# Genomics in the North East & North Cumbria: a briefing paper

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New genomic technologies and knowledge – the ability to incorporate information encoded in DNA into clinical practice – is transforming medical practice. For many years this has been a small-scale enterprise, largely focussed on rare diseases and delivered by tertiary Genetics Services. This is changing rapidly and this paper sets out in summary form some of the activities that will affect you over the next few years.

Earlier in 2015, NHS England commissioned a series of NHS Genomic Medicine Centres (confusing called GMCs), one of which is hosted by Newcastle Hospitals. As part of the application process, all acute Trust Chief Executives signed a letter of support and agreed that their organisations would work with the North East & North Cumbria GMC (NE&NC GMC) – effectively as a network or partner organisations.

## Functions of the NE&NC GMC

*100,000 Genomes Project*

One key reason for creating a national network of GMCs was to find a way of delivering 100,000 whole genome sequences (WGSs) into routine clinical practice. The 100,000 Genome Project is currently the DoH's largest and most high-profile development and is effectively the world test-case for routine introduction of WGS into clinical care pathways.

The Project is divided into 2 work strands: ca. 50,000 'rare disease' genomes (equivalent to ca. 15,000 families) and 50,000 'common cancer' genomes (25,000 patients and their tumours). NE&NC GMC has Rare Disease status (for which I am the Clinical Lead) and is working towards Cancer status (for which I hope we will have an oncology lead). Our role is to coordinate recruitment, sample processing, data analysis and clinical reporting – no mean feat. At present, recruitment is undertaken by a small team of dedicated practitioners, although the goal is to enable clinical teams cross the region to access genomic technologies.

One large component of the Project is to introduce WGS into NHS health care, in order to improve rare disease diagnosis and detect clinically relevant acquired DNA changes in cancers. However, not all patients and families will receive a meaningful result; the second component of the Project is therefore the collection of comprehensive clinical and laboratory datasets which, in combination with whole genome sequence data, will provide a rich research resource to academic and, potentially, commercial organisations. This second component is *not* an NIHR portfolio research project.

*Transformation of the NHS*

The GMCs are effectively being tasked with helping NHS organisations prepare for a future characterised by genomic technology. Some of this 'transformation' is going to be required for patients being recruited into the 100,000 Genome Project: for example, retrieval and export of clinical data (ideally in electronic form), modification of local tumour pathways to enable rapid DNA extraction and processing, 'up-skilling' of the NHS workforce. Again, none of these is a simple challenge.

In addition to the somewhat artificial environment of the 100,000 Genomes Project, it is now time for us as a network of specialities to get to grips with somatic tumour testing (already becoming common- place in some tumours); germ-line genetic testing in some cancer patients (e.g. BRCA1/2 testing in women with ovarian cancer); pharmacogenomics (small scale at present, but set to influence prescribing in most specialties; and migration of microbial diagnostics to rapid DNA-based systems.

## What happens next?

The NE&NC GMC is a network, not just a team based in Newcastle. My team, which *is* based in Newcastle, will need to develop broad and deep links with all NHS organisations in the region and would like to undertake a number of pieces of collaborative work, with approval of each partner organisation. The purpose is to understand current 'readiness' for genomic medicine and encourage the widespread adoption of common systems to facilitate the adoption of genomic technologies across the region.

Some of the areas we will need to explore include:

- local clinical and laboratory data systems and their ability to export data to a common format.
- coding systems being used in clinical data systems (e.g. SNOMED CT)
- local laboratories' progress towards common ISO standards
- local adoption of GS1 bar-coded laboratory systems
- identification of key areas where genomics is likely to impact now and in the future
- workforce skills needs assessment

Importantly, if we can see ourselves as a network, we have the opportunity to share information, to find out what works well, and perhaps what doesn't. We need to work together to prepare ourselves for the inevitable changes in clinical practice that genomics will offer.

So, watch this space: we will be in touch soon to discuss ways in which we can work together with your organisations and specialties, and we are *very* interested to hear you comments, questions or concerns.

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