Demystifying genomics, how do we get started

Anders Rhod Larsen

AGENDA

- 14:00 – 14:15: Introduction to genomics and the story on how Statens Serum Institute (Denmark) started whole-genome sequencing implementation / by Anders Rhod Larsen, SSI (10 min + 5 discussion)
- 14:15 – 14:30: Presentation of ResFinder and MLST Finder tools, how to use the online tools, and where to find the databases for integration into own workflows / by Jette Sejer Kjeldgaard, DTU (10 min + 5 min discussion)
- 14:30 – 14:40: Coffee break (10 min)
- 14:40 – 14:55: National example from Latvia on the implementation of WGS into own workflows / by Reinis Vangravs (10 min + 5 min discussion)
- 14:55 – 15:10: National example from Ireland on the implementation of WGS into own workflows / by Martin Cormican (10 min + 5 min discussion)
- 15:10 – 15:15: Thank you and goodbye / by Birgitte Helwigh, DTU (5 min)

Strategy for implementation of WGS in EU/EEA

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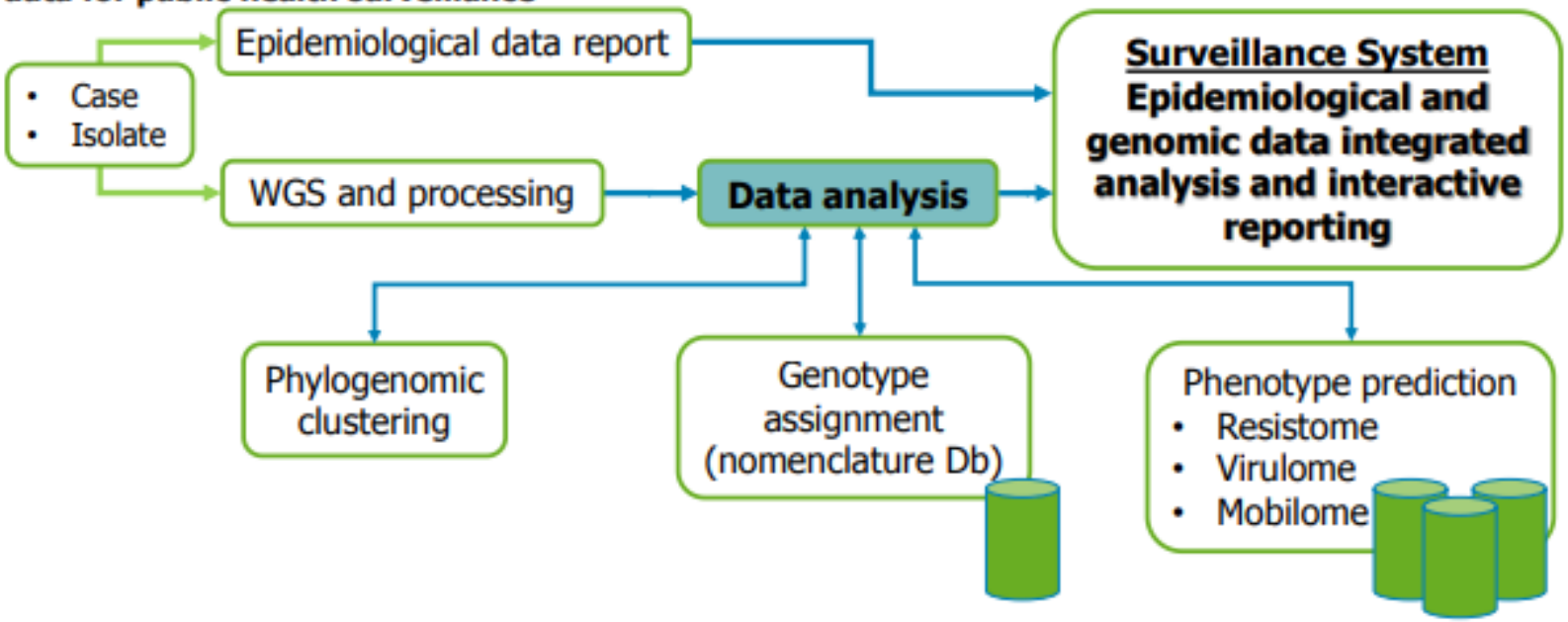
- In 2015, ECDC developed an expert opinion on WGS in consultation with multidisciplinary experts. The resulting strategy envisaged that within five years ECDC would have contributed to the establishment of standards and systems enabling the EU-wide use of WGS as the method of choice for typing of microbial pathogens, replacing other methods.

The main steps involved in the process of WGS data production, bioinformatics processing and analysis, and integration with linked epidemiological data for generic surveillance purposes, are summarised in Figure 2.

Figure 2. Process overview – WGS data production, analysis and integration with epidemiological data for public health surveillance



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NGS implementation at SSI

2013 | 2014 | 2015 | 2016

Listeria
Salmonella
ESBL + CPO
VRE
Haemophilus
VTEC
Campylobacter
Meningococci
Legionella
C. difficile
MRSA
Gr A Streptococci
Gonococci
Virus
TB

Planning | Sequencing and pipeline planning | Evaluation | Routine | ▼ : Real Time

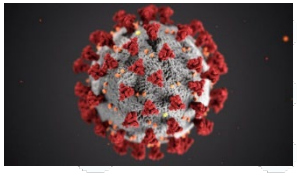
2022

1x NanoSeq

3x NextSeq

3x MySeq

7x I7/Hamilton



5,000 -> 15,000 -> 4,000/ week

CPE in Denmark

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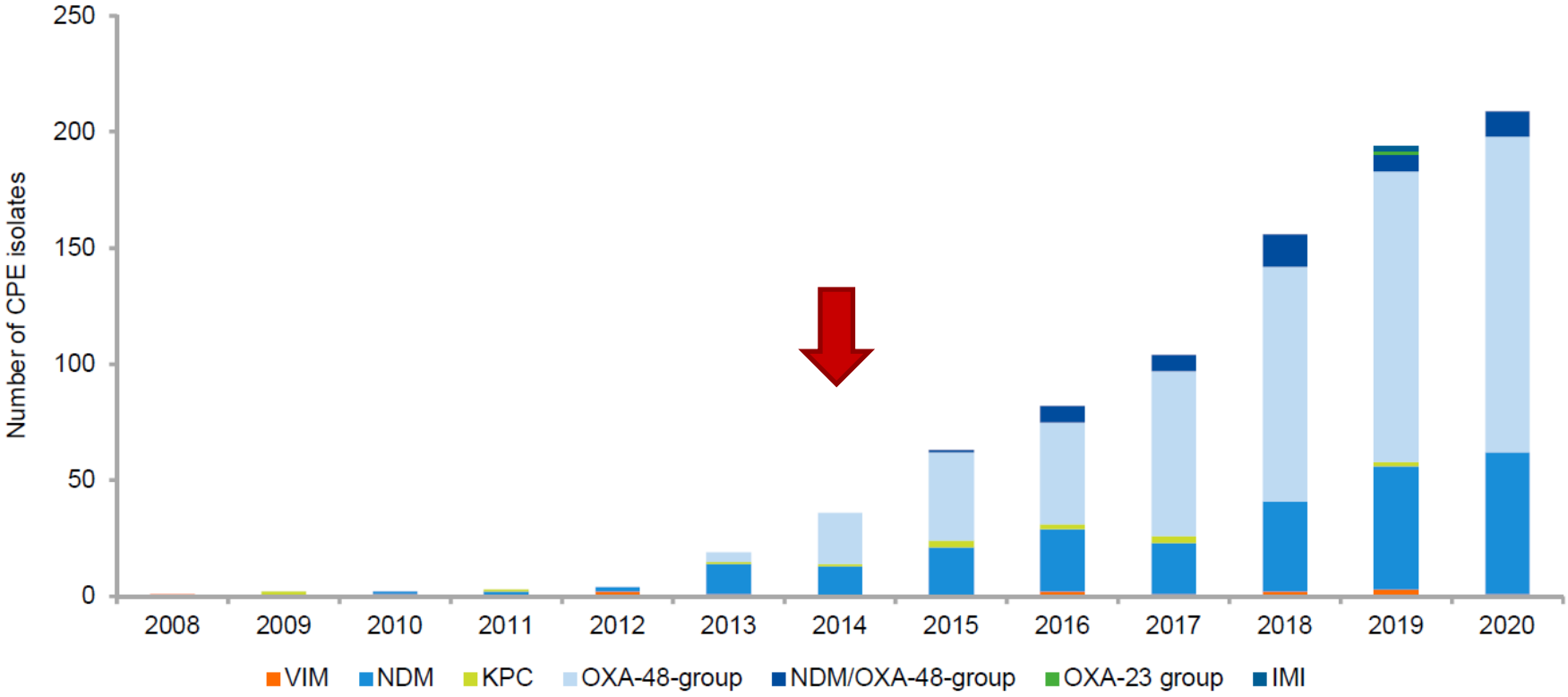
Figure 8.13 Numbers of carbapenemase-producing Enterobacterales (CPE), Denmark, 2008-2020

DANMAP 2020



Cost is a major barrier

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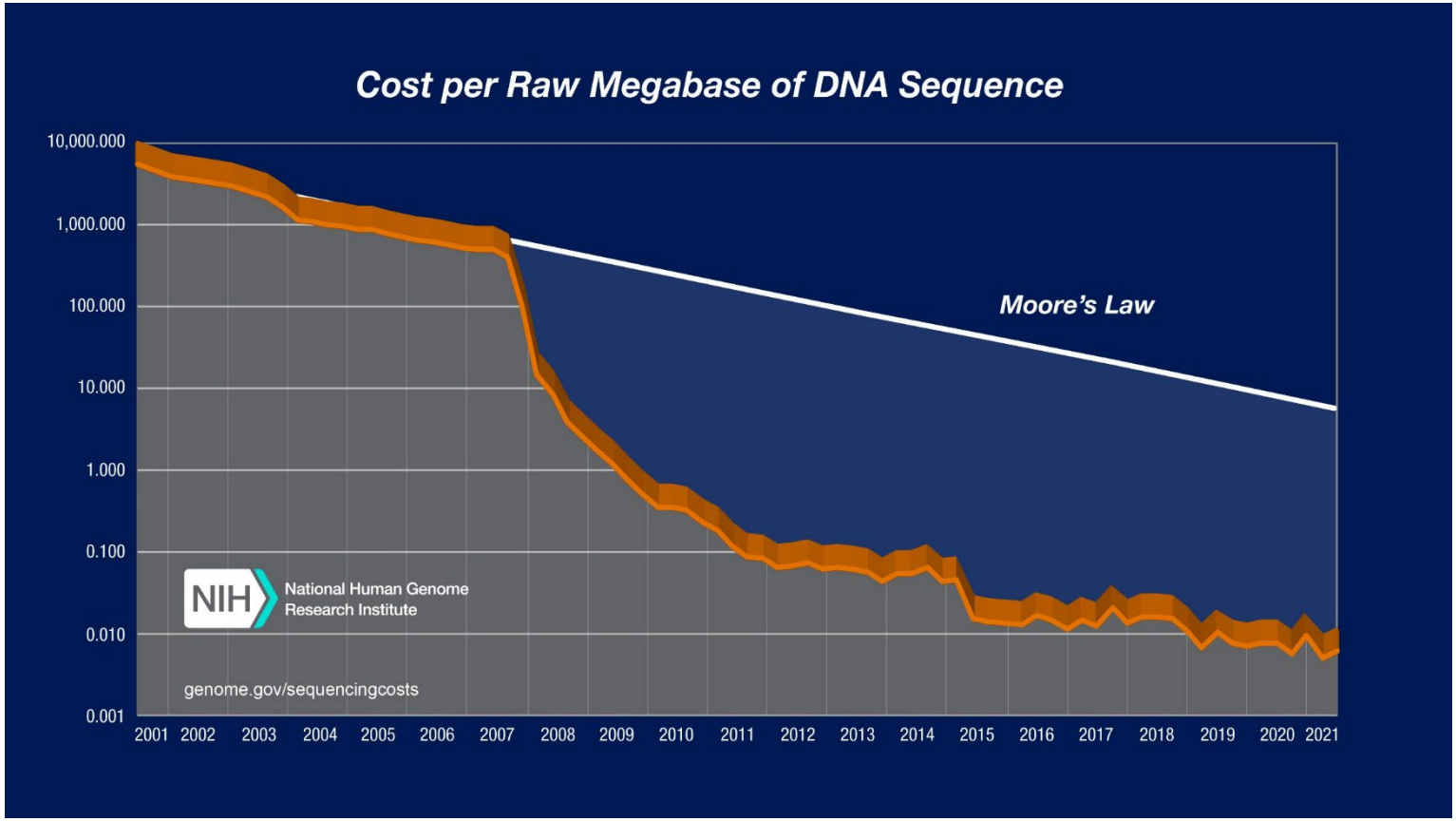
- Hardware, software, reagents
- Bioinformaticians
- Storage



How did we get started in Denmark?

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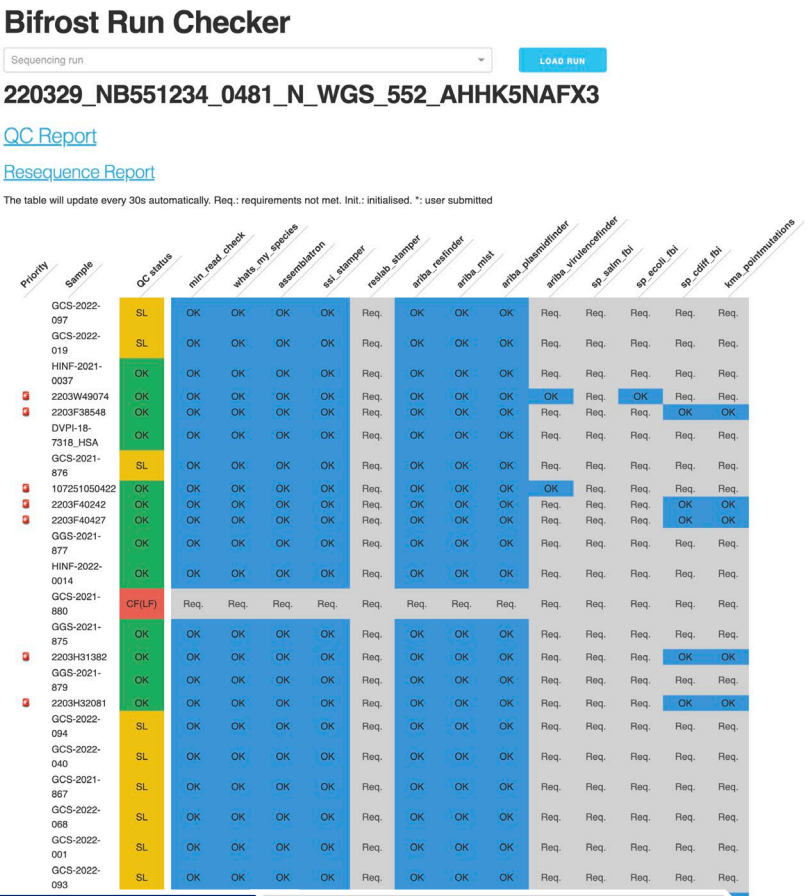
- Sequencing done by research collaborators
  - Building of business case
- Ressources allocated from existing budgets at SSI- shared efforts
  - Replacement of other methods (AST, typing)
- Surveillance initially introduced for less numerous pathogens
- Sentinel surveillance
- Building of own pipeline, including a QC pipeline (Bifrost) and downstream species specific modules for further analysis, e.g. CGE-Finders

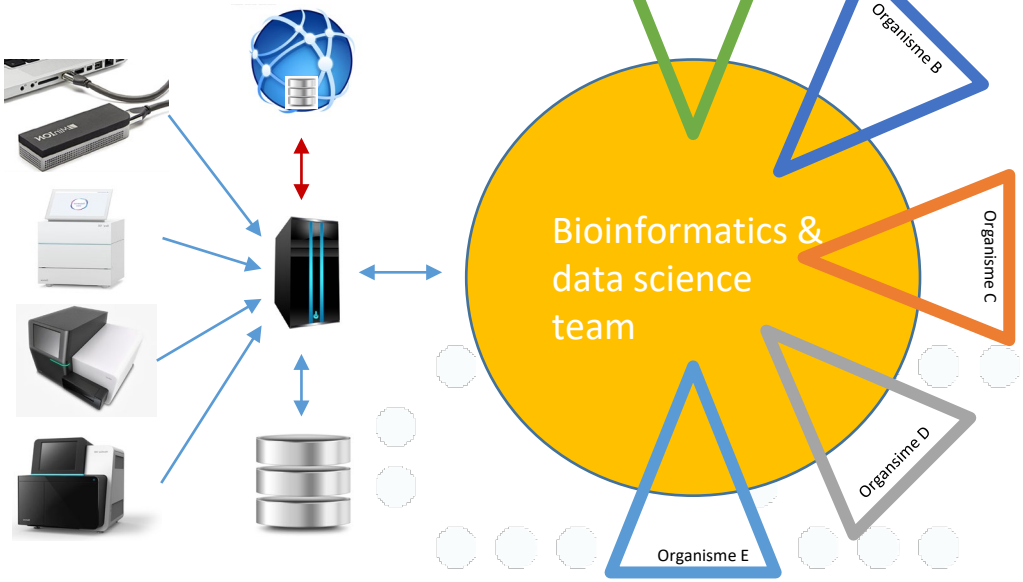
bifrost

- Minimum read check
- Species identification /contaminant check
  - MiniKraken database
- Assemblies
  - skesa
- MLST
- Resistance identification
- Virulence identification (species specific)
- Plasmid identification (species specific)



https://github.com/ssi-dk/bifrost_ssi_stamper

Optimized WGS og bioinformatic flow



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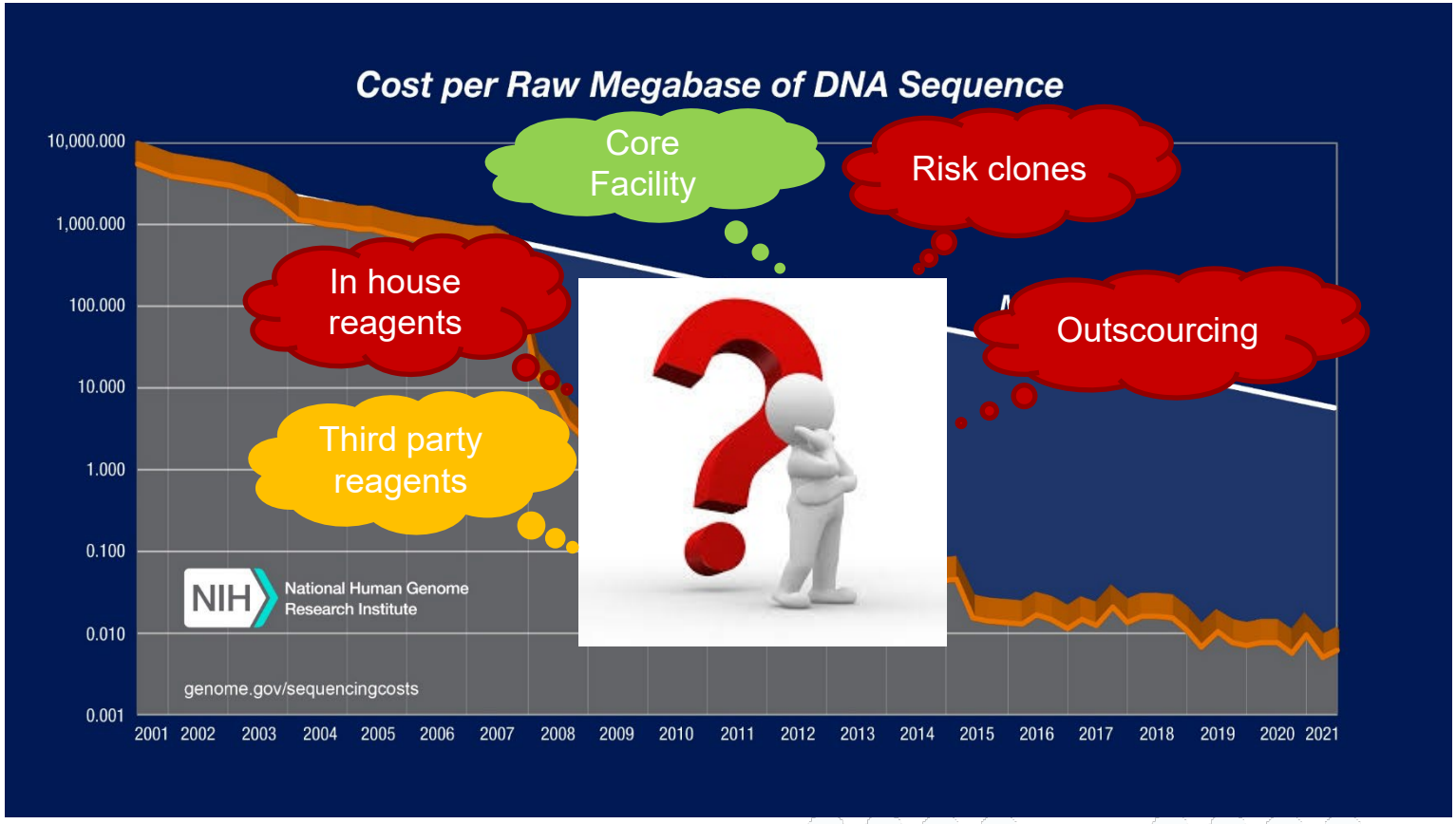
Cost efficiency – where do we go?



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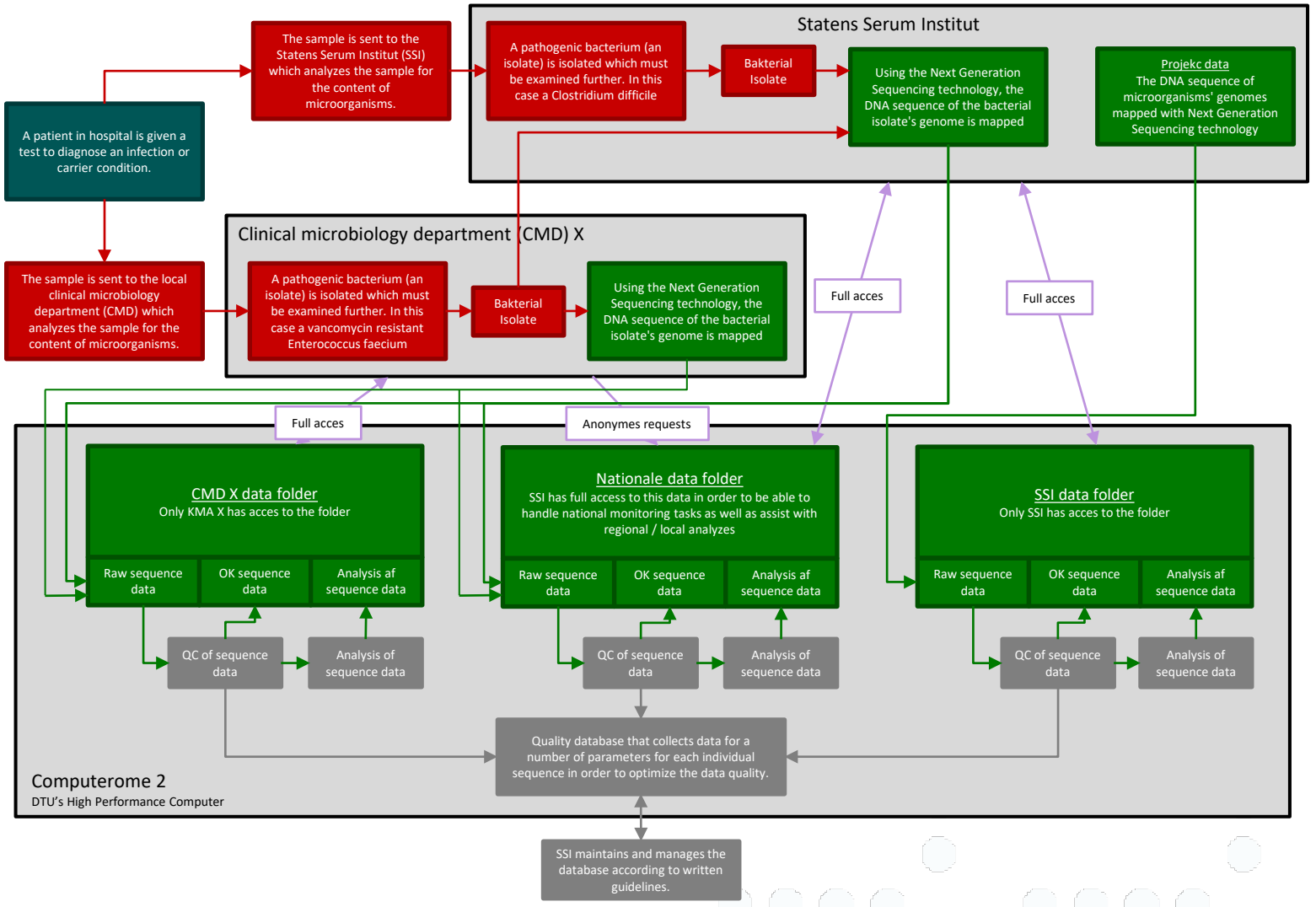

Initiaves to support WGS implementation in EU/EEA

- Covid-19 has boosted the implementation of WGS

- EU4health
  - Incubator- national grants (3 mill + 1 mill €)
  - EURGen-RefLabCap, FWD AMR-RefLabCap
  - More ECDC Tenders have been launched
- EpiPulse established

EpiPulse - the European surveillance portal for infectious diseases

Tool

22 Jun 2021

EpiPulse is an online portal for European public health authorities and global partners to collect, analyse, share, and discuss infectious disease data for threat detection, monitoring, risk assessment and outbreak response.