



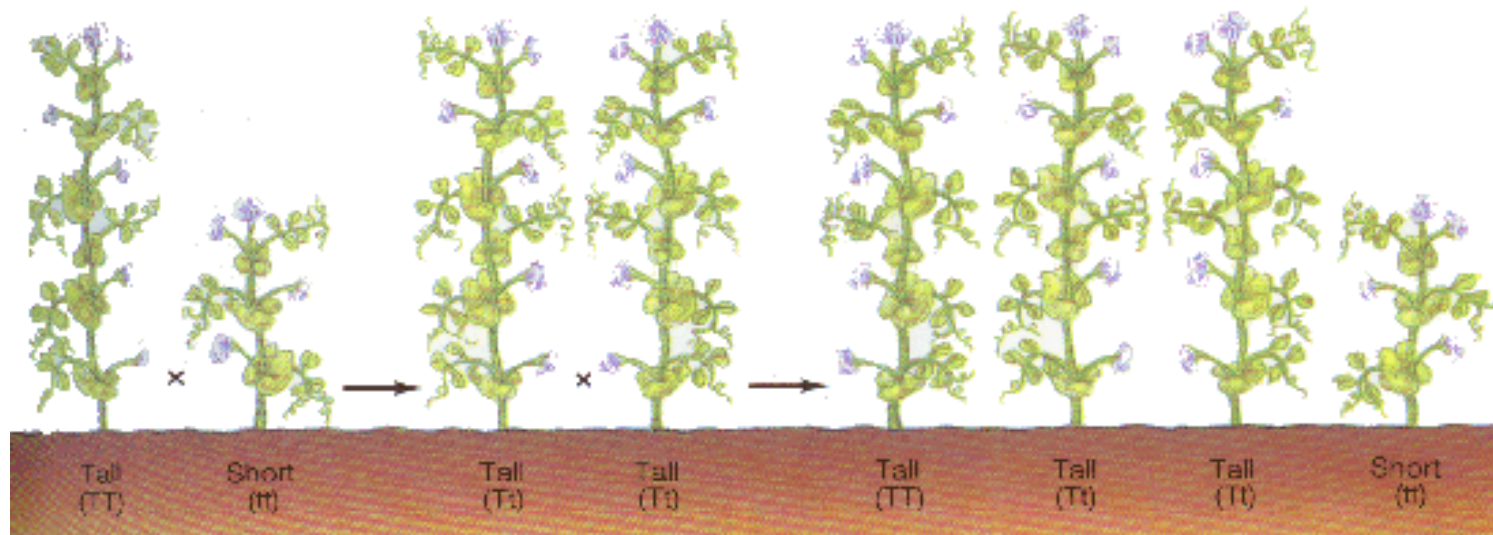
The 100,000 genomes project and you

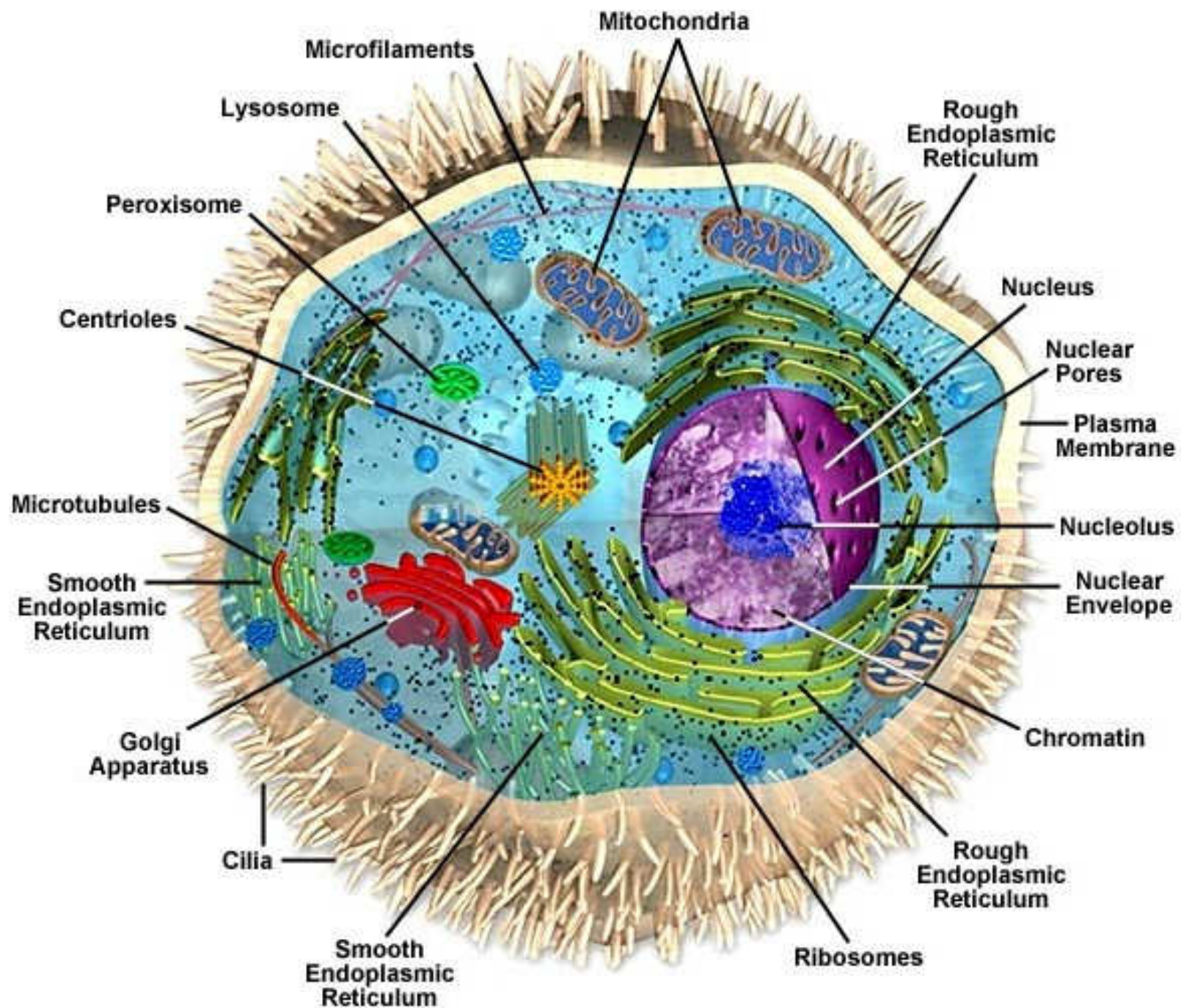
*Patrick Chinnery
Institute of Genetic Medicine*



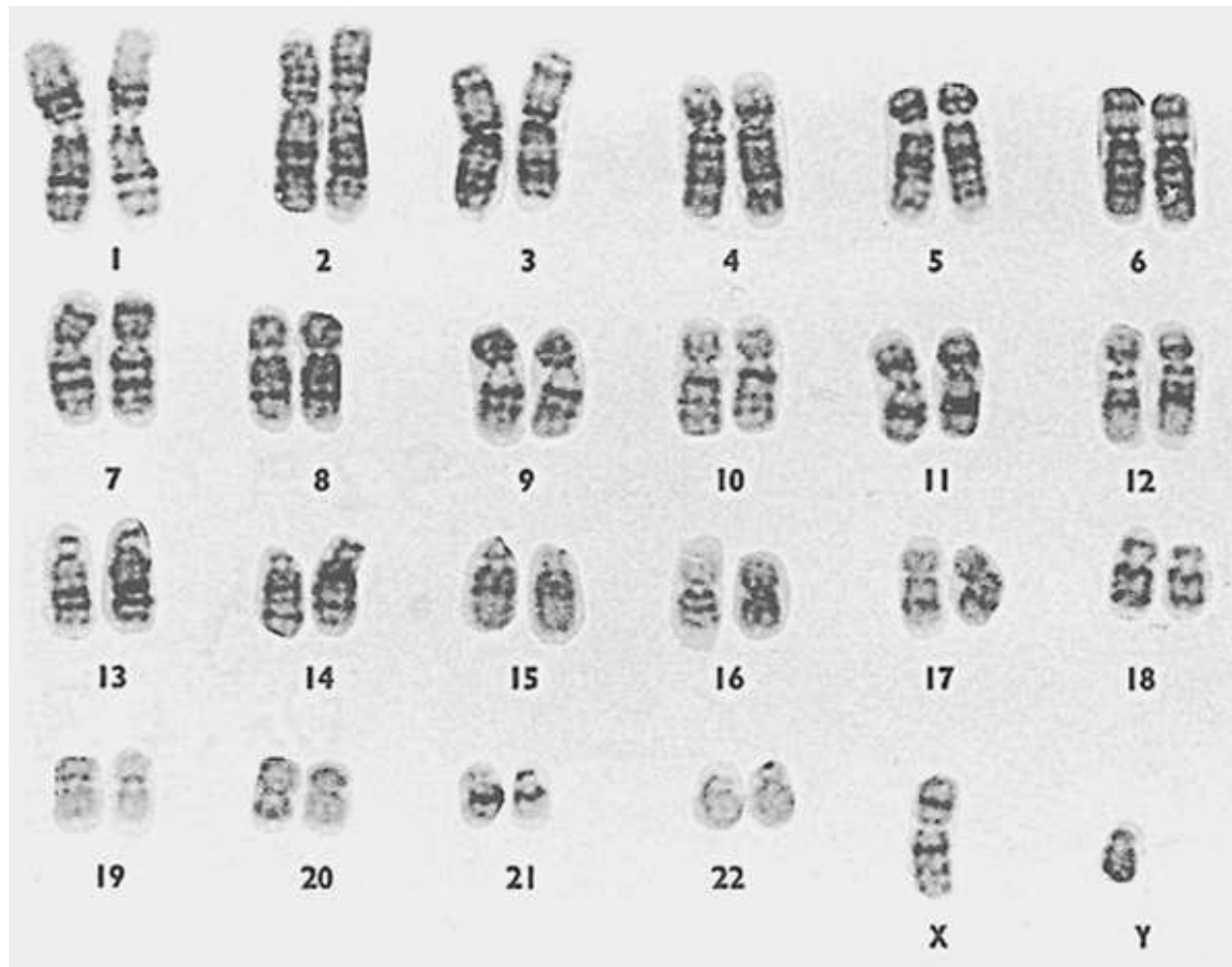


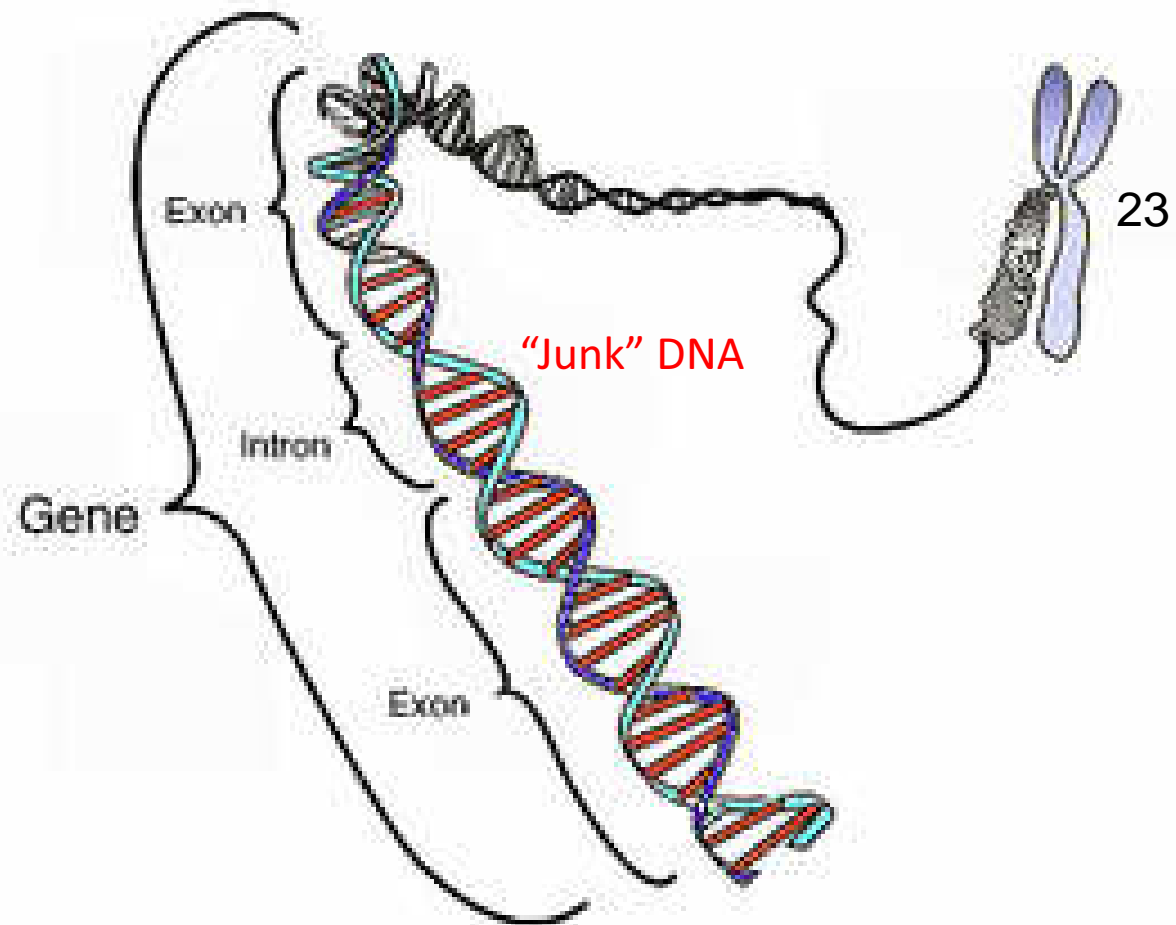
Gregor Mendel
1822-1884





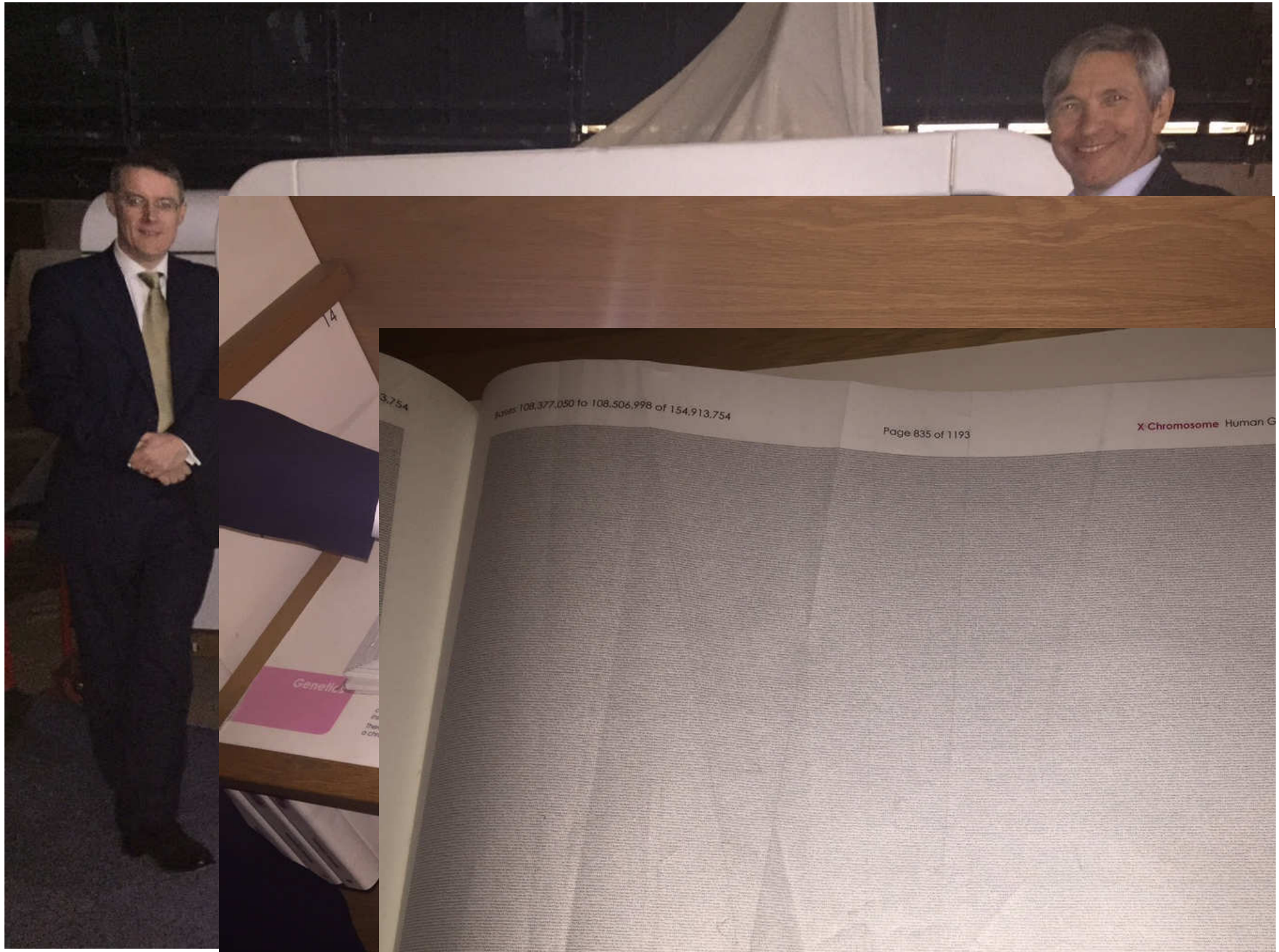
Human chromosomes



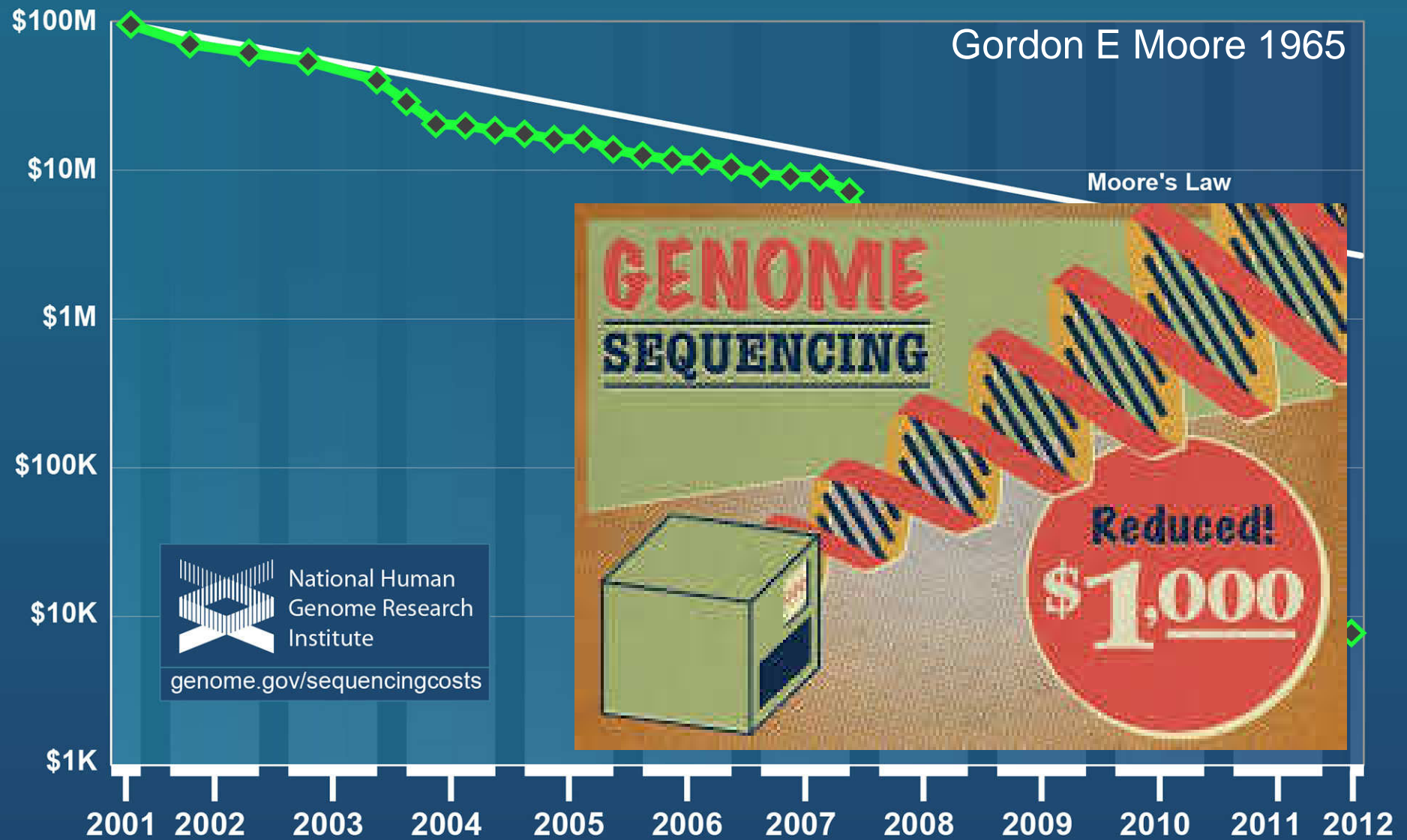




Year 2000 = \$3Billion

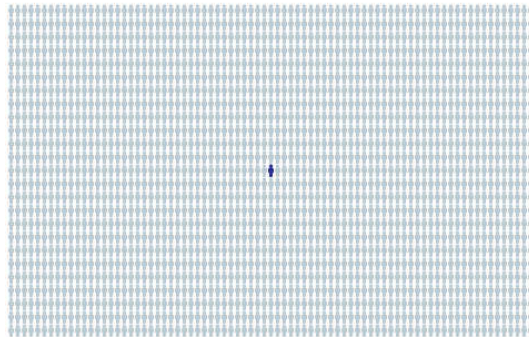


Cost per Genome

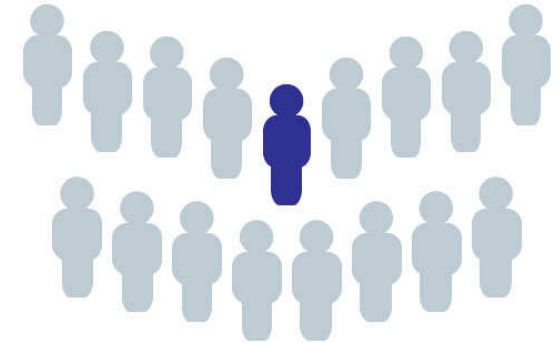


Rare diseases

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



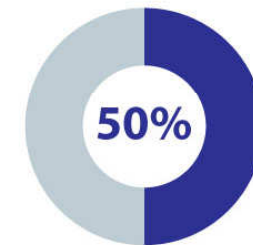
... so, collectively, rare disease will affect **1 in 17** of the population at some point in their life



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



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



There are between **5,000** and

8,000

different rare diseases...

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



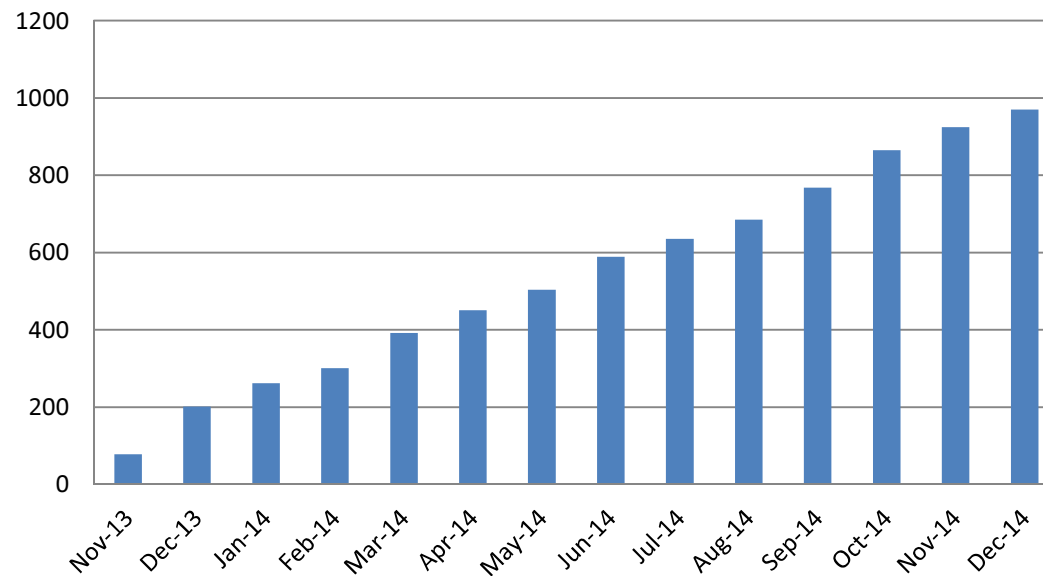
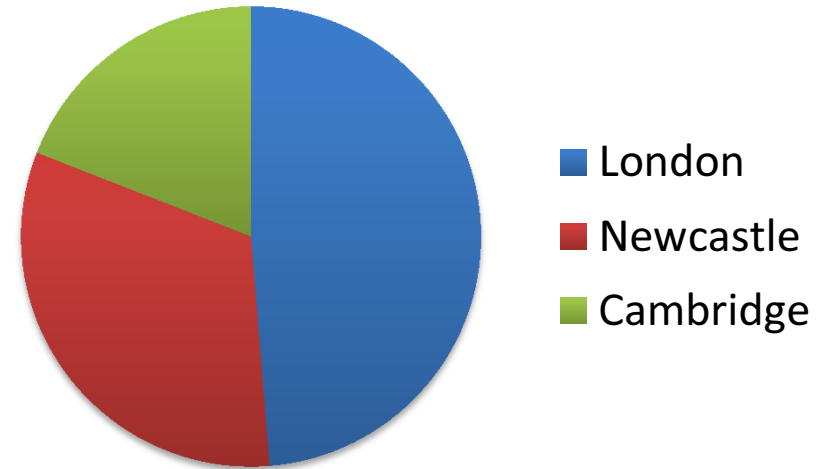
David Cameron unveils plan to map patients' DNA

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

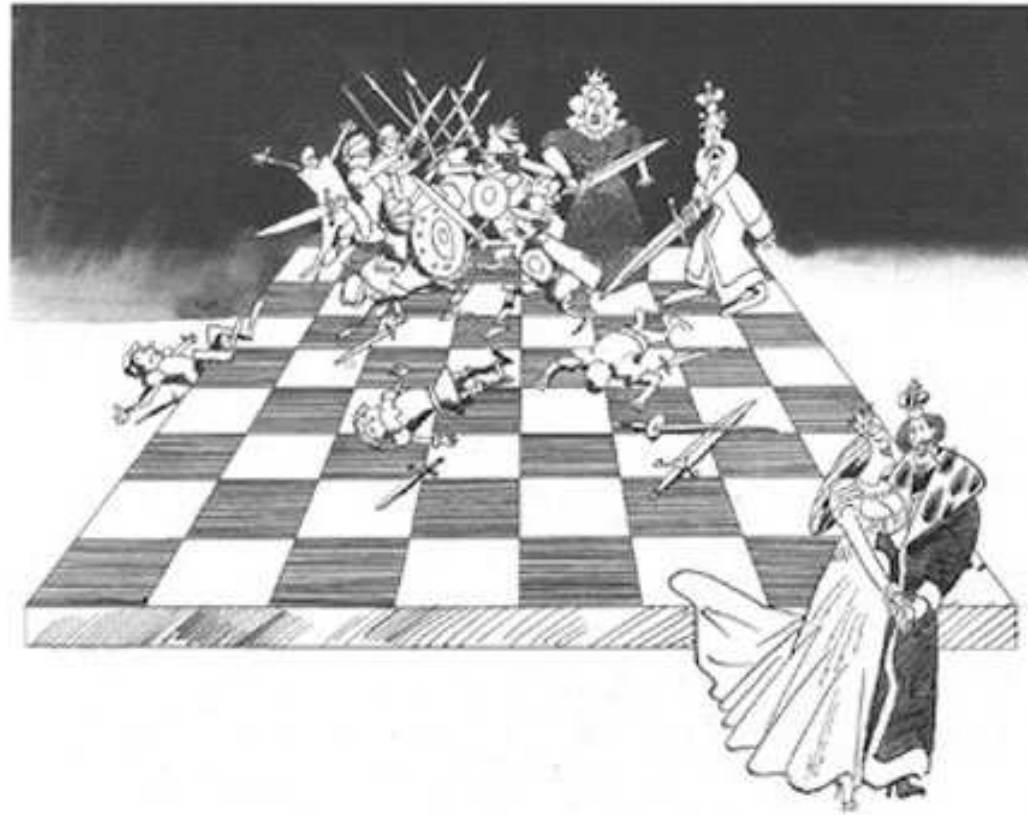


Functional classification of JDW SNPs



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

Ethical hurdles



Unexpected results

Personalised genetic medicine



