Public health approach to birth defects: the Argentine experience

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Abstract Birth defects are a global problem, but their impact is particularly severe in low and middle income countries, where the conditions for prevention, treatment, and rehabilitation are more critical. The epidemiological transition in the infant mortality causes, and the concern of the community and the mass media about the teratogenic risk of environmental pollutants, has made health authorities aware of the importance of birth defects in Argentina. The objective of this paper is to outline those actions specifically taken in Argentina aimed at the prevention of birth defects at a national level. Firstly, we focus on birth defects in Argentina on a general basis, and then we present different laws and actions taken in terms of surveillance and public health programs, primary, secondary, and tertiary prevention. Finally, we present the Teratology Information Service “Fetal Health Line”, and the genetic services organization and health professionals training by the National Center of Medical Genetics and the National Program of Genetics Network. In conclusion, in the country, several programs focus on different approaches to the problem, and the challenge is to coordinate the teamwork between them. Finally, we list tips to address birth defects from the public health perspective.

Keywords Argentina · Public policies in medical genetics · Birth defects surveillance · Genetics services · Training in medical genetics

Introduction

Birth defects are structural or functional defects that originate during the prenatal period. Birth defects affect different organ systems and are etiologically heterogeneous, either multifactorial (predisposing genes that are expressed in the presence of environmental triggers), predominantly genetic (chromosomal or monogenic abnormalities), or predominantly environmental (prenatal exposure to teratogenic agents) (Stevenson et al. 1993).

Globally, birth prevalence is 1–5 %, depending on the inclusion criteria. It is estimated that each year 7.9 million children are born worldwide with major birth defects. Among them, at least 3.3 million die before age 5 and 3.2 million survive with a disability. Birth defects are a global problem, but their impact is particularly severe in low and middle income countries, where the conditions for prevention, treatment, and rehabilitation are more critical (Christianson and Howson 2006). Recently, the World Health Assembly stressed the importance of addressing birth defects to meet the Millennium Development Goal 4 (“to reduce by two thirds, between 1990 and 2015, the under-five mortality rate”) (WHA 2010).

The epidemiological transition that took place in recent decades has led to the reduction of communicable diseases as a cause of death and the relative increase in chronic conditions, including birth defects. In Argentina, as in many low and middle income countries, birth defects have become the leading cause of infant mortality. Moreover, their impact is not only observed in infant mortality but also in spontaneous abortion, lower survival of those affected, comorbidity, high demand for medical and surgical treatments, social and emotional impact, and high economic costs.

The objective of this paper is to outline those actions specifically taken in Argentina aimed at the prevention of birth defects at a national level. Firstly, we focus on birth defects in Argentina on a general basis, and then we present surveillance and public health programs, laws and actions

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taken in terms of prevention (primary, secondary, and tertiary), genetic services, and health professionals training.

Argentine background

Argentina is a middle-income country located in southern South America. It is a federal republic consisting of 24 jurisdictions or provinces. From a historical and geographical criterion, Argentina can be divided into five regions: Central (the most populous), Cuyo (west), Northwest, Northeast, and Patagonia (south). It has a population of 40,117,096 inhabitants, 89.4 % of whom live in urban areas. The continental part has an area of 2,800,000 km², with a low population density: 14.4 people per km². Around 65 % of the population is concentrated in the central region provinces, particularly in the province of Buenos Aires, with 38.95 % of the country population (INDEC 2010).

The health care system in Argentina is divided in three settings: public, social security, and private insurance. The public setting is funded through taxes and is available free of charge to the entire population. This setting serves approximately 46 % of the population, mainly the lower income population, who lacks other health coverage. The social security setting is funded by mandatory contributions from employers and registered workers, covering about 44 % of the population (workers, employees, and retirees). The private insurance setting (for-profit) is funded by specific payments from the insured and serves 10 % of the population, mainly the higher income population. The three systems have little coordination between them, which results in overlapping, inefficiency, and high health expenditure (about 6.61 % of the Gross Domestic Product). The National Ministry of Health (NMoH) has a leading role in public health policy, is responsible for introducing specific regulations, and runs national programs (i.e., immunizations, noncommunicable diseases, sexual and reproductive health, and maternal and child health). On the other hand, provincial ministries of health are autonomous in terms of planning and decision making on human resources, purchases, infrastructure, and other actions to provide services to their people. Given its different settings and administrative jurisdictions, the Argentine health care system could be considered a fragmented model (PAHO 2011).

In 2001, there were approximately 900 health care facilities to serve 400,000 public setting births. Most of them assisted less than 1000 births per year and covered only 30 % of births in the public setting. These mothers and their children had a higher risk of complications and death, since over 60 % of these institutions did not meet the essential obstetric and neonatal requirements (available and suitable operating room, neonatal anesthetic management, safe blood for transfusions, patient transport, and immediate neonatal care) (Speranza and Kurlat 2001). To address this problem, the NMoH developed a regionalization strategy, by concentrating births in health

centers with high number of deliveries, which is associated with better outcomes regarding maternal and neonatal mortality.

In 2012, there were 738,318 live births, with 99.6 % of deliveries occurring in hospitals. The infant mortality rate was 33.3 per 1000 live births in 1980 and dropped to 11.1 per 1000 in 2012. Neonatal deaths (<28 days) represent two thirds of infant mortality (DEIS 2013). The lower the infant mortality rate, the higher the proportional infant mortality attributable to birth defects (Fig. 1). It was approximately 10 % in 1980, rising from 17.9 to 23.6 % between 1998 and 2009 and reaching 26 % in 2012. Among 8227 infant deaths that occurred in 2012, 2175 were due to birth defects. Nowadays, birth defects are the leading cause of infant deaths, surpassing deaths by disorders related to short gestation and low birth weight. Therefore, although infant mortality attributable to birth defects has diminished, this decrease was lower than other causes of infant death. That is the reason why the proportional mortality attributable to birth defects has increased. According to the study on disease burden, in 2005, birth defects in Argentina were the cause of 86,400 years of life lost due to premature death and 104,919 disability-adjusted life year (DALYs) (Borruel et al. 2010). The criteria for reducibility of infant deaths were recently reviewed by the NMoH. The current classification includes many birth defects previously considered irreducible, as reducible, thanks to prevention, diagnosis, and early treatment. This is mainly observed in the newborn through surgical or medical treatment (DEIS 2012). In Argentina, the birth prevalence of major structural birth defects over the 2009–2013 period was 1.62 % (RENAC 2014). The prevalence of selected major structural birth defects is shown in Table 1.

Surveillance

Birth defect monitoring systems were started after the thalidomide tragedy to monitor the frequency of birth defects, detect geographic clusters, and research on risk factors. Until recently, the only epidemiological data available on birth defects in Argentina was the Latin American Collaborative Study of Congenital Malformations (ECLAMC), founded in 1967 in Argentina, and then expanded to other South American countries (Castilla and Orioli 2004). The ECLAMC is a voluntary network of people focused on the study of birth defects risk factors. In the 1960s, infant deaths were mainly attributable to infections and malnutrition. Since birth defects were not health priorities in the region, the ECLAMC developed as a research program without the support of health ministries.

Nowadays, 99.4 % of deliveries in Argentina occur in hospitals and are assisted by neonatologists or pediatricians (DEIS 2013). On this basis, in 2008, members of the National Center of Medical Genetics and the ECLAMC conducted a

Fig. 1 Infant mortality rate per 1000 live births (IMR) in Argentina, 1980–2010. From Annual reports of the Department of Health Statistics and Research, Vital Statistics National Ministry of Health, 1981, 1986, 1991, 1996, 2001, 2006, and 2011

pilot study to develop a public health surveillance system of birth defects (Liascovich et al. 2011). This pilot study led to the development of the National Registry of Congenital Anomalies (RENAC), which was started in November 2009 in the National Center of Medical Genetics as a hospital-based system. At the beginning, RENAC included public maternity hospitals with more than 1000 deliveries per year, and it was designed with the goal of being a sustainable and scalable system at the national level. In 2013, the RENAC began to include nonpublic maternity hospitals and nowadays gathers 134 public and 7 nonpublic hospitals from the 24 jurisdictions. Annual coverage is approximately 280,000 births, which accounts for 65 % from the public setting and 38 % of all births in the country (RENAC 2014). Birth defect surveillance systems have a big potential for early detection of infants with birth defects, thus allowing referral to health services (Botto and Mastroiacovo 2000; Luquetti and Koifman 2011). The RENAC leads to systematic newborn screening of birth defects by physical examination, thus allowing greater awareness of this health problem. Since genetic services are still underdeveloped in some provinces, the RENAC provides information for early clinical management and about local medical genetic resources.

The inclusion criteria are restricted to live births or stillbirths weighing 500 g or more, with major structural birth defects identified from birth until discharge. The reporting neonatologists collect information about the affected cases and send the data to the RENAC coordination through a web-based forum. The website allows sending data, solving operational issues, and providing quality assurance of data. The coordination team suggests diagnosis, the search for associated birth defects, and referral to genetic services. Forum interaction increases social cohesion among participants who feel themselves members of the same team. The RENAC holds an annual meeting, which is essential for face-to-face interaction of members. There is also a blended-learning course for the reporting neonatologists (Groisman

et al. 2013a, b). Since 2013, the RENAC has become an active member of the International Clearinghouse for Birth Defects Surveillance and Research (ICBDSR) and is a world affiliated member of the EUROCAT. Table 2 shows the milestones in the development of the RENAC.

Primary prevention of birth defects

In this section, we include two of the most important prevention measures implemented in the last years: rubella vaccination and folic acid fortification (FAF). Both interventions were intended for the entire community, regardless of intention to conceive. This approach has the advantage of reaching unplanned pregnancies.

Folic acid

Periconceptional folic acid intake through supplementation and fortification prevents neural tube defects. Regarding supplementation, the Argentine guidelines suggest that folic acid should be taken at least one and a half month before conception and up to 12 weeks after. Periconceptional use of 0.4 mg/day of folic acid is recommended to prevent the occurrence of NTDs and 4 mg/day of folic acid to prevent recurrence in women with previous affected children (Department of Maternal and Child Health, NMoH, 2013). However, a survey conducted in two regions of Argentina showed that around 25 % of pregnant women use periconceptional folic acid supplements. Pregnant women with higher levels of education who attend private health centers are more likely to use periconceptional folic acid supplements (Zabala et al. 2008). A survey conducted in one of the Argentine provinces estimated that around 65 % of pregnancies are unintended (Palena et al. 2009). One reason for the relatively low usage of supplements in Argentina is the high rate of unintended pregnancies, and the gaps in knowledge of

Table 1 Prevalence per 10,000 of selected birth defects in Argentina in the November 2009–December 2013 period

Birth defects (ICD-10 codes)	Number	Prevalence (95 % CI)
Anencephaly (Q00)	212	3.0 (2.6–3.4)
Encephalocele (Q01)	97	1.4 (1.1–1.7)
Hydrocephalus (Q03)	536	7.6 (7.0–8.3)
Spina bifida (Q05)	389	5.5 (5.0–6.1)
Transposition of great vessels (Q20.3)	115	1.6 (1.3–2.0)
Tetralogy of Fallot (Q21.3; Q21.82)	128	1.8 (1.5–2.2)
Atrial septal defect (Q21.1–Q21.18)	685	9.7 (9–10.5)
Ventricular septal defect (Q21.0)	1357	19.3 (18.3–20.3)
Cleft palate (Q35)	220	3.1 (2.7–3.6)
Cleft lip ± cleft palate (Q36–Q37)	826	11.7 (11.0–12.6)
Esophageal atresia (Q39.0–Q39.11)	230	3.3 (2.9–3.7)
Intestinal atresia (Q41.1–Q41.9)	103	1.5 (1.2–1.8)
Duodenal atresia (Q41.0)	118	1.7 (1.4–2)
Anorectal malformation (Q42.0–Q42.3)	231	3.3 (2.9–3.7)
Diaphragmatic hernia (Q79.0–Q79.01)	225	3.2 (2.8–3.6)
Ambiguous genitalia (Q56.4)	137	1.9 (1.6–2.3)
Hypospadias (Q54)	87	1.2 (1.0–1.5)
Bilateral renal agenesis (Q60.1)	65	0.9 (0.7–1.2)
Talipes calcaneovalgus (Q66.4)	89	1.3 (1.0–1.6)
Talipes equinovarus (Q66.0)	484	6.9 (6.3–7.5)
Non specified Talipes (Q66.8)	183	2.6 (2.2–3.0)
Limb reduction defect, transverse (Q71.2–Q71.30)	207	2.9 (2.6–3.4)
Limb reduction defect, preaxial (Q71.31; Q72.5)	109	1.5 (1.3–1.9)
Limb reduction defect, postaxial (Q71.5; Q72.6)	29	0.4 (0.3–0.6)
Limb reduction defect, intercalar (Q71.1; Q73.1; Q72.4)	26	0.4 (0.2–0.5)
Non specified limb reduction defect (Q71.8–Q71.9; Q72.8–Q72.90)	163	2.3 (2.0–2.7)
Omphalocele (Q79.2)	167	2.4 (2.0–2.8)
Gastroschisis (Q79.3)	600	8.5 (7.9–9.2)
Down syndrome (Q90)	1220	17.3 (16.4–18.3)

Prevalence was calculated over 703,422 examined newborns in the November 2009 to December 2013 period. From the National Registry of Congenital Anomalies (RENAC)

CI Confidence interval

both the general population and health professionals about the prevention of NTDs (Barbero et al. 2003).

FAF of wheat flour and its derivatives is mandatory (National Law number 25630 (2002)). The fortification includes folic acid, iron, thiamine, riboflavin, and niacin. The amount of folic acid added to flour is 2.2 mg/kg, assuming an estimated intake of 160 g of bread per day, thus yielding a mean intake of 0.35 mg of folic acid. The fortification act was enacted in 2002, and the population began to receive fortified food by the end of 2003. A study conducted in Brazil, Chile,

Table 2 Development of the National Registry of Congenital Anomalies (RENAC) in Argentina

Year	Milestone
2008	A pilot study between ECLAMC and the Ministry of Health was conducted to evaluate the feasibility of RENAC.
2009	RENAC covered 2,549 annual births in four member hospitals from four provinces in the Northeast region. Public setting coverage is 0.6 %. Whole country coverage is 0.3 %.
2010	RENAC covered 39,587 annual births in 31 member hospitals from 3/5 regions. Public setting coverage is 9.6 %. Whole country coverage is 5.2 %.
2011	RENAC covered 140,066 annual births in 78 hospitals from 5/5 regions. Public setting coverage is 34.0 %. Whole country coverage is 18.5 %.
2012	RENAC covered 239,971 annual births in 118 hospitals. Public setting coverage is 56.6 %. Whole country coverage is 32.5 %.
2013	RENAC covered 281,249 annual births in 141 hospitals. Public setting coverage is 65.0 %. Whole country coverage is 38.1 %.

and Argentina (López-Camelo et al. 2010) observed a significant decrease in the birth prevalence of NTDs after FAF: a reduction of 41 % for anencephaly, 41 % for spina bifida, and 17 % for encephalocele. According to the RENAC, the prevalence of NTDs is 1 per 1000 births, which supports effective fortification (RENAC 2014).

Anti-rubella vaccination

Rubella is an asymptomatic or mild acute viral infection. Concerns arise since rubella infection during pregnancy can alter fetal development and lead to congenital rubella syndrome (CRS) (Gregg 1991), which includes serious structural or functional birth defects. Newborns affected by CRS secrete the virus for 1 year, thus being a potential source of spread. The NMoH National Program for the Control of Vaccine-Preventable Diseases (ProNaCEI) aims to reduce morbidity and mortality from vaccine-preventable diseases through vaccination campaigns. This program has implemented several public health measures to prevent CRS (Table 3). Similar steps have been taken in other countries from the region of the Americas with the ultimate goal of eradicating the disease.

Table 3 Prevention of congenital rubella syndrome (CRS) in Argentina

Year	Milestone
1998	MMR vaccine was included in the immunization schedule
2003	Vaccination with measles-rubella vaccine is mandatory in the immediate postpartum or postabortion period. Implementation of integrated surveillance of measles, rubella, and CRS.
2006	Vaccination campaign to women aged 15 to 39 years
2008	Vaccination campaign to males aged 16 to 39 years
2009	Follow-up campaign intended for children aged 1 to 4 years

The definition of rubella eradication from the region of the Americas is the interruption of endemic transmission of rubella virus in all countries of the region for a period not less than 12 months without the occurrence of CRS cases associated with endemic transmission, in the presence of high-quality monitoring (Garcia-Jimenez 2011). There were no confirmed cases of rubella between 2010 and 2013 in the region of the Americas. However, in 2014, a confirmed case was detected in adult male in Argentina (ProNaCEI 2014).

One of the key indicators of good quality surveillance is a notification rate of at least 1 suspected case of CRS over 10,000 live births (PAHO 2009). To achieve this goal, the systematic evaluation of suspected cases is necessary. In Argentina, the CRS is a notifiable disease and must be reported to the National System of Health Surveillance (NMoH) by physicians and laboratories. The notification rate in 2013 was 1.63 per 10,000, reaching the target of 1 in 10,000 (Epidemiological bulletin, NMoH, 2013). However, no notification was received from three provinces in that year. Since the beginning of CRS surveillance in 2003, 2013 was the second year when the notification rate target was reached.

A joint initiative was designed between the ProNaCEI and the RENAC to maintain the reporting rate of CRS in Argentina. The RENAC has trained participating neonatologists in CRS notification. If a case fits the CRS phenotype, the RENAC coordination asks the neonatologist to report to the National Health Surveillance System.

Secondary and tertiary prevention of birth defects

Plan Nacer-SUMAR

Health benefits provided by the public setting to the provinces are not homogeneous but depend on each jurisdiction. Therefore, in 2005, the “Plan Nacer” started as a public health insurance program, intended for the population not covered by social security. This health insurance first covered children less than 6 years of age, and pregnant or postpartum women (up to 45 days after delivery). Since the year 2013, in addition to the coverage provided to mothers and their children, the program started covering children and adolescents aged 6 to 19 years and women up to 64 years of age who had no social security. Thus, this program became into “Plan SUMAR,” which now covers about 8 million people. To those provinces accomplishing specific health goals, this insurance program transfers national funds, which in turn are transferred to health centers, depending on the services they provide to the population. These health centers decide fund allocation: staff incentives and hiring, purchase of input, investment, or maintenance (Plan SUMAR Plan 2013). Thus, the hospitals and

primary care centers in the provinces receive funds from the NMoH.

Regarding birth defects, the Plan SUMAR covers screening for hearing loss in newborns, detection, and treatment of congenital heart defects, and surgery for esophageal atresia, gastroschisis, intestinal atresia, anorectal malformation, myelomeningocele, and hydrocephalus (Plan SUMAR nomenclature 2013). Regarding prenatal care, it covers prenatal visits, screening for toxoplasmosis, and referral to a geneticist, if indicated.

Congenital heart defects

As expected, congenital heart defects are the most common birth defects in Argentina (RENAC 2014) and the main cause of death from birth defects (Bronberg et al. 2009). The estimated years of life lost from premature death attributable to congenital heart defects in children aged 0 to 4 years throughout the country were 29,819 in 2005; DALYs were 39,190 (Borruelet al. 2010). In 2003, congenital heart defect surgeries were only performed in six hospitals, five of which were in the central region of the country. The National Program for Congenital Heart Disease (NMoH) was launched in 2008 to address this potential cause of death with low morbidity after treatment. The Program goals are to increase the access to surgical interventions by improving technological capability of the main cardiologic centers, to reduce the waiting list for surgery, to improve prenatal and postnatal diagnosis, to give medical support to newborns in neonatal intensive care units before being referred to main cardiologic centers, and to organize services through a network with a national coordination. Since April 2010, the comprehensive treatment of congenital heart defects was included in the health coverage provided by Plan Nacer-SUMAR. By 2013, the number of centers included in the National Program for congenital heart disease rose to 17, which were located in ten different jurisdictions. This network allowed patients to be treated locally, thus avoiding the psychological impact of uprooting and potential risks associated with long travels. Between 2010 and 2015, the Program is expected to fund more than USD 170 million for the care (transport, diagnosis, and surgery) of children with congenital heart disease who have no access to social security. Between 2010 and 2013, some 6000 surgeries for congenital heart defects were performed in the context of the Program, and the waiting list for surgeries could be eliminated (Press Department, NMoH, 2014).

Myelomeningocele

Spina bifida in Argentina has a birth prevalence of 5.5 per 10,000 births (Table 1). In 2012, the Myelomeningocele National Group was started by the NMoH (Consulting Group on NTDs 2013). The objectives of this group are to promote a network

for surgical treatment and to overcome the difficulties in the multidisciplinary follow-up of affected children. The group has organized a national meeting of professionals (pediatricians, neurosurgeons, urologists, orthopedists, physiatrists, nurses, etc.) involved in the care of affected children and has drawn up guidelines on “Recommendations for neonatologists on the management of myelomeningocele.” Members of the RENAC coordination have participated in the design of these guidelines, which were distributed among RENAC neonatologists through the RENAC website. At the same time, members of the Myelomeningocele National Group have trained neonatologists in the RENAC annual meeting, and the RENAC also provides the National Myelomeningocele Group with estimates of the expected annual number of cases of myelomeningocele by jurisdiction, which proves useful for planning of services and regionalization of care.

Newborn screening

In Argentina, neonatal screening is available for several congenital metabolic diseases and hearing loss. Table 4 shows these advances in Argentina. Screening includes six metabolic diseases: phenylketonuria, congenital hypothyroidism, cystic fibrosis, congenital adrenal hyperplasia, biotinidase deficiency, and galactosemia (National Law 26279 2007). The NMoH has estimated 95 % coverage in the public setting. However, there are no data on follow-up and long-term management of affected children. The NMoH publishes periodically a procedure manual for endocrine and metabolic screening, which establishes guidelines for laboratory screening, diagnostic confirmation, and treatment. Neonatal screening is organized in a network of local and regional processing laboratories for initial screening, and for those tested positive, there is a referral network for diagnostic confirmation and treatment in a few high complexity hospitals located in large cities (NMoH 2011).

In Argentina, hearing impairment represents 18 % of disabilities and is classified into impaired hearing (86.6 %) and

deafness (13.4 %). The NMoH National Program for Early Detection and Treatment of Hearing Loss was started in 2010. Its aim was to carry out universal neonatal screening for an early diagnosis (between 3 and 6 months of life), thus allowing an early treatment before the first year of life, speech therapy, and follow-up. Neonatal screening through the otoacoustic emissions test is performed before the first month of life. Children who fail to pass the screening test twice are evaluated by an auditory evoked potential of stem, or steady state, and referred to an otolaryngologist, who will do the follow-up. In 2011–2012, the Program provided headphones to 310 affected children and cochlear implants to 31 (National Program for Early Detection and Treatment of Hearing Loss 2012).

Disability

According to the First National Survey of People with Disabilities (INDEC 2003), a total of 2,176,123 persons live with disabilities in Argentina, and 12.8 % (278,482) are born with disabilities. The census taken in 2010 included a question about the presence of difficulty or permanent limitation. Results showed that 5,114,190 people had a visual, auditory, motor, or cognitive difficulty or permanent limitation, which represents a population prevalence of 12.7 % (INDEC 2010). There are several laws and programs focused on disability. The “Protection System of the Disabled” Act (National Law 22431 1981) required that the State should employ 4 % of workers with disabilities. Subsequent amendments to the law have forbidden physical and architectural barriers in urban design, since they could interfere with the disabled. The “System of Basic Services for Habilitation and Comprehensive Rehabilitation for People with Disabilities” Act (National Law 24901 1997) requires that medical care to the disabled should be provided by the social security. National Law 25504 (2001), known as the Unique Certificate of Disability Act, is complementary to previous laws. This law requires that the disability certificate, including the extent and nature of the disability and the potential for rehabilitation, should be issued by the NMoH. This unique certificate provides certain benefits to people with disabilities, such as free access to public transport, free parking, tax exemptions, special education programs, access to job offers in state agencies, and pensions for the disabled or their family.

The NMoH “Incluir Salud” Program transfers funds to the provinces. The provinces must provide health benefits to the disabled (Incluir Salud 2014). The National Advisory Committee for the Integration of the Disabled, under the National Council for Coordination of Social Policies, a State agency comprising various ministries (health, labor, education, justice, science and technology, planning, and finance), jointly with the provinces, aims to develop and coordinate

Table 4 Newborn screening for metabolic diseases in Argentina

Year	Milestone
1986	Screening for phenylketonuria (National Law 23413 1986)
1990	Screening for congenital hypothyroidism (National Law 23874 1990)
1994	Screening for cystic fibrosis (National Law 24438 1994)
2006	Design of the National Program for Strengthening the Early Detection of Congenital Diseases (NMoH Law Decree number 1612)
2007	Screening including six diseases: phenylketonuria, congenital hypothyroidism, cystic fibrosis, congenital adrenal hyperplasia, biotinidase deficiency, and galactosemia (National Law 26279 2007)

programs for the integration of people with disabilities and to monitor compliance with disability laws (Law 1101 1985).

Genetic services

The first genetic services in Argentina were started in the late 1960s by pediatricians and obstetricians trained abroad. The National Centre for Medical Genetics (NMoH) was founded in 1969, and then other genetic services began to be started in the main pediatric hospitals. The development of medical genetics in the country lacked a planned strategy. Until the year 2000, some provinces had no genetic services or lacked updated diagnostic technologies. Moreover, there was functional fragmentation due to the multiple jurisdictions (university, municipal, provincial, national) they depended on. Some practices, such as prenatal diagnosis, were only offered in the private setting. The fact that abortion is illegal (unless the life or health of the mother is at risk, or in case of rape) has delayed the development of prenatal diagnosis in the public setting (Penchaszadeh 2013). There are some limitations to measure the frequency of induced abortions that occurred in Argentina. Nevertheless, some authors refer that abortion is a common practice in Argentina (Mario and Pantelides 2009).

In 2004, a census was taken by the National Center of Medical Genetics to establish the available resources in the country. The study identified 50 medical genetics units in universities and public hospitals, with 67 medical geneticists and 133 laboratory professionals. According to this survey, 10 of 24 jurisdictions had no genetic services in the public setting, while the central region of the country had a large number (Liascovich et al. 2006). In an update in 2007, which excluded those research units only involved in molecular diagnosis for one specific disease, 35 units were counted. Of these remaining units, only 19 had a medical geneticist and a cytogenetic laboratory. Of these, five also performed molecular diagnosis for monogenic diseases. The other 16 units had either a medical geneticist, or a cytogenetic laboratory, but not both (Alba et al. 2007).

In 2008, the National Program of Genetics Network was designed through a decree issued by the NMoH (Resolution 1227/2008). In 2009, this Program coordinated a workshop with representatives of genetic services in the public setting from the whole country. In this workshop, a survey of human resources and necessary equipment was performed. This meeting partly concluded that this topic should be included in the local health agenda of all provinces. Thereafter, training was provided to health professionals across the country (see Health professionals training section). Since 2012, each province has appointed a local health professional to work with the central coordination of the National Program of Genetics Network. The central coordination committed to train locally

selected health professionals in medical genetics, has helped strengthen or develop local genetic services, and has contributed to the expansion of the RENAC. Among new initiatives addressing birth defects in Argentina, there is a new area of rare diseases organized after a national law on the topic was enacted (National Law 26.689 2011).

Teratology Information Services

Since the year 2000, the Fetal Health Line has operated in the National Center of Medical Genetics as a teratology information service, with a toll free number incorporated in 2011. Medical geneticists answer telephone or e-mail enquiries from both the general population and medical professionals about the risks of prenatal exposure to potentially harmful agents (Salud Fetal 2014). To date, this service has answered around 3000 inquiries (Barbero, personal communication) and conducted various research projects (Valdez et al. 2007; Barbero et al. 2008; Barbero et al. 2011; Martín et al. 2014). The Fetal Health Line has promoted training of medical and pharmaceutical professionals about teratogenic agents; it has advised the NMoH on the implementation of standards for controlling the use of teratogenic drugs such as retinoic acid and counseled pregnant women about rubella vaccination (Pardon et al. 2011). The Fetal Health Line is a member of the European Network of Teratology Information Services.

Health professionals training

In 1979, the National Center of Medical Genetics started a medical genetics residency, which was followed by the cytogenetics residency. After that, there were training initiatives in medical genetics: the Annual Course of the National Center of Medical Genetics and other training programs by medical genetics services from different provinces.

A survey showed that health professionals in Argentina had little knowledge of medical genetics (Barbero et al. 2003). In the context of the National Program of Genetics Network, in 2009 and 2010, geneticists from the National Center of Medical Genetics and other genetics services traveled to the Northeast and Northwest regions to train health teams. They conducted 11 training sessions with 925 participants. Among the training activities, the National Program of Genetics Network funded a blended course in the National Center of Medical Genetics for three consecutive years (2011–2013). Moreover, short internships for health professionals from several provinces were carried out at the National Center of Medical Genetics, Hospital J. P. Garrahan and other genetic services, with the objective of training health professionals that later began to work in provincial genetics services.

Regarding training, the Capability project funded by the European Union was carried out at the Hospital Garrahan genetics service between 2007 and 2009 (Barreiro et al. 2013). This project, conducted in the province of Chaco (Northeast region), trained health professionals in medical genetics at primary care and produced online and printed manuals. These manuals are considered reference material in different primary care settings and in health professional training in medical genetics. Garrahan Foundation funded similar programs in three other provinces.

Toolkit for health care needs assessment

In terms of public health-based research on birth defects, geneticists from Argentina, Uruguay, and Brazil participated between 2010 and 2011 in piloting and subsequent implementation of the Public Health and Genomics Foundation (PHGF) toolkit for health care needs assessment of birth defects (Nacul et al. 2014). Assessment of health care needs is a commonly used process for those working in public health (Wright et al. 1998; Williams and Wright 1998) and involves epidemiological, qualitative, and comparative methods to describe the health problems of a population, identify inequalities in health and access to services, and determine priorities for the most effective use of resources (Wright et al. 1998). The PHGF has developed this toolkit to provide users with a stepwise approach, which contributes to the health care needs assessment of birth defects. This proves useful in countries with little experience in needs assessment, service development, and interventions.

In Argentina, the health care needs assessment toolkit was used for neural tube defects (Groisman et al. 2013c) and oral clefts at the national level by the National Center of Medical Genetics group. The Hospital Garrahan genetics service has used the toolkit for health care needs assessment of Down syndrome, congenital heart disease, and genetics services at a provincial level (Bidondo and Barreiro, personal communication). The toolkit has served not only to research policies, programs, and services but also to hold local and regional interest in the topic among the society and to push forward with the inclusion of birth defects in the health agenda.

Conclusions

For an approach to birth defects through a public health perspective, the different stakeholders must be considered. In Argentina, some programs focus on different approaches to the problem, and the challenge is to coordinate the teamwork between these programs. The following are a list of tips to address birth defects from the public health perspective:

- To fight the common prejudice that medical genetics is a discipline for developed countries and is only linked to high technology, research, and genomics. To show that birth defects are public health issues
- To link birth defects to topics already included in the health agenda, such as communicable diseases (i.e., rubella and CRS)
- To consider raising social concerns (i.e., the effect of pesticides or pollutants on the frequency of birth defects, healthcare needs of parents, and patient associations) and the authorities' concern about lowering the infant mortality rate
- To develop a surveillance system to gather information for policy makers and to increase awareness of the problem
- To participate in international projects with expert organizations: the International Clearinghouse for Birth Defects Research and Surveillance, PHG Foundation, etc.
- To use free and open source information and communication technologies (i.e., Moodle for online courses) to overcome geographical barriers
- To develop training programs for health professionals from different levels of care, with an emphasis on primary care

Although in the last decade, Argentina made public health progress in addressing birth defects, the following issues are still pending:

- Most provinces cannot still meet the demand of their population regarding genetic diagnosis and counseling.
- The medical genetics network is being developed, but there are gaps in the interaction between services, with duplication and inefficiencies.
- Argentina lacks a national system for genetics lab quality control.
- Special programs have addressed frequent birth defects, such as congenital heart disease and myelomeningocele, but did not consider other common birth defects, such as orofacial clefts and Down syndrome.

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