Country Report

Public Health Genomics

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Ecuador: Public Health Genomics

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Key Words

Assessment · Birth defects surveillance · Ecuador · Genetic services · Genetic testing · Health technology · Newborn screening · Prenatal diagnosis

Abstract

Ecuador has a heterogeneous population of almost 14 million people and a complex health care system provided through provincial and national health programs by government and private hospitals. There are public health facilities at regional and territorial level. Ecuador has a small cadre of genetic professionals that provide clinical genetic services in a few private medical centers in the main cities. Prenatal screening is offered exclusively in a few individual hospitals, with variable uptake as part of prenatal care. Surveillance of the effect of prenatal screening and diagnosis on the birth prevalence of congenital anomalies is limited by gaps and variations in surveillance systems. Newborn screening programs are almost inexistent. There is broad variation in optional participation in laboratory quality assurance schemes, and there are no regulatory frameworks that are directly pertinent to genetic testing services or population genetics. Health technology assessment in Ecuador is conducted by a diverse collection of organizations, several of which have produced reports related to genetics.

Demographics

Ecuador is located in western South America, bordered by the Pacific Ocean at the Equator, between Colombia and Peru, at 2°00' South, 77°30' West [1]. It has a population of 13,979,416 inhabitants (May 2009 estimate) [2] and an annual population growth rate of 0.935% (2008 estimate) [2]. Ecuador is a multiethnic country with a strong native culture. It is located on latitude zero and has a land area of 283,560 km^2 with a density of 53.6 inhabitants per km^2 [3]. 54% of the residents live in urban zones, especially in the 4 largest cities: Quito, Guayaquil, Cuenca, and Santo Domingo (fig. 1). The country has 24 provinces distributed in 4 main regions: the Highlands (Sierra), Coast, Amazonia, and the Insular region [4]. Ecuador is a representative republic, and the religion is largely Roman Catholic (95%) although religious freedom exists [5]. Spanish is the official language, and indigenous languages are also spoken among aboriginal populations, especially Kichwa [6]. Education is compulsory between 6–14 years of age, but enforcement varies, with school attendance rates through 6th grade of 76% in metropolitan areas and 33% in country areas. Literacy is 92% [6].

Immigration Process

The internal migration in the past 100 years was concentrated in Sierra. Today the population is divided almost equally between Sierra and Costa. This migration,

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Fig. 1. Map of Ecuador with main cities.

particularly to the big cities in all regions, has also increased the urban population. The Amazon region, known as the Oriente (East), is located east of the Sierra and, although it constitutes nearly half of the Ecuadorian territory, remains sparsely populated, containing only 3% of the population, mostly indigenous Amazonians who always have been free and not subject to the Spanish conquest. These Amazonian indigenous nationalities have completely maintained their traditions, culture, and languages and have kept a wary distance to the rest of the Ecuadorian population, especially with the new Mestizos and white settlers who arrived in the interior, particularly after the 1970s when the national government launched a multinational exploitation of oil reserves in the area [7].

In the past 25 years, Ecuador has experienced 2 major waves of emigration, sending 10–15% of Ecuadorians overseas mostly to Spain, the U.S., Italy, Venezuela, and a small but growing number to Chile [8]. Due to an economic crisis in the late 1990s, more than 600,000 Ecuadorians emigrated to the U.S. and Europe from 2000 to 2001 [9]. According to the 2000 U.S. census, there were 323,000 people who claimed Ecuadorian ancestry although it is inofficially estimated that there are approximately 1 million Ecuadorians currently residing in the U.S. [3, 9]. While Ecuador continues experiencing emi-

gration, the number of immigrants into the country, particularly from Peru and Colombia, has increased in the last 5 years. Most Peruvians are economic migrants, while the majority of Colombians are refugees who have escaped armed conflicts and the hardships created by drug eradication programs [7]. The net migration rate is 7.98 migrants/1,000 people (2008 estimate) [2].

Health Care System

With annual earnings per capita of only $5,820, Ecuador is one of the poorest nations in Latin America, ranked 99 in the world [10]. This affects health status, as reflected in quite poor health indices. The general mortality rate is 4.1/1,000 inhabitants, infant mortality rate is 22.1/1,000, life expectancy is 76.81 years, and there was a maternal mortality rate of 90.2/100,00 live births in 2007 [2–4]. Some of the greatest health problems in Ecuador today include malnutrition, diabetes, and heart disease, which are the leading causes of death. Other usual problems include acute respiratory infections and diarrheic diseases, which have risen dramatically since 1990. In addition, there have been current reports of a remarkable increase in illnesses caused by the usage of pesticides, farm chemicals, and petroleum hydrocarbons by the country's large agribusiness industry [11, 12]. Ecuador has one of the greatest inequalities and the lowest investments in health in the region, surpassed only by Nicaragua, Honduras, Bolivia, and Haiti [10].

Segmentation is the characteristic of the health organization of Ecuador, with a general security system financed by contributions from employers and workers in the traditional sector, coexisting with a commercial sector which cares for the population of higher purchasing power and a government sector that cares for the poor [13, 14]. The diverse health care providers and payers include the Ministry of Public Health, the Social Security Institute (IESS), the Military Hospitals, the private for-profit sector, and many non-governmental organizations, which operate independently, sometimes overlapping their actions. Only ~15% of the overall population are covered by social security, whereof 10% belong to Seguro Social Campesino, which is a farmers insurance free of charge. While the government sector network covers 30% of the population, mostly the poor are without insurance. Other providers that cover tiny shares of the population are: Sociedad de Lucha contra el Cáncer (SOLCA), Junta de Beneficencia de Guayaquil (JBG), and the medical services of the armed forces and national police. The private sector provides the minority of the well-to-do population with insurance [10].

The public sector network of health services under the Ministry of Public Health (MSP) is a regional sized structure with 2 levels of decentralization: the provincial and cantonal health areas. There are economic, cultural, and geographical barriers that restrict access to health services and that particularly affect impoverished persons, mostly indigenous, living in country areas. Big cities concentrate health services, with significant inequities in access and privatization by for-profit entities. There is an epidemiological accumulation characterized by the coexistence of diseases of poverty (infectious and nutritional), with a rise in chronic diseases (cardiovascular diseases, cancer, diabetes, obesity, mental illness). Use of contraceptives is low, and induced abortion occurs extensively, with a high rate of unsafe abortions that are responsible for a high maternal mortality [13–15].

At the beginning of the recent millennium, the MSP initiated a practice of health care reform which, however, has been hindered by scattered and clumsy efforts from the state and unconnected from independent organizations, non-governmental organizations, and global agencies. Poor educational levels, minor social status of women, authoritarian traditions, and strict religious directives affect the medical practice of genetics. The major prenatal risk factors for genetic disorders in Ecuador are unplanned pregnancies (70% of all), advanced maternal age, poor prenatal care, rubella, widespread practice of self-medication, alcohol and tobacco use, nutritional deficiencies, labor risks, and reduced access to medical services [16].

Population Genetics Issues

Ethnicity is an essential factor for the epidemiology of genetic disorders as well as for the search of the founder effect of mutations. Currently, 3 primary ethnic groups reside in Ecuador: Mestizos, Amerindian natives, and Afro-Ecuadorian blacks. Mestizos are descendants of Europeans admixed with Amerindian natives and present the largest group, with almost 8 million inhabitants (60% of the overall population). A number of different Amerindian native populations, the most numerous of which are by far the Kichwa (often spelled Quichua), who number ~3 million inhabitants, retain their culture, language, and different identity [17]. They dwell primarily in the Andes highlands although some of them are equally found in the Amazonia region. Finally, half a million Ec-

uadorians descend from African slaves and retain distinct phenotypical features as well as African cultural traits. They live in country areas in 2 different provinces, in Valle del Chota (in the Andes) and in the coastal Esmeraldas province. The extent of the genetic admixture of the 3 primary ethnic groups is not even completely characterized [18, 19].

Genetic Diseases

There are no national programs in public health genomics, although moves towards that goal are beginning to occur. Very few medical institutions have comprehensive clinical genetics services, and patients with genetic conditions are seen by independent specialists in a variety of medical centers, public and private, across the country. These services are located largely in the commercial sector, provided by few clinical geneticists on a fee-for-service basis, and include genetic risk assessments, genetic counseling, clinical genetic diagnosis, and genetic testing for a range of conditions, including familial cancer and prenatal diagnosis. Statistics of genetic consultations performed in these institutions are seldom published and thus are unavailable to quantify the population prevalence of genetic diseases.

The rise of statistics used in this study belongs to the National Register of Hospital Admissions/Discharges from the Instituto Nacional de Estadísticas y Censos (INEC) [2] and data of the MSP [20]. The register is national, community-based, and funded by the government, and it has been extensively used in previous epidemiological studies. The Ministry of Health provides the information recorded that covers the entire country. Ecuador has no public official medical birth registry and neither a range of congenital malformations or disease-specific registries. Tables 1–3 show the absolute number of some genetic diseases and birth defects in the country among admitted patients, as published by the Vital Statistics National Report [2]. These figures should be taken as minimal estimates with uncertain diagnostic accuracy.

ECLAMC Network

ECLAMC is the Spanish acronym for Latin American Collaborative Study of Congenital Malformations. In Ecuador this network has been working since 1973 with 12 maternity hospitals in 6 provinces around the country and covered approximately 4.16% of total live births between June 2001 to June 2005 (66,843/1,605,452 live births). The associated hospitals are Hospital Carlos Andrade Marín (Quito), Maternidad Isidro Ayora (Quito), Hospital Rodríguez Zambrano (Manta), Hospital Napoleón Dávila (Chone), Hospital Miguel Alcívar (Bahía de Caráquez), Hospital Verdi Cevallos (Portoviejo), Hospital Luis Martínez (Cañar), Fundación Mano Amiga (Cañar), Hospital Homero Castanier (Azogues), Hospital San Vicente de Paúl (Ibarra), Hospital Isidro Ayora (Loja), and Hospital Teófilo Dávila (Machala). Table 4 shows the prevalence of major congenital malformations at birth reported by ECLAMC among 66,843 live births [21]. The ECLAMC network has also published 2 studies about cleft palate and holoprosencephaly [22, 23].

Special Conditions

Microtia

According to the ECLAMC study [21], the reported prevalence at birth of microtia/anotia varies from 1.15 to 17.4/10,000 in different countries (table 5), and Ecuador seems to carry the highest prevalence. Quito had a 5 times higher prevalence than the rest of South America (17.4×10^{-4}) and with greater degrees of severity [22, 23]. At the same time microtia/anotia was the most frequent congenital malformation (10.68/10,000 live births) compared with the neighbor countries included in that study. The official data on vital statistics ranked this malformation in 12th place [2]. Further studies are needed to unravel the causes of this higher prevalence of microtia in Ecuador.

Cystic Fibrosis (CF)

Several studies [32–34] reported the incidence of this disease and its 29 most common mutations in Ecuador's population. The 1st study included 62 unrelated patients. The estimated CF incidence in Ecuador was found to be 1:11,252 [13]. The incidence of the different mutations were ΔF508del (37.1%), G85E (8.9%), G542X (2.4%), N1303K (2.4%), G551D (1.6%), and R334W (0.8%). The incidence of CF in Ecuador is similar to other Latin American countries with a large Mestizo population [32]. This study found one of the highest incidences of G85E in the world, while other authors reported lesser frequencies [34, 35].

Hereditary Hemochromatosis

The only study of this disease reported mutation frequencies of 0.0, 0.035, and 0.04 for C282Y, H63D, and S65C, respectively, concluding that the type 1 form was

Table 1. The 25 most common congenital malformations reported among hospital admissions between 2001 and 2007 (absolute numbers)

Rank	ICD-10	Malformation	2001	2002	2003	2004	2005	2006	2007	Total	%
1	Q53	Undescended testes	838	757	699	698	730	896	1,051	5,669	11.03
2	Q25	Great vessels malformations	359	400	329	343	339	367	409	2,546	4.96
3	Q36	Cleft palate	452	437	324	285	357	340	318	2,513	4.89
4	Q35	Cleft lip	352	403	385	312	291	414	341	2,498	4.86
5	Q03	Congenital hydrocephaly	279	317	289	239	280	305	337	2,046	3.98
6	Q21	Cardiac malformations: ASD, VSD, PDA and others related	160	271	212	235	246	355	407	1,886	3.67
7	Q24	Other cardiac malformations	249	236	283	288	244	293	268	1,861	3.62
8	Q66	Feet defects: Talipes equinovarus, calcaneovarus and related	249	284	256	237	244	258	312	1,840	3.58
9	Q20	Congenital malformations of cardiac chambers and connections	309	429	242	213	175	214	256	1,838	3.58
10	Q65	Congenital deformities of hip	197	238	249	265	208	309	325	1,791	3.49
11	Q89	Unclassified congenital malformations	227	149	240	388	223	311	240	1,778	3.46
12	Q17	Microtia/anotia	132	144	148	162	198	210	304	1,298	2.53
13	Q90	Down syndrome	148	190	165	184	166	167	149	1,169	2.28
14	Q43	Other malformations of large and small bowels	136	176	163	150	166	190	162	1,143	2.22
15	Q38	Mouth, tongue, and pharynx malformations	99	123	137	135	135	190	275	1,094	2.13
16	Q75	Bone of skull and face	97	96	159	162	165	172	177	1,028	2.00
17	Q37	Cleft lip and cleft palate	106	150	160	149	157	157	106	985	1.92
18	Q69	Polydactily	134	124	107	97	128	132	138	860	1.67
19	Q04	Other malformations of the brain	87	142	139	92	124	104	141	829	1.61
20	Q79	Osteomuscular system malformations, unclassified	65	63	97	98	253	122	97	795	1.55
21	Q05	Spina bifida	125	162	94	86	106	102	116	791	1.54
22	Q55	Male genitalia malformations	110	57	87	141	189	98	102	784	1.53
23	Q54	Hypospadias	99	121	77	74	78	117	137	703	1.37
24	Q61	Polycystic kidney disease	81	103	109	76	90	96	134	689	1.34
25	Q74	Limbs malformations	75	50	80	78	111	84	101	579	1.13
Total										39,013	75.94

Total admissions during the 7 years analyzed = 5,462,700. Total cases of malformations = 51,375.
ASD = Atrial septal defect; ICD = international classification of diseases (WHO); PDA = patent ductus arteriosus; VSD = ventricular septal defect.

more influenced by the H63D mutation than by the other 2 mutations [36]. More studies are needed in our population to better understand the role of the different mutations in the expression of hemochromatosis.

Other Diseases

The medical literature about genetic diseases and congenital malformations in Ecuador is very scarce. Few genetic diseases have been reported so far. One study shows the relationship between cleft lip and deficiency of folic acid [37], a hydrolethalus syndrome case [38], a neurocutaneous syndrome case [39], a McArdle's disease case [40], and 2 novel mutations in retinoblastoma [41]. Prior studies on public health and genetics in Ecuador have been published [42–45]. More research is needed to better assess the role of genetic factors in health and disease in Ecuador.

Genetic Testing

A range of diagnostic genetic tests are performed in the private sector. As of 2008, more than 60 different genetic tests were available in Ecuador in 15 laboratories

Table 2. Most common chromosome abnormalities reported among hospital admissions between 2001 and 2007 (absolute numbers)

Rank	ICD-10	Chromosome abnormality	2001	2002	2003	2004	2005	2006	2007	Total	%
1	Q90	Down syndrome	148	190	165	184	166	167	149	1,169	72.6
2	Q99	Other chromosome anomalies like chimeras 46,XX/XY, hermaphroditism	12	20	11	35	55	104	9	246	15.3
3	Q96	Turner syndrome	12	6	11	8	8	8	8	61	3.7
4	Q92+Q93+Q95	Other autosomal chromosome abnormalities: trisomies, monosomies, rearrangements (translocations and deletions)	2	16	8	3	7	9	7	52	3.2
5	Q91	Edwards and Patau syndrome	7	3	6	5	8	11	3	43	2.7
6	Q97+Q98	Other sex chromosome abnormalities (Klinefelter syndrome, 47,XXX, 47,XXY)	17	9	4	3	5	1	5	39	2.4
Total										1,610	100.0

Table 3. Most common single gene diseases and metabolic disorders reported among hospital admissions between 2001 and 2007 (absolute numbers)

Rank	ICD-10	Pathology	2001	2002	2003	2004	2005	2006	2007	Total	%
1	E00+E03	Congenital iodine deficiency and congenital hypothyroidism	111	114	133	146	163	163	220	1,050	23.08
2	D58	Hereditary hemolytic anemias	145	132	120	110	137	128	154	926	20.35
3	E78	Familial hypercholesterolemia and hyperlipidemias	56	51	62	64	78	66	57	434	9.54
4	G12	Congenital muscular atrophy	108	102	34	46	32	16	50	388	8.53
5	E83	Hereditary hemochromatosis	42	91	68	41	38	45	54	379	8.33
6	E84	Cystic fibrosis	41	46	46	39	38	50	77	337	7.41
7	G11	Hereditary ataxia	20	27	27	33	29	25	32	193	4.24
8	G71	Muscular dystrophies (Duchenne, Becker, and myotonic)	15	25	27	26	29	33	20	175	3.85
9	E74	Fructosuria, galactosemia	10	15	11	14	15	25	17	107	2.35
10	G60	Hereditary neuropathy	18	20	10	28	13	6	12	107	2.35
11	E71	Maple syrup urine disease and related	13	11	11	18	14	11	7	85	1.87
12	E73	Lactose intolerance	7	9	13	16	13	6	9	73	1.60
13	D82	Genetic immunodeficiency and DiGeorge syndrome	15	11	4	7	8	10	7	62	1.36
14	E75	Gangliosidosis, sphingolipidosis, and related like Gaucher's disease	6	3	5	11	9	11	9	54	1.19
15	D56	Thalassemias and congenital traits	4	4	3	4	6	9	7	37	0.81
16	E72	LCAT/MCAT, ornithine pathway disorders	3	3	7	0	10	6	7	36	0.79
17	E76	Glycosamine metabolism disorders (Hurler and Hunter's syndromes)	2	4	3	4	14	2	4	33	0.73
18	E25	Congenital adrenogenital syndrome	6	5	3	5	3	4	6	32	0.70
19	G10	Huntington's disease	5	2	2	9	3	1	1	23	0.51
20	E70	Phenylketonuria, hyperalalinemia, tyrosin disorders, albinism, and related	2	1	1	0	4	3	8	19	0.42
Total										4,550	100.00

LCAT = Lecithin-cholesterol acyltransferase deficiency; MCAD = medium chain acyl CoA dehydrogenase.

Table 4. Prevalence of major congenital malformations at birth per 10,000 among 66,843 live births in 12 maternity hospitals reported by ECLAMC, June 2001 to June 2005

Rank	ICD-8	Malformation	n	Prevalence
1	749A	Cleft lip	120	18.0
2	7550	Postaxial polydactyly	90	13.5
3	7541	Talipes equinovarus	73	10.9
4	745A	Microtia/anotia	72	10.8
5	7420	Hydrocephaly	37	5.5
6	741	Bifid spine	28	4.2
7	7490	Cleft palate	28	4.2
8	7512	Imperforate anus	26	3.9
9	7400	Anencephaly	21	3.1
10	746SE	Septal defects	20	3.0
11	7550B	Preaxial polydactyly	20	3.0
12	7502	Esophageal atresia	19	2.8
13	7551R	Syndactyly	18	2.7
14	7522	Hypospadias	17	2.5
15	755/2	Limb agenesia	17	2.1
16	7531	Polycystic kidney	15	2.2
17	746TO	Trunk conal defect	14	2.1
18	7542	Talipes talovalgus	13	1.9
19	7431	Microcephaly	13	1.9
20	7532	Congenital hdronephrosis	12	1.8

attached to or affiliated with a private hospital or university (table 6). Most testing requires patients to partially or fully pay out-of-pocket although exceptionally testing is offered free of charge. Most genetic testing occurs out of the context of comprehensive genetic services, and no regulations for genetic counseling are in place. Predictive testing is not available.

Genetic Screening

Genetic screening as such is not practiced in Ecuador. Prenatal screening for Down syndrome, other fetal chromosomal conditions, and fetal anomalies is increasingly being performed as part of prenatal care but only in the private sector, which requires out-of-pocket expense: (a) Fetal ultrasound in the 1st trimester for nuchal translucency is performed only when indicated by the obstetrician. (b) First trimester combined screening by nuchal translucency measurement and maternal blood biochemical markers (pregnancy-associated plasma protein [PAPP-A] and human chorionic gonadotropin [free beta-hCG]) is quickly becoming a standard test for well-to-do women who pay out-of-pocket. (c) Second trimester maternal serum screening (alpha-fetoprotein, unconjugated estriol, free beta-hCG) is accessible only in the large cities. No information is available on quality control or utilization or outcome of these tests. There is no state program of newborn screening. Only a few private hospitals offer newborn screening on a pay-for-service basis for CF, phenylketonuria, galactosemia, congenital hypothyroidism, amino acid disorders, fatty acid oxidation disorders, and organic acid disorders. There are no population-based carrier genetic screening programs for any particular genetic condition in Ecuador.

Genetic Interest Groups

Genetic support organizations exist for a few genetic conditions like Fundación Ecuatoriana de Fibrosis Quística [46] and FEPAPDEM (Federación Ecuatoriana Pro Atención a la Persona con Deficiencia Mental, Parálisis Cerebral, Autismo y Síndrome de Down) [47]. There are neither programs in public education of genetics nor information strategies about rare disorders. Information for patients and families is developed in an ad hoc manner across provinces and generally reflects locally available services.

Professional Involvement and Accreditation

In 1990 the Colegio Médico de Pichincha recognized medical genetics as a medical specialty. About 20 medical professionals practice medical genetics in the country. Although certification is not available, there are no professional education programs in the country for genetic counselors or clinical geneticists.

Discussion

The World Health Organization sponsored a consultation on community genetic services and a regional network of medical genetics in Latin America in 2003 [48]. The main recommendations of the conference included the necessity for government funding of genetic services, research, and teaching in medical genetics, the conduct of epidemiological research on the prevalence and types of birth defects, genetic disorders, and genetic predispositions to common diseases, the training of health professionals in genetics, the education of genetic professionals in community health and public health genetics, the fos-

Table 5. Comparison of the prevalence of microtia around the world

Prevalence[a]	Country	No. of cases	Births	Authors	Year	Reference
17.40	Ecuador, ECLAMC	80	46,041	Castilla and Orioli	1986	[24]
12.00	USA, Navajo Indians	19	15,890	Nelson and Berry	1984	[25]
10.80	Ecuador	72	66,843	Montalvo et al.	2008	[21]
4.34	Finland, FRCM	335	771,425	Suutarla et al.	2007	[26]
3.79	USA, Hawaii	120	316,058	Forrester and Merz	2005	[27]
3.20	Latin America, ECLAMC	175	553,068	Castilla and Orioli	1986	[24]
2.35	Sweden	954	1,950,148	Harris et al.	1996	[28]
2.16	USA, California	636	2,537,099	Shaw et al.	2004	[29]
2.00	USA, California	NA	1,921,698	Harris et al.	1996	[28]
1.46	Italy	172	1,173,794	Mastroiacovo et al.	1995	[30]
1.40	China	453	3,246,408	Zhu et al.	2000	[31]
1.15	France, Central-East	152	1,319,757	Harris et al.	1996	[28]

[a] Prevalence rate by 10,000 births.

FRCM = Finnish Register for Congenital Malformations; ECLAMC = Latin American Collaborative Study of Congenital Malformations.

Table 6. Laboratories that perform genetic testing in Ecuador

City	Laboratory (original name)	Type	Genetic tests offered
Quito	Hospital Metropolitano & Diagen	Pi	Clin, Mol, For, Imm, Micro, Cyt, Onco, Met
	Cruz Roja Ecuatoriana	Pi	For
	Hospital del Cáncer, SOLCA Quito	Pi	Cyt, Onco
	Biomedical Center, Universidad Central	Pu	Clin, Cyt
	Laboratorio Génica	Pi	Clin, Cyt, Met
	Hospital Carlos Andrade Marín	Pu	Clin, Cyt, Met
	Hospital de las Fuerzas Armadas	Pu	Clin, Cyt, Met
	Laboratorio de Genética, PUCE	Pi	Cyt, Mol, For
	Escuela Politécnica del Ejército	Pu	Micro
	Net Lab	Pi	Micro
Guayaquil	Hospital del Cáncer, SOLCA Guayaquil	Pi	Cyt, Mol, Onco
	Centro de Biomedicina, Universidad Católica de Guayaquil	Pi	Cyt, For
	Instituto Nacional de Higiene, Ministerio de Salud Pública, MPH	Pu	Micro
Cuenca	Hospital del Cáncer, SOLCA Cuenca	Pi	Cyt, Mol, Onco
	Laboratorio Biomolecular	Pi	For

Clin = Clinical genetics; Cyt = conventional cytogenetics; Diagen = Diagnóstico e Identificación Genética, LLC; For = forensic genetics; Imm = immunogenetics; Met = newborn screening; Micro = molecular microbiology; Mol = molecular diagnostic; Onco = molecular oncology; Pi = private; Pu = public.

tering of interactions between clinical geneticists, government health personnel, primary health care workers, and community organizations, and a superior planning of regional sized services to avoid duplication and inefficiency.

In Ecuador there is a need for a central coordination of information on service availability of prenatal diagnosis, newborn screening, and genetic testing and also of assessment of modern genetic technologies. As in numerous additional countries, there is no established framework for evaluating genetic/genomic testing and related interventions. Although there is a high level of volunteer participation in quality assurance of genetic testing, current experiences in additional areas of laboratory medicine suggest a call to boost surveillance of molecular genetics laboratories. The prevalence of birth defects and single-gene disorders is hard to determine in the dearth of well-conducted epidemiological studies. Reliance on routine reporting to vital statistics is far from flawless. The picture would be more complete if data from genetic services, largely ambulatory, were reported. While no particular single gene disorder has an outstanding prevalence, microtia/anotia has reportedly a higher prevalence than in other countries, a feature that still requires explanation. There is a gap between genomic science and health applications. Genomics research is not connected to prevention programs or to health services. There are considerable hurdles in the setting and implementation of policies for the prevention and handling of congenital and genetic disorders. Comprehensive clinical genetic services in the household sector are poorly developed and with greatest inequities in approach and emphasis on individual diagnostic services and little focus on genetic counseling. Newborn screening is practically inexistent. There is an insufficient coordination of separate health services, competition for available funds with more programs, and a lack of population-based prevention programs. Programs of primary prevention of congenital defects must be developed and implemented by the Ministry of Health, in particular an expansion of rubella immunization, a broad program of folic acid fortification, strong and better campaigns against alcohol consumption and tobacco abuse in pregnancy, and an increase in the knowledge of health care providers and the general public on avoidance of potential teratogens. It will be necessary to establish a proper linkage between clinical genetic services and basic health care to improve the administration of genetic testing and counseling services and to explain the role of genetic testing in general health. In addition, it will be essential to enhance and enlarge the registry of congenital malformations and to connect it with established policies in health.

Conclusions

Ecuador has a relatively small but genetically diverse population spread over the country. This has significant implications for the successful delivery of genetic prevention programs and clinical genetic services that answer the needs of all Ecuadorians. Although tensions between the public and commercial sectors have prevented further progress, there is a growing need for the Ministry of Health to promote a public policy within a predominantly state-funded system for a range of genetic issues such as newborn screening, genetic testing, and health professional education. Further work is imperative to establish suitable frameworks for the proper regulation and funding of new genetic tests across state boundaries, which will be critical in establishing a nationwide approach to community health genomics policy.

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