The future of genetic diagnostics

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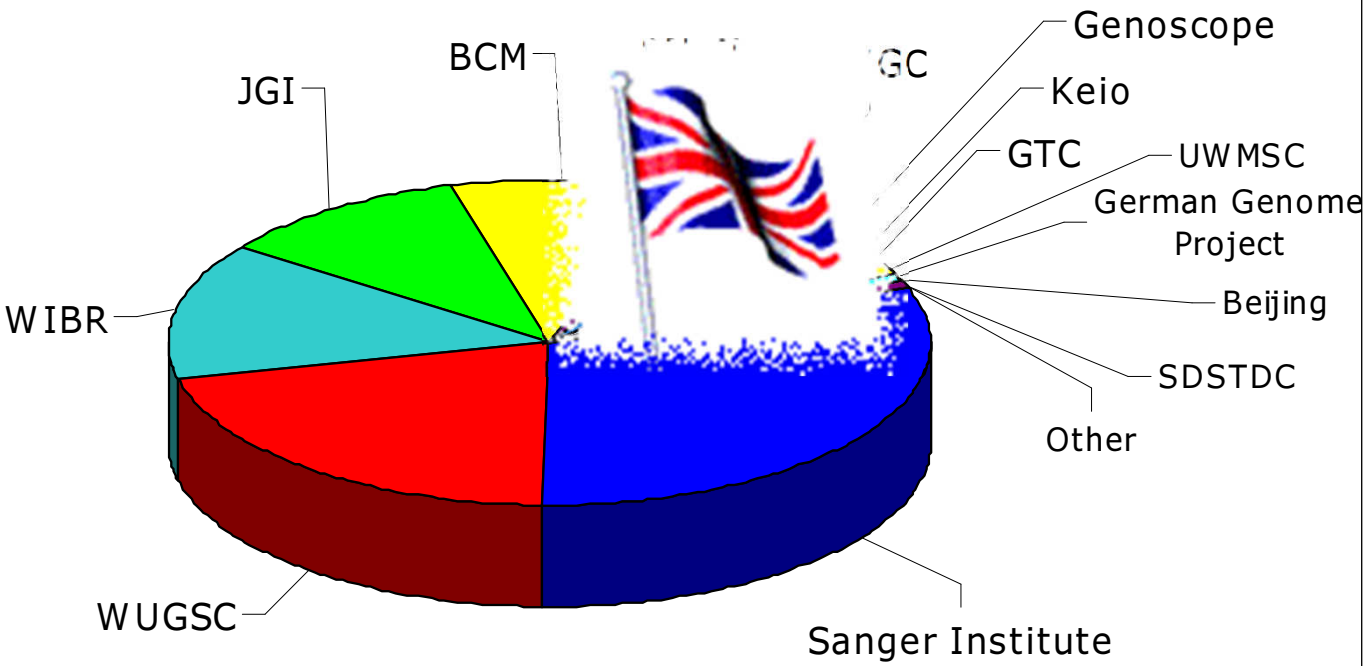
Human Genome Sequence Contributors



Sanger Sequencing

ttagatgagaa

In the A tube, mix A's,C's,T's and G's plus dideoxyA (blank end)

ttagatgagaA

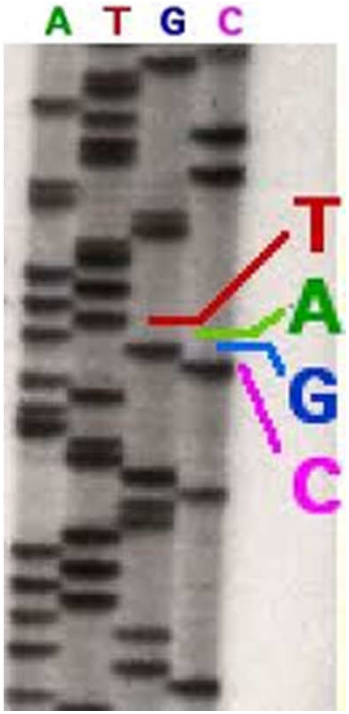
ttagatgagA

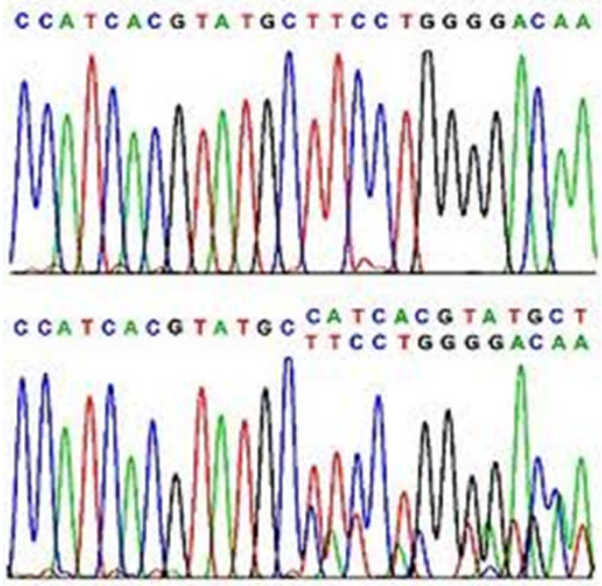
ttagatgA

ttagA

ttA

Sanger Institute





Sequencing 2005

Using ABI 3730 XL*

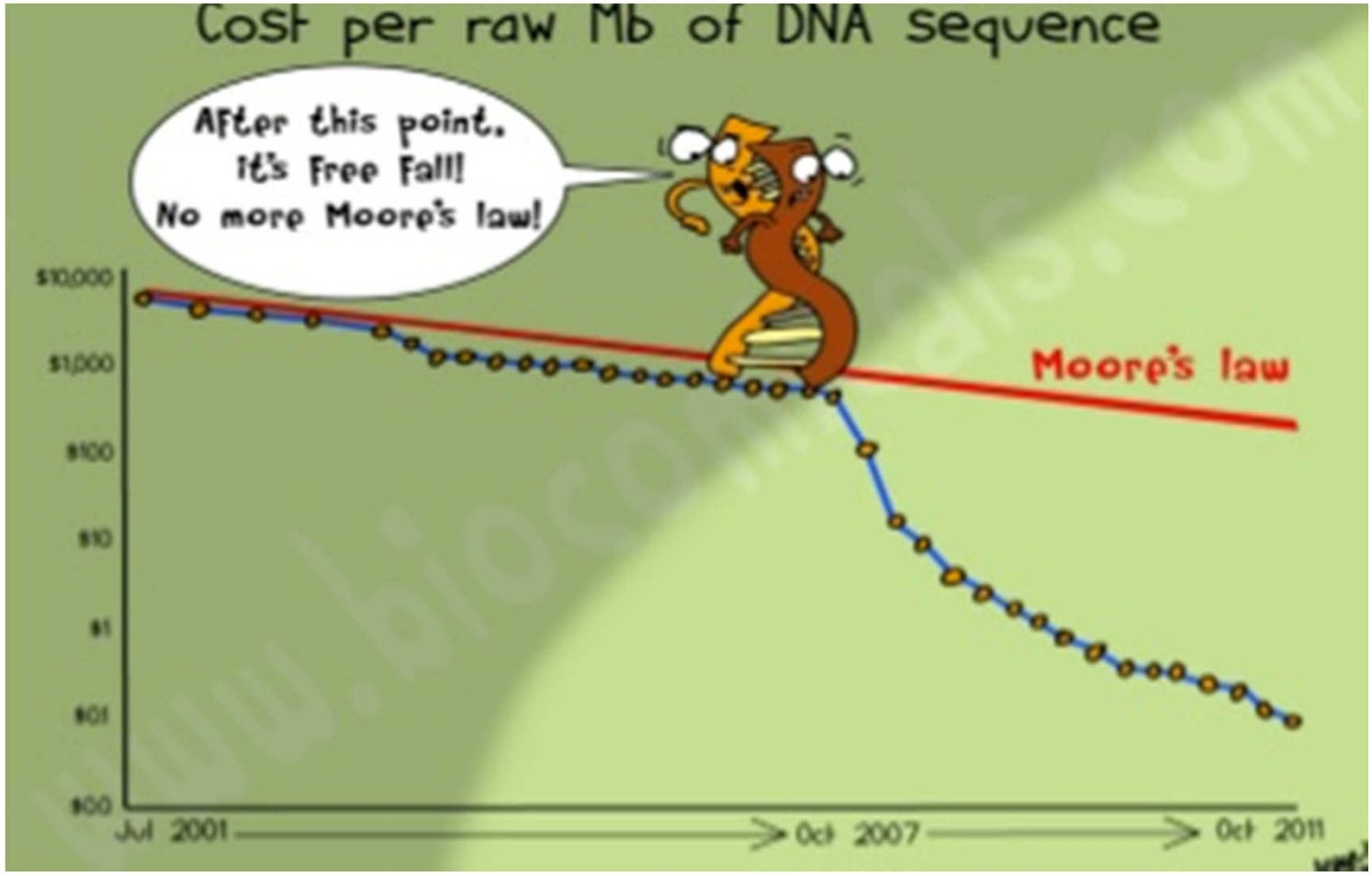
Washington University
Genome Sequencing Center

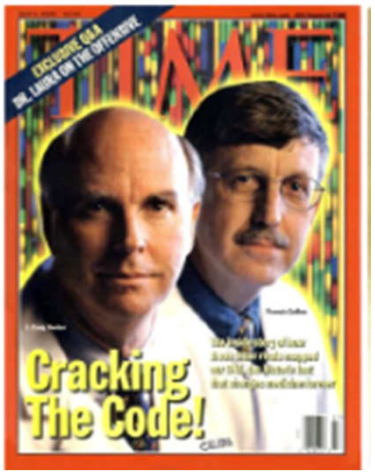
Baylor College of Medicine
Human Genome Sequencing Center

*fluorescent Sanger chemistry
on capillary sequencers

Newcastle University

Penny drops



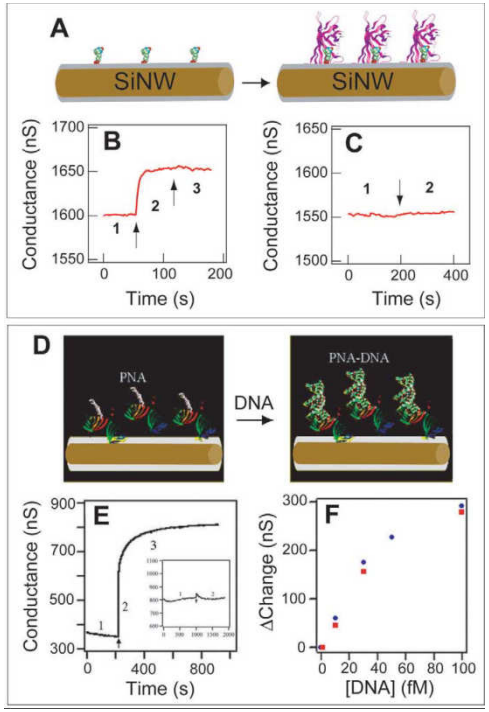


HiSeq & MiSeq (Illumina)

To be used in the 100,000 Genomes Project

Idea for a diagnostic handheld device

Cui, Y., Wei, Q.Q., Park, H.K. & **Lieber, C.M**. Nanowire nanosensors for highly sensitive and selective detection of biological and chemical species. Science 293, 1289–1292 (2001).



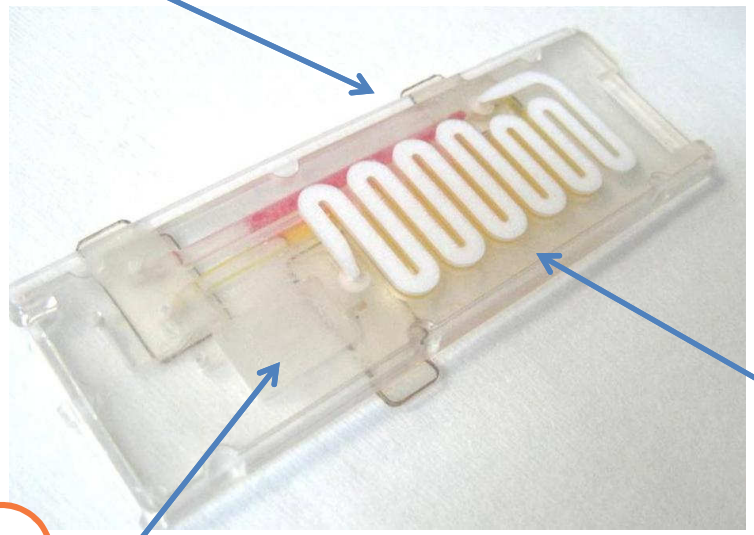
“Devices based on nanowires are emerging as a powerful platform for the direct detection of biological and chemical species, including low concentrations of proteins and viruses.” 1st July 2006 Analytic Chemistry

DNA Extraction/Purification

QuantuMDx

Sample in, DNA out

Sample loading channel

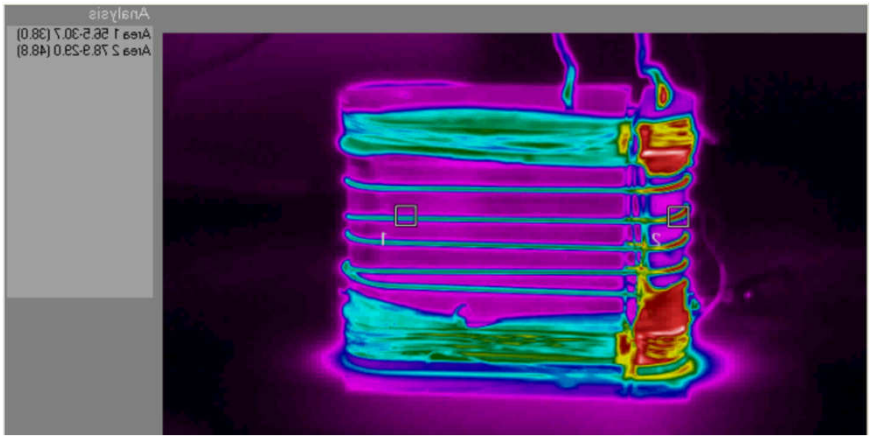


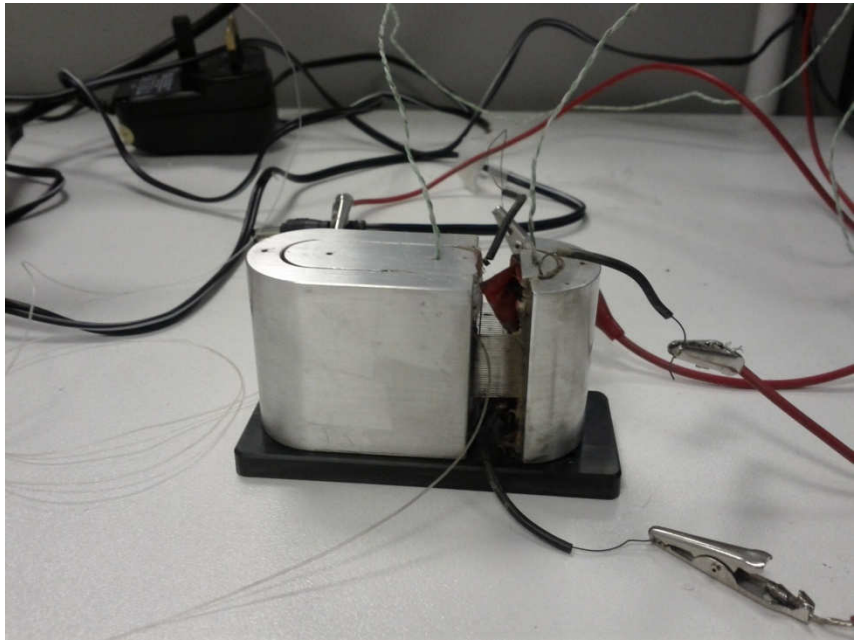
Sample collection well

Sorbent filter in channel

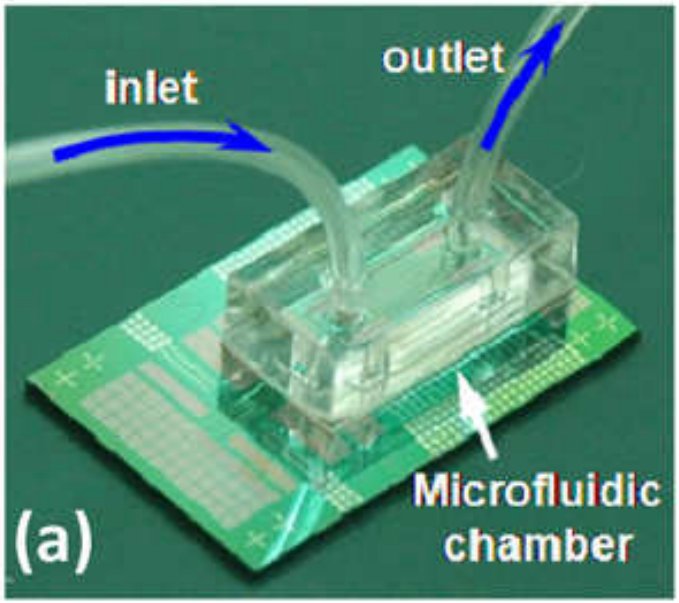
Amplification:
Microfluidic thermal PCR
Test rig – the Cylon

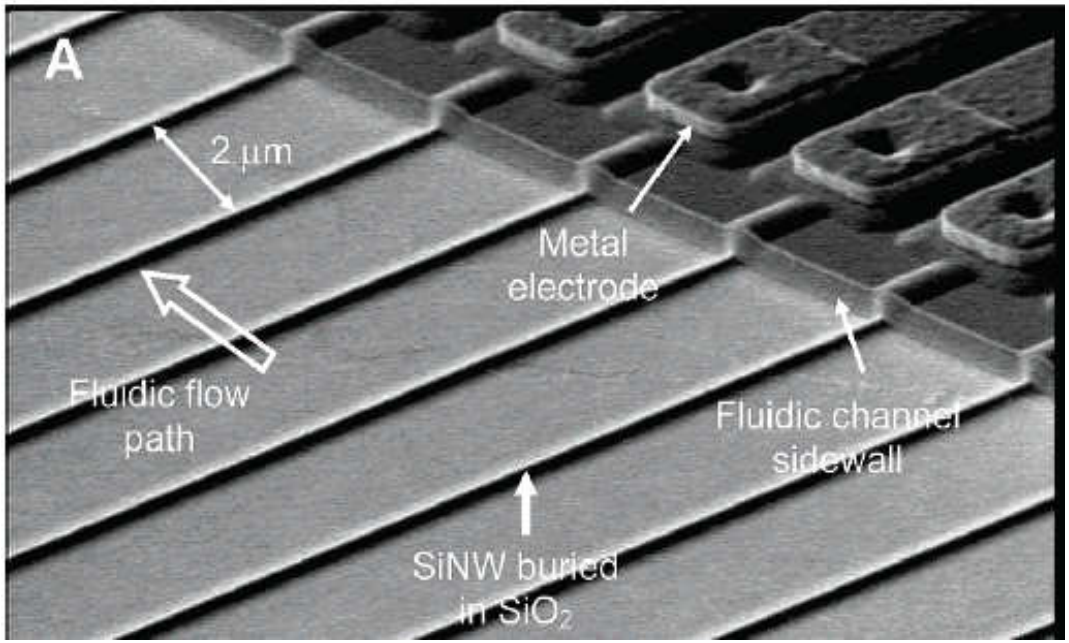
QuantuMD





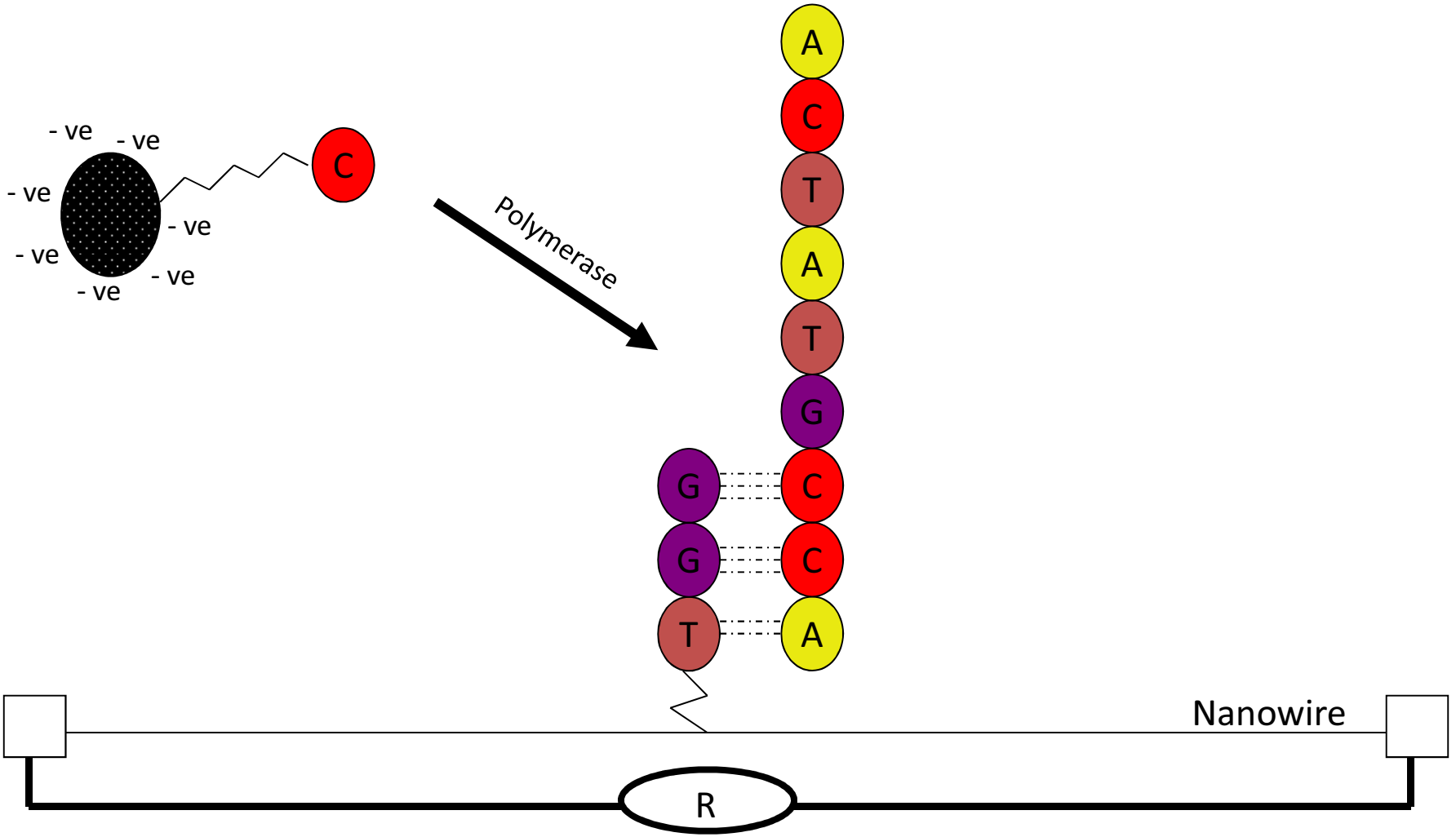
The “cool” bit



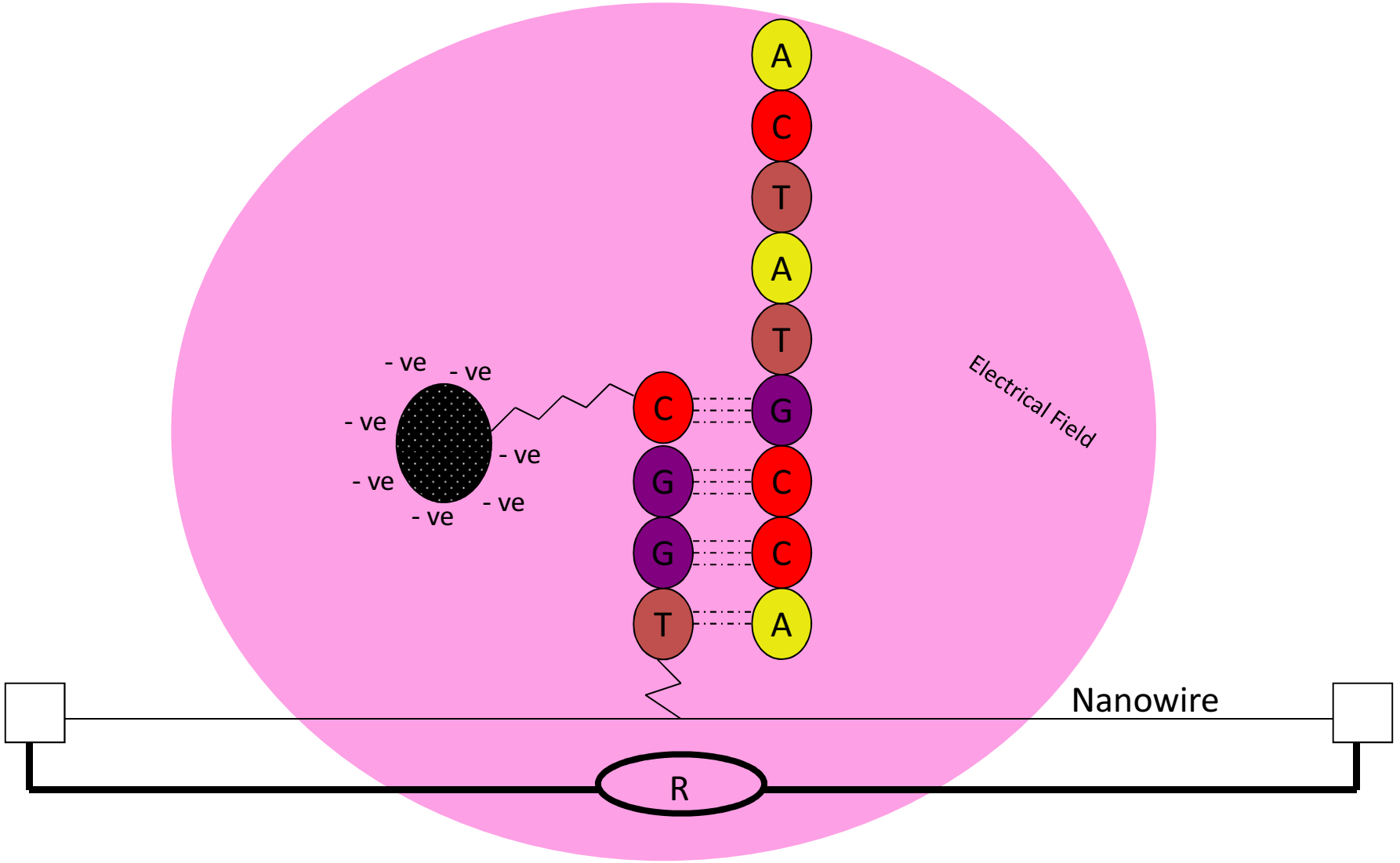


Your fingernails grew 30 nanometres while you were reading this slide

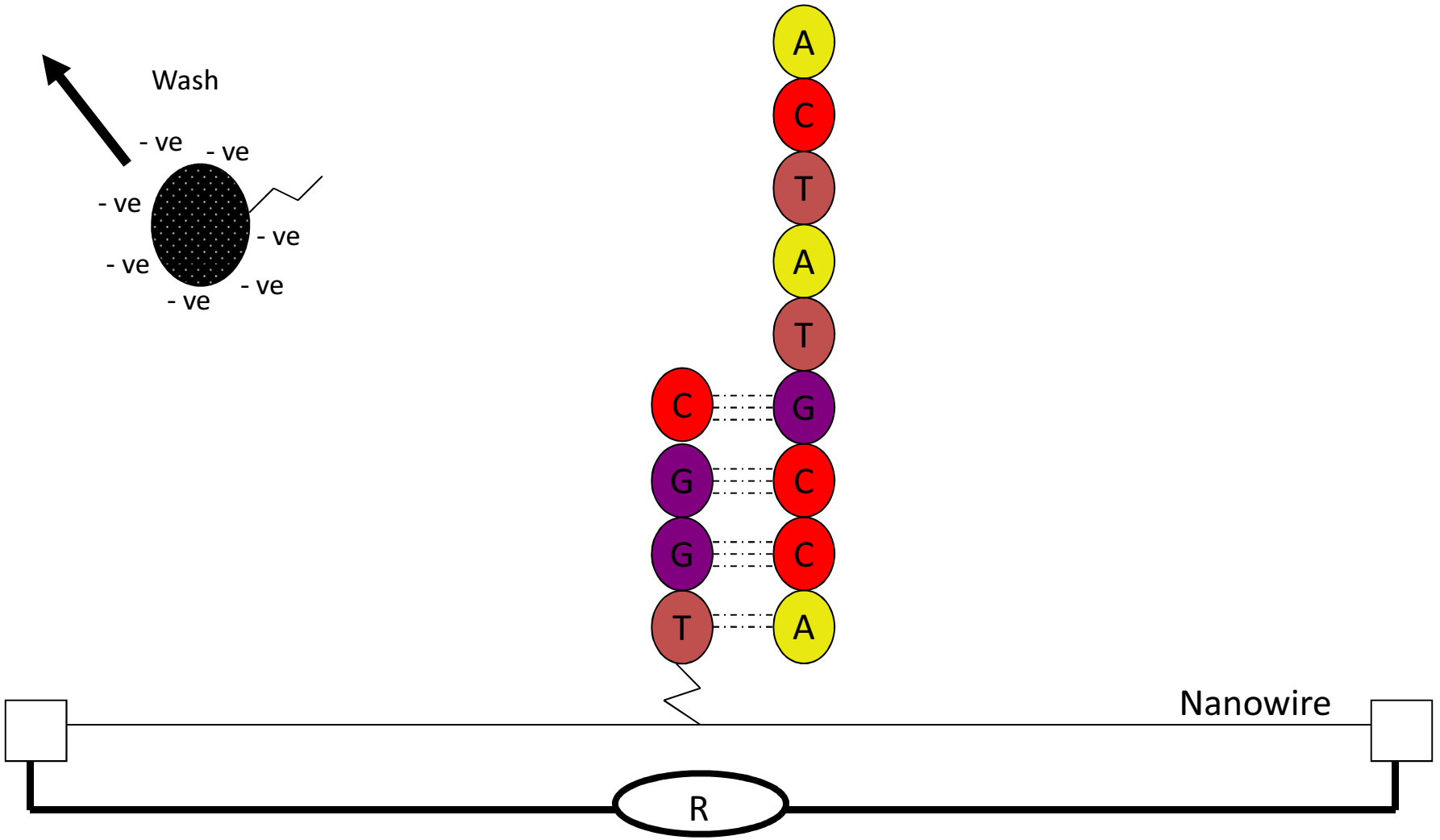
Finding a mutation



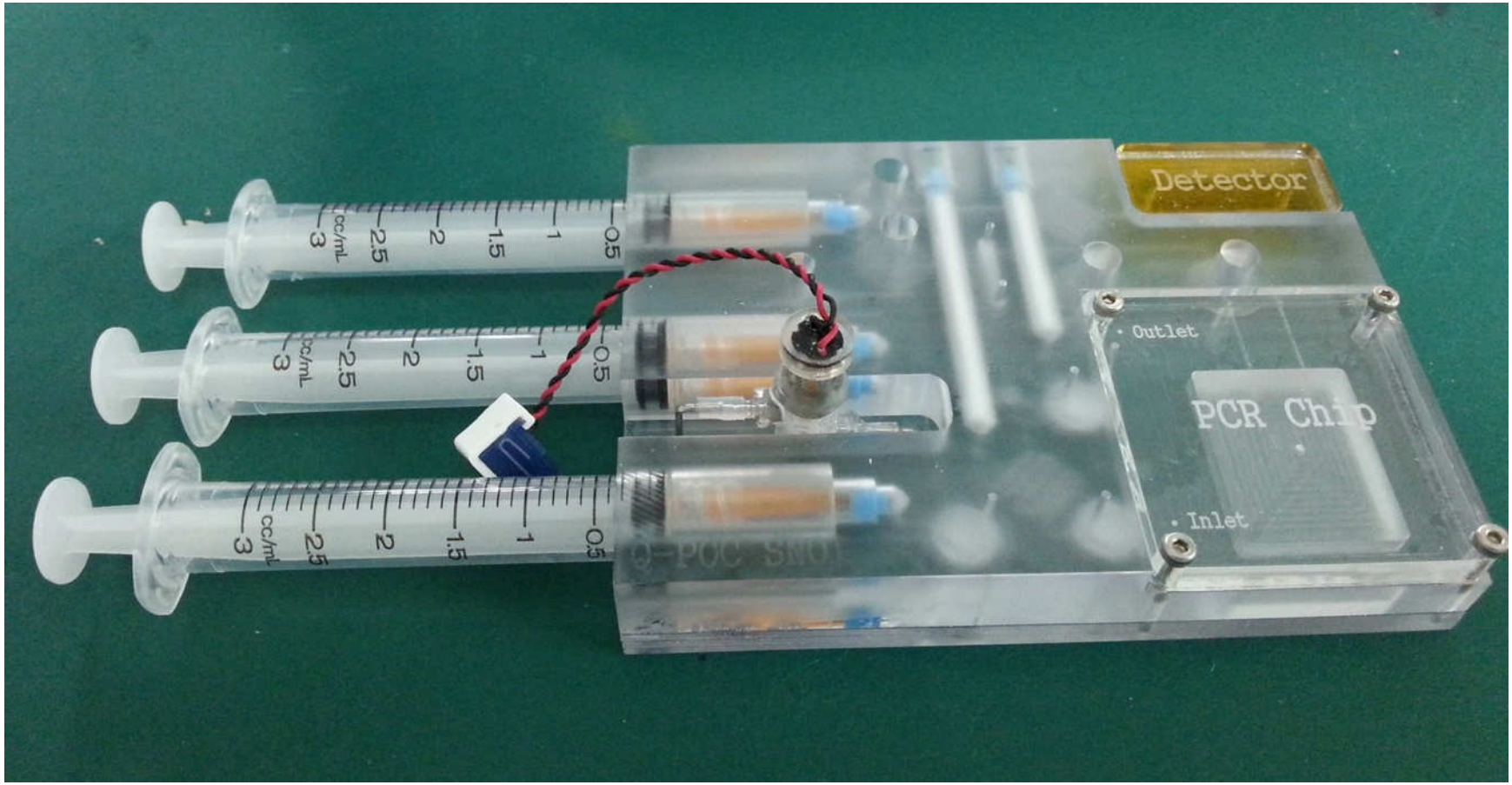
Finding a mutation



Finding a mutation

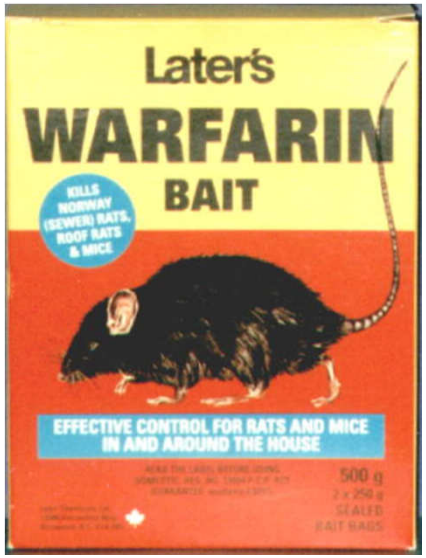


Born April 2014



“Handheld”

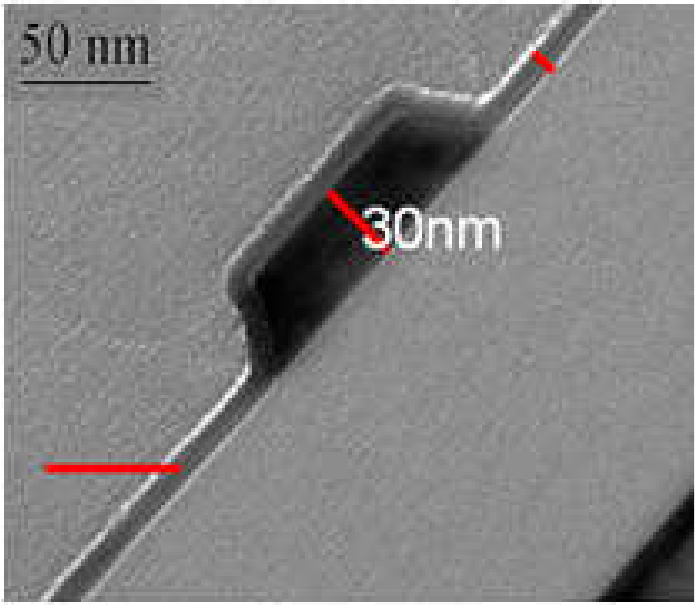
Infectious diseases (TB & malaria), pharmacogenetics, Molecular pathology



Also Coming soon!

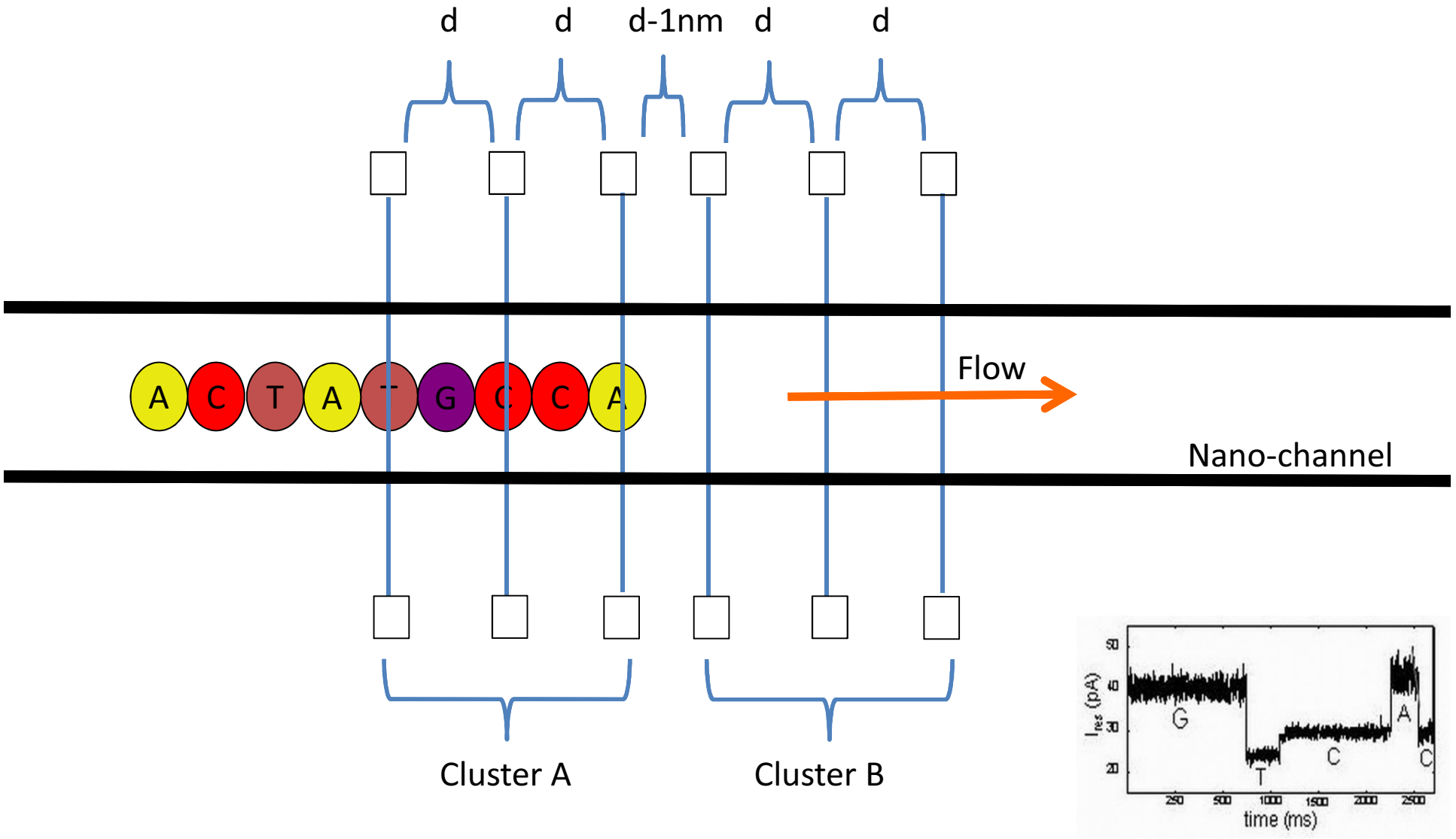
Q Seq Nano-channel Sequencing

Feeding whole DNA molecules into a nanowire-lined channel



High resolution transmission electron microscopy (TEM) images for cross-sectional profile of nanochannel prepared by focused ion beam milling (FIB).

Drive intact DNA molecules
Past nanowire clusters sensitised to A,C,G or T



Summary

- Sequencing technology is getting faster and cheaper
- With better technology, we could diagnose the disease accurately and quickly
- The next best technology is being developed in Newcastle…. So watch the space!

Thank you folks!

Newcastle University

QuantuMD

CANCER RESEARCH UK

ACADEMIC HEALTH SCIENCE NETWORK
NORTH EAST AND NORTH CUMBRIA