# Genomics and the future direction of health research

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# What are the aims of genomic health research?

- Identify the genetic causes of disease
- Predict a person's risk of developing disease - guide prevention strategies
- Diagnose diseases more quickly
- Predict responses to different treatments - precisely tailor treatment
- Classify cancers to guide treatment choices

# Bigger data – international genomic projects

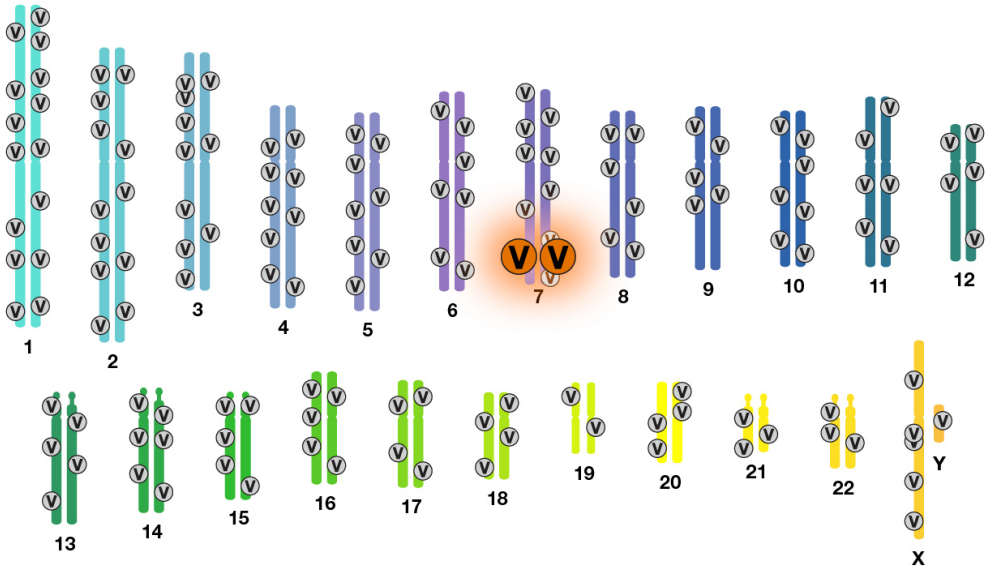
- deCODE (Iceland) – 160,000+
- UK Biobank – 500,000
- *All of Us* Research Program (US, 2018-2023) – 1 million
- European '1+ Million Genomes' Initiative (2020-2022)
- Chinese Millionome Database (CMDB) – ? Million
- National genome projects - Australia, Canada, Dubai, Estonia, France, Hong Kong, Japan, Netherlands, Qatar, Saudi Arabia, Singapore, South Korea, Sweden, and Turkey

# Major UK genomic projects

- 100,000 Genomes Project (2013-18) - 120,239 genomes from 86,618 participants (rare disease and cancer)
- UK Government ambition - 5 million genomes in the UK by 2023/24 (includes 500,000 from UK Biobank and 500,000 from NHS Genomic Medicine Service (GMS) in England.
- *Our Future Health* (2022+) – 5 million recruits. Common chronic disease. Polygenic risk scores.

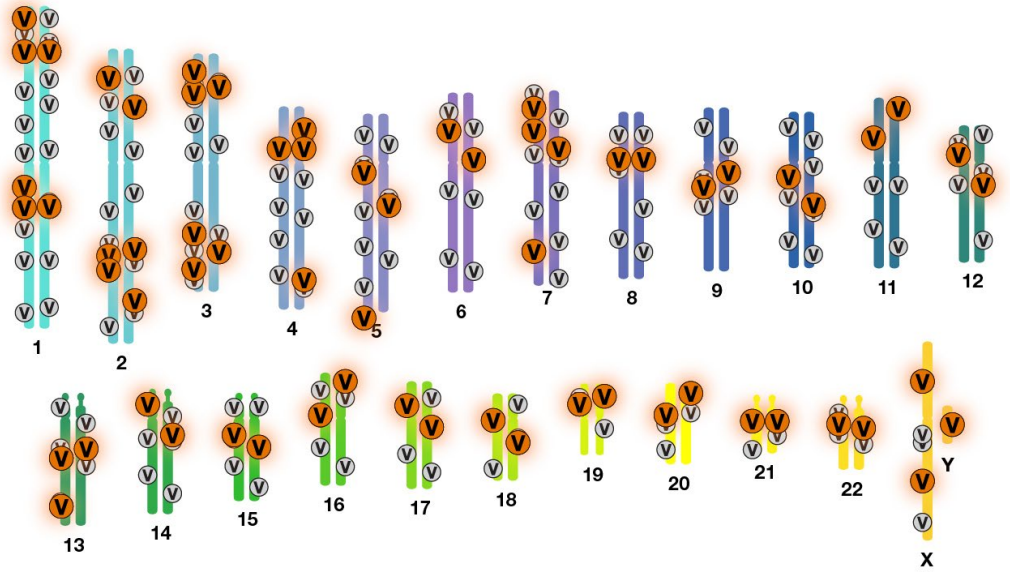
# Monogenic

e.g. cystic fibrosis




# Polygenic

e.g. coronary artery disease




V = genetic risk variant

# Genomic testing techniques for rare and inherited disease

Current:

- Exome sequencing (all genes – but only protein making bits) ~£700

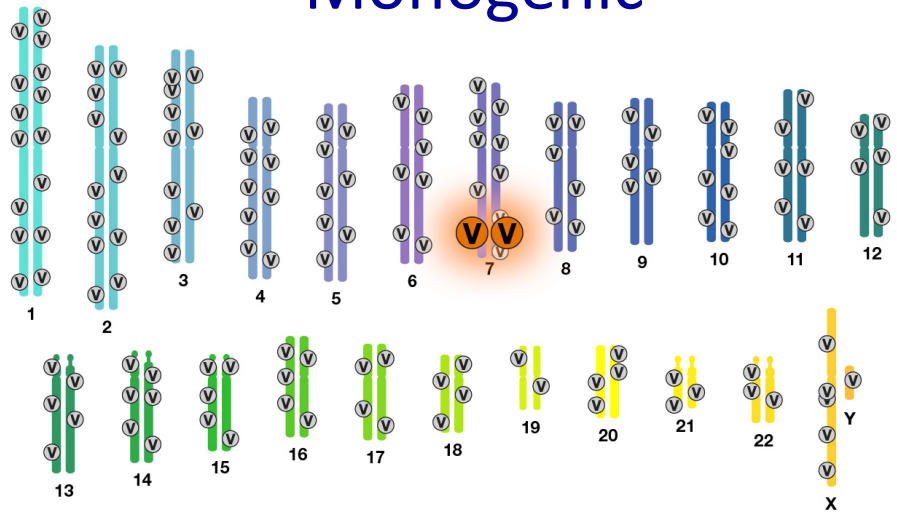
- Whole genome sequencing (all genes and bits in between) = ~£2,000 (50x more data, ~5% more diagnoses!)

R&D stage:

- Transcriptome (RNA sequencing analysis)

- Long-read sequencing (detailed structural information)

Monogenic



NHS firewall

AWMGS

storage

sequencers

Digital infrastructure

CONSENT

Public/patient engagement

Secure research environment(s)

e.g. SAIL, NDR, Genomics England

Researcher access

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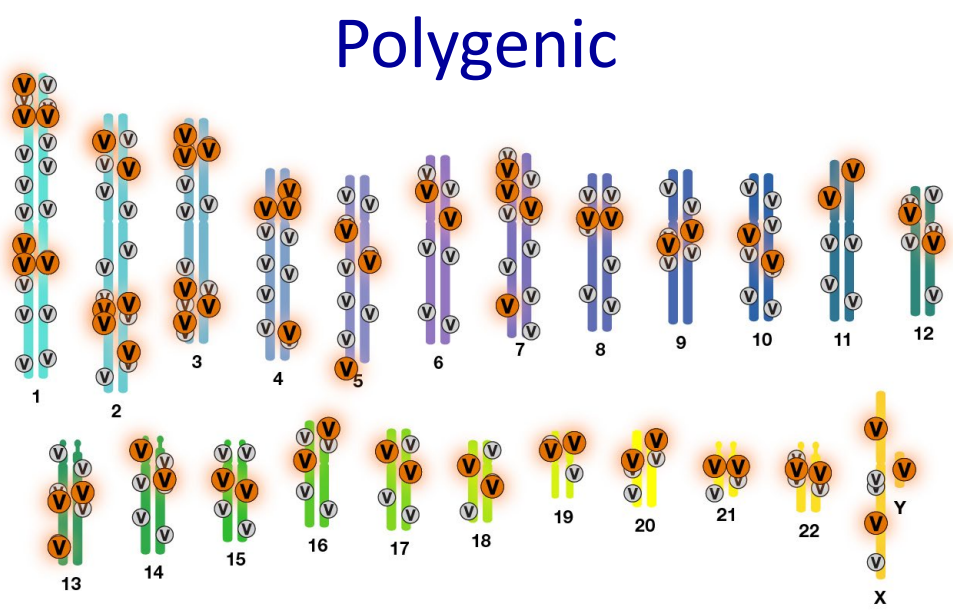
# Polygenic risk scores (PRS) for common disease

- PRS = weighted count of multiple genetic markers
- Risk variants identified by large epidemiological studies (genome-wide association studies)
- PRS available for many conditions such as coronary artery disease, type 2 diabetes, and cancers
- Cheaper than sequencing for monogenic disease – use “SNP-chips”
- PRS struggle to capture full variance of traits – limits predictive power



# Questions?

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