Information Infrastructure for Rare/Intractable Disease Research
(Platform for rare/intractable diseases)

Division of Rare/Intractable Disease Research
Department of Research Promotion
Japan Agency for Medical Research and Development
The construction of the platform is essential for the aggregation and integration of information to facilitate its secondary use.
In February 2017, AMED started research on the construction of information infrastructure (Platform for rare/intractable Diseases) for “Practical Research Project for Rare/Intractable Diseases” supervised by AMED and the “Policy Research Project on intractable diseases” supervised by MHLW (referred to as the “Rare/Intractable disease group”).

“Platform for rare/intractable diseases” is expected to be ready for full-scale operation in 2018. Its objective is to facilitate the maximum effective utilization of information on rare diseases/intractable diseases that “Rare/Intractable disease group” has obtained.
Platform for rare/intractable diseases

(1) Aggregation of information held by “Rare/Intractable disease group” and release of data item (referred to as the “catalog”) out of them

(2) Support for registry construction

(3) Aggregation and integration of information held by the “Rare/Intractable disease group” and promotion of its secondary utilization inside and outside of Japan

(4) Development of artificial intelligence (AI)

Outcomes and Impacts

Promotion of research and development

Improvement and standardization of registry

International collaboration
Overall Design

Research group of Practical Research Project for Rare/Intractable Diseases

Research group of Policy Research Project on intractable diseases

Support of registry construction

Data sharing and provision

Data sharing and provision

Platform for rare/intractable diseases

Center for an Integrated Database of Genomic Information

Center for an Integrated Database of Clinical Information

• Aggregation and integration of information through name-based aggregation with common language
• Creation of added value by reanalysis
• Aggregation of medical needs in rare/intractable diseases
• Release of the catalog

Approval for provision of information by “Platform for rare/intractable diseases” and by the research information resource

Request for secondary use

Provision of data

Third-party institution

Cooperation with other AMED programs by way of data sharing and provision

Program for an Integrated Database of the Clinical and Genomic Information Project for Promoting a Clinical Innovation Network
(1) Release of the catalog

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<th>研究課題名</th>
<th>研究代表者名</th>
<th>レジストリ名</th>
<th>対象疾患</th>
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<td>iPS細胞由来角膜上皮細胞シートのfirst-in-human 臨床研究</td>
<td>西田幸二</td>
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<td>角膜上皮幹細胞破壊症</td>
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<td>自己免疫性肺胞蛋白症に対する酵母由来組換えGM-CSF吸入の多施設共同医師主導治験</td>
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<td>HAMに対する日本発の革新的治療となる抗CCR4抗体の実用化研究</td>
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<td>中性脂肪蓄積心筋血管症に対する中鎖脂肪酸を含有する医薬品の開発</td>
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<td>中性脂肪蓄積症 (NLSD), 中性脂肪蓄積心筋血管症 (TGCV)</td>
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The Rare/Intractable disease group of “Practical Research Project for Rare/Intractable Diseases” and “Policy Research Project on Intractable diseases” supervised by the MHLW has released the catalog information of about 90 registries.
Overall Design

Support of registry construction

Data sharing and provision

Platform for rare/intractable diseases

Center for an Integrated Database of Genomic Information

Center for an Integrated Database of Clinical Information

Collaboration

• Aggregation and integration of information through name-based aggregation with common language
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Program for an Integrated Database of the Clinical and Genomic Information Project for Promoting a Clinical Innovation Network

Cooperation with other AMED programs by way of data sharing and provision
(2) Support of registry construction

Outline

“Platform for rare/intractable diseases” will appropriately provide support for construction of the patient registry to assure the quality of the data (accuracy, reliability, consistency, usability, etc.) through standardization.
Support of registry information

Registry research for Rare/Intractable Disease A

Registry research for Rare/Intractable Disease B

Registry research for Rare/Intractable Disease C

Support of registry construction

Support (1)
Provision of standard operation documents such as standard protocol and standard document for informed consent necessary for each research project

Support (2)
Standardized case data collection system is established to support data collection for each research project

Operational support for registry research

Data provision from registry research

Standard operation documents prepared by “Platform for rare/intractable disease” (e.g., standard protocol and standard document for informed consent) will be customized for each “Rare/Intractable disease group” and used for the research.
Overall Design

Research group of Practical Research Project for Rare/Intractable Diseases

Research group of Policy Research Project on intractable diseases

Support of registry construction

Data sharing and provision

Platform for rare/intractable diseases

Center for an Integrated Database of Genomic Information

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Center for an Integrated Database of Clinical Information

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Cooperation with other AMED programs by way of data sharing and provision

Program for an Integrated Database of the Clinical and Genomic Information Project for Promoting a Clinical Innovation Network
(3) Platform for rare/intractable diseases
Center for an Integrated Database of Genomic Information

Platform for rare/intractable diseases

- Integrate and manage genomic information shared and provided by each “Rare/Intractable disease group”
- Promote collaborative study and secondary utilization through release of profiles of usable information
- Provide incentive to information source (e.g. support of genomic analysis and use of search tool for causal genes)
- Creation of added value by integrating clinical information and omics information
(3) Platform for rare/intractable diseases
Center for an Integrated Database of Clinical Information

Platform for rare/intractable diseases

• Integrate and manage registry information shared and provided by each “Rare/Intractable disease group”
• Promote collaborative study and secondary utilization through release of profiles of usable information
• Provide incentive to information source (e.g. proposal for the research based on the analysis of integrated data)
• Creation of added value by integrating omics information and other registry information
Lastly,

The mission of “Rare/Intractable disease platform” is to support the development of research for rare and intractable diseases through facilitating the maximum effective utilization of information obtained by “Rare/intractable diseases group”.

We ask for your understanding and cooperation for this platform.