive proposals on important projects that will contribute to the future of genome sciences. Such projects are challenging not only for the researchers who proposed but also those in Genome Science who support the projects: the projects encourage progress in analysis techniques, which then enable even more challenging projects to be tackled. This process brings great opportunities for human resource development, and so we welcome proposals for joint research projects. Genome Science thus assists the expansion and progress of life sciences. While contributing to the development of analysis techniques and human resources, and the establishment of an appropriate framework for sharing resources, Genome Science will accelerate the overall development of genome sciences in Japan.

Outline of “Genome Science” activities

With the General Support Program and three other programs described below, Genome Science offers comprehensive support for wet and dry analysis activities in certain projects. The procedures for selecting these activities include open calls for projects to be supported.

【Large-Scale Genome Information Production Program】

This program supports the production of genome information using next-generation DNA sequencers. This includes support for genome DNA sequencing of humans and various biological species, extensive gene expression profiling, epigenomic analysis and metagenome analysis. The program will also develop technologies to improve the supporting ability.

【Medical Genome Science Program】

This program supports genome typing, genome re-sequencing and medical genome informatics. The identification of disease-related genes requires sophisticated analysis of genome information that depends on the integration of polymorphism information based on highly accurate sequence information and clinical information, thus creating the need for concurrent development of necessary technologies. In addition, the program will develop a database that integrates human genome polymorphism information with clinical information.

【Bioinformatics Analysis Program】

To enhance the benefits from other support programs, this program supports bioinformatics analysis. This includes assembling, mapping and annotating (i.e. identification of biological and medical significances) genome sequence data. The program also supports the development of sophisticated information analysis techniques for identifying disease-related genes. Furthermore, we work with wet researchers to accelerate progress in the development of advanced technologies with a hope that they may lead to an increase in the sophistication of our support.

To complement the three support programs, we assist the genome research community under the General Support Program by arranging academic exchanges and interdisciplinary initiatives, acting as an interface for international partnerships, and strengthening interactions with society through discussions on ethical, legal and social

issues (ELSI), which is indispensable for the progress of genome research.

支援課題の公募要項

Open Call for Research Projects supported by “Genome Science”

“Genome Science” is developing next-generation genome analysis systems, and supporting research projects that need such systems to address relevant themes. Genome Science thus promotes the expansion and progress of genome sciences in Japan. We expect to receive proposals on important projects that will contribute to the future of genome sciences.

1. Research projects covered by the “Genome Science” program

The present open call for projects for the “Genome Science” program covers any project entitled to the Grant-in-Aid for Scientific Research (Kakenhi) in fiscal 2015 (new or continuation). When you apply for a Grant-in-Aid for Scientific Research, you might not have been able to include a genome analysis project or a genome-wide functional analysis project in your Kakenhi project. Therefore, the project to be supported need not necessarily be mentioned in the research protocol you have submitted. Nevertheless, you must ensure that the project is closely related to your Kakenhi project. In your application, you must describe how the availability of additional support under the Genome Science program may lead to important research outputs beyond your original expectations, and how it may contribute to the advancement of genome sciences in Japan. We expect to receive proposals on important projects that will contribute to the future of genome sciences. Such projects are challenging not only for you but also those in Genome Science program: the projects encourage progress in analysis techniques, which then enable even more challenging projects to be tackled. Projects that can be achieved by simply outsourcing tasks will not receive high priority in our support program.

We accept applications from the chief researcher or team member of any project (in any research category) that is covered by the Grant-in-Aid for Scientific Research. There is no limit on the number of applications from a single organization, and there is no restriction on duplication with other forms of support under the Grant-in-Aid for Scientific Research. The Genome Science group members will support selected projects (through consultation with applicants) using the budget for the Genome

Science program. Therefore, research funds will not be distributed to the applicants.

2. Screening of research projects to be supported by “Genome Science”

Candidate projects are screened by the Review Committee, consisting of academics (outside of “Genome Science”) and researchers belonging to “Genome Science”. The screening includes reviewing the eligibility of candidate projects as research-intensive academic projects on genomes that cannot be accomplished by simply outsourcing analytical activities, the feasibility of support in terms of expenses, etc., and evaluating the expected contribution to the development of human resources and sciences. This year, GWAS analysis, DNA sequencing by Sanger method and the preparation of library alone is inapplicable. When supporting medical genome sciences, we also consider the ethical aspects of the project. Each successful applicant will be contacted individually by the Support Group of the Genome Science program to discuss the details of providing support. After the final selection of projects to be supported, the respective applicants will be notified on the results and a list of supported projects will appear on the Genome Science website. Information made available to the Review Committee during the screening process is used solely for the screening. The names of members on the Review Committee will be disclosed in the following fiscal year.

3. Disclosure and Sharing of Data and Resources

In view of the increasing amounts of data generated by research activities, we must ensure that data are not only available for analysis by individual researchers but also are shared with the whole research community. Therefore, we require that, after a certain period after the completion of a project, the research outputs must be shared by means of a database, resource bank or the like. We plan to introduce a mechanism to ensure that personal genome information is handled properly.

Analytical outputs from each supported project must be disclosed by appropriate means (article, database, etc.), and the details of the disclosure must always be reported to us. Base sequence data must be transferred to and disclosed on an appropriate database. Research resources such as libraries must be made available for sharing after the publication of articles (arrangements must be made for distribution to the research community, transfer to a repository, etc.).

On the occasion of article publication, please list that you received support from

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“Genome Science”. The English notation of “Genome Science” is MEXT KAKENHI (No. 211S0002).

Except in the case of base sequence data that contain personal genome information, the delivery of base sequence data to the support recipient must be made at the same time as provisional saving of the data to DDBJ (DNA Databank of Japan at <http://www.ddbj.nig.ac.jp/>) or DDBJ-managed DRA (DDBJ Read Archive at <http://trace.ddbj.nig.ac.jp/dra/index.shtml> for new-generation sequence analyzer data). The deposited data must be disclosed immediately after the publication of articles. Even if no article is published, the deposited data must be disclosed by the end of a pre-agreed period (not longer than one year in principle) after the completion of support.

Regarding the handling of data that contain personal information, please read Details of the Medical Genome Science Program.

Support recipients may be requested to permit the use of analytical outputs under appropriate arrangements, the details of which will be discussed with them, for the purpose of contributing to research activities on developing more advanced information analysis systems. For more information, please read Details of the Bioinformatics Analysis Program.

4. Notices for the preparation of application

Because this is the last year of “Genome Science” program, supporting activities for each project should be completed by the end of March, 2016. After the notice of adoption scheduled on the middle of July, sample should be provided for analysis by the end of December. In case of the delay of sample preparation, we may not support your project. Regarding the project that needs approval from the ethical review committee, the project should be approved by the end of October. After the approval from the committee, specimens should be provided for analysis immediately. Please avoid unnecessary requirement in your proposal.

Please note that supporting activities are collaborative researches between support recipients and support group members. If the delivered specimens are found to be unsuitable for analysis in terms of quantity or quality, the support recipient must discuss with the responsible support group members about what action should be

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taken (cancellation of analysis, re-preparation of specimens, etc.). Support recipients are requested to feedback outcomes of supporting activities to support group members, to encourage progress in analysis techniques. On the other hand, support recipients may ask further analysis to support group members, to expand the research further.

We will attack challenging research projects, such as de novo sequencing of higher organisms with new generation sequencers. However, for the large-sized genome sequencing the new sequencing technology development such as the mate pair sequencing of the long insert by new generation sequencers, is necessary. In addition, de novo assembling becomes difficult, and it is expected a high quality draft sequence is not obtained by the heterozygous frequency, the repeat sequence content and the deflection of genomic GC content. Therefore, support from an applicant and researcher community such as the evaluation of the genomic complexity and the choice of an appropriate strain to be analyzed, becomes essential for the large-sized genome sequencing. Furthermore, support by the researcher community becomes required in annotation after the draft sequence completion. For the project of large-sized genome sequencing, please pay enough attention to these points. Number of challenging projects accepted by our program will be limited, and such projects will be carried out as collaborative research between support recipient and support group members.

Since the Genome Science program has a limited budget, not all requests can be fulfilled. To support as many projects as possible, we may request the beneficiaries of our support program to pay some of the expenses (e.g. consumables).

Even though we will do our best in support activities, we do not guarantee success and there is always a risk of unexpected events. Please also understand that the delivered data may not meet your initial expectation. Support recipients have final responsibility for verifying the contents of data, libraries and other research resources provided by the support group.

5. Period of present open call

Proposals of projects are accepted from April 1st, 2015, to April 24th (at midday), 2015.

6. Starting date of supporting activities

Projects to be supported in 2015 are going to be announced on middle of July, and then supporting activities will start after negotiation between support recipients and support group members.

<p>大規模ゲノム情報生産支援活動 Large-Scale Genome Information Production Program</p>

1. Details of the program

Your project may be deemed eligible for one or more of the following types of support under this program. Subjects such as the choice of sequencer and the target accuracy in determining sequences will be discussed between the support recipient and support staff.

A. Genome sequence analysis

Determination of sequences by means of a new generation sequencer

de novo sequencing and re-sequencing of genome, including partial genome

Re-sequencing by the sequence capture method or the like.

Meta genome analysis, including meta 16S rRNA analysis→

Epigenome analysis (methylation analysis, nucleosome analysis, etc.)

ChIP-seq. analysis

Others (only for the whole genome analysis)

Terminal sequence determination of BAC/fosmid libraries

Determination of complete sequences for individual clones (BAC/fosmid clones only)

B. Transcriptome analysis

RNA-seq analysis by a new-generation sequencer

EST library end sequence determination (in a library created by the support recipient except when a full-length library is used)

C. Library creation (only for the whole genome analysis)

BAC/fosmid library creation

Sheared FOSMID library creation

Full-length cDNA library creation by oligo capping

2. Support-Related Requirements

A. Genome and transcriptome analysis

1. The analysis of medical specimens that involve considerations about personal information and/or ethical issues requires coordination with the Medical Genome Science Program.

2. The analysis of pathogenic microorganisms (BSL2 or above, or equivalent) and poisonous/infectious specimens requires discussion between the support recipient and responsible support group members upon its implementation.

3. In principle, DNA specimens required for sequence determination must be prepared by the support recipient. The amount and quality of DNA specimens must be decided by discussion between the support recipient and responsible support group members. ChIP-seq. analysis, for example, requires that the concentration of DNA by a factor of 4 to 8 is confirmed by real-time PCR for a known protein binding region. If the delivered specimens are found to be unsuitable for analysis in terms of quantity or quality, the support recipient must discuss with the responsible support group members about what action should be taken (cancellation of analysis, re-preparation of specimens, etc.).

4. In principle, RNA preparation for RNA-seq analysis must be done by the support recipient. The responsibility for subsequent preliminary tasks before sequence analysis must be decided by discussion between the support recipient and responsible support group members. The support recipient may be requested also to handle basic tasks such as verification of the presence of expected transcription products by means of real-time RT-PCR,

5. For library sequence determination, the support recipient must submit clones, in principle. Preparation of template DNA for sequence will be done by the responsible support group. Experiments that involve the use of recombinant DNA require compliance with the Law Concerning the Conservation and Sustainable Use of Biological Diversity through Regulations on the Use of Living Modified Organisms. If the proposed experiment requires approval by the supporting organization, support activities will begin only after such approval has been

confirmed.

6. The delivery of yielded sequence data to the support recipient will be made at the same time as provisional saving of the data to DDBJ (DNA Databank of Japan at <http://www.ddbj.nig.ac.jp/>) or DDBJ-managed DRA (DDBJ Read Archive at <http://trace.ddbj.nig.ac.jp/dra/index.shtml>). The support recipient will receive the ID and password to the saved data, and will then be able to access the data yielded by analysis and analyze them using the analysis tools provided by the DDBJ. The support recipient may ask for direct delivery of sequence data, in which case the support recipient must provide the storage media (disks, etc.). Also, even in this case, the Bioinformatics Analysis Group will ensure that the same data is provisionally saved to DDBJ.

B. Library creation

1. The created library, together with the starting specimens, will be handed over to the support recipient. In the case of libraries used in the wider context of complete genome sequencing or if the scope of support includes determining terminal sequences, libraries will be hand over after all processes have been completed. At the time of hand-over, the support recipient may be asked to sign a material transfer agreement (MTA), etc.

2. The type and quantity of specimens to be prepared for library creation must be discussed between the support recipient and responsible support group members.

3. Kits are available to facilitate the creation of cDNA libraries. Such library creation tasks are not covered by our program.

3. Bioinformatics analysis

The Large-Scale Genome Information Production Program covers preliminary activities and primary analytical activities: the preliminary activities include the “base call” procedure along with activities such as the addition of quality scores, while the primary analytical activities include routine mapping and assembling activities by means of analytical pipelines that come with the sequence analyzer or that are prepared by the Support Group. Regarding subsequent activities that are more project specific, such as improving the sequence data (e.g. filling gaps and accurately determining sequences), annotation and advanced statistical processing,

arrangements can be made, depending on the request, for such activities to be done as a collaboration or supported by members of the Bioinformatics Analysis Group.

4. Disclosure and Sharing of Data and Resources

1. Base sequence data from each supported project must be deposited to an appropriate database. The deposited data must be disclosed immediately after the publication of articles. Even if no article is published, the deposited data must be disclosed by the end of a pre-agreed period (not longer than one year in principle) after the completion of support.

2. Research resources, such as libraries, from each supported project must be available for sharing after the publication of articles. Arrangements for sharing (distribution to research communities, transfer to a repository, etc.) must be made by the support recipient.

<p>ゲノム医学支援活動 Medical Genome Science Program</p>

1. Details of the program

Your project may be judged to be eligible for one or more of the following types of support under this program.

A. Genome-wide association study (GWAS)

New proposal is inapplicable this year.

B. Linkage analysis of lineage specimens

Parametric linkage analysis

Model-free linkage analysis (affected sib pair analysis, etc., including typing and the analysis of various linkages)

C. Large-scale re-sequencing of genomes

Next-generation sequencers can be used for exome and target sequence capturing or for large-scale re-sequencing of whole genomes. Depending on the request, we may undertake primary analytical activities (mapping, SNV and short-in/del identification, variation listing, etc.), and may also support

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medical/biological interpretation and statistical analysis (e.g. functional annotation of variations).

- D. Functional analysis of human genome using next-generation sequencers
 - RNA-seq. analysis
 - ChIP-seq. analysis
 - Epigenome analysis

2. Support-Related Requirements

A. GWAS

New proposal is inapplicable this year.

B. Linkage analysis of lineage specimens

The informed consent documents to be prepared are similar to those normally prepared for disease-related gene research projects. In this case, we will not deposit data to any database.

C. Genome re-sequencing

In our support program, we carry out the whole genome analysis and deposit data to the public database. Therefore, the informed consent documents for providers of specimens and research proposal to the ethical review committee of your organization must include items indicated in attached documents in addition to the items required by “Ethical Guidelines for Human Genome/Gene Analysis Research”. You may also consult with model form of informed consent documents for our program.

If informed consent documents you have already obtained does not satisfy these requirements, in principle, you must obtain re-agreement to revised informed consent documents.

D. Transcriptome analysis of human genome by means of next-generation sequencers

Expression profile analysis is not officially addressed by the Three Ministries' Genome Guidelines. However, transcriptome by means of next-generation sequencers, for example, involves handling data that contains sequence

information of individual genome. In such cases, the project must be deemed similar to a human genome analysis project in terms of procedural requirements (informed consent from specimen providers and approval from the ethical review committee).

3. Disclosure and Sharing of Data

The result of supported project (all of following A~C corresponds) will be registered with a public database to be able to help a medical study widely. The information that is not personal distinguishable will be accessible without limitation. In the case of personal identifiable information, approved researcher after examination will be accessible to the data(we call the latter "limited access").

Data obtained by the supported project will be registered with NBDC Human Database (<http://humandbs.biosciencedbc.jp/>) located in bioscience database center (<http://biosciencedbe.jp/>).

Human genome sequence data

A. Frequency data

The frequency data on variations, summarized from the whole body of information on genome sequences yielded by the supported project, will be disclosed at the Bioscience Database Center.

B. Reference genome sequences of the Japanese

The Genome Science program will compile the consensus reference sequences of genomes of the Japanese from the whole body of information on genome sequences yielded by the supported project. When sufficient genome sequence information has been gathered and it is considered that the data contains no information that could identify individuals, the accumulated information on reference sequences will be disclosed as reference sequences of genomes of the Japanese.

C. Genome sequences of individuals

Under controlled access, the genome sequences revealed by the supported

project will be made available for sharing with other researchers. The scope of sharing will be limited to genome sequence data and disease (or control) names. Data sharing requires preliminary review and approval by the ethical review committee of the support recipient's organization, and must comply with the following rules:

1. Data must be made available for sharing after being used for the publication of articles.
2. Even if no article is published, the data must be shared after one year has elapsed since the genome sequence data were delivered to the support recipient.

4. Support-Related Arrangements

1. The specimens and data prepared for analysis must be handed over in a format of anonymization in a linkable fashion (to enable analysis at the supporting organization). The tables of code key (used for making the data anonymous), however, should be kept and properly guarded by the support recipient. The supporting organization will not accept such tables.

2. Sequencing operations that require the protection of personal information should be done using computers isolated from the internet in a room to which access is controlled by biometric identification. Data analysis should be done using computers isolated from the internet in a room to which entrance and exit is controlled.

3. All sequence data is handed over to the support recipient only when constant regime * is maintained about the human genome sequence data handling. Otherwise, only summary data is provided and the data analysis is done by supporting group.

* The regime of human genome sequence data handling: Access to analysis room is controlled. For analysis of a personal genome, the dedicated computer is used. Communication and safety measures from the LAN of your organization is managed appropriately. Please refer to NBDC Human Database Security Guideline (For users) for details. However, it is desirable to use computer not connected to Internet when it is not necessary.

情報解析支援

Bioinformatics Analysis Program

1. Details of the program

The following assumes that we support the analysis of data that have already been collected by the support recipient. However, please note that our support under the Bioinformatics Analysis Program can be complete in itself or combined with other types of support under the Large-Scale Genome Information Production Program or the Medical Genome Science Program. In any of these cases, the scope of our support is subject to preliminary discussions.

A. Support for primary analysis

Draft genome sequencing by new-generation sequencers: mapping, assembly, homology search, and annotation

Microbial genome sequencing: assembly, homology search, annotation, and phylogenetic analysis

Meta genome analysis (including meta 16S rRNA analysis): meta genome gene prediction, homology search, annotation, and phylogenetic analysis

Comparative genome analysis: automatic orthologue identification and genome alignment

Genome re-sequencing and polymorphic analysis

ChIP-seq analysis; mapping and region/peak identification

All those analytical activities can be conducted under default parameter settings using a generally available software package (a software package reported by a published academic article and available in the public domain for free-of-charge utilization under GNU-GPL or free software license).

B. Support for more advanced bioinformatics analysis

In the case of large-scale primary analyses or bioinformatics analyses that require the combination of multiple software packages, we may help to create analytical pipelines after discussing the requirements with the support recipient.

If new software needs to be developed for large-scale search for homology, assembly, gene prediction, mapping, etc., or in the case of more project-specific activities such as advanced sequence analysis, highly accurate annotation and statistical processing of analysis data, we may, depending on the request of the support recipient, make arrangements for organizing a joint project with members of the Bioinformatics Analysis Group. However, for certain projects, we may be

unable to find a suitable researcher for you to work with.

2. Support-Related Requirements

1. Support activities that involve considerations about personal information and/or ethical issues will require coordination with the Medical Genome Science Program.

2. Sequence data from the Large-Scale Genome Information Production Program will be deposited provisionally to DDBJ (DNA Databank of Japan, <http://www.ddbj.nig.ac.jp/>). The sequence data will be transferred directly to DDBJ by the Bioinformatics Analysis Group.

3. When we support the analysis of sequence data prepared independently by the support recipient, we request the support recipient to undertake preliminary tasks such as the conversion of waveform and image data into sequence data (“base call” procedure) and the addition of quality scores. The support recipient must provide the storage media (disks, etc.) used for the transfer of data. If the delivered data are found to be unsuitable for analysis in terms of quantity or quality, the support recipient must discuss with support group members about what action should be taken (cancellation of analysis, conducting experiments again, etc.).

3. Disclosure and Sharing of Data

1. Analytical results from each supported project must be disclosed by appropriate means (article, database, etc.).

2. Support recipients may be requested to permit the use of analytical results, under appropriate arrangements to be discussed with them, for contributing to research activities for developing more advanced bioinformatics analysis systems.

支援申請の方法

Flow of the application for support

STEP 1: Using the application form on this website, enter information about the applicant (group representative). You will then receive an email explaining how to obtain an ID and password. Log in using your ID and password, then fill in the application form. You will use the same ID and password to check later the result of your application.

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STEP 2: Again using the application form on this website, submit information concerning your project supported by the Grant-in-Aid for Scientific Research. Note that all information you supply using this form is disclosed on the Database on the Grant-in-Aid for Scientific Research (<http://kaken.nii.ac.jp/>). If the proposed project requires a review by an ethical review committee (such as if your project involves the handling of personal genome information), you must declare this by ticking the box.

STEP 3: Using the application form on this website, submit information concerning outline of activities you wish to be supported by the program. Please specify the time when you can provide sample for analysis.

STEP 4: Your application must describe the research project to be covered by the Grant-in-Aid for Scientific Research, as well as your expectations for the support from the Genome Science program. The first part of this document describes your research project supported by the Grant-in-Aid for Scientific research. Describe the purpose of the research in no more than two pages and add a single-page summary of your publication list. The second part of the document should describe your expectations about the support in two pages, providing the following information: (1) specific details of the support you expect from the Genome Science program; (2) details of research activities that you expect to implement after the completion of the support by the Genome Science program, with details of the research program and research organization that you are going to prepare for maximizing benefits from the support; (3) the significance of the proposed project for genome sciences and medical genome research, and the present state of related research in Japan and overseas; (4) readiness for receiving support in terms of the present availability of materials, etc., and the level of preparedness for maximizing benefits from the support.

STEP 5 (only if the requirement of an ethical review was declared in STEP 3): If you are proposing a research project that requires a review by an ethical review committee (such as if your project involves the handling of personal genome information), and your project has already obtained organizational approval, you must upload the following: (1) application for organizational approval, (2) informed consent document and (3) certificate of organizational approval. If your project has not yet obtained organizational approval, upload only the following: (1) application for organization approval and (2) informed consent document. This Genome Support website provides a sample of an

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informed consent document comprising an information document and a letter of consent ([download](#)). We use these documents for processing applications, and for no other purpose.

STEP 6: Review your application and complete the procedure. Afterward, you can redisplay and correct the details of your application.