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NEWS AND COMMENTARY

Personal genomics

Integrative genomics, personal-genome tests and personalized healthcare: the future is being built today

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Integrative genomics, personal-genome tests and personalized healthcare, require governance models that treat genome-based information as integral part of health information, on the individual, as well as on the population level. Such models must address needs, as disparate as the knowledge necessary to define a clinically useful 'genomic indication', the personal values and preferences of the individuals concerned, and the interests of a large number of different stakeholders.[1] Obviously, healthcare systems are facing the challenge of fundamental reorganization.[2,3]

This is reflected in two articles in the past issue of the *European Journal of Human Genetics* that discuss the need for regulatory frameworks around 'direct-to-consumer genetic testing'. The article by Christine Patch *et al* and the one by David Gurwitz and Yael Bregman-Eschet take very different directions, while addressing very similar topics. That shows, first, the different interpretation of the captivating world of the new genomics, and, second, the relevance of the particular background of health professionals involved, in terms of the solutions they propose.

Christine Patch and co-workers are from a human genetics background and they are members of the Public and Professional Policy Committee of the European Society for Human Genetics. In their article, they argue for regulatory control of direct-to-consumer genetic testing in Europe. They argue, among other things, that the clinical utility of many genetic tests is still unknown, and that, as only few interventions are available, an unfavorable test result will rarely lead to longer and healthier life. Observing the development of an emerging market of commercial genetic services, they ask for guiding principles to reduce the potential harm stemming from these developments to maintain public trust in genetics. The authors also give a comprehensive overview about current statements and regulations in the US, Canada and Australia concerning pre-market review, quality assurance, and advice and advertising. They point out that, whereas in these countries, as well as in the UK, regulators have placed genetic tests into a higher risk category requiring greater oversight, the majority of tests within Europe are classified as low-risk devices (and processes), meaning that claims are not reviewed before tests are marketed and that test marketing is on the basis of a system of self-certification. The authors mention, as a positive example of regulation, the 'Additional Protocol to the Convention on Human Rights and Biomedicine, concerning Genetic Testing for Health Purposes', which has been adopted by the Council of Europe in 2007. They criticize that genetic tests are increasingly offered through private companies and commercial medical services. Christine Patch and co-authors conclude their article with a call for the harmonization of practices and regulations.

The future is being built now, which direction will it take?

As mentioned, the authors of the first article are from the field of human genetics. This is reflected in the argumentation of these authors, which is oriented to contexts of high risks, genetic exceptionalism and genetic determinism. This line of argument pays little attention to the interplay of genomic and non-genomic factors, for example, environmental health determinants. It also fails to recognize that we do have entered the era of integrative genomics and personal-genome tests.[4–6] Integrative genomics provides us with novel ways of (human) disease classification, as it defines disease expression on the basis of its molecular and environmental elements in a holistic way.[7] Although I do not see compelling reasons for regulatory control as proposed by the authors – as that cannot keep pace with the reality of a dynamic science and thus may even imply 'misdirected precaution'[8] – I do support the demand of human geneticists of being involved in building our future around genetics and genomics. However, I also call for other professionals being involved to ensure that we are doing the 'right' things in the 'right' way.[9]

The second article about regulatory frameworks around 'direct-to-consumer genetic testing' in the past issue of the EJHG has been written by David Gurwitz and Yael Bregman-Eschet with a background in biochemistry and law, respectively. The viewpoints of these authors differ from the argumentation of Christine Patch and her co-authors. The short- and long-term recommendations by Gurwitz and Bregman-Eschet have not only been developed based on the current understanding of genomics, its dynamics and future potential, but the authors also include the new realities such as the leading role of industry and the strongly technology-driven provision of personal genomics services. Whereas Patch *et al* promote the idea of 'control' and 'prohibition', Gurwitz and Bregman-Eschet go for intelligent and innovative business

models such as public private partnerships. They suggest self-regulation of the commercial providers like *23andme, deCODEme and Navigenics* by forming an 'Association of Personal Genome Service Providers' whose members must adhere to agreed guidelines or 'Best Practices'. They clearly state the key difference between personal genomics services and 'classical' genetic testing: for example, the offer of personal genomics services over the web and thereby directly available to consumers, outside of a defined clinical context and often without the involvement of a healthcare provider. They also acknowledge that the personal genomics services provide customers with huge amounts of genetic information, to a large extent still meaningless today, but possibly informative for a large range of clinical, physical and behavioral traits in future. However, should regulators protect individuals from their own curiosity?

Interestingly, the authors do no longer use the term 'genetic test', but rather 'genetic information' and 'personal genomics services', and they argue that the 'genetic tests' offered by the commercial sector are mainly not for health purposes, and therefore are not subject to, for example, the 'Additional Protocol to the Convention on Human Rights and Biomedicine, concerning Genetic Testing for Health Purposes' and to the limitations it entails. As a long-term step the authors suggest adding dedicated personal genomics units to existing regulatory bodies whose mandate would include oversight over the direct-to-consumer personal genomics business.

In summary, concrete short–term, as well as long-term recommendations for protective steps against risks arising from 'direct-to-consumer personal genomics services', as, for example, proposed by Gurwitz and Bregman-Eschet may have the power to provide policymakers in and outside Europe with guidance. Such recommendations meet the current view of European policymakers; classifying the majority of tests as low risk leaves enough room for the dynamics of sciences and the use of personal-genome tests for health and non-health purposes. European approaches like 'Health in All Policies', which had been promoted under the Finnish Presidency, take already into account that there are rarely just and purely health-related issues and purposes.[10] The current problems with the application of European law seem to derive from the uncertainty as to how the regulations can and need to be interpreted.

Nevertheless, there are just two points I would like to add to the debate:

First, the OECD can issue recommendations, but these are not legally binding unless they are transposed by national parliaments. In Europe, the Council of Europe is an often forgotten actor that has the responsibility to guide the European member states by dealing with diversity instead of harmonization. As Europe enlarges, it becomes more and more crucial to understand the different cultures and societies.[11]

Second, the future is being built today! Thus, let us invite all relevant professions and disciplines on board of the boat of genetics and genomics – we urgently need an integrative effort to prepare for the shift to a personalized healthcare that will include personal genome services. Embracing integrative genomics implies turning towards systems biology, as this provides the conceptual framework for genome-based sciences that include highly relevant applications in the field of immunity and infectious diseases.

With an eye for genomics, the public health agenda of all healthcare systems demands a novel vision that reaches beyond the research horizon to arrive at broad application and public health impact[9] ■

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References

1 Knoppers BM: Of genomics and public health: building public 'goods'? *CMAJ* 2005; **173**: 1185–1186.
2 Brand A: Public health and genetics – dangerous combination? View-point section. *Eur J Public Health* 2005; **15**: 113–116.
3 Ellsworth DL, O'Donnell CJ: Emerging genomic technologies and analytic methods for population- and clinic-based research; in: Khoury MJ, Little J, Burke W (eds):: *Human Genome Epidemiology. A Scientific Foundation for Using Genetic Information to Improve Health and Prevent Disease*. Oxford New York Tokyo: Oxford University Press, 2004, pp 17–37.
4 Barabasi AL: Network medicine – from obesity to the 'Diseasome'. *N Engl J Med* 2007; **357**: 1866–1868.
5 Motter AE, Gulbahoe N, Almaas E, Barabasi AL: Predicting synthetic rescues in metabolic networks. *Mol Syst Biol* 2008; **4**: 168.
6 Lunshof JE, Chadwick R, Vorhaus DB, Church GM: From genetic privacy to open consent. *Nat Rev Genet* 2008; **9**: 406–411.
7 Loscalzo J, Kohane I, Barabasi AL: Human disease classification in the postgenomic era: a complex systems approach to human pathobiology. *Mol Syst Biol* 2007; **3**: 124.
8 Prainsack B, Reardon J, Hindmarsh R, Gottweis H, Naue U, Lunshof JE: Misdirected precaution. *Nature* 2008; **456**: 34–35.
9 Brand A, Brand H, Schulte in den Bäumen T: The impact of genetics and genomics on public health. *Eur J Hum Genet* 2008; **16**: 5–13.
10 Schulte in den Bäumen T: Governance in genomics – a conceptual challenge for Public Health Genomics Law. *Ital J Public Health* 2006; **4**: 46–52.
11 Bosch X: Group ponders genomics and public health. *JAMA* 2006; **295**: 1762.