Health-RI voor effectieve personalised medicine research
SELL THE PROBLEM YOU SOLVE, NOT THE PRODUCT.
Why Health-RI?

research  care  innovation
If want better treatment for patients, then we must:

- change clinical & public health practices
- change guidelines
- generate high-level evidence
- conduct research providing such evidence
- this requires an excellent research infrastructure
Personalized medicine & health
Personalized medicine: drugs

Diagram showing the relationship between patient, clinical phenotype, disease biology, and improved outcome.
Personalized medicine: drugs
Personalized medicine: biomarkers

the omics revolution
Personalized medicine: pathology

GCP
good clinical practice

CROs
contract research organizations

Improved Outcome

Clinical Phenotype

Patient

Disease biology

etc.
How can big data change science?
Here's how medical research traditionally works:

1. Come up with a question or hypothesis.
2. Design an experiment to test it. Wait for new data to come in.
3. Form your conclusion.

Big data changes step 2
Online, searchable databases provide instant answers, speeding up research.
About 80% of patient information is unstructured, and in turn, unmineable. Advancements in voice recognition and clinical language understanding are enabling the healthcare enterprise to capture information at the point of care, convert patient data into actionable information, and leverage that information for clinical, business, and patient good.
Translating Data to Health

Workshop: 21 - 25 March 2016, Leiden, the Netherlands

Scientific Organizers:
- Niklas Blomberg, ELIXIR Hinxton
- Jan-Willem Bölen, Lygature Eindhoven
- Scott Lusher, Johnson & Johnson Beerse
- Gerrit Meijer, NKI Amsterdam

Topics:
- One Patient - One Petabyte
- Rethinking Data-Driven Translational Research
- Crossing the Research-Clinical Care Line
- Social & Regulatory Aspects
- FAIR Data Sharing

Social!

The Lorentz Center is an internation
the sciences. Its aim is to orga
for scientists in an annual
and collaborative work, discussions a
For registration see www.

This workshop is part of the
program, stimulating multi-
data-intensive research in the
developing and application.

background image: One patient - One petabyte in translational research; Paper design: Srh,
The underlying problem...

FRAGMENTATION of...

• data
• sample collections
• image collections
• regulations
• software tools
• research initiatives
• funding
• expertise
• etc.
researchers want hassle free research...
.... and patients want fast innovation
2016

Health RI
Research Infrastructure

2014

eatris
Biobanking and BioMolecular resources
Research Infrastructure
The Netherlands

200 biobanks

- 2009
Link naar het filmpje:
https://www.bbmri.nl/health_ri/1563/
Internet of Personalized Medicine & Health Data & Things

- Biobanks, cohorts
- Wearables
- MRI center
- Research laboratory
- Twitter data
- Clinical records
- Microscopy facility
# Health-RI value proposition

## Health-RI services and activities lead to an infrastructure that facilitates the research process

The infrastructure set up by Health-RI facilitates the different steps in the research process (non-exhausting) by providing:

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The Health-RI platform offers researchers solutions on demand for each step of the research process. By simultaneously operating on tactical and strategic level, Health-RI connects the needs of researchers and other stakeholders (funders, society, etc.) transforming the total personalized medicine & health research system.
Health-RI value proposition

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**Catalogue**

- data
- samples
- images

**Operational level (platform services)**
- FAIR data management training, education and support
- Compliance tools and legislation
- Biobanking registry support center
- Central catalogue and request tool for data, samples and images

**Tactical level (connecting the network)**
- Connected registries and biobanks
- Single identification, authentication and authorization system
- FAIR data management guidelines
- Citizens / patients a voice in the strategic committee
- Standardize ethical review procedures
- A network of ELSI experts offering ELSI support
- Compliance - by - design

**Strategical level (transforming the system)**
- Increasing citizen participation
- Promotion of the transformation to registration at the source in healthcare
- Promotion of omnibus legislations
- Coordination of investments in data infrastructures via the financing agenda, resulting in sustainable long-term funding
- Coordination of investments in IT tools and facilities infrastructures via the financing agenda, resulting in sustainable long-term funding
- Promotion of active data sharing and open science policy
- Promotion of learning care system

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**Level of services and activities**

**Operational level (platform services)**
- Define research question
  - FAIR data management training, education and support
- Experimental design
  - Compliance tools and legislation
  - Biobanking registry support center
- Access to data and samples
  - Central catalogue and request tool for data, samples and images
- Data generation
  - Catalogue of facilities & expertise centers
  - Interoperable data acquisition tools
  - Linked data-backbone
- Analysis
  - Interoperable analysis tools
  - Federated analysis workspace
  - Helpdesk IT-tools and services
- Translation and dissemination
  - FAIRifier tools
  - Data stewardship training
- Improvement of research/care
  - Researcher-participant interaction App

**Tactical level (connecting the network)**
- Promotion of the opening up of industry trial data
- Promotion of all data to be available in the public domain
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- Coordination of investments in IT tools and facilities infrastructures via the financing agenda, resulting in sustainable long-term funding
- Promotion of active data sharing and open science policy
- Participation in Health-deals
- Promotion of public-private partnerships

**Strategical level (transforming the system)**
- Participation in developing ELSI guidelines
  - Covenants for use of facilities by others
  - Harmonize data acquisition protocols
  - Interoperability standards
  - Interoperable analysis tools
  - Federated analysis workspace
  - Helpdesk IT-tools and services
  - Coordination of IT platforms
  - FAIRifier tools
  - Data stewardship training
  - Connections to Open science initiatives
  - Promotion of learning care system
  - Promotion of active data sharing and open science policy
  - Promotion of public-private partnerships

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Operational shared services

Catalogue
- data
- phenotype
- omics
- wearables
- images
- samples

ELSI services
- compliance by design to regulations
- privacy
- influencing legislation
- mybiobank etc

IT services
- standardisation
- online digital research environment
- “office 365” for personalized medicine & health research
- interfacing to advanced technologies
- deep learning pipelines for image analysis
- multi-omics pipelines
- etc

Personalized Medicine Research Process Management
An ‘office’ suite

Microsoft Office 365
"Office 365" suite for translational research

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Data integration and analysis

- cBioPortal for Cancer Genomics

Deployment and user support

- ServiceDesk

http://www.ctmm-trait.nl/trait-tools
“Office 365” suite for translational research

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Data integration and analysis

- ![cBioPortal](https://example.com) for Cancer Genomics

Deployment and user support

- ![ServiceDesk](https://example.com)

http://www.ctmm-trait.nl/trait-tools
rescue manual curation efforts

clinical trials

cohort studies

Registry

retrospective studies
A practical example:
I would like to perform a study!

**Topic:** breast cancer in men (age 18-50 years)

**Questions:** Where do I begin?
Are there already existing datasets?
“Office 365” suite for translational research

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Data integration and analysis

Deployment and user support

http://www.ctmm-trait.nl/trait-tools
A practical example

**FAIR: Findable**

Study: breast cancer in men

Question: of how many men (age 18-50 years) is material available?
   → data, samples, treatment and follow-up

Step 1: Search and find:

- **Catalogue:**
  - National (cyto)pathological Public Archive (Palga Openbare Databank)
  - Dutch Cancer Registry (IKNL)
  - Central Bureau for Statistics (CBS) statline
A practical example

**FAIR: Accessible**

Step 2: Online request portal

![Image of online request portal with fields for Project leader, Pathologist, Previous contact, and Project title and background information.](image_url)
Step 3: Wait for a linked dataset

“A practical example
FAIR: Interoperable

Available, pseudonymized, dataset and samples for research
A practical example

Step 4: Storage and editing of data, samples, images in Health-RI tools
A practical example

**FAIR: Reusable**

Step 5: Publish and share!

- Clinical data
- Pathology data
- Imaging data
- Biobank data
- Experimental data

Also publish and share the ‘final’ data itself, not just conclusions

Accessible and insightful for non bio-informaticians!
‘Final’ data available for view and query within the data-integration platforms

Possible to view existing data, query and analyze the data within these platforms
Background tranSMART

- Originally designed as a data-warehouse by Johnson & Johnson and Recombinant Data Corporation (2009), used internally for view and analysis

- Software turned open-source in 2012 in the context of IMI ETRIKS

- tranSMART Foundation (2013), a public-private partnership, established an open source community (USA and EU collaborators)

- tranSMART Foundation & i2b2 Foundation merged in 2017
Over 50 implementations
‘Final’ data available for view and query within the data-integration platforms.

Possible to view existing data, query and analyze the data within these platforms.
The trick: apply FAIR to best practices

- BBMRI-NL catalogue (Findable, Accessible)
- Parelsnoer PRISMA data model
- Request portal DNTP developed for PALGA
- ELSI support center BBMRI-NL & COREON
- Molgenis & tranSMART portals for downstream analysis
- cBioportal for intuitive visualisation
- Minimal data sets: Registration at the Source, involving medical associations
- Quality assurance & process optimization: EATRIS
- Standardize ETL between large registries (NKR-oncology; NHR-cardiology, etc.)
- FAIRification expertise: DTL/ELIXIR-NL
- Digital Research Environment Data4lifesciences/ELIXIR
- Harmonized organization of institutional back offices D4LS
- Effective helpdesk for end users
business plan by KPMG started in spring

1\textsuperscript{st} stakeholder meeting june

2\textsuperscript{nd} version draft with project group

will be sent off to stakeholders 5/10

2\textsuperscript{nd} stakeholder meeting 12/10

presentation final version business plan 8/12

establish Health-RI organization Q2 2018
Health-RI wrap up

A) we all share the same problem
   - innovation gap in PM&H research
   - substandard infrastructure

B) solution is clear
   - defragmentation
   - standardization / FAIR
   - bring users & infrastructures together

C) how to organize
   - efficient & effective
   - inclusive

D) how to finance
   - integrated approach
   - with all payers
   - basic component + fee for service
That’s why!

basic research > excellent science > more cure
Endorsed by stakeholders, and the people behind these