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A Framework of Principles for Direct-to-Consumer genetic testing services

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Human Genetics Commission UK

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HUMAN GENETICS COMMISSION

Terms of Reference:

1. To advise UK ministers on the potential legal, social, ethical and economic implications of developments in human genetics including:
 - Need to fill any knowledge gaps
 - Development of national policies
 - Requirement for legislative action
2. Encourage public awareness and engagement
3. Work in partnership with other organisations



Current situation in UK

- Majority of genetic tests provided by the NHS:
 - UK Genetic Testing Network (UKGTN) assesses test and provider before recommending funding
 - Offered within specialist clinical genetic services
 - Tests provided by specialist laboratories
- Genetic tests offered directly to the consumer
 - Limited but growing



NHS framework for evaluation of genetic tests: ACCE

- **Analytic validity**
- **Clinical validity**
- **Clinical utility**
- **Ethical, legal + social considerations**



“ One might assume that Lance Armstrong is having some sort of midlife crisis ” Gabby Logan

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From The Sunday Times

September 7, 2008

Rival genetic tests leave buyers confused

Firms that offer to predict your risk of disease give worryingly varied results

Nic Fleming

Leading genetic testing companies are providing clients with widely divergent and inaccurate predictions of their chances of developing serious diseases. That is the finding from tests conducted by different firms on the same person.

Using my own DNA, I approached three firms who between them provide the majority of genetic tests for common diseases in the UK. They gave contradictory assessments of the risk I faced of developing illnesses, including Alzheimer's and glaucoma, and a confused verdict on my risk of suffering heart problems.

The findings reveal that those paying up to £825 for the tests may be receiving either misleading assurances that they face low health risks or are being caused needless anxiety by warnings of high risks.

Lord Taverne, a member of a Lords select committee investigating genetic testing, said: "This [investigation] confirms that some of the commercial genetic tests can be very misleading and harmful. It may cause unnecessary anxiety and people may take action based on an unreliable diagnosis."

RELATED LINKS

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Premature application of science?



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Nic Fleming in The Times:

‘For example **deCODEme** said my risk of developing exfoliation **glaucoma**, which causes loss of vision, was **91% below average**. Yet according to **23andMe**, I was **3.6 times more likely** to get it than average.’

‘For **age-related macular degeneration**, **deCODEme** put my risk at **20% lower** than average, while **23andMe** said it was **62% higher**.’

‘According to **deCODEme**, my risk of developing **Alzheimer’s** was **74% above average**, while **Genetic Health** said my genes were associated with “a **fourfold increased risk** of developing Alzheimer’s disease by your late 80s”.’



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Engelhardt's profile: deCODEme

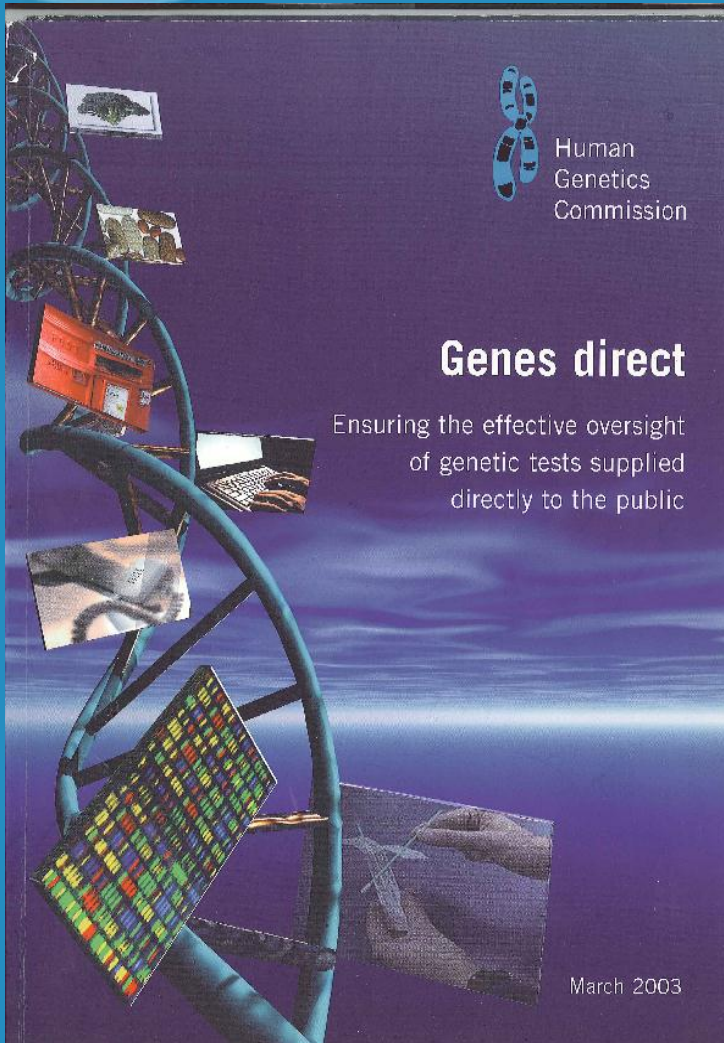
Trait	Relative genetic risk (May 2008)	Relative genetic risk (Dec 2008)
Age-related Macular Degeneration	1.35	0.53
Celiac Disease	0.34	0.32
Colorectal Cancer	0.98	0.87
Crohn's Disease	0.92	0.88
Obesity	1.05	0.77
Prostate Cancer	0.85	0.64
Psoriasis	0.69	0.79
Restless Legs	1.38	1.26
Type 2 Diabetes	1.20	1.25



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Options for Direct To Consumer tests

- Do nothing
- Ban DTC genetic tests
- Regulate in some way





Genes Direct 2003

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

- Defined direct genetic testing as any test to detect differences in DNA, genes or chromosomes that is not provided as part of a medical consultation
- Recommended strict legal controls on the sale of tests and professional self-regulation of those who supply the tests, but no statutory ban



Genes Direct 2003: conclusions

- Predictive DTC tests should only be offered after consultation with a doctor
- Two possible harms identified:
 - Misinterpretation of the significance of results
 - Tests done without informed consent e.g. on children
- Concerns about impact on the NHS



Genes Direct: Framework for regulation proposed

- Medicines and Healthcare products Regulatory Agency (MHRA)
- UK Genetic Testing Network (UKGTN)
- Office of Fair Trading (OFT)
- Advertising Standards Authority (ASA)
- Council for Regulation of Health Care Professionals



More Genes Direct 2007

Report on the developments in the availability, marketing and regulation of genetic tests supplied directly to the public

Recommendations in 3 areas:

1. Pre-market review

- Some tests should not be offered DTC
- IVDD directive classification needs urgent review

2. Advice and Advertising

3. Quality Assurance

- code of practice required



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2008: Meeting to discuss development of a Code of Practice

- Industry representatives from Europe and the USA
- Government
- Consumers
- Charities
- Professionals
- Public bodies



Code of Practice for DTC genetic tests?

- Overwhelming support from all stakeholders
- Tests to be stratified according to risk
- Common standards *re* consent & confidentiality
- Need for quality assurance
- Value of some sort of kite mark



More difficult areas for Code of Practice

- Providing information about clinical validity
- Role of the physician
- How to prevent inappropriate testing of children
- Who would maintain the Code of Practice?
- What sanctions could be introduced for companies not following the Code of Practice?
- Cross border issues



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Framework of Principles instead.....

- Code of Practice for the UK of limited use when tests are available over the Internet
- High level overarching document: a Framework of Principles, could be applicable in many countries and jurisdictions
- Subsequently the UK could develop its own Code of Practice



Framework of Principles

Purpose:

To promote high standards in the provision of genetic tests amongst commercial providers internationally to safeguard the interests of people seeking genetic tests and their families

Identify areas where individual providers, professional organisations, regulatory bodies and/or national jurisdictions should have defined measures in place

Consequences of a DTC test depend on the personality of the subject and their understanding of the result and its implications



Scope

- Tests provided DTC with no intermediary
- Tests provided by non medical intermediary
- Tests commissioned by consumer but where a health professional is involved

(research excluded)



Types of tests covered by principles

- Diagnostic tests
- Pre-symptomatic tests
- Carrier tests
- Prenatal tests
- Susceptibility tests
- Pharmacogenetic tests
- Nutrigenetic tests
- Lifestyle/behavioural tests
- Phenotype tests
- Genetic relatedness tests
- Ancestry tests



Impact criteria

To be considered when determining what additional support is required

- Severity of the condition for diagnostic tests
- Reliability of the prediction for predictive tests
- Likely speed of degeneration for progressive disorders
- Potential for results to influence management
- Likely impact on person's behaviour/lifestyle
- Implications for relatives
- Stand-alone test or one requiring confirmation?
- Potential for test to provide info about 3rd party



Marketing and advertising

- Compliance with local laws & voluntary codes
- Promotional and technical claims must be accurate
- Claims to be supported by evidence



Regulatory information

- Test provider should make available evidence of the association between a genetic marker and disease
- Associations should be validated in more than one large case control study in a cohort that is ethnically/geographically relevant
- Evaluation of the algorithms used to calculate risks figures should be available for scrutiny and review



Information for prospective consumers

- Simple, clear general info about genetics
- Clear explanation about the relative roles of genetics, environmental factors & lifestyle choices
- Specific info about tests offered
- Provision of counselling
- Presentation of results and interpretation
- Confidentiality of records and storage of samples
- Secondary uses
- Handling and resolution of complaints
- Statement about potential of results to reveal unexpected findings
- Potential impact of life insurance
- DNA theft
- Provision for samples and data storage if company ceases trading



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Counselling and support

- Pre and post test counselling required for genetic tests in context on inherited diseases
- Counsellors should have appropriate skills and competencies and be accountable to relevant professional body
- Consumer must be able to withdraw from testing after counselling without incurring further costs



Consent

- Free and informed consent can only be given when a customer has received sufficient relevant information
- Company should take reasonable steps to assure themselves that a biological specimen provided for testing was obtained from the person identified as the sample provided
- Consumers should sign a statement confirming informed consent and that they have read and understood the information provided. Statement should state what will happen to the sample and must be retained



Consent

- Separate, specific consent required for subsequent testing or any secondary uses
- Research must be approved by a Research Ethics Committee
- No testing for adults unable to consent
- Companies must be aware of DNA theft laws



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Genetic testing of children

- Medical tests should normally be deferred until a child/young person has capacity to give consent, unless the management of a clinical diagnosis would be materially influenced by genetic testing, in which case it should be commissioned by a health care professional who will take responsibility for subsequent follow-up



Data Protection

- Highest levels of security and confidentiality required
- No sharing of identifiable samples or data with a third party without consent
- Explicit plans for sample data/storage required in the event of a company ceasing trading



Sample handling

- Nature, purpose and maximum duration of storage should be specified
- Use, storage, transfer and disposal of samples must be in accordance with applicable ethical, legal and professional standards
- Provision for demise of company



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Laboratory processes

- Requirement for high standards as evidenced by appropriate accreditation
- Policies for corrective measures if laboratory performance fails in any way
- Education and training for laboratory personnel



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Interpretation of test results

- Appropriate qualified personnel with recognised training and qualifications
- Results must be accurate and comprehensible to consumer
- No remuneration that allows individual interpreting result to benefit from any particular interpretation (e.g. sale of subsequent services)
- Risk assessment must be based on robust algorithms
- Test providers must regularly review the evidence on which test interpretation based



Provision of results

- Provided to customer is a format that is easy to understand
- Indicate the significance of a results and other relevant (non genetic) factors
- Pharmacogenetic tests: consumer to be advised not to alter medication without professional advice
- Test provider must not overstate the significance of a results
- Distinguish carefully between relative risk & absolute risk
- Security measures required to protect results
- No result to go to 3rd party without consent



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Continuing support for high impact test results

- Companies must provide information about how to access it



Complaints

- Written procedure required
- Designated senior member of staff
- Complaints procedure to be easily accessible
- Complaints to be addressed promptly
- Consumers must be told what further recourse may be available to them if still unsatisfied



Impact of Framework

Well received internationally

- US FDA stated that tests must be subject to pre-market assessment; but Obama wants to relax regulations pertaining to businesses
- Australian National Pathology Accreditation Advisory Council supports principles and is developing guidance in Australian context
- UK coalition government inclined to favour self regulation



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