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A targeted, regulated expansion of genetic services as means to increase health capabilities in Latin America

Due to the recent health transition from infectious diseases to non-communicable diseases, genetic disorders are now a global public health priority. Genetic disorders have a high prevalence and a high burden of disease in developing nations. Services that address these disorders are known to increase health capabilities of individuals. Public health measures that seek to increase access to genetic services can distribute these benefits to underserved populations. This paper focuses on Latin America, as it is a suitable candidate for expansion due to the already established health infrastructure. Through examining previous initiatives in genetics in Argentina, Brazil, Chile and Cuba, this paper argues that a targeted, regulated expansion of genetic services, made easily accessible to low income individuals, will increase health capabilities and overcome the inverse equity law. Tags: genetics, health inequities, access to healthcare, health capabilities, targeted

> An account of two students rotating in the genetics department in Hospital Padre Hurtado of Chile: *We set up in the pediatric outpatient unit and went to see the newborns. Our first visit was with a new baby girl whose was born (39 weeks 2 days) with unfused cranial sutures, hepatomegaly and a tiny little thorax with tiny, tiny ribs. Connie and I examined her as best we could, but couldn't come up with any sort of diagnosis. Dra. Gabriella (who trained at MGH/Children's Boston) came a half hour after us, and we looked at the baby's skeletal survey (on the one computer that can show images) and headed down to radiology to consult with them (nicer computers there). They are running a metabolic panel on her, though I haven't heard of results. Later in the day we saw outpatients, a great spectrum of genetic syndromes, including a case of Neurofibromatosis 1 (in a 1 year old), a boy who was 48,X,X,Y,Y, 2 Down Syndrome's and an adorable girl*

> *with achondroplasia. We saw one 15-year-old boy towards the end of the day for indications of "developmental delay." The delay and odd behaviors that his mother spoke of were clear, and he had a recent history of epilepsy. The karyotype that was recently ordered was "normal" 46,XY but there was likely a genetic cause underlying this, and after the appointment Dra. Gabriella turned to me and said, "If this were the United States he would be a great whole exome candidate." Indeed that is probably true, but in the meanwhile a metabolic panel was ordered to rule out a number of potential issues, many of which are screened for at birth in the US. Fragile-X [testing] is the next step when/if that comes back normal.* (https://asecondwinterinchile.wordpress.com/).

Health capabilities are a basic human right that grant the opportunity to live a happy, healthy life. Without such capabilities, humans would not be able to live comfortably, engage with communities, or achieve their personal goals. Up until recently, efforts to improve health capabilities in developing countries have been focused on controlling the spread of infectious diseases and malnutrition. Further attention needs to be paid to non-communicable diseases. One specific category is genetic diseases, which are highly prevalent in developing countries and contribute significantly to poor health outcomes. Expanding genetic services to reach individuals in poverty can increase health capabilities and decrease health inequities.

The goal of this paper is to assess the costs and benefits of expanding genetic services in Latin America. The development of services in Argentina, Brazil, Chile and Cuba are examined in order to learn from their successes and failures. This assessment will provide public health decision makers with a better understanding of whether and how resources should be devoted in providing this service to improve health outcomes in an equitable manner. Priority in policy making is often awarded to other public health provisions, such as infectious diseases or nutrition, as these are often deemed easier to tackle. Genetic disorders have now transitioned to a worldwide global health problem. Due to this recent transition, a gap exists between services needed and the qualified

health care professionals to deliver these services. Additionally, services are typically only accessible to wealthy individuals. In order to overcome this gap, more services aimed at low-income individuals are needed in developing countries. While the individual health benefits resulting from genetic services include better disease management, psychosocial support, and aiding in family planning, these health gains often fail to reach the population at large. When developing policies, many barriers must be overcome in order to distribute these benefits from an individual level to a population level. Ensuring equity and effectiveness in outcomes involves developing infrastructure, increasing professional capability, and the integration of services into health care systems. However, all of these initiatives could be costly. Government policies and regulation can work to lesson potential inequities associated with the expansion of genetic services. These services must be made easily accessible to everyone through a cost-effective targeted expansion to ensure new investments are widely accessible to individuals in poverty and do not fall victim to the inverse equity hypothesis.

Genetics is a field that, due to technological advancements made in the past few decades, has drastically expanded and is currently at the forefront of public health interventions. The epidemiological goal of genetic services is to decrease the burden of genetic diseases on a population (WHO 1999). Clinical genetic services include genetic screening and testing aimed at helping individuals with possible genetic predispositions manage their risks for developing or passing on a genetic disorder. Examples of these widely utilized public health measures include prenatal and neonatal screening performed by nurses. Prenatal screening involves sampling the mother's blood or conducting an ultrasound to detect abnormalities indicative of birth defects. The American College of

Medical Genetics and Genomics (ACMG) recommends an expanded screening panel of 29 disorders in the United States. According to the ACMG, these expanded newborn screening panels are around the same costs as single gene tests (Grody et al. 2013). Newborn screening involves sampling the baby's blood or conducting a hearing test. Blood samples are then sent to basic laboratories for assessment. These screenings are extremely cost effective. An assessment on prenatal screening for Down syndrome in the United States found that all screening options were more cost effective than not screening, with screening options ranging from $35 to $1200 (Odibo 2005). Yet, these are not diagnostic tests, and it is recommended that abnormal screening results be referred to clinical geneticists or genetic counselors. Clinical geneticists can then diagnose and treat the patient. Further diagnostic options include an amniocentesis, an invasive sampling of the amniotic fluid, or chorionic villus sampling, an invasive sampling of placental tissue. These options require highly technical laboratory assessment and are more costly. Genetic counselors can aid in managing the risks through presenting options of pregnancy termination or providing resources on how best to care for an individual with a genetic disease.

Counseling from a health professional ensures that an individual can continue living as healthy and happy as possible. This counseling component of clinical genetics primarily serves as psychosocial support as well as preventative care to aid individuals in managing their health risks and to diminish suffering. With proper management, many individuals with genetic disadvantages can live happy, healthy lives. Unfortunately, genetic counseling is more prevalent among developed nations than among developing nations, which results in an unequal distribution of services and outcomes. Although

there is no formal data on cost effectiveness, Schüler-Faccini of the Genetics Department of Universidad de Federal do Rio Grande do Sul in Porto Alegre, Brazil, stated that the expansion of services "can represent less of an economic burden than doing nothing, since the burden of congenital anomalies is much higher when not properly treated and prevented" (WHO 2012). Due to the preventative power of these services and potential cost effectiveness, the expansion of genetic services could increase global health outcomes.

Genetic counseling has substantial health benefits to patients in developed countries. A study conducted on the impact of genetic counseling on the prenatal diagnosis of either Down Syndrome or neural tube defects found that "for more than half of the families the availability of prenatal diagnosis was of crucial importance in the decision to plan future pregnancies" (Swerts 1986). These results demonstrate that patients who underwent counseling remembered the relevant risks presented by the genetic counselor and made informed decisions based off of them. Genetic counseling does not just impact the decision-making of patients; it can also increase health outcomes of patients. Dinh et al. (2011) conducted a study on the health benefits of Lynch syndrome (a genetic disorder that increases risks of many associated cancers) and found that careful management of individuals with genetic risks reduced risks of associated cancers by 8.8%-12.4%. The authors concluded, "for a population of 100,000 individuals containing 392 mutation carriers, this strategy increased quality-adjusted life-years (QALY) by approximately 135 with an average cost-effectiveness ratio of $26,000 per QALY." (Dinh et al. 2011). These increases in health outcomes underscore the need to expand genetic services in developing countries. Since current benefits of genetic

services are not globally shared, an analysis of how best to distribute benefits is needed. Services should be made more accessible so everyone has the same opportunity to make informed decisions on their health.

The need for services

There is currently a higher prevalence of genetic diseases in developing countries than developed countries, resulting in significant burdens to communities in developing countries. Globally, more than 7.6 million children are born annually with a severe genetic disorder or birth defect, medically termed congenital anomaly, with 94% of total cases occurring in low- and middle-income countries (WHO 2002). This places high burdens on families under socioeconomic distress who cannot afford the expensive management costs of such disorders without proper access to genetic services (Marques-de-Faria, Ferraz, Acosta, & Brunoni 2004). The higher prevalence of congenital anomalies among the disadvantaged is due to a lack of resources, a lack of a nutritious diet, and environmental factors that increase the risk of negatively affecting fetal development (WHO 2015). What is more, it has been found that low socioeconomic status and lower levels of education are associated with higher emotional burdens from genetic diseases (Penchaszadeh 2013). This emotional burden translates to a financial burden as well. Congenital anomalies and genetic diseases make up "10-25% of pediatric admissions in some urban centers, imposing their burden of more extensive and costly hospital stays" (WHO 1999). Unfortunately, public health measures to address this high burden fall short compared to what is available in the United States. Furthermore, genetic disorders are often under diagnosed in developing countries. This results in lost opportunities for preventative actions, which in turn increases health care costs. The high burden of disease and long-term costs from the disease results in fewer health capabilities

for individuals with genetic disorders, with a concentration of these individuals in lower socioeconomic classes. Therefore, improved public health interventions to address the high prevalence of birth defects among low-income individuals are needed.

As global health outcomes are improving, genetic disorders have risen in prevalence and are now a worldwide epidemic. Developing countries have faced vast epidemiological changes over the past few decades. For example, in Latin America, infant mortality rates declined from 128 deaths per 1,000 to 28 deaths per 1,000 between 1950–1955 to 2000–2005 (WHO 2007). This decrease in infant mortality rates has lead to a "health transition" that constitutes a health risk shift from infectious diseases and malnutrition to birth defects (Christianson, Howson, & Modell 2006). The high prevalence of birth defects and genetic diseases in the world represents a public health priority. For example, the average amount of birth defects per 1,000 live births in Brazil is 57.2, in Argentina is 52.5, in Chile is 52.7, and in Cuba is 47.2 (Christianson, Howson, & Modell 2006). Furthermore, in the past century, there has been an increase in mortality from genetic disorders in Latin America (Penchaszadeh 2004). They are now a major cause of infant mortalities; it is estimated that percentages in some Latin American countries are as high as 25% of total causes of infant morality (WHO 1999). This global health transition illustrates the need to prioritize genetics in public health decisions.

More resources should be devoted to address the growing burden of genetic diseases on populations. However, a scarcity problem in health care exists, and unfortunately there are limited resources to devote to the expansion of health. Initiatives in genetics could be a good allocation of these limited resources due to their high potential to increase health capabilities. The disability adjusted life year, DALY, value

for congenital anomalies in 2010 was 564 per 100,000 individuals. One DALY equates to one lost year of life. Decreasing health burdens through genetic services could save these lost life years. It has been predicted, "70 percent of birth defects could be prevented, ameliorated or treated globally by strengthening primary care services" (Christianson, Howson, & Modell 2006). This strengthening would entail integrating more genetic services into primary care services. The high mortality and morbidity rate from birth defects underscores the need for a target allocation of resources. Global health initiatives have recently begun to prioritize the prevention of birth defects in order to achieve the UN Sustainable Development Goal 3.2, which is "to reduce neonatal mortality to at least as low as 12 per 1,000 live births and under-5 mortality to at least as low as 25 per 1,000 live births" (WHO 2015). The World Health Organization has also declared congenital disorders (birth defects) as a "major health problem worldwide" in a recent report and stated the need for the expansion of genetic services in developing countries (WHO 2011). The WHO maintains that the "societal costs of inaction in genetics, measured in terms of avoidable human suffering and burden to public health, are very high" (WHO 1999). Therefore, in order to evade these high costs, nations should devote more resources towards the prevention and management of genetic diseases.

Genetic Services in Latin America

The development of genetic services in Latin America has expanded over time but still faces many barriers in delivering benefits to the entire population. This region is examined because it is a suitable candidate for expansion due to the already established health infrastructure that can benefit from increasing genetic initiatives. The public sector currently provides the largest amount of genetic services in Latin America (Penchaszadeh

2015). However, "under-financing, poor planning and coordination and deficient regulations and quality assurance have hindered their quality and their responsiveness to population needs." (Penchaszadeh 2015). The services available in the private sector are more developed, "particularly for applications that are commercially profitable, e.g. chromosome analysis, prenatal diagnosis, and DNA paternity testing" (WHO 2003). Foundationally, these base line services already in place are a promising point of expansion. However, the existence of the services does not provide information on the quality or accessibility of services. Four countries, Brazil, Argentina, Chile and Cuba, are further assessed to provide insight on current availability and shortcomings of genetic services. The first three countries, Brazil, Argentina, and Chile all have some established genetic services that unfortunately fall short in reaching a broader population. Cuba serves as a model country due to its primary care based initiatives and integration of genetics into all levels of health care.

As a country classified as high in human development, Brazil makes a suitable candidate for expanded genetic services. Brazil has a Gini index, a metric to measure income inequality prevalent in an economy, of 52.9 (World Bank 2013). This value indicates high inequality in the country. On the Gini index, a value of 0 points towards perfect equality, whereas a value of 100 points towards full inequality. For a point of comparison, Peru has a Gini value of 44.7 and the United States has Gini value of 41.1 (World Bank 2013). Due to Brazil's high development but high inequality, the country can benefit from an equitable expansion of genetic services to improve the health outcomes of a broader population. In 2015, the country spent 9.7% of their GDP on health care expenditures. Universal health coverage has been available in the country

since 1988 and is funded by the Unified Health System (Horovitz et al. 2013). It provides free services at the point of service ranging "from outpatient care to organ transplantation" to those who are insured. Private health insurance is available for those who can afford it, with 24% of the population covered under the private sector (as read in Horovitz et al. 2013).

In regards to genetic services, Brazil is one of the few countries in Latin America that integrates genetics into the national health care system (Penchaszadeh 2013). The need for services is underscored by the high congenital anomaly DALY value (including neural tube defects, cleft lip and cleft palate, Down syndrome, congenital heart anomalies, other chromosomal anomalies, and other congenital anomalies) of 1,117.9 in 2012. These lost life years contributed to a total of 1.82% of total DALYs in 2012 (WHO Global Health Estimates, DALY estimates 2012). This high prevalence translates to high rates of infant mortality as well. In 2006, congenital anomalies accounted for 16.3% of infant mortalities and are now the second cause of infant mortalities (Horovitz et al. 2013). Neonatal screening in Brazil has been available for around twenty years, "comprising tests for phenylketonuria (PKU), congenital hypothyroidism (CH)[1], sickle cell disease, and, in some parts of the country, cystic fibrosis" (WHO Bulletin, 2012). However, services often fail to reach the population at large due to the small number of genetic health care professionals and a lack of regulation. Horovitz et al. (2013) note, "only around 25–30 % of the estimated need in genetics is being cared by specialists in the field". Ideally, a country should have one geneticist per 100,000 individuals (as read in Taucher 2015). However, Brazil has less than 300 clinical geneticists, approximately

[1] PKU is a neurodegenerative disease and CH causes thyroid dysfunction

less than 0.15 clinical geneticists per 100,000 individuals (Passos-Bueno et al. 2014). Therefore, more professionals are needed to meet the need of care. Although Brazil has made recent advancements in expanding genetic services, more continued initiatives must be executed in order to reach the entire population.

Argentina is classified as very high in human development, with a Gini inequality index of 42.3 (World Bank 2013). Like Brazil, the country could benefit from an increase in genetic initiatives in order to diminish health inequities prevalent in the country. Argentina has a high level of established health infrastructure. In 2013, the country had 5,012 health establishments, including hospitals, clinics, and hospice units, with 70% of establishments controlled by the private sector (Wikipedia). In regard to health care, the public insurance covers around 48% of the population, social security funded through employers covers 47% of the population, and the private sector covers 7.5% of the population (Penchaszadeh 2013).

There is a high burden of disease due to genetic disorders in the country; the congenital anomalies DALY value in Argentina was 326.4 in 2012. These lost life years contributed to 2.7% of total DALYs in the country. In order to address this burden, the first medical genetics initiative was developed in the 1960s (WHO 1998). Public health measures have integrated these services into the national health care system; in 2007 there were 41 genetic wards in public hospitals (Penchaszadeh 2013). However, Argentina only has 120 clinical geneticists, approximately 0.29 geneticists per 100,000 individuals (Penchaszadeh 2013). This number falls short of the recommended number of 1 geneticist per 100,000. The most common counseling services currently available include "single-gene disorders and congenital disorders in children and newborns"

(Penchaszadeh 2013). There are fewer services available for adult genetic services. A few special care facilities exist, but access is limited especially to low-income individuals (Penchaszadeh 2013). What is more, misdiagnosis of genetic disorders is common due to a lack of knowledge among health care professionals. Services are also underused due to a lack of regulation in the country. Current initiatives to integrate genetic services into primary care by way of increasing health professional's knowledge of genetics in Argentina has successfully begun to remedy these problems (Barreiro et al. 2013). This program will be further assessed in the recommendations section of this paper. Other recent developments include an expansion of birth defect screening in the past five years, and currently "annual coverage is approximately 280,000 births, which accounts for 65% from the public setting and 38% of all births in the country" (Bidondo, Groisman, Barbero, & Liascovich 2015). Screening currently covers six genetic metabolic disorders. The promising recent genetic advancements in Argentina make the country an appropriate candidate for further expansion in order to increase health capabilities of a wider population.

Like Brazil and Argentina, Chile classifies as a nation well positioned for further developing health capabilities. Chile is classified as high in human development, with a Gini inequality index of 50.5 (World Bank 2013). The country has high inequality and established health infrastructure that makes it feasible to increase genetic services. In 2015, the country spent 7.7% of their GDP on health care expenditures. The health care system in Chile is a two-tiered system with public health insurance covering around 69% of the population and private health insurance covering around 17% of the population (Vargas & Poblete 2008). Due to this divide, most low-income individuals receive their

health care through the public sector whereas high-income individuals receive their health care through the private sector (WHO 2010). Unfortunately, this two-tiered system places low-income individuals at a disadvantage since public insurance provides fewer services due to limited governmental resources. Therefore, those who can afford private health insurance receive better health care.

Genetic disorders have a high burden of disease in Chile. In 2012, the congenital anomalies DALY value in Chile was 97.6, contributing to 2.41% of total DALY values. A Health Reform Commission in the early 2000's determined 56 health priorities to address in reform based on classifications of high burden of disease and cost effective treatments. All health conditions examined had guaranteed treatment and coverage options in the country. Congenital anomalies met the following conditions: health objectives, burden of disease, socio-economic inequality, social preference, rule of rescue (urgency of saving an individuals life), high cost, and cost-effectiveness (Vargas & Poblete 2008).

Health insurance reform addressed these issues in 2005 and led to a guaranteed coverage of congenital anomalies due to their assessment as a burden of disease (Taucher 2015). Newborn screening for phenylketonuria (PKU) and congenital hypothyroidism (CH) has been available since 1990, with high coverage rates of 98.7% of newborns screened from 1992 to 2008 (Taucher 2015). Expanding screening options could increase the rate of early detection, leading to a decrease in infant mortality. Furthermore, a shortage exists among health care professionals specializing in genetics to interpret the screening results. Only one clinical geneticist exists per 500,000 people and Chile holds only 15 clinical cytogenetic laboratories (Taucher 2015). Services range from "prenatal

[risk] assessments, diagnosis of [birth defects], genetic counseling, follow-up of patients with genetic and malformation syndromes, and risk assessment in familial cancer" (Taucher 2004). An expansion of the services that already exist in Chile has great potential to increase health capabilities throughout the population.

Cuba's genetic services serve as a model for many other nations, as services in Cuba have been widely implemented into all areas of the health care system. Cuba is classified as high in human development, with a Gini index range of 31.2-33.7. Universal health care is offered and provides free care at the time of service to all citizens. Health care reform over the last thirty years has focused on community-based primary care preventative measures. This strategy of focusing on preventative care has been extremely cost effective for the country (World Bank). The country invests less money financially on health care than the United States. In 2015, Cuba spent 8.8% of their GDP on health care expenditures, whereas the United States spent 17.1% of their GDP on health.

With a focus on preventative measures, clinical genetic services were established in the 1980's. Today, services expand throughout the primary, secondary and tertiary health care levels (Cruz 2007). National prenatal and newborn screening guidelines were implemented after pilot studies demonstrated their success (Cruz 2007). Prenatal screening includes neural tube defect screening, sickle cell screening, and chromosome disorders, while neonatal screening includes screening for PKU, CH, galactosemia, biotinidase deficiency, and congenital adrenal hyperplasia[2] (Cruz 2007). Coverage has successfully reached majority of the population, with an expansion in neonatal screening

[2] Galactosemia is a disorder of galactose metabolism, biotinidase deficiency is a neuro-metabolic disorder, and congenital adrenal hyperplasia is a disorder that limits adrenal function

rates from 84% in 1990 to 97% in 2007 (Teruel 2009). A total of 837 (approximately 1 per 15,000 individuals) genetic counselors are available to assist in the screening programs. Furthermore, community based initiatives have successfully expanded coverage to rural areas. Provincial hospitals provide services including screening and educational outreach programs (Cruz 2007). Due to these successes, Cuba has the lowest congenital anomaly DALY value of 30.6, contributing to 0.87% of total DALY values in 2012. A further analysis of the country's success in genetic initiatives will be addressed in the recommendations section of this paper.

Although Cuba has successfully been able to equitably expand genetic services, it is important to note the differences between Cuba and the other three countries. The socialist country, ruled by one government party, prioritizes health care, thus a strong political will exists to advance programs in genetics. The highly developed health care system of the country yields health outcomes that are more substantial than the United States. The average life expectancy in 2011 was 79 years, which was slightly higher than the life expectancy in the United States (World Bank). However, Cuba does not have a fast growing economy; with estimates of an almost 0% growth rate (Dominguez 2015). Although no formal poverty rates exist for the socialist country, the Borgen Project claims "poverty in Cuba is severe in terms of access to physical commodities, especially in rural areas" (Mohammed 2014). Researchers term the discrepancy between high poverty rates and remarkable health outcomes as the Cuban Health Paradox. The country has achieved high health indices by way of educating a large amount of doctors and distributing health care centers as evenly as possible throughout the nation. In 2008, the country had 5.91 physicians per 1,000 individuals (Evans 2008). Therefore, the health

outcomes of a nation are not dependent on the wealth of a nation. In the case of Cuba, political will and the large amount of health care professionals contributed more to high health measures.

	Brazil	Argentina	Chile	Cuba
Population (in millions)	202	41.8	17.8	11.3
GINI value	52.9	42.3	50.5	31.2-33.7
Multidimensional Poverty Rate	2.9%	3.7%		
Health care system	public and private	public and private	public and private	all public
Congential DALY value	611.4	175.1	51.5	-
Birth defects per 1,000 live births	57.2	52.5	52.7	47.2
Clinical Geneticist's per 100,000	<0.15	0.29	0.20	1
Number of Genetic Counselors	0	0	1	837

Table 1. A comparison of the countries on different indices demonstrates the success of programs in Cuba.

Divide in Outcomes

One major challenge of developing effective and equitable health care measures in the developing world is overcoming the inverse care law. This law affirms "the availability of good medical care tends to vary inversely with the need to it in the

population served" (Hart 1971). Higher socioeconomic classes receive more and higher quality services than lower socioeconomic classes. Examples of such factors that prevent the poor from obtaining good health outcomes include expenses, an unawareness of health care programs, distance, and low health literacy. The poor often live in remote rural areas, far from health care providers, and do not have adequate funds to pay for health care (Gwatkin 2001). Many social determinants of health, such as expenses, an awareness of health care programs, distance, and knowledge of health, act as mechanisms, which create and exacerbate inequities in health.

Ethically, this inverse care law can be seen as violating John Rawls' well-known difference principle in that it does not provide fair equal opportunities. According to Norman Daniels (1981), health care is a social good that provides opportunities for more normal functionings and capabilities in life. These normal functioning's include participating in and contributing to society. Disease can inhibit an individual from participating in economic, social and political components of society. Using Rawls' difference principle, Daniels argues that health initiatives can only be just if they benefit those with the most health disadvantages. Current health initiatives that fall victim to the inverse care law are an ethical wrong in that they do not improve health capabilities of the least well off. Health resources must be distributed towards individuals most at risk to poor health through providing effective health care services (Daniels 2001). It is an ethical imperative to provide substantial health care to the disadvantaged in order to protect their opportunities and maintain their contributing position in society. Since health services disproportionately benefit the wealthy, they fail to meet the difference principle. Therefore, an expansion of health services is necessary to ensure that the

difference principle is met and all socioeconomic classes are given the opportunity of health capabilities. Lessening the health disparities between high and low socioeconomic classes is of grave ethical concern and needs to be at the foundation of every new health care policy.

As a consequence of the inverse care law, the inverse equity hypothesis explains the effect that new health measures have on health inequities in developing countries. Due to this hypothesis, simply expanding genetic services will not necessarily benefit the poor. In less developed countries where there is room for health improvement among all classes, the inequity gap increases with investments in health care (Victora et al., 2000). Current literature shows that when health care is made more accessible in developing countries, those who need it least are using it more. The World Health Organization has noted that increases in health coverage lead to an exclusion of the poor (WHO 2005). Specifically, universal coverage reforms that at their core seek to provide health care to the poor also fall victim to the inverse care law (Victora et al., 2000). An analysis of fifty countries showed that coverage of maternal and child health services, programs that were aimed at the poor, were concentrated in the highest socioeconomic classes (Gwatkin, Bhuiya, & Victora 2004). The wealthiest groups then reap the benefits before programs become fully accessible to the poor, which can widen inequity gaps and increase social injustice. Considering that those most in need utilize new interventions the least, the poor miss out on a vital means to increase overall capability. Due to this unequal distribution, the least well off are prevented from obtaining the basic human right of living a healthy life. Therefore, more determined health initiatives must be implemented in order to provide the poor with good quality health care and lessen disparities in health.

Understanding this hypothesis and its relation to social injustice can help determine if new policy initiatives will work to improve overall health status.

Applying the inverse equity hypothesis to genetic services, services are likely to be utilized more by the wealthiest. One way to overcome this is to increase the number of services and genetic health care professionals. Unfortunately, there is a shortage of clinical geneticist in Latin America; overall, there is approximately one clinical geneticist per million people (Penchaszadeh 2004). Services fail to reach the broader population as well. Regarding newborn screening, a new Argentinean law in 2007 expanded universal health care coverage to cover screening for galactosemia, congenital adrenal hyperplasia, biotinidase deficiency, retinopathy of prematurity, congenital deafness, Chagas disease and syphilis (Penchaszadeh 2013). Even though services have been expanded in Argentina, only 60% of the population is covered. Furthermore, there are no referral guides in Argentina to direct patients from primary and secondary facilities to specialized genetic services (Penchaszadeh 2013). This results in a decrease in utilization of services due to a lack of awareness, thus furthering the public versus private divide in outcomes. Therefore, reform is needed in order to ensure that genetic services will benefit the lower socioeconomic classes.

As long as Latin America has divides in health outcomes, expansion will have a negligible impact on population health outcomes. While there may be limited benefits among the wealthy, favorable outcomes will not reach the population as a whole. If only a few are benefited by the expansion of services, then distributive fairness will not be met. With such high inequality levels already present in Latin America, failing to ensure accessibility to the poor could further increase disparities. Therefore, the potential

inequities associated with expansion of genetic services are an ethical issue that must be addressed. Government policies and regulation must guarantee distributive fairness when expanding genetic services. Ensuring that low-income individuals receive the same benefit from new initiatives is of utmost importance. In order to be just as well as effective, an expansion of genetic services can and must overcome the inverse equity hypothesis by targeting the poor and ensuring government regulation.

Case studies

A model project named CAPABILITY was developed by experts in Argentina, Egypt and South Africa with a goal to "promote an internationally shared set of basic quality standards for genetic services in middle- and low-income countries" (Nippert 2013). Each program was designed to increase the prevention and care of congenital disorders. The capacity building initiatives were country specific in terms of needs, current health care systems, resources available, knowledge of health care professionals, and cultural factors (Nippert 2013). The project established the GenTEE network to evaluate international genetic screening practices (Nippert 2013). The specific project developed in Argentina is discussed and serves as a model of success.

The CAPABILITY project developed in Argentina, the CHACO outreach project, was designed to increase access to services in remote areas and to integrate services into primary care levels. International support from the European Union CAPABILITY project provided funding. The outreach project was performed in the Chaco province of Argentina, a rural area with little access to health care, with active participation from community members. Developmentally, a community based needs assessment was first conducted. Based off of this assessment, a total of 485 non-genetic health care

professionals in rural areas were then educated in medical genetics. The health care professionals were trained to diagnose and counsel individuals with congenital anomalies in primary and secondary health care facilities. To increase cultural competency in genetics, the training incorporated the PRACTICE mnemonic (prevalence, risk, attitude, communication, testing, investigation, consent, empowerment). Consultation rates for both adult and prenatal genetics rose 3.5 times higher after the training program, successfully increasing interventions in the resource-poor province. In order to ensure sustainability, this initiative was followed up through recurrent educational programs and through the development of a second program "from a public hospital sponsored from within Argentina to build a cytogenetic laboratory in the capital of the Province of Chaco" (Barreiro et al. 2013). Weaknesses of the program include the "limited funding of the project that could not anticipate the fast development of the project and the need for new investment." The overall success of the program demonstrates that building capacity of health professionals in rural areas can increase access to services. A continued investment in the CAPABILITY project will further expand genetic services equitably and effectively to a broader population in Argentina, and can serve as a program of success for other Latin American countries to utilize as well.

A further analysis of Cuba's primary care-based development of genetic services can provide insight into the best manner of how to expand successful services. Cuba integrated a National Genetics Program into their National Health Care System which was fully implemented in 1990, thus ensuring equal access to all. Prior to the national program, only four of fourteen provinces provided clinical genetic services. Provincial programs were also implemented "in every province, staffed with two full time clinical

geneticists, one pediatrician and one obstetrician with training in genetics, both working part time, one technician and two nurses" (Heredero 1992). Only four years after sickle cell carrier testing was integrated into the National Health Care System, more than 90% of couples were covered for the test. An analysis of the effectiveness of the program showed that "the genetics program has contributed to the reduction of infant mortality in 1990 to about 0.7-1.2 per 1000" (Heredero 1992). The strong integration of genetic services into the primary level of health care, along with the other levels, has successfully made genetic services easily accessible to the entire population. These same strategies have the potential to be successfully implemented in other Latin American countries, such as Argentina, Brazil and Chile, in order to reach a broader population.

Pilot programs suggest more genetic counseling training can broaden access. Two genetic counseling training programs have been developed starting in 1995 in Cuba. Since 2013, the two programs have produced a total of 837 geneticist counselors, approximately 1 per 15,000 individuals (Cruz 2013). Beneficial outcomes have resulted, including an increase in prevention and treatment of genetic diseases. For example, infant mortality rates decreased from 7.2 to 4.9 since 2000 to 2011 (Cruz 2013). These increased health outcomes seen in Cuba after the development of genetic counseling training programs call for the development of similar programs in other Latin American countries. In order to ensure equity, training programs need to devote resources to introducing more genetic counselors in rural, remote areas. Increasing the amount of genetic counselors in a region will provide the opportunity to increase health outcomes and decrease the burden of diseases on communities.

Recommendations

New initiatives to expand genetic services must target low-income individuals in order to overcome the inverse equity hypothesis and be equitable and cost effective. To provide fair equal opportunities, new programs should originate in rural areas in order to reach low-income individuals. This can be accomplished through the integration of genetic services into provincial hospitals. Such initiatives will serve to circumvent problems of geographic inaccessibility and will increase equal opportunities. Both the CAPABILITY project in Argentina and the provincial focus of Cuba's National Genetics Program serve as examples of how to successfully grant increased access in rural communities. Furthermore, governments must regulate the advances by way of increasing education throughout all levels of health care, with a focus on primary care.

Increasing the capability of Latin American health professionals is necessary in order to ensure equity and increase health capabilities. To accomplish this, medical genetics should be integrated into undergraduate education. Additionally, required genetic courses in medical schools could increase the capabilities of health care professionals to deliver genetic services. Physician resident programs in genetics should also be expanded to yield more clinical geneticists. Increasing the number of geneticists will result in more consultations and will also serve to increase the knowledge of other health care professionals, by way of presentations and other educational materials. Additionally, the introduction of more genetic counseling training programs can enable health care professionals to educate patients on their genetic risks. In the beginning, these programs can be designed towards individuals who already have an established health career, such as physicians or nurses. Eventually, once a certain amount of genetic counselors are present in a country, genetic counseling training programs should follow

the United States model of training. These programs should then be aimed at developing new careers of recent college graduates. This model, once feasible, saves time and resources for all involved, as an economy will not lose already established health care professionals. Improving the capability of health care providers will serve to increase the effectiveness of health services, thereby working to increase health capabilities of a population.

Aside from advancements in specific training, broader expansive education initiatives should be implemented in Latin America in order to increase both understanding and awareness of genetics in health professionals as well as in the general public. Providing more education to the public is an important strategy to ensure equitable increases in health outcomes. Health promotional efforts with the aim of helping individuals understand their genetic risks are needed. For example, the Argentina CAPABILITY project distributed brochures on genetic diseases to the general population (Barrerio 2013). These supplemental measures work to increase awareness of illness and benefits of treatments in communities. Only once individuals are properly educated can they make informed, autonomous decisions and exercise their health capabilities. Genetic counselors can assist with educational outreach programs, as they are trained to educate others on genetic risks. In addition, these programs should first be concentrated in rural, remote areas in order to reach the populations who are most at risk for poor health outcomes. These targeted services will work to overcome the inverse equity hypothesis and provide equal opportunities to the disadvantaged.

The integration of genetic services into all levels of health care, with a focus on primary care, is necessary in order to increase access and success of such programs.

Penchaszadeh (2015) claims that genetic services should be available at primary, secondary and tertiary levels to take "full advantage of the existing resources, maximize efficiency and avoid duplication of services". Beyond the initial goals stated in this paper regarding universal access and distributive justice, genetic programs should initially be primary care based for the purpose of increasing public education and reaching isolated areas. Primary care, in regard to care in genetics, is defined as "detection of genetic risks in the community by due attention and recording of family history in all patient encounters with the health system" along with cost-effective services such as prenatal screening and genetic counseling (Penchaszadeh 2015). Since primary care based programs are focused on broad reaching policies, these initiatives will benefit the most people, as demonstrated in Cuba. Secondary care is defined as treating more specific, complicated genetic diseases as referred from primary institutions. Tertiary care is defined as a medical genetic center that treats very complex diseases and also has the goal of "develop[ing] research and implement[ing] training programs in the field" (Penchaszadeh 2015). Although both secondary and tertiary care units are important, resources must first be directed at developing primary care based initiatives in order to reach remote areas. Regulating services to be implemented in provincial hospitals will equitably expand health capabilities.

In order to circumvent the issue of limited resources in these developing countries, public-private partnerships could work to build infrastructure in the development of genetic services. Meier, Schöffski, and Schmidtke (2012) claim that the lack of resources, not political will, is the main determinant for developing countries failing to fully implement genetic services. Therefore, initiatives to properly fund

services, such as public-private partnerships, can reap huge benefits. These partnerships serve to bring the government, for-profit organizations, and non-profit organizations (NPOs) together to fund health care initiatives to improve health outcomes. Public-private partnerships are commonly used in other areas of health care in middle- and low-income countries such as programs to decrease HIV/AIDS, malaria and tuberculosis. Known benefits include "mobiliz[ing] private finance, increas[ing] access, introduc[ing] efficiencies in the delivery of public health services and improv[ing] outcomes" (Meier, Schöffski, & Schmidtke 2012). However, since this paper explores a targeted approach to the expansion of genetic initiatives, funding through for-profit companies who focus on accruing financial gain could lead to disproportionately benefiting the rich. A public-private partnership comprised of not for profit NGOs and government partnerships would yield the most equitable results, as not for profit NGOs seek to distribute resources fairly. An application of public–private partnerships, with a focus on not for profit NGOs, to funding genetic services could solve the problem of limited resources and increase access to those who need it most. Aside from obtaining proper funding, another barrier to overcome when expanding services is ensuring the sustainability of programs.

In order for advances to be equitable they must incorporate a long-term plan of sustainability. Such a long-term plan is evident in the CAPABILITY program in Argentina, which proposes a "replication–amplification" approach towards sustainability (Barreiro et al. 2013). This approach constitutes replication as "the establishment of the model in other provinces" and amplification as "feedback of the model in Chaco both in the training areas and in the remaining zones" (Barreiro et al. 2013). Once a program proves successful, it must be implemented into other remote areas. Numerous districts

need to be targeted as to ensure more expansive outcomes. Policy makers must also ensure that the original initiative is augmented by improvements based on community feedback. Input from community members can provide invaluable information about how the program is actually working. These continued efforts will ensure the success of programs in increasing access to underserved populations.

Although advances in genetics might not be easily adaptable in all countries, evidence demonstrates that certain programs can be extremely beneficial to countries in Latin America. The successful case studies in two of the four countries illustrate promising strategies that could effectively and equitably increase health outcomes. Some may claim that the successes of Cuba's programs may not be applicable to the other countries. Although there are substantial differences in geographical size, political structures, and level of development and infrastructure in the four countries examined, there are relevant similarities between health care systems. All four countries have a form of social health care insurance as well as huge inequality gaps. The social insurance prevalent in all four countries can work to ensure that low-income individuals have access to services and do not pay a disproportionate amount for them. Taking these similarities into account, we can expect that the promising strategies implemented in Cuba would benefit the other countries as well. Furthermore, if funding can be carried out by public-private partnerships, then it will be more feasible to implement services in the same manner as Cuba, notwithstanding government resources. Specific initiatives will have to be tailored to each country, but due to the strong similarities between the countries, such tailoring will be minimal.

Some would argue that initiatives focused on treating communicable diseases are a better allocation of resources, as they might be more cost effective and a higher priority in developing countries. Health risks have recently shifted from infectious diseases and malnutrition issues, towards a new emphasis on genetically related birth defects. Due to this health transition, genetic diseases are responsible for a high burden of disease in developing countries. This ensuing health risk merits the devotion of more resources. If these diseases are detected earlier and health care professionals are properly equipped to manage the diseases, costly long-term treatment can be avoided. Furthermore, with more genetic services and a deeper understanding of how to provide interventions, patient care can improve and decrease the burden of disease. An increase in health capabilities will follow, and individuals can then lead a normal functioning life in order to contribute to society. If all resources are concentrated towards infectious diseases, an opportunity is missed to aid in reducing the high burden of genetic diseases, including extreme suffering and even death. It would be unjust to deny equal access to services that have such a high preventative power.

Due to cultural differences, one might also argue that genetic services are not a worthwhile investment in Latin America. Majority of the population is Catholic, a religion that supports pro-life. Certain countries also classify abortion as illegal, including Chile and Brazil. In those locations, effects may be negligible if prenatal screening will not impact decisions to terminate a pregnancy. While this might be true, there is more to prenatal diagnoses than the option of terminating a pregnancy. Families can have adequate time to prepare for the birth both psychologically and systematically, such as arranging the birth to take place in a properly equipped hospital. Therefore, effects will

not be negligible since services seek to diminish the burden of disease through increasing management of care. Genetic counseling can aid in determining the risks of two parents having a child with a congenital anomaly even before they are pregnant. Newborn screening can also provide early diagnoses so treatment interventions can begin earlier. The public health goals of genetic services are not to terminate pregnancies, but to decrease overall suffering.

In conclusion, the successes of programming in Chile and Argentina demonstrate that the expansion of genetic services can maximize health capabilities of the population at large. A targeted, regulated approach of new genetic initiatives should be implemented in middle to high-income countries in Latin America. In order to execute the difference principle, new expansions should be designed to benefit rural, remote areas. Furthermore, government policies must regulate services so they are integrated into all levels of health care, easily accessible to everyone. Expanding services under these criteria can ensure that low-income individuals will have equal opportunities. Successful expansions can be defined by an increase in genetic consultations in remote areas five times fold, providing 1 clinical geneticist per 100,000 individuals, and providing the expanded screening panel of 29 disorders in a nation.

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