

Existing oversight of Direct to Consumer genetic testing in the UK

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UK reports on genetics and genetic testing



Our Inheritance Our Future

Realising the potential
in the NHS

Presented to Parliament
by Command of Her Majesty

June 2003

Ensuring the effective use of
genetic tests and services

Human Genetics Commission

March 2004



**A common framework of principles for direct to
consumer genetic testing services**

Principles and Consultation Questions

Consultation period

The consultation period will run from
1 September 2009 to 30 November 2009.



HOUSE OF LORDS

Science and Technology Committee

2nd Report of Session 2008–09

Genomic Medicine

Volume I: Report



HL Paper 107-I

Regulatory Authorities

- OFT - Office of Fair Trading
- ASA - Advertising Standards Authority
- OFCOM - Regulator of UK communications industries
- MHRA - Medicines Healthcare Products Regulatory Agency

Advisory Bodies

- UK Genetic Testing Network
- National Institute for Health and Clinical Excellence (NICE)
- Human Genetics Commission (HGC)

Relevant statutes

- In vitro diagnostic medical devices (EU, 1998)
- Human Tissue Act (2004)

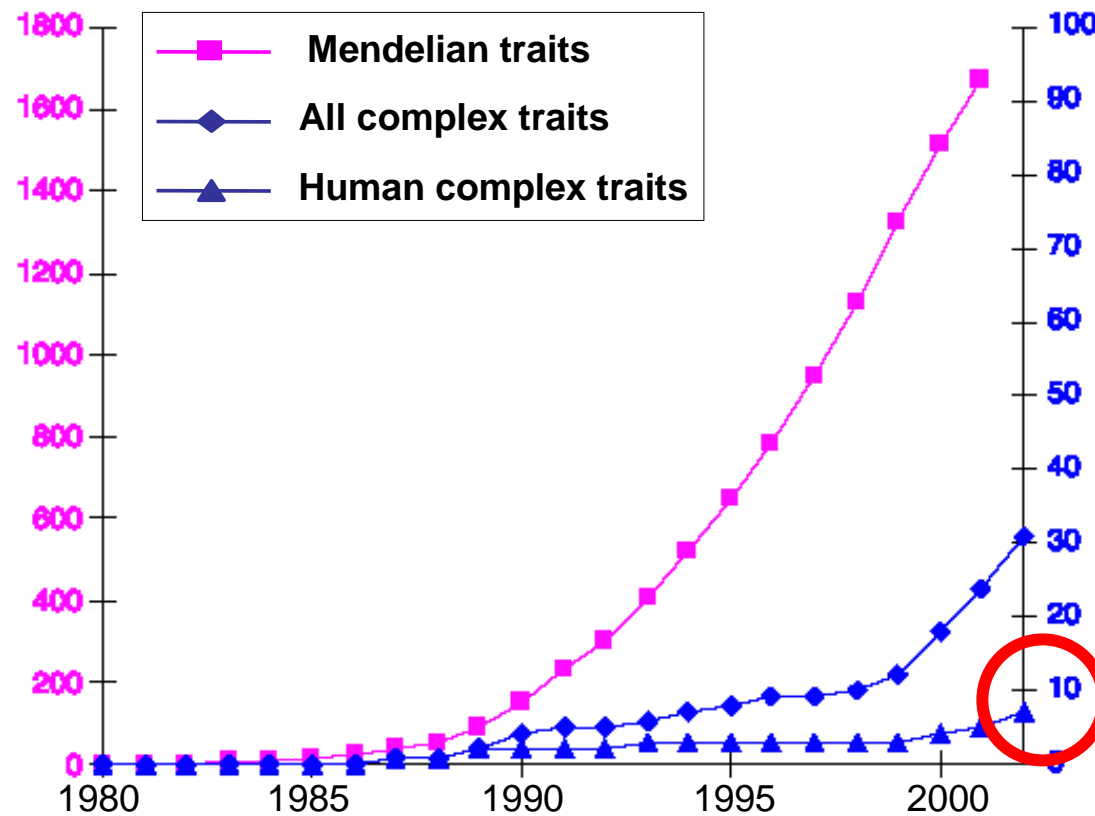
Genetic classification of human disease

- Mendelian
 - Single gene disorders
 - Rare (mostly less than one per thousand)
 - One-to-one relationship between gene and disease
 - Examples: Cystic fibrosis, Huntington's disease, haemophilia
- Genetically complex
 - Multiple genes + environment
 - Common (up to 30% prevalence)
 - Genes not causal – increase disease risk
 - Examples: Obesity, coronary heart disease, cancer

Identification of Genes underlying Mendelian and Complex Traits 1980-2002

Mendelian traits

Complex Traits

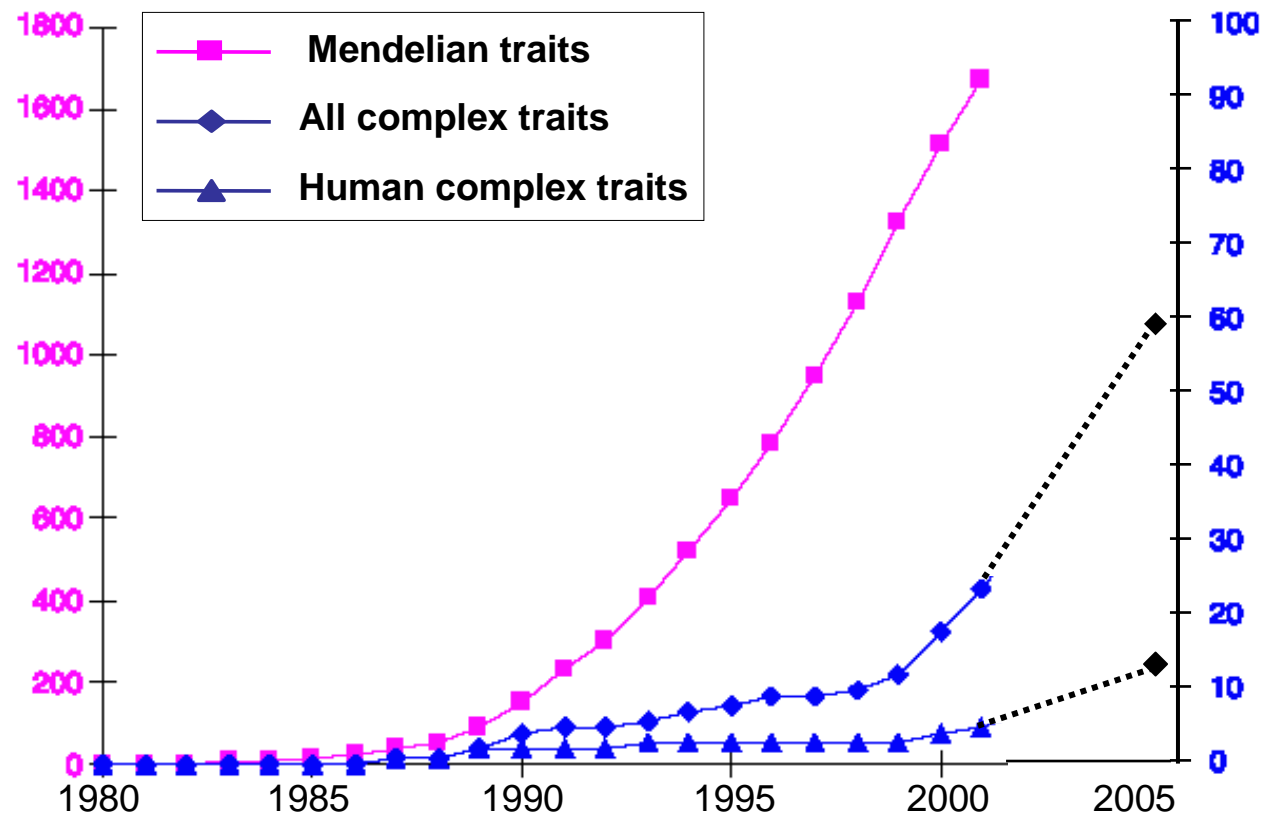


Glazier, Nadeau, Aitman, *Science*, 2002

Identification of Genes underlying Mendelian and Complex Traits 1980-2005

No. of Mendelian traits

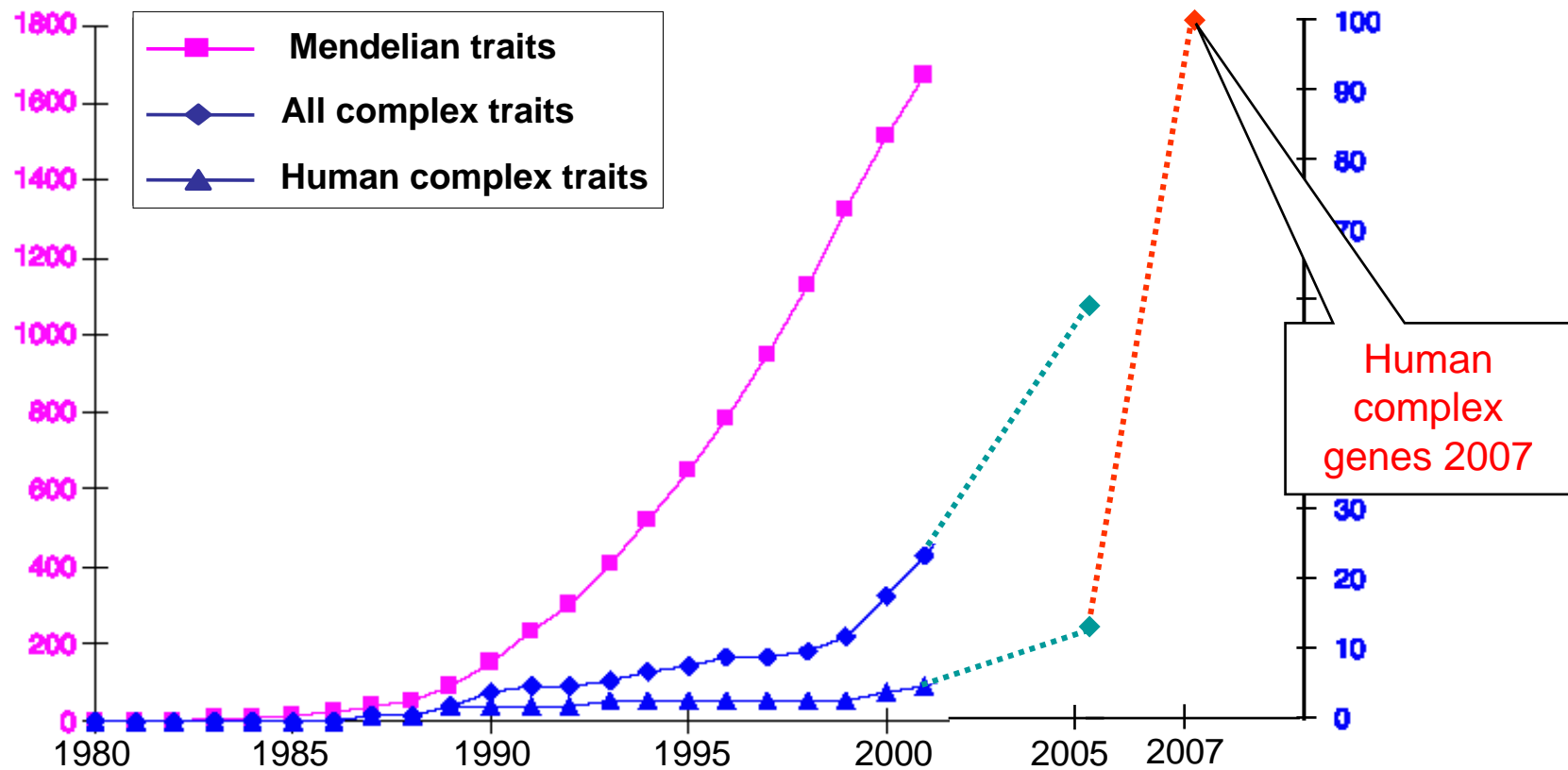
No. of Complex Traits



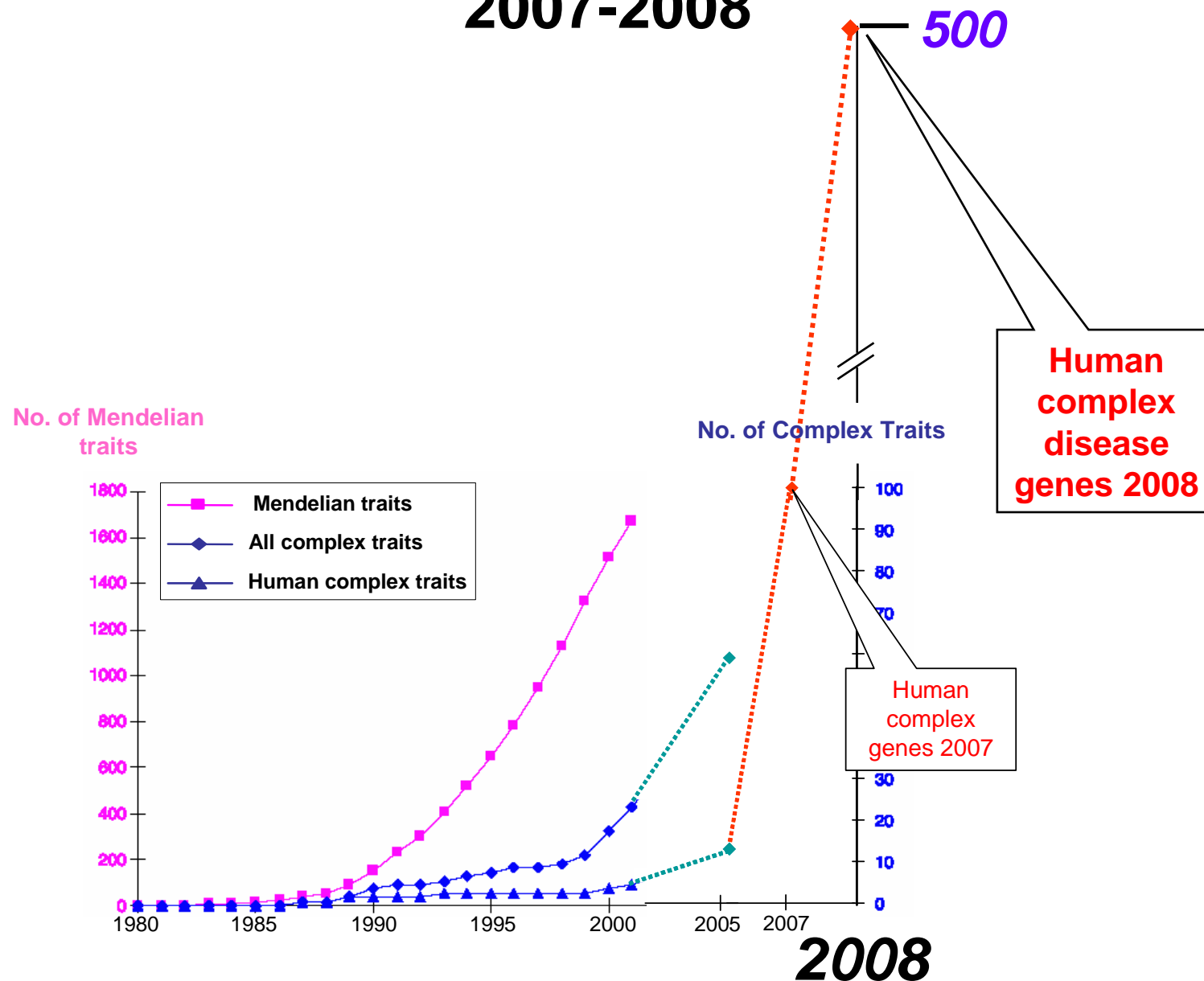
Identification of Genes underlying Mendelian and Complex Traits 1980-2005

No. of Mendelian traits

No. of Complex Traits



Progress in identifying common disease genes - 2007-2008



Advisory bodies on genetic testing

- Monogenic disease – UK Genetic Testing Network
- Common diseases – no formal body



Human
Genetics
Commission

Genes direct

Ensuring the effective oversight of
genetic tests supplied directly
to the public

A report by the
Human Genetics Commission

March 2003

***Genes Direct* report recognised three types of access to genetic tests:**

- Through NHS or private doctors
- Through non-medical intermediaries such as pharmacies of complementary therapists
- Direct to consumer

Despite possible benefits, measures needed to be taken:

- To make sure that only high quality tests are marketed
- To make sure that companies don't misuse the power of modern genetics as a marketing tool
- To avoid possible harms to the consumer



Genes direct

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March 2003

***Genes Direct* recommended:**

- Stricter controls on direct genetic testing, but not a statutory ban
- Well-funded genetic tests through the NHS
- Most genetic tests that provide predictive health information should not be offered as direct genetic tests
- Predictive genetic tests that rely on home testing or home sampling should be discouraged (note risk to children, consent etc.)
- Introduction of a new offence of the misuse of genetic information

Human Tissue Act 2004

“DNA theft”



Human Tissue Act 2004

CHAPTER 30

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PART 1

REMOVAL, STORAGE AND USE OF HUMAN ORGANS AND OTHER TISSUE FOR SCHEDULED PURPOSES

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The Human Tissue Authority

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UK legal concept of “DNA theft”



Human Tissue Act 2004

45 Non-consensual analysis of DNA

- (1) A person commits an offence if—
 - (a) he has any bodily material intending—
 - (i) that any human DNA in the material be analysed without qualifying consent, and
 - (ii) that the results of the analysis be used otherwise than for an excepted purpose,
 - (b) the material is not of a kind excepted under subsection (2), and
 - (c) he does not reasonably believe the material to be of a kind so excepted.

-
- (3) A person guilty of an offence under this section—
 - (a) is liable on summary conviction to a fine not exceeding the statutory maximum;
 - (b) is liable on conviction on indictment—
 - (i) to imprisonment for a term not exceeding 3 years, or
 - (ii) to a fine, or
 - (iii) to both.



Genes direct

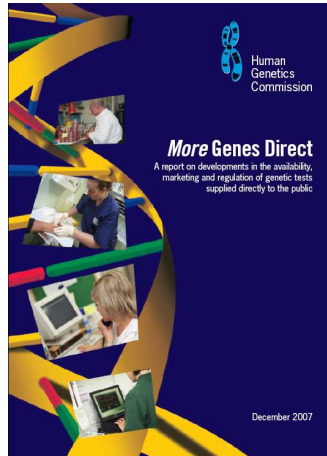
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***Genes Direct* comment on UK regulatory controls:**

- Regulatory approach in EU member states is dependent on implementation of the In Vitro Diagnostics Devices Directive (IVDDD), '98 '02
- In UK, implementation by Medicines Healthcare Products Regulatory Agency
- Uncertainty over status of genetic testing
- Genetic testing aiming to give “lifestyle advice” may not be covered by definitions in IVDDD



***More Genes Direct* updated framework and recommendations of *Genes Direct* and noted:**

- IVDD Directive operates on a risk-based system of regulation in which most genetic tests are classified as “low risk”. Manufacturers are therefore not required to submit a technical file to the National Notified Body (MHRA). Therefore no pre-market assessment of clinical validity or utility. Self-certification sufficient
- None of ASA, MHRA, or OFT had authority to regulate content of internet



***More Genes Direct* recommended:**

- The risk of genetic tests, classified by the IVDD Directive as of “low risk” should be reviewed, with a view to being classified at a higher risk level
- A code of practice should be developed that takes into account the guidelines (on quality assurance) published by OECD and other relevant international standards
- Genetic tests provided overseas via the internet cannot be easily regulated and in order to promote high standards, HGC wishes to liaise with regulators in other countries

HGC SEMINAR, June 30th 2008

“Genetic tests sold direct to the public”

- The seminar included representatives from HGC, UK Department of Health, EC, DCT providers (deCode, Navigenics), consumer groups, representatives of public health and academia
- There was “overwhelming support” for a code of practice on DCT, including support from DCT providers
- Difficulties in national regulation – for an international problem - and absence of an obvious body to take this forward suggested the advantage of developing a “Common Framework of Principles”



Human
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A common framework of principles for direct to consumer genetic testing services

Principles and Consultation Questions

Consultation period

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1 September 2009 to 30 November 2009.



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A common framework of principles for direct to consumer genetic testing services

Summary of Key Principles - 1

DCT genetic testing takes place in an international market that crosses national borders and jurisdictions. To promote consistency, it was decided to initiate a common framework that could have applicability across all jurisdictions

- Test providers should strive to provide a high-quality service that meets customer expectations and safeguards their interests
- Tests for inherited disorders should only be provided with individualised pre- and post-test counselling
- Providers should comply with legislation on advertising



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Summary of Key Principles - 2

- Promotional claims should accurately describe characteristics and limitations of tests, and avoid bias
- Provider should supply easily understood, accurate and adequate information to consumers before obtaining consent for a genetic test, including likely outcomes of the test
- A genetic test should only be carried out after the person concerned has given free and informed consent
- The provider should take reasonable steps to assure themselves that the biological sample was obtained from the person identified as the sample provider



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Summary of Key Principles - 3

- With the exception of paternity tests, genetic tests in respect of children, when according to applicable law, that child does not have the capacity to consent, should normally be deferred until the attainment of such capacity unless other factors indicate that testing during childhood is clinically indicated

Note: Consultation runs 9.1.09 – 11.30.09
<http://www.hgc.gov.uk>



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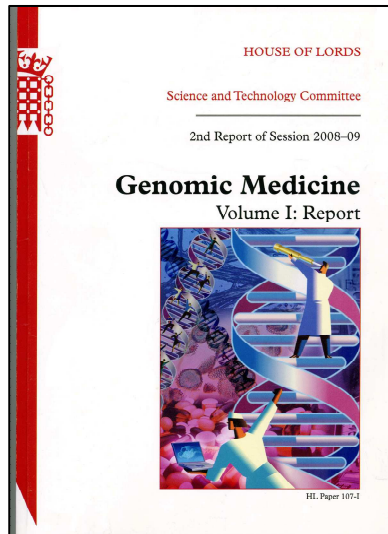
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HL Paper 107-I

**Direct to Consumer Tests
Paragraphs 6.51 – 6.67**

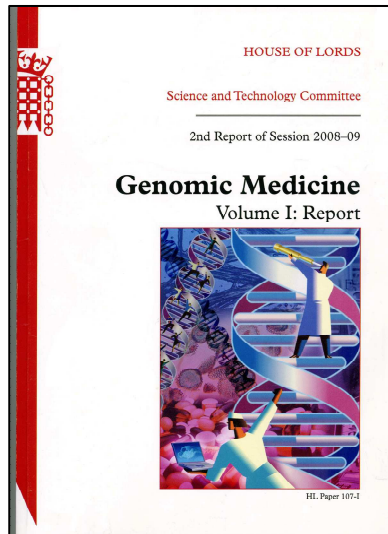


“Witnesses held wide-ranging views about the value of DCTs”

Professor Bobrow (Former Chair in Genetics, University of Cambridge) commented:

“If you look at things like deCODEme and the 23andMe website, a lot of their emphasis is on doing your genome so that you can go and find out whether some chap you have met is your second cousin and other things of that nature. It is scientifically valid, it is medically irrelevant and I think it is very much a question of if you want to blow £1,000 on that, it is your business”.

Dr Ron Zimmern, Executive Director of the PHG Foundation, thought that that companies should not be prevented from selling DCTs and said that he could “see nothing in a free society to suggest that we should stop people from knowing that they have a two per cent higher risk of asthma or a four per cent lower risk of heart disease”. But, he believed that the type of data derived from DCTs was “totally useless information”



On the other hand...

Professor Donnelly (Director, Wellcome Trust Centre for Human Genetics) spoke positively about DCTs:

“DCTs are the first step to a service that would eventually be incorporated into routine clinical practice. There is a possibility ... for people to be able to say, ‘there’s a whole range of diseases, I know from my genetics that for two or three diseases I am at particular high risk, let me focus on the lifestyle changes which will make a difference to those’.

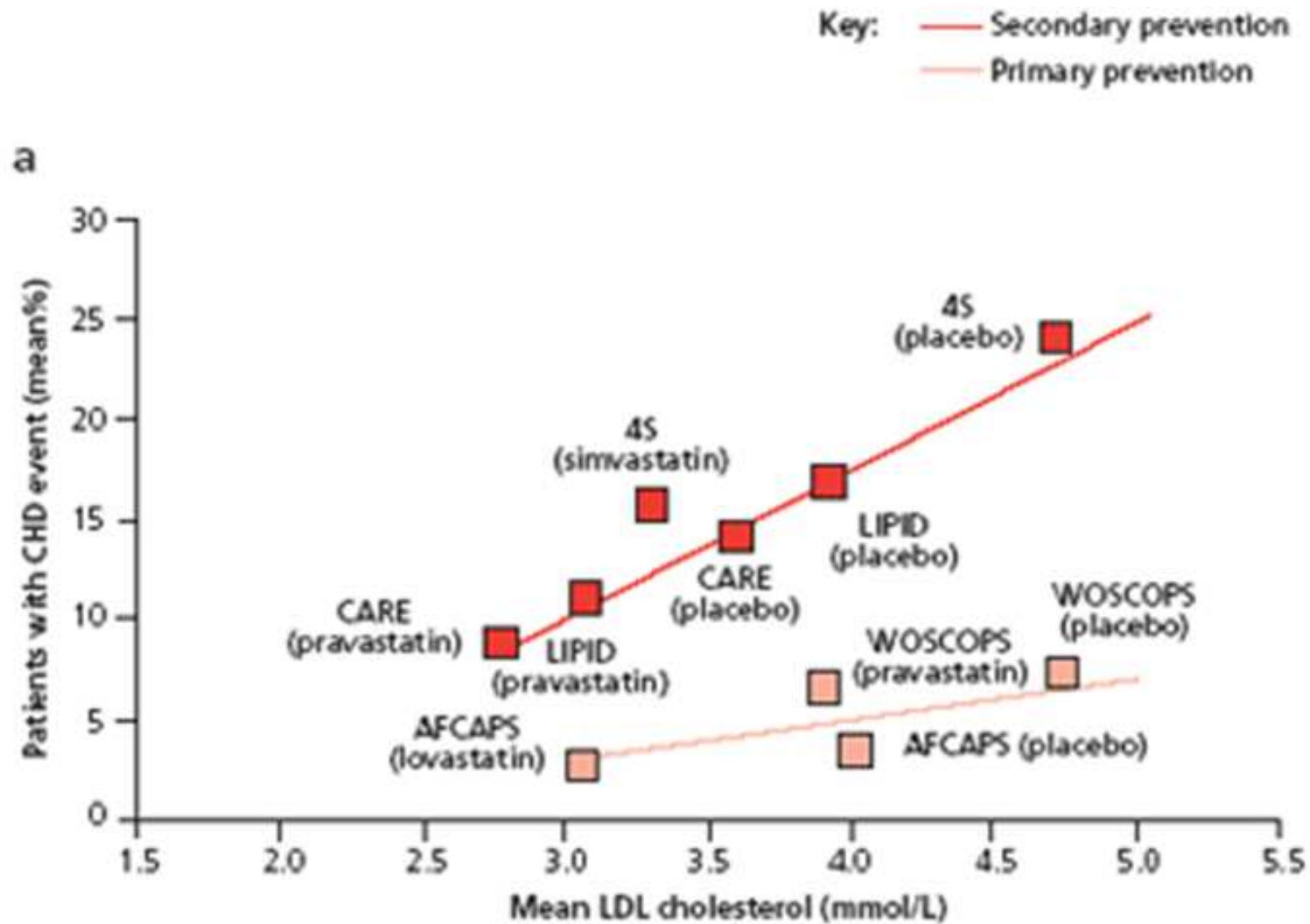
Genome-wide association of early-onset myocardial infarction with single nucleotide polymorphisms and copy number variants

Myocardial Infarction Genetics Consortium*

Table 4 Quintiles of allelic dosage score comprised of nine validated SNPs and risk for early-onset myocardial infarction

Quintile of myocardial infarction genotype score	Odds ratio	95% confidence interval
Quintile 1	1.0 (reference group)	
Quintile 2	1.22	1.04–1.44
Quintile 3	1.43	1.22–1.68
Quintile 4	1.69	1.44–1.99
Quintile 5	2.23	1.89–2.63
<i>P</i> for association of myocardial infarction genotype score with early-onset myocardial infarction: 2×10^{-28}		

Effect of lowering LDL-cholesterol on risk of coronary heart disease



From *Brit J Cardiol*, 2004

Polygenes, Risk Prediction, and Targeted Prevention of Breast Cancer

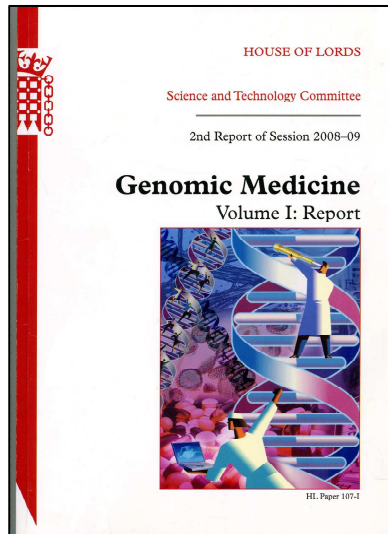
Paul D.P. Pharoah, Ph.D., Antonis C. Antoniou, Ph.D., Douglas F. Easton, Ph.D.,
and Bruce A.J. Ponder, F.R.S.

Table 2. Absolute Risks of Breast Cancer According to Percentile of Population.*

Percentile of Population	Relative Risk	Lifetime Risk† %	10-Yr Risk at 50 Yr of Age† %	Age at Which 10-Yr Risk ≥2.3% yr
5	0.63	6.1	1.5	NA‡
10	0.69	6.7	1.6	NA‡
20	0.77	7.4	1.8	NA‡
40	0.90	8.6	2.1	53
60	1.03	9.7	2.4	49
80	1.20	11.0	2.7	45
90	1.35	12.0	3.0	43
95	1.49	14.0	3.4	41

* The relative risks are based on the risk distribution of seven known breast-cancer susceptibility loci.

† The absolute risks (lifetime risk and 10-year risk at 50 years of age) are estimated from the relative risks and age-specific breast-cancer incidence and all-cause mortality in England and Wales in 2004.



The Inquiry concluded:

We favour a voluntary code of practice. This would offer safeguards for the consumer by encouraging test providers to be open about the limitations of the tests offered, enabling consumers to make an informed decision about purchasing DCTs.

We support the Human Genetics Commission's work on developing, with the industry, a voluntary code of practice for selling genetic tests directly to consumers.

We also recommend that the Department of Health create a web site that should set out up-to-date information on the accreditation schemes with which the DCT laboratories are registered, the quality assurance schemes in which these laboratories participate, and the extent to which the DNA sequence variants used by DCTs for predicting risk of future disease have been validated

CONCLUSIONS

- **Currently no body in the UK has responsibility for oversight of direct to consumer genetic testing**
- **UK advisory bodies recommend a voluntary code of practice based on an internationally applicable framework of principles**
- **Quality assurance should follow international standards such as those in the OECD guidelines**
- **The principles should include consensus on major issues of consent, accuracy of information and marketing, pre- and post-test counselling, data protection and sample handling**

