

The 100,000 Genomes Project

Transforming diagnosis through
genomic technologies

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The future, this year.

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Genomic Medicine

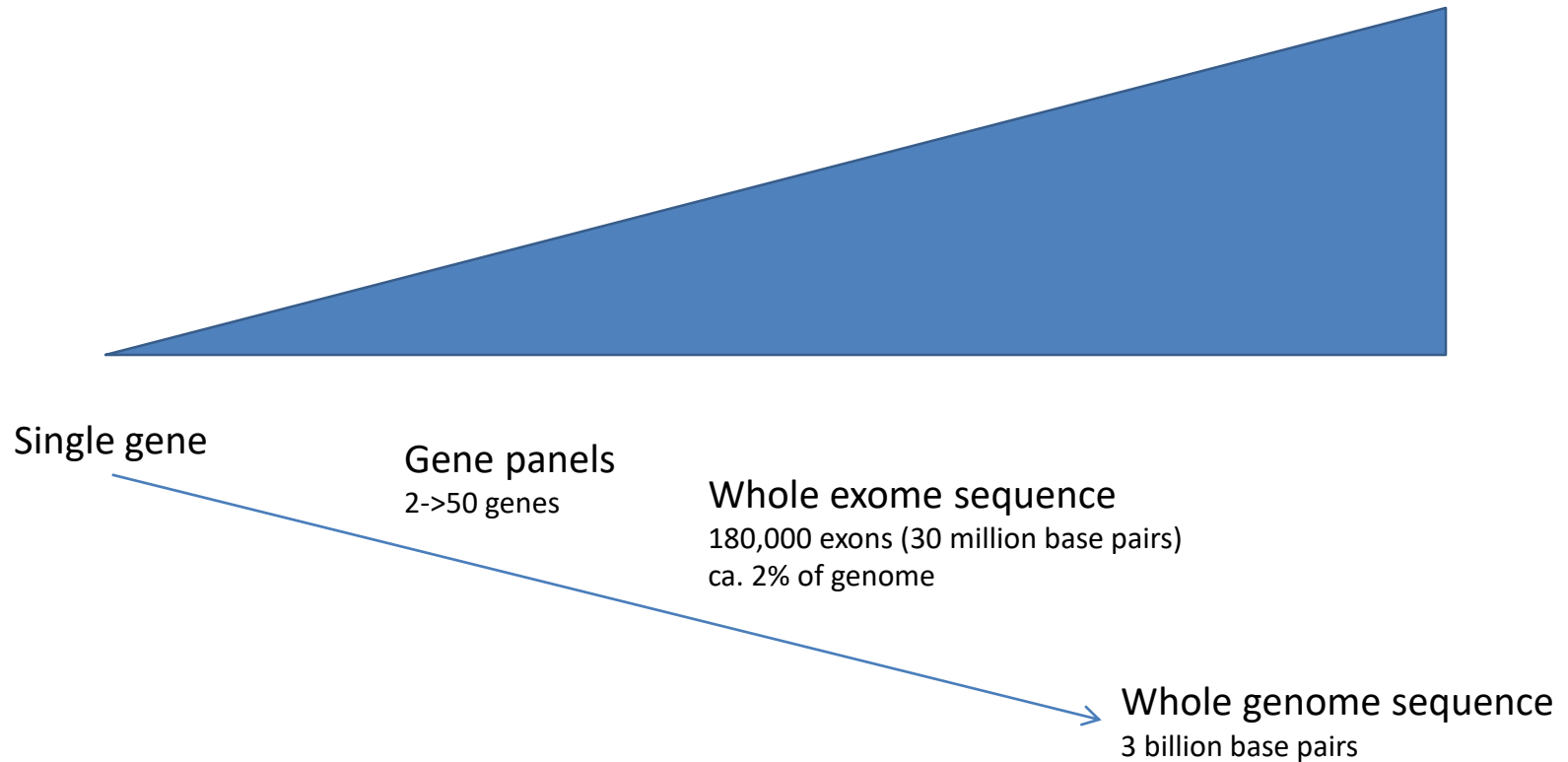
- The use of DNA technology in health care.
- ✓ Detection of genetic alterations in a whole patient (germline).
- ✓ Detection of specific genetic alterations in solid and haematological tumours (somatic).
- Diagnostics
- Prognostics
- Targeted therapy
- 'Pharmacogenomics'
- Personalised medicine

Genomic Medicine evolution

Microarray

FISH

Karyotype

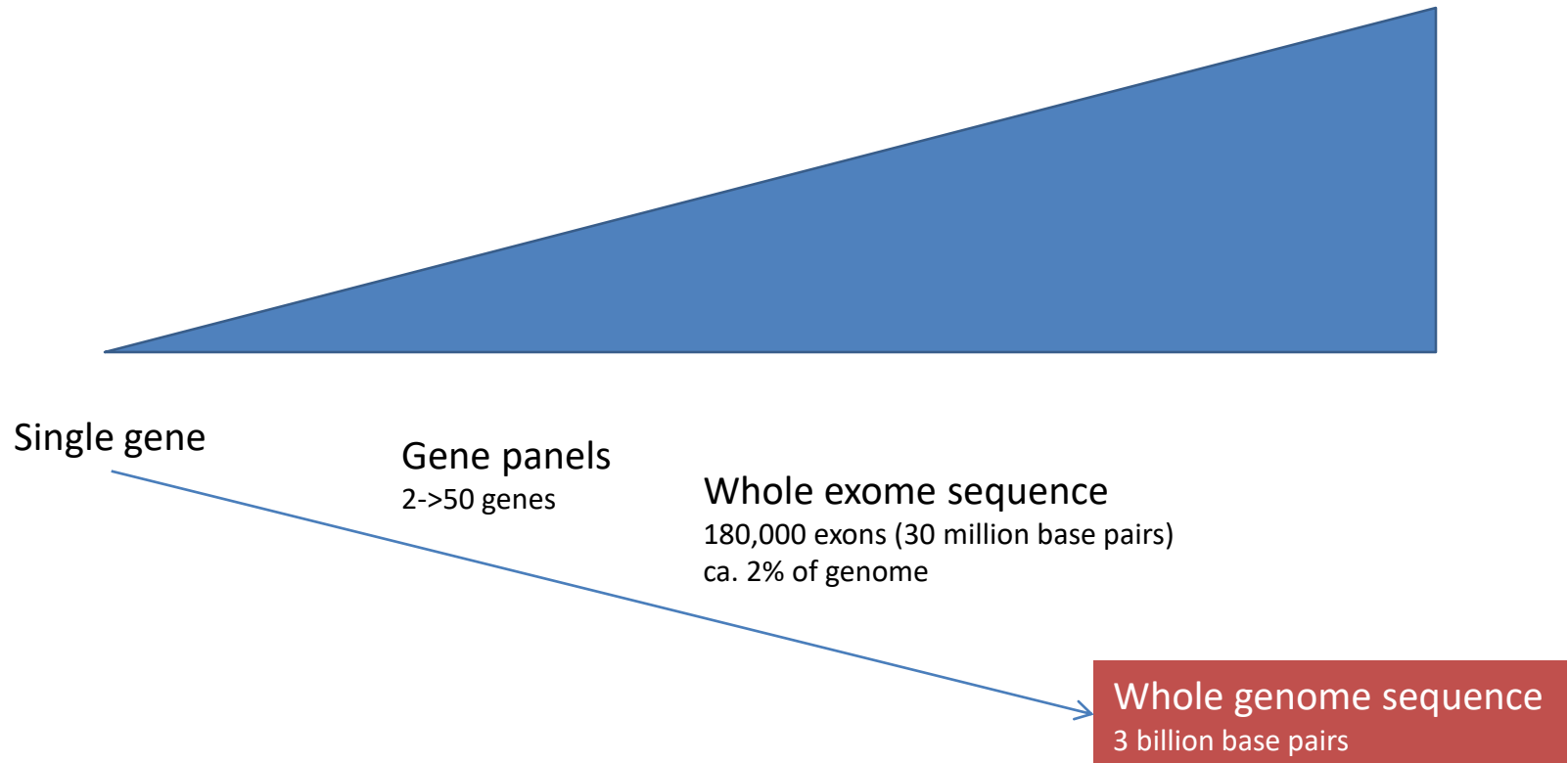


Genomic Medicine evolution

Microarray

FISH

Karyotype



Genomic Medicine

- Most germline testing has been coordinated by tertiary genetics services and restricted to familial disease.
- Development of somatic testing in tumours has been patchy, slow and inequitable.
- The regional budget for germline genetic testing has been a frozen block for several years.
- There is no formal regional budget for somatic testing.
- THIS CHANGES IN 2018.

100,000 Genomes Project

- David Cameron
- National-level biotechnology investment
- Improved diagnostics etc
- Feeds into research
- Feeds into commercial development

- NHS in England is the only health care system in the world doing this at such scale

100,000 Genomes Project

- Focussed only on whole genome sequencing
 - Rare disease sub-project exploring increased diagnostic yield through WGS.
 - Cancer sub-project exploring WGS in tumours (solid and haem)
 - Newcastle University was a Pilot site 2013-15 (ca. 10,000 genomes nationally) – rare disease
 - NHS England commissioned ‘NHS Genomic Medicine Centres’ to deliver the rest
 - Newcastle Hospitals 2015 – 2018 – rare disease + cancer
- >50,000 NHS whole genome sequences now completed

100,000 Genomes Project

Subjects can consent to:

1. NHS diagnostic analysis of genes relevant to their disease
2. NHS diagnostic analysis of genes that cause other serious inherited diseases*
3. NHS carrier testing for recessive inherited diseases (couples)
4. NHS carrier testing for serious X-linked diseases (females of reproductive age)
5. Research analysis of their DNA, 'omics and clinical data
6. (Closed at present) 'Any other finding of potential clinical significance'

* e.g. BRCA1/2, Lynch syndrome, TP53, familial hypercholesterolaemia

100,000 Genomes Project

- Massive transformation
 - Patient consent
 - Phenotypic data collation
 - Local validation & reporting pipelines
 - GeCIPs
 - Initial national focus on FFPE tumour tissue
 - Shift to fresh frozen tissue

100,000 Genomes Project

- You have to stop using formalin in operating theatres*.
- The pathology pipeline has to change**
- Tissue needs a cellularity assessment and biopsy for DNA extraction**

* Thank you, MD, CEO, surgeons, theatre nurses, pathologists, porters...

** Thank you, MRC molecular pathology node, pathologists, technical staff...

100,000 Genomes Project

- Cancer results are being returned within a clinically reasonable timescale (<6 weeks)
- Rare disease results have taken >2 years in many instances.
- There are 6 months of development left.

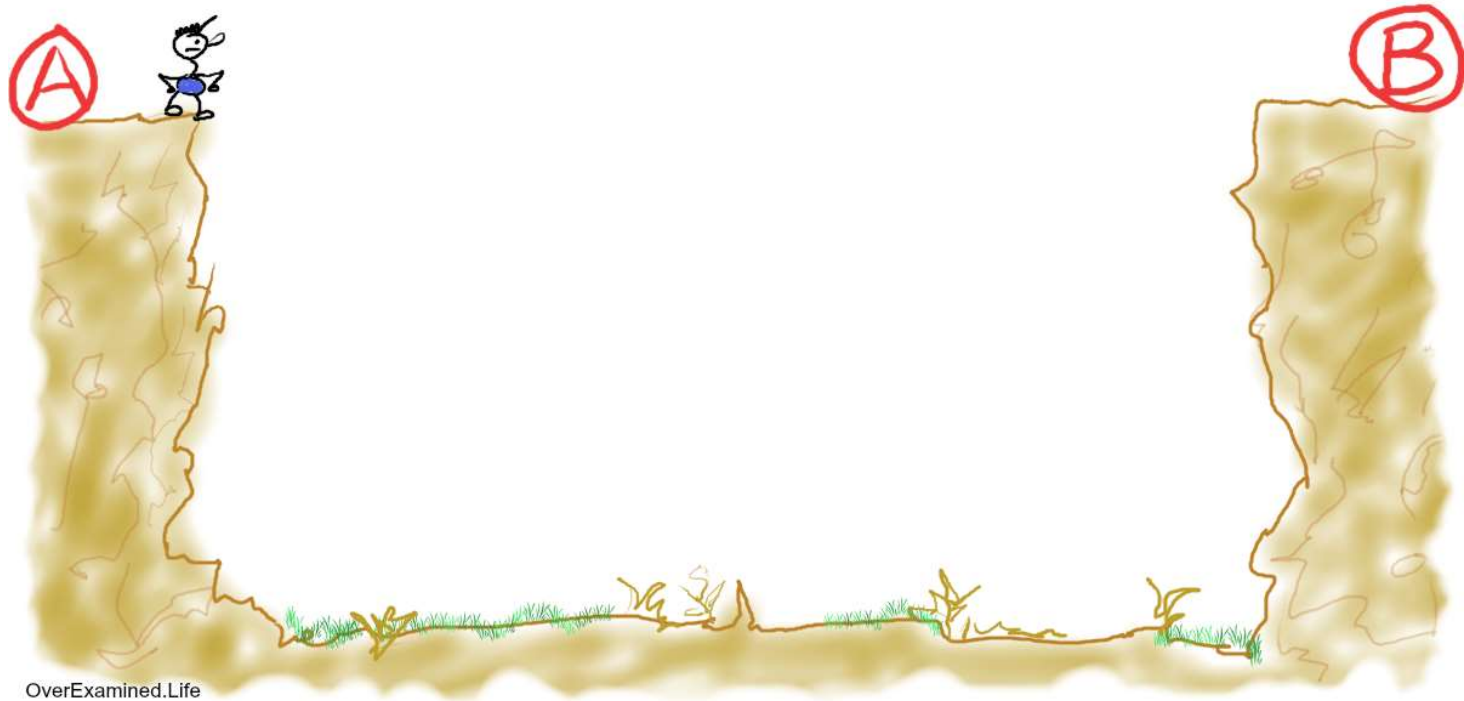
Genomics lab re-procurement

- All genomics labs in England are being re-procured
- There will be 7 Genomics Lab Hubs
- There will be a single national WGS facility (Cambridge)
- Newcastle, Leeds and Sheffield have to submit a single proposal ('GeNEQ'):
 - Central (Leeds) high throughput sequencing lab (mainly germline initially)
 - Labs in Sheffield and Newcastle will mainly process cancer diagnostics
- The tender is due in on 12th April

2018/19: the future happens

Current practice +
100,000 Genome Project

—————> NHS Genomic Medicine Service

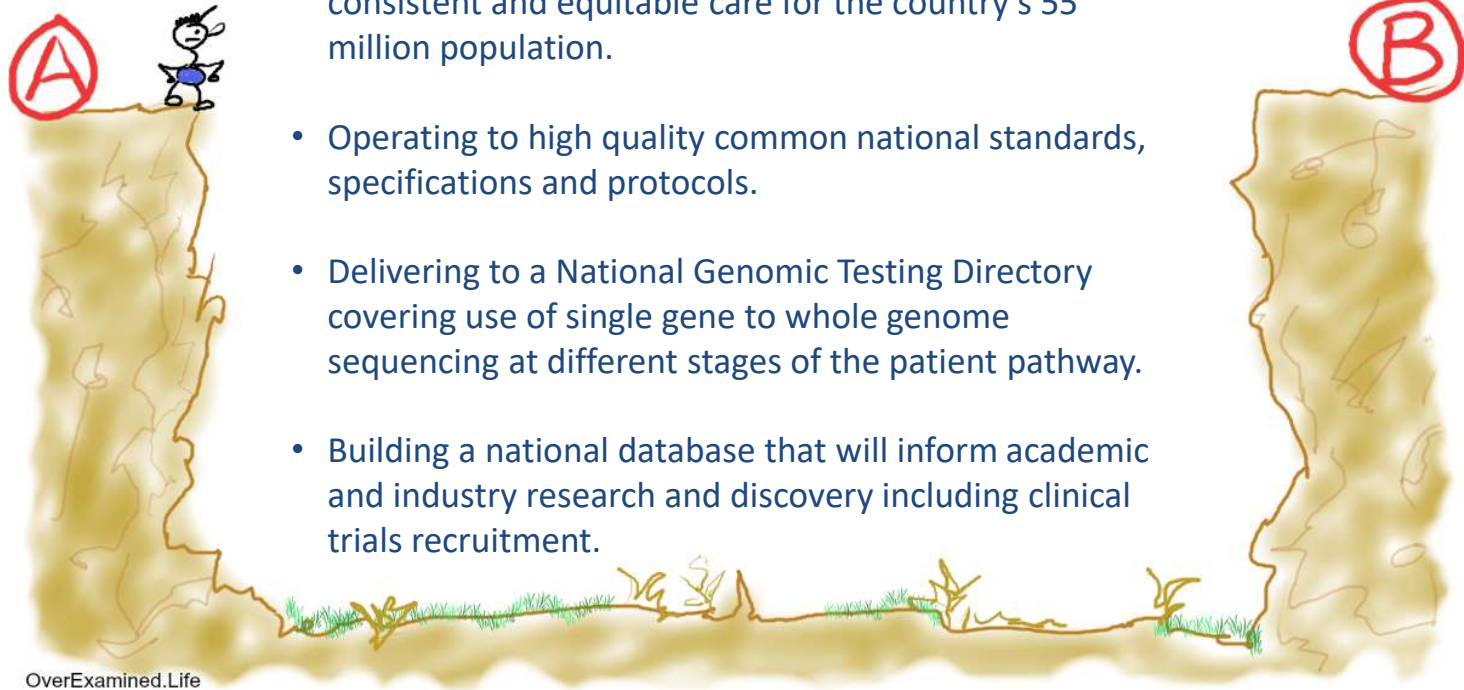


B = 1st October. 2018.

2018/19: the future happens

Current practice +
100,000 Genome Project

→ NHS Genomic Medicine Service



- A national Genomic Medicine Service providing consistent and equitable care for the country's 55 million population.
- Operating to high quality common national standards, specifications and protocols.
- Delivering to a National Genomic Testing Directory covering use of single gene to whole genome sequencing at different stages of the patient pathway.
- Building a national database that will inform academic and industry research and discovery including clinical trials recruitment.

B = 1st October. 2018.

I REALIZE
I'M FEELING PANIC
AND THAT
PANICS ME MORE