







The 100,000 genomes project and you

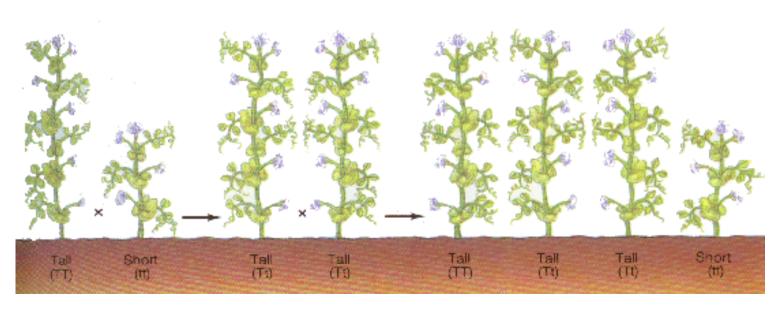
Patrick Chinnery
Institute of Genetic Medicine

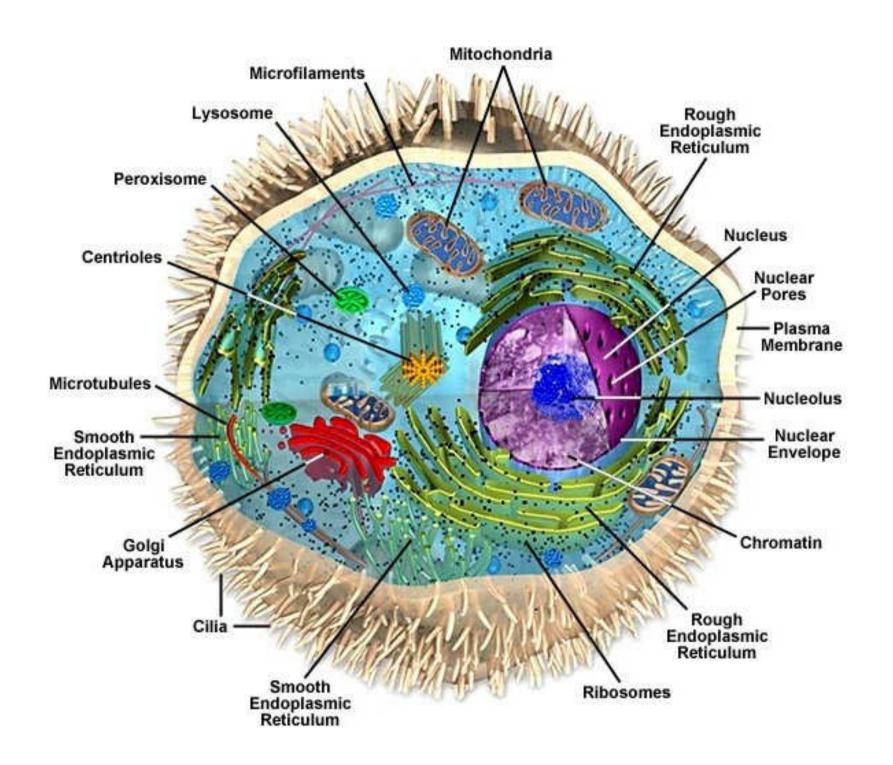




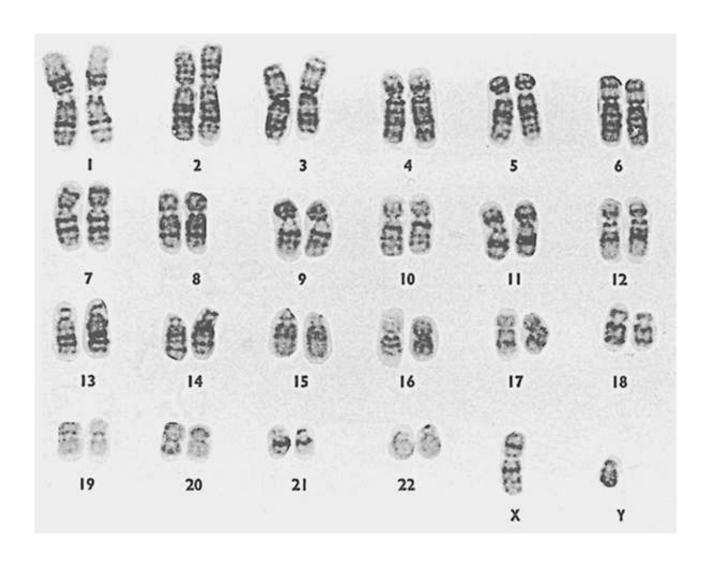


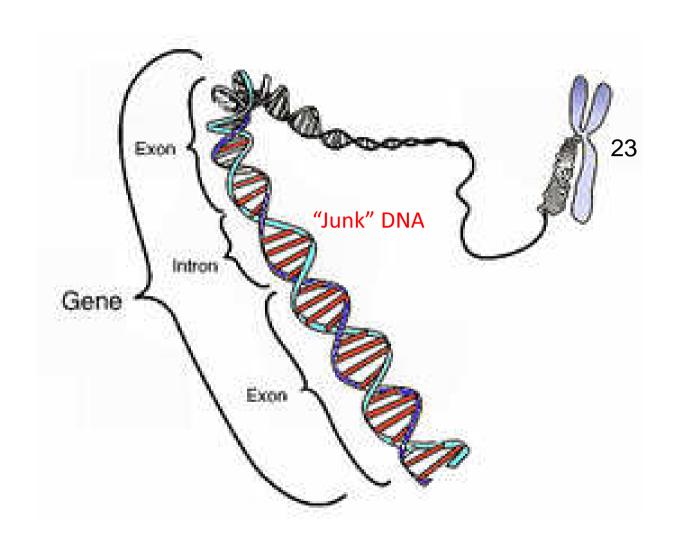
Gregor Mendel 1822-1884

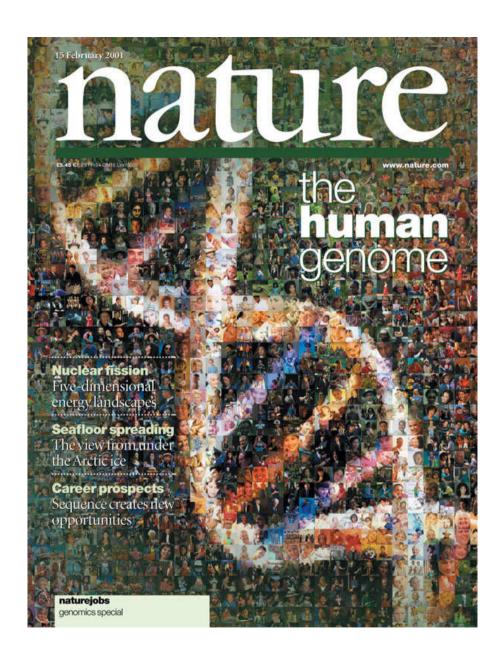




Human chromosomes



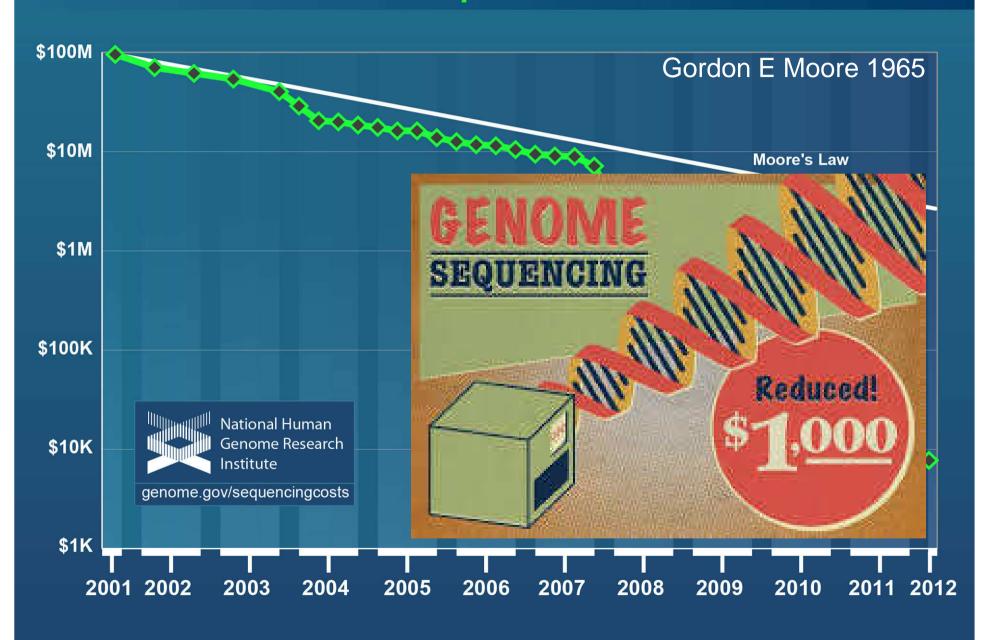




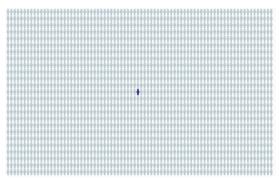
Year 2000 = \$3Billion



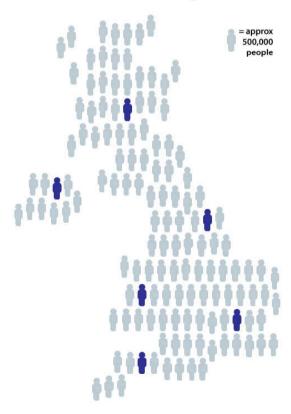
Cost per Genome



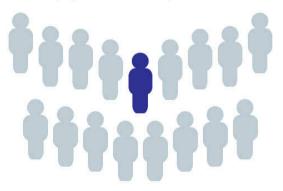
The UK defines a 'rare disease' as one that affects **1 in 2,000** or less of the population...



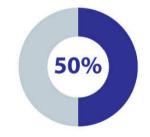
In total, that's about **3 million** people currently in the UK who will be affected by a rare disease



... so, collectively, rare disease will affect **1 in 17** of the population at some point in their life



50% of newly diagnosed cases of rare diseases are in children



There are between 5,000 and



different rare diseases...

... and **80%** of them have a known **genetic origin**



Rare diseases

December 2012

The Telegraph

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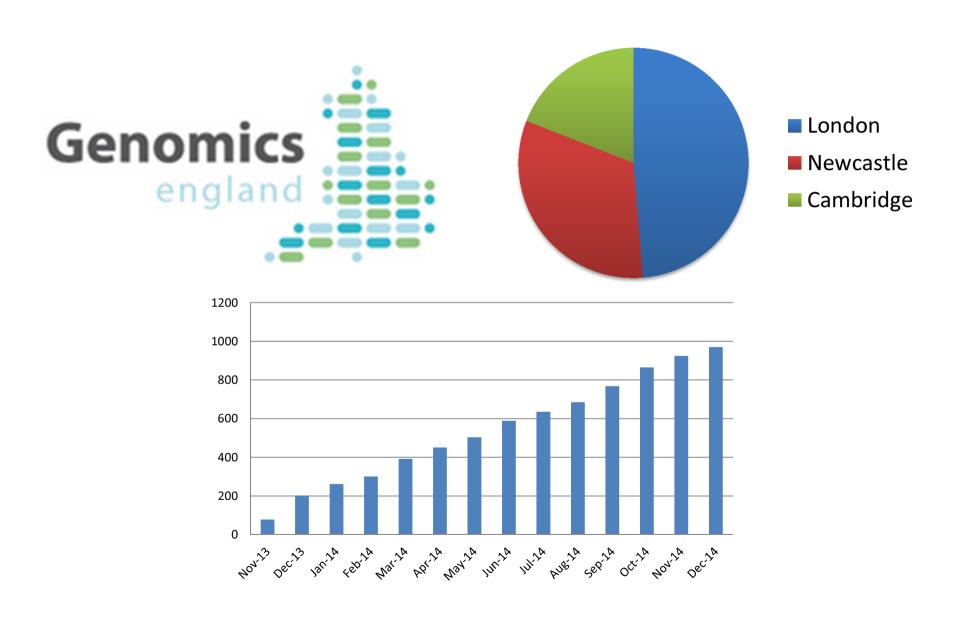
David Cameron unveils plan to map patients' DNA

The Prime Minister says he wants to "push the boundaries" by introducing genetic sequencing to a mainstream health service for the first time.



- Rare diseases
- Infection
- Cancer

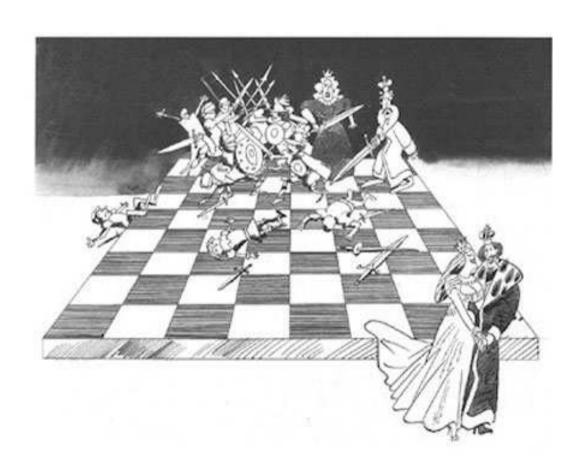
Newcastle recruitment - December 2014



Functional classification of JDW SNPs

Known SNPs	Novel SNPs
8967	2421
44	5
9121	2261
18517	7102
922048	381924

Ethical hurdles



Unexpected results

Personalised genetic medicine



