Commentary: a sociologist's view on community genetics

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Abstract This commentary illustrates and discusses potential research directions for sociologists and anthropologists interested in the field of community genetics and its emerging networks of individuals genetically at risk. Community genetics—the application of medical genetics in community settings for the benefit of individuals—also involves social issues of lay-professional misunderstandings (and more recently also the different perspectives of various expert communities), stigmatization, discrimination, and medicalization. Focusing on a socio-anthropological perspective regarding the views and disagreements surrounding the definition and scope of community genetics, I overview several epistemological, methodological, and practical contributions that such perspective can offer to the study of community genetics.

Keywords Community genetics · Public health genetics · Sociology and anthropology of medicine · Bioethics · Patients' organizations

In this commentary I would like to illustrate and discuss potential issues of concern and research directions for sociologists and anthropologists interested in the field of community genetics and its emerging networks of individuals genetically at risk. Community genetics can be generally defined as the application of medical genetics in community settings for the benefit of individuals. It includes such activities as genetic testing, screening and counseling, DNA registries, and public and health professionals' education on genetic issues. Community genetics thus emerges as a distinct field requiring specific expertise in-between medical

genetics and public health. Moreover, the activities involved in community genetics all have various social, psychological and ethical implications. These activities, which are well-intended and have an important role in the reduction of suffering, can also carry unintended and adverse consequences, especially if health professionals are not fully aware of the social aspects and implications of such activities. These social aspects include issues of lay-professional misunderstandings (and more recently also the different perspectives of various expert and lay communities), stigmatization, discrimination, and medicalization. These are issues that have always interested and engaged sociologists and anthropologists of health and illness. To study these aspects in the context of community genetics requires a paradigmatic shift from the conventional *individual* level of genetic testing concerning, for example, compliance (or resistance), stress (or empowerment), self-image, and so on, to a middle-range level embedding such analyses within local communities rather than restricting them to the counselor–counselee interaction.

Community genetics offers social scientists the opportunity to explore the dynamics of genetic networks of interaction, their social influence, and the agency with which people and groups respond to them in various ethnic, religious, political, and condition-specific contexts. To reduce misunderstandings, we need to understand better how different people and communities interpret what is “a carrier” and what does a “gene for” mean (Condit 1999, 2007). We also need to take into account the blurring of the conventional lay/professional divide, which is occurring as a result of the growing knowledge and involvement of individuals at risk and patients' organizations in genetic research, testing, and education (Novas 2006, 2007; Raz 2009). Studying the consequences of targeting at-risk communities by health professionals can teach us important lessons about genetic stigmatization (Wailoo and Pemberton 2006). To counter-balance medicalization, we need to study how medical

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genetics is embedded by people and communities within their normative practices of marriage patterns, family relations, and family planning (Raz 2005; Featherstone et al. 2006). In order to seriously take into account the disability perspective (Parens and Asch 2000), we need to understand better the views of patients' organizations and the disability community concerning issues of care and prevention (Davidson et al. 2000). This is evidently only a very partial list of socio-anthropological research directions involving community genetics.

This commentary is composed of several sections highlighting inter-related sociological aspects of community genetics. The first section deals with the views and disagreements surrounding the definition and scope of community genetics. It would be unrealistic to expect sociology, where scholars still debate the meaning of “community,” (let alone “community genetics”), to provide a solution to these disagreements. However, a socio-anthropological perspective can provide us with valuable epistemological, methodological and practical contributions. From the viewpoint of the sociology of science, the formation of the field of “community genetics” offers a case study of on-going boundary-work in negotiating the borders and territories of scientific disciplines (Gieryn 1983, 1999). I will ask in this context how community genetics, with its particular aims and scope, is emerging between clinical genetics and public health genetics (Knoppers and Brand 2009; ten Kate 2008).

The following two sections highlight the social and ethical aspects of community genetics and the initiatives conducted within and by genetic communities. Our social and communal narratives of risk and responsibility affect whether and how to implement, regulate, and use genetic testing and screening. Different schemes of genetic screening and testing that take place in community settings are thus influenced by social issues including, for example, individual autonomy; the cultural sensibility concerning “ethnic profiling”; the political and gendered relations of power and authority that underpin patterns of consanguinity as well as the attempts to change them; and the moral and legal standing of the fetus as shaped by communal and religious norms regarding “therapeutic” abortions, which may promote or hinder the use of preconceptional and/or prenatal genetic testing.

The final section focuses on methodology and implementation, suggesting that the use of sociological and anthropological methods is necessary for gaining a comprehensive view of people's motivations, attitudes and behavior within networks of genetic risk. This can further contribute to a more effective implementation of programs and services of community genetics in terms of public understating, individual uptake, informed consent, and informed choice (Marteau et al. 2001; Henneman et al. 2001). Sociology and anthropology can thus help in

sustaining bridges between communities and medical genetics (see also Cowan 2008; Raz 2009).

The aims and scope of community genetics

Community genetics can involve a public health activity, a clinical program, and a grassroots initiative. This field thus involves a plurality of disciplines and stakeholders. In the medical realm, clinical geneticists, public health scientists, and social epidemiologists are debating about what is a good definition of this new hybrid specialty (Knoppers and Brand 2009; Gollust et al. 2005). For some North American proponents of public health genetics, what they regard as a terminological debate will be corrected when community genetics merges into public health genetics. Public health genetics is a term mostly used in the USA, and community genetics is a term mostly used in Europe; however, we should not take for granted that “public health genetics and community genetics could be viewed as one and the same” (Khoury et al. 2000: 5). For European proponents of community genetics, the term denotes an independent orientation that needs to be acknowledged in developing a medical field that should ideally combine the best from clinical genetics and public health (ten Kate 2005; Clarke 1997). The intricacies of this debate, although important, are beyond the scope of this commentary (but see Raz 2009 for elaboration). In its “Aims and Scope,” the *Journal of Community Genetics* presents a clear voice for the community approach, continuing the mission that was carried before by “Community Genetics” (the journal published in 1998–2008 by Karger AG) in studying “the art and science of applying medical genetics to human communities for the benefit of their individuals.”

Such an approach carries a special appeal for sociologists and anthropologists studying the social aspects of genetic testing, screening and counseling. Clinical genetics ideally aims for non-directive individual counseling promoting informed consent and choice, while public health is traditionally population-oriented and promotes the goal of reducing the prevalence of disease (a goal which some clinical geneticists may also identify with). However, the actual application of informed choice into clinical genetics is still in its early development. Down's syndrome, for example, has been viewed for most of its history as a public health problem, with prenatal screening aimed at reducing its incidence (Raffle 2001). As Bryant et al. (2008) show, only relatively recently have there been efforts to promote reproductive choice rather than test uptake as the preferred measure of this screening program's success. A recent report of The Health Council of the Netherlands (2008) claims that the quality of “informed choice” is critical in determining whether screening for Down's gives the

pregnant women taking part in it more advantage than disadvantage.

Importantly, a sociological perspective should remind us that community genetics should be a co-production of medical geneticists, communities and individuals. It thus cannot be owned solely by any one party. While medical science traditionally prioritized the viewpoint of health professionals, a sociological perspective should alert us to the important role of community members and patients' organizations in the co-production of community genetics.

Genetic communities and patients' organizations

The expert perspective of medical scientists has been, and still is, quite dominant in shaping the reality of community genetics. The social construction of community genetics has been conducted, for a long time, primarily through the medical gaze (Novas and Rose 2000; Lemke 2002, 2005). However, in recent years we have seen an increasing influence and involvement of patients and patients' advocates, members of support groups, patients' organizations, and communities at risk (Novas 2007; Novas 2006). This brings us to the important issue of genetic alliances and the needed cooperation with people and associations to promote the concept of community genetics. "Genetic communities" can come into being through the interpersonal mediation of illness experiences, the creation of collective identities, and the political mobilization of such identities.

A necessary starting point is the realization that genetic communities are polymorphous—they come in many shapes and sizes. We used to think about genetic communities mainly as a juxtaposition of a common genetic pool, ethnic bonds, and geo-political circumstances—for example, Ashkenazi Jews or British Pakistanis. In this more "traditional" view of community (I am bracketing the term since there are many manifestations of this phenomenon in "postmodern" societies), pre-existing social categories influence their members' behavior through pre-existing communal norms. For example, work with British Muslim communities showed that families may be reluctant to disclose genetic carrier information because of the stigma attached to disclosing illness in the family, and the links to family honor and marriage prospects (Shaw 2000, 2001; Rozario and Gilliat-Ray 2007; Shaw and Hurst 2008a, b). The use of carrier screening to inform cousin-marriage in consanguineous communities is a complicated issue that raises questions such as whether to support traditional patterns, confront them, or attempt some culturally adapted intervention, such as premarital carrier screening (Raz 2005; Modell and Darr 2002). However, there are various other genetic communities that are in the process of being

constructed and where group members are involved in the construction and negotiation of new norms. Consider, for example, the globalizing Deaf (with an uppercase "D") community, where congenital hearing loss is not regarded as a disability but as a cultural difference that does not require genetic testing (Padden and Humphries 1988). Many deaf people marry another deaf person and many feel that it would be more desirable to have deaf children (Bienvenu and Colonomus 1985). Recently, novel genetic communities are also in the process of being formed around consumerist practices of genetic testing. There can be communities of those similarly at risk and of participants in trials of new therapies. This dynamic array of genetic communities is a fascinating field of study for the sociology of community genetics.

Patients' advocacy organizations and support groups are going to have an increasingly important role in the construction of genetic communities. Sociologists of community genetics must acknowledge this role, study it and give it the voice it deserves. Patients' groups that form "genetic alliances" can influence genetic and medical research according to their preferences by being involved in funding and public relations (Rabeharisoa and Callon 2002; Rabeharisoa 2003). There are many examples that cannot be listed here of grassroots patients' organizations dedicated to initiating and funding genetic research while at the same time providing support, advocacy, and education to affected individuals (e.g., Terry et al. 2007; Terry 2003; Stockdale and Terry 2002; Ekstein and Katzenstein 2001). Such patients' organizations can also manage DNA repositories; acquire patents in relation to methods for diagnosing and treating their relevant genetic condition, and lobby medical authorities to issue guidelines for the genetic testing of the relevant condition (Terry and Davidson 2000; Novas 2006). A sociological focus on this new phenomenon entails various intriguing research questions, such as what kind of a process of organizational maturation do genetic alliances undergo? Do they succeed in preserving a grass-root, democratic organization that represents the range of voices present within patient communities, or are they also subjected to processes of institutionalization, and bureaucratization, especially when commercial and pharmaceutical interests are involved (see Nelis et al. 2007)? Are they becoming increasingly similar to each other as a result of parallel pressures in this organizational field? Does this involve organizational mimicry or normative isomorphism to established medical agencies and hospitals, for example patient advocacy groups which may wish to establish an ethics advisory board for themselves (see for example the case of the Alpha-1 Foundation in Sharp et al. 2008)? What kind of moralities are emerging within such organizations, for example in terms of genetic responsibility? Are they similar, or varying according to disease

severity, availability of treatment, and so on? What are their attitudes towards the conflict of care and prevention, and towards the controversial notion of eugenics?

In addition, a growing number of companies now provide personal genetic information direct-to-consumers. This new form of “collaborative science,” as some see it, offers a basis for a new form of community—an internet-based, virtual network based on beliefs of common genetic susceptibility that links risk, disease, and group identity (Lee and Crawley 2009; Prainsack et al. 2008). There is real potential for collaborative science and consumer empowerment in such initiatives; it would be fascinating to follow, for example, the development of the recent collaboration between PatientsLikeMe, a community-based personalized medicine platform for people with life-changing conditions, and 23andMe, a leading personal genomics company, which have announced a partnership to help people with Parkinson's disease by “recruiting 10,000 people with Parkinson's for a massive study of the disease, and give patients a way to learn more about their personal genetics” (see <http://www.reuters.com/article/pressRelease/idUS165989+09-Jun-2009+MW20090609>). Critical sociologists might argue, however, that the main question here is, as always, *cui bono*? At the end of the day, these virtual genetic “communities” that present themselves as social experiments in the democratization of medical science may turn out to be a sophisticated capitalistic form of “bio-prospecting.” These are intriguing new territories that need to be empirically explored by a sociology of community genetics that is alert to the delicate balance between bio-hope and bio-prospecting.

Bioethics of community genetics

In terms of bioethics, an emphasis on *community* genetics appears to locate questions of beneficence, non-maleficence, autonomy and perhaps even justice in the context of *communitarian* ethics. Genetic programs may be endorsed by *communities* in spite of limits on individual choice (Dor Yeshorim, the ultra-orthodox Jewish community-based program, and the Cyprus screening program are well-known cases in point, see Raz and Vizner 2008; Bornik and Dowlatabadia 2008; Prainsack and Siegal 2006). Such cases seem to violate the universal ethics proposed by liberal individualism and the principlist approach in bioethics. The sociological study of community genetics, where descriptive observation and normative judgment are often interwoven, can thus also join and contribute to the challenge recently made by sociologists, anthropologists and bioethicists who seek to complement the liberal “view from nowhere” with a more socially grounded “view from here” (De Vries et al. 2007).

Furthermore, in shifting from the health professionals' point of view to the lay perspective we also turn from the well-established expert ethics of care to the less familiar lay moralities of responsibility (Raz and Schicktanz 2009). In recent years we see an intriguing convergence of social science and bioethics that is taking place in this context (WHO 2002; Haines 2002; Haines and Williams 2007). Studies of lay moral argumentation concerning new technologies of genetic testing have been conducted, although with a broad public rather than in the context of particular genetic communities (Scully et al. 2004; Banks et al. 2006). A focus on community genetics joins this thrust by stressing two major paradigmatic changes. Firstly, we add to the expert ethical discourse of abstract and formal principles the moral argumentation of lay people in genetic communities, which is usually ambivalent, informal and “unprincipled,” but is nevertheless the morality that people live by. Secondly, we add a contextual focus on social groups as reference points. On the one hand, genetic communities are evidently different from each other and we should examine the effect of such cultural variation in the context of lay (rather than professional) attitudes. On the other hand, genetic communities and networks of individuals affected by genetic risk raise the question of underlying similarities perhaps stemming from the lived experience of being affected (Kleinman et al. 1997; Kleinman 1999). For example, we could ask whether there are similar outlooks shared, by virtue of being affected, by the 600 or more patients' organizations that form the Genetic Alliance in the USA, as well as what are the variations caused by the type of disease, the ethnic composition of the group, its religion, traditional norms, consumerist motivations, and so on. Such questions can lead to broader concerns and comparative cultural narratives underlying lay moralities, thus providing an important contribution to the study of “empirical ethics” (van der Scheer and Widdershoven 2004; Borry et al. 2005).

Methodology and implementation

The contribution of sociology and anthropology to community genetics should go beyond the statistical analysis of uptake, compliance, service delivery, and so on. Although these data are extremely important for establishing an objective baseline for assessment, they do not present the whole picture. Emphasizing only the statistical aspects of behavior, which is in line with the historical positivism of the medical sciences, does not realize the full interpretive potential of social science. Community genetics is an inter-subjective arena of negotiation among different, often contesting, professional and lay languages, moralities and worldviews. While counselees or individuals affected by genetic risk can acquire a commonality of perspective with

health professionals by learning and developing together the symbols by which aspects of the world are identified, still many would define the situation in quite different terms (Michie et al. 1997). The communication of genetic risk, for example, requires that counselors translate statistical and biomedical knowledge into more familiar notions (Rapp 1999; Shiloh 1999). An interpretive analysis is needed in order to better understand the ways in which social and ethical considerations involved in community genetics acquire situated meanings within institutional and political contexts. It is also required for realizing the different meanings of genetic testing for the participants, and the different arenas (public and private) in which these meanings are negotiated within the community or the network of individuals and families that are affected by genetic risk (see for example Atkin et al. 2008; Pilnick 2002; McAllister 2001; Featherstone et al. 2006; Raz 2005).

The issue of implementation evidently deserves a commentary of its own and I can only briefly touch on it here. An important question regarding the implementation of community genetics concerns the issue of health policy and regulation (Knoppers and Isasi 2004). How should we balance the interests of individuals, community leaders and community members, health professionals and health officials, in the context of community genetics? Countries differ greatly in the policies, regulation and legislation they employ in regard to the delivery of carrier screening and testing for reproductive purposes. Israel and the USA, for example, present quite opposite alternatives in regard to carrier screening. In Israel, carrier screening for Tay-Sachs, CF and Familial Dysautonomia is nationally provided to the Jewish population by a governmental body, the Department of Community Genetics in the Ministry of Health, which also provides specific carrier screening and counseling services targeting particular at-risk Arab-Israeli and Bedouin communities (Zlotogora et al. 2009). In the USA, there are no state-administered community-based programs and community-based carrier screening is organized solely by patients' organizations and privately provided (Raz 2009). Whereas the Israeli Ministry of Health administers a national program for preconceptional CF screening (giving couples at risk the possibility to decide between the different options *before* the birth of an affected child), the USA and many other western countries focus on neonatal CF screening (Zlotogora 2009).

Such different policies are influenced by medical concerns as well as social perceptions and economic arrangements. Individual autonomy, being a central social, bioethical, and legal principle in Western countries, explains why these countries avoid mandatory screening which takes place, for example, within premarital genetic counseling in Iran, Tunisia, and Saudi Arabia (Samavat and Modell 2004; Chaabouni-Bouhamed 2008; Alhamdan et al. 2007), and in China until recently (Ebrahim et al. 2006).

The Cyprus thalassaemia screening program represents a mixture of mandatory screening and voluntary testing: When it began in the 1970s the program consisted of Church-mandated carrier screening; then, once prenatal diagnosis became available for thalassaemia, the majority of couples at risk who were detected prior to marriage nevertheless proceeded to marry, and today they also have the option of PGD. In this manner, flexible upgrading of the genetic testing technology was accompanied by increasing personal autonomy (Cowan 2008; Bornik and Dowlatabadia 2008).

Yet another social influence is the cultural sensibility concerning “ethnic profiling,” built on the trauma of sickle cell screening in the 1970 among African-Americans (Wailoo 2001), which has arguably limited community-based carrier screening in the USA. The controversies that erupted over sickle-cell carrier screening continue to have a negative impact even after this program had disappeared. The notion of a “community-based program” is hence frowned upon (Bornik and Dowlatabadia 2008) and even when carrier screening is being recommended by American medical associations, as in the case of CF screening to Caucasian couples, US governmental bodies avoid using the term “program” in this context, so as to emphasize its voluntary, individual and “pan-ethnic” nature. In the Netherlands, initiatives of pre-conceptional carrier screening for CF and hemoglobinopathies are being promoted but usually without directly targeting particular ethnic communities (such as immigrants from Surinam and the Netherlands Antilles, or from Turkey and Morocco), even though members of such ethnic communities have a higher risk of being carriers (Lakeman et al. 2008, 2009). Furthermore, many of the immigrant or indigenous ethnic communities that are at higher risk for recessive genetic diseases are also characterized by high frequencies of consanguinity; however, education and counseling aimed at such communities must be very sensitive regarding the political and gendered relations of power and authority that underpin patterns of consanguinity as well as the attempts to change them (Shaw 2000, 2001; Shaw and Hurst 2008a, b; Raz 2005).

Health insurance is an additional important socioeconomic factor. Who picks up the bill for medical genetic services? In the USA, if two engaged orthodox Jews seek to get tested for Tay-Sachs through the public health system, their health maintenance organization (HMO) might tell them that it does not cover such testing unless the woman gets pregnant. In many cases, people have to pay for the test out of pocket. In Israel, some of the tests (Tay-Sachs, CF, Familial Dysautonomia) are offered free of charge through the National Carrier Screening Program; other tests are offered free of charge to communities at risk; and private genetic testing done by individuals is subsidized by HMOs. In the USA, this is not standardized.

Yet another social factor that influences genetic screening consists of the moral and legal standing of the fetus as reflected, for example, in the pro-life vs. pro-choice debate. Communal and religious norms influence attitudes regarding “therapeutic” abortions, which may promote or hinder the use of pre-conceptional and/or prenatal genetic testing. This may explain why some countries prefer newborn screening to pre-conceptional screening, which may increase the choice of preventive abortion. The question of whether and how to implement, regulate and use genetic testing and screening is thus also contingent on our social and communal narratives of risk and responsibility.

Comparative sociological analysis of such schemes and their antecedents carries practical contribution for the planning and carrying out of genetic services in the community. For example, such sociological analysis can be valuable in developing strategies for interventions for carriers identified through newborn screening programs, as this is not a well-defined or well-understood community (Hayeems et al. 2008). When it is difficult to categorize the people on the front end of screening, it is also difficult to customize a program for them. Should it be a public health activity or a clinical program, and how will that constrain or facilitate delivery? These are practical issues of implementation that can be greatly helped by a sociological perspective. The government should protect people from genetically-related health insurance discrimination, but people also need protection from the utilitarian ethics of governments, whose public health officials may be tempted by the cost-effectiveness of preventive genetic testing. At the end of the day, communities—as well as individuals—should hold the power to accept or reject the offers of clinical genetics, but for that they need to learn about it. The field of community genetics is still in search of regulatory and moral models. Hopefully, this commentary will promote further socio-anthropological research into community genetics.

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