

#### **Chairs: David Lomas and Jim Smith**

Mike Barnes, Ines Barroso, Ewan Birney, Phil Beales, Mark Caulfield, Dave Hawkes, Harry Hemingway, Martin Hibberd, Tim Hubbard, Nick Luscombe, Seb Ourselin, Charlie Swanton, Richard Trembath

UCL Partners, Crick, Sanger, EBI, QMUL, LSHTM, KCL

#### Medical Bioinformatics: Data-Driven Discovery for Personalised Medicine

P.L. Bezies (UCLP), M. Caulfield (UCLP), P.V. Coveney (UCLP), D. Hawkes (UCLP), H. Hemingway (UCLP), T.J. Hubbard (Sanger), D.A. Lomas (UCLP), N.M. Luscombe (UCLP, Crick), J.P. Overington (EBI), L. Smeeth (UCLP), J.C. Smith (Crick), C. Swanton (UCLP, Crick)

- 1. Objectives
- 2. The Partnership
- 3. Disease Types
- 4. eMedLab Hardware Infrastructure
- 5. Research and Training Academy
- 6. Coordinating Analytics Research: Academy Labs
- 7. Strategic Issues
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- 9. Metrics for Success

#### 1. Objectives

Our vision is to maximise the gains for patients and for medical research that will come from the explosion in human health data. To realise this potential we need to accumulate medical and biological data on an imprecedented scale and complexity, to coordinate it, to store it safely and securely, and to make it readily available to interested researchers. It is vital to develop people with the skills and expertise to exploit these data for the benefit of patients. Together, UCL Parmers, the Francis Crick Institute. Sanger Institute and the European Bioinformatics Institute shall deliver the following:

#### 1.1 Create a powerful eMedLab E-infrastructure (lead: Smith)

We are hampered in our work to generate new medical insights because of the fragmented accessibility of fundamental clinical and research data, and the lack of a high-performance computing (HPC) facility in which to analyse them. We shall build eMealLab, a shared computer einster to integrate and share heterogeneous data from personal healthcare records, imaging, pharmacoinformatics and genomics. Through co-location, we will eliminate the delays and security risks that occur when data are moved. It also provides a platform to develop analytical tools that allow biomedical researchers to transform raw data into scientific insights and clinical outcomes, eMedLab will store data security and its modular

"We will maximise the gains for patients and for medical research that will come from the explosion in human health data"

How do we achieve this?









# eMedLab infrastructure

- Shared computer cluster
- Integrate exchange heterogeneous data
- Methods and insights across diseases

# eMedLab academy

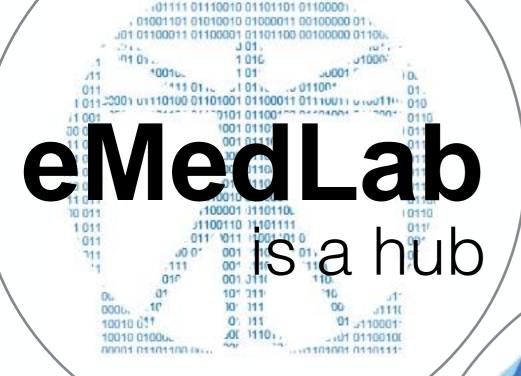
- Recruit outstanding Career Dev Fellows
- Training workshops and courses
- Promote collaborations via "Labs"

# Research operations

"We will maximise the gains for patients and for medical research that will come from the explosion in human health data"

How do we achieve this?

# eMedLab enables projects through infrastructure and people



## 3 disease areas

>6M patients



partners









genomic



electronic health records













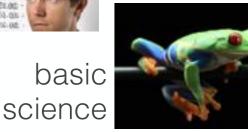
EMBL-EBI



clinician scientists

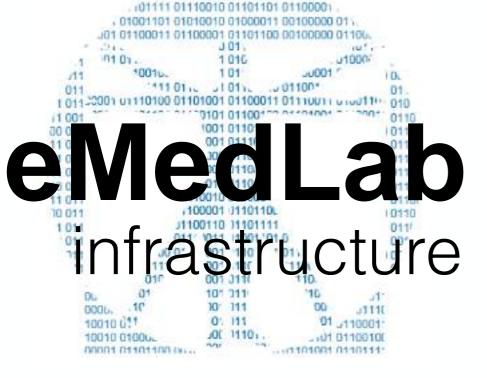


analytics



3 expertises

Infrastructure



# Private cloud computing

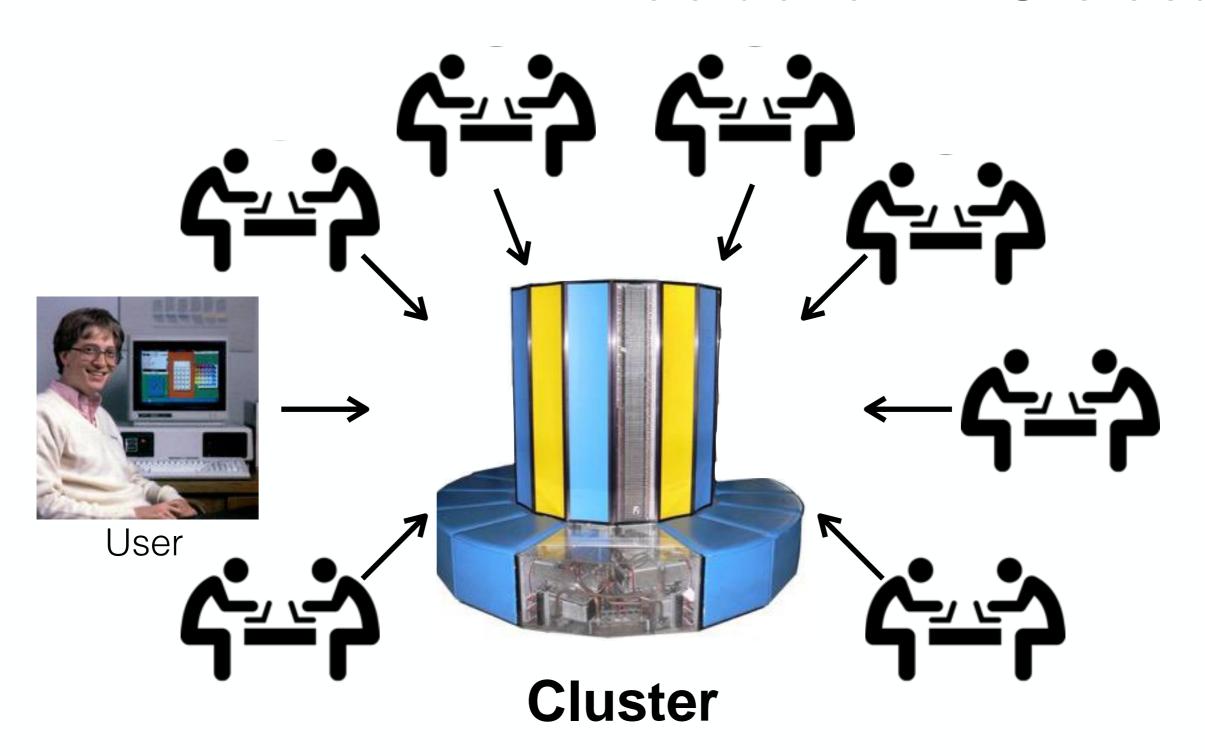
for medical bioinformatics

- One of the largest high-spec private cloud computing systems
- Very high specifications
  - 6048 cores
  - 5.5PB data storage
  - Fast interconnects
- Flexible, transferrable, accessible





# Traditional HPC cluster

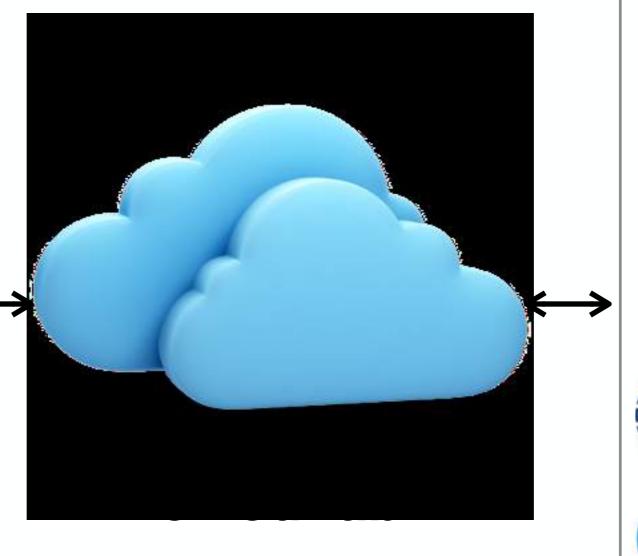


# eMedLab users Virtual machine environment

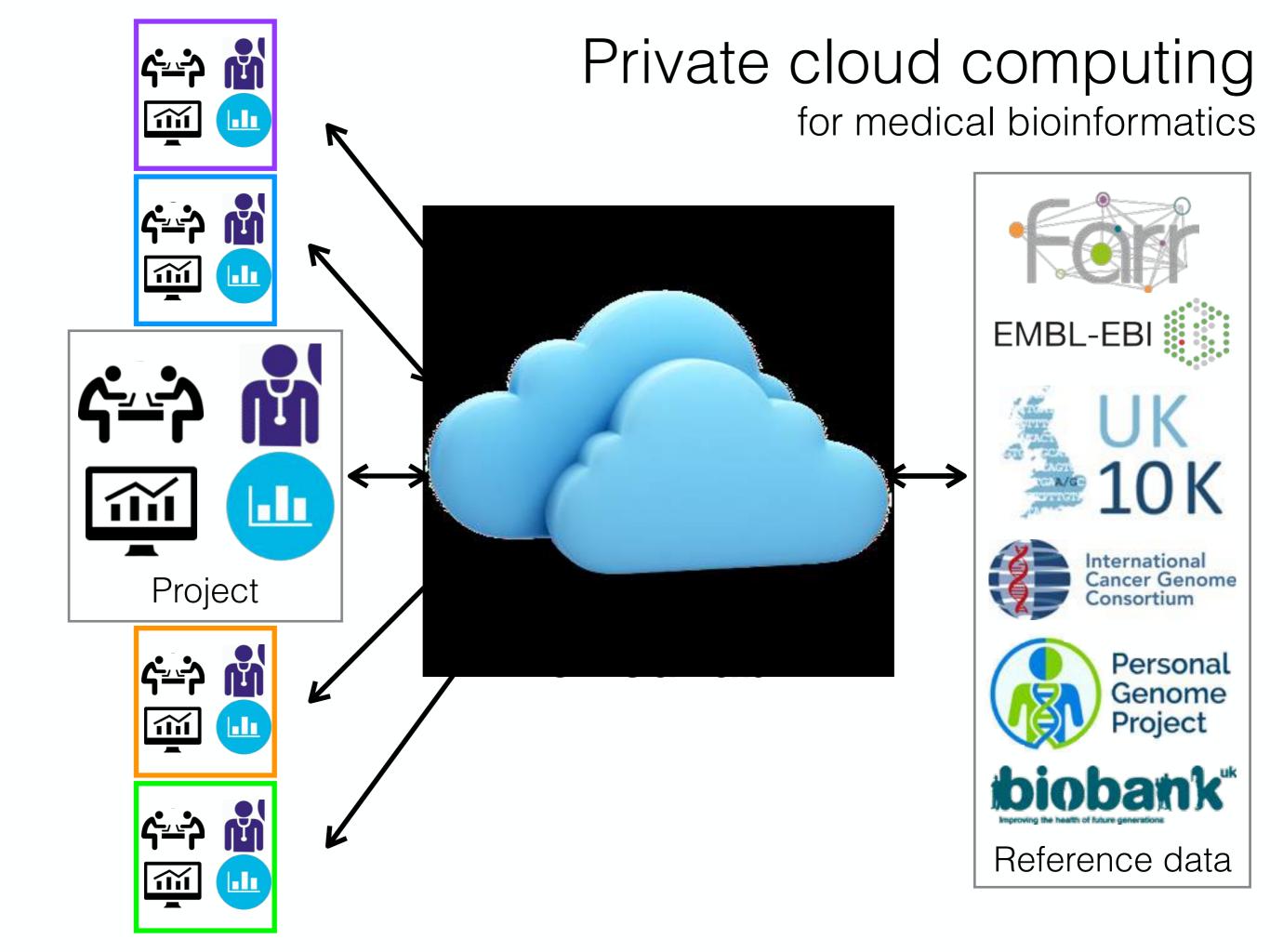
User data

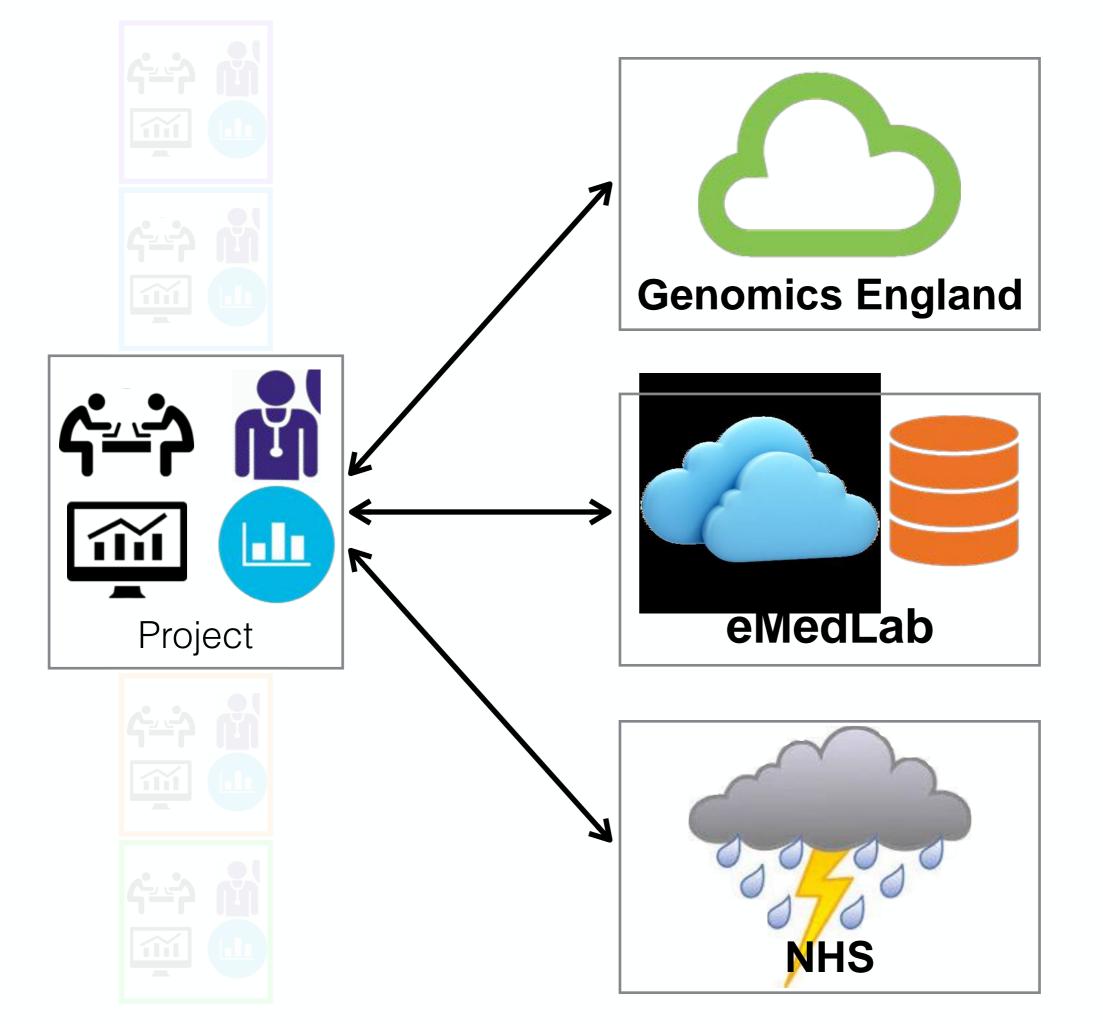
# Private cloud computing

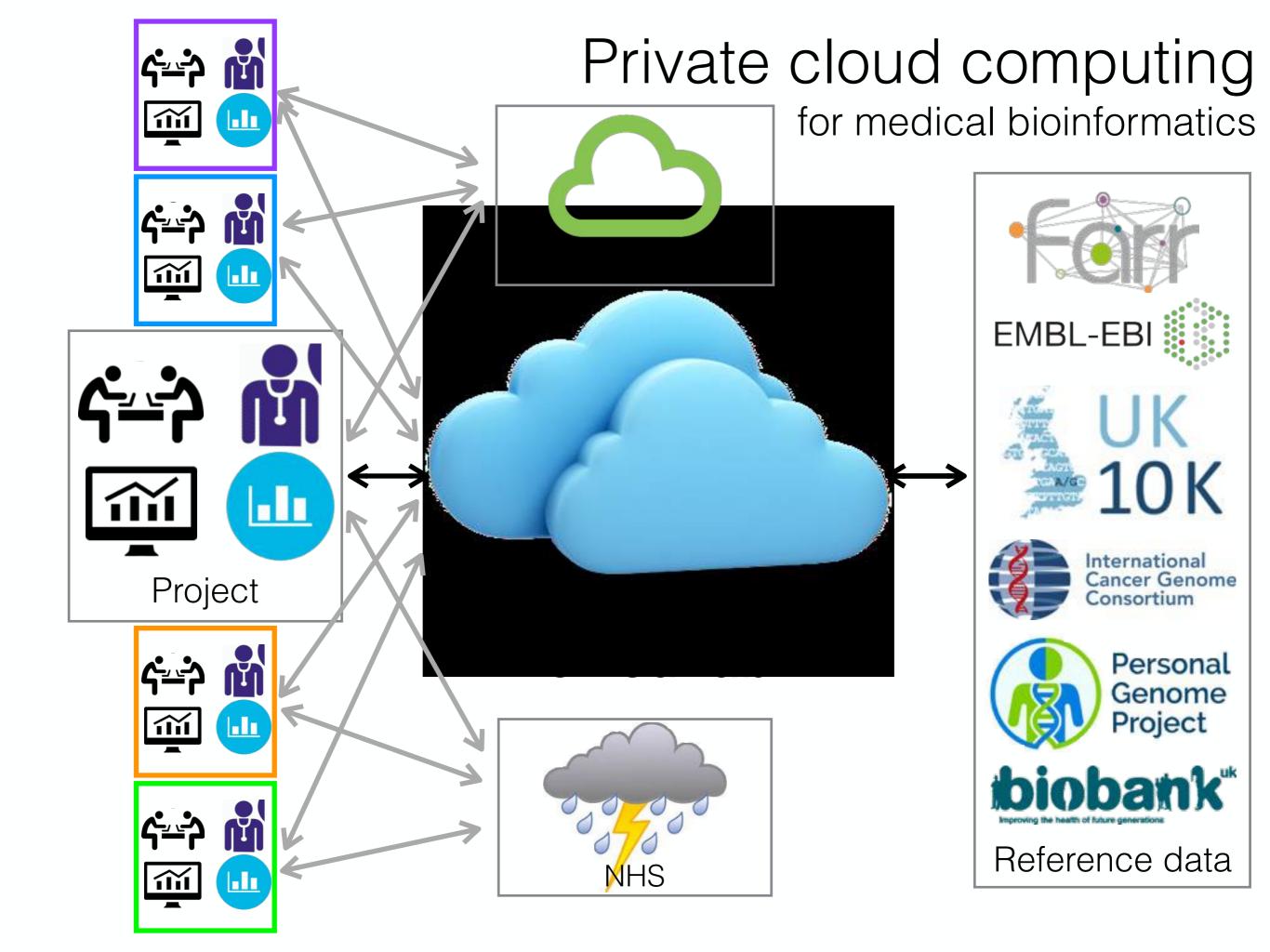
for medical bioinformatics

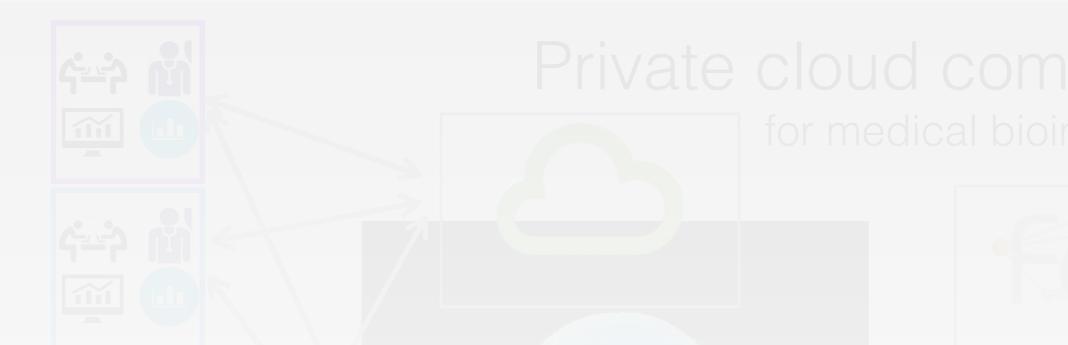








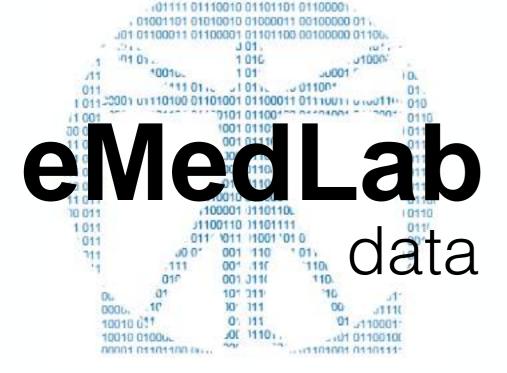




Flexible, transferrable, accessible

"This is very exciting"
Computer Weekly

Data management



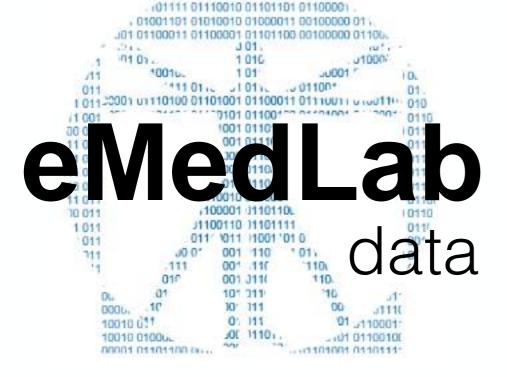
# Principles

#### Anonymised data:

eMedLab can host anonymised clinical data subject to local ethics approval

#### Pseudoanonymised data:

 eMedLab can host pseudoanonymised data from patients who have given consent for research



# Principles

#### • Reference data:

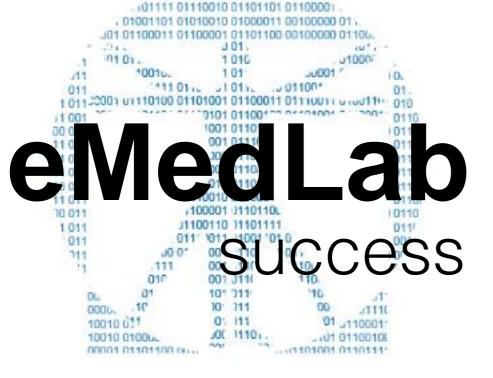
- Suitable for data with high levels of commonality across projects
- Users are responsible for approvals to access
- Access managed through VM configurations

#### Specific data:

- Uploaded to eMedLab via project-specific VMs
- Extend access to other projects through VM cross-talk

#### Data manager:

Full-time manager for curation and access of reference data



# Challenges

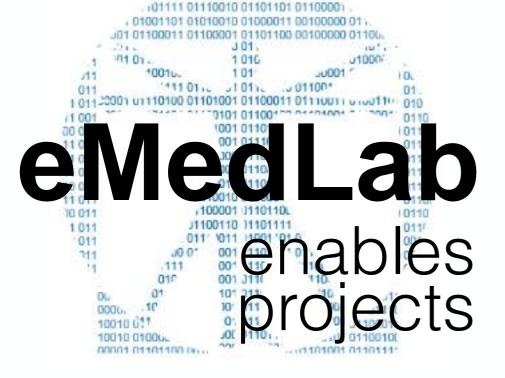
#### Linking with identifiable data:

- eMedLab will not host identifiable data
- Technical solutions:
  - Virtual Machines go to the data
  - Link remotely with identifiable data in Farr Safe Haven and others
  - A Safe Haven Virtual Machine that hard-codes ISO 27001 requirements?

#### Disparate data models:

- Harmonise data models across centres
- Global Alliance for Genomics & Health (https://genomicsandhealth.org/)
- Observational Medical Outcomes Partnership (http://omop.org/)

**Projects** 

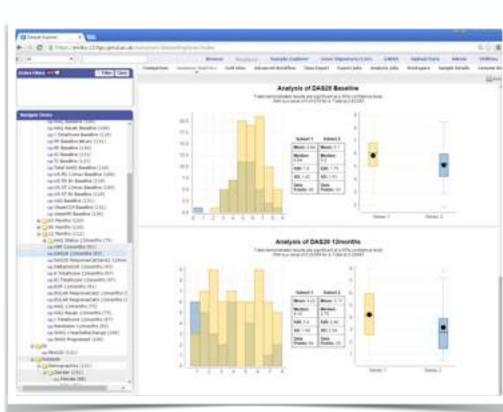


# StratMed platform MRC Matura Consortium



- Rheumatoid arthritis doesn't always respond to biologics
- Data: -omic and clinical trial data for 10,000 patients across 21 partners
- How: Integrate and present information to clinician researchers
- Who: clinicians, bioinformaticians, pharma
- Outcomes

Biomarkers to predict responses to biologics Effective treatment programmes for patients

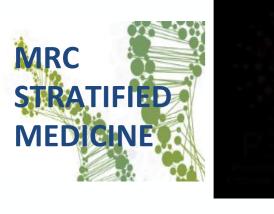


# StratMed platform

Joined up MRC Infrastructure







**2** Farr London

Hosting and prototyping enabled by Farr-funded Virtual Machine (VM) cluster at QMUL









Stratified medicine VMs
Transferred to eMedLab
to enable HPC and
Machine Learning
applications

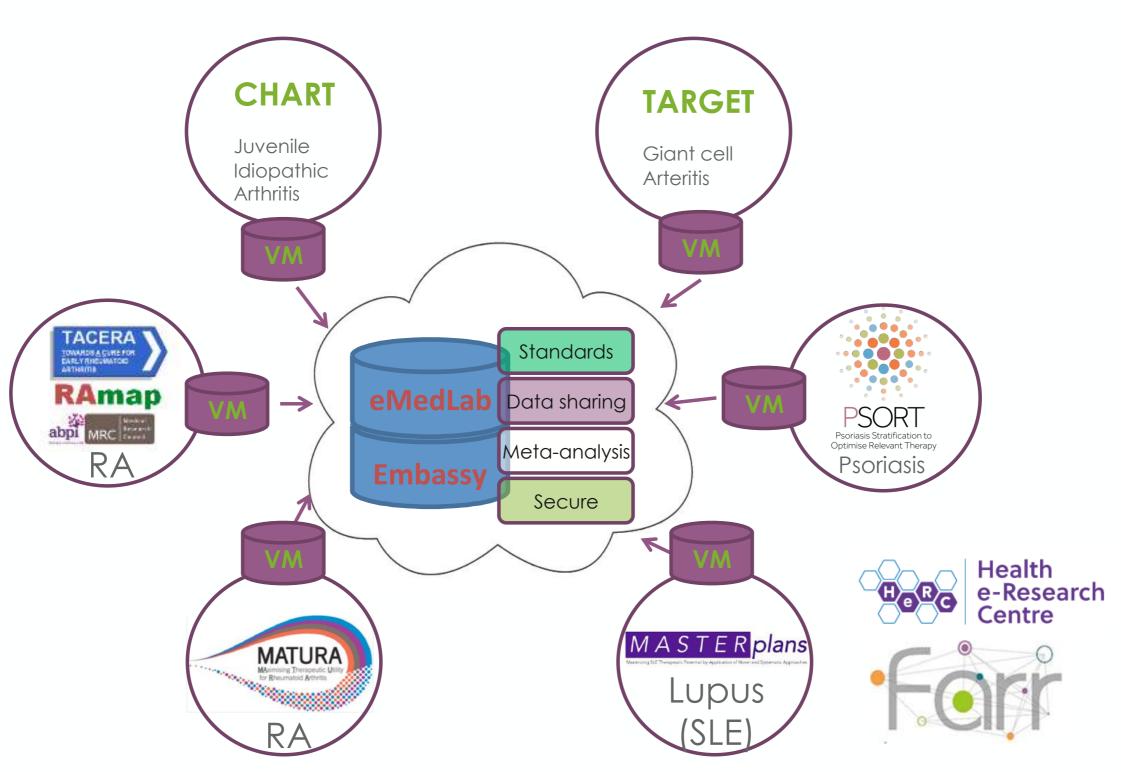
3 eMedLab

RA-Map, MATURA & PSORT adopt a federated TranSMART/i2b2 infrastructure

Strat Med

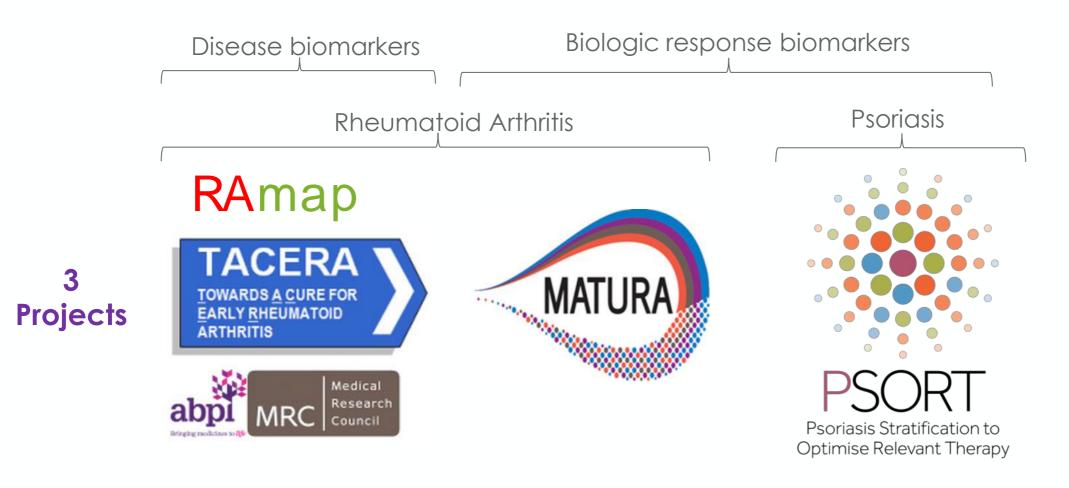
# eMedLab is building synergy

and facilitating data sharing



# eMedLab is building synergy

and facilitating data sharing





















**MANY PARTNERS** 























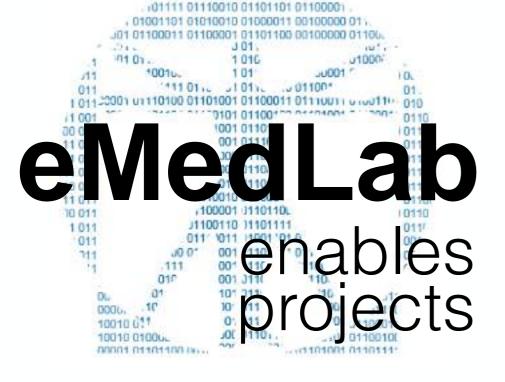












# Cancer genomics Swanton & Van Loo

Cancers evolve heterogeneously

• Data: genomes, MRI, histopathology

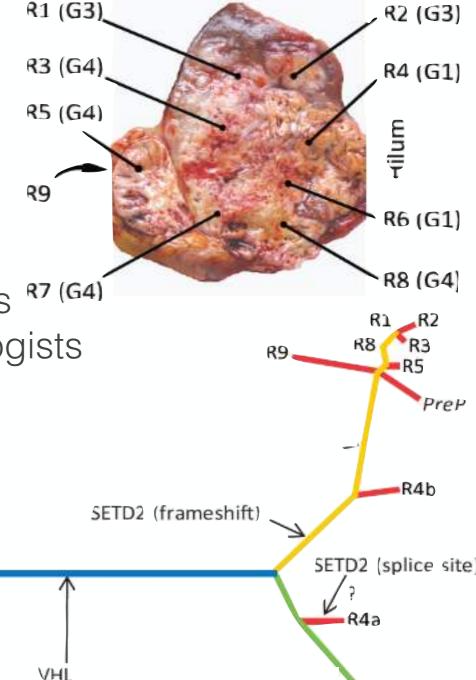
• How: Phylogenetic analysis of cancer genomes

• Who: clinicians, statisticians, evolutionary biologists

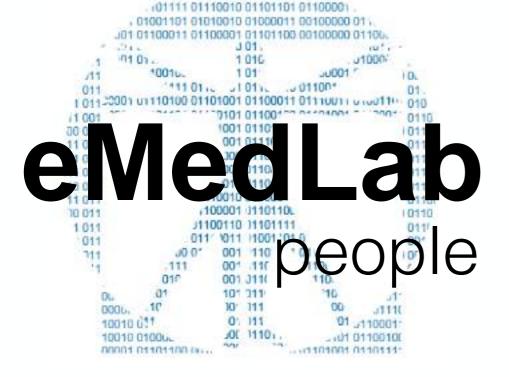
#### Outcomes

Mechanisms of cancer progression

DARWIN II clinical trial to target clonal drivers



People



# career development fellows have been recruited internationally



#### **Andre Altman**

- Neurodegenerative diseases
- Stanford to UCL
- Altmann, Science 2015



### Helena Kilpinen

- Rare diseases and HiPSCI
- EBI to GOSH/Sanger
- Kilpinen, Science 2013



### **Alan Hodgkinson**

- Mitochondrial diseases
- McGill to KCL
- Hodgkinson, Science 2014



#### **Borbala Mifsud**

- Non-coding mutations
- Crick to QMUL
- Mifsud, Nat Genet 2015



# Research operations underpinning eMedLab

**Bruno Silva** 

Kuba Purebski Richard Christie

Tom King David Fergusson

David Ocana John Bouquiere

David Wong Mike Atkins

Rich Boyce Andy Cafferkey

Ric Passey Faruque Sarker Jackie Stewart

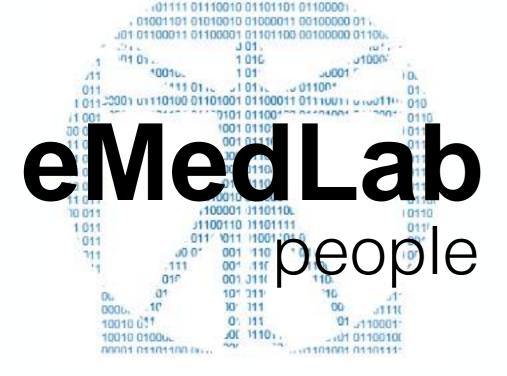
Luke Sudbury Tom Jones Gianni Dalla Torre

Pete Clapham James Beale Steve Whitbread

Adam Huffman Tim Cutts Luke Raimback

Stefan Boeing Josh Randall William Hay

- Federated support across partners
  - procurement
  - core infrastructure
  - data management
  - virtual machines...



# UK technical network for national infrastructure

- Future proofing technologies
  - team coordination
  - compatibility
  - secure data transfer
  - regulated data access...

Crick, EMBL-EBI, Sanger, Farr, Medical Bioinformatics, Genomics England, ADRC, JISC



**Jacky Pallas** 

Simon Thompson

Tim Hubbard

Jeremy Sharp

Alan Real

John Ainsworth

Mark Parsons

Malcolm Teague

Steve Pavis

Steve Newhouse

Tim Cutts

David Fergusson

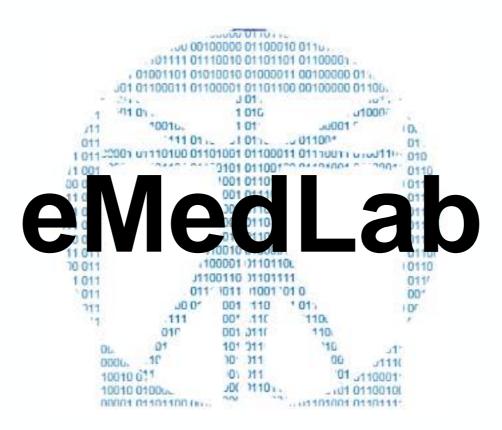
Success



# we've made progress but it's not enough!

- Sustainable infrastructure: enable data intensive clinical research
- <u>Capacity</u>: network of scientists, clinicians and research operatives
- Cross disease boundaries: exploit commonalities in data analysis
- Strategic alliances: Farr UK, Genomics England, Turing etc...

Clinical outcomes: scientific & clinical success through genomic, imaging and EHR data



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