

Qualification and provision of physicians in the context of the National Policy on Comprehensive Care of People with Rare Diseases in the Brazilian National Health System (SUS)

Débora Gusmão Melo^(a)

Carla Maria Ramos Germano^(b)

Carlos Guilherme Gaelzer Porciúncula^(c)

Isaias Soares de Paiva^(d)

João Ivanildo da Costa Ferreira Neri^(e)

Lucimar Retto da Silva de Avó^(f)

Marcelo Marcos Piva Demarzo^(g)

Marcial Francis Galera^(h)

(a,b,f) Departamento de Medicina, Universidade Federal de São Carlos (UFSCar). Rod. Washington Luis (SP-310), Km 235, Campus da UFSCar. São Carlos, SP, Brasil. 13565-905. dgmelo@ufscar.br; cgermano@ufscar.br; lucimar@ufscar.br

(c) Faculdade de Medicina, Universidade Federal de Alagoas (Ufal). Maceió, AL, Brasil. cggael@uol.com.br

(d) Hospital Universitário Pedro Ernesto. Rio de Janeiro, RJ, Brasil. ispaiva.gen@gmail.com

(e) Curso de Medicina, Universidade Potiguar. Natal, RN, Brasil. jineri@uol.com.br

(g) Departamento de Medicina Preventiva, Universidade Federal de São Paulo (Unifesp). São Paulo, SP, Brasil. marcelodemarzo@gmail.com

(h) Departamento de Pediatria, Faculdade de Medicina, Universidade Federal de Mato Grosso (UFMT). Cuiabá, MT, Brasil. fgalera@uol.com.br

This theoretical essay reflects on the qualification and provision of physicians in the context of the National Policy on Comprehensive Care of People with Rare Diseases in the SUS. To carry out this analysis, we introduce the Policy and its guidelines, with a focus on discussion of two integrated strategies: the provision and settlement of geneticist physicians; and training of primary care professionals with regard to genetic diseases and congenital defects. Finally, in view of the Brazilian National Curriculum Guidelines for undergraduate medical courses, we suggest a profile of minimum competencies required in Genetics, developed in order to provide resources to undergraduate courses in Health, in particular, medical courses. We provide a theoretical reference to support the outline of programs of education and training in Health, contributing to including Genetics in the SUS.

Keywords: Rare diseases. Public policy. Medical Genetics. Education in Health. Clinical competence.

Introduction

Medical Genetics deals with rare diseases, which altogether make up a significantly large group with a prevalence of 31.5 to 73.0 per 1,000 individuals¹. The incidence of genetic diseases and congenital defects in Brazil is no different from that found in other parts of the world; about 3% to 5% of Brazilian newborns have congenital defects that are totally or partially determined by genetic factors².

As health indicators improve, genetic diseases and congenital defects account for a larger proportion of deaths among children³⁻⁵. The impact of congenital defects is particularly noticed when child mortality rates reach figures below 40 per 1,000⁶. That is what has happened in Brazil, where these diseases, recorded in Chapter 17 of the International Classification of Diseases (“Congenital malformations, deformations and chromosomal abnormalities”), have been the Number 2 cause of child mortality in all regions of the country since 2005⁷.

Since 1998, the World Health Organization (WHO) has constantly reminded developing countries of the need to begin activities of promotion and protection of Health in the field of primarily genetic diseases and congenital defects⁸⁻¹¹. In this context, in January 2014 the Brazilian Ministry of Health implemented the National Policy on Comprehensive Care of People with Rare Diseases (PNAIPDR) in the SUS¹², ensuring health care services for a wide range of rare genetic diseases that are usually chronic, progressive, degenerative, and often present a death risk¹³.

This paper is a theoretical essay that reflects on the qualification and provision of physicians in the context of PNAIPDR. In order to carry out this analysis, we will first introduce the Policy and its guidelines, and then we will focus the discussion on two integrated strategies: the provision and settlement of geneticist physicians; and training of primary care professionals with regard to genetic diseases and congenital defects. Finally, in view of the Brazilian National Curriculum Guidelines for

undergraduate medical courses, published in 2014¹⁴, we will suggest a profile of minimum competencies required in Genetics, developed by the Teaching Commission of the Brazilian Society of Medical Genetics to provide resources to undergraduate courses in Health, in particular, medical courses.

We provide a theoretical reference, based on competence, to support the outline of programs of education and training in Health, contributing to including Genetics in the SUS.

The National Policy on Comprehensive Care of People with Rare Diseases: the historical perspective and guidelines

Conceptually, a disease is considered rare when its prevalence is equal to or lower than 50 to 65 per 1000,000 individuals^{15,16(1)}. There are about five to eight rare diseases^{15,17} and, in Brazil, it is estimated that 13 million people have a rare disease¹⁸. Nearly 80% of rare diseases have a genetic etiology; the remaining 20% include immune diseases, some types of cancer, and uncommon infectious diseases¹⁵.

About 50% of rare diseases affect children¹³ and 30% of patients die before the age of five¹⁵. In Brazil, surveys carried out by patient associations indicate difficulties in diagnosing such diseases, with patients seeing several physicians, sometimes for decades (Brazilian Alliance of Genetics, personal communication). Although there is not a specific treatment for most rare genetic diseases, the implementation of adequate care services can improve quality of life and increase life expectancy of patients, besides providing family genetic guidance^{15,17}.

Formal discussion of the implementation of a public health policy for Genetics began in Brazil in October 2004, with the creation of the Clinical Genetics Work Group¹⁹, which was composed of specialists in medical genetics and technicians from the Ministry of Health. The work of this group resulted in the publication of the National Policy on Comprehensive Care in Clinical Genetics in SUS in 2009, and its objective was to build a local, tiered service network that granted access to

⁽¹⁾ In the context of PNAIPDR, a disease is considered rare if it affects up to 65 people per 100,000, that is, 1.3 per 2,000 individuals.

comprehensive care in Genetics²⁰. As is often the case, some aspects had to be regulated and this policy was actually never implemented.

The Policy approved in 2014 was the result of work carried out by another group, created in 2012 by the Department of Specialized and Thematic Care/Coordination of Medium and High Complexity, with representatives of civil society, specialists in medical genetics and technicians from the Ministry of Health²¹. To include all known rare diseases, it was decided to classify diseases in PNAIPDR according to their nature, as being genetic or non-genetic. Therefore, two classifications of rare diseases were designated. The first included three groups of genetic diseases: (1) congenital abnormalities or those with late onset, (2) intellectual impairment, and (3) inborn errors of metabolism. The second includes rare non-genetic diseases¹².

According to the PNAIPDR guidelines, care services must be structured according to a systemic logic, focused on the acknowledgment of the needs of patients and dynamic and continuous care to meet those needs. The SUS must then ensure comprehensive care, organized at all levels, from prevention, embracement, diagnosis, treatment (ensuring access to available technologies and genetic counseling) support and help, to resolution, follow-up and rehabilitation²¹. This policy includes specific functions for primary health care (PHC) and for outpatient and hospital specialized care, and also includes implementation of “specialized care services for rare diseases” and “reference services for rare diseases”²¹. For PHC, nine specific assignments were defined that included mapping of people who have congenital abnormalities and/or genetic diseases, or are at risk of having them, for regulated referral; promotion of education in health with the aim of prevention; providing clinical follow-up after diagnosis; genetic counseling (counter-referral); and home care in specific cases²¹. Specialized care services and reference services are responsible for diagnosis, and therapeutic and preventive actions, for people who have rare diseases or at risk of developing them, which includes specialized clinical multidisciplinary monitoring and non-directive and non-coercive genetic counseling²¹.

About genetic counseling in PNAIPDR

Genetic counseling is defined as a communication process that deals with human problems associated with the occurrence or recurrence of a genetic disease in a given family. This process involves the attempt, by one or more people who are properly trained, to help individuals or their families: (1) understand medical facts, including diagnosis, the most likely course of the disease (prognosis), and measures (treatments) available; (2) analyze how heredity contributes to the disease and the risk of recurrence in specific members of the family; (3) understand the options available in the case of recurrence, with regard to the family's reproductive life; (4) choose the most suitable actions, considering the risks and objectives of the families, and act accordingly; and (5) adapt in the best possible way to relatives' disease and/or to the risk of recurrence²².

The Policy recommends that genetic counseling be offered to individuals and families with rare genetic diseases, or those are at risk of developing them, with assistance and education as a primary goal, allowing them to understand all aspects of the disease, from its etiology, evolution, and prognosis to decision-making concerning reproductive rights²¹.

The guidelines for the Policy also state that genetic counseling must be performed by a qualified multiprofessional team, including a geneticist physician and/or a qualified health professional who has a undergraduate degree, a master's degree or PhD in Human Genetics or specialized in Human Molecular Biology or Human Cytogenetics, awarded by the Brazilian Society of Genetics, or a specialist degree in Genetics awarded by the Federal Council of Biology, and proof of an 800-hour minimum of professional experience or internship in genetic counseling²¹.

In European countries, the United States, Australia, and Canada, where genetics are more rooted in healthcare practice, there is an occupation called "genetic counselor," which usually corresponds to a professional with a degree in a life sciences field (more frequently Medicine, Nursing or Psychology) and with specialized training, typically a master's degree, that includes knowledge of Clinical Genetics, Population

Genetics, Cytogenetics, and Molecular Biology, as well as skills related to Psychology and communication skills^{23,24}.

Although the PNAIPDR is aware of the need for genetic counseling in comprehensive care of individuals and families with rare genetic diseases, the occupation of genetic counselor not yet been regulated in Brazil. Currently, the Brazilian federal agency for Coordination for the Improvement of Higher Education Personnel (CAPES) recognizes 36 graduate programs in Genetics, 17 of which are PhD's, 16 master's degrees, and three vocational master's degrees²⁵. Most of these programs have researchers with experience in Human Genetics and/or Medicine, and they are responsible for training professionals in the healthcare and life sciences. There is only one vocational master's degree specialized in genetic counseling; it is offered by the Institute of Biosciences of the University of São Paulo, and it has been in place since 2015²⁵.

Medical Genetics in Health Care in Brazil and new training needs in view of PNAIPDR

In Brazil, Medical Genetics was recognized as a medical specialty by the Federal Council of Medicine in 1983. In the search for its consolidation, the Brazilian Society of Clinical Genetics was founded in 1986; it was renamed the Brazilian Society of Medical Genetics (SBGM) in 2006²⁶.

Services in Medical Genetics began to develop in the country in the 1960s and 1970s, often related to undergraduate courses in Human and/or Medical Genetics, with greater interest in research on specific diseases or groups of diseases. During the 1970s and 1980s, services with greater care capacity were implemented, in association with hospitals and/or public universities²⁷. Records made by SBGM in 2000 reported the existence of 64 care services in Medical Genetics: 37 (58%) in the Southeast (75.7% in the state of São Paulo), 17 (26%) in the South, 7 (11 %) in the Northeast, and 3 (5%) in the Center–West. At that time, no service was found in the North. The types of

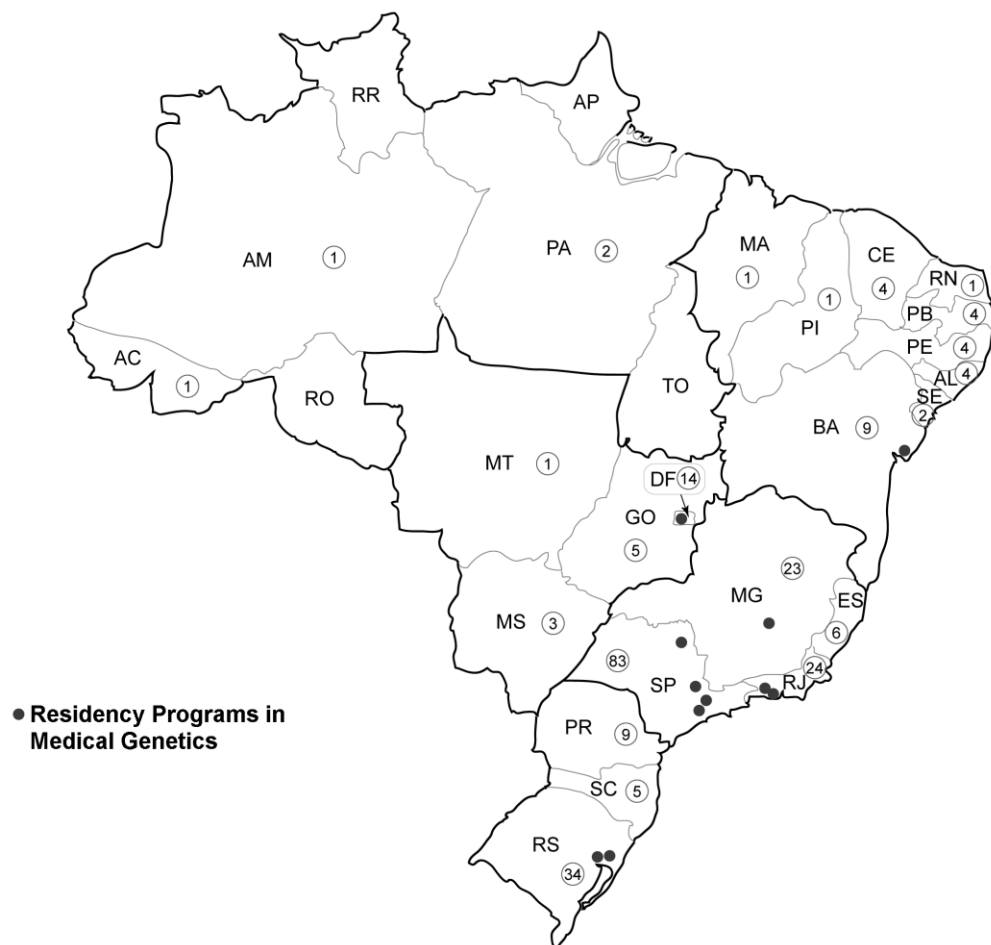
service offered varied greatly; some were very comprehensive (clinical consultation, laboratories and research), whereas others only offered genetic counseling³.

The number of services and human resources involved in Medical Genetics in Brazil is considered insufficient to meet the demand. It is estimated that most patients and families who die from genetic diseases are not provided with proper care^{28,29}. Data from the survey Brazilian Medical Demography, carried out by the Federal Council of Medicine³⁰ and published in November 2015, report the existence of 241 physicians specialized in Genetics[Ⓜ] distributed unevenly in the territory (Fig. 1). The geographical distribution of genetics professionals and services is related to population density and the human development index of the regions, and in poorer and less populated areas (particularly the North and Northeast) there is a greater need for professionals.

There are only 11 residency programs in Medical Genetics in the country, which offer a total of 22 new vacancies per year to resident physicians, and, except for a program associated with the University of Brasilia and another with the Federal University of Bahia, all the programs are located in the South and Southeast (Fig. 1)³¹.

Figure 1. Distribution of the 241 physicians specialized in Genetics and the 11 residency programs in Medical Genetics in Brazil.

[Ⓜ] The total of 241 specialists in Medical Genetics includes 17 (7.1%) with duplicated records.



Additionally, it is estimated that about twenty nurses are working in this field, most of them with PhD's in Genetics. They are currently engaged in consolidating the Brazilian Society of Nurses in Genetics and Genomics, founded in June 2015 (Flória-Santos, personal communication).

Due to the concentration of professionals and services in the Southeast and South regions, there is significant migration of patients (especially from the North and Northeast) looking for health care. Lack of specialized human resources is an obstacle to the inclusion of Clinical Genetics in SUS^{5,28,32} and an obstacle to the implementation of PNAIPDR³¹.

In addition to this lack of specialists, there is also underdeveloped training of health professionals who work in PHC with regard to Clinical Genetics^{31,33,34}. Generally speaking, medical professionals who work in PHC are capable of understanding situations in which it is necessary to refer patients to a genetics specialist, but they have difficulties with collecting and assessing family background information and identifying patterns of genetic heritage³¹. Successful experiences in Genetics training with PHC professionals have been reported in the country^{35,36}, highlighting the potential of the Family Health Strategy to contribute to the implementation of PNAIPDR.

Care focused on individuals, which is a practice encouraged by the Family Health Strategy, favors a comprehensive and longitudinal approach to individuals, considering them as unique subjects, and also taking into account their families and sociocultural integration. Therefore, professionals who work in PHC can have more opportunities to identify hereditary diseases in families and map situations of environmental risk of congenital defects, such as exposure to teratogens. In addition, with regard to genetic diseases, which are usually multi-systemic and require the participation of large numbers of professionals, the coordination of care services carried out in PHC may ensure effective coordination between different levels of care^{37,38}. Education of generalist health professionals who work in PHC is essential to properly translate recent findings in Genetic and Genomic Medicine into benefits to patients and families, contributing to reducing inequalities in Health^{31,34,39,40}.

Although it is still limited, education and training in Genetics has been implemented in undergraduate medical courses in Brazil, but the Genetics knowledge proposed in curricula varies greatly with regard to the topics presented and their depth^{28,41,42}. Usually, this learning is based on a subject that is often on its own, with limited clinical practice, and restricted to the study of diseases that are rare in the general population, but common in teaching hospitals⁴³.

The current Brazilian National Curriculum Guidelines (DCN) for undergraduate medical courses, published in 2014, establish as part of the skills profile for graduates: "Being able to propose and explain the diagnostic investigation to the person

being cared for or the person in charge, so as to broaden, confirm or rule out diagnostic hypotheses, including recommendations for genetic counseling”¹⁴. It is not clear in DCN which knowledge and skills are necessary to achieve this competency, which encouraged SBGM to position itself on the subject.

Minimum competencies in Genetics required of health professionals in Brazil in the context of PNAIPDR

Groups of specialists in human and Medical Genetics in Europe and the United States have been working on the development of educational guidelines and the establishment of genetics skills for health professionals⁴⁴⁻⁵¹.

Using the material produced by these groups in March 2015 as a reference, five geneticists, university professors, members of the SBGM Teaching Commission during the 2014-2016 term, and the authors of this paper met for two days with the purpose of developing a proposal for a profile of minimum competencies in Genetics, adapted to the Brazilian reality, considering PNAIPDR and the latest DCN for undergraduate medical courses. The material produced by this group of specialists was validated by other SBGM members on the Internet, and it is now presented in this paper.

The theoretical references of two organizations, the National Coalition for Health Professional Education in Genetics (NCHPEG) in the U.S.⁵⁰ and the European Society of Human Genetics (ESHG)⁵¹ were chosen as benchmarks for discussion. NCHPEG is a nonprofit organization whose purpose is to promote health professional education and access to information about advances in Human Genetics. In 2007, it published *Core Competencies in Genetics for All Health-Care Professionals*⁵⁰. As for ESHG, in 2008 it defined specific competences for physicians, nurses, obstetricians, and dentists who are not specialists in Genetics, and also for specialized physicians and nurses who work in the European Union⁵¹.

As a result of the work done by the SBGM, four core competencies were established for all health-care professionals: (1) examine their own clinical competence regularly, acknowledging any gaps and advances in Genetics and

Genomics over time, understanding the need for continued education; (2) identify individuals who have or may develop genetic diseases and know how and when to refer them to medical genetics specialists; (3) handle patients with previously diagnosed genetic diseases or congenital defects, using clinical guidelines already established in the scope of their professional work; and (4) promote and encourage clinical practices and training in Health with the aim of preventing genetic diseases and congenital defects. Table 1 presents the specific set of necessary skills, knowledge, and actions that were defined to gain these competencies.

Table 1. Profile of minimum competencies in Genetics for all health professionals in Brazil, as proposed by the Brazilian Society of Medical Genetics.

Knowledge
Acknowledge the importance of genetic diseases and congenital defects in local and national epidemiological contexts.
Learn the terminology and basic concepts used in Medical Genetics.
Know the classic patterns of heritage in families and in communities.
Recognize the importance of genograms when assessing predisposition/susceptibility and genetic disease transmission.
Have basic notions of Morphogenesis and Human Physiology and the role of Genetics in these processes.
Understand how the interactions between Genetic, environmental and behavioral factors act in susceptibility, in the beginning and in the development of diseases, as well as in responses to treatment and health maintenance.
Identify the main teratogenic agents and related preventive measures (especially alcohol and illegal drugs).
Identify the main genetic risk factors – advanced parental age, inbreeding, recurrence in the family.
Know preventive measures related to genetic diseases and congenital defects: folic acid before pregnancy, maternal immunization, healthy life habits.
Recognize that genetic diseases are frequently multi-systemic disorders that need an interdisciplinary and multiprofessional approach.

Know the principles and guidelines of the National Newborn Screening Program.
Know the official and compulsory forms for recording genetic diseases and congenital defects: Statement of Live Birth and Death Certificate.
Know genetic diseases and congenital defects that are not rare, that is, that have prevalence above 1.3:2,000 individuals.
Know the main genetic tests used in clinical practice.
Know the health care service network available at the three levels for individuals with genetic diseases and congenital defects and their families.
Know the duties of geneticist physicians in the acknowledgment and handling of genetic and congenital diseases, with the purpose of making the reference and counter-reference system operational.
Skills
Gather information about the genetic history of families, including the construction of a genogram for at least three generations.
Recognize the variations in normal phenotypes and their morphological and functional alterations.
Complete reference and counter-reference documents for patients with suspected genetic diseases and congenital defects or with established diagnoses.
Use appropriate communication skills; show awareness of the need for confidentiality and a non-directive approach to patients and their families.
Use available technology properly to obtain updated information about Genetics and Genomics.
Actions
Respect non-directive and non-coercive genetic counseling.
Take into account cultural and religious beliefs of patients with regard to their genetic heritage when providing care to people with or at risk of developing genetic diseases.
Be sensitive to the importance of and need for privacy and confidentiality.
Show awareness of the importance of the social and psychological impact of a genetic diagnosis on patients and their relatives.
Be able to work in a cooperative and collaborative manner in an interdisciplinary and multiprofessional team.

This competency profile is expected to be included in undergraduate medical courses, for which DCN has already acknowledged the importance of Genetics in health care processes. Likewise, other undergraduate courses such as Nursing are expected to include Genetics in their curriculums and benefit from this proposed competency profile.

It is known that training of health professionals, especially physicians, on genetic diseases and congenital defects includes the suitability of training during courses, but also involves capturing graduate professionals^{28,31,34}. Therefore, it is necessary to permanently structure and promote courses of continued education for these professionals, within a time frame that will keep them up to date. One possible strategy is to take advantage of the period of one or two years of residency in family and community medicine as a training opportunity. The More Doctors in Brazil Project is currently set to make these residencies compulsory beginning in 2018, before entrance into other medical specialties⁵². The success of this strategy depends on possible developments in the program, which has been going through a period of institutional instability and uncertainty regarding its future.

Final considerations

By introducing a proposal for a profile of minimum competencies in Genetics for health professionals that is suitable to the Brazilian reality, we hope to provide a theoretical reference to guide the curriculums of health care courses, especially in Medicine. This very same competency profile can support policies for continued professional training in the field of Genetics, so as to enable the SUS workforce with regard to genetic diseases and congenital abnormalities, preparing professionals for implementation of PNAIPDR.

It is worth noting that the More Doctors Program aims to keep Medical Genetics as a specialty to which the residency program has direct access⁵². It is our understanding that it is the role of the Ministry of Health to launch more residency

programs in Genetics, in order to ensure the training of sufficient specialized human resources to implement PNAIPDR in SUS.

Finally, it is essential to acknowledge the work of genetic counselors as health professionals and to clearly define their training and duties.

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Collaborators

J.I.C.F. Neri, C.G.G. Porciúncula, D.G. Melo, I.S. de Paiva and M.F. Galera developed the profile of competencies in Genetics presented in this paper. D.G. Melo developed the initial version of this paper. M.M.P. Demarzo, C.M.R. Germano and L.R.S. de Avó contributed greatly to critical discussion of this paper. All authors contributed to the writing and review of the final text.

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