PHG FOUNDATION

making science work for health

CDC Public Health Genomics 25th Anniversary Mini Symposium

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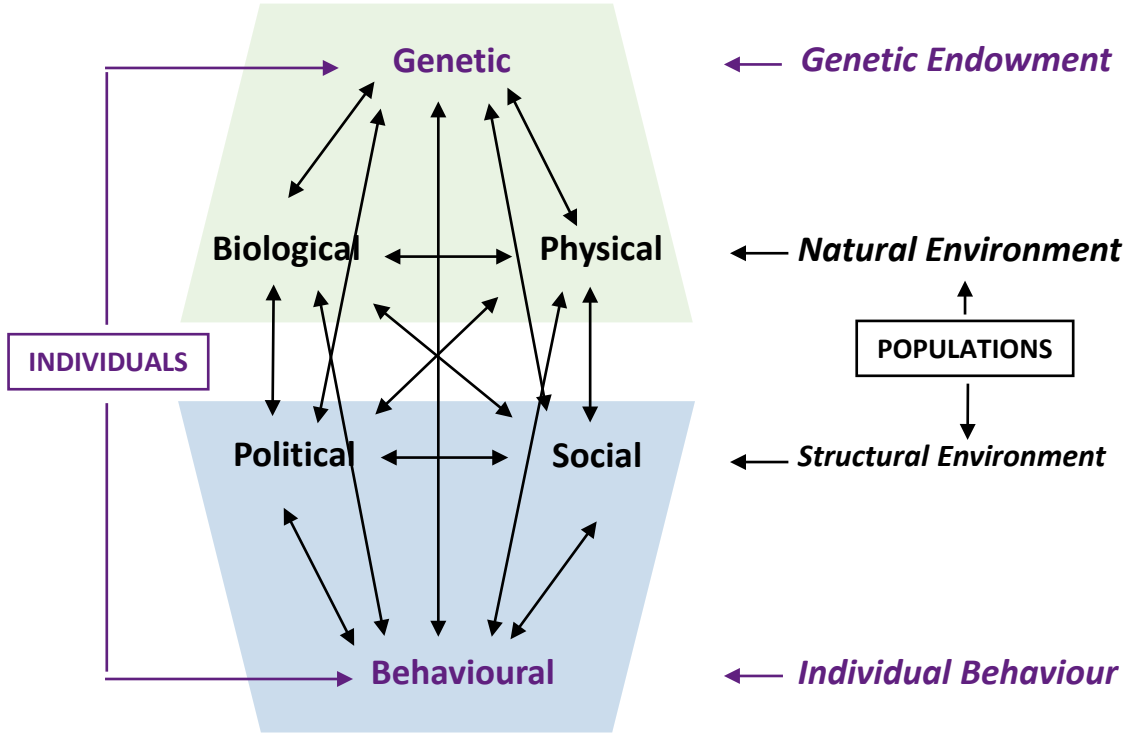
Chair, Board of Trustees, PHG Foundation

Webinar 9 February 2023

UNIVERSITY OF CAMBRIDGE

Early Years
Basic Principles

Determinants of Health



Biological versus social

External versus internal

Individual versus population

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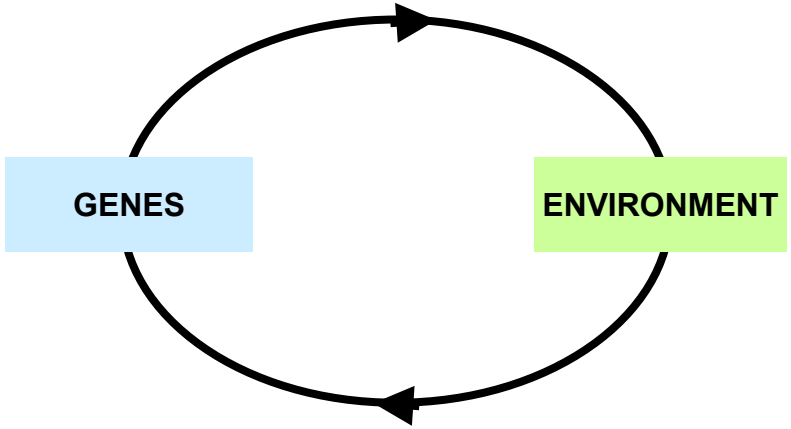
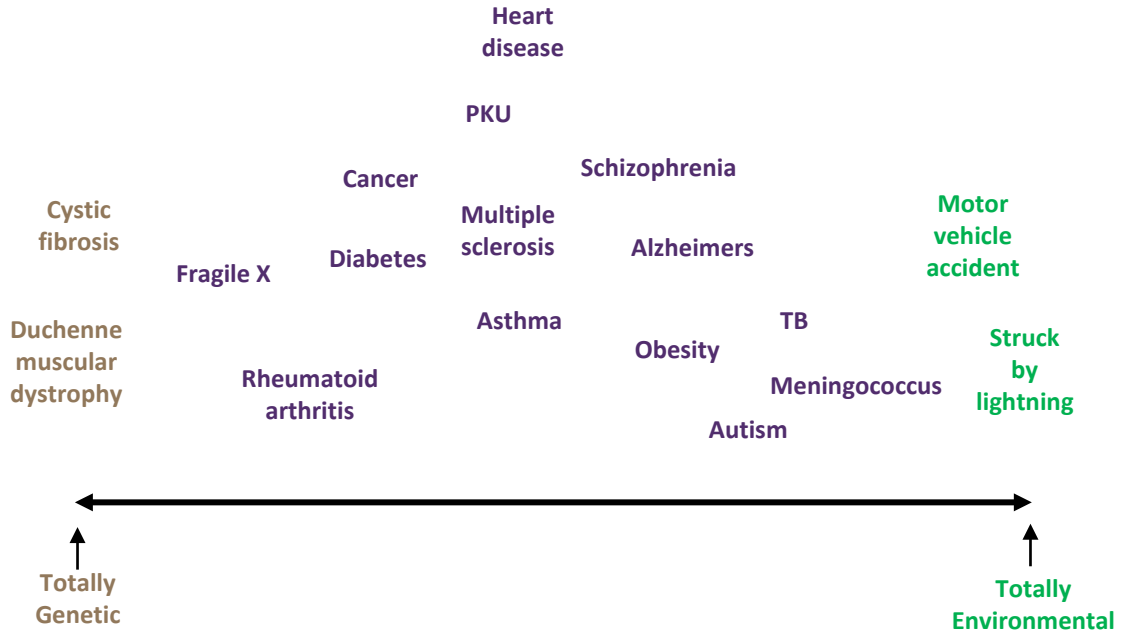
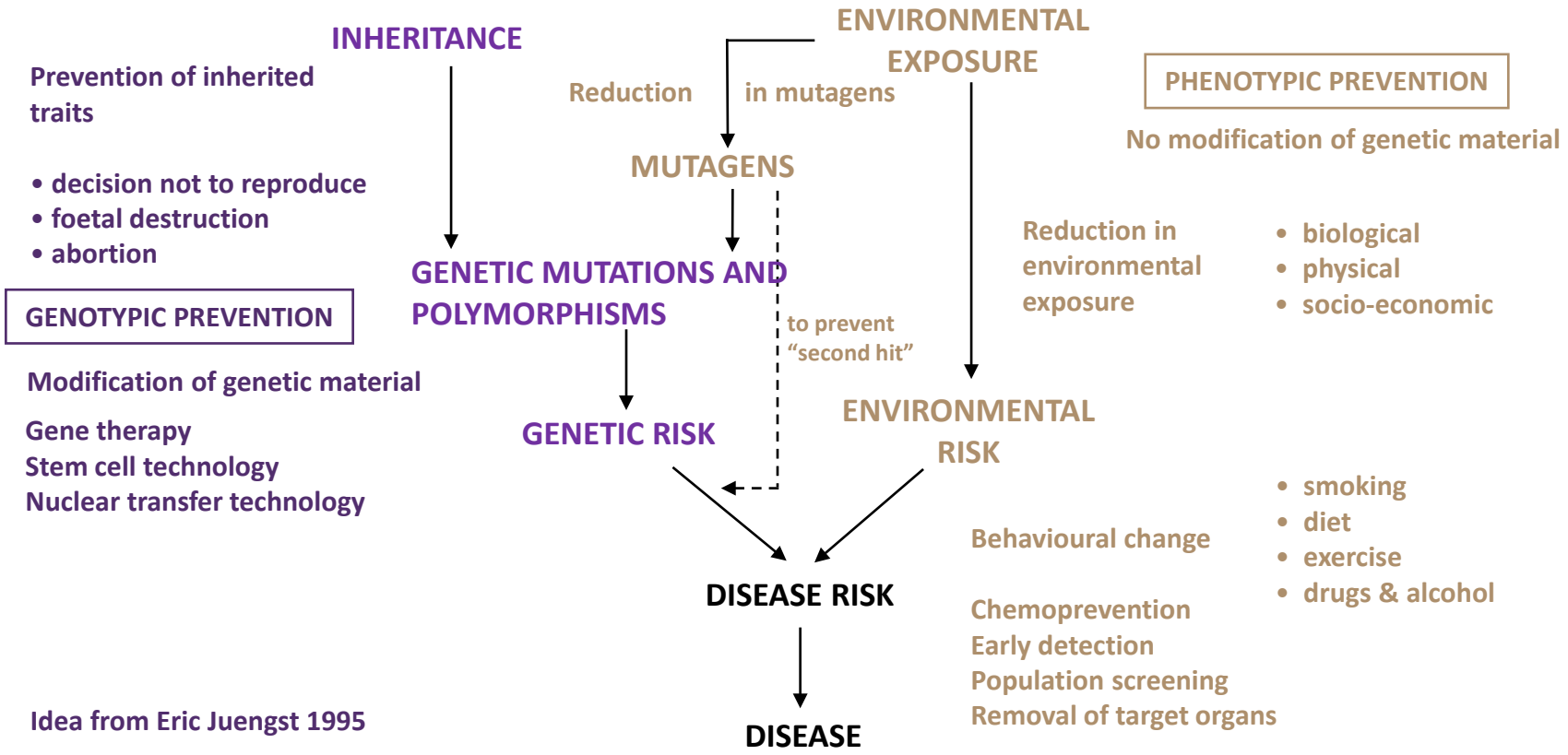
Gene-Environment Interaction





A Model of Disease Prevention



The Nuffield Trust
Scenario Project 1999

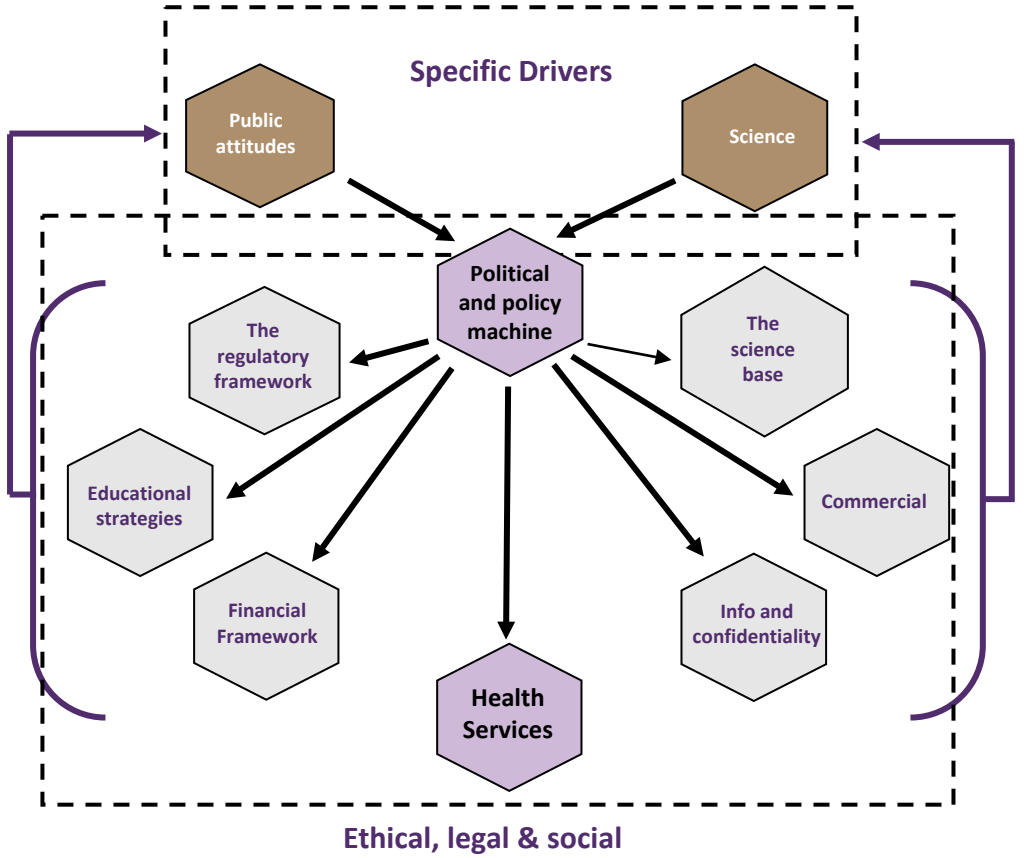
Issues Set Out in the Report

A. Two specific drivers

1. Science and its capacity to improve human health
2. Public attitudes towards genetics

B. Six policy issues

1. The regulatory framework
2. Educational strategies
3. The science base
4. Commercial considerations
5. Information and confidentiality
6. Financial framework for health



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Principal Recommendation

The UK government should take the lead in developing a policy framework that would provide a context within which scientific developments in genetics and molecular biology and their clinical and public health applications might be assessed.

The framework should:

a) **be informed by a set of core values**
b) **take into account the issues set out in this report**
c) **be developed in consultation with all relevant stakeholders and with the public**
d) **ensure partnership arrangements between industry, the universities and the health service**
e) **emphasise the need for co-ordination across all government departments**

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Bellagio 2005

The Bellagio Initiative (2005)

1. Expert meeting convened with funding from the Rockefeller Foundation
2. Eighteen participants from the UK, USA, Europe and Canada
3. Interdisciplinary group
4. Unanimous agreement about the definition and scope of the enterprise known as public health genetics
5. Establishment of an international collaboration known as GRAPH Int - Genome-based Research and Population Health International Network
6. Commitment to take on a leadership role

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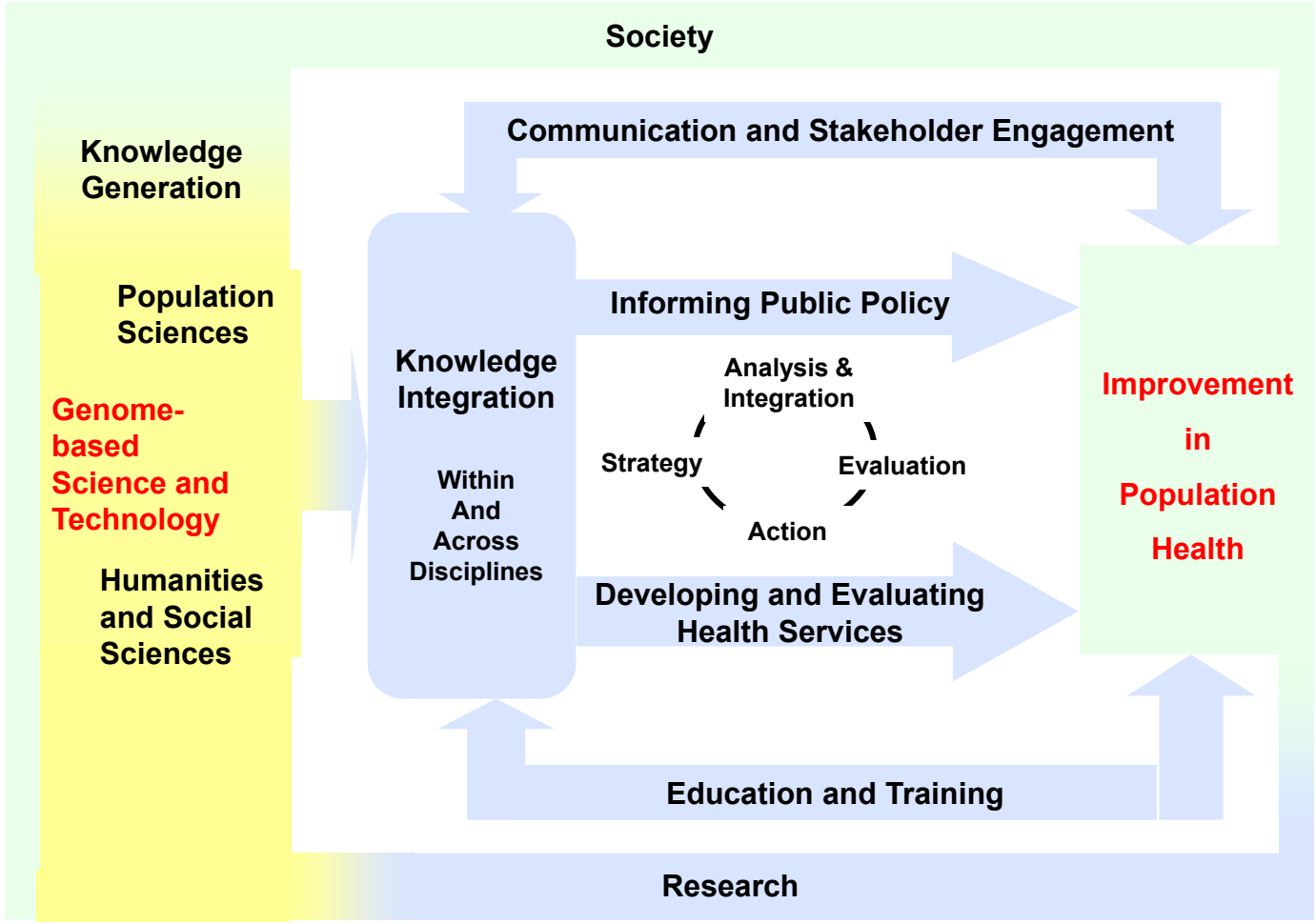
The Enterprise



Knowledge Base for Public Health Genetics

GENETIC SCIENCE

- Basic concepts of mendelian genetics
- Family histories and pedigrees
- Risk assessment and communication
- Principles of genetic epidemiology
- Principles of molecular genetics
- Genetic testing and screening
- Genetics of common disorders
- Gene-environment interaction

POPULATION SCIENCES

- Epidemiology
- Biostatistics
- Environmental health sciences
- Infectious diseases
- Social and behavioural sciences
- Health economics
- Health services research
- Management science
- Information science

HUMANITIES

- Sociology
- Anthropology
- Law
- Economics
- Ethics
- Metaphysics and epistemology
- Theology
- Political philosophy

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Knowledge Integration

The activity that we call knowledge integration is the driving force or 'engine house' of the endeavour

It is the process of selecting, storing, collating, analysing, integrating and disseminating information both within and across disciplines for the benefit of population health and includes methodological development

It is the means by which information is transformed into knowledge

Interdisciplinarity is a key feature

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Public Health Genetics

The effective translation of genome-based knowledge and technologies for the benefit of population health

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Key Policy Developments in the UK (1)

2003 Addressing Genetics, Delivering Health

Our Inheritance, Our Future: Realising the Potential of Genetics in the NHS

2005 Bellagio Meeting

2007 PHG Foundation established as independent Charity

Genetics, Health Care & Public Health

2009 Cell Free DNA for Non-Invasive Prenatal Diagnosis

2010 Genomic Medicine: *HL Science & Technology Committee*

2011 Next Steps in the Sequence

2012 Launch of 100,000 Genomes Project

Building on Our Inheritance *Report of Human Genomics Strategy Group*

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Key Policy Developments in the UK (2)

2015 Pathogen Genomics into Practice

2017 Generation Genome: Annual Report of the Chief Medical Officer

2018 Launch of NHS Genomics Medicine Service

Genomics and Genomic Editing in the NHS *Science & Technology Committee*

2020 Genome UK: The Future of Healthcare. *Government Policy Paper*

2021 Genome UK: 2021 to 2022 Implementation Plan

2022 Genomics Beyond Health. *Government Office for Science Report*

Accelerating Genomic Medicine in the NHS. *NHS England 5 Year Strategy*

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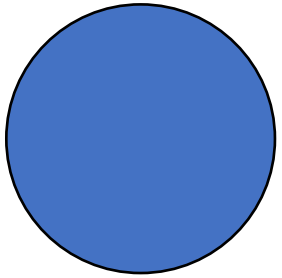
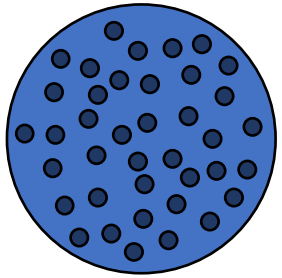
Some Implications

Populations and Individuals



Homogenous population

Single ontological entity



Heterogeneous population

A set of individuals

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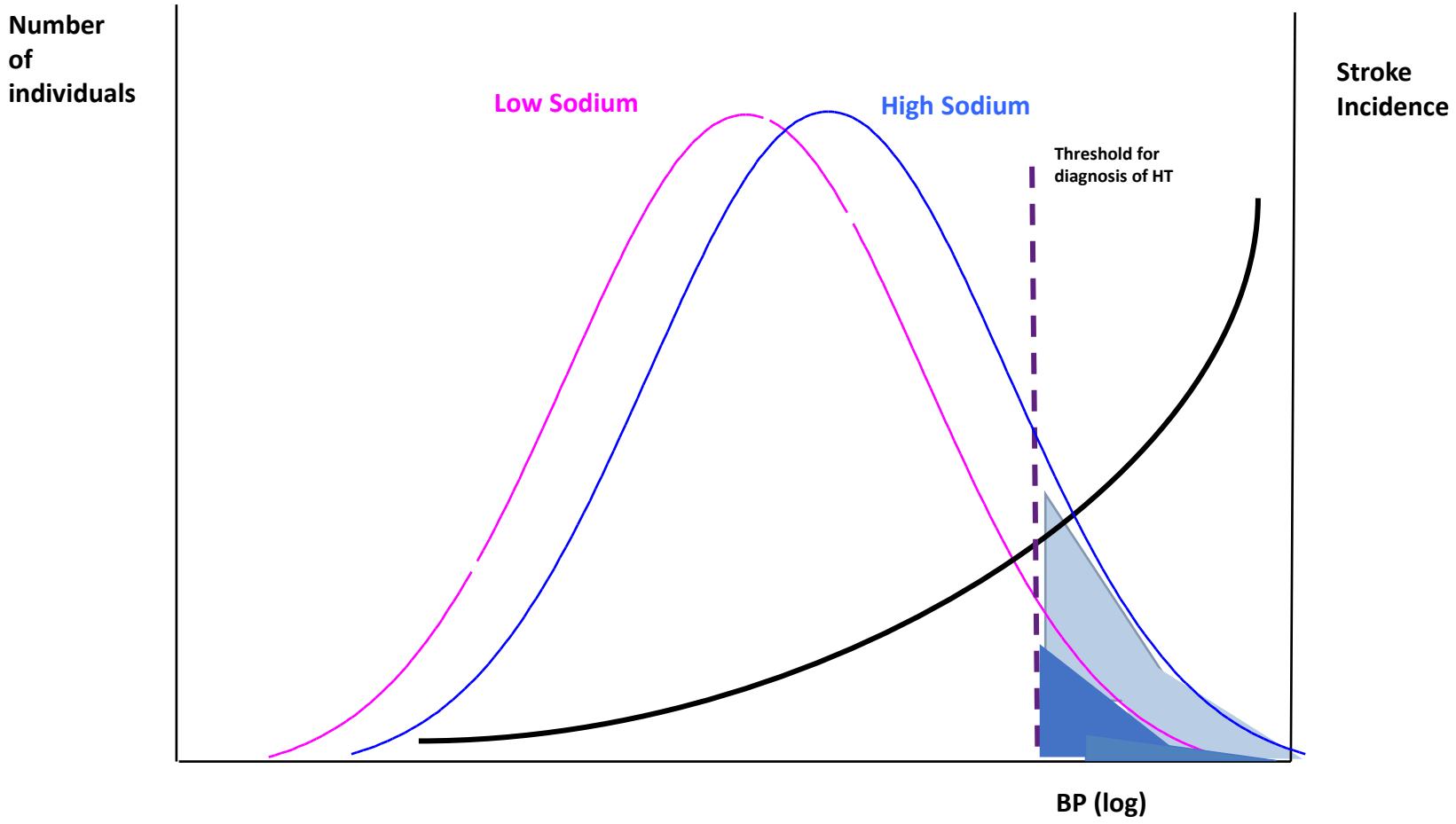
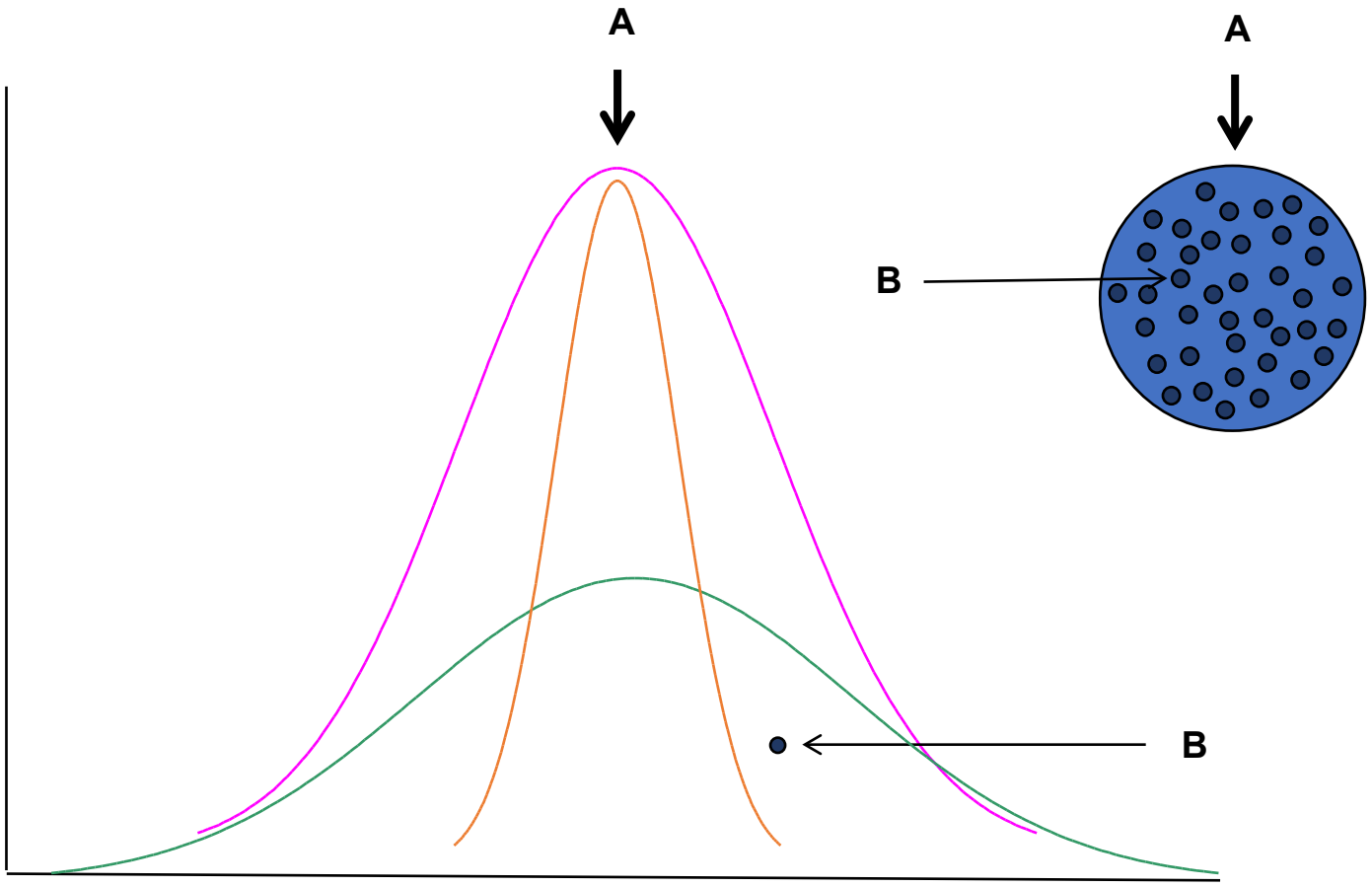
Geoffrey Rose: Population versus High-Risk Prevention



Geoffrey Rose: Causes of Incidence versus Causes of Cases



Exponential Growth of Scientific and Medical Data

1. Analysis and synthesis of data
2. Accessibility of data
3. Transparency of information
4. Empowering individuals

Growth of

- Scientific data
- Scientific publication

Bioinformatic Support

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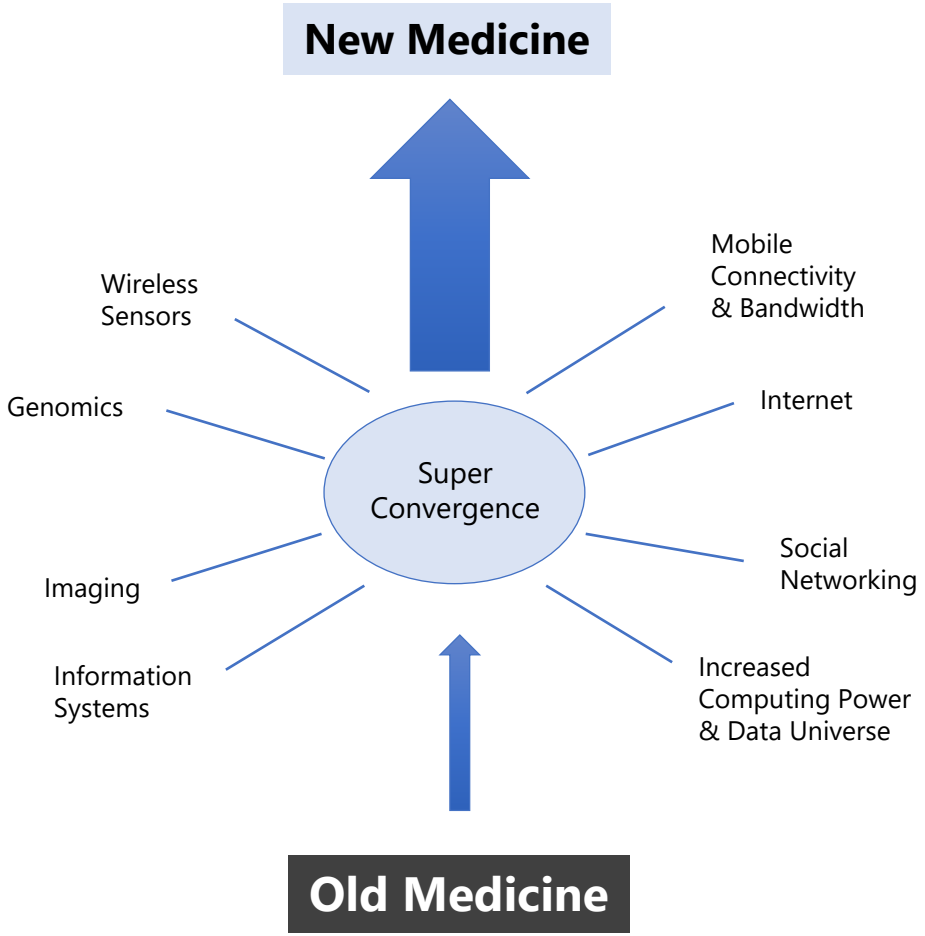
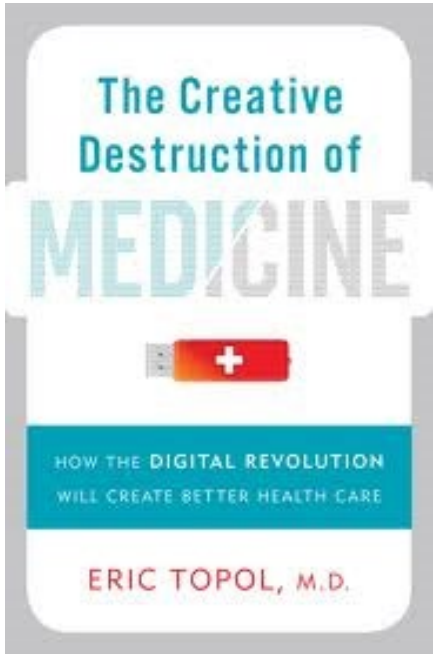
The Creative Destruction of Medicine





Adapted from Fig 1: The transformation of medicine today to new individualised medicine enabled by digitising humans. Eric Topol (2012)

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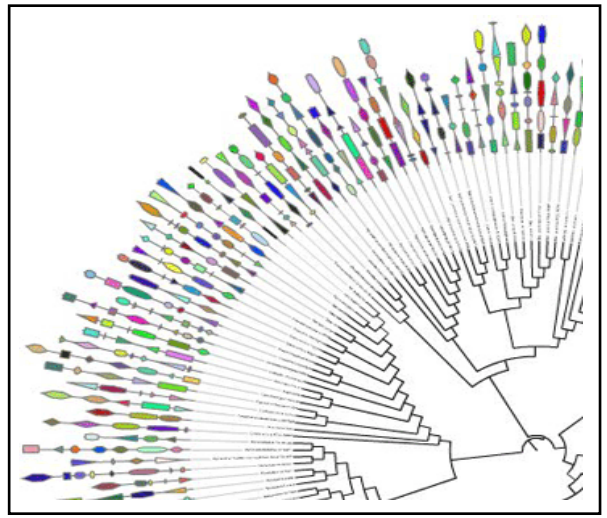
From Sewerage to Bioinformatics



19 C Sanitary Engineer

20 C Social Engineer

21 C Information Engineer

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PHG Foundation 2007

PHG Foundation

A policy think tank focused on genomics and related technologies and how they can be used for better, more personalised healthcare

Active since 1997

A linked exempt charity of the University of Cambridge since 2018

Funded by philanthropy, grants, and commissioned work

At the heart of the Cambridge biomedical hub

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Our Vision

A society that makes the best possible use of advances in life sciences to promote health and improve the prevention and treatment of disease by making responsible changes in health policy and the organisation of health systems.

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What We Do

We help policy makers understand how new technologies could improve healthcare, and the actions needed to put innovations into practice

Provide expert assessment of scientific basis, clinical relevance and legal, regulatory and social impacts

Bring together stakeholders to explore issues, build consensus and create robust health policy solutions

Look ahead to a future where bioscience and technology enable more effective and personalised prediction, prevention and treatment of disease

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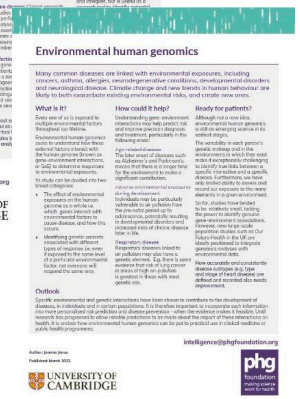
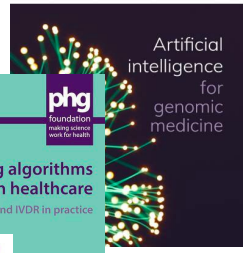
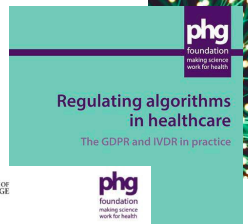
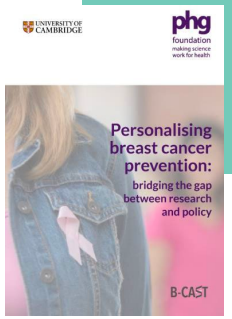
One side explainers

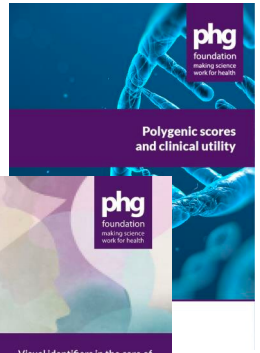


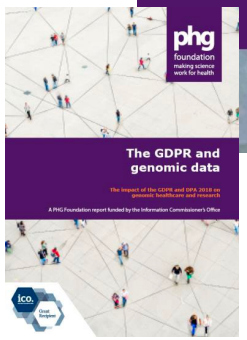






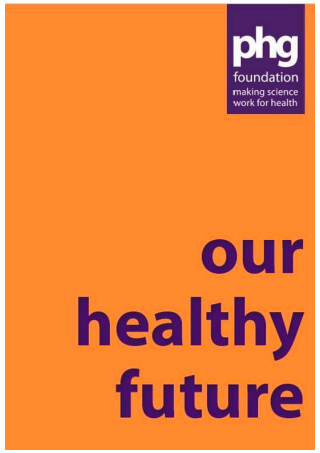
Reports

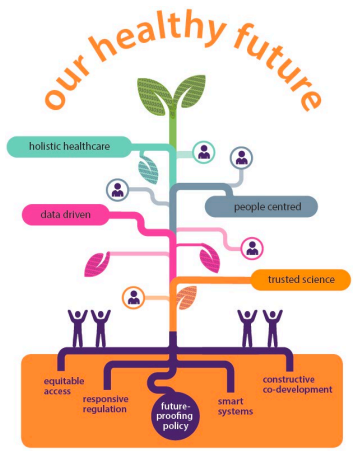




- Blogs
- Consultations
- Working Groups
- Lectures
- Committees
- Discussion dinners
- Meetings

Policy Briefing Notes





- Impact of **new & emerging science and technology** on the future of healthcare
- Implications and issues for **policy & public**

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making science work for health

Where next?

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