phg foundation
making science work for health

2 Worts Causeway Cambridge CB1 8RN (UK)
Tel +44 (0)1223 740200 **Fax** +44 (0)1223 740892
www.phgfoundation.org

Liberating the NHS: Commissioning for Patients

Response from the PHG Foundation

Introduction

The PHG Foundation (Foundation for Genomics and Population Health) is the successor body to the UK Public Health Genetics Unit. Its overarching purpose is to foster and enable the application of biomedical science, particularly genome-based technologies, for the benefit of human health. The Foundation has a particular interest in the way that new technologies are translated within health services, in genetic research and its impact upon clinical and public health services.

Relevance to commissioning

The Foundation's remit is a public health focussed organisation that has specific expertise in the context of inherited disease. It has recently been involved in the development of commissioning advice and quality standards in the areas of genetic ophthalmology and inherited cardiovascular conditions as exemplar conditions for genetics within mainstream clinical services.

Consultation Question 3.33

How can the NHS Commissioning Board develop effective relationships with GP consortia so that the national framework of quality standards, model contracts, tariffs and commissioning networks best supports local commissioning?

Are there other activities that could be undertaken by the NHS Commissioning Board to support efficient and effective local commissioning?

The PHG Foundation has been concerned for the last decade with considering how NHS services need to embed genetics and genomic technologies into clinical practice in order to deliver the best outcomes for patients. This includes patients with inherited disorders, and also a great many patients and relatives who may have these conditions and whose risk needs to be assessed at a primary or district general hospital level. As such, these patients do not necessarily fall into the category of rare disorders; they include, for example, the child with learning disability presenting to community paediatricians who needs a more detailed diagnosis (*e.g.* from a genetic test) in order to inform management and provide advice to the parents on recurrence risk.

As a further example, this group would also include the child with profound hearing loss who needs an early diagnosis of a genetic condition known as Usher syndrome, in which vision is also lost, in order to consider cochlear implantation at an early stage because dual sensory loss compounds communication problems. There are almost 80 groups of rare single gene disorders that affect the eye - with about 1,500 new cases presenting each year. A similar range of conditions is also present in a large number of other specialties, including, for example, cardiology, renal medicine, neurology, haematology and many others.

The critical point at issue is that these individually rare inherited conditions are extremely numerous and collectively quite common. They will usually present to general mainstream services and have to be considered first in that context. They may need to be fully assessed in relatively specialised services that integrate genetics and mainstream clinical elements.

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Both parts of the pathway of care need to be properly commissioned. Except for rare inherited cardiovascular conditions (such as inherited cardiac arrhythmias) the specialised elements are not currently covered in the national specialised definitions sets. However, their number is increasing as our understanding of inherited diseases increases, and they will be found in every area of clinical medicine. In addition, primary and secondary care elements such as initial recognition, referral and sometimes long-term follow up also need to be embedded by commissioners in the patient pathways that begin in primary care, and include a variety of specialities in district hospital.

In this rather complex area, it is self-evident that GP consortia will need advice, including:

- **Public health advice** - what conditions, how many patients; effectiveness of interventions?
- **Expert clinical and laboratory advice** - how are patients assessed, diagnosed, and treated? What specialist expertise and access to investigations or management is required? What is the expected effect on outcomes?
- **Commissioners** - how much do these services cost? What quality standards are agreed?
- **Patients and families** - what are the attributes of a good service and how can they be achieved?

Although some advice for the more common conditions (such as familial hypercholesterolaemia) or on the provision of genetic tests will be available via NICE and the UKGTN respectively, our concern is that there are no organisations to give commissioning advice, in the round, to GP consortia in this area of genomic medicine. For local consortia, although it might be expected that public health specialists would give such advice, this field is relatively specialised, requires expert input from professionals, laboratories, patients *etc*, and there are very few individuals or organisations across the country that would have the necessary knowledge or experience. Further, as consortia struggle to take on the new agenda, it is inevitable that attention will be focused on the more common conditions and bigger contracts and this area will get lost.

Such advice should be coordinated by the National Commissioning Board. However, it may not have the capacity to do so, or see this as a priority.

We believe that it will be important that the National Commissioning Board explicitly considers how it will procure commissioning advice in the context of inherited disease and genomic medicine more generally. Examples of supportive commissioning frameworks are provided on the PHG Foundation website (www.phgfoundation.org/pages/recentwork.htm); see *Commissioners Guides* under Cardiovascular conditions and Ophthalmology headings.

Genetic and genomics will grow steadily in importance throughout clinical medicine as basic scientific and clinical research progress. It is critical that innovations are embedded in commissioning at this stage, in an evidence- and population-based manner. DH should put in place a structure that will support this through the National Commissioning Board and enable commissioners to be partners in development rather than finding at a later stage that they are technology and provider led in a field in which they have no experience or independent advice.

PHG Foundation

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Contact details: http://www.phgfoundation.org/contact/hilary.burton