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Mainstreaming genetic testing: Current issues in the construction of an 'external regime' for the UK

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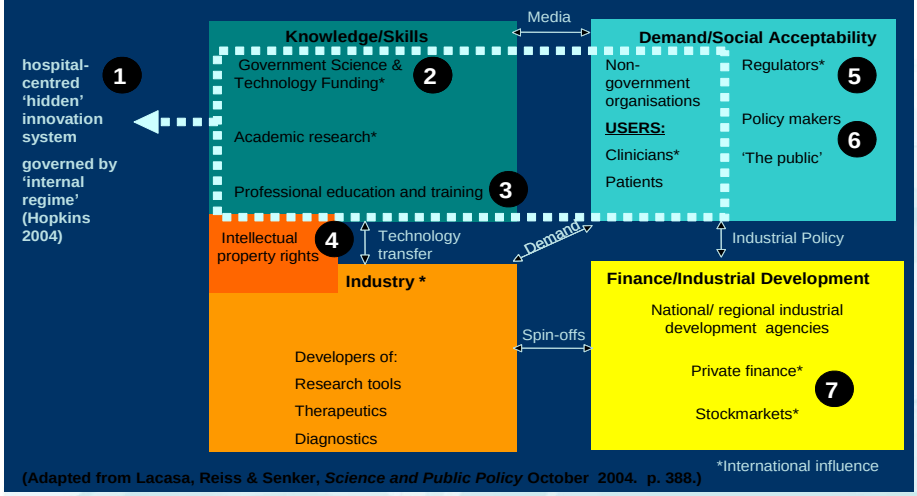
Since the publication of the draft Human Genome and the subsequent UK Government White Paper *Our inherence, our future* (2003) , there has been a surge activity around the development and provision of genetic testing services in the NHS. Building on a doctoral study (Hopkins 2004) of the evolution of genetic testing in the UK, this poster utilises a National System of Innovation framework (Cf. Lacasa, *et al.* 2004) to chart key changes to socio-technical system for UK genetic testing.

Figure 1 sets out the key domains of a national system of innovation relevant for high technology: The base of knowledge and skills, demand/ social acceptability, industry, and finance. Importantly, historically, genetic testing has not relied on all of these domains to reach the clinic. Instead the emergence of UK genetic testing technologies has been characterized as a 'hidden' innovation system (Hopkins 2004 & 2006, NESTA 2006) where governance of innovation takes place within an *internal* regime (dependent on informal self-regulation, ad-hoc funding, and bottom-up initiatives) within hospital-centred networks of researchers, specialist clinicians, charities and later clinical scientists and professional bodies representing the aforementioned groups (See white box by point 1 in **Figure 1**).

This poster illustrates how, in recent years, construction of a new *external* regime (including laws, regulations, social norms) has commenced to accommodate the positions of wider groups (e.g. policy makers, commercial providers, primary care professionals) , transforming the socio-technical system for genetic testing and making its problematisation processes (Blume 1992) more politicised, scrutinised and mainstream than in the previous *internal* regime. Robertson (2007) suggests these changes represent a shift in the governance model, disrupting the established innovation system as the existing expert-elite are increasingly have to share power over the technology.

The most obvious signs of the construction of an *external* regime followed Alan Millburn's (then Secretary of State for Health) announcement, in 2001, that a programme of investment was planned. He said "*I have learned a lot about both the potential and problems associated with developments in genetics….it is time for politicians and the public as well as scientists to engage with the issue*".(speech at International Centre for Life, 19.04.01, Newcastle-upon-Tyne). The Government's subsequent initiatives described in the 2003 White Paper *Our inheritance, our future* attempted to support the new enlarged innovation system by targeting a range of key processes shown in **Figure 1** (especially points 2, 3, 6).

FIGURE 1: KEY ELEMENTS OF A NATIONAL SYSTEM OF INNOVATION



(Adapted from Lacasa, Reiss & Senker, *Science and Public Policy* October 2004. p. 388.)

These include: The creation of the Genetic Knowledge Parks to aid translation of genetic research; Training and initiatives for medical staff; Greater co-ordination of the clinical testing laboratories through the establishment of the UK Genetic Testing Network; Increased support for screening services and commissioning of services amongst other activities. Other earlier efforts to establish key elements of the *external* regime for genetic testing such as the establishment of the Human Genome Commission and efforts to establish and maintain a moratorium on the use of genetic testing in insurance may be thought of as actions necessary to generate public trust (point 6).

The Human Genetics Commission also have called for legal protection against genetic discrimination. This could provide reassurance to individuals and facilitate participation in genetic research (such as UK Biobank) and help to ensure patients are dissuaded from having tests that might benefit them or other members of their family. A public consultation on the 'Single Equality Bill' which move these proposals forward has recently finished.

Since the1990s commercial efforts to identify and patent genes and offer testing services have led to the problematisation of further issues centred on Regulation (point 5), intellectual property rights and the related challenge of technology transfer (point 4). As commercial testing has grown (notably in the USA) concerns of a regulatory 'gap' in the *external* regime have emerged (Hogarth *et al. forthcoming*). Non-Governmental Organisations such as GeneWatch and Which? have become important actors in this debate, pushing for increasing tightening of the regulatory framework for in-vitro diagnostic tests - a major element of the *external regime*. There has also been much concern that innovation will be hindered by a proliferation of patents on disease genes (Kaye *et al.* 2007, Hopkins *et al.* 2007). In particular Kaye *et al.* suggest there is a need for an infrastructure to assist researchers in negotiating the legal aspects of technology transfer.

Finally, with additional public sector funding for translation as yet unidentified (Hopkins 2007) , and a risk of market failure in the commercial incentivisation to develop tests (Martin *et al.* 2006) the mobilisation of sufficient capital from the UK private sector remains a challenge (point 7). As the medical application of genetics has become of wider interest (and perhaps of more relevance to mainstream medicine), so the construction of the *external* regime as led to the generation and reshaping of institutions, practices and networks. We might speculate that the UK has now have developed a new field of *politicogenetics* (Cf. Patternson, 1998) where politics and genetics meet once again after a long period of separation due to the legacy of eugenics (Kevles 1985).

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