This vision does not represent government policy but provides useful insight into how genetics services might develop over the next 5 years

Annex N

Cancer Genetics Services 2015

Background

1. In July 2009, the House of Lords Science and Technology Committee published the findings of its Inquiry into Genomic Medicine. Whilst the report from the inquiry recognized the advances that had been made since the White Paper *Our inheritance, our future - realising the potential of genetics in the NHS* (June 2003) it contained 84 recommendations on how the NHS, and the UK, could take advantage of new technologies in genetics and genomics. In response, the Government reaffirmed its commitment to the development of genetic and genomic technologies and services in the NHS and maintaining the leading position of the UK in this field.

2. The Government also announced it would establish the Human Genomics Strategy Group (HGSG), a cross-departmental committee with a remit to monitor advances in genetic and genomic research and to evaluate their benefit to healthcare services in the NHS. The HGSG is also responsible for oversight of progress against the recommendations contained in the House of Lords report.

3. The NHS Cancer Plan stated the Department of Health would work with Macmillan Cancer Support to develop new services to improve cancer genetic risk assessment and counseling. This work began in 2001, with the production of the Kenilworth Model of service delivery. Seven pilot sites were selected to make up a partnership programme with Macmillan. These pilots have now finished and learning from the pilots can be taken forward.

Assessment and referral

4. All individuals concerned about an inherited risk of cancer will be swiftly assessed, classified into high, moderate or general population risk, and then directed onto a clear pathway where they are either appropriately reassured or referred on to secondary or tertiary care. There will be clear access to services for concerned individuals and healthcare professionals, and effective triage of people concerned about inherited risk will enable specialists to see the most appropriate referrals. There will be no one model for achieving this although local areas will draw on the learning from the Department of Health/Macmillan pilots.

5. More healthcare professionals from outside the specialty of clinical genetics will be trained and supported in taking and drawing detailed family histories in order to support cancer risk assessment of patients in primary and secondary care. The aim is that family history services meet key criteria and are staffed by trained and updated health professionals closely linked to clinical genetic services.

6. Greater equity in accessing services will be achieved through careful planning and promotion of services to take into account the demographic profile of the local population across all socio-economic and ethnic groups. The anticipated growth in cancer-related referrals in certain areas with high proportions of ethnic minority

populations will need to be fully resourced.

7. The development and commissioning of appropriate local services will be informed by and involve people at an increased inherited risk of cancer, leading to improved design, delivery and sustainability.

Clinical genetic services

8. The support and expansion of tertiary cancer genetic services will continue, as it is acknowledged that this activity is yet to plateau. All people in England at an increased inherited risk of developing cancer will have appropriate access to quality, evidence based cancer genetic services. Clinical genetic services will have close ties with primary and secondary services, delivering a coordinated service and improving access to appropriate assessment, genetic testing and surveillance.

9. Clinical genetic services will provide genetic counseling, diagnostic and predictive testing for patients as appropriate for their risk level.

10. People with inherited, strongly predisposing cancer gene mutations will benefit from the increased availability of coordinated follow-up care via multidisciplinary carrier clinics to address their specific cancer risk management needs, which may include early tumour surveillance, risk reducing surgery (for example mastectomy and bilateral salpingo-oophorectomy), dedicated psychological support and access to relevant research trials.

Education

11. The work of the NHS Genetics Education and Development Centre (NEGDC) on developing a set of UK Workforce Competencies for Genetics in Clinical Practice for Non-Genetic Healthcare Staff will be an integral part of all appropriate curricula. Clinical genetic services will play an increasing role in the education and training of non-specialised practitioners. Resources will be provided to support sufficient genetic education facilitators to train all healthcare professionals who require a working knowledge of cancer risk assessment in primary and secondary care.

12. The National Healthcare Science School of Genetics will have established a single entry education programme for healthcare scientists specializing in genetics. It will also be at the forefront of a national programme for the teaching of bioinformatics (the use of IT to analyse, visualize, catalogue and interpret genomic information) to establish the discipline as an integral part of the education pathway for healthcare professionals and scientists.

Surveillance of people at raised risk or greater

13. Unnecessary surveillance of people at general population risk or marginally raised risk of developing cancer will be ceased. Communication plans will be developed and implemented so that those who will no longer receive surveillance understand the reasons for this and receive appropriate reassurance and support.

14. Based on advice from the Advisory Committee on Breast Cancer Screening, the NHS Breast Screening Programme will develop and implement appropriate surveillance protocols for women identified as at increased risk of developing breast cancer, including MRI surveillance where appropriate. This will reduce geographical inequalities and allow proper quality assurance and evaluation of effectiveness. Each local area will have clear pathways for identifying women eligible for enhanced breast screening and for referring them into the programme. Identification of the highest-risk women eligible for enhanced surveillance will occur primarily through clinical genetic services as these will identify women with BRCA1, BRCA2 or TP53 mutations and facilitate cascade testing and subsequent surveillance of close relatives.

15. The NHS Bowel Cancer Screening Programme will develop and implement appropriate surveillance protocols for people identified as at increased risk of developing bowel cancer. This will ensure appropriate audit, capture and follow-up of these patients.

Genetic testing

16. Diagnostic and predictive gene testing for high risk cancer predisposing disorders will be available with shortened turnaround times as technology improves. There will be ongoing regular investment to replace outdated molecular testing equipment to ensure turnaround times can be reduced in line with service needs.

17. The capacity for genetic testing will expand significantly to meet the needs of patients with an inherited cause for their cancer. As personalised cancer treatments for those with specific genetic mutations become increasingly available and more clinical trials are undertaken in this group, there will be increasing demands on the availability and capacity of rapid genetic testing to determine eligibility and allow optimal management of cancer patients. Continued investment in genetic testing and services and closer ties between clinical genetics and oncology will mean that cancer patients who require testing will have swift access to their results and the genetic counseling support they need to make decisions about their care.

Stratified medicine in the use of personalised healthcare

18. Pharmacogenetics, the study of the way in which genetic variation across the genome affects an individual's response, or lack thereof, to specific drug treatments, and its application as part of the diagnostic and treatment pathway, will become more commonplace. A more targeted approach to treatment based on higher quality and better understood genetic information will see more effectively applied treatments delivering better patient outcomes. The development of companion diagnostics will further refine the ability to tailor therapies to the individual.

Advances in genetic knowledge

19. The discovery of new lower-penetrance genes associated with familial disposition, and increased understanding of the role of existing known genes and their interaction with lifestyle factors, will allow better and earlier identification of those at risk of developing cancer, opening up more opportunity for prevention and early detection.

Developments in the technologies to deliver testing and the rapid integration and commissioning of these advances will enable quicker and more cost-effective testing. Effective translation of research findings into clinical practice will require detailed bioinformatics knowledge, since many low penetrance genes exert their effects only in a specific genetic and/or environmental background.

The Concordat and Moratorium on Genetics and Insurance

20. The current Concordat and Moratorium on genetics and insurance runs to November 2014 with a review in 2011. Whether or not it is extended further, there will be measures in place to ensure that patients are not deterred from taking predictive genetic tests by fears that insurers may use their adverse test results in making decisions about insurance premiums.

A national register for families with known genetic mutations

21. A national register for families with known genetic mutations is desirable to enable long-term follow-up of these families. This would allow the collection of data on pathogenic mutations and unclassified variants in single cancer genes (such as BRCA1, BRCA2 and TP53), surveillance outcomes, cancer incidence and outcomes from risk reducing surgery, supporting the continued development of evidence-based management of people with these mutations. However, the difficulties of establishing a national (rather than local or regional) register as well as the appropriate consideration of consent and confidentiality of results would need careful consideration, as would security and access rights. By 2015 it is more likely that surveillance through the national screening programmes will allow long-term follow-up of people eligible for enhanced surveillance.

***Improving Outcomes: A Strategy for Cancer* Stakeholders**
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