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Risk and utility in the governance of diagnostic testing: the case of genetic screening, 1960 to the present

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Routine collection of blood samples from neonates – often using so-called Guthrie cards (pictured) – began in the 1960s when a number of North American and European countries set up screening programmes for phenylketonuria, a rare single-gene disorder which leads to developmental delays and early death if untreated. Such programmes have since been introduced in many other countries around the world. At the same time, refinements in laboratory technology – especially the development of tandem mass spectrometry from the early 1990s, followed by the inception

of increasingly powerful new genomic technologies – have made it possible to detect a growing range of disorders from the same blood samples. These proliferating possibilities have been accompanied by often intense discussion about just what diseases should be included in newborn screening programmes.

During the 1970s and again in the 1990s, these discussions took place in the context of debates about the implementation of genetic screening programmes more generally, including prenatal foetal screening for conditions such as Down syndrome, and adult carrier screening for single-gene disorders such as Tay-Sachs disease and cystic fibrosis. This context was important in setting the terms of debate. Until recently, adult and prenatal genetic testing was chiefly undertaken with the aim of informing reproductive decision-making. Clinical geneticists and genetic counsellors argued that such tests involved significant risk of harm to those tested, including the distress of deciding whether or not to proceed with a pregnancy when faced with merely probabilistic information about the possible outcomes, as well as the danger that clinicians might knowingly or unconsciously pressure patients to make reproductive choices they would later regret. This perception that reproductive tests could cause harm as well as benefit in turn fuelled deliberation about the possible risks and benefits of genetic testing more generally, including newborn screening. By the 1990s, they included a pervasive fear of genetic discrimination, as well as concern that the commercialisation of genetic testing would lead to the introduction of tests offering little or no benefit to patients. In North America and Europe, this resulted by the end of the decade in calls to extend the regulation of diagnostic tests and devices – including but no longer necessarily confined to genetic tests – to include assessment of clinical utility.

Despite sharing common starting points, these debates have played out in different ways in different national and regional settings, reflecting divergences in clinical and public health provision, the relative power of patient organisations in medical policymaking, and the willingness of regulatory authorities to intervene in the marketing of diagnostic tests. There is much here that invites sociological and anthropological investigation, with potentially far-reaching implications for thinking about how diagnostic and other medical tests might be governed in the best interests of individual and public health and wellbeing. So far, this invitation has gone largely unanswered.

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is interested in the development of medical knowledge and medical practice from the mid-nineteenth century to the present. In particular, he uses insights from the sociology of scientific knowledge to examine how developments in medical science have informed and been informed by wider changes in medical practice and medical policy. He currently holds a Wellcome Trust Senior Investigator Award in Medical Humanities for a research project entitled Making Genomic Medicine. This project aims to disentangle the scientific, technological, social and political processes that have led, over the past forty years or so, to the current ferment of activity around medical genomics and so-called genomic medicine.

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