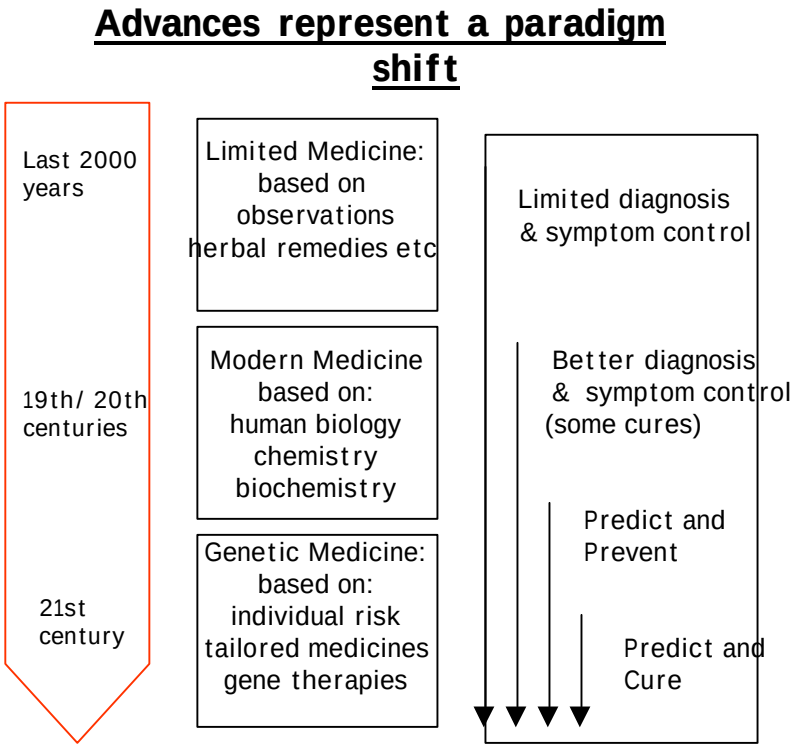
POLICY IMPLICATIONS OF THE GENETICS WHITE PAPER

INNOVATIONS IN GENETICS
TRANSLATION INTO SERVICE DELIVERY

PETER GREENAWAY
Research and Development Directorate
Department of Health

LIKELY ADVANCES REPRESENT A STEP-CHANGE



- Genetic medicine could enable the prediction, prevention and cure of disease as well as the diagnosis and management of symptoms
- This could lead to changes in the way in which we classify illness e.g. Type I diabetes being subdivided on the basis of new knowledge of genetic cause
- There are likely to be significant advances over the next 5-10 years, although the exact size and timing of these remains unclear

RISK TESTING AND DIAGNOSIS FOR GENETIC DISORDERS CAN BE DIVIDED INTO THREE GROUPS BASED ON THE BALANCE BETWEEN GENETIC AND ENVIRONMENTAL FACTORS

Single-gene disorders are highly penetrant - if you have the gene mutation you are very likely (90 -100%) to get the disease in your lifetime, e.g. Cystic Fibrosis, Huntingdon's.
Testing is often used to diagnose the condition

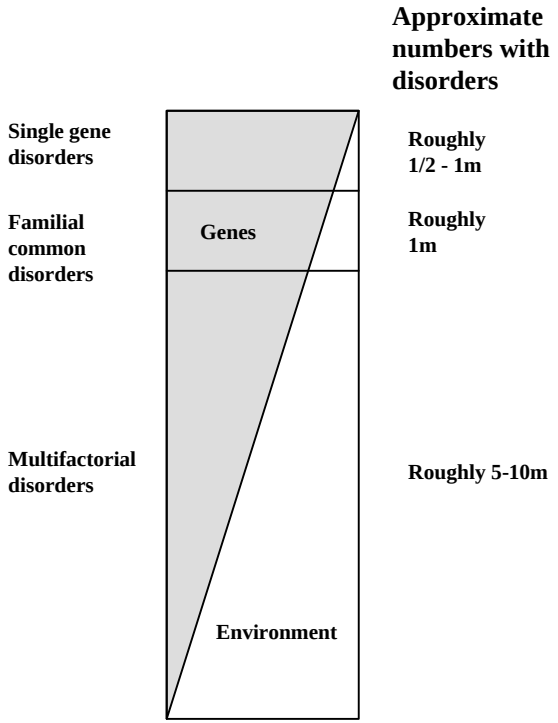
Familial subsets of common disorders are a genetic sub-set of many common disorders such as cancer. They are relatively highly penetrant (e.g. mutations in the BRCA1 gene give a lifetime risk of breast cancer of 35 - 80%)

Tests are used to predict future condition

Multifactorial common disorders are more influenced by environmental factors. Having a gene mutation gives an elevated lifetime risk of developing the condition, but is much less predictive than for the other two groups. Conditions include various cancers, CHD, diabetes, depression.
Tests could be used to determine susceptibility to condition

Tests for single gene disorders/ familial subsets already exist and there are likely to be more in the immediate future.
Tests for multifactorial disorders are expected later

The relative importance of genetic make-up and environmental factors varies between groups(1)



(1): Numbers based on Royal College of Physicians Report 1991 and expert opinion (John Bell of the John Radcliffe Hospital, Oxford).

SINGLE-GENE DISORDERS AFFECT ROUGHLY 700,000 PEOPLE AND COST HEALTH AND SOCIAL SERVICES ABOUT £2bn PER ANNUM

About 6000 single gene disorders identified to date. Affect about 700,000 people[1] (but estimates vary between 1-5% of the population)

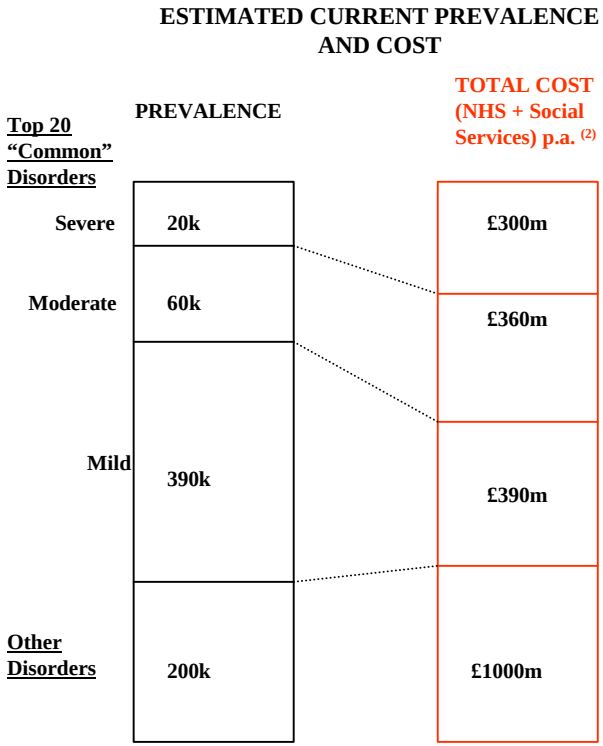
- top twenty "common" disorders account for about half a million people
- impact on health ranges from mild, to moderate, to severe
- total cost to health and social services approximately £2bn p.a.[2]

Currently tests available for most "common" disorders, but not for the many rarer ones

- but many disorders diagnosed using non-genetic-based diagnostic tests, e.g. Phenylketonuria

Over next 5-10 years likely to identify some new single-gene disorders

- and develop genetic tests for more rare single gene disorders (where the genes are already known)



1. Based on Royal College of Physicians: Working Group of the Clinical Genetics Committee (1991)
2. Department of Health: internal analysis

ABOUT A MILLION PEOPLE ARE AT PARTICULARLY HIGH RISK OF SOME COMMON DISORDERS[1]

Many of the common disorders (cancer, CHD etc) have a subgroup of people at particularly high genetic risk (e.g. 35 - 80% lifetime risk)

Unclear how many will fall into this group - but estimated to be about one million people or 10% of those with common disorders (CHD, cancer, diabetes etc)[1]

Best understood at present are BRCA I & II for breast cancer and genes connected with familial colon cancer

- NHS offers testing for both conditions in some places (although significant potential to improve services for both conditions)
- BRCA also offered by Rosgen/Myriad - private US based company offering test with greater sensitivity than that on offer in NHS, but at much higher price
  - could be used on those patients with more difficult to detect variations

Tests shortly coming on line for other conditions

- tests for risk of skin, colon and bladder cancers may be commercially available in next few years
- but predictive power of new tests remains to be assessed

Testing offers the potential to target monitoring and preventive regimes at those at high risk and to reduce level of intervention for those at low risk

1: John Bell (John Radcliffe Infirmary)

THE GENETICS WHITE PAPER

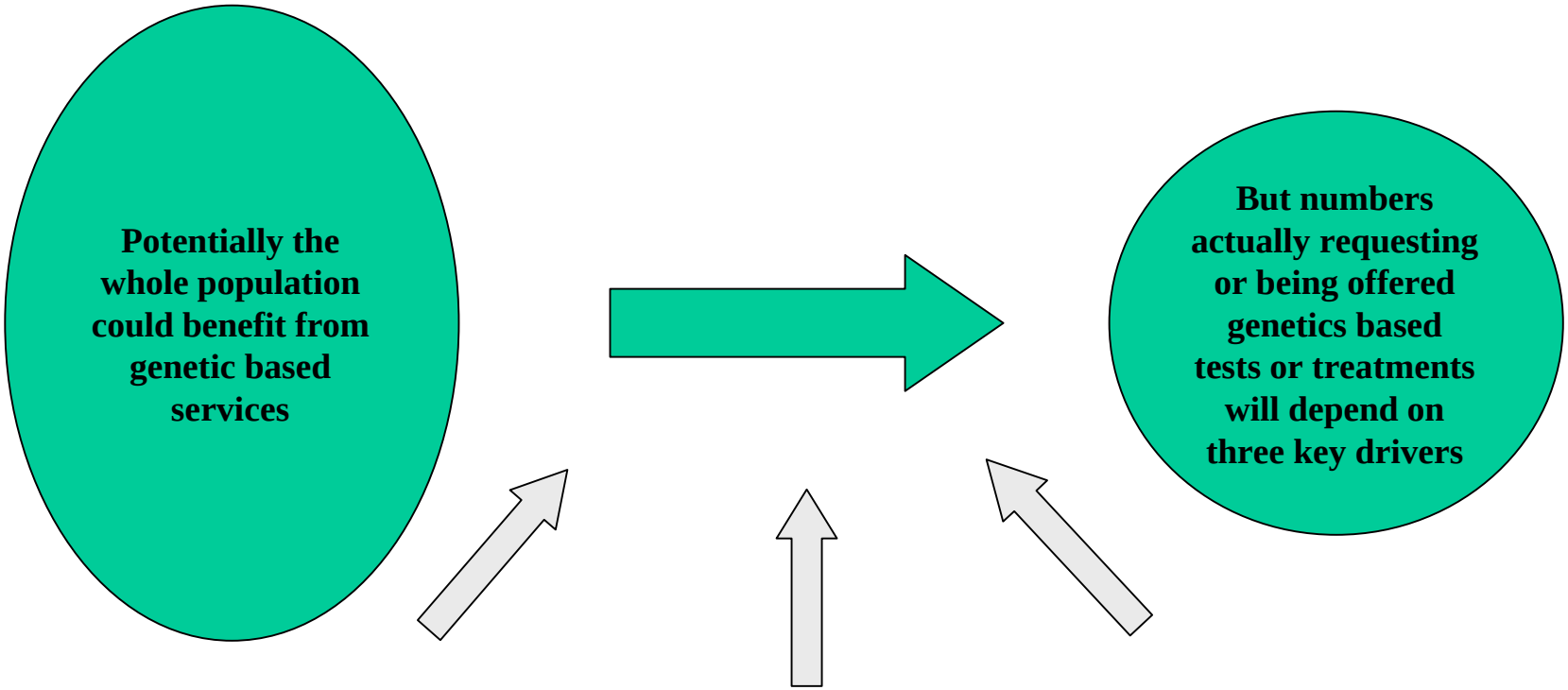
OUR INHERITANCE, OUR FUTURE
Realising the potential of genetics in the NHS

- A vision of how people will benefit in future from advances in genetics
- Strategies for realising these benefits
- Safeguards to prevent inappropriate use

KEY MESSAGES

- Genetics offers enormous potential to improve health and healthcare
- NHS should lead the world in harnessing these potential benefits
- Government will continue to support the current research initiatives underway in the UK, both in the public and private sectors
- The UK will maintain its role at the cutting edge of genetics research and development

DEMAND DRIVERS FOR THE PROVISION OF GENETICS BASED SERVICES



1. Technology
- affecting pace and nature of service developments

2. Policy/ Supply Of Services
- availability of services
- marketing of services
- cost (free, payment)
- regulation/legislation

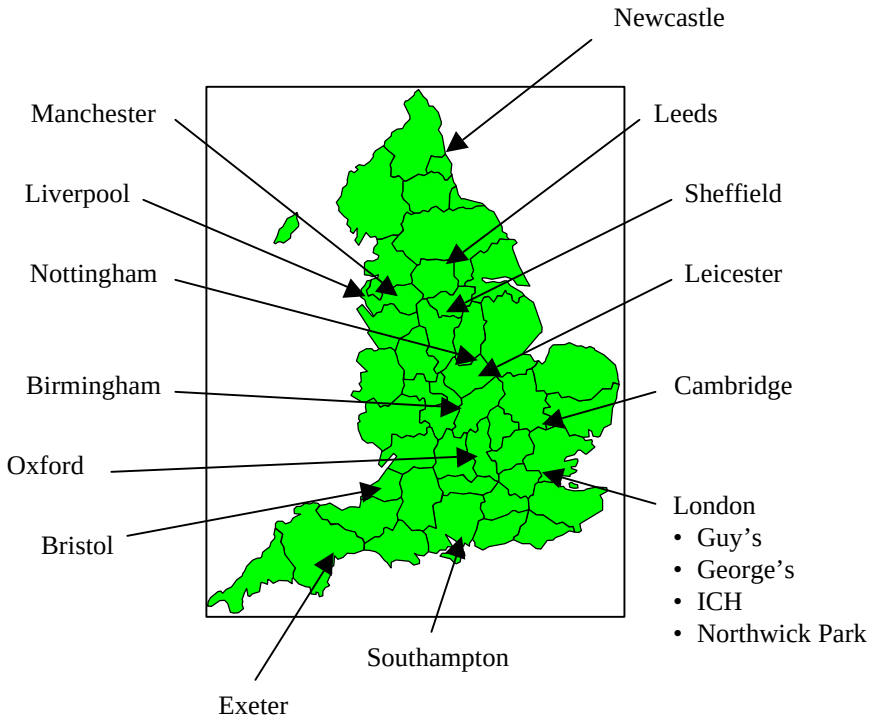
3. Public Reaction to genetics-based technology

£50 million over 3 years

- Upgrading genetics laboratories
- More scientists, counsellors, consultants
- Initiatives to bring genetic-based healthcare into mainstream services
- New legislation banning DNA theft
- NHS Genetic Education and Development Centre
- Consideration of screening at birth
- Research programmes

BULK OF CURRENT GENETICS SERVICES ARE PROVIDED BY A SPECIALISED REGIONAL GENETICS SERVICE

Regional Testing Centres



- Management and testing of most single gene disorders currently based in regional genetics services
- Also do some testing for familial subset of common disorders e.g. BRCA test for breast cancer
- 17 main genetics centres and many more smaller units. In addition some genetic testing is also done in other specialities e.g. haematology and obstetrics
- In total about 90 consultant geneticists, 120 nurse counsellors, 200 lab scientists
- Provide integrated testing, counselling and diagnostics services
- Work on hub-spoke basis, undertaking sessions in other peripheral hospitals/ clinics

IMPROVEMENTS TO CURRENT SERVICES AND THE INTRODUCTION OF SERVICE IMPROVEMENTS

Need to make significant improvements to current services to ensure they can deal with current demand

- develop education and training in primary care
- reduce variation in level and quality of specialist services
- ensure clear referral protocols and guidelines for care are in place throughout system

Need to assess potential for widespread introduction of technologies currently available, for example pharmaco-genetic testing, risk testing, tumour typing, DNA microbial analysis

And we need to establish the infrastructure and flexibility to deal with future advances

- consider potential to invest more in key groups who will be able to manage/ train greater numbers quickly eg genetic consultants and counsellors
- undertake further work into future service models
- put in place management structures to ensure service can maximise potential benefits of future developments

STRENGTHENING SPECIALIST SERVICES

- Upgrading NHS genetics laboratory facilities
- Working towards CPA accreditation
- Modernising services to reduce reporting times (3 days for prenatal diagnosis;2 weeks where the likely mutation is known; 8 weeks for index cases)
- Introduction of more screening schemes - hypercholesterolaemia, sickle cell disease
- Improving IT services for better handling of work and communications

THE WORKFORCE

- 50 Genetic Counsellors
- 90 Grade A trainees Clinical Scientists
- 10 full-time trainers
- 10 specialist GPs
- Visiting fellowship schemes
- Genetic Education and Development Centre

NHS GENETICS EDUCATION AND DEVELOPMENT CENTRE

- Identifying learning needs and skill requirements for different staff groups - developing curricula
- Developing skills and competency frameworks
- Producing learning support materials, running seminars, workshops etc
- Raising the profile of genetics

IMPROVED COMMUNICATION OF GENETIC KNOWLEDGE

- Genetics Knowledge Parks
- Decision support systems
- Data Sheets
- NHS Direct
- National Electronic Library for Health

ROLES FOR PRIMARY CARE IN GENETICS

- Managing patients' concerns and expectations
- Identifying genetic conditions
- Assessing and managing risk
- Screening and testing
- Disease prevention programmes
- Providing and co-ordinating long term care
- Gatekeeping to specialist care

RESEARCH

- £1.5 million for health services research
- £0.5 million for piloting near patient testing
- £3.0 million for gene therapy for single gene disorders (including safety)
- £2.5 million for gene therapy for CF
- £4.0 million for gene therapy vectors
- £4.0 million for pharmacogenetics
- Chair in pharmacogenetics plus staff

GENE THERAPY RESEARCH

- Single gene disorders (none specified)
- Phase II trials of gene therapy procedures
- Translational research
- Safety of gene therapy vectors and interventions (GTAC/CSM priorities)

- Gene therapy for cystic fibrosis - separate funding arrangements

PHARMACOGENETICS RESEARCH

- Make use of medicines more effective
- Reduce adverse side reactions
- Increase the number of medicines available
- Focus on commonly used or expensive drugs
- Increase research capacity (Chair + Department)

ACHIEVING THE VISION

- Strengthening specialist genetics services
- Improve public perception/understanding of genetics
- Develop an appropriate evidence base

STRENGTHEN SPECIALIST GENETICS SERVICES

- Modernising/reconfiguring service provision
- Capturing and analysing genetic information
- Developing the workforce

IMPROVE PUBLIC PERCEPTION/UNDERSTANDING OF GENETICS

- Perception/understanding
- Risk management
- Health promotion

DEVELOP AN APPROPRIATE EVIDENCE BASE

- Genetics based health services
- Gene Therapy
- Pharmacogenetics