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EDITORIAL

A reply to community genetics: 1998–2009... and beyond

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Dr. Stemerding is to be commended for his paper in which he seeks to reflect on *community genetics* "in the light of the emerging field of *public health genomics*". His evidence comes from an appraisal of the contents of the journal *Community Genetics*. His conclusion is that a tension exists between the "professional endeavour" of community genetics and its function "as a programme aiming at individual empowerment" which, he says, has significance not only for that discipline but also for public health genomics (Stemerding 2010).

So what are these two disciplines, and how do they differ, if they do at all? The founders of community genetics clearly see their's as a "unique concept" with "its own place besides clinical genetics and public health genomics (ten Kate 2008)". They suggest that the "aims of community genetics and public health genetics are not the same, although they have much in common" (Schmidtke and ten Kate 2010).

In my reply, I agree with the author that the tension referred to in the paper applies to both disciplines, but I suggest not just to them alone, but across all medical specialties. I also seek to dispel the notion that, apart from a slight difference in emphasis, community genetics is unique and different from public health genomics. I shall argue that they are in essence one single discipline. Their histories are, of course, clearly different, the one coming from the practice of clinical genetics, with an emphasis on inherited and heritable disorders, the other from the practice of public health, with perhaps a greater interest in common complex diseases, such as diabetes, heart disease and cancer. But I suggest that, history aside, both fields have the same aims, the same tensions, the same problems, and the same aspirations for improving the health of individuals and populations, notwithstanding the fact that emphasis and areas of interest may be slightly different.

Community genetics gave itself a new definition in 2010 (ten Kate et al. 2010) which in essence uses much the same language as the earlier definitions of public health genomics:

> "the art and science of the responsible and realistic application of health and disease-related genetics and genomics knowledge and technologies in human populations and communities to the benefit of individuals therein"

The definition of public health genomics, agreed at Bellagio in 2005 (Bellagio Report 2005; Burke et al. 2006; Zimmern and Stewart 2006) was:

> "the responsible and effective translation of genome-based science and technologies for the benefit of population health"

and prior to that, when the discipline was called public health genetics, an adaptation of the Acheson definition of public health:

> "the application of advances in genetics on the art and science of promoting health and preventing disease through the organised efforts of society"

To the extent that there are differences in language, these focus on the words "to the benefit of individuals therein" as compared to "for the benefit of population health". But this is not, I suggest, a real difference, but one based on a misunderstanding of what is meant by *population health*.

I digress here with a little philosophical musing about populations and individuals. Public health practitioners in

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the nineteenth and the early part of the twentieth century did of course regard *the population* as a single ontological entity. They manipulated the environment which then had an effect on the health of the population, an entity which was conceptually treated as if it were a thing in itself, and by and large homogenous. However, by the latter half of the twentieth century, it was clear that much public health effort and interventions were being directed at individuals through health promotion strategies. Individual behaviour and the idea that individual behaviour was an important determinant of health was very much part of public health thinking and practice. The implication of this was that there came into being an implicit change in the ontology of the population, shifting from being an entity in its own right to being the description of a set of individuals.

This change in conceptualisation has particular relevance to the genomic era, when we now all recognise the heterogeneity of populations and the role played by individual genetic variation. No external determinant will have the same effect on an individual in exactly the same way as it will on another. Biological mechanisms, as was recognised by the great zoologist Ernst Mayr, occur in individuals. The population, he argued, is no more than an abstraction, an average of the individuals within it (Mayr 2004).

Public health practitioners have, in recent decades, recognised the complex relationship between populations, sub-populations and individuals, and have seen their role as one which seeks, both in policy formulation and service provision, to balance appropriately the needs of populations with those of individual citizens and patients. This is of course the tension to which Dr. Stemerding refers, one which has been recognised and dealt with by public health practitioners for many decades. At the heart of this is the emphasis on "autonomy and self-determination as fundamental values" for individuals, but as with all ethical principles, they have to be invariably balanced against other values, some of which may be inconsistent or even directly at variance with the requirements of individual autonomy. Principles are there, but in the real world, choices and judgments have to be made, as individual examples present themselves to us, even if they conflict with each other.

But lest this be thought to be just a dilemma for public health practitioners, it is a fact that clinicians and clinical geneticists in both developed and developing countries have encountered this across their working lives. Whether implicitly (as was the case in earlier years) or explicitly, they have had to balance the service they deliver to the individual patient in front of them with the needs of the larger population that they serve. The prioritisation of resources, whether of time, skills, services or money, in order to achieve the proper balance between populations and individuals, and between one individual and another has been part of clinical practice for many decades.

In the context of clinical genetics, this tension is often played out over the issue of reproductive choice. Informed consent is now a driving force, one accepted by public health practitioners and by the public health genomics movement. The reduction of the birth prevalence of inherited disorders will be welcomed by both practitioners of public health genomics and community genetics (whether they regard it as the primary aim of a programme or merely a consequence), but both will insist that such reduction is legitimate if and only if this comes about as a consequence of real parental choice, without coercion and without deception. Indeed, the experience that public health practitioners have in the balancing of values has enabled them to participate in debates surrounding reproductive choice and other matters such as consent for genetic testing, genetic testing for minors and the establishment of biobanks. They participate in these discussions with as much knowledge and understanding as clinical geneticists, and holding, I would suggest, the same set of ethical values.

The community genetics community embraces the need for evidence and for the responsible application of genomic knowledge for the benefit of their patients. This again is no different to the attitude of public health genomics and their requirement for evidence-based practice and policy. But rather than this being seen as a tension between evidence-based decision making and "individual decision making" (as it is termed in the paper), evidence-based medicine should be regarded as an aid, as a piece of data input, to help inform the judgments of clinicians and policy makers.

The quote from Laberge in the paper, that "in public health genomics too, personal responsibility and empowerment are promoted as final objectives, making public health eventually the result of individual decisions of citizens" is a concept that I thoroughly agree with. Indeed, I have spoken in public of a view that public health practice in future years may revolve around the information engineer (as distinct from the sanitary or social engineer of the nineteenth and twentieth centuries) whose role is to provide, in a readily understandable format, the evidence for the effectiveness of interventions so that individual citizens may take responsibility for their own health and health care, with clinical professionals as advisers who will guide them in the interpretation of the evidence.

It may perhaps be useful also to reflect on the distinction between the words *genetics* and *genomics*. There are no absolutes in the use of words, so I make no absolute claim about the correctness of my usage. But I find it helpful to understand that the word genetics has historically referred to matters that pertain to inheritance, so that genetics is primarily about inherited or heritable disorders and conditions: hence, the specialty of clinical genetics. By contrast, the word genomics is, for me, about the broader matter of DNA and the genome, and primarily focuses on the part played by

genetic variance and its role in health and in the pathogenesis of disease. It is for this reason that people speak of the new specialty of medical genomics, rather than medical genetics. Clinical geneticists will always be needed to pronounce on decisions about inheritance and the management of family members rather than just the patient in front of the clinician. But as we understand more and more about cellular and molecular mechanisms of disease, physicians in all specialties will need to use genomic concepts in their diagnosis and management of their patients.

When I last wrote about the relationship between community genetics and public health genomics, I conceptualised community genetics as that subset of public health genomics that concerned inherited disorders and the practice of clinical genetics in a community setting. The new definition (ten Kate et al. 2010), supplemented by Dr. Stemerding's findings, appears to go beyond its historical roots and what I took at the time to be its focus. As set out now, the definition accorded to it appears to be indistinguishable from public health genomics, a discipline which has come of age, and with its own tradition of literature (Khoury et al. 2000; Burke et al. 2006; Stewart et al. 2007; Stewart et al. 2009). My own reading of the journal *Community Genetics* is that its focus (although not entirely) continues to be on the subject matter of inherited disorders, but I welcome the notion that it seeks to take on a wider brief.

I therefore welcome the aspirations of the community genetics community, I welcome their expertise and focus, and I welcome the fact that in them we have close colleagues. To unite gives greater power and increases our chances of achieving our goals. I am thus perplexed as to why they seek to divide and claim that their discipline is unique and different from public health genomics. If there are differences, surely they are only a matter of emphasis.

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