難病情報のData-sharing による医療および行政・産業への波及効果について

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泉田欣彦

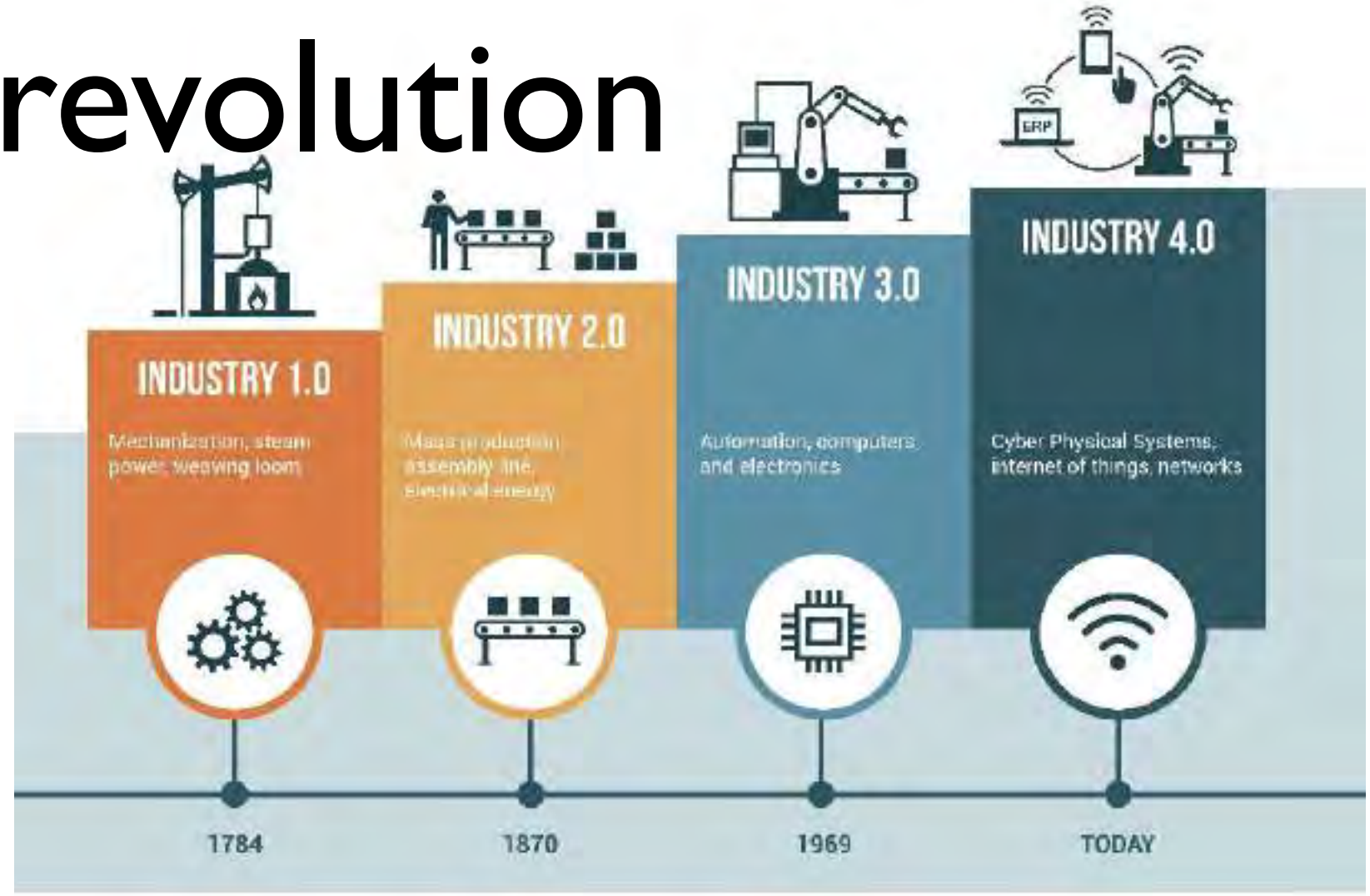
Information is Drug

Gene therapy

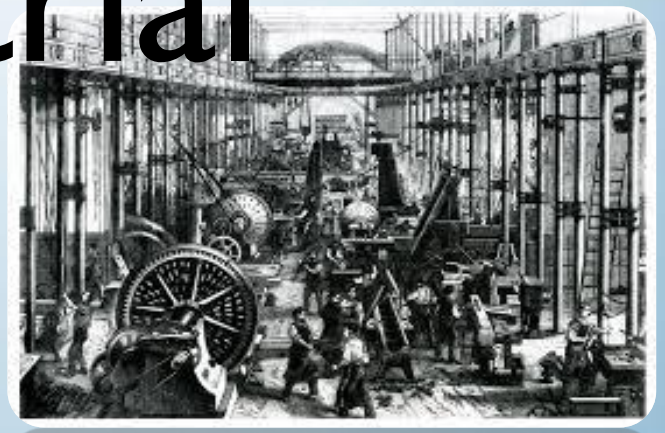
Evelyn Villarreal suffered from Spinal Muscular Atrophy

Evelyn Villarreal, treated for spinal muscular atrophy type 1 with a new gene therapy, is nearly 3. Few children with her condition reach age 2

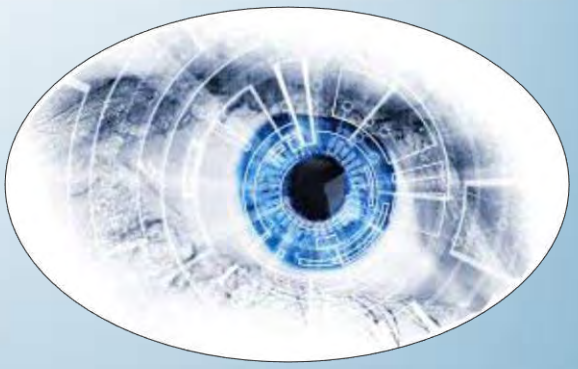
Industrial revolution



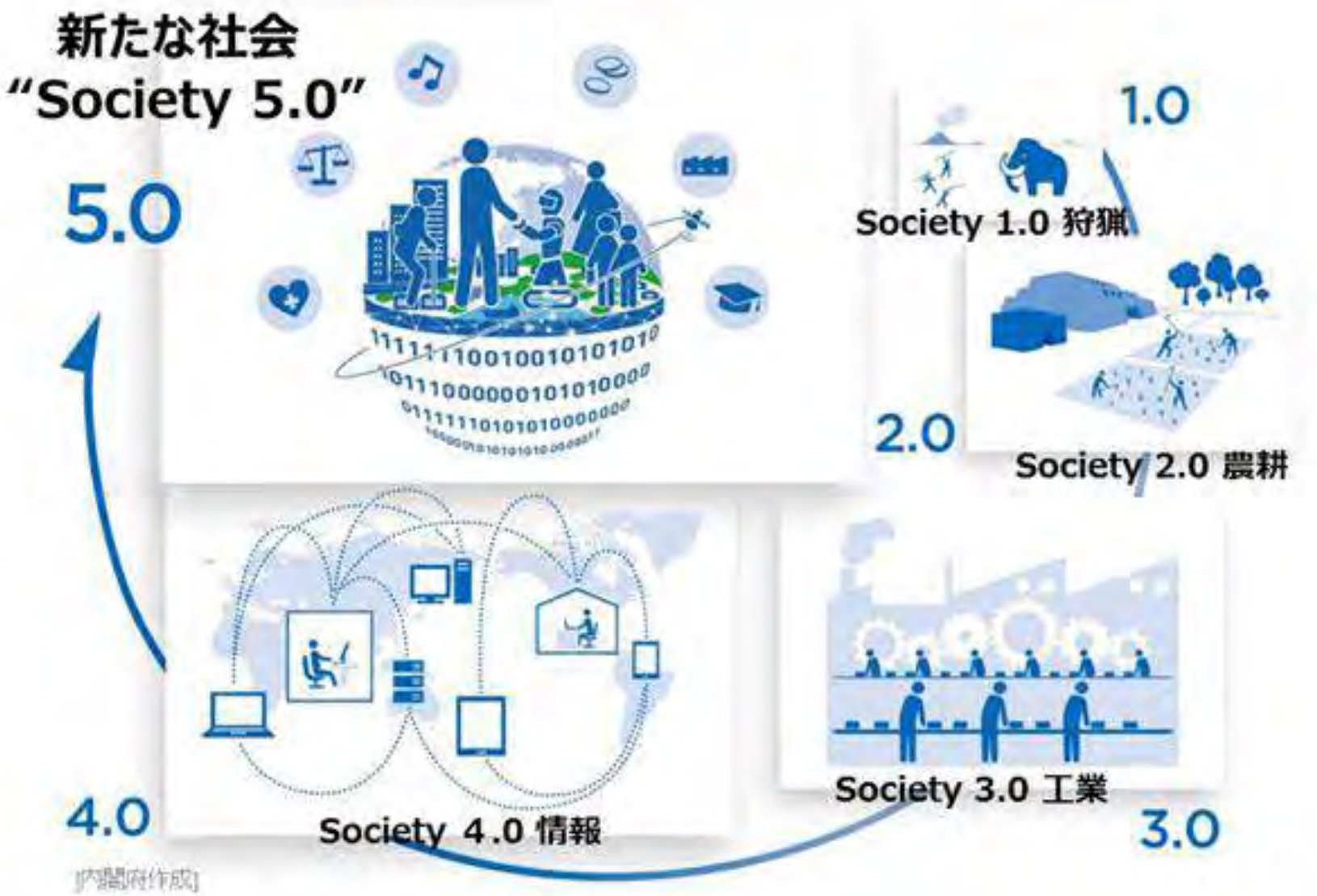
First industrial revolution



4th industrial revolution



第５期科学技術基本計画
Society5.0



Breaking Barriers to Health Data

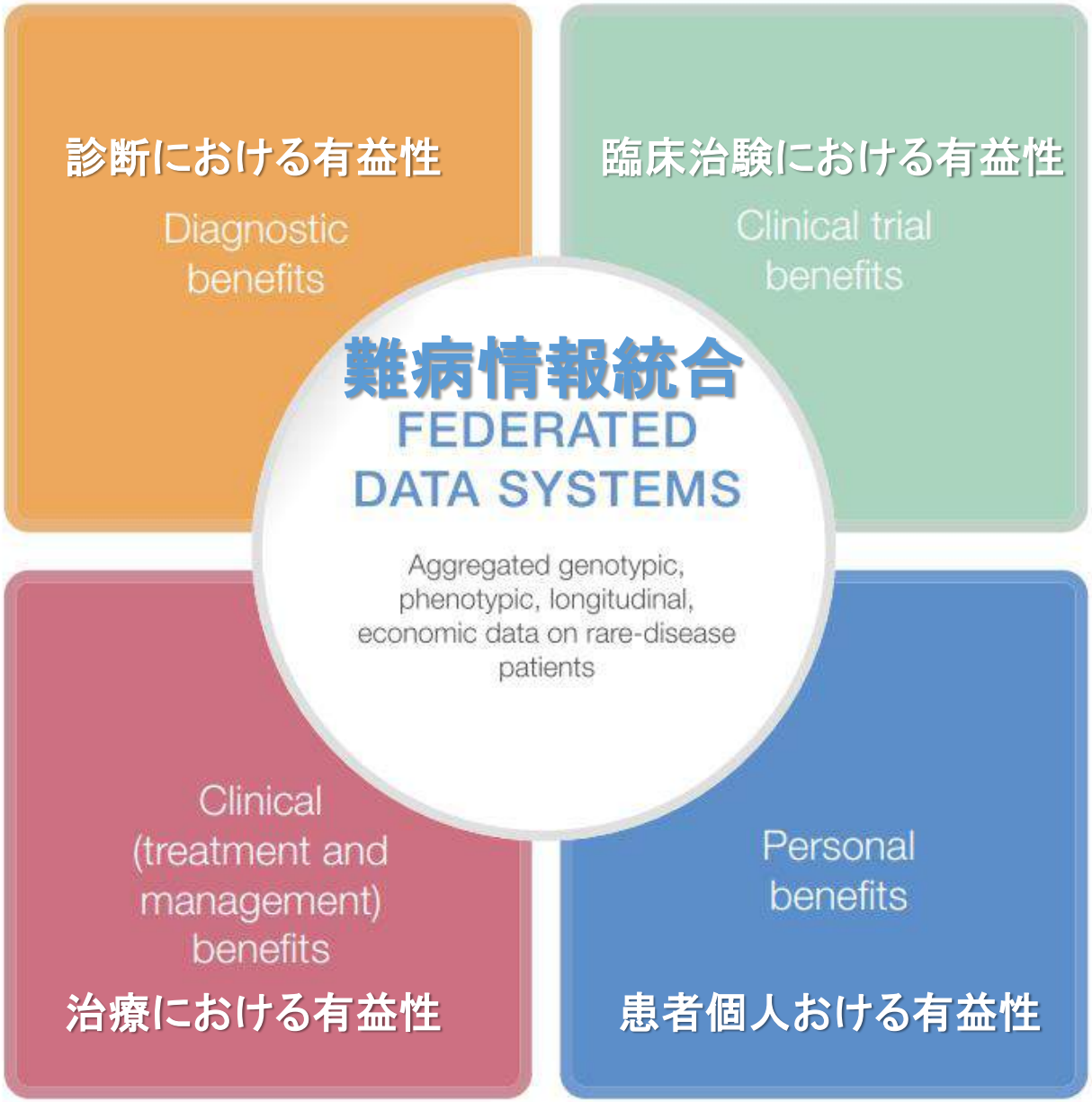
https://www.weforum.org/projects/breaking-barriers-to-health-data-project

Electronic Data Capture in clinical research

英国NHS

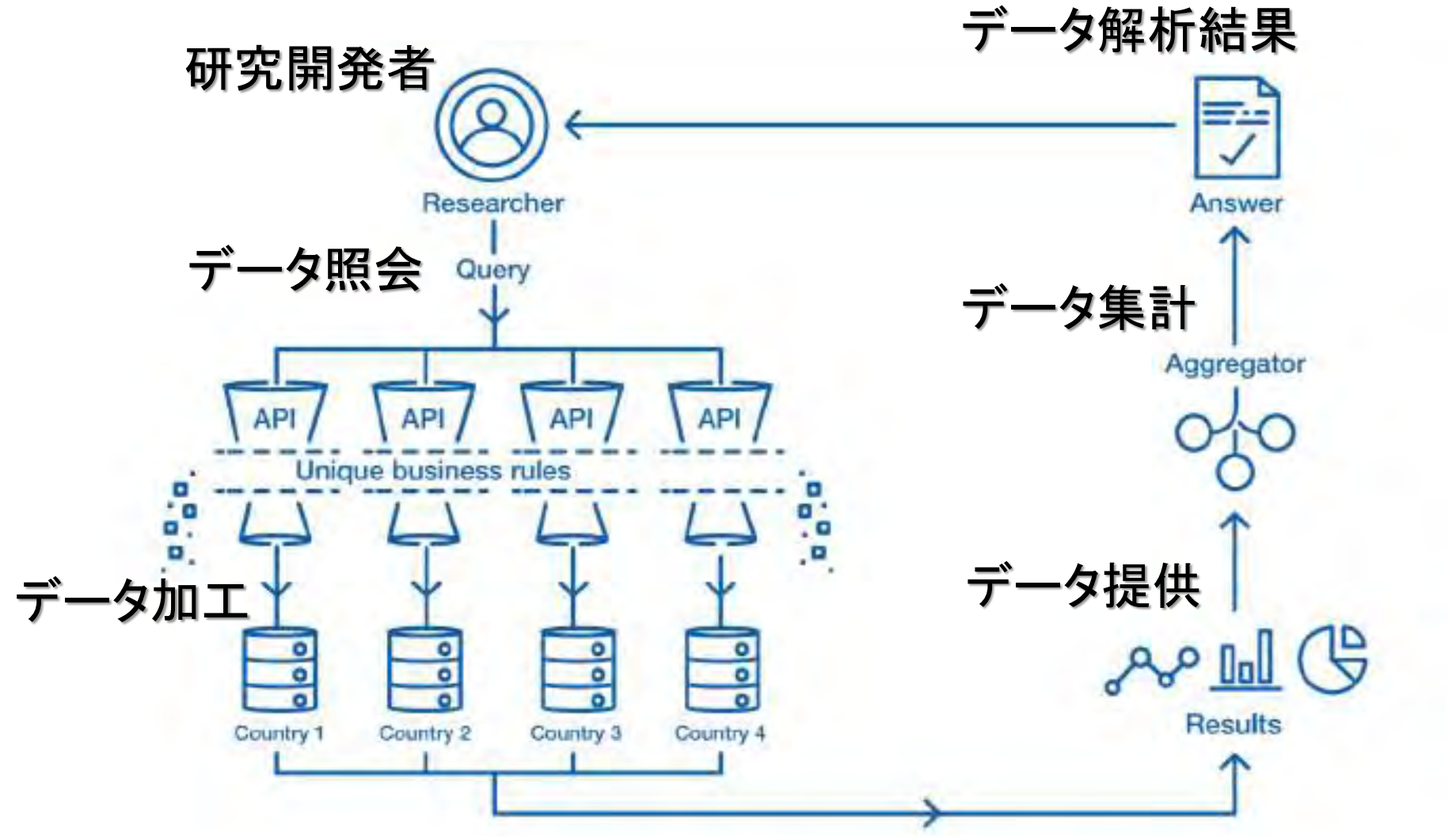
米国Vanderbilt university

VMC



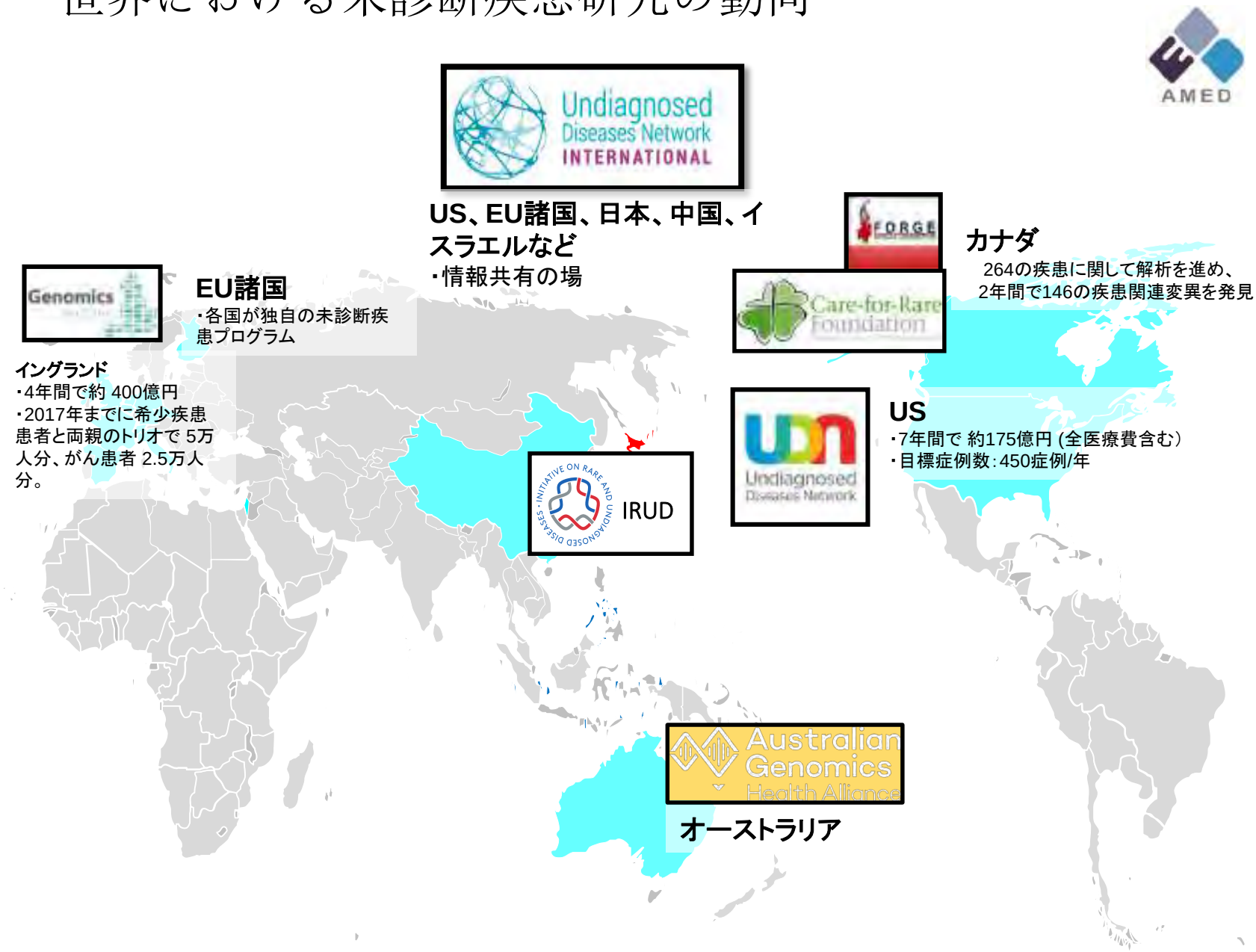
http://www3.weforum.org/docs/WEF_Global_Data_Access_for_Solving_Rare_Disease_Report_2020.pdf

難病関連情報統合の社会実装



Source: World Economic Forum, *Federated Data Systems: Balancing Innovation and Trust in the Use of Sensitive Data*

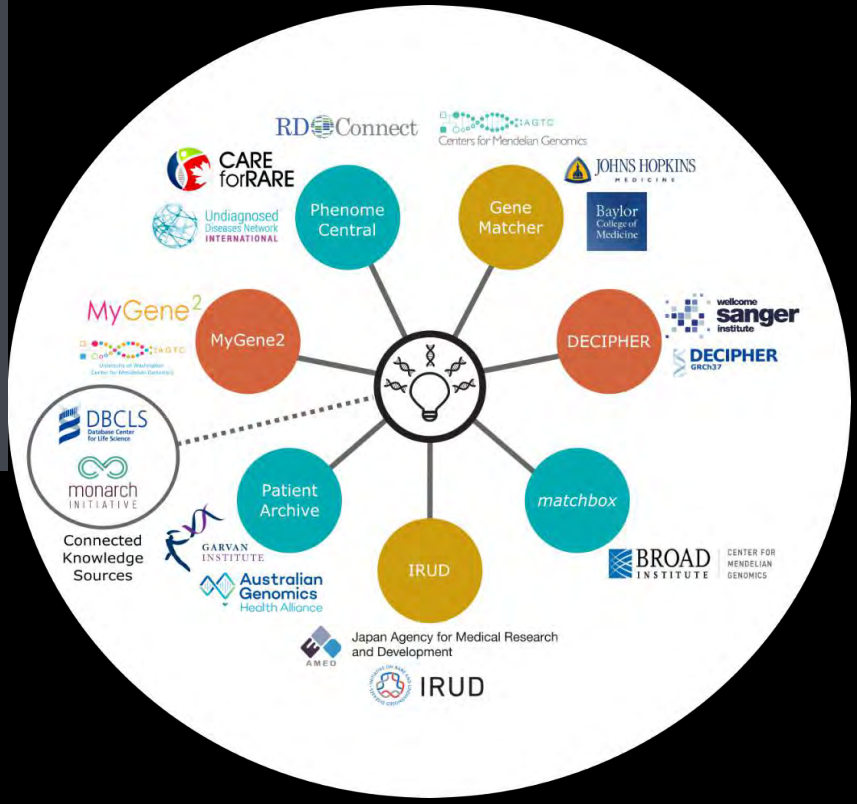
世界における未診断疾患研究の動向



The Initiative on Rare and Undiagnosed Diseases (IRUD) became an official connected node within the Matchmaker Exchange (MME) network as of December 2017.

GLOBAL DATA SHARING

This API was launched as a federated system in July 2015. A special issue of Human Mutation in October 2015 described the MME, its API, connected matchmakers, and a few examples of successful disease gene discovery. The MME is a driver project of both the Global Alliance for Genomics and Health (GA4GH, ga4gh.org) and the International Rare Disease Research Consortium (IRDiRC.org).



National Institutes of Health

The Precision Medicine Initiative

Source: NIH

From 100,000 to 5 million

News story

Matt Hancock announces ambition to map 5 million genomes

The NHS Genomic Medicine Service is the first national genomic healthcare service in the world and will allow faster diagnosis and personalised care.

Published 3 October 2018
From: Department of Health and Social Care

Related content

National Health Service

Research, testing and star

Health and Social Care Secretary Matt Hancock said:

"I'm proud to announce we are expanding our 100,000 Genomes Project so that one million whole genomes will now be sequenced by the NHS and the UK Biobank.

I'm incredibly excited about the potential for this type of technology to improve the diagnosis and treatment for patients to help people live longer, healthier lives – a vital part of our long-term plan for the NHS.

From 2019, the NHS will offer whole genome analysis for all seriously ill children with a suspected genetic disorder, including those with cancer. The NHS will also offer the same for all adults suffering from certain rare diseases or hard to treat cancers.

Today's commitments form part of our bold aspiration to sequence 5 million genomes in the UK, using ground-breaking technology to do this within an unprecedented 5-year period."

Genome Medicine Alliance Japan, GEM-J

Data sharing

between
- Japanese (homogeneous genome and healthcare) and the Global
- Research and Clinical practice
- Common diseases and Rare diseases

GA4GH

Sharing of Data, Ideas, Experiences and Technologies

Research

Japanese variants

TOGOVAR

Japan Human Genome DB

Cooperation

QC check and standardization

Clinical practice

Japanese disease variants

MGeND

Polygenic Diseases

DM | CeVD | SycD | CVD

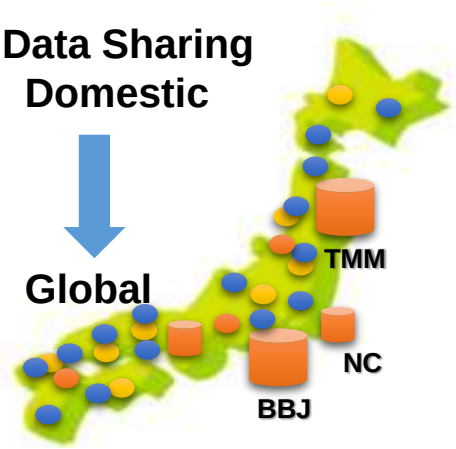
MEXT

BBJ

TMM

Data Sharing Domestic

Global



Rare diseases and Cancer

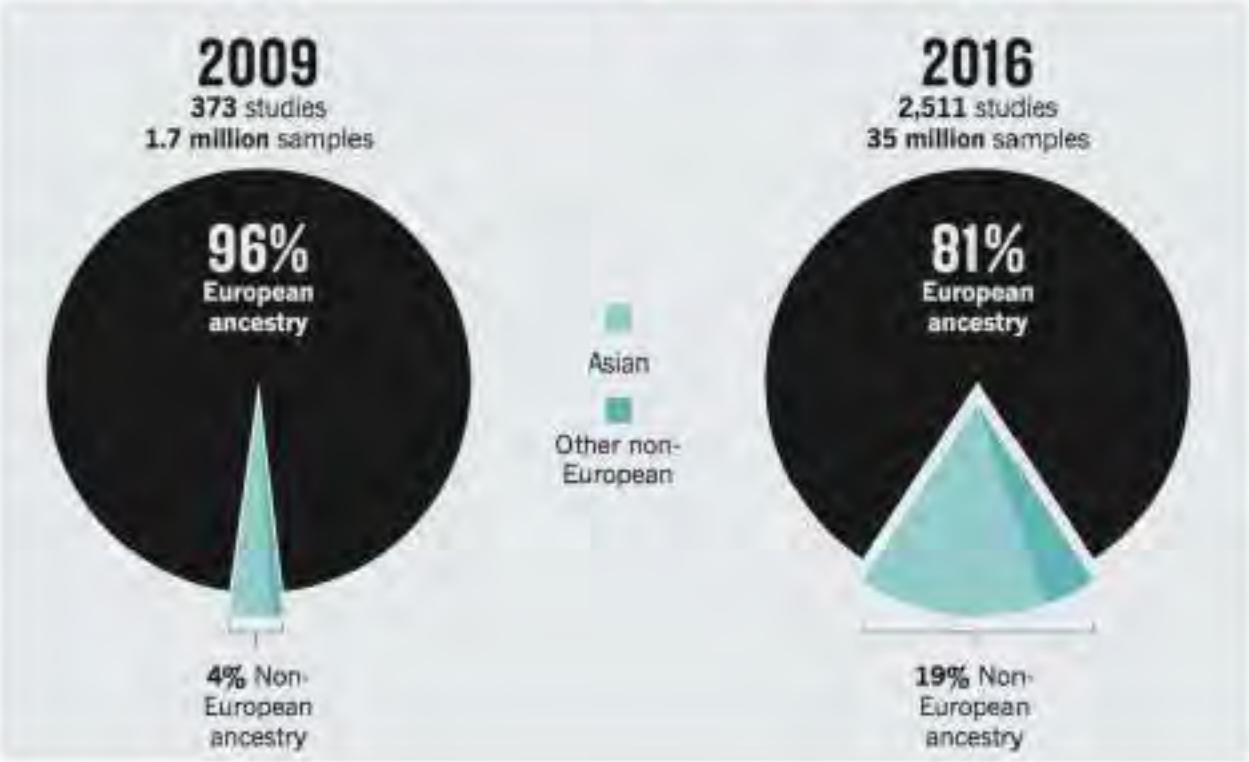
RD | Cancer | Infection | Dementia

IRUD

RD

MHLW

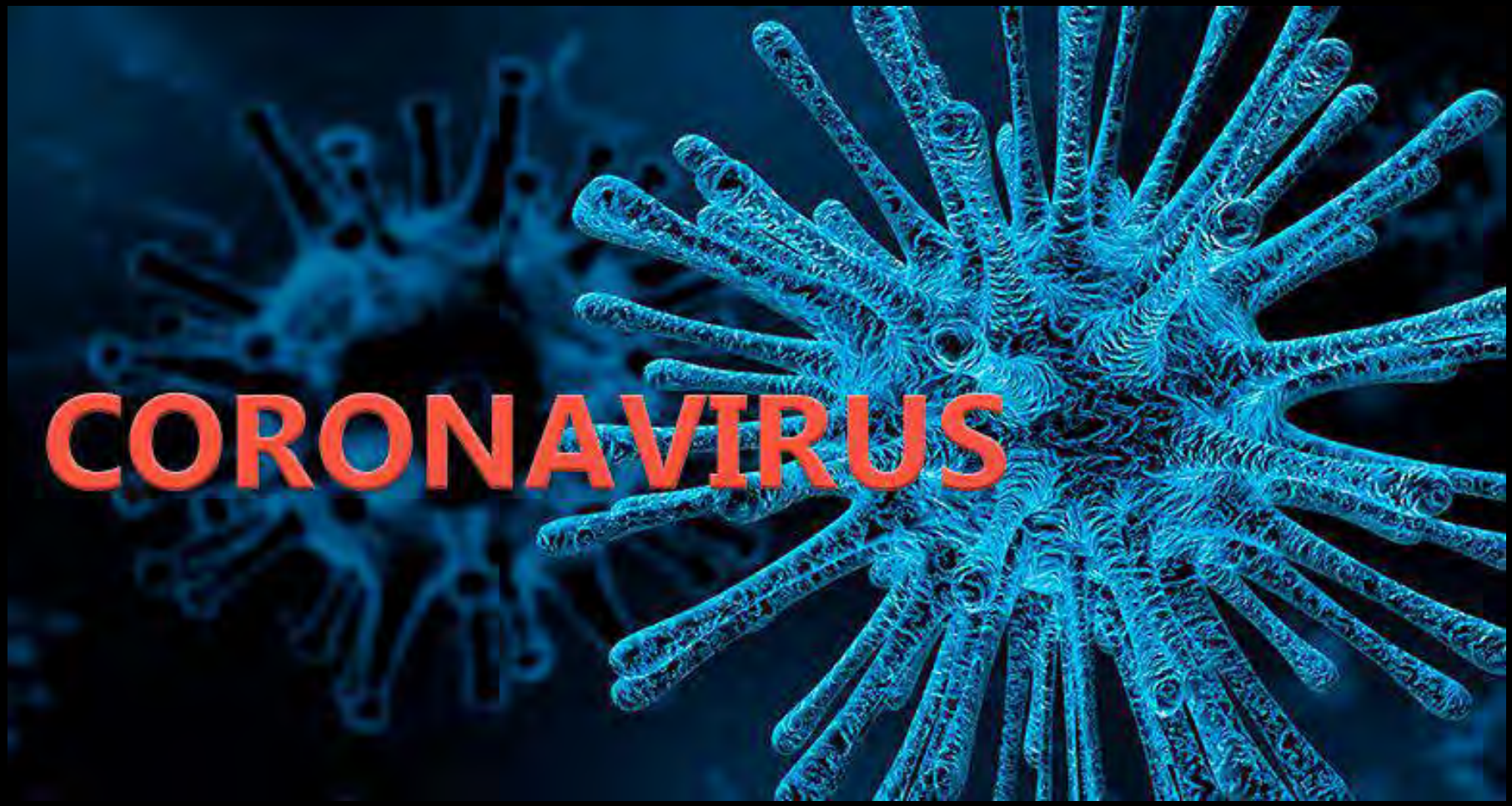
Genomics is Failing on Diversity

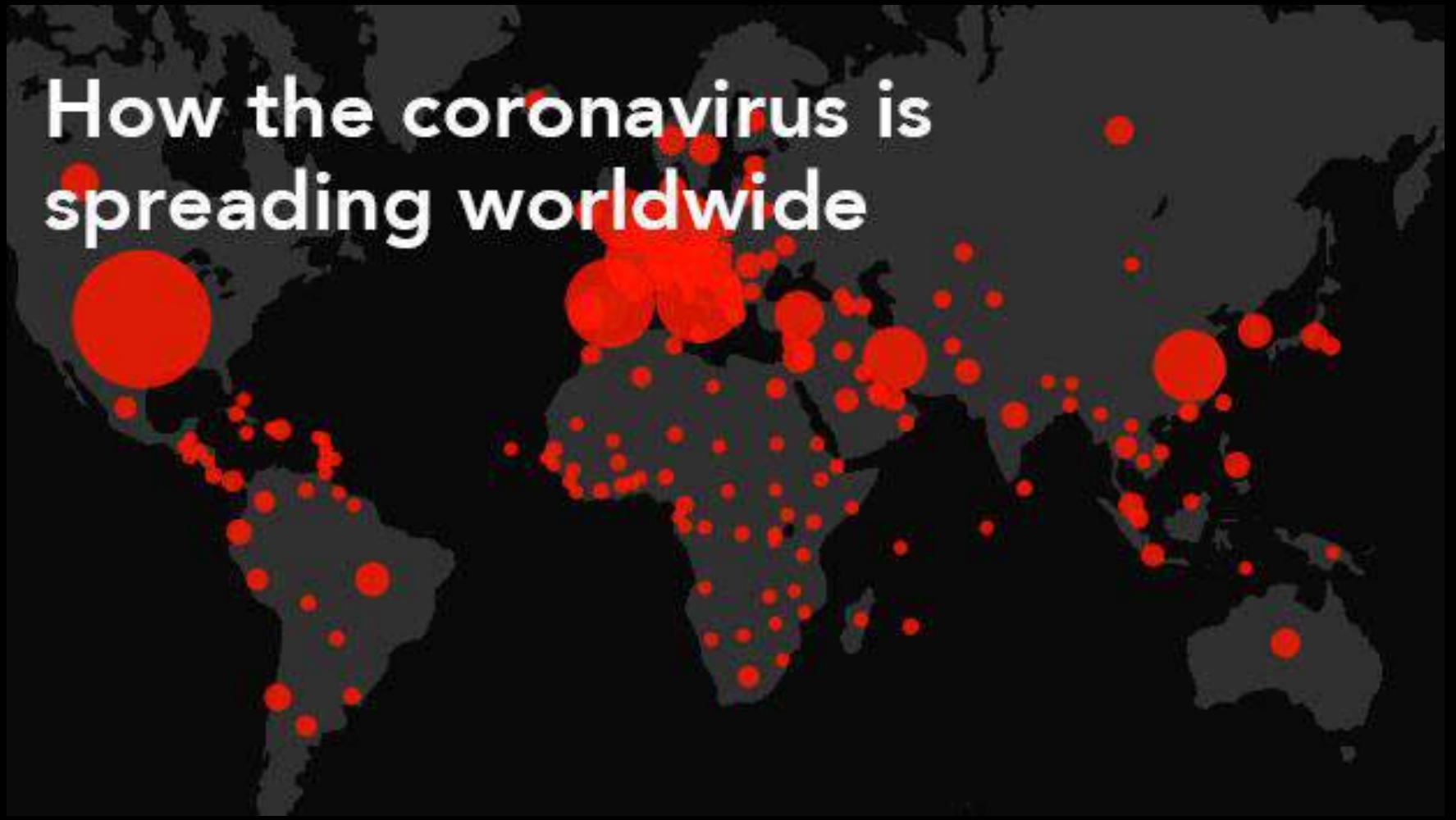


(Need & Goldstein, 2009)
(Bustamante et al., 2011)

(Popejoy & Fullerton, 2016)

Stanford MEDICINE


CORONAVIRUS


How the coronavirus is
spreading worldwide

Global Cases

49,237,148

Global Deaths

1,241,378

https://coronavirus.jhu.edu/map.html

Genetics or Governance?

COVID-19国際研究組織が発足している

数多くの世界の難病ゲノム研究組織が参加

The COVID19-HGI in numbers

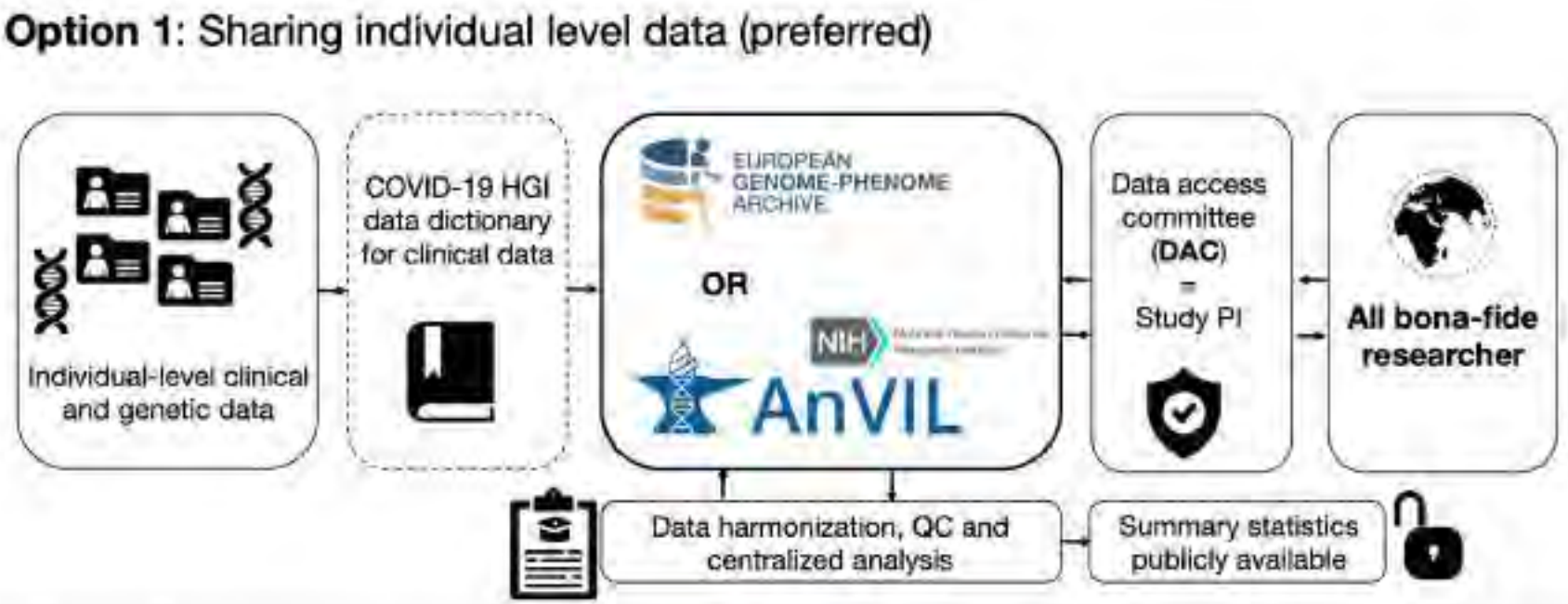
- 928 members (last call) → **1027 members**
- 51,000 unique users on the website
- 176,845 page view

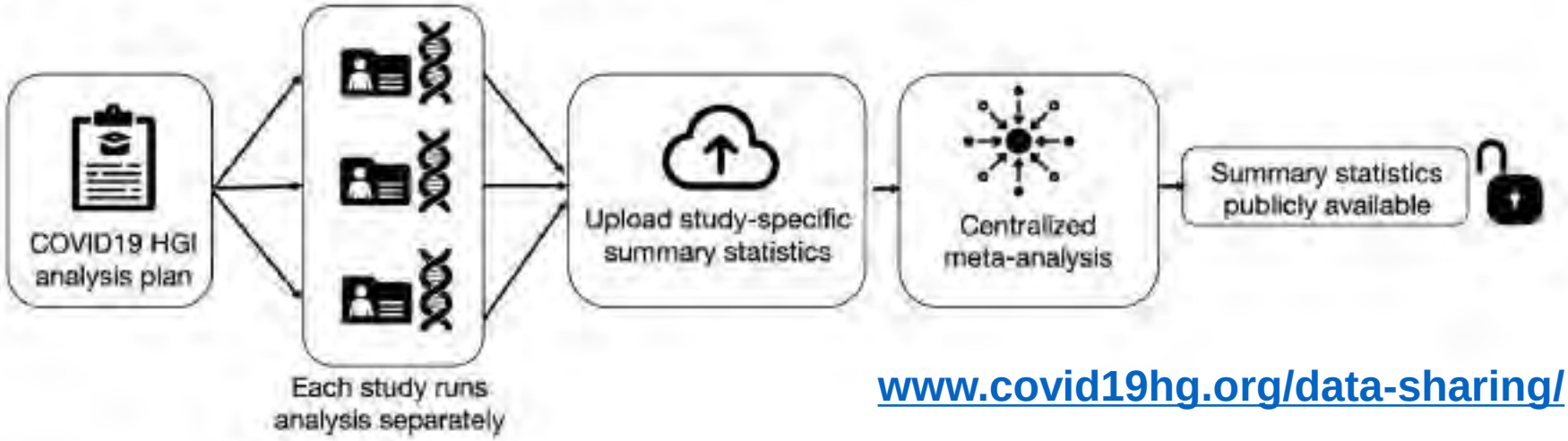
- **183 studies from 46 countries**
- **10 ”contributing studies”**

Lifelines, UK Biobank, Genes and Health,
BioMe, Helix Exome+, Partners Biobank, Finngen, Netherlands Twin Registry,
Generation Scotland, INTERVAL

https://www.covid19hg.org/

難病研究で培ったノウハウで新型コロナの遺伝学的研究が可能

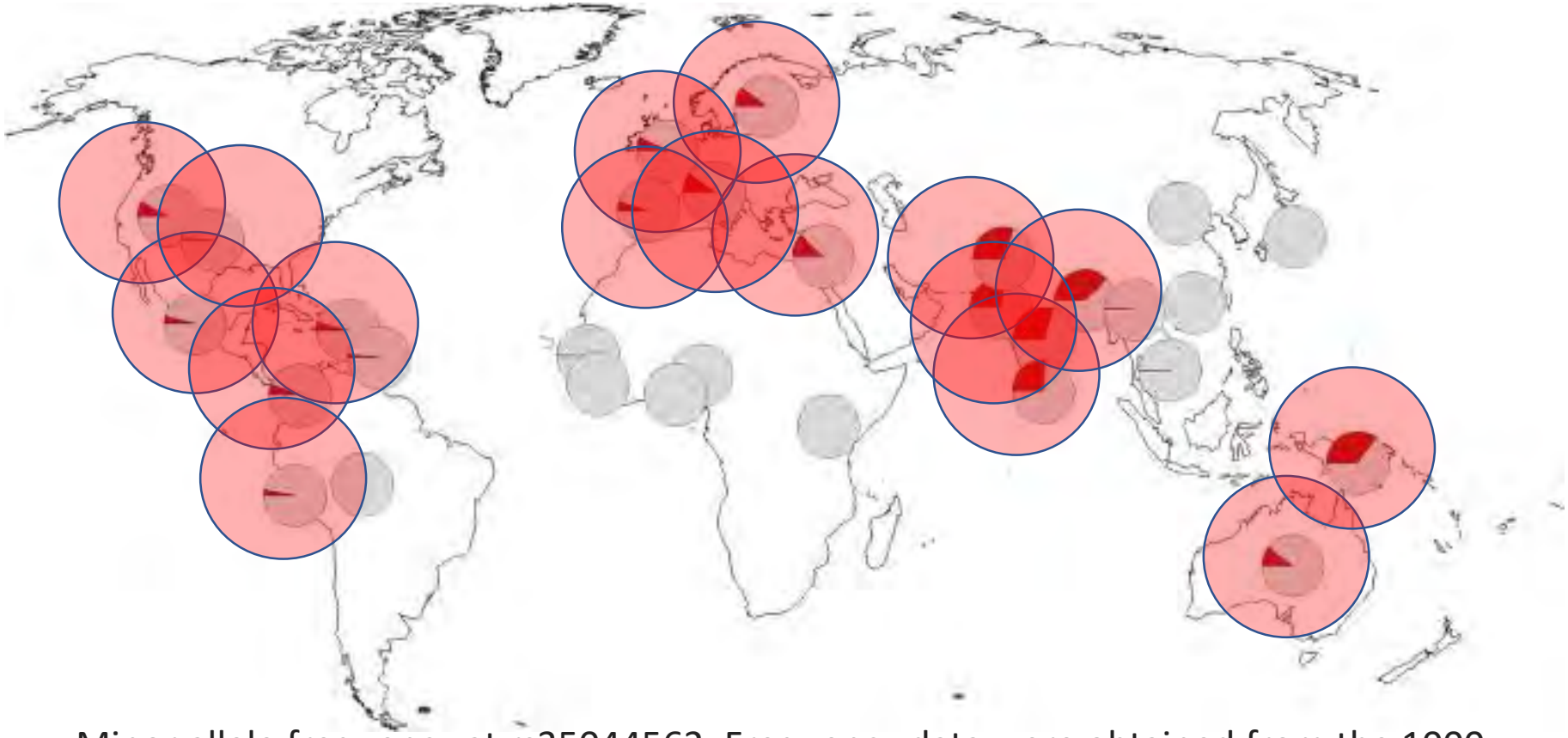




www.covid19hg.org/data-sharing/

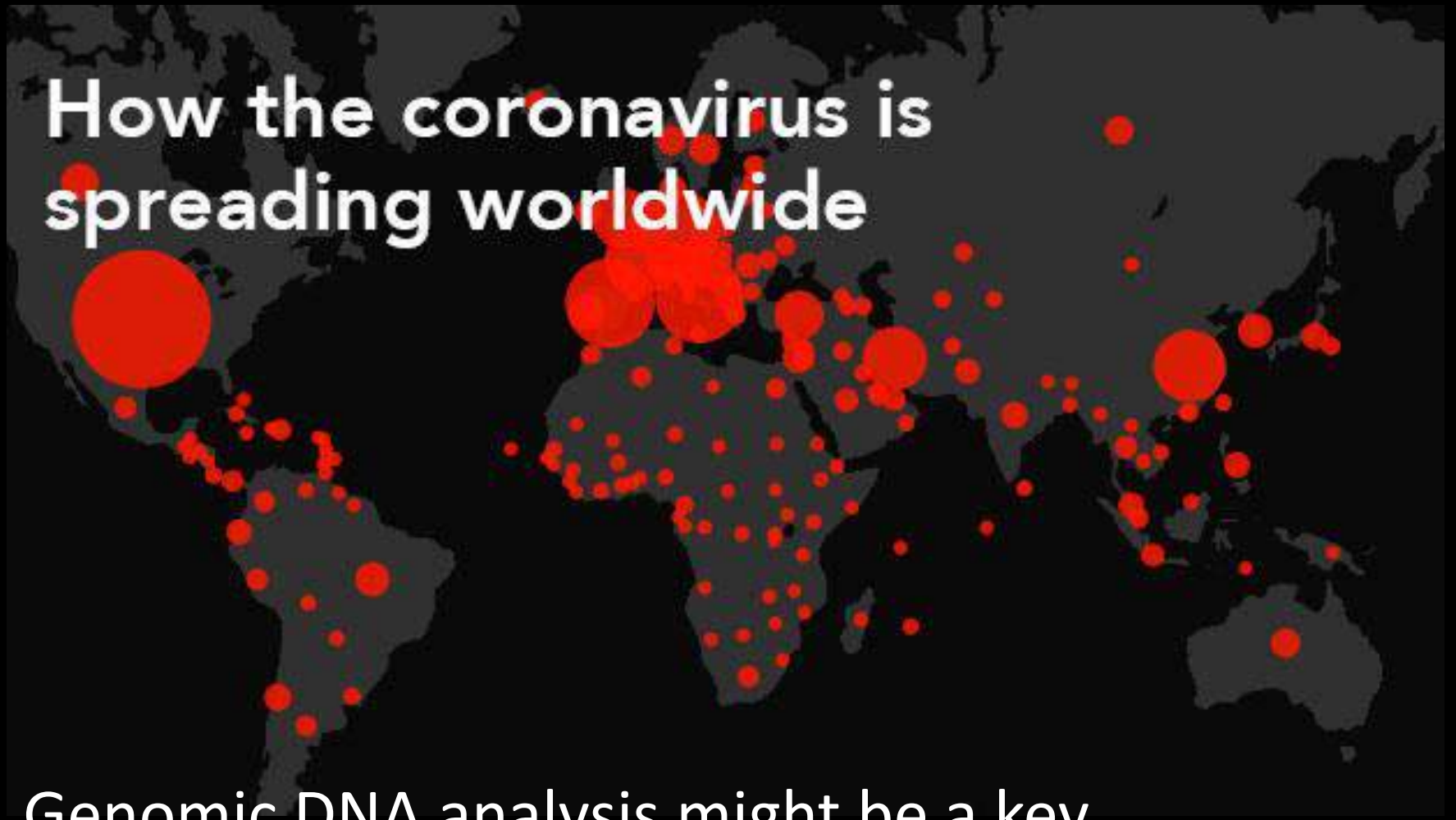
Covid-19の重症度の主な遺伝的危険因子はネアンデルタール人に起源がある

COVID-19 Host Genetics Initiative



Minor allele frequency at rs35044562. Frequency data were obtained from the 1000 Genomes Project[22]. Map source data were obtained from OpenStreetMap[23].

Nature. 2020 Sep 30. doi:10.1038/s41586-020-2818-3.



Genomic DNA analysis might be a key to survey Covid-19 spreading

英国NHS-digital医療情報統合システムにより新型コロナに合併する難病の予後予測が可能

-先天的代謝異常
重度の複合免疫不全症
ホモ接合型鎌状赤血球症

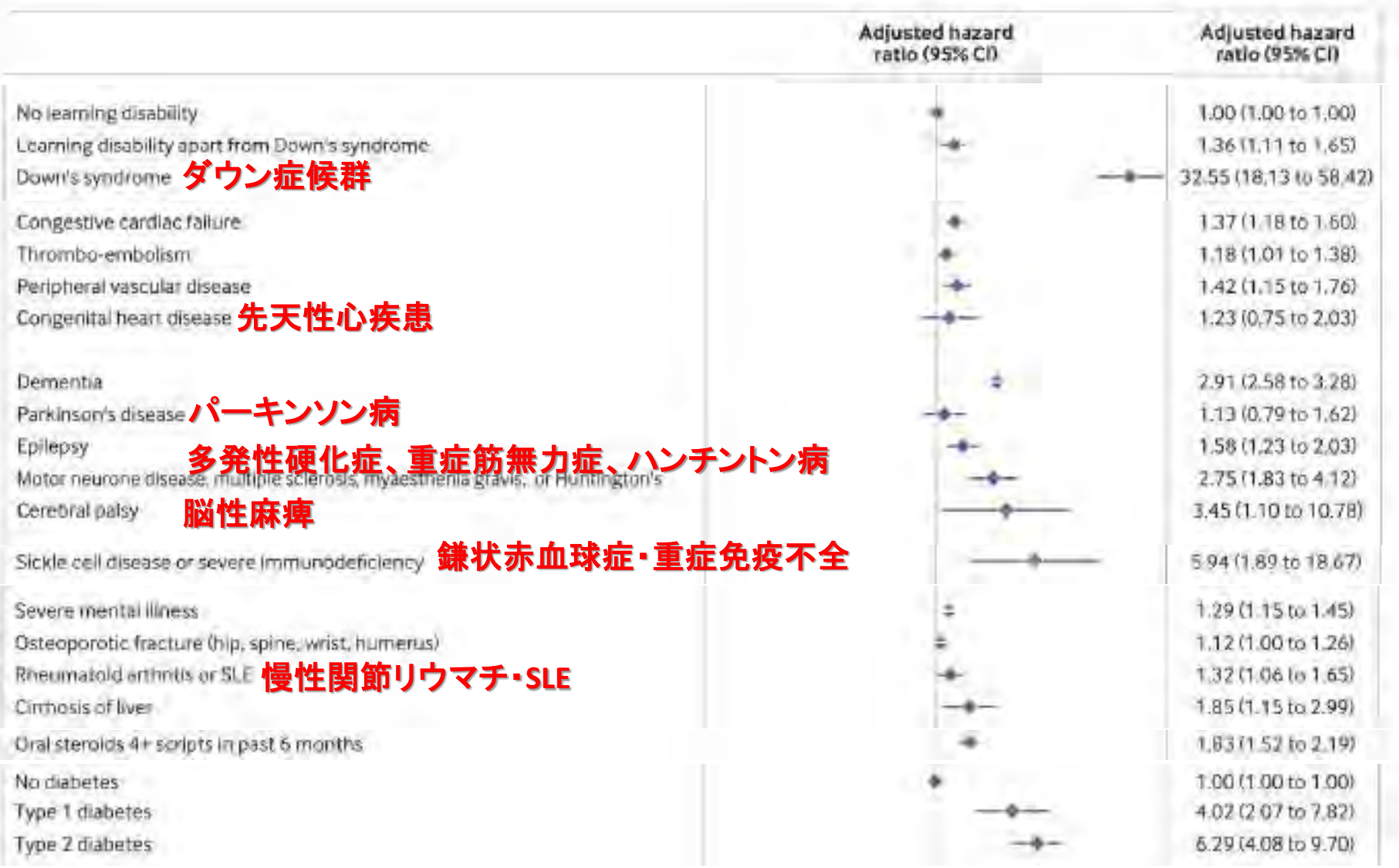
-慢性肝疾患
一次胆汁性肝硬変
一次硬化性胆管炎
ヘモクロマトーシス

-慢性神経疾患
てんかん
パーキンソン病
運動ニューロン疾患
脳性麻痺
ダウン症

- 自己免疫性疾患
関節リウマチ
全身性エリテマトーデス
強直性脊椎炎またはその他の炎症性関節症（例：乾癬性関節炎）
結合組織疾患（例：エーラーズ・ダンロス症候群、強皮症、シェーグレン症候群
多発性筋炎または皮膚筋炎
血管炎（例：巨細胞性動脈炎、結節性多発性動脈炎、ベーチェット症候群）

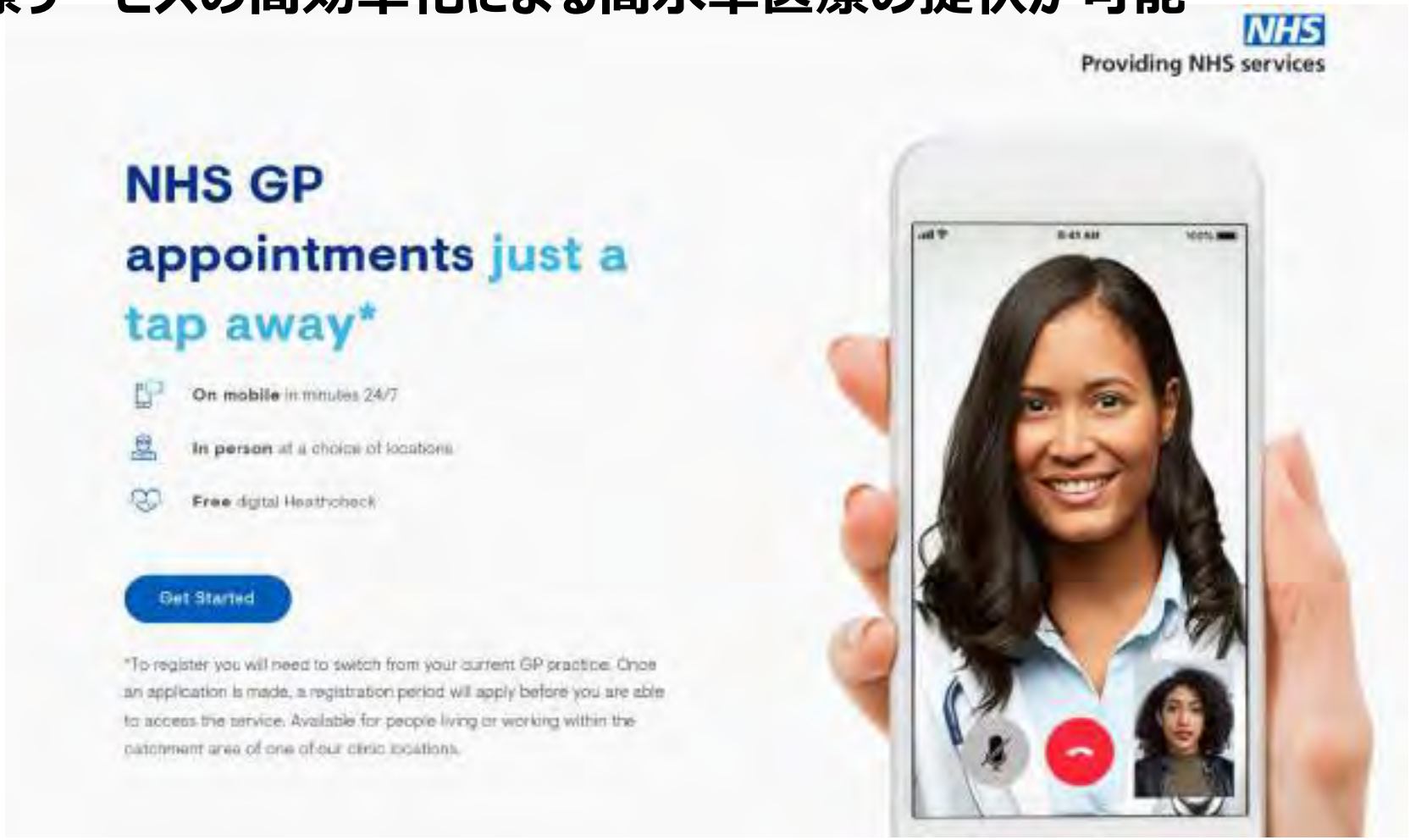
BMJ 2020;371:m3731 | doi: 10.1136/bmj.m3731

新型コロナによる難病の疾患別死亡の相対危険度

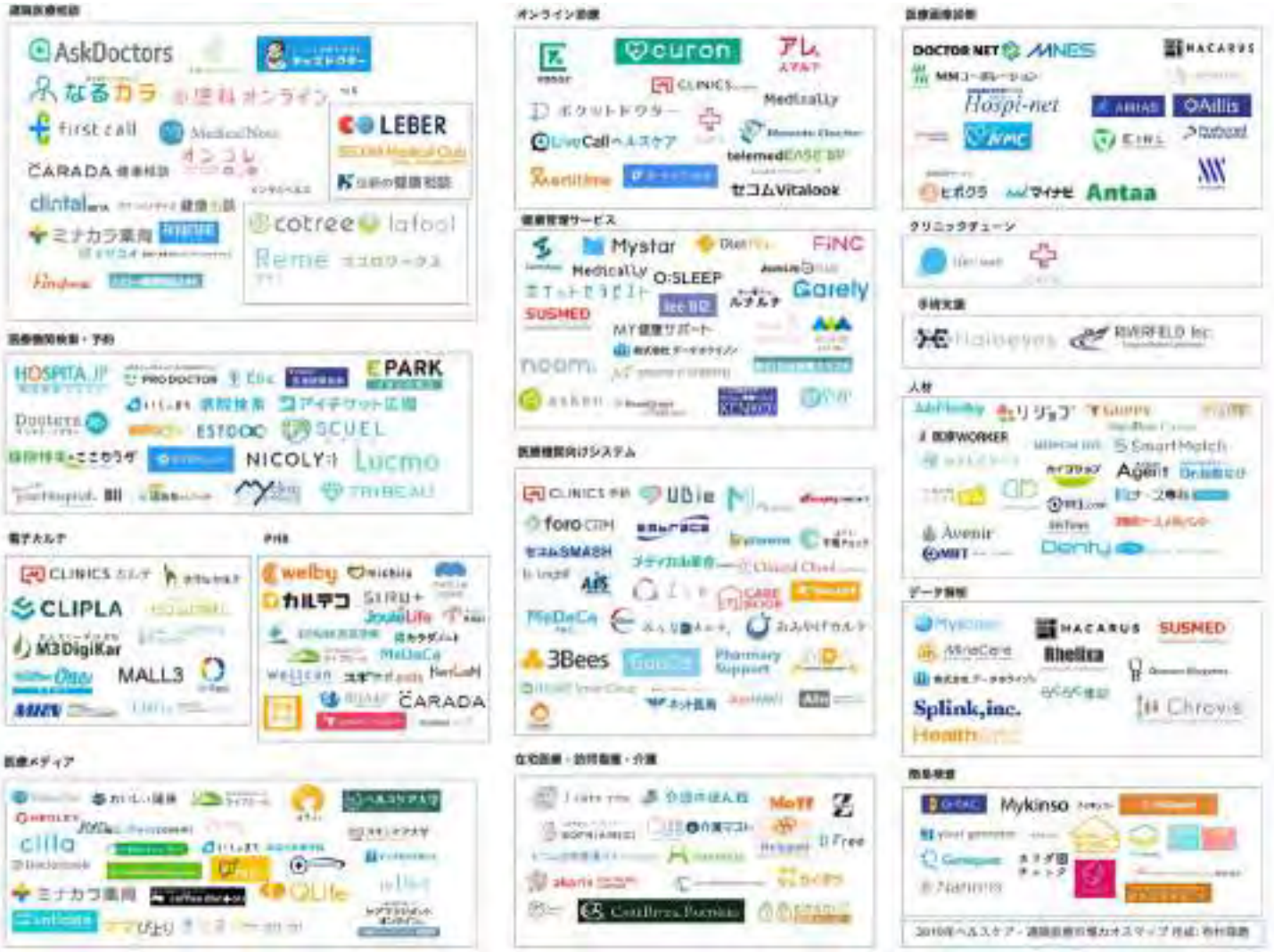


BMJ 2020;371:m3731 | doi: 10.1136/bmj.m3731

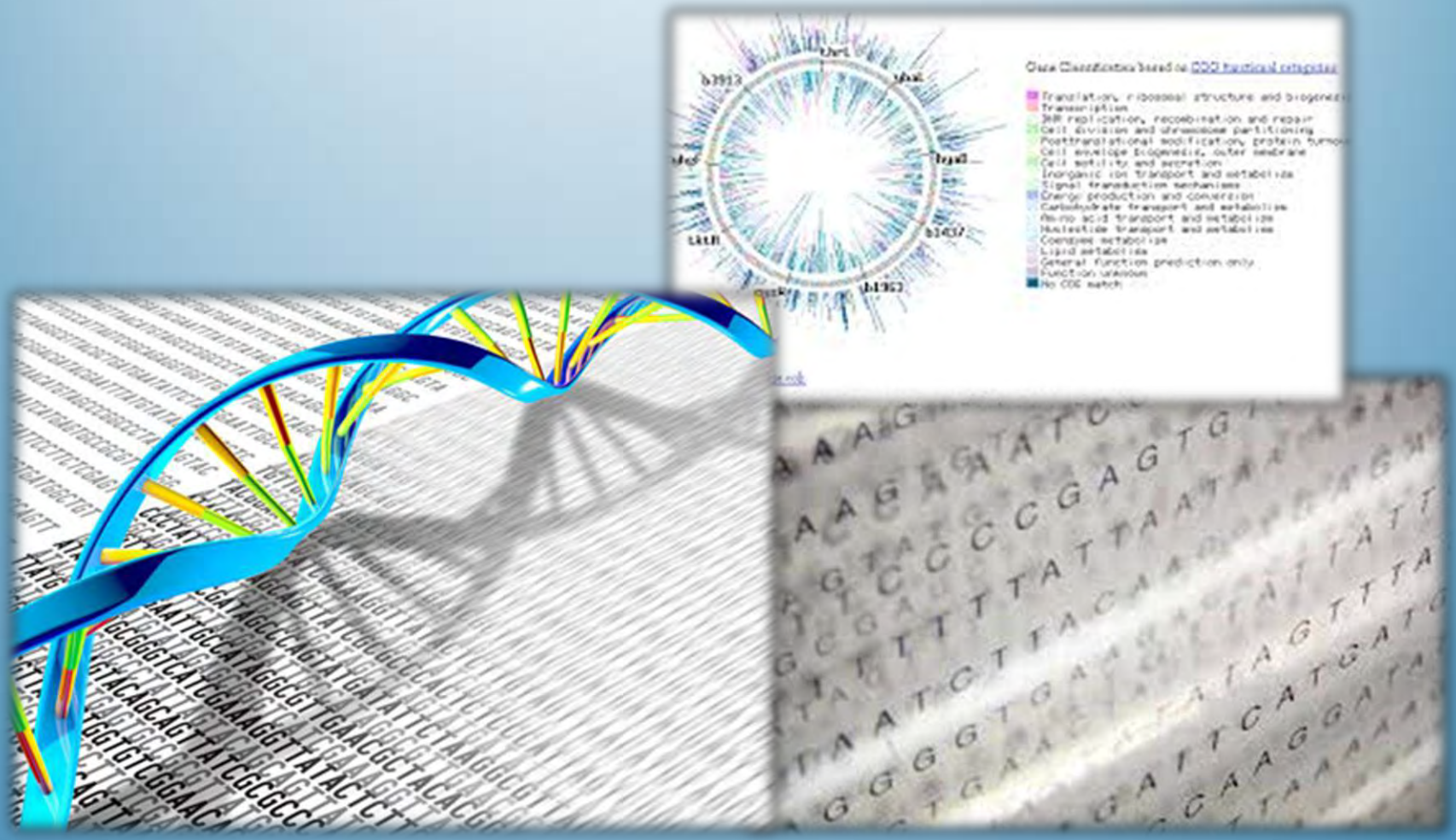
・NHS加入者は無料で事業公共性の高いデジタル医療を受診可能
・医療サービスの高効率化による高水準医療の提供が可能



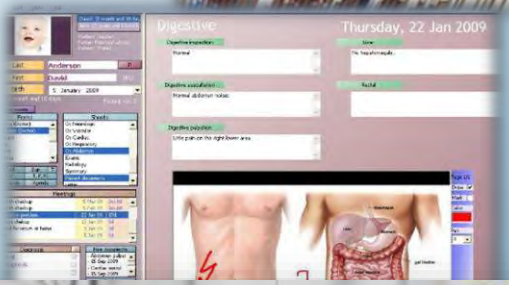
国内には未だ一定の標準規格化がなされておらず、社会医療情報の分散統合は未完のまま

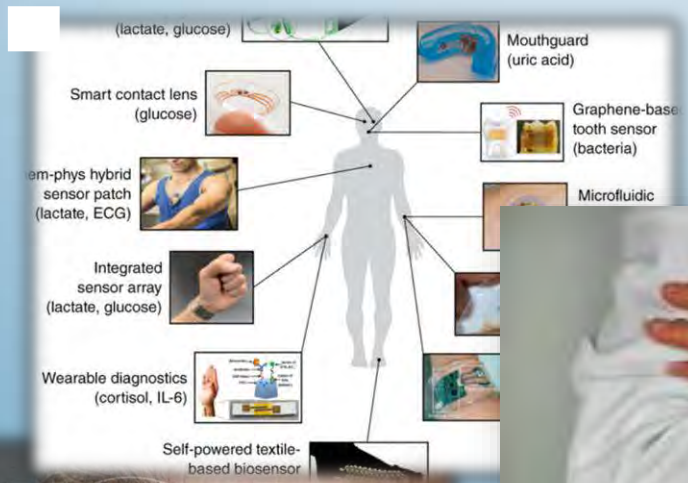


遺伝子情報

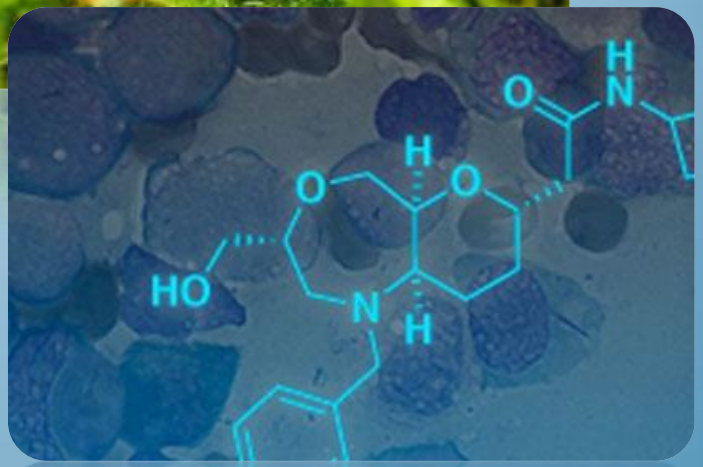


臨床情報・センサー情報


Digestive
Thursday, 22 Jan 2009


(lactate, glucose)
Mouthguard
(uric acid)
Smart contact lens
(glucose)
Graphene-base
tooth sensor
(bacteria)
em-phys hybrid
sensor patch
(lactate, ECG)
Microfluidic
Integrated
sensor array
(lactate, glucose)
Wearable diagnostics
(cortisol, IL-6)
Self-powered textile-
based biosensor

環境因子



感染・ワクチン

Precision medicine

Information on gene, life style and circumstance for individual healthy life course

Precision Public Health

Providing the right health intervention to the right people at the right time reducing health disparities.

Data-driven biosocial medical service

- 積極的な国際標準化政策誘導及び政府開発投資
- 持続可能な産学連携技術開発の推進

Life Stage

Maternal & Child | Healthy | Monomorbidity | Multimorbidity

Social

Personal

Molecular

Maternal and Child registry

Healthy-control registry

Non-Communicable diseases registry

Healthy longevity/Death registry

Genome-Phenome data sharing

Personal health record data sharing

Social-networking data sharing

Open data & open access

Open Science

Academia **Industry**