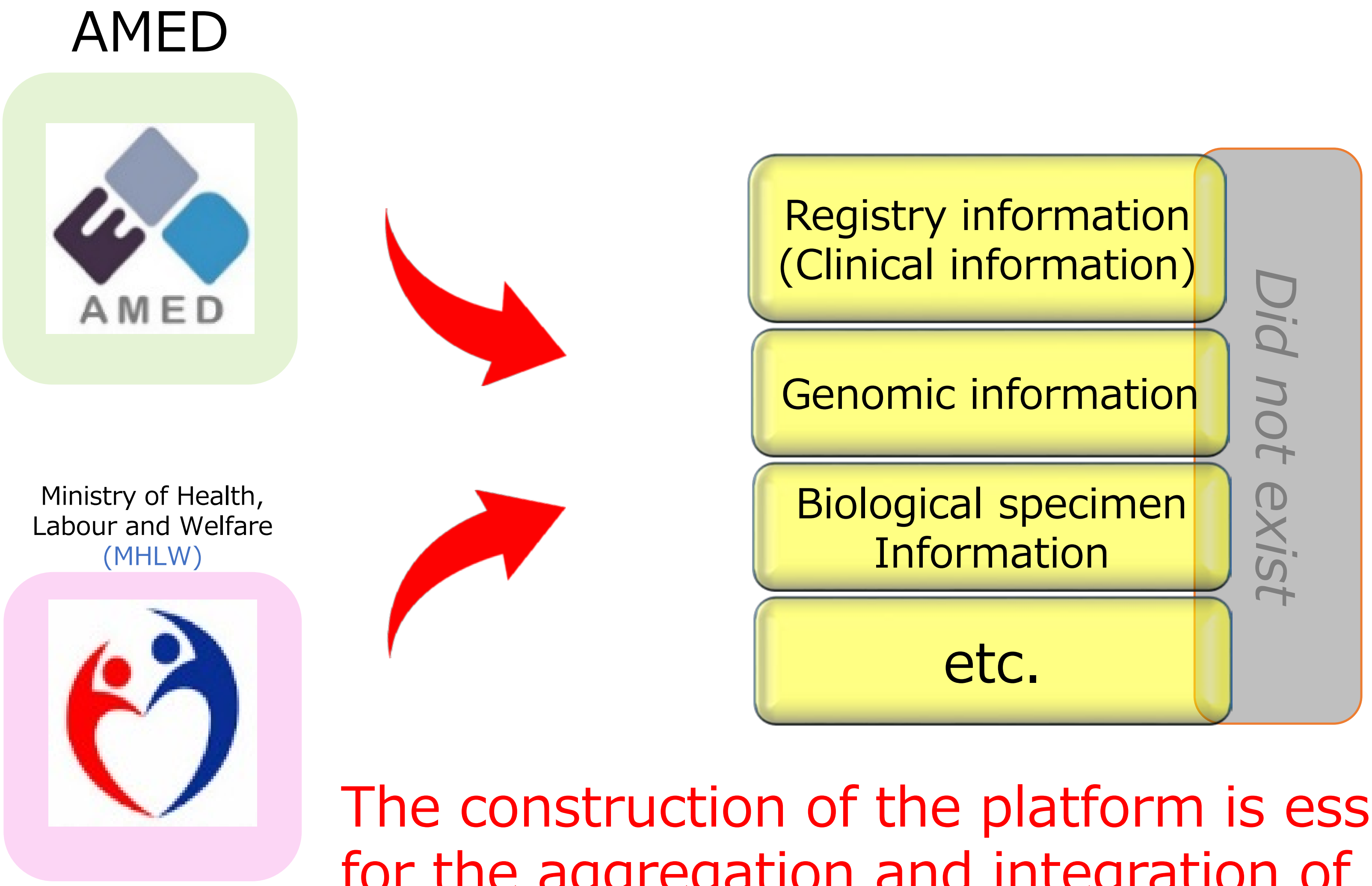
国立研究開発法人日本医療研究開発機構
Japan Agency for Medical Research and Development

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

Need for Shared Information Infrastructure



AMED

Ministry of Health,
Labour and Welfare
(MHLW)

Registry information
(Clinical information)

Genomic information

Biological specimen
Information

etc.

Did not exist

The construction of the platform is essential for the aggregation and integration of information to facilitate its secondary use

Platform for rare/intractable diseases

In February 2017, AMED started research on the construction of information infrastructure (Platform for rare/infractable 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)

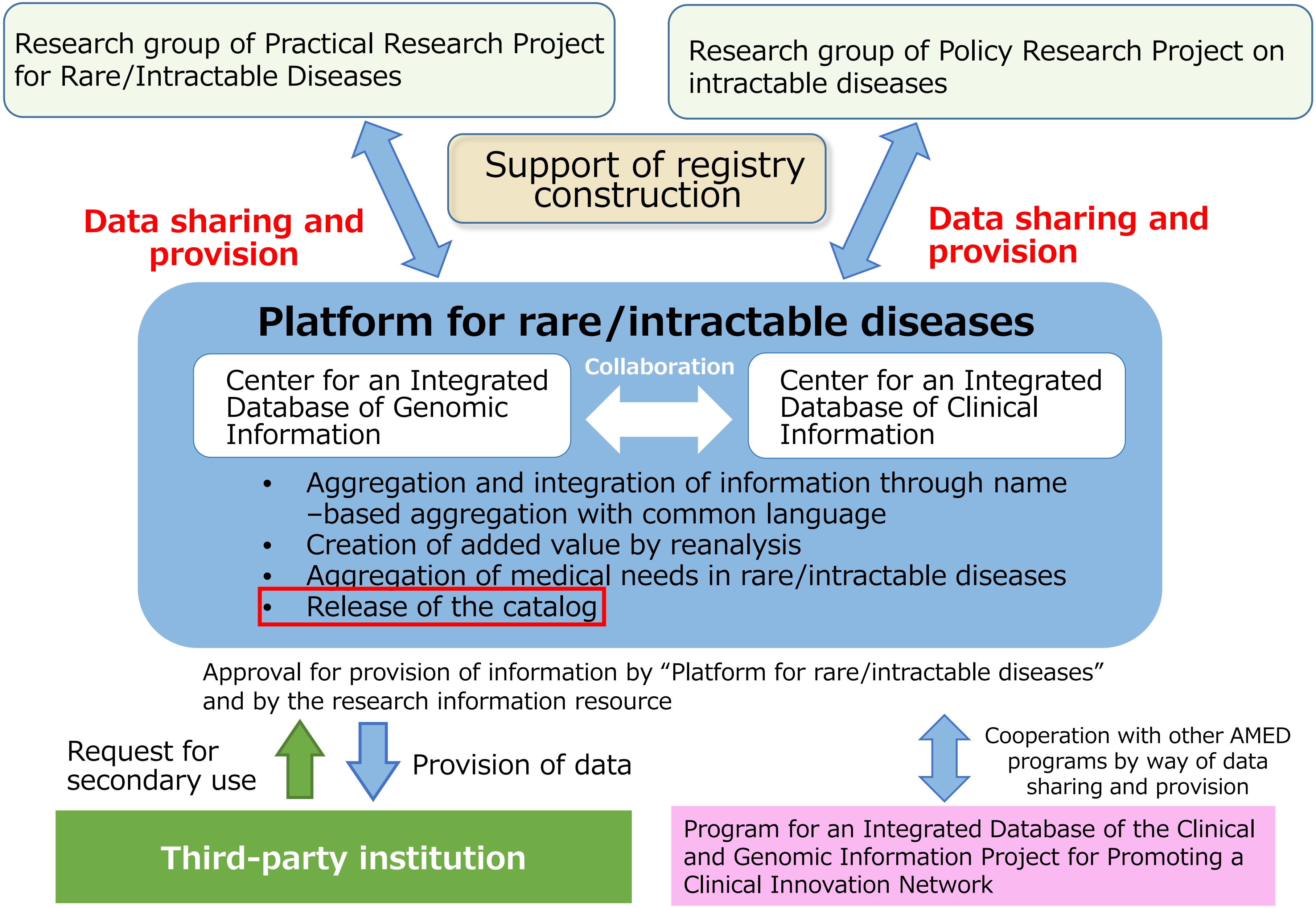
Outcomes and Impacts

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

Collaboration

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

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

Third-party institution

Program for an Integrated Database of the Clinical and Genomic Information Project for Promoting a Clinical Innovation Network

(1) Release of the catalog

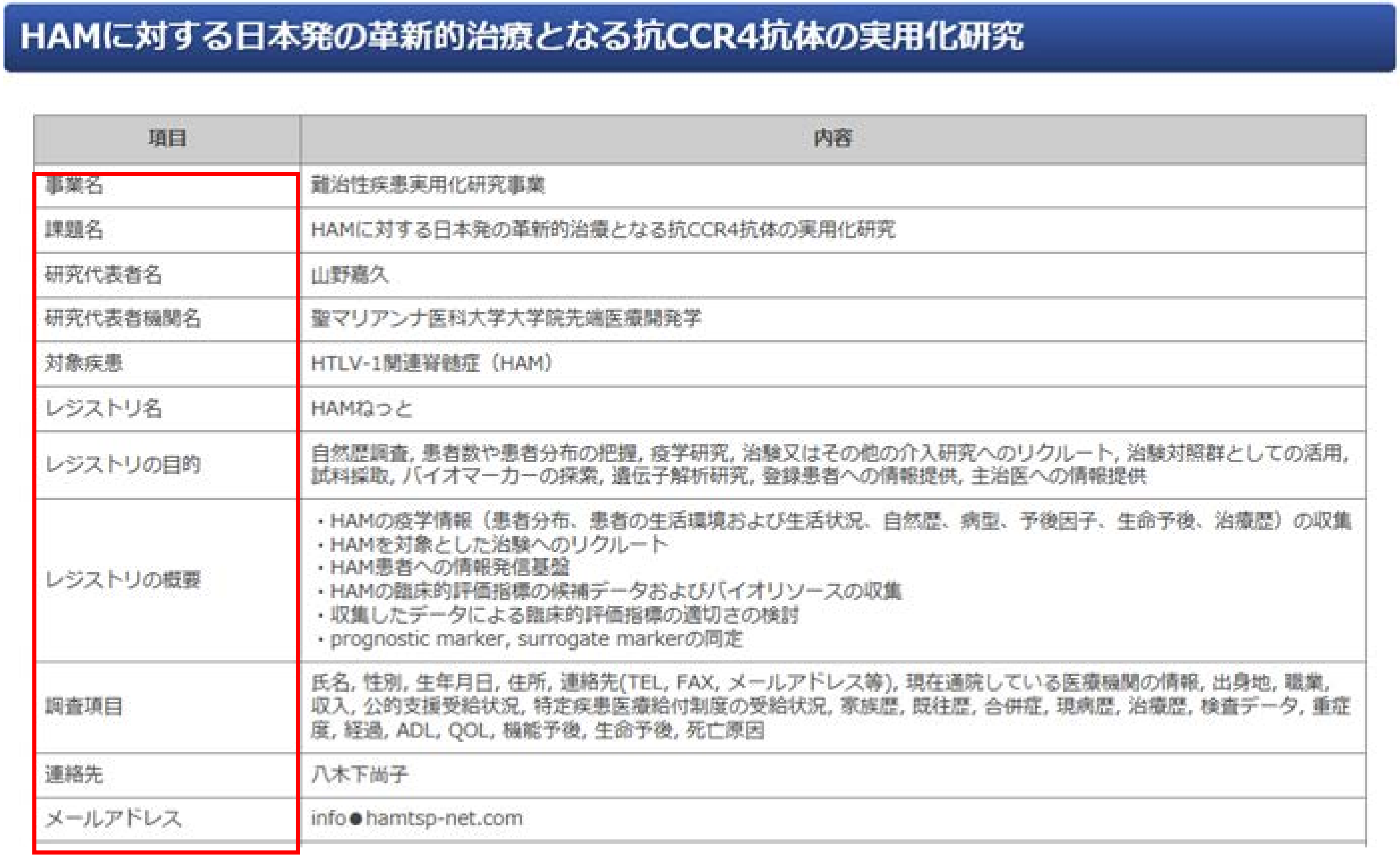
Registry list

日本医療研究開発機構（AMED）の「難治性疾患実用化研究事業」

本事業は、「発病の機構が明らかでない」、「治療方法が確立していない」、「希少な疾病である」、「長期の療養を必要とする」の4要素を満たす難病に対して、病因・病態の解明、画期的な診断・治療法の開発を推進し、全ての難病患者が受ける医療水準の向上を図ることを目的としています。

研究課題名	研究代表者名	レジストリ名	対象疾患
iPS細胞由来角膜上皮細胞シートのfirst-in-human 臨床研究	西田幸二		角膜上皮幹細胞疲弊症
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	中田光	Leukine吸入療法後のAPAP患者追跡データベース	自己免疫性肺胞蛋白症
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	山野嘉久	HAMねっと	HTLV-1関連脊髄症（HAM）
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	平野賢一	International Registry Study of Neutral Lipid Storage Disease (NLSD) / Triglyceride Deposit Cardiomyovasculopathy	中性脂質蓄積症 (NLSD), 中性脂肪蓄積心筋血管症 (TGCV)

(1) Release of the catalog

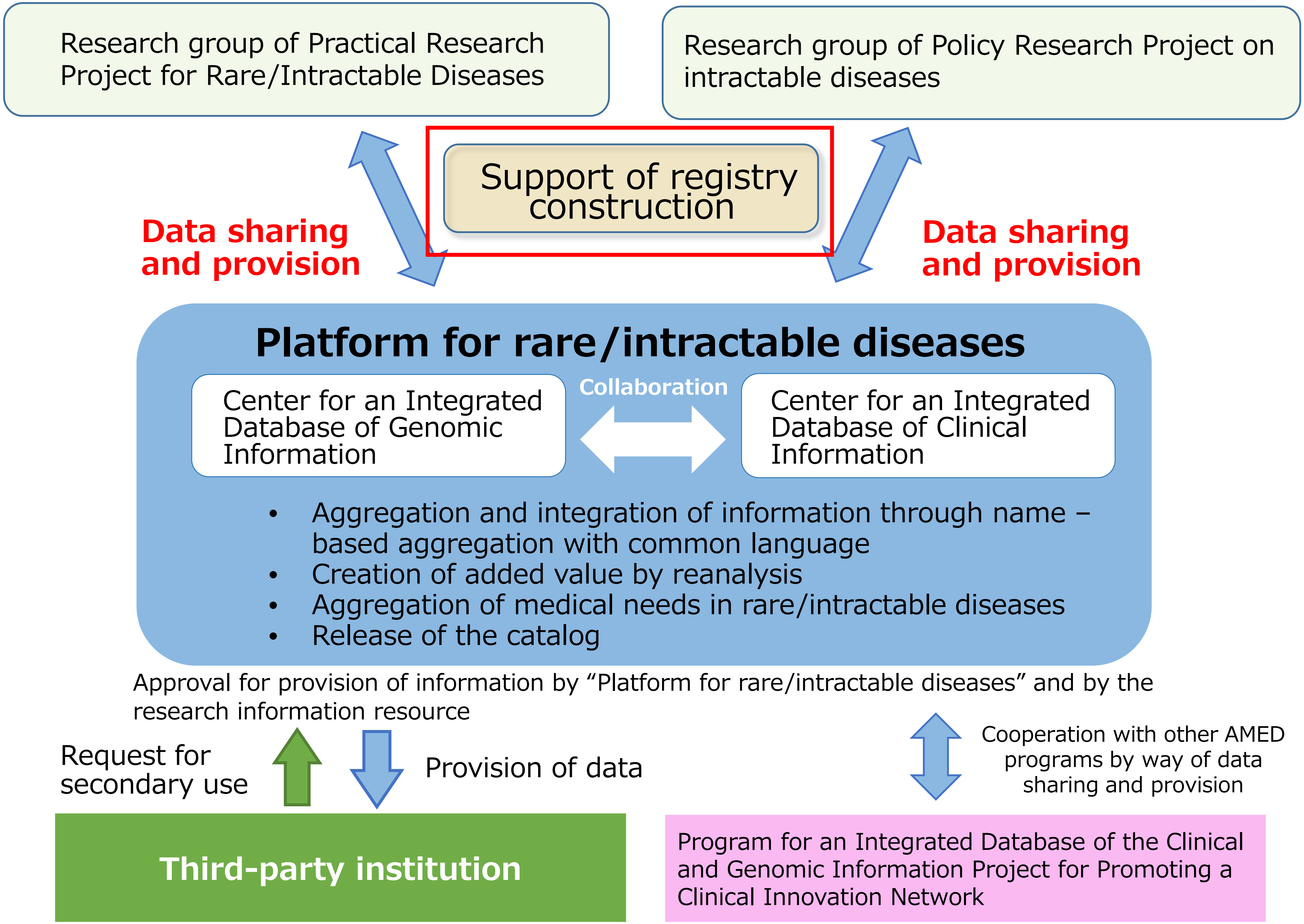


HAMに対する日本発の革新的治療となる抗CCR4抗体の実用化研究

項目	内容
事業名	難治性疾患実用化研究事業
課題名	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
研究代表者名	山野嘉久
研究代表者機関名	聖マリアンナ医科大学大学院先端医療開発学
対象疾患	HTLV-1関連脊髄症（HAM）
レジストリ名	HAMねっと
レジストリの目的	自然歴調査，患者数や患者分布の把握，疫学研究，治験又はその他の介入研究へのリクルート，治験対照群としての活用，試料採取，バイオマーカーの探索，遺伝子解析研究，登録患者への情報提供，主治医への情報提供
レジストリの概要	・HAMの疫学情報（患者分布、患者の生活環境および生活状況、自然歴、病型、予後因子、生命予後、治療歴）の収集 ・HAMを対象とした治験へのリクルート ・HAM患者への情報発信基盤 ・HAMの臨床的評価指標の候補データおよびバイオリソースの収集 ・収集したデータによる臨床的評価指標の適切さの検討 ・prognostic marker, surrogate markerの同定
調査項目	氏名，性別，生年月日，住所，連絡先(TEL, FAX, メールアドレス等)，現在通院している医療機関の情報，出身地，職業，収入，公的支援受給状況，特定疾患医療給付制度の受給状況，家族歴，既往歴，合併症，現病歴，治療歴，検査データ，重症度，経過，ADL，QOL，機能予後，生命予後，死亡原因
連絡先	八木下尚子
メールアドレス	info●hamtsp-net.com

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



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

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Collaboration

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Request for secondary use

Provision of data

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Third-party institution

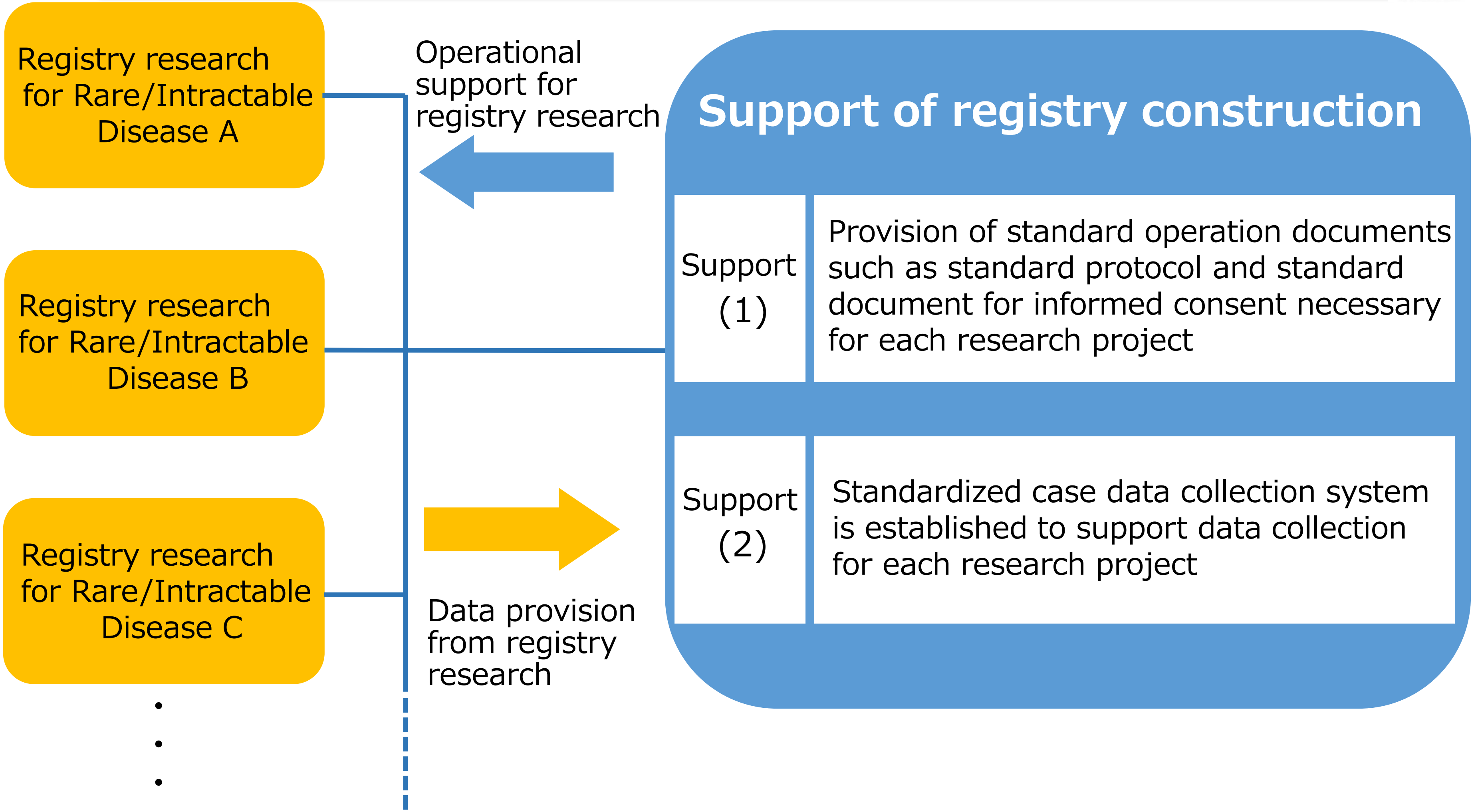
Program for an Integrated Database of the Clinical and Genomic Information Project for Promoting a Clinical Innovation Network

(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

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Operational support for registry research

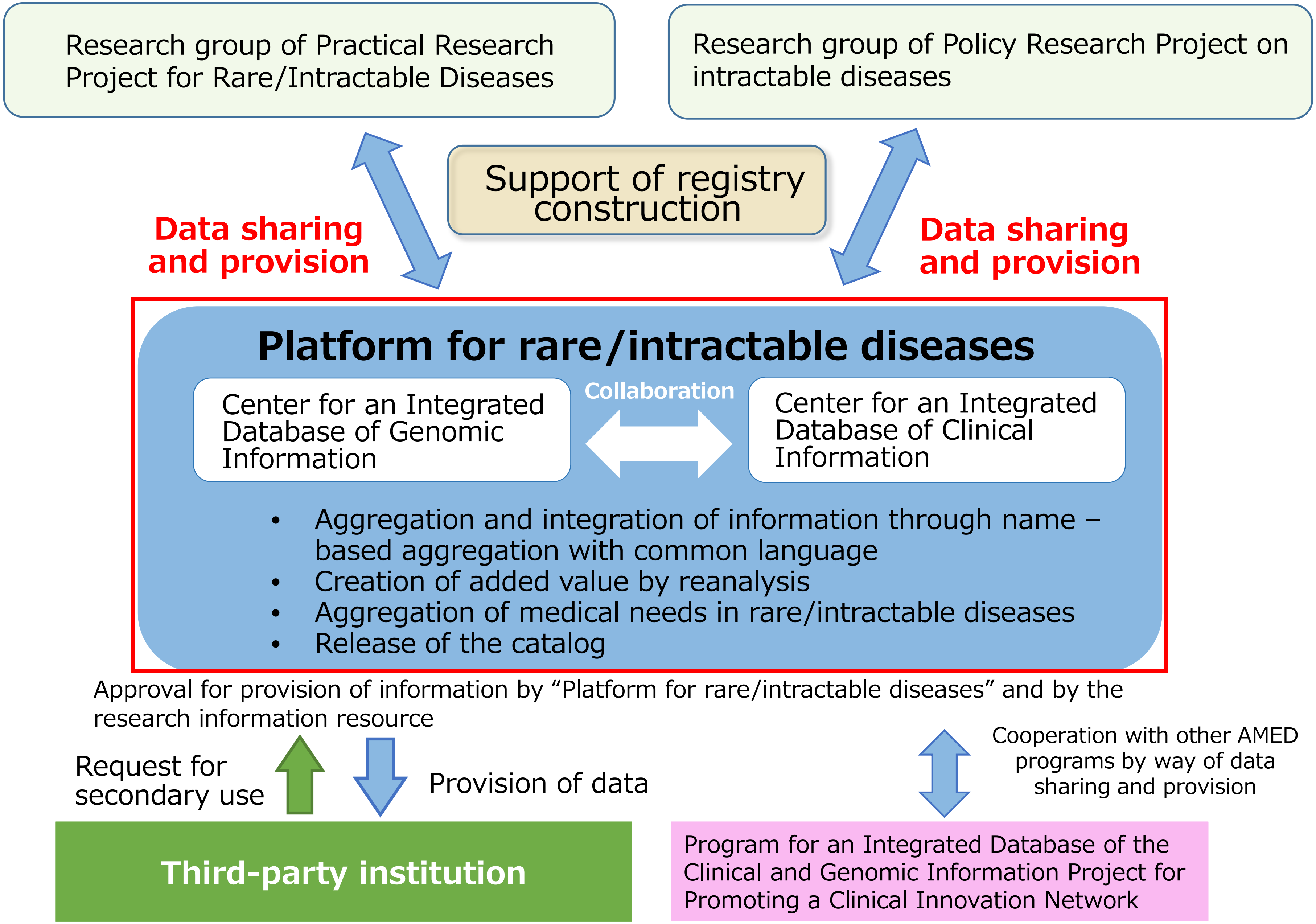
Data provision from registry research

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

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



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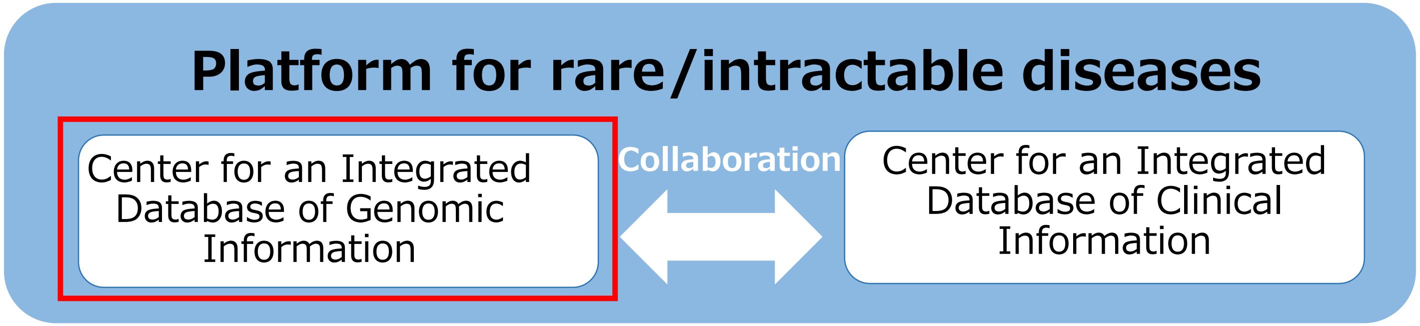
(3) Platform for rare/intractable diseases Center for an Integrated Database of Genomic Information



Platform for rare/intractable diseases

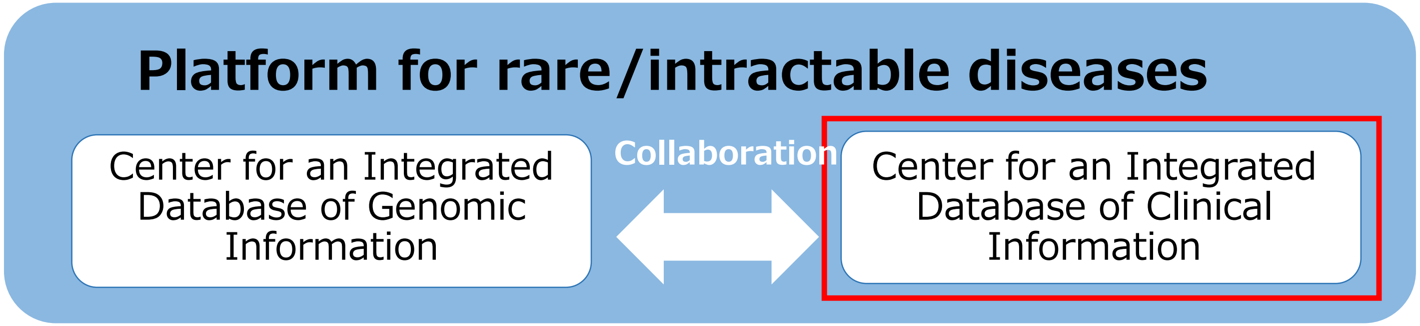
Center for an Integrated Database of Genomic Information

Collaboration

Center for an Integrated Database of Clinical Information

- 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

Center for an Integrated Database of Genomic Information

Collaboration

Center for an Integrated Database of Clinical Information

- 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.