

Community genetics: 1998–2009...and beyond

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In recent years, public health genomics has been introduced in the scientific literature as a new endeavour, aiming at the translation of genome-based knowledge and technologies into health interventions and public policies for the benefit of public health (Brand and Brand 2006; Zimmern and Stewart 2006; Gwinn and Khoury 2006). In 2009, *Public Health Genomics* started to appear as an international journal and a new signpost of the emerging field; however, as the editors pointed out, the new journal builds on an earlier version which was already founded in 1998, published as *Community Genetics* (Knoppers and Brand 2009). Thus, as a new and emerging field, public health genomics does not only embody promises and expectations for the future. It is also rooted in a history of past attempts and achievements, constituting “community genetics” as a bridge between genetics and public health (ten Kate 2005). In this context the relationship between public health genomics and community genetics has become a matter of debate. As becomes clear from the establishment of the new *Journal of Community Genetics*, there is a continuing interest in community genetics, defined by aims independent from public health genomics. In an interesting sociological commentary in the first issue of this journal, it is indeed observed that we should not take for granted that “public health genetics and community genetics could be viewed as one and the same” (Raz 2010). In a farewell editorial, published in the final issue of the former journal *Community Genetics*, Leo ten Kate likewise emphasized that community genetics “is not just a name but a unique concept, which has

its own place besides clinical genetics and public health genetics or genomics” (ten Kate 2008, see also Schmidtke and ten Kate 2010; and ten Kate et al. 2010).

In this commentary, I will take a closer look at the uniqueness of the concept of community genetics, using the 11 volumes of the former journal *Community Genetics* as my primary source material.¹ My aim is not a complete review of the contents of this journal, which would be an impossible task, but a discussion of some aspects and questions which I see as particularly interesting and significant for our understanding of the concept and agenda of community genetics. What can we learn from the history contained in this former journal about the particularities of community genetics and its relation with the emerging field of public health genomics? Most revealing in this history is the tension between a conception of community genetics as a professional and regulated endeavour and as a programme of individual empowerment. Although we can see this tension as a unique feature following from the concept and agenda of community genetics, it is also highly significant, as I will argue, for the future prospects of public health genomics.

The agenda of community genetics

The ambitions of community genetics as a field can be defined in terms of four movements or shifts which characterize the activities of its practitioners as distinct from the traditional practices of clinical geneticists (ten Kate 1998; Brisson 2000). The first of these movements is

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¹ The journal *Community Genetics* started to appear in 1998 and has been succeeded in 2009 by the journal *Public Health Genomics*. In total, 11 volumes have been published, including 46 issues.

a shift in focus away from individuals to populations, bringing genetic services to the community as a whole. Implied by this movement is a shift from people with symptoms to people without symptoms, whereby the initiative is coming from the care system. The third movement is a shift from reproductive choice as a main focus to options for prevention of disease, and, in relation to this movement, we might also mention a fourth shift, from rare monogenetic disorders to multi-factorial forms of common diseases. This latter shift, however, seems at present more a prospect than reality (ten Kate 2001; Brand et al. 2006).

Although the first two shifts are clearly defining the agenda of community genetics, it is the third shift—from reproductive choice to prevention of disease—which brings us to a question that is most revealing and significant for the ambitions of the field. Traditionally, reproductive choice has been a major focus of clinical genetic services and community genetics, likewise, has its roots in population carrier screening programmes offering reproductive choice (Modell and Kuliev 1998). Accordingly, in the volumes of *Community Genetics* we see a continuing interest in developments of carrier screening and prenatal screening. Community genetics, however, is also clearly inspired by notions of public health, aiming at health promotion and prevention of disease. Thus, as some authors in the field have argued, programmes offering reproductive choice should not be part of the community genetics agenda because the aims of such programmes cannot and should not be understood in terms of prevention (Khoury et al. 2000; Holzman 2006). In the journal *Community Genetics*, a tension between the aims of prevention and reproductive choice has indeed been noted as a point of discussion and concern (Nordgren 1998; Lippman 2001), but more importantly, the journal has also been instrumental in attempts to reconcile these different aims by emphasizing *informed choice* as a key concept in community genetics (ten Kate 1999, 2000, 2005; Henneman et al. 2001). This principle is of crucial importance, as I will argue, for our understanding of the impact of community genetics in society.

An examination of the variety of practices that are discussed in *Community Genetics* again reveals that the aims of the field do not correspond in any straightforward way to a public health agenda in a strict sense. The practices described in the different volumes should not be understood just in terms of traditional public health aims, but rather as a new way of working which involves the system of health care as a whole. Thus, we find not only discussions about the ways in which advances in genetics may be integrated in public health. We also find discussions about genetic service provision in clinical care, focussing on common diseases like cancer and heart disease, and as

the most important subject, we find quite a lot of papers about ways in which genetics relates to practices and perspectives in primary care.²

The new way of working that is promoted by community genetics can be defined as involving the identification of genetic risk groups in the community. In this approach, individuals who may not be aware of being at risk can be offered information about their genetic status and potential options for prevention. This way of working indeed marks some of the more salient shifts characterizing the ambitions and activities of community genetics. Instead of waiting for people coming with complaints to the consultancy room, individuals now have to be actively approached by professionals in the care system (ten Kate 1998). This brings me to another observation about the contents of the first 11 volumes of *Community Genetics*. It is interesting and significant that a large share of the papers published in the journal is devoted to questions relating to the users that community genetics should serve.³ Obviously, because individuals are targeted without symptoms or complaints, the needs, experiences and wishes of these prospective users have become a highly relevant concern for the proponents of community genetics. Again, this is a point of crucial importance for our understanding of the future impact of the field.

Future prospects of community genetics

Taking these observations as a starting point, I will now consider two possible scenarios as potentially relevant futures for community genetics. The future that is implied in the agenda of community genetics, obviously, is a future in which it is the health care system through which new applications of genetic knowledge are made available to individuals in the population in an ‘evidence-based’ way (Blancquaert 2000; Baird 2001; Gwinn and Khoury 2006). Accordingly, it is the professional who should decide for whom particular applications might be needed and useful; however, in discussing the role of community genetics in society, several authors also refer to the possibility of another future scenario. In this scenario, genetic tests are becoming more easily available through commercial providers offering their products on the market direct to ‘consumers’ who are willing to pay for it

² As a rough estimate, we can say that of the 430 items that appeared in *Community Genetics* from 1998 to 2009, 8% was explicitly devoted to the role of genetics in public health, 5% to genetics in clinical care and 7% to genetics in primary care. Not included in these figures are the items focusing on genetics in reproductive care (13% of the total number). See also ten Kate (2007) for an overview of the contents of the first nine volumes.

³ Indeed, of all the 435 items mentioned in note 2, 14% explicitly focused on the variety of users in terms of particular risk groups, minorities or communities to be served by community genetics.

(Holzman 1998; Williams-Jones 2003). From the point of view of community genetics, this prospect is clearly seen as a threat that has to be averted by sound policies of regulation (Ronchi et al. 2000; Guilloid 2000; Holzman 2006).

Community genetics, in other words, will have to be developed in a societal landscape offering a variety of contexts in which applications of genetic knowledge may become available to future users, both inside and outside the health care system. One element in this landscape which will shape future applications is governmental regulation. Another element is the growth of commercial services, offering genetic tests on an international scale through the internet. What is the relevance of these observations for our understanding of the future impact of community genetics? There are two points which I see as most important here, one of which goes down to the heart of community genetics itself. The first point is that it will be very difficult, if not impossible, to resist by governmental regulation a growing commercialisation of genetic services on a global scale. Moreover, and this is my second point, a scenario like this will become all the more probable in a world governed by a principle of informed choice, the very principle adopted by community genetics as its key concept.

Community genetics, we may say, is based on an individual rights perspective, emphasizing autonomy and self-determination as fundamental values. Traditionally, individual rights have been conceived as a way to *protect* individuals against interventions—medical or otherwise—that may be harmful or unwanted, but as we may learn from the contents of *Community Genetics*, individual rights can be understood in terms of *empowerment* as well. Thus, empowerment has been intimated in the journal as serving the aim of equal access to services, information and choices for individuals from diverse communities. In a world where respect for individual autonomy is not universally accepted and where we find many disadvantaged populations and communities, both protection and empowerment are of course highly relevant concerns (Wertz and Fletcher 2004), but as observed in *Community Genetics*, in a particularly thought-provoking contribution, the notion of empowerment may also take on a different, more radical and problematic meaning (Caulfield and Wertz 2001). In this guise, it serves as a perceived *right of access* to services for everyone who—for whatever reasons—might want to. From this perspective, reasoned attempts to restrict access or protect individuals may easily be branded as paternalism. Needless to say, this notion of empowerment fits nicely with the aims of commercial providers of genetic tests (Parthasarathy 2003).

A tension between regulation and empowerment

Let me sum up at this point what I see as some of the more striking issues emerging from the first 11 volumes of

Community Genetics. In my discussion, I focused on the agenda of community genetics involving a quite complex picture of a broadly conceived entrenchment of genetics in the system of health care. I added to this picture some observations about the societal landscape in which this agenda will have to be realised. From this picture emerged an important tension between regulation of health care services on one hand and empowerment of individual health consumers on the other. This tension not only characterizes our modern health care landscape. It is also manifested in the community genetics agenda itself, revealing a clear ambivalence between community genetics as a professional and regulated endeavour and as a programme of individual empowerment. Another, interesting and significant manifestation of this ambivalence is the way in which prospective users are represented in the volumes of *Community Genetics*. As I noted, the needs and wishes of users appear in the journal as a highly relevant concern, but what is most revealing in this respect are the various ways in which users are defined, ranging from patients (Emery et al. 1998) to publics (Henneman et al. 2004), citizens (Godard et al. 2007), clients (Detmar et al. 2008) and, indeed, consumers (Terry and Davidson 2000).

What about public health genomics?

The starting point of my commentary and the exploration of the contents of the journal *Community Genetics* was the question of the uniqueness of the concept of community genetics, especially in relation to public health genomics as an emerging field. One way to understand this uniqueness is in terms of the origin of the field. Community genetics has been positioned as a bridge between clinical genetics and public health (ten Kate 2005). In other words, community genetics is rooted in a tradition of individual care embodied in clinical genetics, and it shares with this tradition informed choice and individual empowerment as the major aims of the field. In this respect, community genetics may be contrasted to public health genomics, even though both fields share the aim of integrating genetics in public health. Firmly rooted in a public health tradition, public health genomics emphasizes the improvement of population health as its key objective. Indeed, the focus on health from a population perspective is exactly the reason why proponents of the field prefer to name it ‘public health genomics’ instead of ‘community genetics’ (Knoppers and Brand 2009).

In adopting informed choice as a key concept, community genetics not only distinguishes itself from public health genomics, but it also highlights an important tension between professional regulation and individual empowerment; however, in this latter respect, community genetics involves a

challenge that is also highly significant for our understanding of the future prospects of public health genomics. Moving from opposite starting points, community genetics and public health genomics, in a common endeavour to integrate genetics into public health, to some extent are heading for a similar approach. I have described the agenda of community genetics in terms of different movements, including a shift in focus away from individuals to populations. In similar terms, we can describe the programme of public health genomics as a movement from the population level to a more individualised approach. Thus, it is stated as the “holy grail” of public health genomics that, based on a fuller understanding of genetic and environmental factors involved in the causation of disease, it will be possible to devise effective preventive interventions targeted at individuals with specific genotypes (Zimmern and Stewart 2006). In other words, instead of the traditional “one size fits all” stance underlying whole-population strategies in public health, public health genomics promises a more nuanced approach that incorporates differences in individual susceptibility as opportunities for individualised prevention (Bellagio report 2005). Accordingly, we can observe 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 (Laberge 2002).

Another more obvious point, on which community genetics and public health genomics agree, is the belief that genome-based information or interventions should be introduced only in an ‘evidence-based’ way. In this regard, the endeavour of public health genomics obviously also involves a potential tension between the aim of evidence-based interventions and a focus on individual decision making and personal responsibility. Compared to community genetics, this tension may become even more challenging because in public health genomics, as authors about the field contend, “it may be several decades before the scientific basis for the ‘predict and prevent’ scenario can be adequately evaluated” (Stewart et al. 2007). In other words, the so-called “translation highway” of genomics in public health appears to be a long and winding road to a distant and uncertain future (Gwinn and Khoury 2006; Khoury et al. 2007 and 2008). In this context of high expectations and major uncertainties, the more immediate future of public health genomics will not be shaped by evidence-based professional strategies of personalised prevention, but will primarily depend on the initiatives of commercial providers of genetic information and, of course, on the appeal of their services to individual health consumers. In this context, we may also expect ongoing conflict between those developing new genome-based technologies for the health care market and those who have to evaluate these technologies from an evidence-based public health point of view (Woodcock 2008).

Facing the challenge

In my account in this commentary of the concept and agenda of community genetics, I have revealed a tension which also points to an important future challenge for the emerging field of public health genomics. Is there anything for us to learn from the experiences in the field of community genetics that might suggest ways to bridge potential conflicts between policies of regulation and the empowerment of individual users? This seems to me a most interesting and critical question for community genetics in the future.

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