

Centre for
Personalised
Medicine

Ethical and Public Health Implications of Genomic and Personalised Medicine

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The Wellcome Trust Centre
for
Human Genetics



Ethical and Public Health Implications of Genomic and Personalised Medicine

- What is 'personalised medicine' and what is in a name?
- Public Health Genomics
- Ethical Implications of Genomic Medicine
 - Resource allocation
 - Health inequalities
 - The nature of evidence
 - Genomic medicine and big data

What is 'personalised medicine'
and what is in a name?

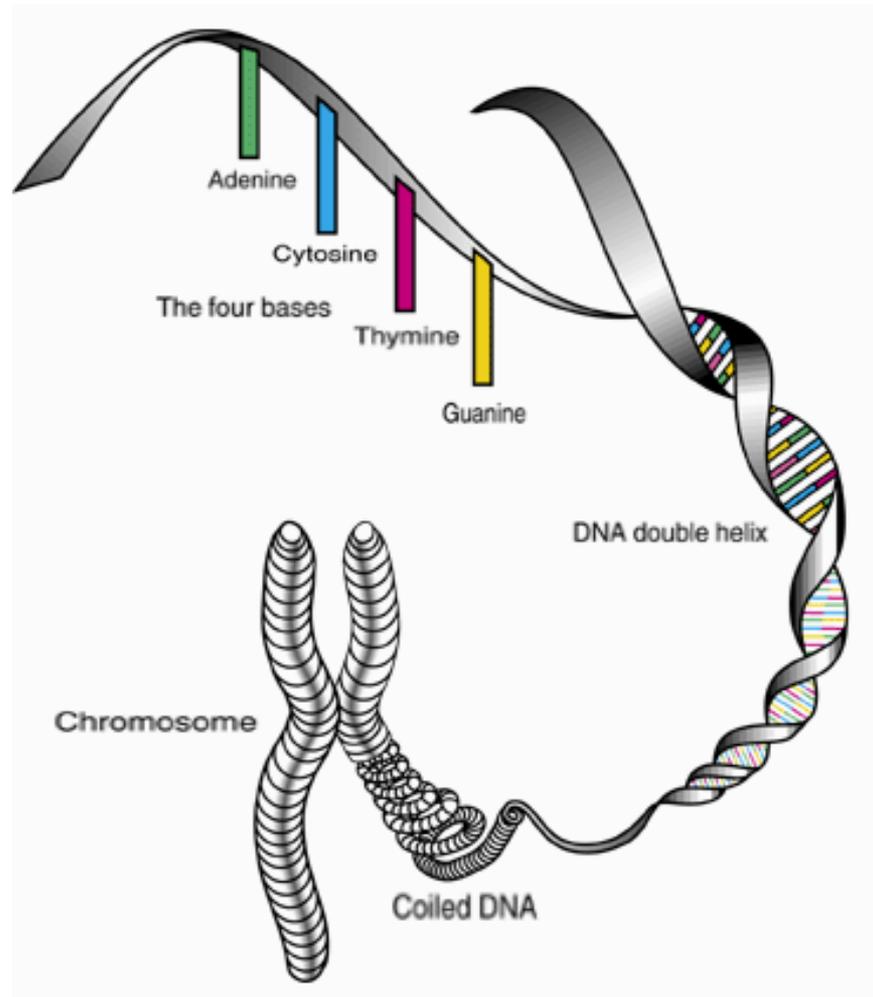
Medical Practice

- Defining nature of human variation
- Application in
 - Risk assessment
 - Diagnosis
 - Prognosis
 - Treatment
- Individuals and populations



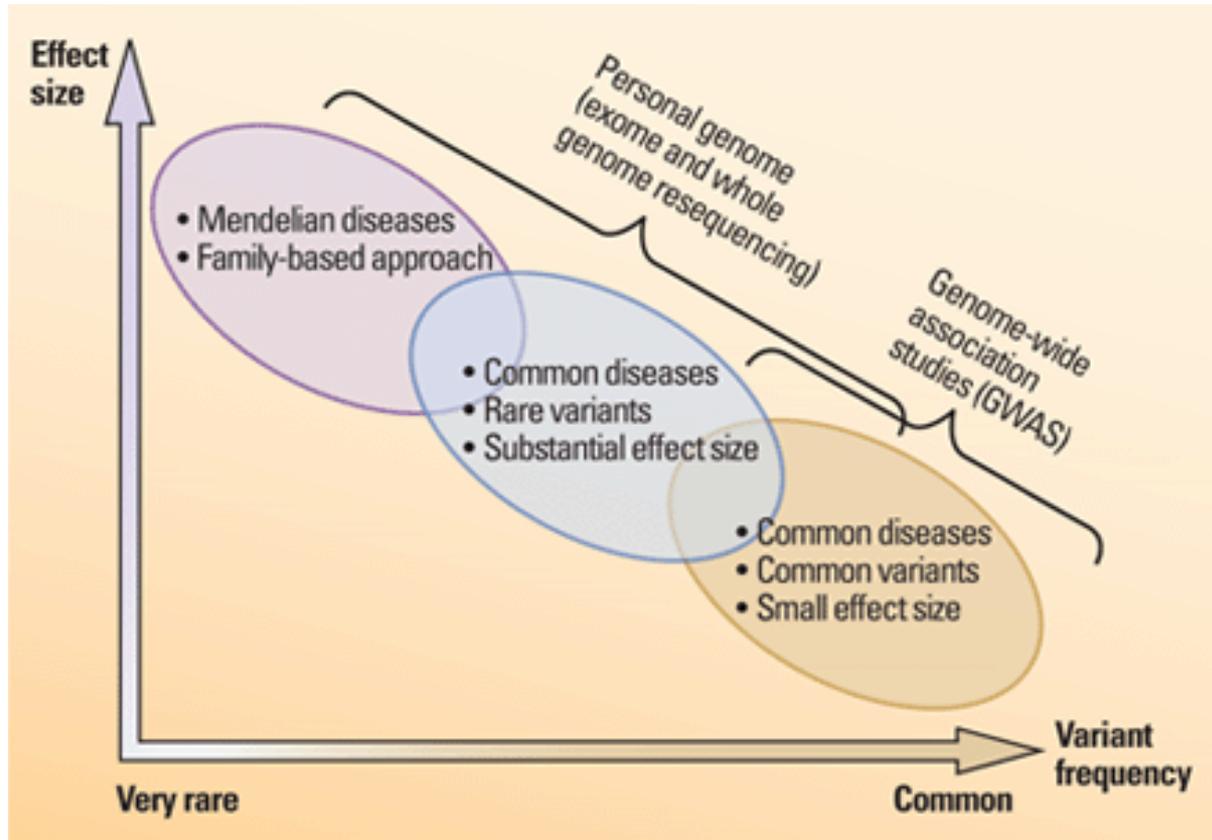
Nature, February 2001

Genomics



Source: Jr. <http://www.kerchner.com/books>

Human Variation

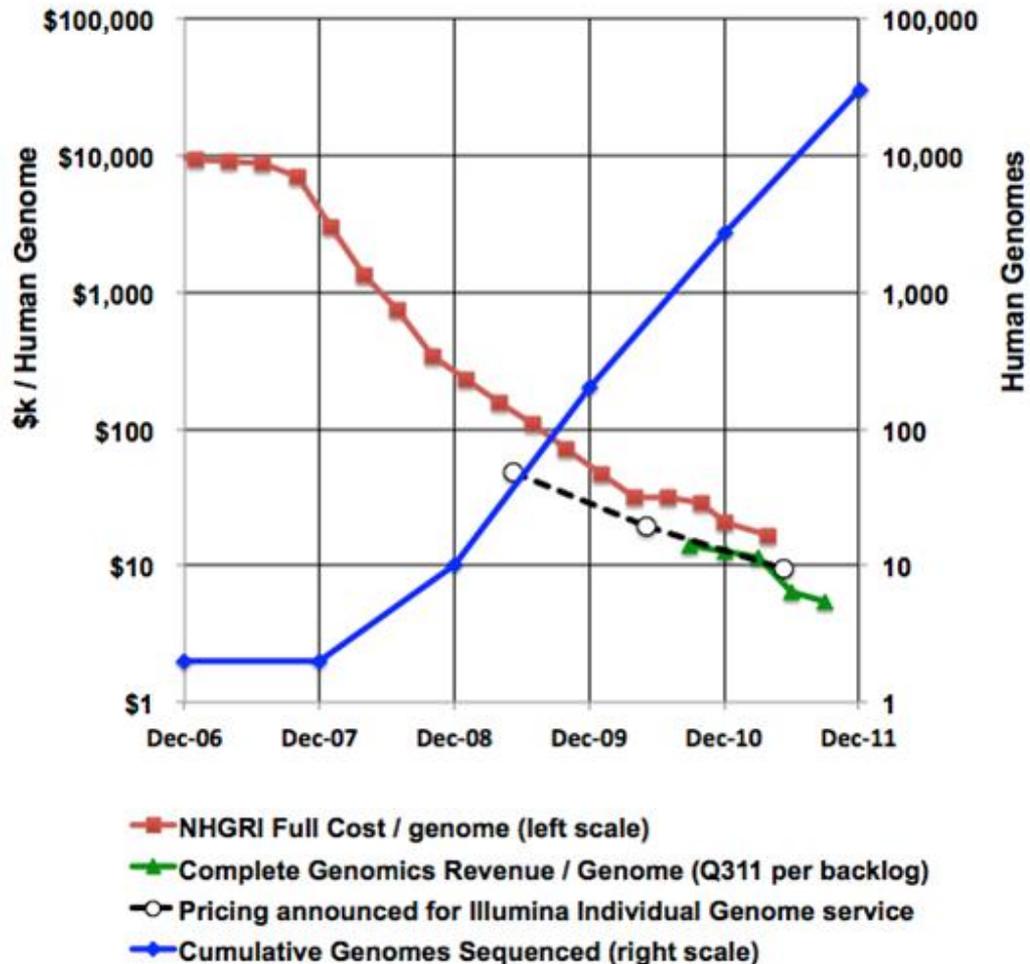


Kaiser, J. *Science* 23 November 2012: Vol. 338 no. 6110 pp. 1016-1017

CREDIT: ADAPTED FROM S. TSUJI ET AL., *HUM. MOL. GENET.* **19** (15 APRIL 2010)

Technology Driving Change

Rapidly Decreasing Sequencing Costs Drive Exponential Growth in Genomes Sequenced





www.illumina.com

Current top-end DNA sequencing machines can each sequence a human genome in 24 hours.

Costs for sequencing an individual's genome are now around £2,500. They should drop below £1,000 within a few years.

What is personalised medicine?

- 2003 250 peer reviewed articles published
- 2012 nearly 2500 peer reviewed articles annually
- What does it mean and does it matter?

What's in a name?

- First use of the term was in reference to pharmacogenomics
- Three components
 1. Evolution of medicine incorporating new scientific knowledge
 2. Distinct, holistic form of healthcare centered around the individual needs of the patient
 3. Treatment targeted at stratified subgroups of patients

What's in a name?

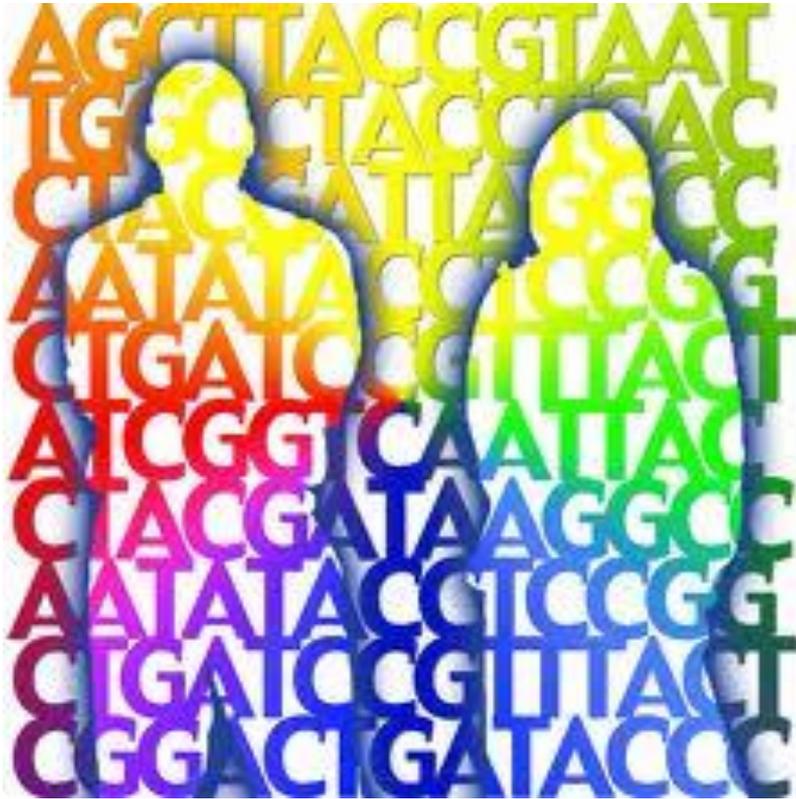
- Two versions of personalised medicine
 - The use of up to date science and technology to define disease at the molecular level accounting for the unique biological need of a person
 - A new form of holistic healthcare centred on an individual patient's preferences
- “Orchestrating labels” – involvement, partnership and now personalised
- Personalised medicine vs personalised healthcare

What is personalised medicine?

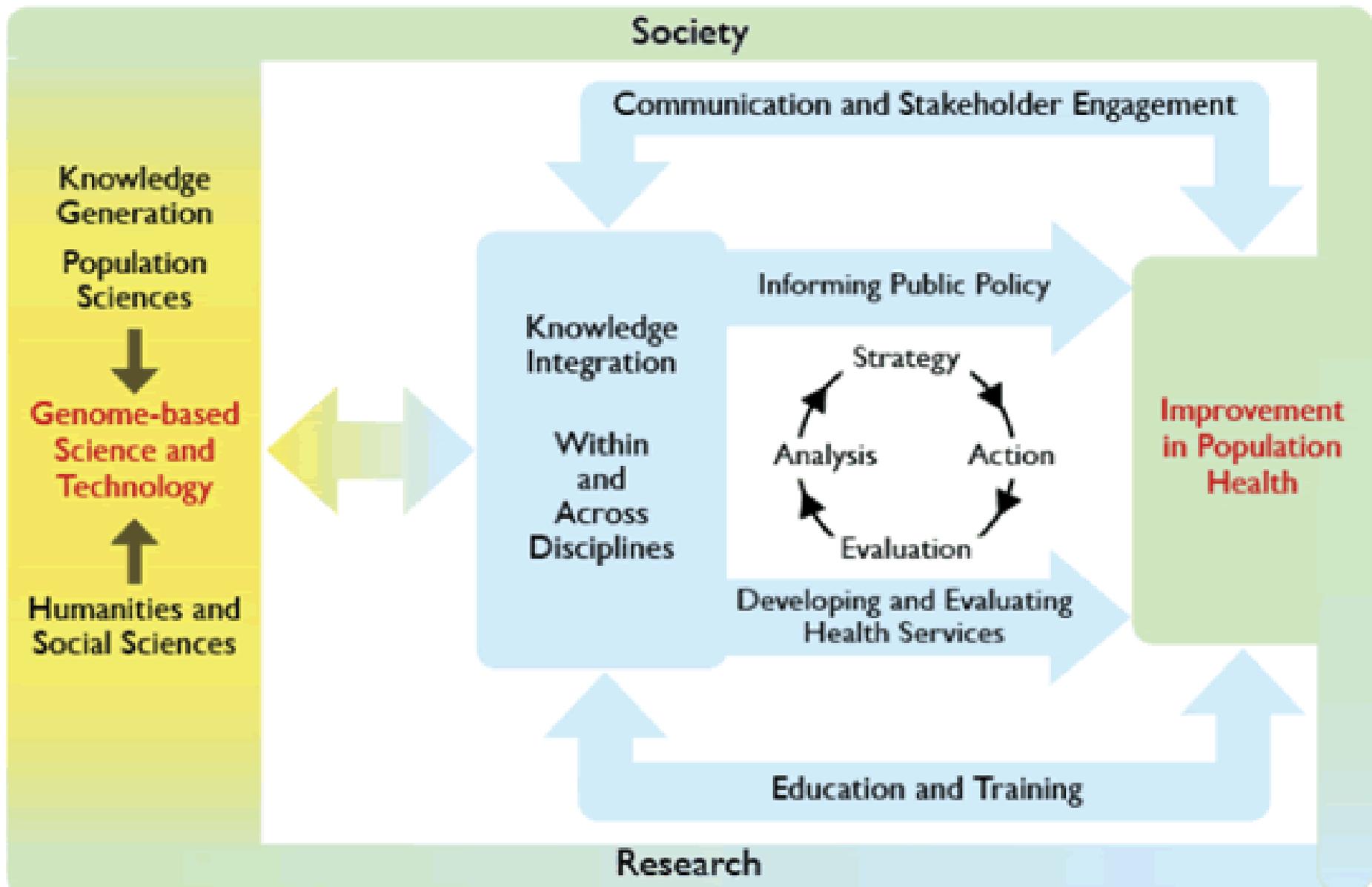
- Medical care incorporating new and up-to-date science and technology to define disease at a molecular level accounting for the unique biological make-up of a person

What is public health genomics?

Public Health Genomics



- **The responsible and effective translation of genome-based knowledge for the benefit of population health. (Bellagio workshop, April 2005)**



Faculty of Public Health

The science and art of promoting and protecting health and well-being, preventing ill-health and prolonging life through the organised efforts of society.

- Health Improvement
- Health Protection
- Healthcare Public Health (improving services)

Healthcare system



What are the challenges.....?

Challenges

- Inputs
 - Equity of access
 - Education and training
 - Initial costs
 - Consent processes
 - Clinical validity and utility
- Process
 - Standardisation of testing and clinical service
 - Computing capacity – data analysis and storage
- Output
 - Delivery
 - Acceptability
 - Ethics

Genomic Medicine in Practice - Challenges

- Translation
 - Basic science
 - Genotype/phenotype
 - Uncertain significance
 - Data challenges
 - Study design
 - Research/clinical interface
- Implementation
 - Workforce education
 - Data collection/storage
 - Service delivery
 - Service financing
 - Cultural change
 - Financing
 - Regulation
 - Licensing

Ethical Implications of Genomic Medicine

- Resource Allocation
- Health Inequalities
- The Nature of Evidence
- Genomic Medicine and Big Data

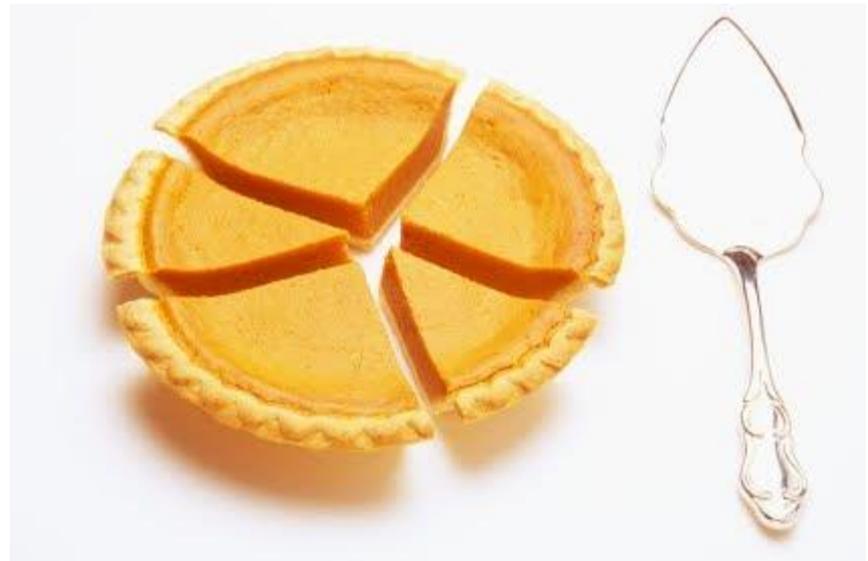
Genomic Medicine and Resource Allocation



Starting point

Rationing: The healthcare budget is limited and decisions need to be made about what gets funded

This is not a matter
of autonomy or
patient choice



Just Allocation of Resources

What should be the general ethical principles for the allocation of healthcare resources?

- General answers might include:
 - Cost
 - Opportunity costs
 - Clinical effectiveness
 - Benefit
 - Cost effectiveness
 - Need / Ability to Benefit

Just Allocation of Resources

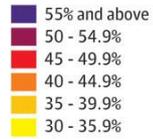
- Broadly fall into two categories ...
- Equality of Outcome
 - Funding is based on treatments being equally effective or cost-effective
 - Best use of resources
- Equality of Persons
 - Individuals are treated equally as individuals
 - Required resources do not count against an individual
 - Equal opportunity at the best health

Issues in Practice

1. Evidence and reasons
2. Cost-effectiveness
3. Age
4. Self-inflicted Illness
5. The boundaries of health

England's health inequality mapped

Local authorities by % of children not achieving a good level of development

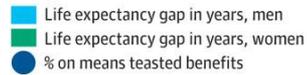


Greater London

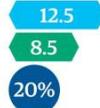
Westminster 53.4%



Life expectancy gap shows the difference between the richest and poorest in each council



Redcar & Cleveland 49.0%



Tower Hamlets 54.4%



Wirral 42.1%



Solihull 30.7%

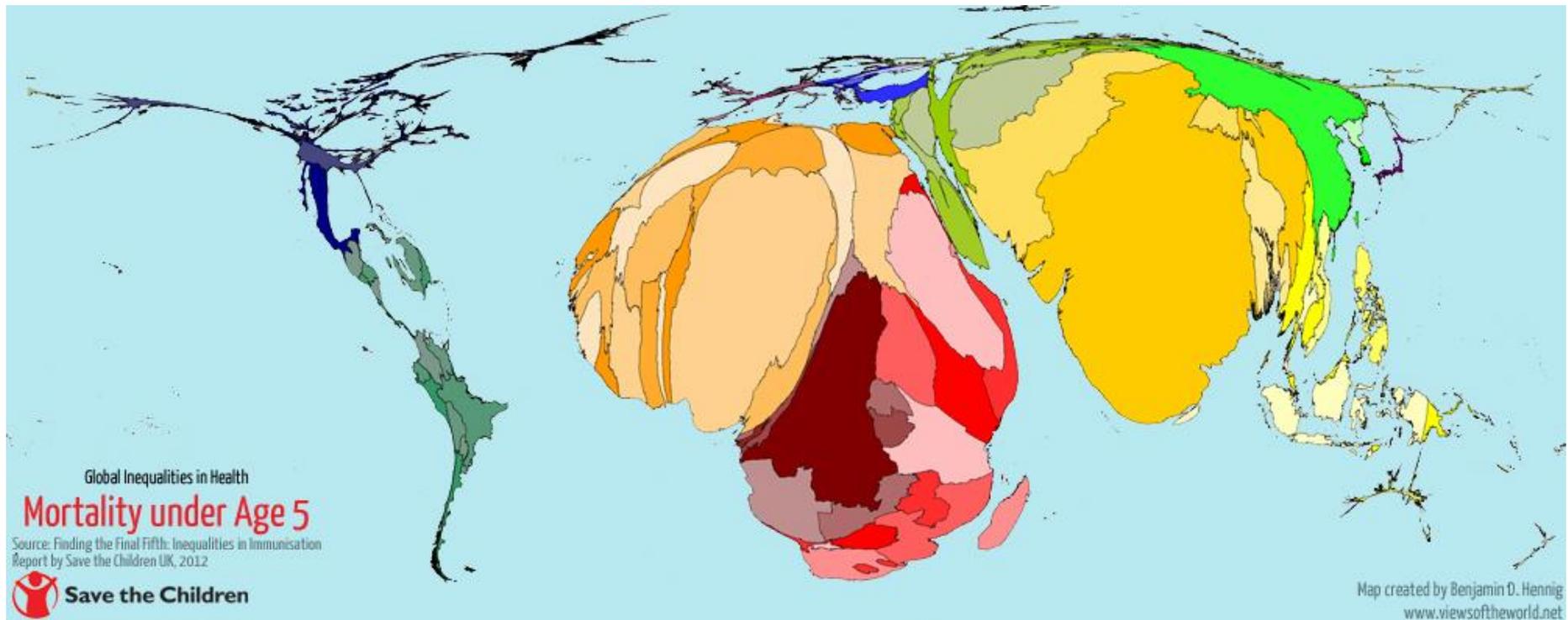


Cornwall 44.2%



Health Inequalities

Health Inequalities



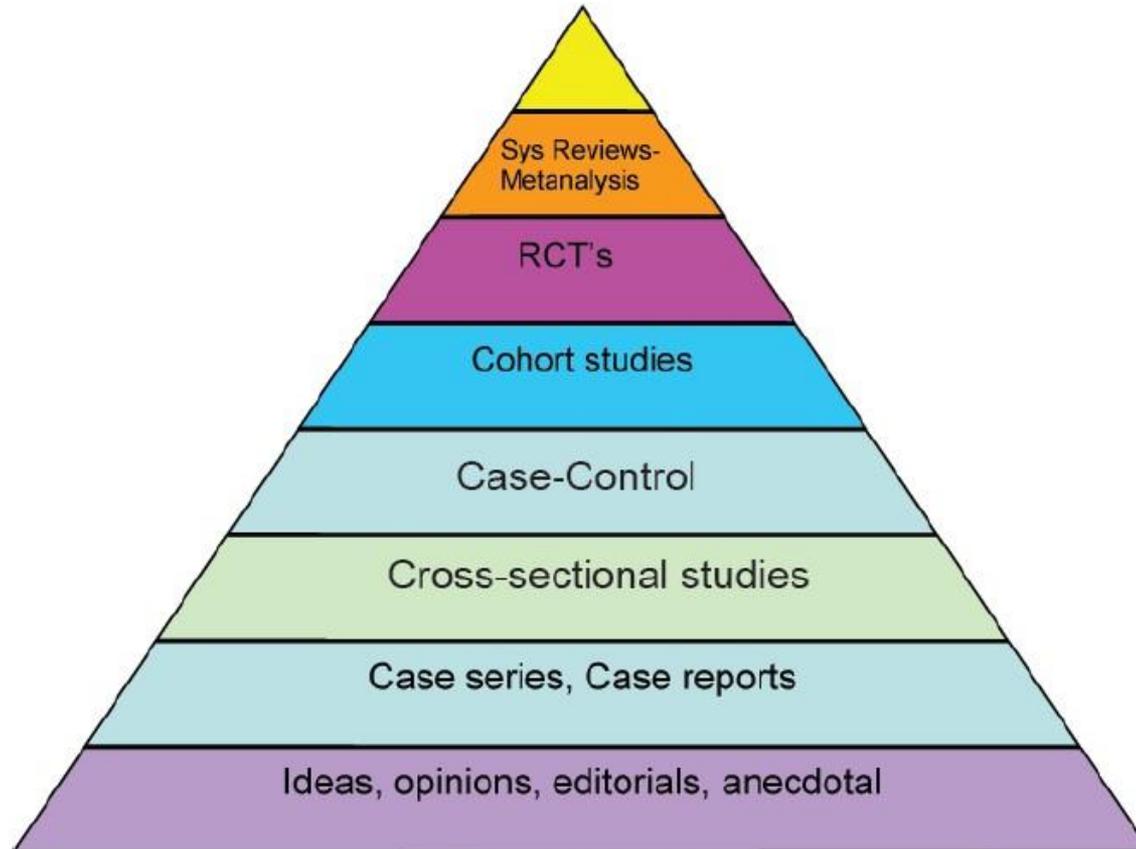
Genomic Medicine and Health Inequalities

- Will genomic medicine increase or decrease health inequalities
 - Within countries
 - Between countries
- Is there a moral obligation to implement advances in biomedical science within healthcare with the aim of reducing health inequalities?

Genomic Medicine and Health Inequalities

- Considerations
 - Personal responsibility
 - Educational inequalities
 - Models of delivery and effect on health inequalities
 - Genomic medicine in screening and health promotion
 - Resource allocation in genomic medicine as a method to decrease health inequalities
 - Stratification vs discrimination

Genomic Medicine and the Nature of Evidence



Genomic Medicine and the Nature of Evidence

- EBM developed in medical education
- PM developed in clinical pharmacology
- Evaluation of interventions vs discovery and explanation
- Public health vs individual health
- Empirical observation vs mechanistic disease models
- The average patient vs outliers

Genomic Medicine and Big Data

- Are there inherent differences between forms of data; genetic data, health data and social data?
- Will increasing use of big data in healthcare reduce these differences and 'normalise' healthcare and genetic data?
- What is the value or role of family history in an era of big data?
- Is there an obligation to collect family history information to contextualise big data sets?

Genomic Medicine and Big Data

- Considerations
 - Trust
 - Data governance and data ownership
 - Electronic patient records
 - Data linkage
 - Patient privacy
 - Individual consent as a barrier to progress
 - Institutionally vs centrally held data

Not to mention.....

- Direct to consumer testing
- Variants of uncertain significance
- Consent
- Genetic testing in children
- Genetic data and insurance
- Problems of hype
- What about those patients which don't have biomarkers that offer targeted treatment options?

The future



The future



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Cambridge

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