Royal College of Physicians

The Royal College of Pathologists
Pathology: the science behind the cure

Submission

Science and Technology Committee

Commercial Genomics

This is a joint submission between the Royal College of Physicians, the Royal College of Pathologists and their Joint Committee on Genomic Medicine.

Summary

1. We are excited by the opportunities for genomics to advance medicine and improve health. Genomics has already transformed the lives of many and promises much more in the future.

2. Genomic tests should only be approved for use in healthcare if there is evidence of benefit to the individuals being tested. When used for screening this means that before implementation they must meet the same requirements as other types of screening test.

3. Genomic tests can lead to confusing and/or worrisome results. Commercial providers do not, at present, support patients who are left confused and anxious. NHS providers are left dealing with unexpected or difficult outcomes, placing additional strain on the NHS whilst the commercial providers take their profits.

4. Selling whole genome sequencing to those that can afford it will create a two-tier health service and be of limited research value.

5. **We advise creation of a regulatory body similar to the Human Fertilisation and Embryology Authority (HFEA) to oversee genomic testing in the UK. This would protect the public and the NHS, and support the UKs continuing global leadership in collaborative genomic research and delivery of a high-quality genomics clinical service.**

Inquiry questions:

Any health or other benefits that consumers can derive from using commercially available genomic testing

Most commercially available direct-to-consumer genomic tests imply that they evaluate a much greater proportion of an individual's overall risk than they do. Many diseases - especially those occurring most commonly and in mid-late life - have complex origins in genetic predisposition, environmental exposure and random events. For many such disorders the genetic contribution is modest and incompletely understood, and for many there few (or no) satisfactory strategies to reduce risk.

Misinformation is common. It is not possible to evaluate the extent to which current commercial testing can provide health benefit without a regulatory framework to agree quality parameters by which this can

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be judged. NHS genetic services have seen many examples of inaccuracy and error with potentially serious consequences for the patients affected.

Pre-conceptual couple-based carrier testing, eg for cystic fibrosis, through reputable providers who have published their data in the peer-reviewed literature could potentially be conducted in partnership with NHS-services, although concerns about preferential access to care by those who can afford to pay for such testing would have to be recognised and mitigated.

The extent to which currently available genomic sequencing and interpretation can provide accurate and unambiguous health results, for healthy and ill sections of the population

Genetic testing can be very beneficial in the diagnosis and treatment of diseases that are proven to have clear genetic causes and interventions known to improve outcomes, hence genomic sequencing is most effectively targeted towards those people who show symptoms, or have strong family history of, an inherited disease.[1]

The interpretation of the health impact of genomic sequencing is very dependent upon context and clinical evaluation of a patient's symptoms and signs (if any), their family history, and the reasons underlying the test request.

For healthy people, genomic tests will usually only adjust individual risk by modest amounts. Lifestyle factors such as obesity and smoking often make a far larger contribution to overall disease risk than genomic susceptibility.

The counselling or other support offered for those receiving, or considering asking for, commercial genomics test results, and whether this is to the standard required

In general, genetic tests in healthy people only adjust individual risk by modest amounts, but in all individuals extensive genomic sequencing will throw up abnormal results that are most unlikely to be of clinical significance, but which will provoke considerable anxiety in many. Any worrisome or confusing results are generally left for the NHS to evaluate and interpret, creating an uneven playing-field where the commercial providers take profit from offering tests whilst contributing nothing to the NHS providers who are left dealing with unexpected or difficult outcomes.

The potential benefits and risks for the NHS that arise from the increasing availability of commercial genomic testing

The UK has great strength in public health and genetic epidemiology and has internationally-renowned academics and researchers who could deliver real benefits to the nation by developing rigorously-evaluated genomic screening that is integrated with existing systems (where appropriate) and meets the criteria required by the UK National Screening Committee.

[1] BMJ 2019;364:l789

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The unrestrained and unregulated use of commercial genomic testing by the public is a major risk to the NHS. When uncertainty arises in medical practice, the default approach is for patients to demand and the NHS to provide 'just in case' investigations and care, even though the likelihood of harm may be very low. The 'worried well' with anxiety around confusing or misleading commercial genomics test results are likely to create a serious drain on a resource-constrained system, with articulate low-risk patients competing for places in clinics with those with significant genetic diseases. Cascade testing of relatives or enthusiastic encouragement for relatives to also participate in commercial genomic testing will amplify the problem further.

What data obtained from genomic testing could be used for and if sufficient protection is in place for consumers using commercial genomic tests

Offering fee-for-service whole genome sequencing to healthy individuals will generate data of doubtful research value. It would be better to invest in more sequencing of UK Biobank or other well-phenotyped cohort studies.

The regulations or standards that commercial genomic tests are currently subject to, and if any new or strengthened regulations or standards should be introduced to mitigate any perceived risks associated with commercial genomic testing

It is rarely possible to determine the performance characteristics of a commercial genomic test and how it compares in quality and accuracy with other similar offerings or those available within the NHS.

We advise a similar regulatory approach to that adopted for the pharmaceutical industry. Pharmaceuticals are approved as 'prescription only' or 'over the counter' medicines. Genomic tests could be approved for 'access via a registered health professional' or 'direct to consumer' use. Companies offering tests should display a minimum amount of information for example:

- the scope of the test
- who it is intended for
- the potential benefits and harms
- the sensitivity and specificity of the test
- the likely range of results
- the recommended strategies for managing the results

Thought must also be given to ethical concerns, such as submission of DNA samples from people who lack capacity, and the implications of genetic results for family members who have not been tested.

The potential benefits and risks, for individuals and for the NHS, and the ethical implications of the NHS offering genomic testing to healthy individuals willing to pay and share their data anonymously

This approach has the potential to result in two-tier access to genetic services, with the wealthy worried well that harbour variants of dubious significance identified on whole genome sequencing competing for clinic space with patients with serious genetic diseases. A fundamental tenet of the NHS is that care is delivered on the basis of clinical need and not on the basis of ability to pay.

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About the RCP

The RCP plays a leading role in the delivery of high-quality patient care by setting standards of medical practice and promoting clinical excellence. We provide physicians in the UK and overseas with education, training and support throughout their careers. As an independent body representing over 36,000 fellows and members worldwide, we advise and work with government, the public, patients and other professions to improve health and healthcare. Our primary interest is in building a health system that delivers high-quality care for patients.

About the Royal College of Pathologists

The Royal College of Pathologists is a professional membership organisation with more than 11,000 fellows, affiliates and trainees, of which 23% are based outside of the UK. We are committed to setting and maintaining professional standards and promoting excellence in the teaching and practice of pathology, for the benefit of patients.

Our members include medically and veterinary qualified pathologists and clinical scientists in 17 different specialties, including cellular pathology, haematology, clinical biochemistry, medical microbiology and veterinary pathology.

The College works with pathologists at every stage of their career. We set curricula, organise training and run exams, publish clinical guidelines and best practice recommendations and provide continuing professional development. We engage a wide range of stakeholders to improve awareness and understanding of pathology and the vital role it plays in everybody's healthcare. Working with members, we run programmes to inspire the next generation to study science and join the profession.