Pathogen Genomics: from research to service, and beyond

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Supported by

MRC Cloud Infrastructure for Microbial Bioinformatics

Partneriaeth Genomeg Cymru Genomics Partnership Wales

Llywodraeth Cymru Welsh Government

BBSRC bioscience for the future

Conflict of Interest

No Conflicts to declare!

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MRC Cloud Infrastructure for Microbial Bioinformatics

MRC CLIMB

UKRI Medical Research Council

COG-UK, ConCOV

wellcome

SP3

BBSRC bioscience for the future

Microbes in the Food Chain

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Llywodraeth Cymru Welsh Government

Welsh Government

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Public Health Wales NHS Trust

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The ARCCA team

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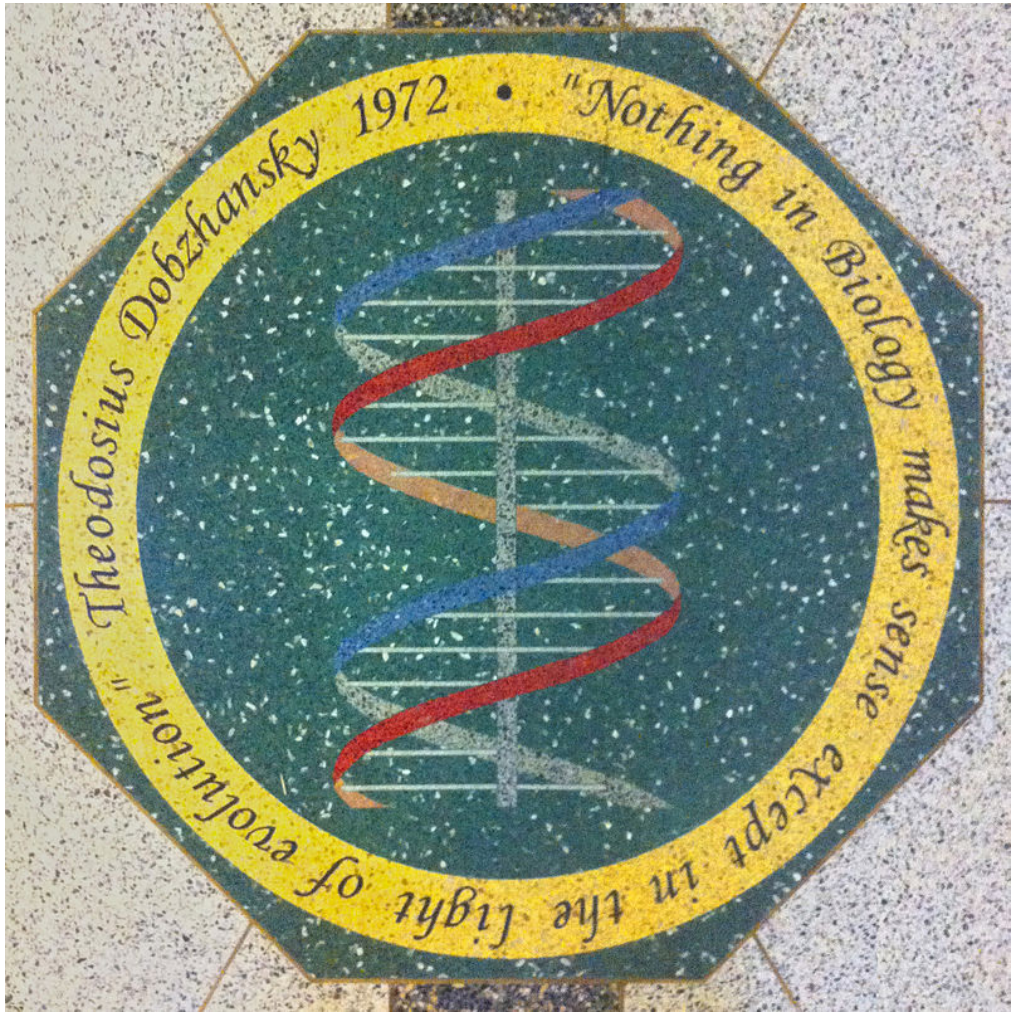
Dr Catie Williams

Amy Gaskin

The wider COG-UK Consortium : https://www.cogconsortium.uk/

Overview

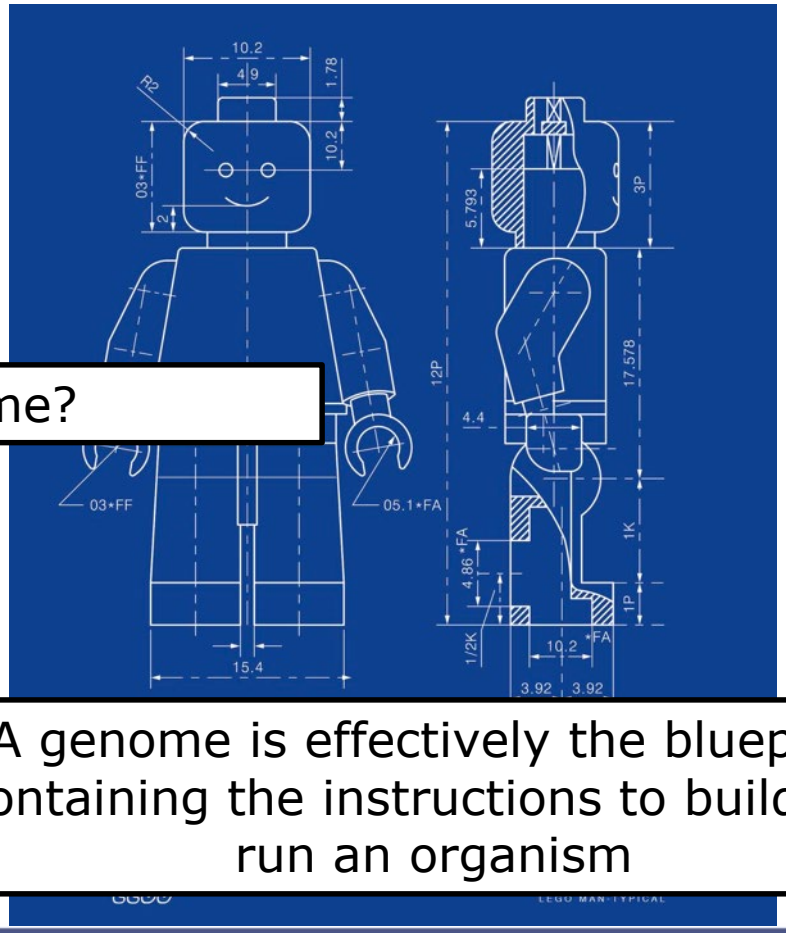
- Introduction to genomics
- Potential
- Key challenge
- Research and service in action
- The future



So, what is Genomics?

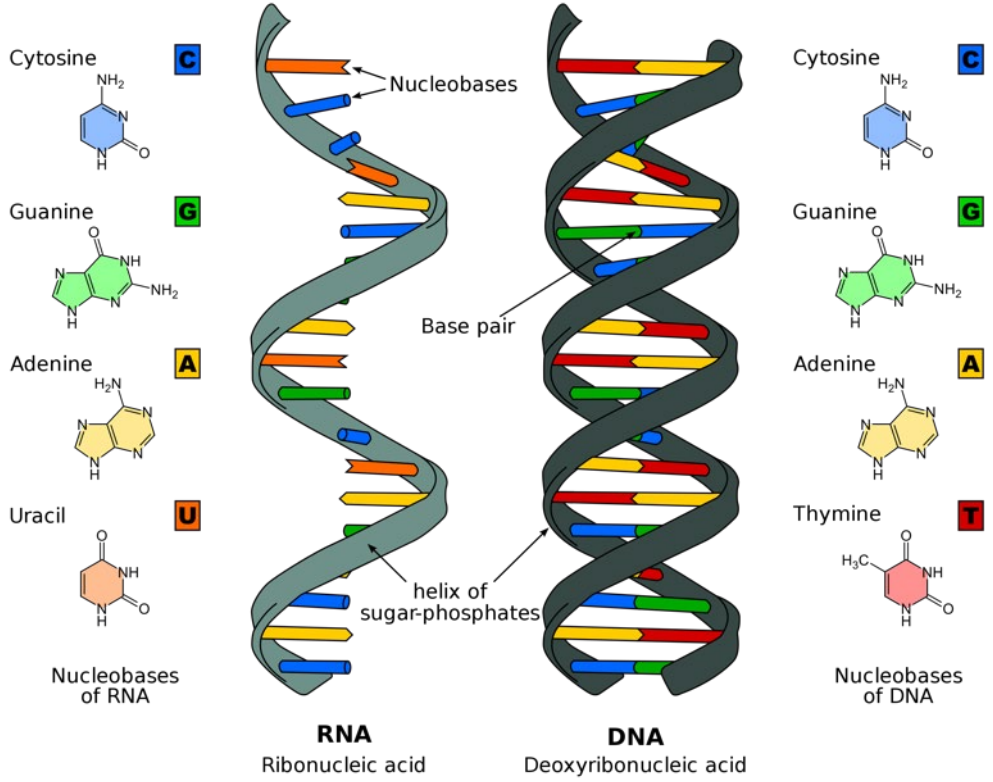
Put simply, genomics is the branch of biology that is concerned with studying genomes

But what is a genome?



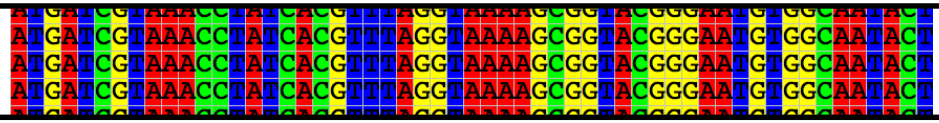
A genome is effectively the blueprint containing the instructions to build and run an organism

Idea is simple

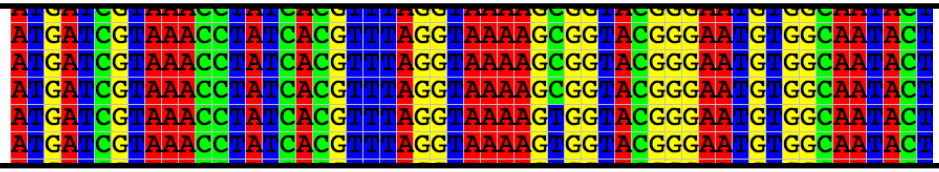


1. The blueprint is 'written' in nucleic acids – DNA and RNA and is unambiguous

2. We can use that 'text' to examine organisms of interest



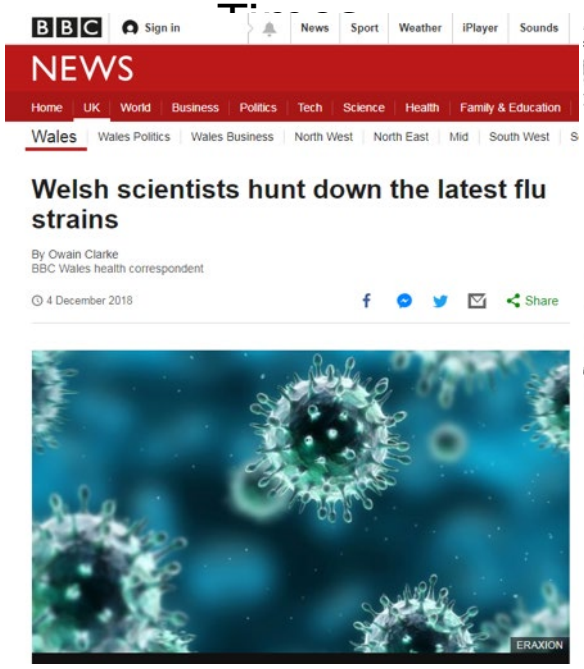
3. Changes in the blueprint can result in changes in the organism

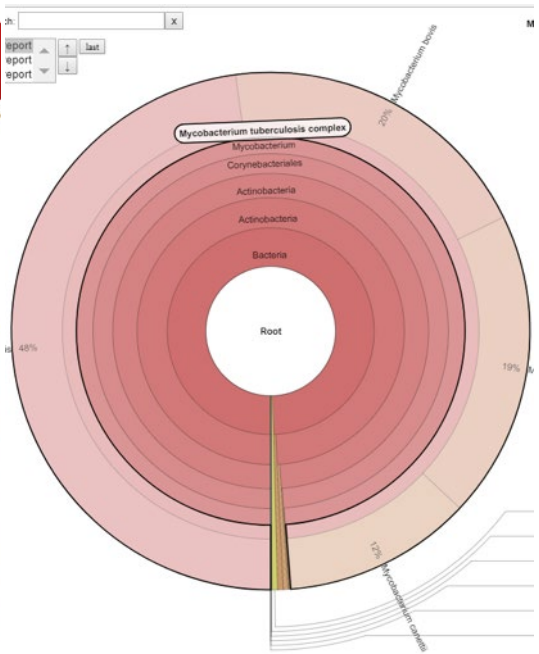


4. This means that genomics could replace a whole slew of tests and underpin new services

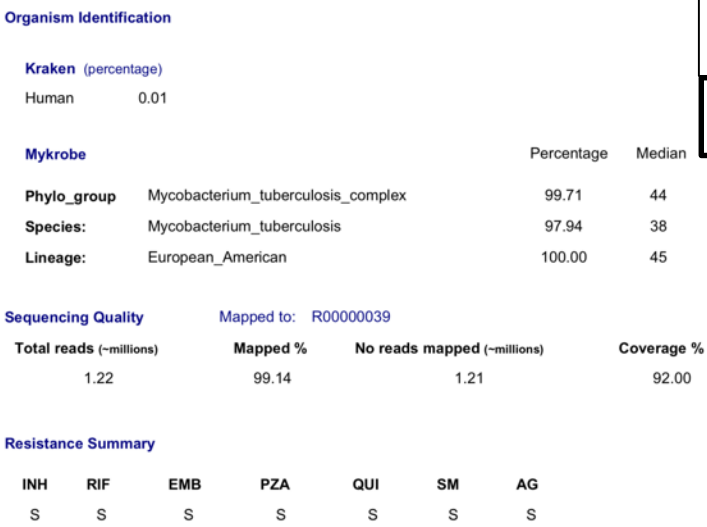
Pathogen genomics has massive potential

High throughput = Faster turnaround Times





Unambiguous = clearer diagnostics



Precision medicine

Whole Genome = More tests possible simultaneously

Common data types = Diagnostic data can be used for (population-level) surveillance in real time

Precision healthcare

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Vast potential, but genomics greatest strength is also its greatest challenge

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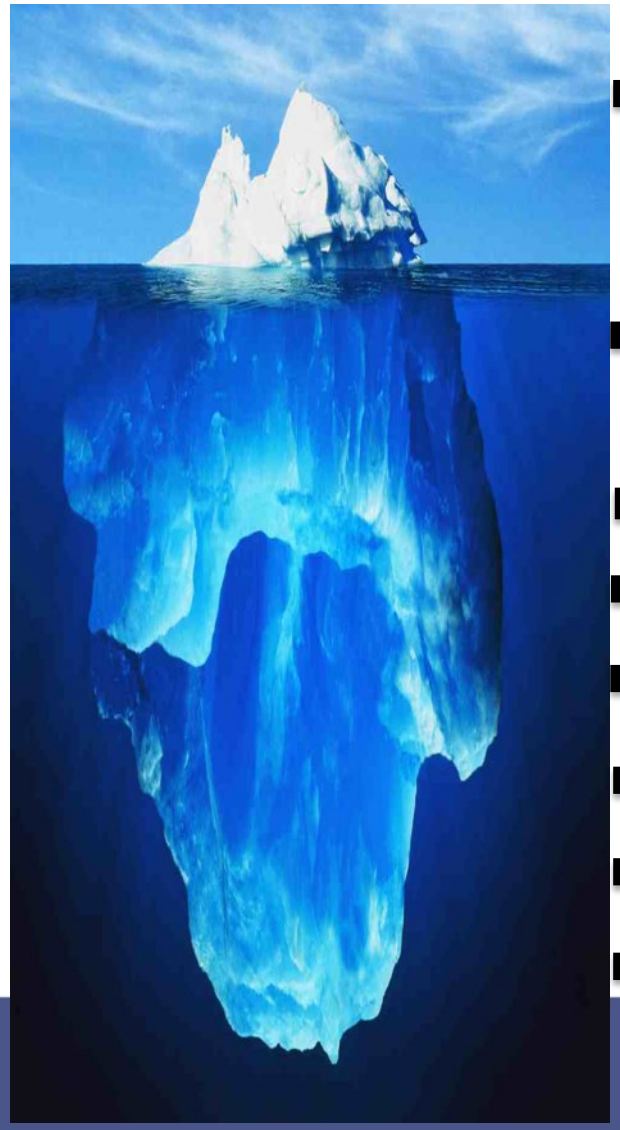
The sequencing iceberg: data and translational challenges of moving genomics from research to service



Sequencing is relatively cheap, and sequencing data is the 'same'

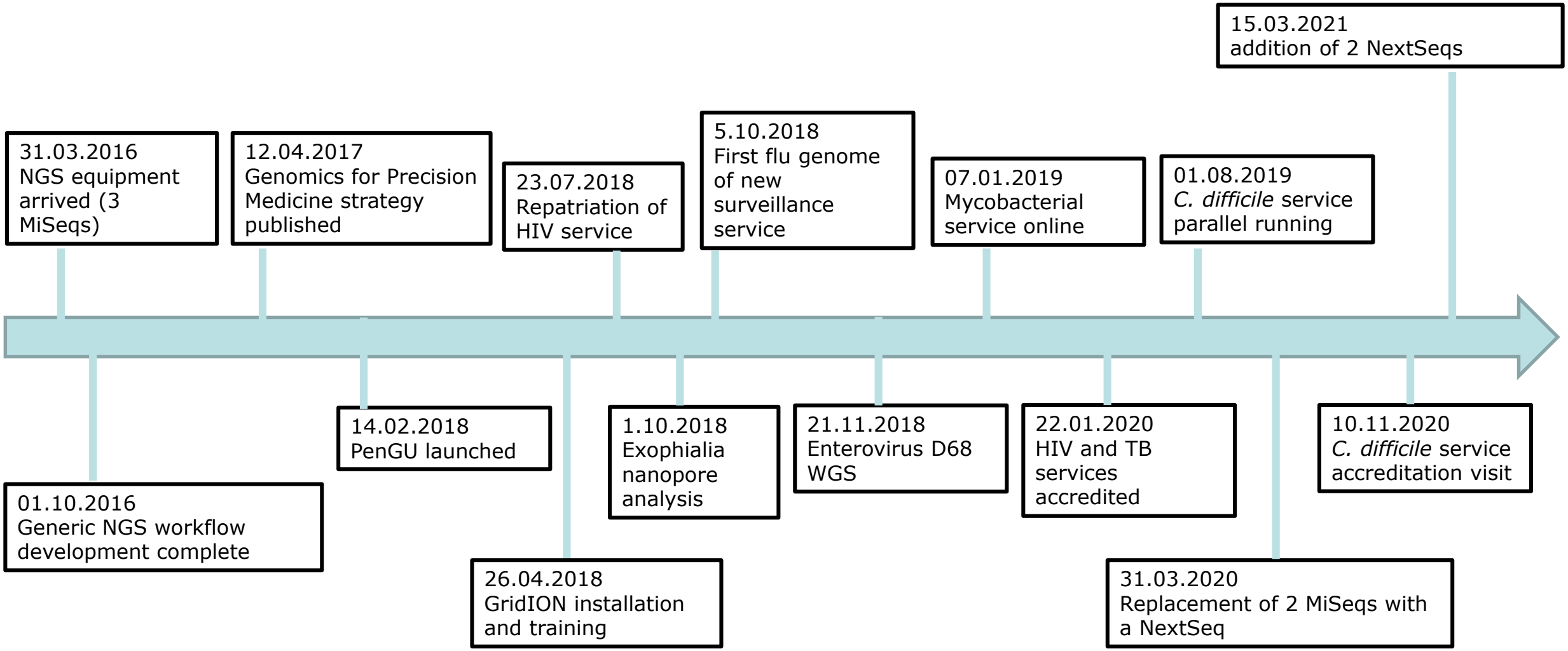
However, the key challenges and costs are hidden

- Bioinformatics expertise
- Network capacity
- User accessibility of software/hardware
- Appropriate compute capacity
- Software development
- Storage availability

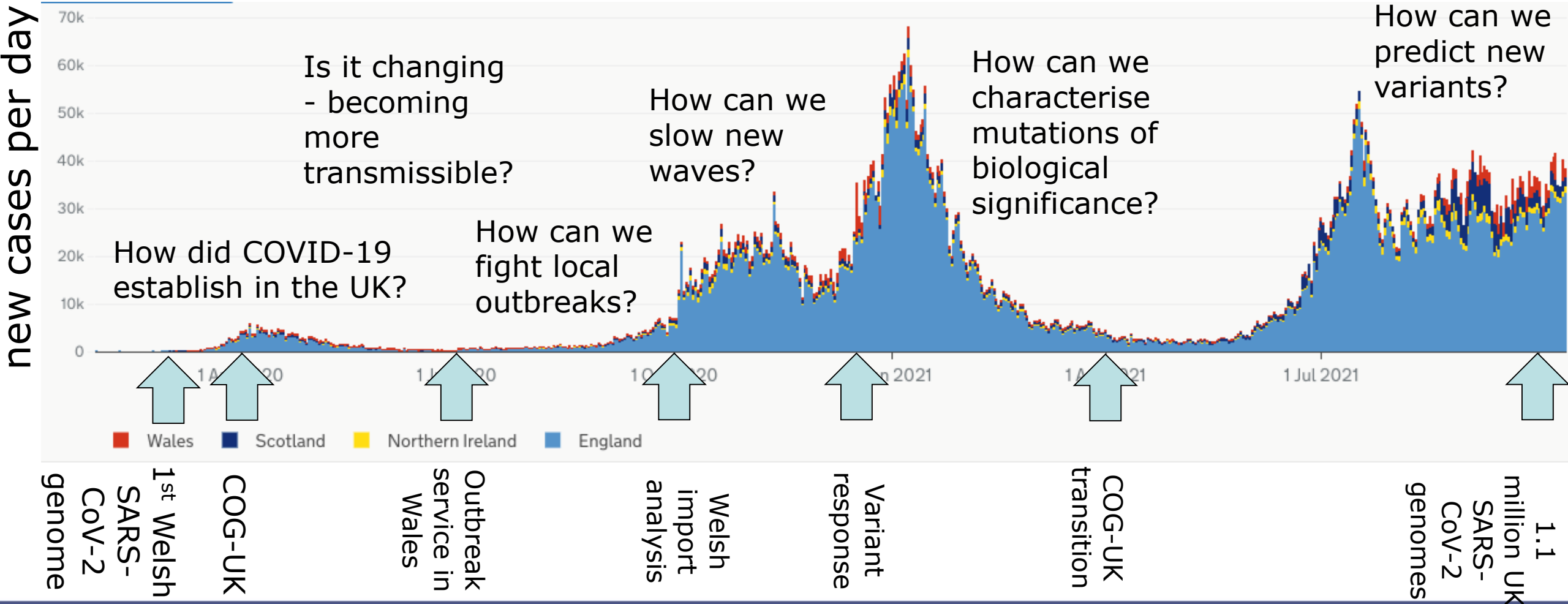
Requires research, infrastructure, expertise

In research, these can account for up to 90% of the costs of doing genomics work

Sequencing iceberg breaking in Wales



Vision of the future: Evolution of precision healthcare services from research – SARS-CoV-2



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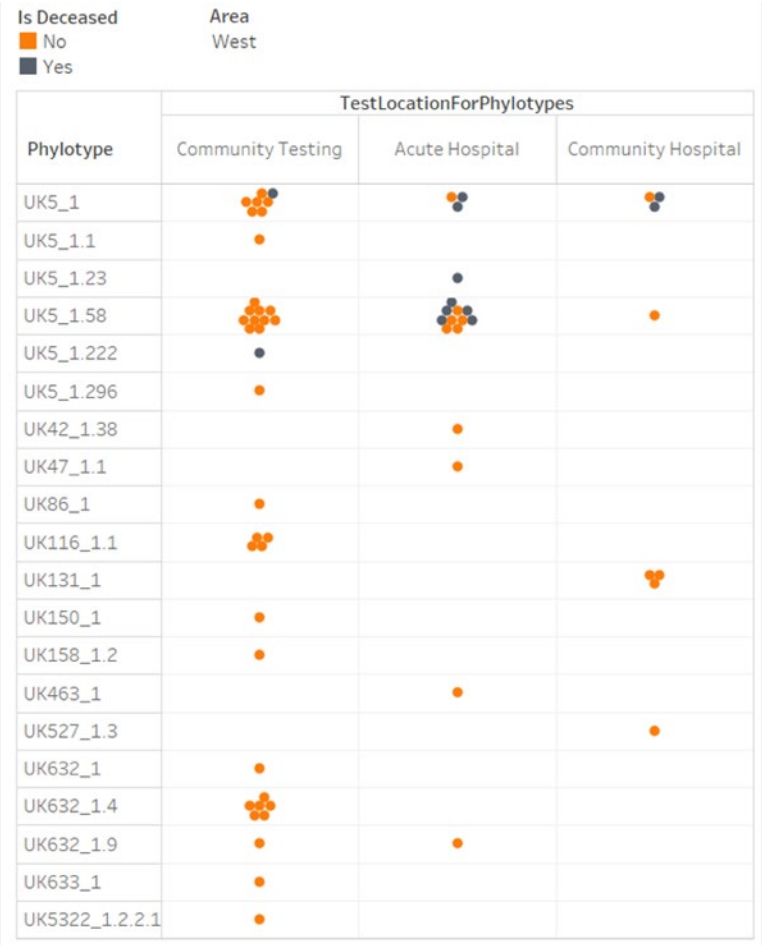
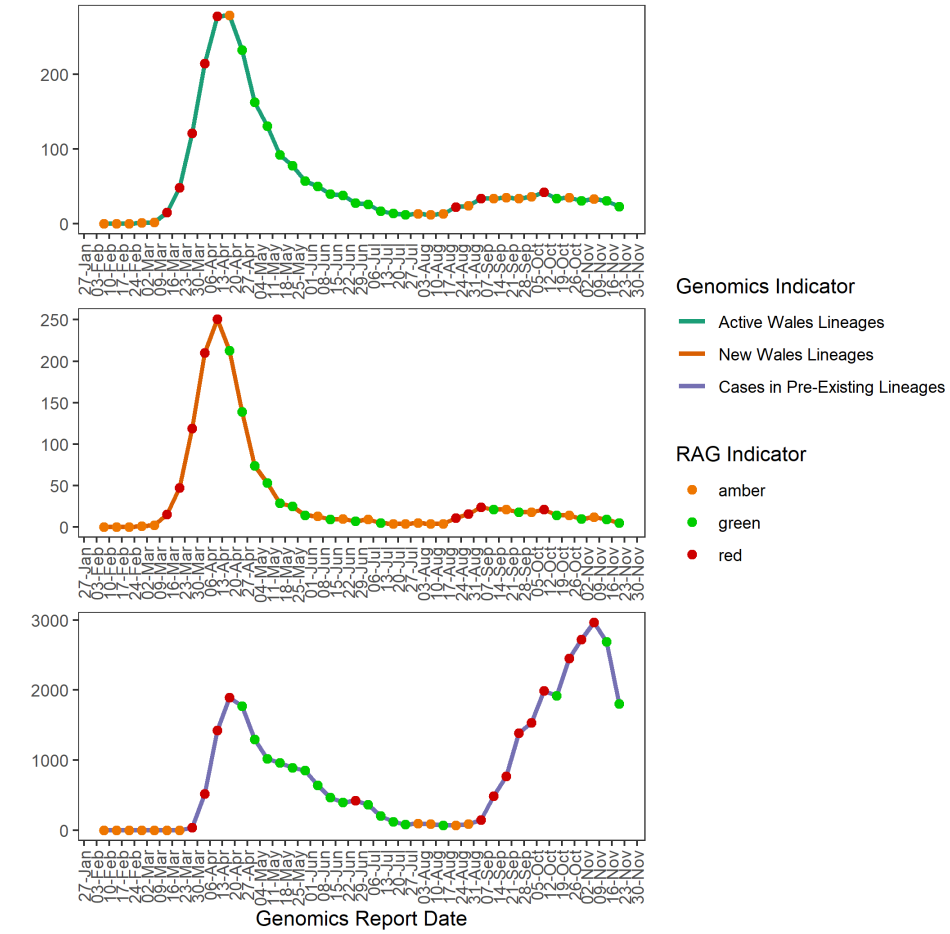
Pathogen genomics research translation enables precision public health





Local Level

National Level

Supporting Government

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Pathogen genomics in Wales

- PenGU has translated pathogen genomics research into service
- Key is co-creation and collaborative working
- Current development areas
  - **AMR bacterial surveillance and characterisation**
  - **Cystic Fibrosis polymicrobial infection diagnostics**
  - **Enterovirus surveillance**
- Production systems
  - ***C. difficile* surveillance and outbreak support**
  - ***Mycobacteria* identification and characterisation**
  - Influenza surveillance
  - **HIV susceptibility testing**
  - COVID-19 outbreak support and surveillance

Accredited Services
System design/needs assessment in progress
Pilot system development

COG UK COVID-19 GENOMICS UK CONSORTIUM
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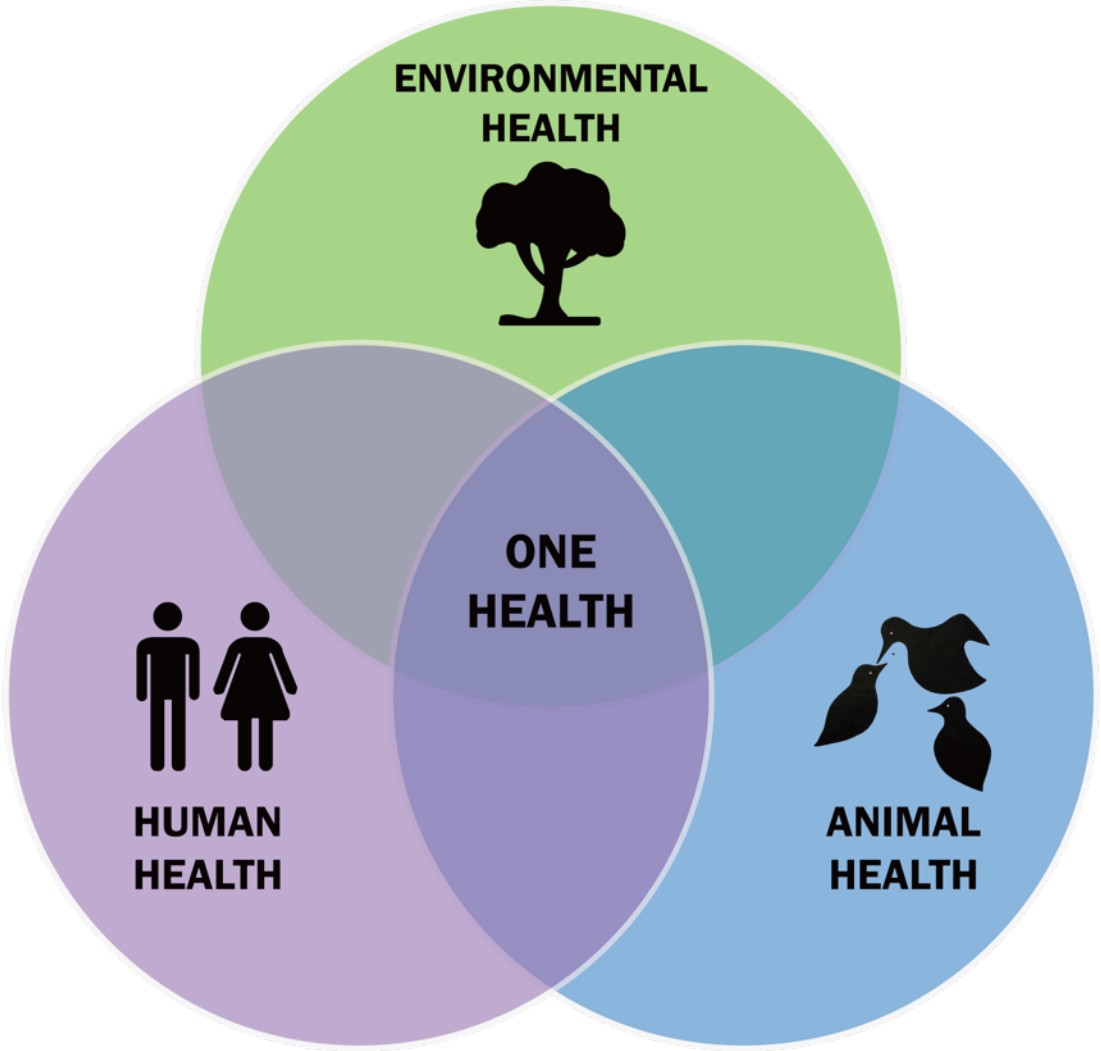
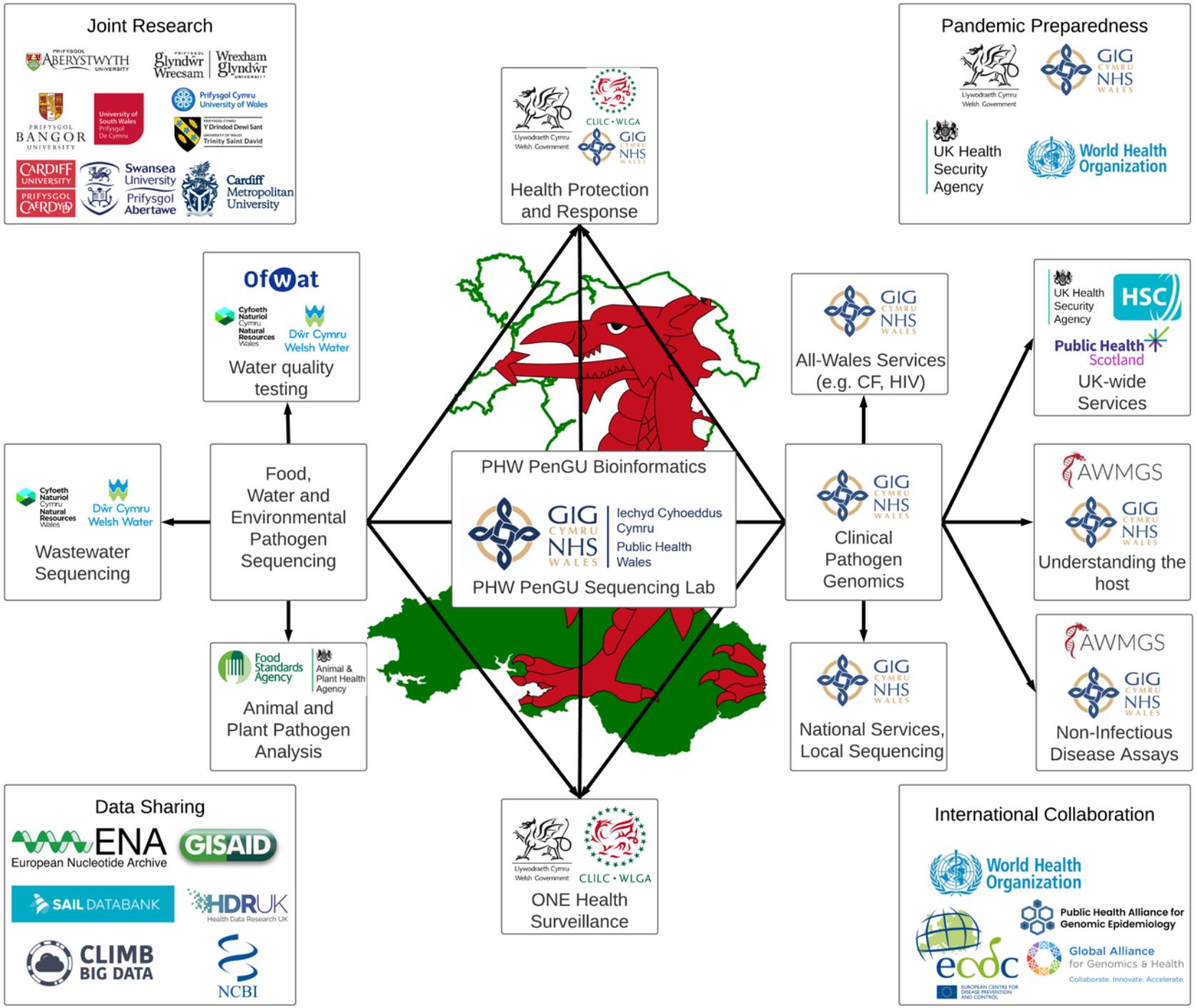
Going further – ONE Health

- Next step is to move beyond single services to new levels of data integration and data use
- This will require integrating research from other areas
- End goal is enabling a holistic view of healthcare that integrates pathogen, human, animal and the environment





Genomics is the lynchpin of future service development, which unlocks the possibility of ONE Health and precision healthcare/public health

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Conclusions

Genomics is a foundational technology which allows precision healthcare

Genomics in service has grown from genomics research, and the translational pathway for this remains critical

SARS-CoV-2 has demonstrated how the potential of genomics can be realised

Challenge now is to imagine that new future, and what research and service development work is required

Genomics isn't just another typing tool, it is a field which will give birth to new services and enable a totally new approach to healthcare, integrating patients and populations