J Genet Counsel (2013) 22:869–874
DOI 10.1007/s10897-013-9607-1

Medical Genetics and Genetic Counseling in Chile

Sonia B. Margarit · Mónica Alvarado · Karin Alvarez · Guillermo Lay-Son

Received: 18 July 2012 / Accepted: 13 May 2013 / Published online: 7 June 2013

Abstract In the South American Republic of Chile genetic counseling is not currently recognized as an independent clinical discipline, and in general is provided by physicians with training in clinical genetics. At present only one genetic counselor and 28 clinical geneticists practice in this country of over 16 million inhabitants. Pediatric dysmorphology constitutes the primary area of practice in clinical genetics. Although the country has a universal health care system and an adequate level of health care, genetic conditions are not considered a health care priority and there is a lack of clinical and laboratory resources designated for clinical genetics services. Multiple educational, cultural and financial barriers exist to the growth and development of genetic counseling services in Chile. However, during the last 10 years increased awareness of the importance of identifying individuals at risk for inherited cancer syndromes led to growing interest in the practice of cancer genetics.

Keywords Genetic counseling · Clinical genetics · Chile · Cancer

S. B. Margarit (✉) · G. Lay-Son
Center for Human Genetics, Facultad de Medicina Clínica Alemana-Universidad del Desarrollo, Av. Las Condes 12438, Santiago, Chile 7710162
e-mail: Sonia.Margarit@gmail.com

M. Alvarado
Regional Genetic Services, Southern California Permanente Medical Group, Pasadena, CA, USA

K. Alvarez
Clínica Las Condes, Santiago, Chile

G. Lay-Son
Hospital Padre Hurtado, Santiago, Chile

Introduction

Chile is a long narrow country located in the southern cone of South America, isolated from neighboring nations by the Andes mountain chain. The country is divided from North to South into 15 administrative regions expanding over 292,000 mile^2. The Metropolitan Region includes the capital city of Santiago, the most densely populated region of the country, and comprises over 40 % of the population. The official language is Spanish.

Demographics

Chile's current population is approximately 16.5 million inhabitants (INE Chile 2012). The majority of the population (86.1 %) resides in the cities, whereas a small minority live in rural areas (INE Chile 2004). Ninety-five percent of the Chilean population self-identifies as Caucasian and "mestizo" (mixed Caucasian and Amerindian). Chilean mestizos are descended from an admixture between the conquering Spaniards and local natives groups. About 5 % of the population reports indigenous ethnicity, predominantly Mapuche(INE Chile 2003).

70 % of Chileans self-identify as Roman Catholic, 15.1 % Evangelical, 1.1 % Jehovah's Witnesses, 1 % other Christian, 4.6 % other, and 8.3 % are Agnostic or Atheist (INE Chile 2003). Chile has experienced a demographic transition which has led to an increase in the proportion of people aged 65 years and over. That age group made up 5.5 % of the population in 1980 compared to 9.2 % in 2010 (ECLAC/CEPAL 2009).

The United Nations' Human Development Index (HDI) provides a composite measure of health, education and income in member nations since 1990. The HDI calculated for Chile in 2011 was 0.805 which is above the Latin-American

average and places it in 44th place out of 187 countries. Table 1 compares development indicators for Chile compared to the U.S. and other countries of South America (UN Development Program 2011).

Health Topics

The current health problems in the Chilean population are similar to those in developed countries, due in part to the demographic transition mentioned above. The adult mortality rate is 87 per thousand. Non-communicable diseases (NCD) are prevalent, with cardiovascular disease (30 %) and cancer (23 %) being the main causes of death at all ages. Diabetes and other NCD contribute another 24 %(WHO 2012a).

The infant mortality rate has dropped to around 8 per thousand live births (INE Chile 2011), while congenital anomalies are the main cause of death in children under 5 years of the age (WHO 2012b). Chilean law prohibits termination of pregnancy following prenatal diagnosis of fetal abnormality or under any other circumstances. Prenatal diagnosis (amniocentesis) is performed usually to confirm the presence or absence of a chromosomal abnormality or a suspected genetic syndrome by ultrasound. This enables the healthcare team to anticipate a delivery at a tertiary care hospital and also prepares parents for the birth of a child with special needs.

The Chilean newborn screening program began in 1984 and was expanded nationwide in 1998. It includes phenylketonuria (PKU) and congenital hypothyroidism and covers 98.7 % of newborns. A recent pilot project explored the feasibility of an expanded program that could include a total of 9 inborn errors of metabolism (Cornejo et al. 2010).

The prevalence of genetic disorders is difficult to establish with certainty because there is currently no national registry of birth defects or official records for genetic conditions. However, data are available since 2001 from the Maule region (south-central region of the country) and also since 1960 from some hospitals participating in the Collaborative Study of Congenital Malformations in Latin America (ECLAMC). These data suggest that the global rate of major congenital anomalies in live births is 3.1 % (Castilla and Orioli 2004). The most common congenital anomalies per 10,000 births are: congenital heart disease (57.4), talipes (20.7), polydactyly (18.6), and cleft lip and/or cleft palate (18.1). Down syndrome has an incidence of 24.7 per 10,000 (Nazer and Cifuentes 2011). PKU has an incidence of 1 per 18,916 live births (Cornejo et al. 2010). Since 2000 wheat flour has been fortified with folic acid, and this is reported to have led to a decrease of 50 % in the rate of neural tube defects, from 17.1 to 8.6 per 10,000 births (Cortés et al. 2012).

Healthcare System

Chile has a complex, multifaceted health care system involving financing, health insurance and service delivery (PAHO 2007). Total health expenditures in Chile represent 8.2 % of the gross domestic product (GDP)(WHO 2012c). Since the 1980s, the National Health Trust (Fondo Nacional de Salud or FONASA) public insurance program coexists with many private insurance companies (known as Institutos de Salud Previsional or ISAPRES). FONASA beneficiaries comprise about 78.8 % of the population while 13.1 % belong to an ISAPRE. The remainder of the population is covered under other types of plans (e.g. armed forces insurance) or has no health insurance at all (Ministry of Social Development and Chile 2009).

The latest healthcare reform came in 2005 when an "explicit guarantee in health care" system (GES, "Garantías Explícitas en Salud") was established which includes a "Universal Access Plan with Explicit Guarantees" of service (AUGE Plan) for all health insurance plans. It includes guaranteed health services for 69 diseases relevant to the Chilean population (Nunez Mondaca 2011; PAHO 2007). Among them are congenital anomalies (congenital heart disease, cleft lip and/or palate, and neural tube defect), a number of adult cancers (breast, gastric, leukemia, testicular, cervical, prostate, brain) and all infantile cancer diagnosed less than

Table 1 International Human Development Indicators – United Nations Development (UN Development Program 2011)

2011	Chile	Argentina	Perú	Bolivia	Brazil	USA
Ranking of 187 countries	44	45	81	108	85	4
HDI	0.805	0.797	0.725	0.663	0.718	0.910
Mean years of schooling (adults)	9.7	9.3	8.7	9.2	7.2	12.4
Adult Literacy Rate	98.6	97.7	89.6	90.7	90.0	n.a.
Education Index	0.797	0.806	0.704	0.749	0.663	0.939
Health Index	0.932	0.882	0.852	0.735	0.844	0.923
Income GDP per capita*	13,057	13,202	7,836	4,013	9,414	41,761

GDP gross domestic product, *HDI* human development index

*GDP per capita (2005 PPP $): GDP derived from the purchasing power parity (PPP) expressed as constant 2005 international dollar

15 years of age. Two monogenic diseases (hemophilia and cystic fibrosis) are also covered by the AUGE plan (Ministry of Health and Chile 2012; Nunez Mondaca 2011).

Current Status of Genetic Counseling in Chile

In Chile, as in most Latin American countries, there are no formal post-graduate programs in genetic counseling, and no recognition of genetic counseling as an independent clinical discipline. Clinical Genetics became recognized as a medical specialty in 1997, however the profession is still underdeveloped with only one clinical genetics fellowship program for physicians (Castillo Taucher 2004). This is still true in 2013. Consequently, genetic counseling services are primarily provided by the few trained clinical geneticists available and other physicians involved in the care of patients with genetic disorders.

The Center for Human Genetics, Facultad de Medicina Clínica Alemana-Universidad del Desarrollo in Santiago, is to our knowledge the only center in the country that has a U.S.-trained, ABGC-certified genetic counselor (first author), working since 2004 as part of the clinical, research and educational team. She has been providing genetic counseling services for the division pediatrics and obstetrics at Padre Hurtado Hospital, a public hospital in Santiago since 2004. The most common indications for genetic counseling are diagnosis or positive family history of chromosomal alterations, skeletal dysplasia and recurrent miscarriages. Genetic counseling is well received by these families. Patients and their relatives express gratitude for the opportunity to learn more about genetics, risk of recurrence and management of the disorder and to also have the opportunity to talk about their concerns. The genetic counselor also provides hereditary cancer risk assessment at Clínica Alemana, a private clinic, since 2009, assisting providers and their patients with informed decisions about their cancer screening or treatment plans. The genetic counselor also offers genetic counseling to patients and families from support organizations such as the Center of Movement Disorders for Huntington's disease patients (CETRAM) and Dystrophic Epidermolysis Bullosa (Debra Chile). Furthermore, the Center of Human Genetics team at Padre Hurtado Hospital includes a genetics nurse trained at the Center who is responsible for the development of the early intervention program for Down syndrome.

Clinical and Laboratory Geneticists in Chile

To our knowledge there are 28 clinical geneticists currently practicing in the country, or about 1.6 geneticists per 1,000,000 inhabitants. Twenty-two out of 28 (79.1 %) genetics providers reside in the capital city of Santiago, and 6 geneticists reside and work in the rest of the country. Clinical Genetics is considered a primary specialty in Chile, but 12 of 28 practitioners (42.9 %) are also pediatricians. According to the UK Royal College of Physicians the recommended ratio of clinical geneticists to population is about 0.75 full-time equivalent (FTE) per 250,000 people (The Royal College of Physicians 2011). Based on that recommendation, Chile should have 49.7 full-time equivalents (FTE) of clinical geneticists. Considering that the public health system has 13.2 million beneficiaries (FONASA 2012), 39.6 FTE would be recommended, but we estimated that currently there are only 12.5 FTE, or 32 % of the ideal number. By comparison the recommended FTE for physician geneticists in the U.S. would be 1,232 yet there are only 540 FTE or 44 % of the ideal number (SACGHS 2010).

In order to learn more about the practice of clinical genetics in Chile, including an assessment of the current clinical genetics "workforce", type and hours of practice, demand for services, and barriers encountered, we recently conducted a brief survey with members of the Chilean Genetics Society and the Genetics Branch of the Chilean Society of Pediatrics. We received a total of 20 responses; the main results of this survey are summarized in Table 2. The vast majority of clinical geneticists work in the public and private healthcare systems, and most see up to 70 patients per month. The survey responses identified the key practice areas as pediatric dysmorphology and prenatal diagnosis. However, due to the few available specialists, most clinical geneticists practice in a general genetics clinic. Many also do teaching and work in research and clinical laboratories. The median length of professional practice among our respondents was 20 years. Since the rate of new graduates from the Chilean genetics fellowship program is 0.7 per year, we project that the number of clinical geneticists in Chile will have no significant changes over the next few years.

The country also faces a shortage of trained and certified laboratory geneticists. Currently there is no certification program similar to the Clinical Laboratory Improvement Amendments (CLIA) in Chile, however general regulations exist for quality and competence of all clinical laboratories (Instituto Nacional de Normalización 2003). There are over 15 clinical cytogenetic laboratories, mainly offering karyotype and FISH analysis, and some molecular genetics services for a limited number of genetics diseases (Castillo Taucher 2004). Recently, chromosome microarrays (array CGH) analysis became available for clinical diagnosis.

The majority of diagnoses for monogenic conditions are based on clinical evaluation. Confirmation by molecular genetic testing for certain genetic conditions can be performed by local clinical laboratories on case by case basis, and on occasion samples are sent outside of the country for genetic testing. Most genetic testing is not covered by health insurances and must be paid out-of-pocket.

Table 2 Results of survey of clinical geneticists

Professional experience (median)	20.5 years
Clinical practice (n=16)	
Patient care, public system	87.5 %
Patient care, private system	68.8 %
Working hours (n=16)	
Patient care, public system (mean)	20.9 h per week
Patient care, private system (mean)	10.8 h per week
Number of patients per month (n=20)	
Fewer than 20	26.7 %
21–50	33.3 %
51–76	26.7 %
76–100	6.7 %
More than 100	6.7 %
Areas of practice (main) (n=15)	
Dysmorphology	53.3 %
Clinical cytogenetics & molecular genetics	20.0 %
Teaching	13.3 %
Inborn errors of metabolism	6.7 %
Research	6.7 %
Areas of practice (all) (n=15)	
Dysmorphology	86.7 %
Prenatal diagnosis and Reproductive medicine	67.7 %
Teaching	67.7 %
Research	67.7 %
Clinical cytogenetics & molecular genetics	60.0 %
Neurogenetics	53.3 %
Inborn errors of metabolism	33.3 %
Clinical cancer genetics	33.3 %

Challenges to Development of Genetic Counseling Services in Chile

In view of the rapid changes that have taken place in medical genetics in the past 10 years there has been an increasing recognition of genetics advances among Chilean medical professionals and the public at large. However, there are still several barriers to the development of genetic counseling and clinical genetics services in Chile (Table 3).

One of the main barriers for the development of the genetic counseling profession in Chile is the lack of familiarity of Chilean physicians and the public with the concept and practice of allied health professionals like genetic counselors. Medical providers are either “doctors”, “nurses” or “midwives”, and the concept of a clinical genetics expert who does not fit into one of those categories is foreign to most, not just in Chile, but in Latin America in general. The lack of genetic counseling training programs in Chile and Latin America is another huge barrier which is unlikely to disappear in the near future. Both of these barriers contribute to a lack of understanding of the potential role and contributions of genetic counselors to the delivery of genetic services in Chile.

The lack of understanding of what genetic counseling entails, and of the level of clinical expertise that genetic counselors can provide, may lead some Chilean physicians to believe that genetic counseling should only be provided by physicians. Physicians in Chile are regarded as authority figures, and may be very directive in the advice they provide to their patients, consistent with a paternalistic doctor-patient relationship that is found particularly in communities of low social-economic status. Furthermore, some physicians in Chile may believe that genetic counseling focuses mainly on

Table 3 Barriers for genetic counseling and clinical genetics in Chile

Barriers for genetic counseling:

- Lack of awareness and recognition of the value of genetic counseling by the medical community and the public
- Belief that genetic counseling should only be provided by physicians
- No academic equivalent (post graduate degree)
- Currently only one genetic counselor in the country (first author)
- Little integration of genetics/GC within multidisciplinary health care team
- Lack of resources: inequity of health care services, especially to underserved populations
- No official guidelines for genetic counseling practice

Barriers in clinical genetics:

- Shortage of experts in the area
- Little or no funding for genetics fellowships
- No consensus in the integration of genetics in undergraduate/postgraduate medical curricula
- Not recognized as priority in clinical practice. New positions are scarce in the public health system
- Lack of appreciation of anticipatory medical care and role of clinical geneticist.
- Few local laboratories and limited assays available
- Disparate in the quality control regulation of clinical laboratories (such as CLIA)

educating their patients on the medical issues of their conditions, but little attention is given to the counseling aspects of the patient encounters and the emotional and psychological needs of patients and families. Taken together these attitudes and beliefs may lead to the idea that training in genetic counseling is not necessary, diminishing its importance and depriving patients of the benefits it could provide. Integration of genetic counseling into a multidisciplinary health care team also seems to create difficulties to health care providers. Some physicians are concerned that having another person provide genetic counseling might contradict the primary care provider and actually confuse instead of help the patients.

Another important barrier is the disparity of health care resources. The majority of the Chilean population is covered under a public health insurance plan with limited access to genetic services. Since genetic counseling is not recognized as an independent clinical discipline, health insurance coverage is not foreseen as a priority. Therefore, genetic counseling remains available only for patients who can afford to pay out of pocket for the consultation.

During the last decade increased awareness of the importance of identifying individuals at risk for inherited cancer syndromes for the purpose of screening and risk reduction fueled clinical and research interest in cancer genetics among Chilean healthcare providers. Enthusiasm generated by recent international cancer genetics conferences held in Santiago, motivated a group of clinical, laboratory and academic leaders to formulate a plan to develop an online training program to provide oncology health professionals in Chile and South America an opportunity to acquire clinical training in genetic counseling in the field of cancer genetics. The program is not intended to train health care professionals as future genetic counselors similar to post graduate degree in the U.S; however the organizers hope that it will improve access to cancer genetic counseling services, interest and support throughout the continent.

Conclusions

Healthcare providers in Chile are beginning to understand the value of genetic counseling. Given the numerous challenges to increase access to clinical genetics and genetic counseling services in Chile, there is a tremendous need to educate health professionals in these fields. The need for genetic education becomes even more relevant in the face of new advances of the human genome and personalized medicine that eventually will influence the practice of all health care providers. Development of a training program for hereditary cancer risk assessment may be the first step in raising awareness and interest in the practice of genetic counseling in Chile.

Acknowledgments We would like to acknowledge the Chilean clinical geneticists and cytogeneticists who participated in a mini-survey for this report.

References

Castilla, E. E., & Orioli, I. M. (2004). ECLAMC: the Latin-American collaborative study of congenital malformations. *Community Genetics, 7*(2–3), 76–94. doi:10.1159/000080776.

Castillo Taucher, S. (2004). Genetic services in Chile. *Community Genetics, 7*(2–3), 121–125. doi:10.1159/000080781.

Cornejo, V., Raimann, E., Cabello, J. F., Valiente, A., Becerra, C., Opazo, M., et al. (2010). Past, present and future of newborn screening in Chile. *Journal of Inherited Metabolic Disease*. doi:10.1007/s10545-010-9165-8.

Cortés, F., Mellado, C., Pardo, R. A., Villarroel, L. A., & Hertrampf, E. (2012). Wheat flour fortification with folic acid: changes in neural tube defects rates in Chile. *American Journal of Medical Genetics. Part A, 158A*(8), 1885–1890. doi:10.1002/ajmg.a.35430.

ECLAC/CEPAL. (2009). *Population projection. Latin America and the Caribbean. Demographic Observatory (Vol. IV)* (ECLAC/CEPALth ed.). Santiago de Chile: United Nations Publication.

FONASA. (2012). Informacion Corporativa / Estadisticas Institucionales. Retrieved from http://www.fonasa.cl/.

INE Chile. (2003). *Censo 2002, Síntesis de resultados*. Santiago de Chile: Empresa Periodística La Nación S.A. Retrieved from http://www.ine.cl/cd2002/.

INE Chile. (2004). *CHILE: Estimaciones y Proyecciones de Población por Sexo y Edad. País Urbano-Rural 1990–2020*. Retrieved from http://www.ine.cl/canales/chile_estadistico/demografia_y_vitales/demografia/demografia.php.

INE Chile. (2011). *Compendio estadistico 2011*. Retrieved from http://www.ine.cl/canales/menu/publicaciones/compendio_estadistico/compendio_estadistico2011.php.

INE Chile. (2012). *Resultados Preliminares Censo de Población y Vivienda 2012*. Retrieved from: http://www.censo.cl/.

Instituto Nacional de Normalización. (2003). *Norma Chilena Oficial NCh 2547 Of. 2003* Laboratorios Clínicos - Requisitos Particulares para la Calidad y Competencia. Retrieved from: http://www.inn.cl/.

Ministry of Health, Chile. (2012). *Acceso Universal Garantías Explícitas (AUGE)*. Retrieved June 28, 2012, from http://www.redsalud.gov.cl/portal/url/page/minsalcl/g_gesauge/presentacion.html.

Ministry of Social Development, Chile. (2009). *CASEN 2009*.

Nazer, J., & Cifuentes, L. (2011). Malformaciones congénitas en Chile y Latino América: Una visión epidemiológica del ECLAMC del período 1995–2008. *Revista Médica de Chile, 139*(1), 72–78. doi:10.4067/S0034-98872011000100010.

Nunez Mondaca, A. (2011, November 22). *Assessing equity in health system finance and health care utilization: the case of Chile, and a model to measure health care access*. Oregon State University. Retrieved from http://hdl.handle.net/1957/25931.

PAHO. (2007). *Chile. Health in the Americas: Vol. II. Countries, PAHO Scientific Publication* (pp. 196–217). Washington, D.C: Pan Amer Health Org.

SACGHS. (2010). *Genetics Education and Training of Health Care Professionals, Public Health Providers, and Consumers. Draft*

Report of the Secretary's Advisory Committee on Genetics, Health, and Society. Retrieved from:http://oba.od.nih.gov/SACGHS/sacghs_home.html

The Royal College of Physicians. (2011). *Consultant physicians working with patients. The duties, responsibilities andpractice of physicians in medicine* (5th ed.). London. Retrieved from www.rcplondon.ac.uk.

UN Development Program. (2011). *Human Development Report 2011. Sustainability and Equity: A Better Future for All* (p. 195). New York: United Nations. Retrieved from http://hdr.undp.org/en/data/profiles/.

WHO. (2012a). *Chile: Noncommunicable diseases country profile*. WHO. Retrieved from http://www.who.int/gho/countries/chl/country_profiles/en/index.html.

WHO. (2012b). *Chile: Health profile*. WHO. Retrieved from http://www.who.int/gho/countries/chl/country_profiles/en/index.html.

WHO. (2012c). *Global Health Observatory Data Repository*. Retrieved from http://apps.who.int/ghodata/.