

## Community Genetics: a new discipline and its application in Brazil

Genética Comunitária: uma nova disciplina e sua aplicação no Brasil

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**Abstract** *Community genetics is a new discipline which aims to provide genetic services to the community as a whole. As a science, community genetics encompasses all research needed to develop and evaluate its application. There is no question that the development of community genetics is necessary in Brazil. The implementation of such programs in our country, especially for hemoglobinopathies, has been recommended by the World Health Organization and other international organizations. Apart from the need for and appeal of community genetics programs, some aspects require serious review. This article discusses various cultural, social, psychological, and economic factors that can make genetic screening an invasion of individual privacy.*

**Key words** *Community Genetics; Genetic Counseling; Genetics*

**Resumo** *A Genética Comunitária é uma nova disciplina, que tem por objetivo o fornecimento de serviços de genética para a comunidade como um todo. Enquanto ciência, engloba todas as pesquisas necessárias ao desenvolvimento e à avaliação das suas aplicações. Indiscutivelmente, o desenvolvimento da disciplina no Brasil é muito necessário e a implantação de programas brasileiros de genética comunitária vem sendo recomendada pela Organização Mundial de Saúde e por outras organizações internacionais, sobretudo para as hemoglobinopatias. Apesar da necessidade e do lado atraente dos programas comunitários, alguns aspectos destes devem ser seriamente considerados. No presente artigo, são discutidos alguns fatores culturais, sociais, psicológicos e econômicos que podem transformar a triagem genética em uma invasão da privacidade dos indivíduos.*

**Palavras-chave** *Genética Comunitária; Aconselhamento Genético; Genética*

The emerging possibilities of applying new diagnostic techniques to genetic diagnosis and population screening indicate the need for a community genetics discipline (Modell & Kuliev, 1998). As with many other disciplines, community genetics has both scientific and applied aspects (Ten Kate, 1998). Its aim is to deliver genetic services to the community as a whole. As a science, community genetics encompasses all research needed to develop and evaluate its application. Community genetics as a science is a broad field which is better characterized as *meta-genetics* than by its contribution to genetics itself (Ten Kate, 1998).

Population screening is an essential part of community genetics. There are two major types of genetic screening tests. The first aims at the early recognition of affected individuals for whom medical intervention will have a beneficial effect. This group includes fetal screening, with prenatal diagnostic tests for conditions like Down's syndrome, and neonatal screening, of which the classic example is the phenylketonuria survey. The second major form of genetic screening, more common in community genetics programs, is the identification of individuals at risk of transmitting a genetic disease. Classic examples are carrier detection tests for diseases such as Tay-Sachs, cystic fibrosis, sickle cell anemia, and the thalassems (Gelehrter & Collins, 1990).

Among the hereditary disorders, the hemoglobinopathies are the ideal field for Brazilian community genetics programs, for three reasons: they are the most common serious single-gene disorders in this country, they are relevant to health, and screening programs for hemoglobinopathies have been initiated in some universities. Because of the ethnic composition of the population, sickle cell syndromes, hemoglobin C, and  $\beta$ -thalassemia are common in Brazil and constitute a public health concern (Ramalho, 1986). In an attempt to ameliorate this situation, an optional genetic counseling program for hemoglobinopathies has been developed by the authors at the Blood Center of the State University of Campinas (UNICAMP) over the last 15 years (Paiva e Silva et al., 1993; Ramalho et al., 1995).

Implementation of community hemoglobinopathy programs in Latin America has been recommended by the World Health Organization (WHO, 1983), the Third World Academy of Science (TWAS, 1986), and the Pan-American Health Organization (OPS, 1987). The WHO Committee for the Prevention and Control of Hemoglobinopathies has again recommended the implementation of community hemoglo-

binopathy programs in Latin America, especially in Brazil (Penchaszadeh, 1993). Other examples of genetic disorders amenable to community programs in Brazil are cystic fibrosis in the white population, Tay-Sachs disease in Jewish communities, and glucose-6-phosphate dehydrogenase deficiency in the population as a whole. However, a community program's success depends on the population's receptivity to such studies. Evaluation of results is thus important, since receptivity depends on socioeconomic, psychological, and cultural factors. Indeed, as commented by Bowman (1991), population programs are often conceived in a theoretical, idealized world, but are carried out in the very different real world.

Apart from the need for and appeal of community genetics programs, some aspects relevant to Brazil and other Latin American countries should be considered:

- Community programs must be offered on a voluntary basis, as recommended by the Medical Code of Ethics. The ability to achieve prospective carrier screening depends on the availability of motivated individuals, level of development of health services, and rate of change in social attitudes in each country (Modell & Kuliev, 1998). In an extensive survey of a Brazilian population, Teixeira & Ramalho (1994) encountered a good community response to an optional program on hereditary hemoglobinopathies. However, many researchers tend to routinely perform involuntary screening, generally not consulting the screened individuals about their desire to participate.
- Community programs require adequate infrastructure to provide population screening, genetic counseling, and information and education to professionals and the public at large. In Brazil, such infrastructure is still highly precarious in some regions and requires urgent development.
- Genetic counseling is a communications process dealing with human problems associated with the occurrence (or risk of occurrence) of a genetic disorder in a family. This process involves an attempt by one or more appropriately trained professionals to help the individual or family understand the medical facts and make the best possible adjustment to the disorder. Since genetic counseling is a communications process, several aspects of information transfer must be considered. One must assess the emotional, cultural, and intellectual receptivity of individuals seeking such counseling. The information communicated is often emotionally loaded and may contain medical information that is difficult for individuals to

understand and/or accept (Gelehrter & Collins, 1990). Thus, in many community genetics programs, genetic counseling is provided by a multiprofessional health team, including physicians, medical geneticists, psychologists, social workers, and specifically trained genetic counselors.

- Communities should be consulted about the application of new genetic knowledge (Modell & Kuliev, 1998). However, in Brazil such public consultation is hampered by people's limited knowledge about genetics.
- Community programs in the Northern Hemisphere include prenatal diagnosis and the possibility of "therapeutic" abortion in their methodology. The aim of prenatal diagnosis is to offer parents the assurance of having unaffected children when the risk of having an affected child is unacceptably high. Prenatal diagnosis also allows one to convert a probability statement about the risk of a specific disease to a certainty. However, Brazilian legislation forbids abortions in such cases. Therefore, the sheer application of new genetic techniques to clinical problems may clash with religious, socioeconomic, and legal factors that cannot be ignored.
- Population screening of asymptomatic heterozygotes in order to provide genetic guidance entails the risk of labeling, discrimina-

tion, stigmatization, and loss of self-esteem by screened individuals (Wilkie, 1994). The effect of a community program varies according to the circumstances of screening and genetic counseling. Social, cultural, and psychological aspects must thus be considered in each case. The effects of genetic counseling for Brazilian blood donors with the sickle cell trait (hemoglobin S heterozygotes) has been investigated (Paiva e Silva & Ramalho, 1997). The theoretical risks of genetic screening did not appear to be a cause for concern in most of the cases examined. However, despite the ethical and psychological precautions taken, signs of stigmatization, labeling, discrimination, and loss of self-esteem were evident in a few cases. These patients received psychological support. We wonder whether other Brazilian community programs evaluate the psychological effects of their counseling.

The above-mentioned difficulties are obviously not insurmountable in Brazil. However, this word of caution is intended to help prevent Brazilian community genetics programs from invading individual privacy under the auspices of a new discipline. Several Brazilian research centers currently fulfill the technical, professional, and ethical conditions needed for the development of high-quality community genetics programs.

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