DH consultation on UK Plan on Rare Diseases

1. I write with regard to the DH consultation on the UK Plan for Rare Diseases.

2. The Royal College of General Practitioners is the largest membership organisation in the United Kingdom solely for GPs. Founded in 1952, it has over 44,000 members who are committed to improving patient care, developing their own skills and promoting general practice as a discipline. We are an independent professional body with enormous expertise in patient–centred generalist clinical care. Through our General Practice Foundation, established by the RCGP in 2009, we maintain close links with other professionals working in General Practice, such as practice managers, nurses and physician assistants.

3. The College welcomes the opportunity to respond to this consultation and our responses to the questions posed in the consultation document are as follows:

1. Do you agree that commissioners of services should explore the potential of expert clinical systems to reduce diagnostic delay, particularly in neurology and genetics?

The evidence is not yet conclusive that expert clinical systems used in a primary care setting reduce diagnostic delay. We believe that the integration of these
systems into primary care will take some time and attention to detail to get right and evaluate. Stand alone clinical expert systems do however exist and serve as a useful educational tool. The systems that might be studied should of course not just cover neurology or genetics alone.

It is time consuming, but worthwhile to use a diagnostic algorithm with a patient.

We would also like to emphasise the importance of avoiding referral 'ping pong' where a negative diagnosis from one specialist rebounds to the group for referral to another specialist and further waiting time to be seen and for diagnostic tests. A failure to diagnose should prompt lateral inter consultant referral to a high point in the second waiting list. Commissioning contracts in the future should permit this.

2. Can you suggest ways of rare disease featuring more prominently in speciality training for doctors?

Rare disease training needs to start earlier than at the level of speciality training. Pressures of service delivery and meeting portfolio training needs will compete. Undergraduates should learn the value of recognising signs and symptoms which don’t ‘fit’ to a common disease. One way which is known to be educationally effective is ‘case-based’ or ‘problem-based’ learning where a variety of different illnesses are taught based on commonly presented symptoms. When this is coupled with meeting patients with different conditions, whether rare or common, this can be very powerful and medical schools often use rare problems to teach about normal and abnormal symptoms.

The value of putting together a case report around a patient who has suffered diagnostic delay is quite powerful and should be encouraged including through speciality training.

Training also needs to take in the value of learning how to manage co-morbidity. This will, and is, becoming a major factor in how we interact with patients and manage them and, by dealing with patients with a shared understanding of the major underlying condition or presenting problem, will aid diagnosis.

The value of significant event audit is important and every rare disease must be recorded as a significant event and this method of quality improvement ingrained into the mind set of any healthcare professional.
Universities have an important role to play in supporting the continued education of GPs, other doctors and medical professionals in rare disease issues. Improved opportunities for networking and sharing expertise and the development of innovative ways of delivering these to busy professionals would be helpful. This should be in collaboration with, for example the Medical Royal Colleges and the relevant structures in the NHS to achieve effective access for all who need it.

Other ways that rare diseases could feature more prominently in speciality training for doctors are:

- students to research one rare disease and report back to the group;
- GP attachment;
- meet a family with a rare disease patient; and
- meet an expert patient and ask them to describe their illness, its effects and their experiences at non-specialist clinics etc.

General Practitioners do benefit from the RCGP and BMJ e-learning modules as well as case discussion in small learning groups.

3. Do you agree that the UK National Screening Committee should take into account the benefit of screening in reducing the ‘diagnostic odyssey’ and in allowing informed choice for subsequent family planning?

An example of a generalist is someone who has a sufficient breadth of skills and competencies to handle undifferentiated symptoms safely, and to start initial assessment and treatment. Using this description in the UK, the first point of contact for any person seeking care for a new problem is nearly always with a ‘generalist’. Medical generalists are important in the ‘front line’ where problem mapping and diagnosis is essential.

GPs see large numbers of people for many different conditions, often very early in the development of their illness. The epidemiology is very different in primary care compared to secondary. The GP will make an initial assessment and refer as appropriate. We agree that early diagnosis is important.

With the possible expansion of the newborn screening programme, and antenatal screening there will inevitably be greater awareness of the screened diseases.
This will occur at both public and professional level. Informed choice is mandatory to make reproductive choices.

However, we would suggest that whilst screening is certainly valuable if treatment is available, a positive screening test in an area where treatment is not available and no specialist care is available, can be cruel and possibly destructive, particularly regarding new parents bonding with a new baby. The best value for new born screening is when treatment can start soon and improve lives.

The issue of implications for future family planning is very important, but must take into account family attitudes to termination of pregnancy. Selective IVF may be an option in some localities but funding for this is not clear cut, and may have regional variation. Early genetic diagnosis is very important if any choice about family options is to be possible.

4. How can the NHS best ensure research in rare diseases carried out by the NIHR biomedical research centres and units is rapidly transferred into practice for the benefit of UK patients and their families and carers?

The College suggests that translational studies and quality improvement work can rapidly translate research into service delivery, therapeutics and better diagnosis.

On NIHR biomedical research, there is a need to link research funding to specific clinical centres which have specialists with the relevant clinical skills to be able to conduct research. Localities need to have robust and easily accessible ethical committees to approve research. Patient registers are necessary to identify patients who may benefit from the research.

The RCGP supports the development of, and strategic funding for, clinical research networks (CRNs) focused on rare disorders. A CRN is a multidisciplinary multi-centre network, with agreed aims and objectives, researching a particular condition or group of conditions. Support for the development of these networks would enable experts to collaborate nationally and internationally, leading to a pooling of knowledge and expertise. There needs to be formal and informal methods to assist collaboration between researchers and reasonable adjustments made to ensure there is adequate funding for research into rare diseases.
The UK Plan for Rare Diseases should also address current inequalities in access to medicines for rare diseases, and lay out a clear framework for the process by which new medicines are to be evaluated and the basis for decisions as to whether or not they are to be provided by the NHS made clear so patients and professionals can contribute to the decision making process alongside other stakeholders.

5. Do you agree that commissioners of care for people with rare diseases should assess options for improved care coordination, including named care coordinators?

Yes, the College believes this should be mandatory. However it is unlikely that there will be a care coordinator for every rare disease within the geographical boundaries of a clinical commissioning group. However the principal of care coordination would be a necessary commissioning outcome. The emphasis on community care, integrated care and the utilisation of the MDT, would be welcomed by both patients and healthcare professionals.

A care coordinator would need to have the necessary competencies to be effective, including good knowledge of the disease, the natural history, the organisation of care that is available including knowledge of where the expert centres exist.

6. Do you agree that this list of criteria for expert centres should be the basis for future shaping of services?

- Co-ordinated care
- Adequate caseload for expertise
- Not dependent on a single clinician
- Arrangements for the transition from children’s to adults’ services
- Engaged with people with rare conditions
- Research active.

Yes. All these parameters will be considered by commissioners of such services and are highly relevant. We welcome the setting up of the Centre for Rare Diseases and personalised care at the University of Birmingham which is research active. Good examples of expert centres include the Motor Neurone
Disease (MND) centres which work to provide holistic care. The RCGP are working with the MND association and will help to promote the good work being done. With rare diseases we recognise that affected patients and their families will need to travel great distances to expert centres. Perhaps access to care needs to be provided in new and imaginative ways including the use of telehealth technology.

7. Do you agree that each expert centre must know its network of local hospitals, and the local hospitals must know the pathway to the expert centre which will offer help, support, advice and assistance?

Yes. Not only local hospitals, but also General Practitioners and the clinical commissioning groups. It is inconceivable that through the commissioning framework that the network members are not known to each other.

The advice offered by expert centres must be flexible, responsive and if necessary available 24 hours each day. The advice could be offered by telephone, but again there is an opportunity to use secure web based systems such as video links to access care.

8. In England, how best might this be facilitated with the introduction of Local HealthWatch and HealthWatch (England)?

HealthWatch (England) should have an explicit scrutiny role over services commissioned by the NHS Commissioning Board rather than advisory. Local HealthWatch should have explicit responsibility to oversee local Clinical Commissioning boards and specialised services based in that locality, with regard to their importance for patients based outside of that locality. In addition Local HealthWatch must oversee and scrutinise access to services based outside that locality for patients based within that locality.

Healthwatch should have the power so that action is taken by the Care Quality Commission (CQC) when there are concerns about health and social care services. Healthwatch and CQC will need independent specialist advice on the quality of care of people with rare diseases. In the annual report of Healthwatch there needs to be a standing section on rare diseases.
Patient voices also need to be heard through patient participation groups. Local patient representatives on CCGs will need to ensure feedback is secured and appropriately actioned.

It might also be worthwhile considering appointing a local clinical champion for each area, to take in the consultation and turn it into clinically meaningful changes in practice, procedures to improve care. Linkage of electronic records, involvement of patient bodies, thinking of establishing generic care pathways would be helpful.

9. Do you agree that the United Kingdom should continue to participate in the Orphanet project?

Yes. The Orphanet site is an excellent resource and one of the roles that the RCGP Clinical Champion for Rare Diseases could take on is assist to publicise this to the members of the RCGP. It is likely that awareness and use of the Orphanet site is low in primary care.

10. What sources of patient information and support are available which are not listed in this plan?

Patient information systems attached to GP computer systems are useful when the diagnosis is made.

The College uses online learning resources such as Essential Knowledge to provide educational updates to our members. An example of a good patient online site is the DIPEX project which provides video narratives of the experiences of patients accessing healthcare. Good validated sources of patient information must be provided by the expert centres and a recognisable portal.

Patient UK is the primary source of information for patients but it lacks information for patients on rare diseases or in accessible formats for patients with learning disabilities and those with sensory impairments. There needs to be UK accredited online rare disease portal drawing together information with a review of the information needed by health professionals. There needs to be face-to-face information appointment at a designated time after diagnosis with involvement of Designated Care Coordinators.
11. Do you agree that registers are an important tool in rare disease and could be a core component of the service specification of an expert centre?

Yes, but they must be properly resourced. Good outcome data on patients being looked after by expert centres is, of course, central. Although this could be a core service specification as an incentive, it is important for expert centres to publish their data to demonstrate value for money, effective care and disseminate good practice.

12. Are there any areas of work that the UK Plan on Rare Disease needs to pay particular attention to in order to advance equality?

Mention must be made of the difficulty during the transition phase between adolescents and adults. There needs to be coordination of care that picks up on the physical and psychological needs of such individuals.

Travel to access expertise will be an essential element of the provision of care for most rare disease patients unless they happen to live close to the relevant centre of expertise for those with their condition. It is hoped that the plan will make it easier to do this, and also make better use of these visits to address all relevant issues in one visit. The development of “virtual” clinics, telemedicine and other innovative measures may help if integrated with locally provided services.

The NHS in each of the devolved nations is responsible for making arrangements for meeting the needs of rare disease patients living there. It is not automatic that they will choose to send their patients to centres of expertise in England, just as it is not automatically the case that English patients will be referred to centres of expertise in other nations. A close integration of mechanisms across the whole of the UK would help so that patients and families wherever they live can be confident that they will be able to access the expertise they need whether it is in England, Scotland, Wales or N Ireland – or even further afield if this is the best option.

4. We gratefully acknowledge the contributions of College members in formulating this response and attach a table setting out the individual views of some of our members which we hope you will find useful.

Yours sincerely
Professor Amanda Howe MA Med MD FRCGP

Honorary Secretary of Council