

Genetics in Primary Healthcare in Brazil: Potential Contribution of Mid-level Providers and Community Health Workers

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Abstract

Objective: This study aimed to investigate genetic care competencies of mid-level health providers and community health workers in Brazil.

Methods: It is a descriptive survey study, whereby mid-level health providers (nursing auxiliaries and technicians, and auxiliaries in oral health) and community health workers were invited to participate by answering a questionnaire including 10 structured genetics-oriented issues. Data were presented as percentages. Differences between groups were assessed by the Fisher exact test, with the level of significance set at $p < 0.05$.

Results: Concerning knowledge, most of the participants valued important aspects of family history like the distribution of a disease over successive generations and presence of consanguinity, and 79% of them recognized that there are families at increased risk of developing cancers. They also had good knowledge about exposure to teratogens and neonatal screening. Regarding clinical skills, practitioners were able to recognize facial dysmorphias and 63% showed predisposition to gather information about genetic disease in the family history. Regarding attitudes, 65% believed that patients with rare diseases require interdisciplinary approach and 61% showed initiative to refer patients for specialist. In general, there was no significant difference between both professional groups on the investigated knowledge, skills and attitudes, except for the skill to distinguish the phenotype of Down and Crouzon syndromes, which was more present among the mid-level health providers ($p = 0.04$).

Conclusion: These findings may contribute to developing an ongoing education program for mid-level health providers and community health workers, leading to a strategy to overcome the challenges of integrating genetics into primary healthcare in Brazil.

Keywords: Brazil; Medical genetics; Rare diseases; Clinical competence; Competency-based education; Continuous professional education

Introduction

In 1988, during the promulgation of the current Constitution of the Federative Republic of Brazil, it was instituted the Unified Health System (*Sistema Único de Saúde*, SUS), which offers to all Brazilian citizens integral, universal, equalitarian and free access to health services of different levels of complexity (from simple outpatient to highly complex procedures, as organ transplants). In addition to democratization, the SUS also represented a change in the healthcare paradigm, with the adoption of a decentralized management model, and with popular participation, that values the health protection and promotion, the disease prevention and the comprehensive care [1].

In the course of reorientation of the healthcare model, the primary healthcare has been strengthened with expansion and qualification measures. In this context, since 1994, the Family Health Strategy (FHS) has been the main policy for structuring municipal health systems. The FHS consolidates a model of community-oriented primary care, including comprehensive care, cultural competence and social

accountability, focusing on local family and community-based healthcare [2]. The FHS prioritizes coverage of vulnerable low-income populations and now reaches about 63% of Brazilian citizens (about 123 million people in November 2015).

An important aspect of the FHS is the establishment of a multidisciplinary team (family health teams) at least composed of: general practitioner or specialist in Family and Community Medicine, nurse, auxiliaries nurse or nursing technicians, and community health workers. The oral health care professionals may be added to this composition: dentist, auxiliaries or technicians in oral health. Family health teams are assigned to specific geographical areas and defined populations of 600-1,000 families. The team provide a first contact with the local health system, coordinate care, and works towards diagnostic integration between specialist and hospital care. Health services and health promotion activities take place at health facilities in patients' homes and in community [1].

In January 2014, the Ministry of Health in Brazil established the National Policy on Comprehensive Care for People with Rare Diseases in SUS. As 80% of rare diseases have a genetic aetiology, this has created an opportunity to include genetics in SUS. This Policy assumes that healthcare should be integrated into a region-based context, structured both by primary and specialized healthcare services.

Primary healthcare was established by the Policy as being responsible for nine initiatives and specific actions to promote health, including, early diagnosis, monitoring, patient follow-up, appropriate specialist referral and care coordination, including at the family level. Specialized healthcare should consist of outpatients and hospital services that support and complement primary healthcare services [3].

There is a shortage of physicians and nurses specialists in genetics in Brazil, which undoubtedly is an obstacle for universalization of clinical genetics in SUS [4,5]. Furthermore, doctors, nurses and dentists working in primary healthcare still have insufficient genetic care competencies to support the implementation of the National Policy on Comprehensive Care for People with Rare Diseases [6]. It will be a challenge to design a feasible and affordable training program for primary healthcare professionals, and it will be particularly important to engage the members of family health teams in this training.

Given the background, the main purpose of this study was to investigate genetic care competencies of mid-level health providers and community health workers in Brazil. We have not identified other studies in the literature that investigated this particular subject among health professionals who do not have higher education, which motivated this research. Our initial hypothesis was that, in Brazil, these particular professionals do not received any kind of training about medical genetics or prevention of birth defects. However, we believe we are in a timely moment to implement an educational program in Genetics, which can reduce health disparity.

Methods

Setting

The study was conducted in São Carlos, a city located in the São Paulo State's geographical centre, in southeast Brazil. There are approximately 220,000 inhabitants, and the birth rate is approximately 2,700 births/year [7]. In 2010, its human development index was 0.805 and, therefore, it was considered the 28th most developed city in the country [8]. Its gross domestic product per capita is R\$ 20,519,06 [7]. Over the last four years, São Carlos has had an average of 8.57 deaths per 1,000 births, 41% lower than São Paulo State's average, the rate of which was 12.15 deaths per 1,000 births in the same period [9].

São Carlos primary healthcare is based on 12 Basic Health Centres and 15 Family Health Centres. Both have family health teams. The Basic Health Centres has, additionally, medical doctors, paediatricians, gynaecologists and obstetricians.

The only public clinical genetic service in the city is an outreach activity at the Federal University, established in 2006. This Medical Genetics Outpatient Care is located in the local Speciality Health Centre and meets the demand of the SUS for this speciality in the city, as well as for other cities in the nearby region.

Characteristics of participants

The study included 52 health workers, being 25 mid-level providers (12 auxiliaries nurse, 8 nursing technicians and 5 auxiliaries in oral health), and 27 community health workers. All 162 mid-level health providers and 123 community health workers of city's primary healthcare at the time of data collection were invited to participate of the research, but approximately 18% of them agreed to participate.

About 65% of participants worked in Family Health Centres, and the remainder 35% worked in Basic Health Centres. Most participants (92%) were female. A profile of the sample can be seen in Table 1.

Characterization	N	%
<i>Mid-level health providers</i>	25	48
Auxiliary nurse	12	23
Nursing technician	8	15.4
Auxiliary in oral health	5	9.6
Community health workers	27	52
Work at Basic Health Centre	18	34.6
Work at Family Health Centre	34	65.4
Female	48	92.3
Male	4	7.7
Time working in Primary Healthcare	7.2 (SD ± 4.6)	

Table 1: Profile of the study sample (N=52).

Design of the study and data collection procedures

This is a descriptive cross-sectional comparative study, in which mid-level health providers (MLHP) and community health workers (CHW) were invited to answer a questionnaire including demographic information and 10 structured genetics-oriented issues developed by the authors. Data collection was carried out during the years of 2014 and 2015.

The structured genetics-oriented issues were developed taking as reference the "Competencies in Genetics common to all health professionals", identified by the U.S. National Coalition for Health Professional Education in Genetics (NCHPEG), in 2007 [10]. The NCHPEG recommend that at a minimum every healthcare professional should be able (1) to examine a patient's competence of practice on a regular basis, identifying areas of strength and areas where professional development related to genetics and genomics would be beneficial; (2) to understand that health-related genetic information can have important social and psychological implications for individuals and families; and (3) to know how and when to make a referral to a genetics professional [10]. They also defined knowledge, skills and attitudes (Table 2) required to achieve these competencies.

A number of 10 structured genetics-oriented issues were defined considering that the questionnaire should have an adequate size to be answered in about 30 minutes. Eight of the 10 presented issues were built based on news published by the Brazilian press on websites, and included a small report and 4 to 8 sentences related to the described situation, which could be marked if the participant agreed with the option. Therefore, it was intended to obtain real daily situations for analysis and reflection. Questionnaires addressing this subject have not been developed and validated previously; the authors (experts on genetics and/or primary healthcare) developed initial evidence on this questionnaire validation using content validation only (expert and semantic analyses). Structural validity was not performed.

1 Knowledge
1.1 basic human genetics terminology
1.2 basic patterns of biological inheritance and variation, both within families and within populations
1.3 how identification of disease-associated genetic variations facilitates development of prevention, diagnosis, and treatment options
1.4 the importance of family history (minimum three generations) in assessing predisposition to disease
1.5 the interaction of genetic, environmental, and behavioural factors in predisposition to disease, onset of disease, response to treatment, and maintenance of health
1.6 the difference between clinical diagnosis of disease and identification of genetic predisposition to disease (genetic variation is not strictly correlated with disease manifestation)
1.7 the various factors that influence the client's ability to use genetic information and services, for example, ethnicity, culture, related health beliefs, ability to pay, and health literacy
1.8 the potential physical and/or psychosocial benefits, limitations, and risks of genetic information for individuals, family members, and communities
1.9 the resources available to assist clients seeking genetic information or services, including the types of genetics professionals available and their diverse responsibilities
1.10 the ethical, legal and social issues related to genetic testing and recording of genetic information (e.g., privacy, the potential for genetic discrimination in health insurance and employment)
1.11 professional role in the referral to or provision of genetics services, and in follow-up for those services
2 Skills
2.1 gather genetic family history information, including at minimum a three-generation history
2.2 identify and refer clients who might benefit from genetic services or from consultation with other professionals for management of issues related to a genetic diagnosis
2.3 explain effectively the reasons for and benefits of genetic services
2.4 use information technology to obtain credible, current information about genetics
2.5 assure that the informed-consent process for genetic testing includes appropriate information about the potential risks, benefits and limitations of the test in question
3 Attitudes
3.1 appreciate the sensitivity of genetic information and the need for privacy and confidentiality
3.2 seek coordination and collaboration with an interdisciplinary team of health professionals

Table 2: NCHPEG's core competencies in genetics.

The first question brought the story of a boy with sickle cell anaemia. The aim was to evaluate knowledge on the basic terminology used in human genetics (for example, the distinction between genetic disease and hereditary disease), and the skill to gather information about the family history.

The second question presented epidemiological data about trisomy 21 in Brazil. The participants were questioned about their approach before the birth of a child with trisomy 21 in the community that they work at. The options allow to evaluate the attitude of correctly refer patients that can properly be benefited by a consult with a medical geneticist.

The third question used the disclosure of an event to support rare disease patients. The options allow to evaluate the disposition to collaborative working with an interdisciplinary team of health professionals.

Questions 4 and 5 discussed teratogens: question 4 warned about the dangers of consuming alcohol during pregnancy, while question 5

explained the thalidomide fetal syndrome. The aim of the options was to evaluate the knowledge about the side-effects of alcohol and misoprostol, and about the importance of the prenatal care on congenital defects prevention.

The sixth question addresses the newborn screening. There's a National Neonatal Screening Program (*Programa Nacional de Triagem Neonatal*), coordinated by SUS, which includes mandatory screening of all newborns for phenylketonuria, congenital hypothyroidism, hemoglobinopathies, cystic fibrosis, biotinidase deficiency and congenital adrenal hyperplasia. The options allow to evaluate the participants' knowledge about the Brazilian newborn screening program.

Questions 7 and 10 were the only that weren't based on news published on the Internet. Question 7 showed two pictures: one with a trisomy 21 child, and the other with a Crouzon syndrome child. The goal was to evaluate the skill on recognize facial dysmorphias and differentiate the two phenotypes. The tenth question presented a

pedigree, a technique used in the FHS, and the sentences allow to evaluate the participants knowledge on regarding the importance of family history, including the valuation of recurrence of disease over successive generations, consanguinity and recurrent pregnancy loss.

Question 8 brought the announce of the prophylactic mastectomy that the American actress Angelina Jolie had undergone. The options evaluated the knowledge about hereditary cancer and the importance of the family history.

Question 9 was about a craniofacial stenosis case and analyzed the attitude of properly refer patients for expert evaluation.

Data analysis

The answers were analysed and grouped into three categories: knowledge, skills and attitudes. Descriptive statistics were made using Microsoft Excel® and results presented as percentages. Differences between two groups (MLHP and CHW) were assessed by the Fisher

exact test, using Graphpad prism 6®. The level of significance was set at $p < 0.05$.

Ethical aspects

The project was approved by the Human Research Ethics Committee at the Federal University of São Carlos (process 447,484). All participants provided a written informed consent at the beginning of the questionnaire and all questionnaires were anonymized.

Results

Table 3 synthesizes the obtained results. In general, there was no significant difference between the two professional groups in the investigated knowledge, skills and attitudes, except for the skill to distinguish the phenotype of Down and Crouzon syndromes, which was more present among the MLHP ($p 0.04$).

Categories		Mid-level providers (N=25)		Community health workers (N=27)		P value
		Yes		Yes		
		N	%	N	%	
Knowledge	Able to differentiate genetic disease and hereditary disease	24	96	22	81	0.19
	Values the distribution of a disease over successive generations	23	92	26	96	0.60
	Values the inbreeding/consanguinity in a family history	21	84	25	93	0.41
	Values spontaneous miscarriages in a family history	12	48	13	48	1.00
	Recognizes misoprostol as teratogenic	20	80	21	78	1.00
	Considers the prenatal care important to prevent congenital defects	25	100	27	100	1.00
	Considers important to completely abstain from alcohol during pregnancy	23	92	25	93	1.00
	Recognizes that neonatal screening identifies congenital diseases	23	92	25	93	1.00
	Considers possible to diagnose Down syndrome by neonatal screening	2	8	5	19	0.42
	Recognizes that there are families at increased risk of developing cancers	20	80	21	78	1.00
Skills	Gathers information about a genetic disease in the family history	15	60	18	67	0.77
	Recognizes facial dysmorphias	22	88	27	100	0.10
	Able to differentiate Down and Crouzon syndromes' phenotypes	23	92	18	67	0.04*
Attitudes	Refers patient with rare disease for evaluation with interdisciplinary team or specialist	14	56	20	74	0.24
	Refers family to diagnosis confirmation before a suspected case of Down syndrome	14	56	18	67	0.57

* Asterisk indicates the skill where there was a statistically significant difference between distinct groups of professionals.

Table 3: Results regarding knowledge, skills and attitudes investigated in both groups of professionals.

Regarding the knowledge, in a family history, 94% of respondents valued the distribution of a disease over successive generations, 88% valued consanguinity, 48% valued the presence of spontaneous miscarriage and 79% recognized that there are families at increased

risk of developing cancers. About prevention of birth defects, 100% valued pre-natal care, 92% considered that alcohol abstention during pregnancy is important and 79% recognized misoprostol as a teratogenic. Around 92% recognized that neonatal screening identifies

congenital diseases and that abnormal results require early intervention, but 13% considered possible to diagnose Down syndrome by neonatal screening.

Regarding skills, most of the professionals recognized facial dysmorphias in a syndromic patient and, facing a patient with a genetic disease, 63% showed predisposition to gather information about genetic disease in the family history.

Of the attitudinal perspective, 65% believed that patients with rare diseases require interdisciplinary and/or specialized approach and 61% showed initiative to refer patients for specialist.

Discussion

The contribution of MLPH and CHW in the health system is variable, and there's a discussion about the kind of care that these professionals can provide [11], including the fact that their training is different from country to country [12,13].

In Brazil, the professional practice of Nursing was regulated in 1986 and, at the time, three professional categories were defined: auxiliary nurse, nursing technician and nurse [14]. To become a nurse, it's necessary to take a 5 year course of higher education; the nurse is the most qualified of these professionals and the one with the most complete activity field. To auxiliaries nurse or nursing technicians are assigned support functions to nurse and direct care activities to patients without complexity, which include medication, vaccination, making dressings, provide hygiene and comfort [15]. Despite the limits of the activities of the three professional categories of Nursing defined in law, concerning to auxiliaries easiest functions than to technicians, the practice has created a reality in which there was no difference between the work done by nursing technicians and auxiliaries, allowing to question the relevance of this division [16]. In 1999, the National Curricular Guidelines were instituted for the professional education of technicians from the health area (which includes the nursing technicians and oral health technicians, etc) [17] and, in theory, the category of auxiliary nurse was extinguished. The professionals belonging to this class were encouraged to continue training until they become nursing technicians, which many did. On the other hand, auxiliary nurses who have graduated before the new legislation had kept their rights and may exercise the office until they retire. So we live in Brazil a transition period of work in Nursing, with the existence of auxiliary and technicians MLHP [18]. Currently, the technical course in Nursing has a minimum of 1,200h theoretical workload and more 600h clinical supervised internship, lasts 24 months and requires prior completion of high school [19].

The auxiliaries and technicians in oral health have emerged in Brazil after SUS in 1988, in an attempt to establish a new model of collective oral healthcare [20]. A significant change occurred after 1994, with the inclusion of oral health in the FHS, expanding the population's access to promotion and recovery of oral health [21,22]. However, only in 2008, the practice of the auxiliaries and technicians in oral health was regulated [23]. Both professions have similar skills, related to educational activities, promotion and prevention of oral diseases as teaching oral hygiene techniques, dental biofilm removal and topical application of fluoride, leaving the technical more complex than auxiliaries tasks. It is possible that over time, as has happened in Nursing, only remain a technical profession in oral health that currently requires minimum course workload of 1,200h, and to complete 600h training professional becomes assistant in oral health.

In Brazil, the CHW emerged as a professional in 1991 with the creation of the Program of Community Health Agents (*Programa de Agentes Comunitários de Saúde*), aimed at reducing the alarming indicators of child and maternal mortality in the Northeast. In 1994, with the strengthening of the FHS, the Program of Community Health Agents was extinguished and the CHW were absorbed as members of family health teams [1]. The CHW profession is regulated since 2002 [24] and requires that the professional has completed primary education, resides in the community in which it operates and has attended successfully an introductory course in specific training [25], which usually has total workload of 40 hours.

The general duties of MLHP and CHW at Primary Health Care, especially in the FHS, are strongly associated with prevention and education promotion for individual and collective health. To CHWs, in particular, concern periodic home visits to monitor risk situations to families. These professionals maintain direct relations with the community, collect data on family composition, the conditions in which these families live and the health needs of the population in their area [1]. We believe that these are tasks which, if well used, would contribute to implementation of the National Policy on Comprehensive Care for People with Rare Diseases in the SUS.

In addition to NCHPEG [10], many groups have worked on creating core educational guidelines and competencies in Genetics for health professionals [26-32], but all these recommendations are addressed to professionals with higher education. We have not identified other studies in the literature which investigated genetic care competencies of MLHP and CHW.

With proper training, MLHP and CHW can conduct active searches for suspected cases of probable genetic aetiology diseases in the communities in which they work. Therefore, it is important to identify people with sensory disabilities such as deafness and blindness, as well as physical and intellectual disabilities, comorbidities that are often part of the phenotype of genetic syndromes [33,34]. The MLHP and CHW can also help identify risk factors for genetic disorders and birth defects such as inbreeding/consanguinity couples, maternal and paternal advanced ages [35-37], exposure to teratogens [38-40], and positive family history [41,42].

Overall, the primary healthcare professionals in Brazil already have the skill to collect family history and even already they are working with pedigree in the FHS. So, it is required to enable them to understand the importance of certain aspects of clinical history, such as consanguinity, recurrent abortion and family history of cancer, in the identification of families at risk of having genetic disease.

Some experiences of direct involvement of MLPH and CHW in cancer screening programs at primary healthcare [43-45], including Brazil [46], suggest that these professionals can help identify people at higher risk of developing cancer from the family history. There are also reports of successful training MLHP and CHW to work in genetic counselling for specific diseases, such as cystic fibrosis [47] and sickle cell anaemia [48,49].

In the current Brazilian scenario, we believe that is important to value education activities aimed at preventing exposure to teratogens, especially alcohol, which is probably responsible for a significant proportion of cases of intellectual disability and neurobehavioral disorders [40,50,51]. Other actions to prevent birth defects that should be encouraged are related to rubella vaccination and the use for preconception folic acid [52]. More recently, the association reported in Brazil, and not fully proven yet between Zika virus infection and

microcephaly has startled public health experts [53,54]; every effort to combat the *Aedes aegypti* (mosquito vector of Zika virus) should be undertaken by the primary healthcare professionals.

Interestingly, our results did not show significant differences in competencies between MLHP and CHW, except for the largest clinical skill of MLHP to distinguish the Down and Crouzon syndromes. This may indicate that both MLHP as the CHW had no access to specific training in Genetics during their training courses as health professional. Indeed, we suspect that the knowledge of Genetics that these professionals have has been acquired during the primary and secondary education, or through the media and in clinical practice.

Our results indicated that professionals dominate better knowledge relating to government health programs already established, such as the National Neonatal Screening Program and the Prenatal Care Program. Thus, we believe that the imposition of a specific program of training related to rare genetic diseases will be well received by MLHP and CHW, and will have a positive effect on healthcare.

During the training, it should be emphasized the importance of patient's follow-up in a multi-professional team. Therefore, it is necessary to expose the services and resources available, including the specialized healthcare in clinical genetics, which patients can be referred if necessary. About this, Pletcher et al. [55] designed a practice guideline to help primary healthcare providers to indicate for genetic referral, which may be useful.

Limitations and strengths of the study

The results of this study reflect the effect of a non-probabilistic convenience sample, which has a limited size and is embedded in a specific context. Furthermore, data were collected using a novel questionnaire, which has not yet fully been validated. On the other hand, to the best of our knowledge, this is the first study investigating the contribution of MLHP and CHW on care in medical genetics. Further studies should address this issue.

Final Considerations

Education in order to develop genetics healthcare competence is crucial in translating the benefits of medical genetics and genomics to patients and their families, and has potential of reducing disparity and inequity in populations' health needs. The findings of this study may contribute to developing an ongoing education program for MLHP and CHW, leading to a strategy to overcome the challenges of integration genetics into primary healthcare in Brazil.

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Compliance with Ethical Standards

This survey was supported by CAPES (Coordination for the Improvement of Higher Education Personnel) through the Young Talents for Science Project. The research was approved by the Human Research Ethics Committee at the Federal University of São Carlos (process 447,484). All participants provided a written informed consent.

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