Editorials

Genomics in general practice:

Generation Genome

Genetics (the functioning and composition of a single gene) and genomics (all genes and their inter-relationships identifying their combined influence on the growth and development of the organism)[1] are developing at a rapid rate. The impact this will have on general practice is uncertain. However, the recent publication of Dame Sally Davies's 2016 Chief Medical Officer's (CMO) report *Generation Genome* outlines how genomic medicine can be utilised and developed to become available to more patients within the NHS and the delivery of the genomic dream.[2] GPs may progressively be expected to consider genomics in their diagnosis and treatment decision making, and patients are likely to increasingly seek from GPs explanation for tests they have had in secondary care or from direct-to-consumer testing that they obtained privately.

A recent editorial in the *BJGP* looked at the clinical areas where GPs are most likely to come across genomics in practice and asked what is needed from general practice in order for it to become 'genomics ready'.[3] This article looks at some of the barriers and inequalities to accessing genomic medicine, and considers how genomics might be incorporated into general practice.

BARRIERS AND INEQUALITIES TO ACCESS OF GENOMIC MEDICINE

Dame Sally Davies's report describes the '*tendency in some parts of the NHS to think of genomics as a thing far in the future or even worse, a potential burden rather than a boon*'. While acknowledging where this view comes from, she highlights that in the long term it will affect equality and access to genomic care.[2] Though a third of all referrals to genetic services are currently made by GPs, there is wide variation in the extent to which this occurs, with some GPs never referring.[4] Such variation reflects differences in access, knowledge, and confidence within primary care. From the GP's perspective, there may be concerns about genomic assessment increasing patient anxiety and providing uncertain benefit to the patient. There may also be ambivalence about offering genomic counselling, and whether such advice is part of their role. They may feel lacking in test-specific knowledge and counselling skills, and that they lack the 'back up' of access to genetic services or advice. GPs may be concerned that the speed at which genetic testing is being implemented into clinical practice is being technology driven as opposed to patient or clinician driven. Limited time within a standard consultation is an obvious barrier; 10 minutes does not lend itself to taking a detailed family history and certainly not to explaining the implications of genetic testing.[5,6] GPs may fear opening up a 'Pandora's box' of potential extended family risk, particularly in the absence of rapid access to a genetic counsellor. Even a short 7-item family history questionnaire to identify at-risk relatives of a genetic condition was found to be unlikely to be implemented in primary care due to concerns about workload implications, duplicating roles with secondary care, and potential ethical implications of its use.[7]

> *"... 10 minutes does not lend itself to taking a detailed family history and certainly not to explaining the implications of genetic testing."*

From the patient's perspective, there may be limited understanding of how to interpret genomic evidence in relation to decision making about lifestyle and health behaviour, and limited awareness of their own personal risk due to lack of knowledge about their family history.[6] However, it is likely that, with increasing marketing of direct-to-consumer genetic testing, patients will increasingly initiate such testing.

Inequalities of access are already being recognised in terms of referral to services; for example, referrals to a secondary care familial cancer genetics clinic found that white, married females with a higher level of education and the presence of children were by far the most likely to be represented.[8] It is also likely that genomic services will be clustered around tertiary services, with limitations in access to those living distant from such centres.

Ethnic disparities in the use of cancer genetic services are also recognised.[9,10] People from black and minority ethnic groups are substantially under-represented in those attending cancer genetic services.[10] There is also the complication of cross-cultural communication and that messages about risk may be difficult to convey through translation.[10]

GENOMICS IN GENERAL PRACTICE

Faced with these challenges, how should genomic-informed care be incorporated into 21st-century general practice? Educational initiatives to increase the confidence of providing genomic care within general practice have been and are being considered, but to date these have had mixed outcomes. A systematic review of such interventions found that, although there was insufficient evidence to draw firm conclusions as to how to provide the education required, short-term educational encounters were viewed as being unlikely to cause significant practical change in identifying genetic risk and appropriate management.[11]

Given the extent to which GPs are feeling overloaded, more in-depth education is unlikely to have widespread take-up. An alternative approach that is being considered is for 'just in time' education, as in the Genomics Education Programme (GEP) resource (https://www.genomicseducation.hee.nhs.uk/news/item/156-just-in-time-resources-for-healthcare-professionals), which can be downloaded during a patient encounter. Currently, this consists of factsheets for genetic conditions, but it is anticipated that the resource will expand to include much more 'downloadable' and 'virtual' help for GPs.[3] Although this type of support may be beneficial to GPs who gain familiarity with using it, it is likely that many GPs will see this approach as lacking feasibility within the constraints of a general practice consultation.

There is a risk that the blurring of responsibilities between genomic care in primary and secondary care risks the creation of technical systems that support 'non-specialists' to deliver 'expert care', resulting in a system that potentially puts patients at risk and overburdens practitioners.[12]

LOOKING TO THE FUTURE

Future generations of doctors will be trained from an undergraduate level to incorporate

Box 1. Resources

Unique
Support for families living with a rare chromosomal disorder
http://www.rarechromo.co.uk/html/home.asp

Antenatal Results and Choices
Help for families when a genetic/chromosomal difference is found through antennal screening
http://www.arc-uk.org/

SWAN
Supporting families affected by a syndrome without a name
https://www.undiagnosed.org.uk/

Genetic Alliance
Supporting families living with a genetic disorder
https://www.geneticalliance.org.uk/

RCGP Genetics in Cancer
Education module for GPs
http://www.rcgp.org.uk/learning/online-learning/ole/genetics-in-cancer.aspx

UK Genetic Testing Network
Advisory organisation that provides commissioning support to the NHS and DH for NHS patients in the UK; it advises about which laboratories perform which tests
https://ukgtn.nhs.uk/

genomic medicine into 'personalised care' for patients and at postgraduate level put this into practice. As such, there is likely to be a 'generation gap' as the world of medicine changes from one with little consideration of the patient's genome to one in which the genome is essential in tailoring prevention, diagnosis, and treatment. The CMO's report recommends that the royal colleges place emphasis on continuing professional development and revalidation for those clinicians who lack undergraduate training regarding genomic medicine.[2]

How might services be designed in order that general practice genomics care has equity, sustainability, affordability, and adaptability to the rapid progression of technology? The advent of new, large-scale primary care provider organisations in the NHS is creating opportunities for hub-and-spoke genetic services based around community genetics counsellors. This might improve access to genomic medicine. Such community-based care can improve access and minimise some of the geographical barriers to care.[4] Such services should be accessible to address the needs of patients from ethnic minorities and specific cultural groups, with the presence of appropriately trained, culturally sensitive interpreters. A community-based hub could also provide improved access between GPs and specialist providers (in particular genetic counsellors), with provision of virtual advice. However, without a much needed expansion in the numbers of genetic counsellors it is unlikely that the service will be able to be provided wholly from primary care.

In addition, access to genomic services could become directly available to patients, or direct from secondary care, only involving the GP where additional information about the patient is required. This would help prevent time delays for the patient and avoid extra administration for the GP. Even when referral takes place from secondary care directly to clinical genetic services, however, patients may well seek ongoing care about these issues from a primary care setting.

GPs with a special interest in genomics may help locally to provide a point of contact for GPs, patients, and genetic counsellors, and provide ongoing continuity of care. Nationally, primary care genomic 'champions' may provide links between Health Education England and the Royal College of General Practitioners, and so help GPs to keep abreast of new advances through access to guidance and review articles.[13]

In conclusion, genomics is an often complex and rapidly progressing specialty, which has the potential to support 'personalised medicine' and aid the GP in tailoring care to each patient's specific needs. However, rather than providing stopgap solutions there is a need to consider service re-design to provide community services, and increase GPs' access to advice. For individual GPs, being aware of the limits to your knowledge and where to find information, resources, and signposting for patients is likely to be crucial (Box 1).

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