

1. Introduction

A number of Latin American countries are experiencing an epidemiological transition, with increasing proportion of morbidity and mortality due to genetic disorders and birth defects. However, health systems in these nations have not responded appropriately to the rising needs of genetic services. Many misconceptions abound, e.g. that genetic services are always high-tech and prohibitively expensive, that very little can be done to improve the health and quality of life of affected individuals and their families, and that genetics is closely linked to abortion. In addition, there is a scarcity of human resources trained in clinical genetics, genetic counseling and laboratory genetics, albeit with marked regional differences. The World Health Organization has supported the development of genetic services in developing countries, convened meetings of experts in medical genetics from these nations, and published recommendations (Penchaszadeh and Beiguelman, 1998; WHO, 1999, 2000, 2002).

Following the recommendations of the Meeting on Collaboration in Medical Genetics held in Toronto, Canada, 9-10 April 2002 (WHO, 2002), the WHO Human Genetics Programme organized a consultation in Latin America to assess the status of genetic services in the region and to devise mechanisms of cooperation to further their development, including issues of genetic research, training and education. The main objectives of the Consultation on Community Genetic Services in Latin America and Regional Networks on Medical Genetics were as follows:

- to update the state of development of medical genetic services in selected countries in Latin America;
- to update the major lines of research in medical genetics taking place in the region;
- to assess the major ethical, legal and social implications of the development of medical genetics in the region;
- to establish a regional network of cooperating centres to plan research and services on the application of genetic/genomic knowledge to improve health in the region.
- to delineate an actionable agenda for the implementation of above proposals.

Twenty recognized experts in medical genetics (see List of Participants) from Brazil, Chile, Colombia, Costa Rica, Ecuador, Mexico, Paraguay, Peru and the United States convened on 19 June 2003 in Porto Alegre, Brazil during the Congress of the Brazilian Society of Clinical Genetics. Each participant contributed a working paper on the situation of medical genetics in his or her place of work (city, state or province, or country). Invited participants from Argentina and Cuba were unable to attend but sent their papers for discussion. The Consultation followed the proposed objectives and the following reports on the common denominators of working papers and the conclusions of the discussions.

2. Medical Genetics Latin America

2.1 Overview of the economic, social and health situation in the Region

An analysis of the working papers written by the participants in the Consultation revealed a number of similarities in the demographic, socioeconomic, and health systems issues in the various countries of the region. At the same time, marked regional differences and unequal distribution of clinical and research resources within and between countries was evident.

The average GDP in Latin America is approximately 10% of that of industrialized countries with almost half of its population living in poverty. The population continues to grow due to reductions in mortality

and increases in life expectancy at birth and in spite of significant reductions in fertility and birth rates. The region is becoming more urbanized and, by 2025, it is estimated that 85% of the population will live in urban areas.

Between 1960 and 1996, the average infant mortality rate fell from 105 to 35 per 1000, with 12 of 22 Latin American countries having rates under 30 per 1000 and 3 having rates below 15 per 1000. An epidemiological transition is occurring, with an increasing proportion of morbidity and mortality due to birth defects, genetic diseases and common chronic conditions associated with genetic predisposition. Congenital anomalies now rank 2nd to 4th as a cause of infant mortality in several countries. Similarly, the share of mortality from malignancies and cardiovascular diseases in Latin America is 8%-25% and 20%-46%, respectively. Access to family planning services and reversible contraceptives is limited and maternal mortality is still unacceptably high (average 190 per 100,000). The average annual per capita health expenditure in Latin America in 1995 was only US\$105, compared with US\$1,860 in industrialized countries. Health resources are unequally distributed, favoring excessive specialization and high technology medicine in tertiary centres rather than cost effective primary care-centred medicine. The privatization of health care that occurred in the last decade is increasing the inequities.

2.2 Overview of genetic services within the health systems

The last time Latin American medical geneticists met to discuss collectively the state of genetic services in the Region was in 1996 in Rio de Janeiro, Brazil, at a satellite meeting on the occasion of the 8th International Congress of Human Genetics (Beiguelman and Penchaszadeh, 1997). A recent review (Penchaszadeh, 2000) outlined the major characteristics of the development of medical genetics and genetic services in Latin America.

Genetic services, defined as health services focused on the diagnosis, treatment, follow-up and counseling of patients affected or at risk for genetic conditions and their families, have had an incipient and fragmentary development in Latin America. Presentations at the Consultation showed the development of medical genetics to be uneven within and between countries, essentially following socioeconomic lines; more developed in wealthier, urbanized areas, and less developed in poorer and rural areas. Most services are concentrated in large urban centres and have a restricted scope, determined largely by the interest of individual clinicians and researchers and the resource limitations. In general, genetic services are characterized by marked inequity to their access, as they are underdeveloped in the public health sector. Genetic services, including laboratory tests, are usually covered by private health insurance plans; however the largest provider of health services in Latin America is the government, which still does not recognize genetic services and testing as priorities in public health programmes. The private sector possesses a significantly more developed infrastructure, particularly for applications that are commercially profitable, e.g. chromosome analysis, prenatal diagnosis, and DNA paternity testing. However, these services are accessible only to a wealthy minority.

A common theme in the working papers submitted by participants was the shortage of trained clinical geneticists and of hospital positions for those with training. In some of the region's countries there have been difficulties in recognizing clinical genetics as a medical specialty.

Reasons for the slow development of genetic services in the region include the following (Penchaszadeh, 2000).

- There is still a burden of unmet needs in other areas of health, such as infectious diseases, malnutrition, prenatal care, labour and delivery care, and neonatal care.
- The medical profession and public health officials do not consider genetic conditions a priority.

- Genetic services are misperceived as expensive and concerned only with rare diseases.
- Their preventive value is erroneously associated with the interruption of affected pregnancies, which is opposed by traditional sectors.
- The public is largely unaware about genetic risks and the possibilities of prevention.

Departments of health at city, provincial, and national levels usually do not have explicit policies or programmes in genetic health care, except for neonatal screening for phenylketonuria and congenital hypothyroidism, which tends to be mandated by law. Even so, coverage rarely surpasses 30-50 % of births in a few major cities of the region. Furthermore, programmes tend to lack financial resources and organization for follow-up and treatment of affected infants. Food fortification with folic acid as an explicit government policy is slowly gaining ground in some Latin American countries, most notably Chile.

Induced abortion is prohibited in most Latin American countries, with the exception of Cuba, and some local jurisdictions such as Mexico City and others. Allowed exceptions include pregnancies resulting from rape or when the life of the mother is at risk. Legal prohibition, however, does not prevent abortions, as there is little or no government interference: 95% of the 4.5 million induced abortions that take place yearly in Latin America are illegal and take place largely in unsafe conditions, particularly for low income women. The consequence is that 20 % of the unacceptably high maternal mortality of the region (190 per 100,000) is caused by unsafe abortions (UNFPA, 1998). In this context, while prenatal diagnosis is an accepted and demanded service by those with higher educational and socioeconomic status (irrespective of religious affiliations), it receives no public funding, resulting in socioeconomic barriers to access, distortions in their rational utilization, service fragmentation and lack of quality control.

In spite of the problems mentioned above, Latin America counts several centres of excellence in clinical genetics, cytogenetics, prenatal diagnosis, and the management of haemoglobinopathies. Molecular diagnosis is slowly becoming available for a limited number of conditions. Genetic services are slowly becoming more organized and are expanding their activities and coverage to primary care settings.

2.3 The medical genetics professionals

Largely physicians trained in medical genetics who practice clinical genetics and genetic counseling constitute the workforce of medical genetics. Genetic tests (cytogenetics, biochemical and molecular) are performed by specially trained physicians or, more commonly, by Ph.D. geneticists. There has recently been an increasing awareness of public health officials, medical professionals and the public at large about the role of genetic factors in health and disease. Clinical geneticists are developing successful local postgraduate training programmes in medical genetics and there are approximately 500 trained clinical geneticists in the region, who provide an array of medical genetic services, including genetic counseling.

The specialty of medical genetics has been recognized in Mexico, Cuba, Brazil, and Argentina. Virtually all countries have professional and scientific societies of human or medical genetics. Recently a Latin American Network of Human Genetics (acronym in Spanish and Portuguese: RELAGH) has been constituted as a loose federation of human genetic societies of Latin American, and is the official representative in the Steering Committee of the International Federation of Human Genetic Societies.

2.4 Training and education in medical genetics

Genetic education of health professionals is clearly deficient both at undergraduate and postgraduate levels. While basic genetics is generally taught in most medical schools, this is not the case with clinical and community genetics. Most papers from participants stressed the need to teach medical genetics in the

clinical years of medical students' and nurses' undergraduate education. Some countries have postgraduate training programs for physicians and scientists, leading to Master and Doctorate degrees. Clinical training in genetic residencies is scarce, however. Almost no country in the region trains non-physician allied health professionals to perform genetic counseling, which remains a practice restricted to physicians. Equally of significant concern is the lack of education in genetics of policy makers, judges and the public at large.

2.5 Research in medical genetics

In line with the lack of governmental support for biomedical and epidemiological research in general, and the scarcity of resources and funds, genetics research in the region is far from the potential level it could be according to the talents, abilities and ingenuity of Latin American medical scientists. Several important exceptions were noted in the presentations and discussions.

2.6 Ethical, legal and social issues

Participants in the Consultation outlined the major ethical, legal and social issues in medical genetics in the region as follows.

- There are inequities in access to genetic services since public sector genetic services are scarce and under-funded. Private health insurance offers poor coverage of genetic conditions, and private services can be afforded only by the higher socio-economic classes
- Because termination of pregnancy for foetal reasons is illegal and there is lack of support in the public sector, prenatal diagnosis is restricted to the private sector. Only those with economic resources can access prenatal diagnosis and terminate an affected pregnancy safely.
- Problems exist surrounding the respecting of the privacy of genetic information.
- Discrimination exists towards people with disabilities, to patients with genetic conditions, to indigenous peoples and to carriers of the sickle-cell trait.
- There exists a lack of governmental regulation of genetic tests, resulting in the introduction in countries of the region of predictive genetic testing by commercial interests with neither clinical validation nor genetic counseling.
- Genetic research is conducted without proper informed consent.
- Genetic research is conducted by international scientists without technology transfer and capacity building in the host countries.
- Genetic exploitation of indigenous peoples exists and involves the taking of DNA samples for study abroad without any benefit sharing with communities where research was performed.

3. Recommendations

The Group recognized that recommendations per se do not necessarily contribute to change. Indeed, there has been no shortage of recommendations by groups of experts in medical genetics on how to develop genetic services in developing countries in general, and in Latin America in particular (PAHO, 1984, 1987; Beiguelman and Penchaszadeh, 1997; WHO, 1999, 2000, 2002). The challenge is to go beyond recommendations and to develop an actionable plan to bring about changes in the structure, financing and priorities of health services in the region, with particular emphasis on conditions with genetic influence. The Group proposed the following actionable recommendations and the strategies to implement them.

3.1 Medical genetics research

In order to further medical and human genetics research in the region, the Group proposed the following actionable recommendations.

- Develop countrywide accessible databases of existing research projects in medical genetics in research institutes, universities and other academic institutions of the region.
- Develop a system to disseminate results of research to the general public and policy makers.
- Improve efficiency and avoid overlaps by promoting collaborative research projects among academic institutions of the region and prioritizing research projects on diseases of impact and significance to the public's health.
- Identify and disseminate possible national and international sources of funding for medical genetics research in the region.

3.2 Epidemiology of genetic disorders and congenital malformations

The Group identified the study of the epidemiology of genetic disorders and congenital malformations as a high priority, in order to possess the necessary data to identify causes, plan prevention programmes and implement the care of affected children and their families. The specific actionable recommendations in this area are.

- Determine prevalence at birth, type, and medical and psychosocial cost of genetic disorders and congenital malformations in the countries of the region.
- Establish programmes to prevent exposure to teratogens in pregnancy.
- Fortify staple foods with folic acid, taking into account regional diets.
- Conduct feasibility studies to implement ultrasound in gestation for the detection of foetal malformations.
- Conduct feasibility studies to implement prenatal screening of Down syndrome by maternal serum biochemical markers and ultrasound, followed by prenatal diagnosis in positive cases.

3.3 Pre- and post-natal health services

- Implement reproductive health clinics at primary and secondary care levels, for comprehensive care of pregnant women.
- Establish a Task Force at the national level in each country of the region to establish the criteria for neonatal screening and recommend programmes, which use reliable and low cost technology for conditions meeting the defined criteria.

3.4 Common diseases

States should devote attention to the emergent epidemics of common diseases in the region's countries particularly cancer, cardiovascular conditions, diabetes and obesity conditions. The possible role of genomics in uncovering genetic susceptibilities and in helping prevent these conditions will require in-depth epidemiological studies and cost/effective analyses.

3.5 Genetic testing

Genetic testing should always occur within the context of health care, and be preceded by genetic counseling.

- The publicly supported health system, on which the majority of Latin American populations relies exclusively for medical care, should include in its services genetic counseling and testing (including prenatal diagnosis) and include provisions to ensure proper reimbursement.
- States of the region should develop appropriate simple regulatory systems for the assessment of safety and effectiveness of genomic technologies and discoveries before introducing and applying them in the health system.
- Genetic services should develop expertise in the technical, medical and ethical aspects of predictive genetic testing.

3.6 Ethical, legal and social issues

- Establish guidelines to acknowledge and promote respect of traditions, culture, religious beliefs and social expectations of individuals and communities in all aspects related to genetics, including reproduction, abortion, disability, race and ethnicity.
- Promote the creation of ethics committees in medical and research centres to address these aspects.
- Informed consent should be transformed into a meaningful educational process by which patients are fully informed about all implications of genetic testing, whether for research or medical care, in a manner devoid of any coercion.
- Centres that perform genetic testing should have norms to respect confidentiality and privacy of genetic information.
- Organize fora with participation of geneticists, ethicists and jurists to identify and address legal issues in genetics research and the provision of genetic services.
- Organize interdisciplinary relationships with policymakers to study legislation for the regulation of different genetic activities, including the marketing of predictive genetic tests, use of genetic testing in health insurance and the workplace, in vitro fertilization, gene therapy, stem cell research and cloning.

3.7 Genetic Laboratories

Undertake collaborative actions to.

- Establish a directory of genetic laboratories in the region that would include information on infrastructure, tests performed, methods and techniques, human resources and quality control.
- Establish networks of collaborating laboratories to avoid duplication of services, reduce costs and perform quality controls.
- Establish minimal guidelines for quality control of genetic testing.
- Enforce the appropriate use of biological samples, based on informed consent and confidentiality.
- Promote the inclusion of genetic testing, including prenatal diagnosis, in the coverage of health insurance plans.

3.8 Training of health professionals and the general public in genetics

- Recommend that health authorities create a special task force of specialists in the field of clinical, medical and human genetics, preferably chosen among members of the different genetics societies, in order to: (a) help publicly funded tertiary hospitals to create, expand and maintain medical genetics services with trained personnel and infrastructure to provide adequate services linked to primary health care at affordable costs for the local government; (b) promote initial training and continuing medical education for health professionals in the fields of clinical genetics, cytogenetics, biochemical genetics, and molecular genetics; (c) effectively integrate medical genetics in the public health system by creating jobs, within the system, for clinical geneticists, cytogeneticists, biochemical geneticists, molecular geneticists, and ancillary personnel in medical genetics.
- Establish programmes for the teaching of medical genetics to all health professions (physicians, nurses, psychologists, public health professionals, etc), both at undergraduate and postgraduate levels.
- Organize workshops for medical school leaders and faculty in developing countries to develop a core-curriculum in genetics tailored to national and regional needs.
- Target public health professionals and health policy makers for special courses and workshops on the relationship between genetics and public health. In addition to health professionals, important targets of workshops in genetics education include: policy makers and opinion leaders, patient/parent organizations, journalists, and the general public.
- Promote the development of educational materials for different targets according to local needs. Specific recommended actions are the development of training modules on topics such as genetic counseling and the application of genetics and genomics in clinical practice. Such educational materials should be culturally appropriate and harness the advances in information technology (CD ROM's, Internet, etc).
- The ethical, legal and social implications of medical genetics services and their supporting genomic technologies should be an integral part of the education in genetics at all levels and for all targets.

4. Proposal for a Latin American Task Force in Medical Genetics

The Group decided to form a permanent Task Force on Cooperation in Medical Genetics in Latin America, which will work in conjunction with the Latin American Network of Human Genetics (RELAGH). The Steering Committee for the Task Force will be constituted by Dr R. Giugliani and Dr A. Giraldo from RELAGH, and Dr V. Penchaszadeh, Chairperson of the Consultation. The following Working Groups were proposed as part of the Task Force:

4.1 Working Group on the Epidemiology and Prevention of Congenital Malformations and Folic Acid Supplementation Programmes (Coordinator: Dr Eduardo Castilla)

This Group will describe the frequency, characteristics and burden of congenital malformations in the region's countries, and will contact public and community health services to advise on the strategies for the prevention of birth defects. Special emphasis will be given to folic acid fortification of local foods for the prevention of neural tube defects.

4.2 Working Group on Pre and Post-Natal Care and Neonatal Screening (Coordinators: Dr Manuel Saborio and Dr Marcos Aguiar)

This Group will:

- promote measures to improve the access of women to reproductive health services and prenatal nutrition;
- promote actions for the detection of increased risks for birth defects and genetic diseases at the primary care level;
- delineate criteria for the inclusion of specific conditions for safe, cost/effective and equitable neonatal screening.

4.3 Working Group on Genetics Research (Coordinator: Dr Silvia Castillo)

This group will develop a database of current research projects being conducted in the region in basic and medical genetics, disseminate research themes and findings and strengthen the network of collaborative projects.

4.4 Working Group on Training of Specialists in Medical Genetics (Coordinator: Dr Decio Brunoni)

This Group will identify training centres in medical genetics in each country of the region, their training programmes, their curricula, their resources, and relationship with other academic and medical institutions.

4.5 Working Group on Education of Genetics to Health Professionals and General Public (Coordinator: Dr Joao Pina-Neto)

This Group will study the curricula in genetic education for undergraduates, postgraduates and health professionals, and will attempt to produce a basic curriculum. The Group also will address education and dissemination of genetic knowledge to the general public

4.6 Working Group on Genetic Laboratory Issues (Coordinators: Dr Roberto Giugliani and Dr Iscia Lopes-Cendes)

This Group will establish a directory of laboratories in the region performing genetic testing, for clinical and research purposes, including a list of tests performed by each laboratory.

4.7 Working Group on Ethical, Legal and Social Issues (Coordinators: Dr Alejandro Giraldo and Dr Jose M Cantu)

This Group will:

- study ethical, legal and social issues of research in medical genetics as well as the application of genetic knowledge to health promotion and protection;
- study the use of informed consent for genetic testing in research and clinical practice, privacy and confidentiality and genetic discrimination;
- analyse and produce recommendations on the ethical requisites for conducting predictive genetic testing;
- analyse legislation in the region on genetic issues and offer advice to legislators.

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