

Is primary care the right place for genetic diagnosis?

The inaugural meeting of the Primary Care Genetics Society (PCGS) in London on 14 February 2007 was host to around 100 people, including over 40 GPs. Similar grass-roots professional bodies such as the Clinical Genetics Society, have previously played a central role in the shaping of genetic testing services in tertiary care.¹ The founders of the new PCGS hope it will also become an important force for change in the use of genetics in health care, by helping primary care professionals to apply genetic knowledge in their practice and put forward their needs in policy debate. A central feature of the meeting's programme was one such debate around the motion: 'This house believes that primary care is well placed to diagnose genetic conditions'.

It is now more than 3 years since the Department of Health outlined plans to encourage wider exploitation of genetic knowledge in the NHS,² including in primary care, and the debate reported here, was an informative thermometer of current opinion.

Dr Nigel Starey (for the motion) based his argument on the holistic approach that GPs have traditionally played in the care of patients. Despite today's more mobile population, people still form longer relationships with their GPs than with those in more episodic-based secondary care. GPs are therefore well placed to help patients to navigate the system and balance their different needs. Even though some may not see the relevance of genetics to primary care practitioners, Starey, a GP with a special interest (GPwSI) in genetics, elaborated on how colleagues at his practice support patients with a wide range of genetically relevant matters: managing patients with chronic genetic conditions such as Down's Syndrome; discussing issues around amniocentesis and or CVS (chorionic villus sampling); and routine

pre-natal (Down's Syndrome) and postnatal screening tests with young families. Overall, genetics has been estimated to be relevant to perhaps 10–15% of consultations but Starey believes this to be an underestimate, with queries ranging from young patients concerned with acne or stature, to adults concerned about male pattern baldness, family history of cancer, or drug response to common drugs such as codeine and warfarin. Starey sees the role of primary care practitioners as being able to diagnose, inform patients and aid in their decision making, refer them further to specialists, to support, and to manage ongoing care.

Dr Ian Robinson (who was required to hide his GPwSI hat in speaking against the motion), suggested that although GPs are ideally placed to make genetic diagnoses, coping with current workload was their main priority. Genetics is 'mostly off the radar' for GPs. They did not have the 30–45 minute sessions genetic counsellors could offer, and had little time to adapt their 10-minute sessions by getting relevant materials prepared for genetic counselling, or have time in the session to undertake tasks such as taking pedigrees. GPs were also not supported with delegating these tasks to others and, despite trials and IT-based systems to enhance genetics, knowledge-based decision making had not yet become available. Furthermore, as GPs become increasingly target driven, there is no QOF-based mechanism to promote such activities.

Emery and Hayflick³ had previously suggested that education, specialist GPs, IT systems, and counsellors should be used in combination, but Robinson believed the role of a new breed of primary care geneticists was the most crucial measure. These specialists, (rather than uninitiated GPs or GPwSIs) should deal with genetics-related enquiries,

fielding these locally.⁴ This could reduce the need to make referrals to tertiary regional genetics centres, which were considered to be overloaded already.

These views were elaborated in a 30-minute discussion (anonymised comments are summarised here) which began with a challenge of how primary care geneticists (whether counsellors or GPs) could discover which patients needed specialist advice and arrange referral, even within the PCT, if GPs were not aware of the relevance of genetic factors raised.

Research was cited as showing that 'many GPs are confidently incompetent' (that is, they do not know what they do not know about genetics).⁵ Furthermore, where patients in need could be recognised by GPs, there was concern that the additional funding for appropriate management of rare genetic conditions, although worthy, was difficult to build a case to support in cost-benefit terms, given anything other than a long-term view. Such difficulties, a counsellor suggested, may make the lower cost of counsellors than GPs a relevant factor in moving towards primary care-based genetic practice. The 'holistic GP' was further criticised as not being 'a sustainable model'. One GP referred to the 10 GPwSIs in genetics funded by the Department of Health's 2003 genetics white paper, as an 'expensive experiment', implying that evidence in support of the usefulness of this (still ongoing) initiative is unavailable. However, even if the initiative is successful, GPwSIs are a self-selected group, many of whom have a prior interest in genetics, including some with genetics training at degree level. Overall, their capabilities are unrepresentative of GPs as a whole.

The status quo was seen as unacceptable with GPs referring patients they could not help to tertiary care, who

then 'bounce back' either as inappropriate referrals or, following counselling, for ongoing care. One GP suggested local referral triage systems could help GPs to learn from each other. However, more broadly, there was concern that GPs would not receive the educational support to properly establish genetic knowledge to improve the current situation. The clinical genetics departments of regional genetics centres were unable to provide greater levels of education given the current resources, (it was subsequently noted that only £15 million had been made available for dedicated initiatives relating to capacity building in primary care across the whole country in the wake of the genetics white paper).²

New monies had yet to be identified to maintain or supersede existing efforts beyond the next 1–2 years. The prospect that genetic services could once again become 'Cinderella services' raised patient group concerns that some strategic health authorities would champion services, while others would not, leading to a postcode-lottery situation for those in need. A non-GP expressed hope for a national network of GPs with competency in genetics. In the longer term this may become more realistic as it was noted that the RCGP curriculum will include a genetics module from 2007.⁶ Some people at the meeting saw justification for genetics education on such a scale, as correct diagnosis through early testing (especially where genetic testing could better guide treatment) was perceived as likely to be 'massively cost-effective'. The chair, Dr Anna Stone, concluded that:

'... the whole work force needs to understand their role in the system ... [there is a] need for more genetic education for all.'

Despite the high level of interest in

genetics shown by this audience, the vote was against the motion. Primary care was not viewed as the place to undertake genetic diagnosis, at least for the time being. Indeed, slightly fewer people supported the motion than at the start of the debate. It seems clear that primary care professionals will require much greater support to facilitate the wider use of genetics than they have received to date, if the Department of Health expects genetic knowledge to have a wider role in primary care.

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The Primary Care Genetics Society aims to support and facilitate the educational needs of primary care practitioners to help translate the continuing advances in clinical genetics into practice. Membership is open to all with a professional interest in primary care genetics. Please visit www.pcgs.org.uk for further information.