Understanding Antenatal Genetics Services in Sri Lanka:

Current Landscape of Screening and Diagnostic Services and Contextual Factors Influencing Their Availability and Uptake

by

Jenae Elaine Logan

Duke Global Health Institute
Duke University

Date:_____

Approved:

_____
Subhashini Chandrasekharan, Co-Supervisor

_____
Truls Ostbye, Co-Supervisor

_____
Robert Cook-Deegan

Thesis submitted in partial fulfillment of
the requirements for the degree of
Master of Science in the Duke Global Health Institute
in the Graduate School of Duke University

2016

ABSTRACT

Understanding Antenatal Genetics Services in Sri Lanka:

Current Landscape of Screening and Diagnostic Services and Contextual Factors

Influencing Their Availability and Uptake

by

Jenae Elaine Logan

Duke Global Health Institute
Duke University

Date:_____
Approved:

_____
Subhashini Chandrasekharan, Co-Supervisor

_____
Truls Ostbye, Co-Supervisor

_____
Robert Cook-Deegan

An abstract of a thesis submitted in partial
fulfillment of the requirements for the degree
of Master of Science in the Duke Global Health Institute
in the Graduate School of Duke University

2016

Abstract

Background: Too little information is available on Sri Lanka's current capacity to provide community genetic services—antenatal genetic services in particular—to understand whether building that capacity could further improve and reduce disparity in maternal and child health. This qualitative research project seeks to gather information on congenital disorders, routine antenatal care, and the current state of antenatal screening testing services within that routine antenatal to assess the feasibility of and the need for scaling up antenatal genetics services in Sri Lanka. Methods: Nineteen key informant (KI) interviews were conducted with stakeholders in antenatal care and genetic services. Seven focus group discussions were held with a total of 56 Public Health Midwives (PHMs), the health workers responsible for antenatal care at the field level. Transcripts for all interviews and FGDs were analyzed for key themes, and themes were categorized to address the specific aims of the project. Results: Antenatal genetic services play a minor role in antenatal care, with screening and diagnostic procedures available in the private sector and paid for out-of-pocket. KIs and PHMs expect that demand for antenatal genetic services will increase as patients' purchasing power and knowledge grow but note that prohibitive abortion laws limit the ability of patients to act on test results. Genetic services compete for limited financial and human resources in the free public health system, and inadequate information on the prevalence

of congenital disorders limits the ability to understand whether funding for services related to those disorders should be increased. A number of alternatives to scaling up antenatal genetic services within the free health system might be better suited to the Sri Lankan structural and social context. Conclusions: Scaling up antenatal genetic services within the public health system is not feasible in the current financial, legal, and human resource context. Yet current availability and utilization patterns contribute to regional and economic disparities, suggesting that stasis will not bring continued improvements in maternal and child health. More information on the burden of congenital disorders is necessary to fully understand if and how antenatal genetic service availability should be increased in Sri Lanka, but even before that information is gathered, examination of policies for patient referral, termination of pregnancy, and government support for individuals with genetic disease are steps that might bring extend improvements and reduce disparity in maternal and child health.

Contents

List of Tables

Acknowledgements

First, I would like to thank my in-country collaborator Dr. Amila Chandrasiri for his invaluable contributions to my thesis project – from drafting the initial proposal, to scheduling interviews and facilitating focus group discussions, to revising a map of MOH areas in the Galle district. I am extremely grateful to Dr. Chandrasiri not only for making my project happen, but also for making me feel welcome and comfortable in a new country. I would also like to thank the staff and students of the University of Ruhuna Faculty of Medicine: Dr. Vijitha Desilva for his mentorship and kindness; my research assistants Nadeesha Jayasekhara, Nethma Ellawella, and Srimali Fernando for their attention to detail and friendship; and Ruvini Kurukulasooriya for her hospitality and assistance with the logistics of life in Sri Lanka.

Next, I would like to thank my thesis committee chair and advisor, Dr. Subhashini Chandrasekharan for providing the guidance and insight that made this work possible. Thanks also to Dr. Chandrasekharan's family for welcoming me on its pilgrimage through the island nation and leaving me with incredible memories. Thanks to my other committee members Dr. Truls Ostbye and Dr. Robert Cook-Deegan for their mentorship throughout my undergraduate and graduate careers. I must also express my gratitude to Lysa MacKeen for helping me (and my family) to troubleshoot the challenges of planning and implementing a fieldwork project.

1. Introduction

Sri Lanka has charted rapid economic growth and social development over the past decade. The country's gross domestic product (GDP) grew by an average of 6.1% each year from 2000 to 2014 [1], and in 2010 the International Monetary Fund graduated Sri Lanka from its status as a low-income country to a middle-income emerging market [2]. Since then, this small island nation has matched and even outperformed other middle-income countries on key economic and health indicators. Sri Lanka is praised for reducing its infant and under-five mortality rates by two-thirds and its maternal mortality by three-fourths before the deadline set by the Millennium Development Goals [3].

However, the fact that there has been progress does not mean it has been uniform or sufficient. There is high regional variation in infant and maternal mortality rates, and recent data indicate that improvements have decelerated and even stagnated in recent years [3]. The Ministry of Health recognizes this in its National Strategic Plan for Maternal and Newborn Health, stressing that since many deaths are being prevented, the nation must ensure high quality of lives for individuals who now survive [4]. In order for Sri Lanka to continue to improve maternal and child health and target residual disparities, it must characterize the remaining contributors to maternal and child mortality and understand the morbidities faced by children who would not have survived decades ago.

1.1 A Shift in the Burden of Disease

In other low- and middle-income countries (LMICs) charting socioeconomic improvements, when neonatal and maternal mortality decline as infectious disease control, nutrition, maternal and child healthcare access, and family planning availability improve, the relative contribution of congenital disorders[1] to the overall disease burden rises [7, 8]. Indeed, from 1990 to 2010, congenital disorders rose 25% in their contribution to Sri Lanka's total DALYs (disability-adjusted life years)[9]. As Sri Lanka's economy continues to improve and its health indicators approach those of high-income countries, congenital disorders are likely to become even more prevalent and require a larger proportion of health resources.

To deal with the rising burden of congenital disorders and combat the resultant pressures placed on health systems, the World Health Organization has highlighted the need for building capacity in community genetics services to diagnose, manage, and prevent the subset of congenital disorders with underlying genetic causes in (LMICs)[5].

---

[1] The World Health Organization considers the term congenital disorder interchangeably with congenital anomaly, birth defect, and congenital malformation. It defines these as "structural or functional anomalies (e.g. metabolic disorders) that occur during intrauterine life and can be identified prenatally, at birth or later in life" [5]. Congenital heart defects, neural tube defects, and Down syndrome are the most common serious congenital disorders worldwide. The International Classification of Diseases, 10th revision (ICD-10) breaks down congenital anomalies into malformations, deformations, and chromosomal abnormalities and does not categorize hemoglobinopathies (such as thalassemia and sickle-cell disease) with these conditions [6]. For this reason, "congenital disorder" will be the term used in this report; hemoglobinopathies account for 6% of congenital disorders worldwide and have been reported as problematic in Southeast Asia. Congenital disorders therefore includes conditions caused by environmental toxins or maternal infections (congenital syphilis and rubella, HIV, toxoplasmosis) and conditions caused by changes in genetic information, including single gene disorders, chromosomal abnormalities, and multifactorial genetic causes. These changes lead to malformations such as cleft lip/palate and club foot, to genetic syndromes, and to metabolic disorders [7].

Community genetics services include pre-conception care, newborn screening, carrier screening, and antenatal screening and diagnosis of congenital disorders and genetic disease through laboratory or clinical means.

Pre-conception care includes those basic services related to adequate nutrition, infection control, folic acid supplementation, and family planning; many of Sri Lanka's past gains in maternal and child health can be attributed to proper implementation of this type of community genetic services [10]. As for the other community genetic services, we found no reports of carrier or newborn screening programs taking place in Sri Lanka. We did, however, find a report stating that antenatal community genetic services have existed on a limited scale in Sri Lanka for decades but that they haven't been integrated well into the public system of antenatal care [8]. We therefore chose to focus our study on antenatal genetics services as the next logical aspect of care in which scaling up community genetics services might bring gains in maternal and child health beyond those already achieved. In light this consideration, and of Sri Lanka's economic growth and shifting disease burden, we were led to ask: what is Sri Lanka's current capacity to provide antenatal genetics services and could building that capacity lead to further improvements in maternal and child health?

1.2 Congenital Disorders and Antenatal Care in Sri Lanka

To understand whether and how antenatal genetic services can improve maternal and child health and determine both the feasibility of and the need for scaling

up antenatal genetics services in Sri Lanka, more information is needed about the prevalence of congenital disorders in Sri Lanka. Information is also needed about antenatal care. Our literature review provided few data on the prevalence of congenital disorders in Sri Lanka and on their social or economic impacts. An Annual Health Bulletin released in 2007 estimated congenital malformations, deformities, and chromosomal abnormalities broadly accounted for 89.5/100,000 deaths in infants under 1 year of age, but no individual disorders were included in the mortality and morbidity data [8]. Aside from one report that a registry for patients with thalassemia would be established in Kurunegala [11], limited information is available on if and how data on congenital disorders are collected and analyzed. While sufficient data are available to detect a recent rise in the contribution of congenital disorders to DALYs, more granular information is needed to capture trends in the prevalence of specific conditions. A robust genetic epidemiology system for surveillance, diagnosis, and reporting of congenital disorders can provide this information and guide efforts to build capacity for community genetic services. However, in order for data collection to occur uniformly and sustainably, it must be prioritized in comprehensive national health strategies. Characterizing Sri Lanka's current system for genetic epidemiology is therefore necessary to understand which services, if any, could be developed and sustained.

Information on Sri Lanka's health system and antenatal care in general on the other hand is more abundant than is information on genetic services. While the public

sector delivers the majority of inpatient care, the private sector delivers a large fraction of outpatient care: 15.0% of respondents to the 2012-13 Household Income and Expenditure Survey (HIES) reported seeking outpatient care at a private hospital in the past month, compared with 17.4% seeking outpatient care at a public hospital [12]. Private health expenditure as a percentage of total health expenditure rose from 51.9% in 1995 to 56.3% in 2008 [13].

The growing role of the private sector aside, the bulk of antenatal care takes place in the government sector, where services are free and generally trusted [14, 15]. Sri Lanka's institutionalized health system includes both primary and referral hospital facilities as well as field-based services [14]. The field-level health workers credited with much of the reduction in maternal mortality are the Public Health Midwives (PHMs) who play a critical role in maternal healthcare delivery. Their extensive geographic coverage has greatly improved access to care [10, 14]. In fact, they have helped make access to antenatal care nearly universal. In 2013 the Sri Lanka Medical Association reported that 99% of the population is covered by antenatal care [16]. The components of that routine antenatal care, the services that all women should be provided as a matter of policy, are laid out in an antenatal care package published by the Family Health Bureau of the Ministry of Health [17].

1.3 Screening and Diagnosis of Congenital Disorders

The antenatal care package however makes no mention of gestational services, apart from basic pre-conception services, related to the detection of congenital disorders with underlying genetic causes. Lack of information and in some cases unclear information leads to uncertainty over which services are available and whether they are utilized. The Sri Lanka College of Obstetricians and Gynecologists releases its own guidelines, and according to them, antenatal care should include ultrasound and maternal multiple-marker screening for structural anomalies. Other sources report that invasive tests for chromosomal abnormalities and single gene disorders (carried out using amniocentesis or cord blood sampling) are available in the private sector [8, 18]. However, information on the quality and prevalence of screening and diagnostic services is limited. Dias et al. suggest that the skill of technicians performing the procedures is inadequate and that despite the availability of multiple screening methods, knowledge of Down syndrome screening options is low among not only patients but also health care workers [19, 20]. Clearer information on the components of antenatal care in practice and the role that screening and diagnosis of congenital disorders plays in that care is also necessary to understand the need for and feasibility of building capacity for antenatal genetic services.

1.4 Non-Invasive Prenatal Testing (NIPT)

Non-invasive prenatal genetic testing (NIPT) is of particular interest to our research group. NIPT is a relatively new genetic screening technology that some suggest is well suited to LMICs, where genetic disorders and hemoglobinopathies are often prevalent and technical resources are constrained [21-23]. NIPT uses cell-free fetal DNA (cffDNA) present in a pregnant woman's bloodstream to detect fetal sex, chromosomal disorders, and in the future, single gene disorders including beta-thalassemia, though the technology is not currently at the point of detecting them [24, 25]. The test requires only a blood draw, can be performed as early as 10 weeks into pregnancy, and detects Down syndrome more accurately than traditional biochemical screening tests commonly used to screen for chromosomal abnormalities; the lower false positive rates of these new tests thus reduce the need for technically demanding invasive procedures that carry a risk for miscarriages [26]. This is especially important in LMICs where facilities and trained physicians for performing the procedures are often limited and present only in urban areas. However, NIPT's utility in a given LMIC depends on access, technical capacity, and cost, and country-specific research is necessary to identify the challenges or advantages NIPT might present in particular social, structural, and legal contexts [18, 23].

1.5 Ethical, Legal and Social Considerations

Ultimately, for any antenatal genetic services—NIPT or the more traditional methods of screening or diagnosis—to be implemented or expanded, they must be appropriate for the unique considerations of Sri Lanka's healthcare system and its legal and social climate. Abortion is illegal in Sri Lanka except when a mother's life is at risk [27-32]. This constrains what expectant mothers can do in response to a positive screening result. Debate surrounds the question of whether it is ethical to offer antenatal screening and diagnosis in countries where abortions are prohibited [33]. Over recent decades, bills have been drafted to amend abortion laws, spurred by the recognition that diagnostic methods are available to detect fetal abnormalities. Despite abortion being illegal, it is nonetheless widely practiced and unsafe abortion was the second highest contributor to maternal mortality in Sri Lanka [31, 32, 34]. In 2005 the Ministry of Health estimated that 7% to 16% of women's hospital admissions were due to complications of abortion, a range that likely underestimated the actual rate since many women are reluctant to disclose their involvement in an illegal procedure [31]. Recent data indicate a rise in unsafe abortions [35], with estimates ranging from 260,000 to 290,000 abortions per year in a nation where there were only 365,792 live-births reported in 2013 [36, 37].

None of the attempts to change the law has been successful thus far, and many attribute the failures to religious opposition from the significant Buddhist and Catholic populations. Religious beliefs help explain the current atmosphere surrounding

termination of pregnancy, and they also help explain current practices in antenatal screening and diagnosis. A better understanding of religious beliefs and other sociocultural features of Sri Lanka's population is necessary, as these important contextual factors will continue to influence the availability and uptake of community genetic services.

1.6 Research Objective and Specific Aims

The overarching goal of this research is to characterize current antenatal care for congenital disorders in Sri Lanka, to evaluate the need for, and feasibility of, altering it—including the introduction or extension of screening and diagnostic services such as NIPT—to further improve maternal and child health. In order to accomplish this goal, the specific aims of this study are:

Aim 1: To understand the current landscape of antenatal care. We placed particular emphasis on screening and diagnostic services, and on the availability of resources for informed decision-making about genetic testing, including genetic counseling resources. To address this aim, I gathered the perspectives of key stakeholders in antenatal care and community genetics services. Stakeholders include:

- Experts and authorities including maternal care specialists in public and private hospitals, health administrators, government officials, medical

geneticists and representatives of genetic/genomic service providers as well as Down syndrome support organizations.

- Field-level maternal healthcare providers known as Public Health Midwives (PHMs), health professionals responsible for the majority of routine antenatal care in Sri Lanka.

Aim 2: To characterize prevalence of congenital disorders and genetic disease in Sri Lanka. To address this aim, I gathered information on current practices for the surveillance, diagnosis, and reporting of congenital disorders, and learned how key stakeholders evaluate the prevalence of such disorders and their impact on health systems and on society.

Aim 3: To understand contextual factors influencing the availability of antenatal genetic services within the public health system. I assessed the perspectives of key stakeholders on the possible utility of antenatal genetics services and identified ethical, legal, and social issues that guide availability, uptake, and future implementation patterns of these services in Sri Lanka.

2. Methods

This report describes the results of a qualitative study conducted between May 2015 and August 2015. We conducted nineteen Key Informant Interviews (KIIs) with representatives of stakeholder groups engaged in the delivery of antenatal care and genetic services and seven Focus Group Discussions (FGDs) with Public Health Midwives (PHMs).

2.1 Setting

We (the PI and co-PI, Jenae Logan and Amila Chandrasiri) conducted research in two primary locations: the city of Colombo, Sri Lanka's capital and largest city, and the district of Galle, the most populous district of Sri Lanka's Southern Province.

Colombo is the headquarters for the Ministry of Health and the Family Health Bureau. It is home to Sri Lanka's highest concentration of experts in antenatal healthcare policy and delivery. Large public and private hospitals, numerous field antenatal clinics, and Sri Lanka's only reported public and private genetic testing facilities are located in Colombo. Colombo is where new genetic testing technologies are most likely to be marketed, and many key informants live and/or work within the Colombo municipality [8, 32, 38].

Though Colombo is home to specialized antenatal services, much of routine antenatal care is spread throughout rural and urban areas in the rest of the nation. The Galle District is home to more than a million people, primarily Buddhists of Sinhalese

origin but with representation from other ethnic and religious groups. Table 1 breaks down the demographic characteristics of residents of the Southern Province and the Galle District compared to the nation overall.

Table 1: Demographic Features of Residents of the Galle District

	Ethnic Group				Religion				Literacy		
	Sinhalese (%)	*Tamil (%)*	*Moor (%)*	*Other (%)*	*Buddhist (%)*	*Hindu (%)*	*Islam (%)*	*Other (%)*	*Literate (%)*	*Computer-Literate (%)*	**Total Population**
Sri Lanka	74.9	15.3	9.3	0.5	70.1	12.6	9.7	7.6	95.7	24.2	20,359,439
Southern Province	95.0	1.7	2.9	0.4	94.7	1.3	3.2	0.8	95.3	22.4	2,477,285
Galle	**94.4**	**1.9**	**3.6**	**0.1**	**93.9**	**1.5**	**3.7**	**0.9**	**96.2**	**24.3**	**1,063,334**
Matara	94.3	2.6	3.1	0.0	94.1	2	3.2	0.7	94.8	21.7	814,048
Hambantota	97.0	0.4	1.1	1.5	96.8	0.2	2.5	0.5	94.1	20.0	599,903

Source: Sri Lanka Department of Census and Statistics. Census of Population and Housing – 2012. Final Report – Southern Province [39].

With respect to the provision of Maternal and Child Health services specifically, the Galle District is subdivided into 20 Medical Officer of Health (MOH) areas, where Public Health Midwives and their support staff deliver care. The Galle District is also home to Karapitiya and Mahamodara Teaching Hospitals and a number of Southern Province administrative health offices. A subset of key informant interviews were conducted in Galle.

2.2 Participants

Key Informants (KIs) were selected by purposive sampling for their ability to provide a range of perspectives on antenatal and genetic services in Sri Lanka. KIs were initially identified through review of official government documents and published research articles. Among those selected and contacted via email with requests for participation were experts, thought leaders, and authority figures in prenatal care delivery, health policy, and community genetics services. While used snowball sampling during interviews to identify additional KIs, all but one of the informants KIs identified were already in our list, so our initial list was already near saturation. The KIs fell under three broad categories: 1) Healthcare Providers, 2) Health System Administrators, and 3) Genetic/Genomic Experts. Table 2 further describes participants. Inclusion criteria were that KIs had relevant expertise and agreed to be interviewed. Those who could not speak English or Sinhala would have been excluded, but all KIs were English-proficient.

Table 2: Key Informant Interview Participants

Key Informant Type and Subtype		Interviews	Session Identifiers
Healthcare Administrators	Government Officials	4	64A, 630A, 612A, 612B
	Hospital Administrators	2	69A, 616A
Healthcare Providers	VOGs[§]	2	64B, 68A
	Fetal Medicine Specialists	1	724A
	Pediatricians	1	715A
	Clinical Embryologists	1	719A
Genetics Experts	Cytogeneticists	1	711A
	Medical Geneticists	2	619A, 619B
	Industry Rep.	3	612C, 612D, 711B
Other	Bioethicist	1	626A
	DS Support Org. Rep.	1	612E
Total:		**19**	

[§] VOG: Visiting Obstetrician & Gynecologist; the terminology used in place of OBGYN

Focus Group Discussions (FGDs) took place in 7 MOH areas selected for their geographic dispersion and varied socioeconomic and ethnic compositions. See Appendix A for a map of the Galle District broken into MOH areas. To recruit FGD participants, a Research Assistant (RA) contacted the Medical Officer of Health for each MOH area with a request for participation, and he or she then to gathered 8 PHMs assigned to that area. PHMs were required to play an active role in the delivery of antenatal care; those not currently practicing would have been excluded, but we did not encounter this situation. Table 3 shows the number of participants participating in the FGD in each MOH area selected.

Table 3: Focus Group Discussion Participants

MOH Area	Number of PHMs	Session Identifier
Balapitiya	8	FGD7
Bope-Poddale	6	FGD2
Galle MC	9	FGD4
Gonapinuwala	8	FGD1
Imaduwa	8	FGD3
Niyagama	9	FGD5
Rathgama	8	FGD6
Total:	**56 participants**	

2.3 Procedures

All study procedures were approved by the ethical review boards of Duke University (Protocol #B0958) and the University of Ruhuna Faculty of Medicine. Written informed consent was obtained from Key Informants who interviewed in person, and verbal informed consent was obtained from those who interviewed over Skype. Consent was obtained prior to turning on a recording device. All Key Informant interviews were

conducted in English by the PI (Jenae Logan) and the co-PI (Dr. Amila Chandrasiri), with the PI as the lead interviewer. Semi-structured interview guides were developed for each broad category of informant, and additional questions were asked depending on a KI's given expertise. KIs received no compensation. Interviews lasted 30 minutes to an hour.

Written informed consent was also obtained from all PHMs prior to recording and discussion. FGDs were facilitated in Sinhala by the co-PI. The PI and an RA fluent in Sinhala were in attendance, and they were responsible for operating the recorder (PI) and taking detailed notes (RA). Sessions lasted 40-70 minutes per site and ended with an opportunity for PHMs to ask questions about any of the topics covered. Participants were each given 3 box folders for use in their offices. Refreshments were provided to all staff present at the MOH office.

2.4 Measures

2.4.1 Key Informant Interviews

In-depth interviews with expert stakeholders were conducted with the following goals:

1. To gain insight into the state of antenatal care and antenatal genetic services, the state of genetic services in general and especially of community genetic services, and resources for genetic counseling and patient informed decision-making.

2. To characterize diagnosis, surveillance, and reporting practices related to congenital disorders as well as the public health and societal impact of those disorders in Sri Lanka.
3. To identify contextual factors influencing the implementation and uptake of antenatal genetics services especially in the public health system.

Semi-structured interview guides were used to ensure that major topics were addressed. Guides included questions and probes about routine and specialized antenatal care; congenital disorder diagnosis, surveillance, and reporting; social acceptance of and support resources for individuals with congenital disorders; genetic technologies available, including new non-invasive prenatal testing; and contextual factors (practical, legal, ethical, and social) shaping availability and utilization patterns for antenatal genetics services. Examples of questions asked during KI interviews are in Appendix A.

2.4.2 Focus Group Discussions

FGDs were conducted with public health midwives in Galle with the following goals:

1. To understand the components of routine antenatal care in practice
2. To further characterize diagnosis, surveillance, and reporting practices of congenital disorders
3. To understand PHM's knowledge of and experiences with congenital disorders and antenatal screening for and detection of those disorders

4. To learn how PHMs perceive mothers' knowledge of congenital disorders and antenatal screening for and detection of those disorders

A focus group discussion guide was designed and used to ask about (1) routine antenatal care components, (2) antenatal services requested by patients, (3) PHM knowledge of and experiences with congenital disorders and screening and diagnostic tests, and (4) mothers' knowledge of genetic disease and screening and diagnostic tests. Examples of questions asked during FGDs are listed in Appendix A.

2.5 Analysis

Each interview was recorded, and the PI transcribed each audio recording within 48 hours of interview completion. Analytic memos were written for each interview for ideas about initial codes. FGDs were recorded and transcribed by one of two RAs as soon as possible after FGD completion and then translated into English. All recordings and transcripts were stored in a secure folder in Duke University's HIPAA-compliant cloud-based storage system. Transcribed and translated documents were stripped of identifying information and then uploaded to NVivo qualitative analysis software (QSR International Pty Ltd. Version 10.2.1).

Two preliminary codebooks were developed for KIIs and FGDs based on questions from interview and FGD guides as well as analytic memos. Preliminary codes were entered into NVivo and applied to a KII transcripts and FGD transcripts by the PI. A separate analyst independently coded the first four KII transcripts and two FGD

transcripts, blinded to the codes applied by the PI, to assess inter-coder reliability. Coding disagreements were discussed amongst the researchers, and codes were eliminated, combined, or clarified until agreement was reached. The constant comparative method was used to identify themes that emerged from the data during the coding process so that they could be added to the coding framework [40].

After coding, data excerpts were grouped under their coding assignments and then collapsed into larger categories based on content similarity. Through an iterative process of thematic analysis, both KII and FGD categories were grouped into the themes and subthemes to address the study's specific aims. Some of the aims were addressed using only information gained in a subset of KIIs based on respondents' unique expertise, and others were addressed by information obtained from KIs and PHMs. For aims of a more descriptive nature (for example, which services are included in routine care?), summaries were constructed from coded sections of text extracted from NVivo using summarizing content analysis [41]. For other aims in which there was value in knowing the opinions of multiple stakeholders, illustrative quotations were extracted from transcripts. Literature searches and review of any public documents mentioned by study participants were used to confirm and supplement information.

3. Results

To meet the overarching objective of our research project, we present our findings according to the Specific Aims laid out in the beginning of this report. Section 3.1 corresponds to our first specific aim and characterizes the current landscape of antenatal care in Sri Lanka, with a particular focus on the role of antenatal genetic screening and diagnostic services and the current infrastructure in place to support patients' informed decision-making about antenatal genetic screening and diagnosis. In Section 3.2 we address our second specific aim; we characterize the landscape of genetic services by describing how congenital disorders including genetic diseases are diagnosed and reported and by discussing their impact on Sri Lanka's health system as well as on individuals and families. Finally, in light of our findings about the current state of antenatal care (Section 3.1) and the impact of congenital disorders (Section 3.2), we report findings that address our third specific aim in Section 3.3, namely participants' views of the utility of antenatal genetics services and contextual factors influencing the availability and uptake of these services. We also report a number of possible alternatives to scaling up antenatal genetics services they suggested to address the burden of congenital disorders.

3.1 Understanding Antenatal Care and Care Related to Congenital Disorders

In Section 3.1, we present information addressing Specific Aim 1. While our primary objective with this aim is to understand the role of antenatal screening and

diagnosis of congenital disorders, in order to do that we needed to first understand the state of antenatal care in general. We therefore begin in Section 3.1.1 by describing current components of antenatal care in Sri Lanka in the public and private sectors.

We then present in Section 3.1.2 information gathered on methods of screening and diagnosis of congenital disorders, and particularly genetic diseases, currently available in antenatal care. We discuss the types of screening and diagnosis that are performed in Sri Lanka and the patient population that uses them. In Section 3.1.3 we report PHMs' and KIs' perceptions of patients' knowledge of the antenatal screening and diagnostic services available in Sri Lanka and of mothers' primary concerns during pregnancy. In Section 3.1.4 we describe current resources available in Sri Lanka to promote patients' understanding of the results of genetic screens and diagnostic tests.

Finally, to investigate whether and how new non-invasive methods of antenatal screening (NIPT) could be implemented within public sector care, we devote Section 3.1.5 to a discussion of the current availability and utilization of NIPT in Sri Lanka.

3.1.1 Components of Routine Antenatal Care

Activities in the routine provision of care are decided by the antenatal care package laid out by the Family Health Bureau. The standard package of services is summarized in Table 4 as background needed to frame reports from KIs and PHMs.

Table 4: Routine Maternal Care Guidelines

Pre-Conception Care	
Registration of Married Couples and Educational Sessions	Eligible couples register. They receive folic acid supplements and rubella immunization for rubella. They are screened for risk for a number of conditions. They receive information on family planning and on symptoms of pregnancy.
Antenatal Care Clinic Activities (Booking Visit, 6-8 weeks)	
Registration of Pregnancy	If not already registered in the field
Confirmation of Pregnancy	Clinical or laboratory confirmation
History Taking	Personal history (past and present), medical history, family history, pregnancy history (past and present)
Nutritional and Clinical Examination	Height and weight measured; BMI calculated. Blood pressure taken and recorded.
Routine Investigations (some repeated at another visit from 24 to 28 weeks)	
Urine	To measure urinary glucose
Hemoglobin (Hb)	To detect anemia
Blood Sugar	To detect gestational diabetes
VDRL (HIV & Syphilis)	Venereal Disease Reference Laboratory
Blood Group & Rh Status	May use results from previous pregnancies.
Tetanus Immunization	
Folic Acid Prescription	During first 12 weeks of period of amenorrhea (POA)
Micronutrient Supplementation	After 12 weeks into POA; folic acid, worm treatment, vitamin C, iron, calcium, malaria prophylaxis
Follow-up Antenatal Care Clinics	
The FHB recommends each woman have 9 clinic visits during her pregnancy.* The "Maternal Care Package – Guide to Field Health Staff" guides management decisions for each visit. At each visit, PHMs track the progression of pregnancies, make risk assessments, and discuss the results of any tests performed since the last visit, among other specific activities.	
Antenatal Care Classes	
Pregnant women should be able to participate in 3 antenatal classes (1 per trimester) along with their husbands. PHMs are given guidelines on the content and scheduling of antenatal classes.	

Source: Constructed using information obtained during Key Informant Interviews and supplemented by a circular released by the Family Health Bureau in 2014 specifying the most up-to-date components of the routine system of antenatal care as well as by the "Maternal Care Package–Guide to Field Health Staff," a manual guiding management decisions for clinic personnel.

*Note: Care differs for high- and low-risk pregnant women. The conditions that are classified as high-risk by the field guide are listed in Appendix B. High-risk women are referred to consultants, who advise a plan for clinic visits. PHMs increase the frequency of home visits to high-risk women. The field guide provides a detailed breakdown of how care should differ for each high-risk condition. For women of advanced maternal age, field staff are instructed to explain to her the risk of chromosomal abnormalities and first trimester miscarriages associated with advanced age.

Key Informant Perspectives

Key Informants confirmed that service delivery actually follows the antenatal care policies in the manual. Healthcare administrators report that registration of pregnancy takes place as early as possible and no later than 8 weeks into pregnancy. After a mother is registered at a local clinic, she receives routine home visits from her specific PHM. She attends regular antenatal clinics for basic investigations, risk-assessment, and routine follow-up.

The routine package for all mothers includes recording multiple types of clinical history (see Table 4), physical examination, screening for familial diseases, identification of consanguinity, urine samples, and blood tests for glucose and proteins. Screening for potential antenatal infection includes VDRL testing for syphilis and HIV. Diabetes screening takes place at the first antenatal visit (before 12 weeks) to identify pre-existing diabetes and then again at 28 weeks to identify gestational diabetes. Mothers are immunized against tetanus and rubella, and their weights and BMIs are monitored throughout pregnancy.

KIs called attention to the aspects of the care package relevant to genetic disease; they mention history-based screening (including family history and individual history) in particular. Women also receive pre-pregnancy counseling and antenatal educational sessions, where they are advised to take folic acid to prevent structural anomalies and made aware of the risks of consanguineous marriages. Beta-thalassemia carrier

screening for soon-to-be-married couples takes place in districts where the hemoglobinopathy is more common. A basic ultrasound scan is recommended 11-14 weeks into pregnancy to assess fetal growth, head circumference, and overall wellbeing. One provider, a pediatrician, reported that almost all mothers have this scan at some point between 12-20 weeks, but that it is performed by a general obstetrician who can detect significant malformations but not minor abnormalities (715A). Ultrasound scanning for fetal abnormalities is available in the private sector. One healthcare provider noted that in the government sector this and other specialized screens are only offered to high-risk patients:

> "[It's a] low-resource setting, so we can't provide the prenatal care for all the women, but based on their needs, basically high-risk people we try to deliver care" (64B).

Healthcare administrators noted that the national program focuses on preventative activities rather than on screening for existing conditions. One healthcare provider remarked that Sri Lanka's antenatal care programs focus on the health of the mother:

> "The baby, not really…it's there, but we don't have much concern about …fetal abnormalities" (724A).

PHM Perspectives

The services described by PHMs as falling within routine antenatal care align with those reported by KIs. All PHMs reported making their first home visit no later than 8 weeks into pregnancy, and most reported visiting as early as 5-6 weeks, as soon as they were informed of a pregnancy. As noted in Table 4, care differs for high-risk

mothers. PHMs reported that the characteristics defining high-risk pregnancies are: (1) advanced maternal age (>35); (2) high or low maternal BMI (<18 or >25 kg/m^2); (3) maternal hypertension or diabetes; (4) history of subfertility, miscarriages or other complications during previous pregnancies; (5) consanguineous marriages; (6) maternal blood group incompatibility; (7) pregnancies in unmarried women; (8) unexpected or teenage pregnancies; (9) twin pregnancies; and (10) suspected breech presentation.

PHMs in all FGDs reported that when they encounter a mother with a high-risk pregnancy, they increase the frequency of home visits. They discuss with mothers and their family members strategies to manage risk during a pregnancy and describe additional investigations or treatments that might be required. Some PHMs direct mothers to the MOH, who then refers mothers to relevant specialists or clinics at tertiary government hospitals. Others advise mothers to register directly in a hospital with a consultant obstetrician.

A comment that emerged repeatedly in discussions was that not all women accept PHMs' referrals to government hospitals. The share of women electing to utilize private hospitals over government facilities varied from one MOH area to another. PHMs from FGD5 speculated that nearly 90% of women seek care in the private sector to avoid long waits in the government hospital, but PHMs in FGD4 reported that very few mothers used private hospitals. Mothers of low socioeconomic status seek care only in the government sector. Reasons cited for mothers' preferences for the private sector

included that consultations were faster and reports were returned more quickly, that husbands could be more involved (since some government hospitals prohibit husbands from attending consultations), and that people place more value on services for which they pay. Although most women ultimately deliver in public hospitals, consultants who have public appointments and private practices perform deliveries. PHMs in nearly all FGDs said that mothers feel they will be given more time during delivery if the consultant performing their deliveries has seen them in the private sector.

3.1.2 Current Role of Screening and Diagnostic Services

As noted in Section 3.1.1, antenatal screening and diagnosis of genetic disease are not a part of the routine care package laid out by the national program. The government does not provide facilities for antenatal screening and diagnosis nor promote the procedures for the majority of women in Sri Lanka. However, key informants and PHMs reported that antenatal screening and diagnostic procedures do play a limited role.

Healthcare administrators reported that consultant obstetricians (VOGs) have the discretion to perform or refer patients for specialized antenatal screening and diagnostic services (612B & 616A). VOGs act according to guidelines laid out by the College of Obstetricians and Gynecologists for maternal and child healthcare when choosing which patients they will refer for specialized services beyond the routine care package (616A). The VOGs we interviewed reported that the patients they opt to send for more detailed

investigations by a consultant radiologist or a fetal medicine specialist are patients in whom they have detected an abnormality during a routine ultrasound scan (68A).

According to one VOG, the most common investigation performed to detect congenital abnormalities in general is a specialized ultrasound referred to as an anomaly scan. It is most commonly performed in the private sector but is available to some extent in the government sector. Other available screens for genetic disease include triple testing; first trimester combined screening (FTS, which combines nuchal translucency ultrasound with a blood test); and NIPT. Available antenatal diagnostic services include karyotyping via chorionic villus (CVS) or amniotic fluid sampling.

Respondents provided several informative comments on the trends they see in the utilization of antenatal screening and diagnostic technologies (Table 5). All screening and diagnostic services are performed primarily for patients with high-risk pregnancies, with strong family histories of certain conditions, or who have encountered problems during previous pregnancies (64B). Even within the subset of patients who are high-risk, cost and geographic availability limit the number of tests and screens performed. Patients who live near the centers where services are offered, whose providers are familiar with the services, and who are able to pay out-of-pocket are the ones utilizing services.

We heard very few estimates of the absolute number of tests performed. One informant affiliated with a private hospital offering antenatal testing estimated that his

facility performs 500 triple tests and receives 5-10 amniotic samples for karyotyping per month (619B). The facility offers five-mutation screening for thalassemia but the KI noted little uptake, speculating that the private genetics company Genetech might receive more samples for thalassemia testing (619B). Genetech also receives amniotic fluid samples to be tested for trisomies 21, 18, and 13 as well as for sex chromosome abnormalities, but its representatives provided no estimates of test volume (612D).

Table 5: Comments on Antenatal Screening/Diagnostic Utilization Trends

612B	It's not equally a thing. So those in the Colombo-based, Kandy-based, even the Galle-based people will receive these kind of [antenatal screening and testing] services, but not for the people in the other areas.
69A	So if they want to go for the more advanced treatments it's available in the private sector. Things are available, but for a fee.
64B	Nowadays I'm prescribing these prenatal testing, specially ultrasound, first trimester, 20 weeks, for different people who have money. Otherwise, high-risk patients in other group.
68A	But all women unfortunately they don't get this anomaly scan…. If they can afford it they get it done. Otherwise unless we detect something abnormal they don't really get it.
711A	The reality is that they [antenatal testing and screening technologies] are so expensive…the vast majority of people in the public sector just cannot think about the cost of anything, of amniocentesis.

3.1.3 Knowledge of Screening and Diagnostic Services

Key Informant Perspectives

KI perceptions of lay knowledge of antenatal screening and diagnostic tests are listed in Table 6. In the experience of the healthcare providers interviewed, the majority

of people come into consultations with no knowledge of or expectation for genetic screens or tests. They simply wish to be assured of the overall well being of their babies (630A). One KI attributes patients' lack of demand to their religious and cultural backgrounds and high regard for doctors (724A). However, he and other KIs note that there are differences among social groups. Educated people who know about the problems with certain types of pregnancies (advanced maternal age, consanguineous marriages) know about and request specialized testing services (64A & 619A). Individuals of lower socioeconomic status are particularly unaware and undemanding; not only do they have less access to the Internet to learn about tests, but they also feel that specialized tests and screens are too expensive to consider.

Genetics experts interviewed also believed that people aren't aware of the details of screens and tests. Genetic experts and providers say, however, that knowledge of genetic diseases and the tests that can detect them is increasing in the lay population (68A). As buying power also increases, KIs expect that demand for antenatal screening and diagnostic services will rise (619B). Another provider suggested not only that knowledge about tests is increasing but that inability to pay is becoming less of a deterrent; patients with strong family histories of congenital disorders in particular find ways to access tests even if they are poor (719A).

Table 6: Comments on Lay Population Knowledge of Antenatal Screening/Diagnostic Services

630A	Some parents…would like to know about gender of the child, and that he is fine, he is having all four limbs, those

	things. They…want to know whether the child is okay with everything.
724A	Our people I think, because of religious and the cultural background, whatever we say--because they have high regards on doctors--whatever we say, they don't question you back. They ask only one question: whether my baby is all right or not, that's it. If I say all right, that's it.
68A	There are people who want to know about everything. But there are people who, even [when] we tell them, they don't understand.
724A	Now people are getting more information and they just come by [to utilize services] their own.
719A	So some people [who] have the family history of Down syndrome or some abnormality, they think everything is related to Down syndrome. So they come, right? Whether they are socially…wealthy or not. They come.

PHM Perspectives

PHMs, who have the most frequent contact with mothers during the antenatal period, echoed the view of KIs that mothers are primarily concerned with the growth and overall wellbeing of their babies. Mothers ask questions about nutrition, pregnancy monitoring charts, due dates, expected modes of delivery, and sometimes, fetal sex, but not as often about the risk of genetic disease or fetal abnormalities:

> "They ask us about different myths regarding nutrition. They ask whether treacle, kurakkan, red color foods, gahala, pineapple are not good to eat" (PHMs in FGD2).

While most mothers wish to know that their children are without deformities, mothers who have given birth to children in the past with abnormalities or malformations like cleft palate and mothers over 35 are particularly concerned about congenital disorders. PHMs agreed with KIs that knowledge of congenital disorders and screening and diagnostic services remains low except among mothers with higher education levels.

PHMs noted that even knowledge of the basic services included in the routine care package is low. Though mothers are educated about routine investigations (Hb, VDRL, FBC, blood tests) during the antenatal period through health programs at antenatal clinics, they ultimately know the investigations simply as blood and urine tests and are unaware of their purposes, limitations, or timelines for administration:

> "They know that blood investigations are done. But they don't know for what those are done" (PHMs in FGD2).

Mothers are generally aware that a scan can inform them about the sex, growth and well-being of their fetus, but most think that a routine ultrasound scans can rule out all abnormal conditions. Though a few mothers confuse scans with X-rays and ask whether a baby can be harmed or even develop abnormalities from the scan, PHMs reported that mothers do not generally fear scans and ask about them:

> "Most of them like to get the scan done. They frequently ask about the scan from us. Rarely some mothers ask whether it is good to do a scan and whether [the] baby can get affected"(PHMs in FGD2).

Mothers who are more familiar with screens and tests for abnormalities get their information from the Internet, from PHMs, from doctors or midwives they've encountered in previous pregnancies, or from acquaintances who've had personal experiences.

3.1.4 Resources for Informed Decision-Making

The next step in achieving our overall research objective is to understand the resources available to ensure that patients understand the results of antenatal screening

and diagnostic tests and make informed decisions in response. In Section 3.1.4., we share the perspectives of Key Informants and PHMs on the volume and quality of genetic counseling for the screening and diagnostic procedures that already take place. We also investigate PHMs' perceptions of their knowledge of congenital disorders and antenatal genetics services, in order to assess whether they might be utilized as a resource to promote informed decision-making if the genetics services were introduced into routine antenatal care.

Key Informant Perspectives

KI views were mixed on whether Sri Lanka currently has the infrastructure to address ethical concerns surrounding genetic screening and return the results of screening and diagnostic tests in a manner that enables patients to make informed decisions. Since the national program does not promote screening and testing, it does not include any counseling services (612B). Administrators report that the most senior health provider, usually a consultant obstetrician, performs any counseling related to suspected abnormalities (630A). Healthcare providers confirmed that they are the ones responsible for counseling (64B), and many commented on the limited amount of time they can devote to counseling in both their public and private roles (715A). Genetics expert KIs agreed that time and lack of training restricts many providers from adequately counseling their patients. One cytogeneticist also highlighted physician attitudes as a contributor to inadequate counseling infrastructure in Sri Lanka:

> "So I have also heard stories about people who have had tests and have not been told the test [results] and then their child has been born with a genetic disorder… Somewhere between the test being done and the baby being born, something has broken down, communication… People want to give good news but are not so happy to give bad news. They do a test to tell the family [good news], but the bad news will also be there" (711A).

Other participants' comments on the resources for counseling to enable informed decision-making are listed in Table 7.

Table 7: Comments on Resources to Promote Patients' Understanding of Results

715A	I see them in the private sector as well, you know, in private sector you might have about 5-7 minutes to see the patient. So I will ask one of those patients to come to a separate… come and meet me at a separate time so that I can spend about one hour with them.
619A	[At the postgraduate level] they give a good training to the ones in clinical genetics on counseling, and to the genetic diagnostic people on the basis of counseling, a little bit, because properly speaking a good genetic counseling session should have a genetic counselor, a clinical geneticist, a genetic diagnostician, the family, the nurse, all around one table, that's the idea of one.
619A	But the biggest problem in Sri Lanka is, there is no pre-test counseling. And post-test testing. With regard to prenatal diagnosis. No one gives it. It should be done by the obstetricians and gynecologists who perform these prenatal tests. But they have no time. So if they don't have time, they shouldn't do it.

Though most KIs perceived a lack of proper counseling infrastructure, they did note some exceptions. One KI offering a range of specialized tests (724A) employs a genetic counselor whom all his patients must see. If he feels that patients traveling from far away will not have adequate follow-up counseling, he refuses to see them and perform screening or diagnostic procedures. Some private sector genomics companies also

incorporate counseling into the delivery of their services. Credence Genomics (711B) has a counselor on staff and trains doctors on the principles of Mendelian inheritance that they may use when explaining the results of tests to their patients. Genetech returns a well-explained report to the consultant physician who orders a test and does not return results directly to patients (612D and 612C).

PHM Perspectives

PHMs confirm the KI reports that consultant obstetricians, not field staff, are responsible for the limited amount of counseling related to antenatal screening and diagnosis. In fact, PHMs stated that they do not often discuss congenital disorders and related investigations during antenatal visits. They bring congenital disorders up for discussion only when discussing folic acid supplementation, hypothyroidism, or consanguinity, preferring instead to focus on nutrition, mental health, and breastfeeding. PHMs reported feeling uncomfortable when mothers asked where certain investigations were available and PHMs were unable to answer.

Some of their discomfort is superstitious: in two FGDs, midwives revealed that they fear that talking about disorders at the time of fetus formation may affect the fetus. Mothers are also afraid of this, and one PHM reported advising them "not to watch confusing things even on television." They reported that they tell mothers to avoid viewing photos depicting the possible complications during pregnancy, and that if they encounter a baby with an abnormality, they should avoid looking at it so that their own

will not be affected. However, another part of PHMs' discomfort stems from their lack of expertise. PHMs in all focus groups agreed that just like mothers' knowledge, their own knowledge of congenital disorders and the screens and tests able to detect them is inadequate.

PHMs were somewhat unfamiliar with the screening and testing technologies available. Nearly all mentioned the routine ultrasound scan (USS) used to identify certain conditions, but unlike mothers they distinguished between this early ultrasound scan performed at 12 weeks and a specialized ultrasound (anomaly scan) performed at 18-22 weeks. PHMs knew that this scan was available in Colombo, and some mentioned it being available in Galle. A few reported that certain investigations could only be done in private hospitals. PHMs brought up amniocentesis but were not sure how it differed from an anomaly scan. Some PHMs, like some mothers, were unclear about the distinction between X-rays and ultrasound scans. Very few mentioned blood tests to detect chromosomal abnormalities. One PHM brought up "alpha protein" saying that she had read about it in a book but did not really understand the test.

PHMs reported learning about congenital disorders and new or specialized investigations largely from the mothers in their fields or from discussions with fellow PHMs or doctors:

> "In my clinic there was one mother who had gone through the scan, then it was that mother who explained it to all of us" (PHM in FGD6).

> "When a mother comes to the clinic with new investigations, we ask about them from the doctor" (PHM in FGD3).

PHMs, like mothers, also use the Internet to gain information, particularly to answer mothers' questions about topics with which PHMs are unfamiliar. One PHM mentioned "Sapatha" magazine as a good source of information; others cited TV shows that they watched while visiting houses in the field. One PHM reported that her mother watched the shows and described them to the PHM when she returned from the field.

Though a few PHMs reported that they had learned more about congenital disorders by attending health conferences, no PHM had received formal training or an education module geared to congenital disorders and the tests that detect them.

> "Our knowledge isn't enough. We educate mothers with what we know. If not we refer to MOH" (PHMs in FGD2).

They would like more guidance on identifying babies with congenital disorders, caring for children with disorders, and understanding the reports of tests so that they can explain results to mothers:

> "Sometimes we also haven't received much knowledge on this. So, when mothers ask us different questions, we feel uncomfortable" (PHMs in FGD1).

Many expressed a desire for a program to educate PHMs as well as the mothers they serve about these aspects of pregnancy.

3.1.5 Non-Invasive Prenatal Genetic Testing (NIPT)

Current Use of NIPT

As mentioned in Section 3.1.2, non-invasive prenatal testing is available in Sri Lanka in a very limited way. However, during the course of our study an individual who contracts with Beijing Genomics Institute (BGI) to offer its non-invasive prenatal test (The NIFTY™ Test) was identified and interviewed. We therefore devote this section to a discussion of current adoption of NIPT in Sri Lanka.

The KI sends samples to Hong Kong, China and receives test results 10 days later. Few people are aware that the test exists; he estimated handling 15 cases per month. Most of his patients work in the health sector, have married late, or have had babies with Down syndrome. Some are from other countries, including the nearby Maldives. He charges his patients 68,000 rupees (approx. 470 USD) but pays BGI 350 USD; courier fees comprise the additional cost. He performs the test as early as 10 weeks of pregnancy, and advises patients to have CVS at 22 weeks in the case of a positive result. BGI will pay for CVS as an optional follow-up procedure (valued at 400 USD). At the time of the interview no samples had been found positive for Down syndrome. If NIFTY™ ever returns a false negative, BGI will pay a family 68,000 USD; for the KI this "insurance policy" was an important factor influencing his decision to contract with BGI. Other companies have approached him to offer their non-invasive tests, but finding higher prices and no comparable insurance policies he remains with BGI. He reports that BGI is considering starting a lab in India to shorten the time it takes to learn results.

Fewer than half of the other key informants had heard of non-invasive prenatal testing. Genetic experts were most familiar with the test. No health administrators were familiar with it and roughly half of health providers were familiar. Healthcare providers who did know that NIPT is available in Sri Lanka reported that it is more expensive than biochemical screening and ultrasound scans (68A). One speculated that he did not think there was much uptake because the price of the test was too high (619B). One provider who refers his patients for NIPT estimated that he had referred nearly 100 patients, but only those in Colombo, and they usually didn't know about the test prior to seeing him (724A). He reports that his patients were satisfied with their results. He saw potential for NIPT to play a greater role in the future but thought cost might be prohibitive, as amniocentesis cost half as much at that point. He mentioned a recent audit of complications of procedure-related losses that found that all babies lost had some sort of abnormality, suggesting that invasive procedures carry minimal risk. Most people he encountered therefore chose amniocentesis for its cost and diagnostic ability (724A).

Current Use of Other Genomics Services

To understand how knowledge and utilization of NIPT might change in the future, we asked KI about the state of genetics and genomics services beyond NIPT. They reported that availability and utilization of such services is low.

Providers were familiar with the fact that karyotyping is available at the Human Genetics Unit (HGU) at the University of Colombo. The HGU offers cytogenetic screens

for roughly 2500 rupees (17.40 USD) as well as thrombophilia screens for women with recurrent pregnancy losses for 5000 rupees (34.80 USD). The HGU does not offer genomic tests for breast and colon cancer risk since the volume of tests requested is not large enough to rationalize stocking test reagents; however, these tests are offered in what one informant called the HGU's private sector "sister lab", the Asiri Hospital Reproductive Genetics Laboratory (619A).

Representatives of commercial genetics companies described a number of other services they or their competitors offer in Sri Lanka. Credence Genomics (711B) uses next-generation SNP-based methods for whole-genome and whole-exome sequencing on the Ion torrent PGM (Life Technologies) sequencing platform. It has not ventured into antenatal testing but does offer both Credence Carrier and Credence Newborn Screening. Another company, Genetech, was founded in 2002 and gained recognition for its pioneering work in forensic DNA testing and paternity testing. It performs a range of molecular diagnostic tests that it develops in-house based on published research. Like Credence it does not market antenatal testing, but it does have the capability to perform PCR-based trisomy tests (21, 18, 12) on amniotic fluid and chorionic villus samples (612C). Genetech also has tests for thalassemia, Duchenne Muscular Dystrophy, leukemia testing, and chromosomal mini-translocations (612D).

One KI reported that people from other countries take advantage of Sri Lanka's high quality tests offered for relatively low prices. He expects that the role of genomics services in Sri Lanka will grow:

> "You see every specialty there is a part, and now it is becoming more and more prominent. I mean, pathology, microbiology, parasitology, immunology, you name it. You know, pathology might be entirely genetics in a few years time" (711B).

Genomics services, including those for antenatal and neonatal testing, are currently targeted to Sri Lanka's upper middle and middle classes (711B).

3.2 Congenital Disorders in Sri Lanka

Since our overarching objective was to understand whether there is a need to scale up screening and diagnostic services especially for antenatal care, we needed to establish a basic understanding of prevalence of congenital disorders, and more specifically genetic disease, in Sri Lanka. We discussed in Section 3.1 that services for antenatal screening and diagnosis of genetic disease are neither widely available nor widely utilized in Sri Lanka. Here in Section 3.2, we situate the availability of these services within the context of disease prevalence in Sri Lanka.

We asked: how common are congenital disorders, and what are their relative contributions to Sri Lanka's overall burden of disease? How does caring for a child with a disorder affect a family? To answer the first of these questions in Section 3.2.1 we describe resources and methods used in Sri Lanka for surveillance, diagnosis, and reporting of congenital disorders. A new registration system was only recently put into

place and has not been a source of information on the prevalence of disorders in the past, so we also report in Section 3.2.2, how KIs and PHMs perceive congenital disorder prevalence based on their own experiences. Finally, to truly understand the value of screening and diagnostic procedures, we must better understand the social and economic impacts of congenital disorders on families. We must also understand whether support systems are available to help mitigate those impacts. These issues are addressed in Section 3.2.3.

3.2.1 Surveillance, Diagnosis, and Reporting of Congenital Disorders

Healthcare providers indicated that diagnosis of many congenital disorders has been particularly problematic in the past. It was not uncommon for parents to take a child home and begin to suspect that something was wrong but receive no medical confirmation for more than a year (626A). One pediatrician reported that about half of the parents he has encountered who have a child with a genetic disease have a proper diagnosis; most simply suspect that there is a problem with their child (715A). Since the government system lacks institutional resources for genetic diagnosis, cost proves prohibitive: he described an encounter with a child with clinical features of Patau Syndrome and noted that to obtain a definitive (genetic) diagnosis would cost 10,000 to 20,000 rupees (roughly 69 to 138 USD). Another informant stated lack of specialized training among healthcare workers as another contributor to unsatisfactory diagnoses. Though it is possible for obstetricians to detect some congenital disorders through a

routine ultrasound scan, to do so reliably requires special training in fetal medicine, a subspecialty that is not yet available in Sri Lanka (724A).

Other key informants speculated that diagnosis of more common disorders and of Down syndrome in particular has improved with better education among both parents and health professionals. One cytogeneticist (711B) who sees complicated cases agreed that diagnosis is improving but that many patients still come in having exhausted all other tests available. For them, the ultimate diagnosis is a rare genetic defect. Another genetic expert agrees with this observation:

> "The problem with many of the other conditions is that it's not so apparent at birth and you have to wait until the other physiological manifestations are there" (711A).

Beyond diagnosis, surveillance and reporting of congenital disorders have also been problematic. KIs reported that Sri Lanka lacked an integrated system for the surveillance, diagnosis, and reporting of birth defects (as mentioned earlier, a term used interchangeably with congenital disorders) until only recently. As infant mortality has dropped in recent decades, the Ministry of Health has recognized the importance of understanding how birth defects contribute to infant mortality.

> "If you take the under 5 deaths…the second leading cause is birth defects. 18.4%. That means that every 5 deaths, 1 is due to birth defects. It's high time for us to focus on these" (612A).

In 2010 the Ministry of Health formulated a birth defects national action plan to raise awareness, promote proper management, and improve surveillance to inform

prevention and control programs. It also introduced the National Birth Defects Surveillance Program to remedy a previously fragmented approach to data collection. The program is headed by the Maternal & Child Morbidity and Mortality Surveillance Unit of the Family Health Bureau. It was piloted in the Southern Province, and Galle was one of the pilot districts. In the new program, data on birth defects are taken primarily from morbidity and mortality records and newborn examination. One KI affiliated with the program cautioned that these sources of data alone are insufficient:

> "So remember now, birth defects: to capture 100% you have to have your eye until 6 years. By 2 years you will capture only 96%...1st year, 80%; and 2 years, 96%; and 100%, 6 years" (612A).

In order to register the defects not visible at birth, the program brings different service providers into the system to capture data at multiple time-points. Field-level staff must investigate all infant deaths and report whether a defect was the root cause. PHMs must register defects they find during routine service encounters with children under 5 years of age, and Public Health Inspectors (PHI), another category of field-level health worker, must register those they find during school medical inspections for children above 5. The KI reported that compliance with registration requirements is good and that it will be easy to integrate new activities into the web-based system as the program scales up.

Among other KIs interviewed, we found that health administrators in Galle (64A &630A) were well informed about the national program. One noted that doctors are not uniformly aware of their responsibilities within the program, a lack of awareness she

attributed to staff turnover as consequence of the government health system's policy that health workers be cycled between the field and hospital levels. One government official agreed that compliance is good but cautioned:

> "…[I]n some hospitals overloaded with work, this leads to burden to them [medical staff], but they are not refusing, they are not complaining, they are doing it" (630A).

Among our focus group participants no PHM was familiar with the reporting practices specified by the National Birth Defects Surveillance Program. All stated that most abnormalities are identified at hospitals during newborn examinations, and that when they encounter a baby with an abnormality that becomes apparent later, they inform the MOH and/or note their finding on a mother's "card."

The National Birth Defects Surveillance Program requires that those registering birth defects in newborns complete a notification form immediately and a complete case abstraction within 14 days after birth. Completion of this form is guided by a manual describing ten priority conditions and how to recognize them. The ten conditions include (1) neural tube defects, (2) congenital heart disease, (3) thalassemia, (4) cleft lip/palate, (5) limb defects, (6) congenital rubella syndrome, (7) congenital hypothyroidism, (8) congenital syphilis, (9) chromosomal abnormalities, and (10) unspecified severe deformities. The KIs we interviewed did not share their experiences with these forms or comment on whether they think the program has improved diagnostic accuracy.

3.2.2 Prevalence of Congenital Disorders

Key Informant Perspectives

Since systems of data collection have been fragmented until now, Key Informants base their perceptions of the burden of congenital disorders on their own experiences and on the limited data that have been publicly available up to this point. When we asked healthcare administrators about the prevalence and impact of congenital disorders in Sri Lanka, they focused on the public health significance of the disorders compared with other health conditions. Some considered the prevalence of birth defects in general low and argued that the government health system is capable of managing most conditions:

> "When you consider…the epidemiology of birth defects…all are not lethal. So with the development of the science and medicine so that most of them can be managed easy, and within our heath system" (612B).

Another KI praised Sri Lanka for achieving "almost first-world indicators" with a "third-world investment," reducing IMR to roughly 9 deaths per 10,000 live births. Only 18% of those 9 are related to genetics, and in his eyes that is just one life:

> "…[W]hen you're trying to determine how to save it [one life], there may be more cost-effective ways" (619B).

Others KIs disagreed. One thought in absolute rather than relative terms and viewed the prevalence of birth defects as significant:

> "In 2011 we carried out a survey and we found 321 children with birth defects at birth. And we extrapolated this to the 2011 actual births, number of births. And every year that means…there is a caseload of 6400 children…with birth defects.

So it's a big burden" (612A).

With regard to a thalassemia, a genetic condition reported by many sources to be common in Sri Lanka, one KI viewed the burden as relatively small and better addressed by funding individual treatment rather than mass screening:

> "Because in Sri Lanka, if you really look at the numbers, we are not talking of large numbers. We are talking of about 80 thalassemics born in Sri Lanka every year. 15% are the second child in a family…With reproductive counseling, and donor insemination or something, you can prevent those. It is cheaper for us to actually treat the other[s] with bone marrow transplantation and maybe give them a cure, rather than for us to set up a large program of screening and pregnancy termination" (619B).

PHM Perspectives

PHMs did not speculate as to the absolute or relative prevalence of congenital disorders, but most reported that they had encountered a variety of disorders during their careers. The ones PHMs reported as being most common varied from one area to another, with some reporting cleft lip and palate as the most common, and others reporting heart defects or Downs syndrome, which was brought up by PHMs in all focus groups. Other conditions mentioned included polyhydramnios, autism, heart defects, spina bifida, imperforate anus, kidney problems, dextrocardia, hydrocephalus, and anencephaly. When we asked whether the number of congenital disorders PHMs encounter has increased or decreased over time, we heard mixed views. One reported that the number of Down syndromes in particular has increased; others reported that the

number of most disorders had decreased and attributed this decrease to improved awareness of the importance of folic acid.

3.2.3 Social and Economic Impacts of Genetic Disease

The final aspect of our characterization of congenital disorders is their impact on individual families. An important mediator of that impact is the availability of social and financial support systems for those families.

One healthcare provider we interviewed spoke of the large financial burden placed on parents whose children have congenital disorders. They may need to travel to the hospital for special treatments or therapies, requiring them to miss work and to hire expensive transportation (715A). Other KIs spoke of the social strain of having a child with a disorder. One who had been involved in research exploring perceptions of disorders with genetic causes in particular found that understanding of the conditions is very poor, especially in rural areas (724A). Lack of understanding may lead parents to view their children as sources of embarrassment (612E).

The PHMs participating in our focus groups, like the KIs, reported that having a child with a congenital disorder places strains on a family:

> "When there is such child in family it causes harm to the whole family. It is good, if that baby dies at birth. If not it is a pain to others in the family as well. Even the other living children won't be looked after well" (PHMs in FGD3).

Mother may be restricted to their homes and fear having more children. They are often poor and live far from available treatments. Leaving work adds to their financial strain.

KIs clarified that embarrassment and shame are not universal. A KI who had been involved in separate research into societal perceptions found acceptance rather than embarrassment among some groups, particularly in Christian circles.

> "Their view was, we have been given a Downs syndrome child as…in a sense as a challenge from God. We have been singled out to be given this special child, and it's our kind of duty to God and duty to the child to do what we can" (626A).

The KI had found different sentiments in the Buddhist families he encountered, reporting that husbands often deserted their families or resorted to alcoholism:

> "There's a tremendous amount of shame attached to having a Down syndrome child because that signals all kinds of karma" (626A).

As for supportive resources to lessen economic and social burdens, KIs reported limited availability. One KI noted that while there are government funds allocated to assist families, the funds are insufficient and difficult to access.

> "For example, to buy a [wheel]chair or something like that will cost you at least about 10,000 rupees and that's a very big expense for them. On top of their daily needs. And the government will…'officially' support. But it's very difficult to get this support. You have to go through lots of offices, fill out lots of forms, go behind the people, and get it done" (715A).

This KI does not believe that NGO support is sustainable or has the necessary reach.

> "It's a problem in the whole country. [It can't be] just some non-governmental organization in Galle supporting Galle children" (715A).

But regardless of their reach, there are NGOs serving a subset of the population affected by congenital disorders. Some genetics experts with patient contacts attempt to link families with one another or with one the few supportive agencies operating in Sri

Lanka (711B). Some suspect that supportive resources are improving, particularly for individuals with Down syndrome. For those with conditions that are not apparent at birth, support remains inadequate. And while there are more facilities opening to educate children with Down syndrome, it is still difficult to support the children all the way through education and then find places for them to work (711A).

An informant representing one organization that provides support to children with Down syndrome and their families reported that the organization's goals are to bring children into society, to train health professionals and caregivers, and to change societal perceptions, which he agreed have been unfavorable throughout history.

> "In our country especially these children were looked upon as a burden to the rest of society. There are parents who hide these children, don't bring them into society… We want to change that perception. To say that they are not an embarrassment, you know you can enjoy this sort of child" (612E).

His organization operates a center that provides basic education to twenty-five children and holds workshops for parents and caregivers. It also holds an island-wide celebration for World Downs Syndrome Day. Since the organization was established, the KI has noticed progress in the children the center serves but has no data on changes in societal perceptions. He, like other KI's, agreed that the government must get more involved. He specifically called for them to educate health professionals and provide opportunities for employment to children who have received vocational training.

3.3 Evaluating Possible Utility Within the Current Context

In Section 3.3, we address our final Specific Aim, gaining the perspectives of stakeholders on the possible utility of introducing antenatal genetic screening and diagnostic services into routine antenatal care and identifying the various contextual factors that influence patterns of implementation and uptake. We begin in Section 3.3.1 by presenting informants' views on the advantages and disadvantages of antenatal screening and diagnosis of congenital disorders. We also discuss their views of the potential utility of non-invasive prenatal testing. We proceed in Section 3.3.2 to discuss the features of Sri Lanka's legal and social context that our participants see believe influence availability and uptake. We end in Section 3.3.3 by presenting a number of alternatives to scaling up antenatal genetics services that KIs suggest might be better suited to the Sri Lankan context.

3.3.1 Utility of Screening and Diagnosis

Key Informant Perspectives

Key informants had differing opinions of the utility of antenatal screening and diagnostic services and their place within Sri Lanka's health system, as illustrated by comments in Table 8. Healthcare administrators recognized the value of such services but stressed that for them to be offered in the government health system, they must be offered free of charge to the entire population. They also stressed that people must be

able to do something in response to the results they receive, and restrictive abortion laws currently limit their actions (64B).

Healthcare providers saw antenatal testing and screening services as potentially useful. The main reason was that for certain conditions antenatal detection enables earlier intervention and may increase a child's chance of survival or quality of life (64B & 724A). They also noted that in light of recent improvements in maternal health, continued progress may require scaling services up to the level of developed countries worldwide (64B). Antenatal genetics services are one area in which Sri Lanka falls below OECD countries. Providers nonetheless recognized practical constraints, saying that while they would like to offer screening to all patients, it would place a substantial burden upon them and add to already heavy workloads.

One molecular geneticist discouraged the use of antenatal genetics services due to his own religious beliefs, but he conceded that "from a service point of view," he believed that tests should be available (619A). Another geneticist felt that offering testing might be useful in some regions of the country—for instance, in areas with higher prevalence of thalassemia—but not in others (612D). Other experts felt that services should be scaled up simply to make access more equitable than it is now (711A).

Table 8: Comments on the Utility of Antenatal Screening/Diagnosis

616A (administrator)	I'm not saying we don't need those kinds of services. We do need them. For those who can, for whom it is appropriate. Yes, we do need to improve, you know, the kind of diagnostic facilities for antenatal testing. But then, in

	conjunction with that, what do we do with that? Are we going to do something? That's very important.
64B (provider)	Yeah so I think that… with the recent development, that prenatal screening has to be available, especially in the major centers…so we can detect these things and we can prepare and treat or whatever, because there are so many conditions now we can treat, right?
68A (provider)	What we tell them [couples who ask, "what's the point?"] is at least if we detect something [that] can be corrected, then we can plan the delivery… time and mode of delivery, and then we can deliver when the pediatric surgeon is available.
64B (provider)	So it is timely and needed thing for Sri Lanka [to offer genetic testing and screening], that when you consider the maternal health and the prenatal health, the prenatal screening is one of the compulsory components, because we have achieved the maternal care level, quality of maternal care level, close to developed countries. (64B)
724A	If you ask a midwife, probably they might come out with, "even if you diagnose an abnormality, or if you diagnose the Down syndrome, where's the remedy?" And the answer for me is--Down syndrome…yeah I know, termination is illegal. But Down syndrome babies can have some nasty structural problems… And always I justify it, asking everybody, if you have a right to know what's happening with [your[baby. If it's healthy, or…not healthy. I think it's the right of the patient. And… some conditions, if you diagnose them early, then you can do something prenatally… or at the time of delivery, to optimize the condition. But the problem is I can't do it for everybody. Though ideally, screening should be for everybody. Selective screening is not recommended…in this field.
711A	Well I think it has to be incorporated into the public sector health provisions. I think that…the fact that the children are born to any social class means that if you really want to help people it has to be in the public sector.

PHM Perspectives

A question posed to PHMs asking whether the saw a need for additional antenatal services was excluded from analysis. We had not considered that the presence of a community medicine specialist (our FGD facilitator) asking about antenatal genetic

services might imply to participants that we were advocating for the use of the services and lead them to report greater utility of genetics services than they truly perceived.

3.3.2 Contextual Factors Influencing Availability and Uptake

In relaying their perspectives on the current system of antenatal care and the utility of antenatal screening and diagnostic services including NIPT, KIs highlighted a number of contextual factors that had limited availability and uptake in the past. These factors are important things to consider in a discussion of whether these services can or should be offered to a larger fraction of Sri Lanka's population through the government programs.

3.3.2A Legal/Ethical Context

Prohibitive abortion policy and the ethical predicaments it presents are strong barriers to availability and uptake for many KIs, and their specific comments on legal and ethical issues are listed in Table 9. KIs suggest that legal issues have prevented greater uptake of antenatal testing and screening technologies and have deterred Sri Lanka's genetics and genomics companies from really entering the world of prenatal testing (719A, 612D). Since abortion is not permitted unless two consultants agree that a mother's life is in danger, healthcare providers said that patients and they themselves question the purpose of screening and testing (64B, 612B, 68A). The fact that abortion is still widespread despite its illegality introduces additional considerations to providers who debate offering tests and screens. Many do not want to risk legal ramifications for

providing information that led to an illegal abortion (616A and 619A). Others though, suggest that in some cases testing can actually reduce the number of illegal abortions that occur, since many tests will return negative results and reassure parents of the absence of an abnormality (619B). A new non-invasive method of testing, one KI reported, will not overcome legal issues regardless of whether it offers advantages over other screening tests, because the issue is one of termination and not the difficulty of performing tests (626A).

Table 9: Legal/Ethical Context

719A	In Sri Lanka we don't have legal provision to do termination. So because of that, the sector [antenatal screening and diagnosis] is not developed actually.
612D	We have the technique, and if someone inquires we do the test. But because of the ethical issues we do not promote the test in Sri Lanka. Because there are the ethical issues with the prenatal diagnosis: after you detect the chromosomal abnormalities then what is the next step?
64A	In our country, termination of pregnancy is not yet approved by any means. So even if we diagnose a congenital abnormality during the antenatal period, then you end up in a deadlock.
612B	I think the public health specialists, the one thing that we need to think about is the screening and the confirmation and the management. So if only the screen component is there, if you can't do the diagnosis at this time, you can't do the management, then it's a problem.
68A	Even we feel upset, because there are fetuses with multiple abnormalities, we know that this baby's not going to survive, but still we can't do [anything about] it.
616A	I mean, imagine the heartache. Going through those nine months. And what do they do? What they might end up doing is pushing them into illegal abortion, which is happening. There is no question, it is happening.
619A	Why get involved in controversy? I mean, you can get sued. If something goes wrong you can be sued and it's an abortion, it's termination. And it's a crime.
619B	I always relate a story of…a patient comes into my office

	and says look, I came to meet you because I was told by this consultant that you have saved 17 babies from thalassemia families. So I was wondering, what do you mean by saved 17 babies? I haven't saved 17 babies like that….I knew who had referred the patient and I went back to see [him], this particular person has referred 20 couples, and out of them 17 of them have ended up with the child being normal, having no mutation or being a carrier. So my interpretation of that was…in the absence of genetic testing, all of those 17 babies would have been terminated. But because the genetic test was there, and they were detected to be either carriers or normal, they were not terminated.
626A	So something like non-invasive testing people think, "Oh this is wonderful, this is fantastic, this is progress, this is moving forward." But I think in the Sri Lankan context in particular…that is not the issue. The issue is one of termination, and there are no routes, no legal routes to termination unless in extreme circumstances.

3.3.2B Financial Context

Legal and ethical predicaments aside, there are important financial considerations to scaling up antenatal genetics services, and KIs reports of those considerations are detailed in Table 10. As noted in Section 3.3.1, many KIs commented that to extend services to the entire population would require that the government sector incorporate them into the routine antenatal care package and offer them free of charge. Government hospitals have a limited budget and must prioritize services that are effective and sustainable (612B, 616A).

Health providers and administrators know that the services are available in the private sector and are regulated by the Directorate of Public Health Services, and for many, it would be acceptable for the services to remain exactly where they are (69A).

The national program focuses more on the preventive aspects of birth defect management, offering supplements and immunizations that are arguably more cost-effective than are screening programs (612B). However, other KIs argued that investing the resources to train healthcare workers in procedures to detect congenital disorders would save money in the long run, even if the cost of offering services to the entire population is currently prohibitive (64B).

Table 10: Financial Context

616A	I mean, for me, there's lots we can do antenatally, right, and our VOGs go abroad, get trained in some of these in-utero things that they are doing, and that's fine. But at the end of the day, is it cost-effective for a country like Sri Lanka?
612B	So then most of these things are available, but in the free health services…if we start we have to provide for 400,000 pregnancies per year, whatever the things. Even if we are going to introduce a small sheet of paper, again we have to provide it for 400,000. Our health indicators are so good because of the free health services…if you overburden the health services…it will collapse.
69A	So if they want to go for the more advanced treatments it's available in the private sector… things are available, but for a fee. We are giving all these [routine] services free of charge.
612B	There are so many hundreds of things, the new things coming out, new tests and everything is coming up every day. Then as a medical person, or as a country, do we need to implement all these things? If the people who can afford those kinds of things, [if] they can get them from the private sector?
64B	Policy makers and health planners must recognize and provide opportunities for the postgraduate trainings, and provide facilities, like ultrasonic scanners, and then provide training, especially from local trainers…so then, at the beginning it is like a big cost, but long run it is more cost effective. You can improve maternal health, and neonatal health.

3.3.2C Resource Limitations:

Many KIs raised the point that there are not enough specialized providers to see all women, let alone perform specialized services. Representative quotations on resource limitations are listed in Table 11. One KI (724A) estimated that Sri Lanka's health system handles 300,000-370,000 deliveries per year, and that fewer that 200 consultant obstetricians handle those deliveries. With relatively few obstetricians serving so many patients, it is unsurprising that specialized services have been restricted to women at high risk (64B). One KI certified by the Fetal Medicine Foundation to do invasive and non-invasive procedures notes that the few who are trained cannot offer NT scans and anomaly scans to the entire patient population. This KI requires referrals for patients he sees in the sites he visits to each month outside his primary location to keep his workload manageable (724A). He expects that the human resource scarcity may become less severe: a proposal is "in the pipeline" to introduce fetal medicine as a subspecialty, and there is a training module in place for new registrars (post-MBBS trainees) to do basic scans so that they will have background knowledge when they become consultant obstetricians.

As discussed in Section 3.1.4, genetic counselors are as few and far between as fetal medicine specialists. There are insufficient numbers to serve the patient population. This situation is faced in countries around the world, but one KI reports that he expects

it will improve as newly introduced training programs for genetic diagnostics and clinical genetics turn out batches of health professionals. Genetic diagnosticians are expected to set up laboratories, and clinical geneticists are expected to move into clinical units to assist with counseling (619A).

Some KIs noted a scarcity of material resources, though not as severe as human resource constraints. Many facilities are equipped with scanners, but providers reported that there are not enough scanners and that the resolution of the scanners is poor. One health administrator also reported that the scanning equipment available in her hospital is too basic to detect anomalies and perform high definition scans (69A). The KI trained in fetal medicine countered that the scanners could all be optimized to detect the conditions of interest if people were trained to do such optimization, though possibly not as efficiently as higher-resolution scanners (724A). Other testing and screening technologies beyond ultrasound scans are also limited by material resource constraints. One KI called attention to the fact that some screens and tests require specialized reagents and facilities choose not to offer them if there is not sufficient demand (616A).

Table 11: Human and Material Resource Limitations

724A	At the moment, if I start to do routine screening I will not be able to go out of this hospital.
64A	Overburdened is a big problem [for PHMs]. Overburdened the routine program will carry on but the final things like, to go and counsel about congenital abnormalities, and those things? Cannot be done when you are overburdened with work.
619A	We can match any equipment now, anywhere in the world. But the difference is that there are people available to translate that into lay language that they can understand.

	We have trained clinical geneticists and we have trained genetic diagnosticians. People in genetic diagnostics are expected to go and set up laboratories. People in clinical genetics are expected to move to clinical units and help with counseling and other matters. So we have trained a lot of people. Who we can—and we will—use in training here.
616A	Some of these tests are extremely specialized; they are only for…one or two patients. So it's not worth having test kits, stocking them for certain purposes.

3.3.2D Sociocultural Context

Key informants identified certain features of Sri Lankan society and culture that influence availability and uptake of antenatal genetics services, as illustrated by comments listed in Table 12. The first we alluded to in Sections 3.1 and 3.2, there is poor knowledge about congenital disorders and the services that detect them in the patient population and among health workers at all levels of the healthcare system. One KI who has surveyed pregnant women and their providers in different parts of the country found that knowledge about screening for Down syndrome was extremely low among women and only about 40% among health professionals. He noted that especially among older consultant obstetricians, the fetal medicine subspecialty was not popular during their training. New consultants may have had some exposure during training and are therefore more likely to refer patients for specialized services (724A).

Another KI, a molecular geneticist, agrees that training has improved in later batches of medical students. They attend talks on genetics and have a reproductive health module that includes genetics, sensitizing them to the topics of congenital

disorders and relevant tests. However, he thinks that despite their growing knowledge their ability to interpret test results remains inadequate, as does their ability to perform proper scans (619A).

One KI praised field-level staff (PHMs) as a potential resource to promote patients' informed decision-making but stressed that PHMs must receive adequate training and information in order to properly inform patients (724A). Though PHMs receive some information and training related to congenital disorders, it is mostly in the form of information on the risks of advanced maternal age, consanguinity, and thalassemia. KIs agree with PHMs that their training must be expanded, but they also caution against overburdening them (64A).

Parents are even less aware than health workers of congenital disorders and the services that detect them (724A). Some are even afraid of tests, believing that if a health professional advises a patient to seek a scan for anomalies, he or she must suspect that there is a serious problem with the pregnancy. KIs reported that though few people come in with pre-existing knowledge of scans, knowledge is growing. They noted that women must be taught that any pregnancy carries a risk and that scans aren't necessarily only for high-risk women (64B).

Religion

KIs describe religion as preserving the current situation in which antenatal screening and diagnosis are available but patients are restricted from acting on results

by opting for a termination. Buddhists and Christians account for roughly 90% of the population, and neither religion condones abortions. KIs differed in their views as to which religious group—Buddhists or Catholics—has been more resistant to changing attitudes about termination of pregnancy (619B, 612B). As mentioned in Section 3.2.2, some KIs perceive differences in the ways Christians and Buddhists regard the birth of a child with a congenital disorder, and other KIs perceive differences in the way the religions approach screening, testing, and termination when abnormalities are found (711A). One cytogeneticist thought that NIPT would face the same religious opposition just as it would still face legal opposition; when the underlying decision is based on a religious belief, the timing of the test is unimportant.

Patient Attitudes/Behaviors - Patient Empowerment:

Many KIs described the Sri Lanka patient population as being reluctant to disagree with health professionals or to request additions to the free health system, and they think that such patient empowerment is necessary for any large-scale changes to be made in the legal system or in the free health system. Another informant described the typical Sri Lankan personality as being reluctant to hear bad news, making patients unlikely to request antenatal genetics services in case the services return a truth they aren't prepared to face.

Table 12: Sociocultural Context

Knowledge	
724A	And then again, the patient awareness and the awareness among healthcare workers about available screening, tests,

	strategies for noticing aneuploidy and also structural aneuploidy is also very, very poor.
64B	So they usually ask, "is there any problem?" if we are asking that they have these scans. They get terrified. Why are we asking? But then we explain that other countries, like other developed countries, European countries, across the board everybody is having this kind of scan. Everyone should know what it is….not only high-risk patients.
Religion	
619B	There is no big trend in this country for prenatal diagnosis. According to Buddhism if you terminate pregnancy at…whatever period of gestation, that is killing.
612B	Everyone thinks that because this is a Buddhist country it's difficult to change. But always it's not the Buddhist monk who are against this thing. It's the Christian church.
711A	My impression, even with them, is that they are more interested [in termination] if they are Buddhist. The Catholic families, I think they are left feeling shocked about it, and I think that together they are not willing to talk about it. If they are alone with me, if only one of them is there, and would probably consider [termination]. But [in] the body language that they indicate is that they are not interested in talking about a termination of a pregnancy, [indicating that] "God gives my child, I will take whatever God gives."
619A	Surprisingly I've found that the Christians and the Catholics, Buddhists to a lesser degree, in one of those papers [that the KI had authored regarding views toward termination], and Hindus, even lesser…they were for prenatal diagnosis and termination of pregnancy, because there was a direct question in that, if your child was having prenatal diagnosis and shows up as a Down syndrome, will you terminate? The majority of Catholics, the majority of Christians, a fair amount of Buddhists, and a fair amount of Hindus…the Islamic cohort was a little smaller, but they were also close to the Christians and Catholics.
Patient Attitudes/Behaviors – Patient Empowerment	
616A	There might come [a time] when they [patients] will also say, "you know, why not?" But still in our situation, the patients' rights groups, the kind of lobbying power or whatever, it's not strong. Whereas in the West it's far more developed. Whereas here the patients still accept that okay, the care provider knows best, he will do what is best for me. It's a very prescriptive type of mentality.
724A	Our people are not broad-minded. They are very narrow-minded… It's difficult when you convey something and ask them to all come, come up, come out and then ask for it. They will never do it. Therefore as long as they don't do it,

	this [the push to change laws] will never develop into anything.
711A	But they do know. They have heard through maybe the newspapers or the TV, about antenatal testing. So they, they're familiar with the concept. But I think most live along with the Sri Lankan personality… They just don't want to face the truth. They're not willing to think through the consequences. They'd rather sort of wait and hope for the best.

3.3.3 Alternatives to Scaling Up Antenatal Genetic Services

Key Informants brought up a number of alternatives to scaling up antenatal genetics services that might be better situated to the Sri Lankan context. One healthcare administrator against making genetic testing services freely available across the country suggested that certain government facilities could serve as centers of excellence (616A). Another administrator felt that the current system could be made stronger if the government adds to and more effectively utilizes field staff (64A). One healthcare provider also felt that strengthening the current system should be considered rather than introducing new services. He noted the importance of prevention and called for improved understanding of which aspects of the routine program are not being implemented effectively or adhered to by pregnant women (715A).

A molecular geneticist (619B) suggested that it would be more cost-effective for the government to cover the cost of curative or therapeutic interventions for children born with certain conditions rather than implementing large-scale screening programs. Another molecular geneticist remarked that other specialized technologies could minimize congenital disorders and might be more acceptable to Sri Lankan society (619A). A cytogeneticist thought along similar lines, suggesting that pre-implantation diagnosis, if the technology could be developed, might be more acceptable given the religious and legal context (711A). The informant affiliated with a Down syndrome support organization spoke to the need for increasing governmental support for

individuals with congenital disorders, regardless of whether or not screening and testing are scaled up (612E).

Table 13: Proposed Alternatives to Scaling Up Antenatal Genetic Services

Key Informant	Summary of Suggested Alternative	Key Informant Comments
616A	Establish Centers of Excellence where specialized services are available and providers are adequately trained.	We can't set it up everywhere; we shouldn't have these scans everywhere. Some center, we have these other institutions in like catchment areas, send to that center. And the specialists there will do that. That's how I think it should work. Not spending money, throwing money everywhere, because there happens to be a specialist there who wants this particular service…. We have to work together. We share our resources, and that's the best possible way of doing it.
64A	Better utilize field staff to deliver counseling related to congenital abnormalities.	Overburdened, the routine program will carry on but the final things like, to go and counsel about congenital abnormalities, and those things, cannot be done when you are overburdened with work, so now it's… the government is thinking of making some policy changes to recruit more PHMs and also to distribute them equally.
715A	Improve implementation of components of routine antenatal care.	You are going to surveille [sic] the birth defects, but prevention is more important than surveillance, I would like to see… in the post-natal clinic what they now know. Because they have gone on to the [antenatal] clinics and now, what they know about these things [basic prevention practices]. Because we all know that folic acid has to be taken a couple of months prior to the pregnancy but I'm sure if you ask 100 mothers, more than 50% did not have it.

619B	Incorporate curative/therapeutic interventions rather than implement large-scale screening programs.	My line of thinking for thalassemia was, thalassemia is a curable disease if you give them a bone marrow transplant. I advocated for establishment of bone marrow transplant services in Sri Lanka…. I always advocate for… doing prenatal diagnosis with a view to saving a baby rather than other side.
619A	Offer alternative, less controversial means of minimizing the risks of genetic abnormalities.	Sperm banks and ovum banks, things like that… might have a little bit of help to people. Even in consanguine[ous marriages], ovum donation or sperm donation might, or some people who are carriers of thalassemia…maybe a sperm donor can [prevent the birth of a child with an abnormality].
711A	Develop pre-implantation genetic diagnosis as a more acceptable "preventative" measure.	The other thing is to go back, rather than [to] prenatal diagnosis… is to really develop the technology and the expertise to… do the pre-implantation diagnosis. Which is the way most families [with history of genetic disease] probably would prefer to go ahead … Again, there are few patients who can keep up with the cost [of pre-implantation diagnosis] in this country. But that is maybe really a more acceptable alternative for Buddhist families…., the technologies may develop so that you could have better prevention or maybe management.
612E	Increase support for and acceptance of individuals with Down syndrome.	I think the government has to get involved more in these things. An organization like us cannot [do it all]…one thing is we don't have the capacity to do [things] island-wide. We won't be able to spend that kind of money. The government should play a role [in supporting individuals with Down syndrome].

4. Discussion

This exploratory qualitative study provides insight into current antenatal care practices and the impact of congenital disorders in Sri Lanka.

First, our results indicate that among field staff, administrators, and specialized providers there is uniform awareness and understanding about which services are part of the routine antenatal care package delivered to all women in the free health system (Table 5). However, there is limited awareness about antenatal screening and diagnostic services available beyond the basic care package. While participants confirmed that antenatal screening and diagnostic services are to some extent available, they drew attention to inequity in access to such specialized antenatal services. Antenatal screening and diagnostic procedures are performed within the private sector and primarily for high-risk women. Even within the high-risk population, access to services varies across socioeconomic levels and geographic regions.

That specialized services are utilized disproportionately by the wealthier members of a society is the very concern that was raised by a 2002 WHO report addressing the role of genomics in health systems worldwide [42]. While the report referred more broadly to the growing disparity that Singer and Daar [43] call a 'genomics divide' between developed and developing countries, it stressed that disparities grow within countries as well [42, 44]. Inequitable access to antenatal genetic services leads to inequitable distribution of psychological distress over the outcomes of

pregnancy and of the financial burdens of caring for children with congenital disorders [33]. This may ultimately contribute to, or at the very least sustain, the disparities that persist in Sri Lanka's maternal and child health indicators.

A second, but highly related finding of our study is that there is a severe shortage of the human resources that could serve to reduce the current inequity in access to specialized services. There are too few skilled technicians to offer antenatal screening and diagnostics services to a larger portion of the population. Furthermore, the nation lacks the genetic counseling infrastructure to deliver the results of screens and tests in a manner that promotes informed decision-making. Pre- and post-test counseling is left to consultants who are too busy to spend sufficient time with patients and have received no special training to communicate the results of genetic tests. Patients are left with great uncertainty, attempting to interpret for themselves reports that are highly specialized or turning to PHMs for interpretation. Unfortunately, PHMs themselves are not familiar with the antenatal screens and diagnostic services available.

Further compromising informed decision-making is that neither PHMs nor patients want to discuss possible complications during pregnancy because they fear that discussing the possibilities will make them realities. However, despite their fears, our study found PHMs eager to learn about congenital disorders and the technologies that detect them. While utilizing PHMs to provide antenatal genetics counseling does not seem viable with PHM knowledge of congenital disorders as it currently stands,

education modules on the most common disorders and procedures available to detect them would not only be well-received by PHMs, but it might also improve knowledge enough that this valuable workforce could be utilized to improve patient capacity to make informed decisions. At the very least, education modules would improve PHMs' knowledge of which women should be referred for genetic services and what those women can expect of such services. In other countries, midwives have undergone training to provide counseling on the results of antenatal screening tests [45]. In Sri Lanka itself, Vithana et al. recently demonstrated that simple, community-based training programs geared to specific conditions or activities lead to significant improvements PHM knowledge, attitudes, and practices [46].

Third, participants in our study confirmed that information on the prevalence of congenital disorders is limited. This stands in the way of understanding the impact of specific conditions and the possible benefits of early detection. Guidelines published for the newly piloted National Birth Defects Surveillance Program published since our study took place indicate that the system will collect information on both structural and functional birth defects. The structural conditions include spina bifida, cleft lip, heart defects, club foot, Achondroplasia, and Down syndrome, and the functional conditions include developmental disabilities (cerebral palsy), metabolic diseases (phenylketonuria), and hematologic disease (including thalassemia) [47]. As the project grows and data accumulate, policymakers will be better able understand the relative

burdens of each condition and prioritize scaling up methods of screening and or diagnosis that can detect them.

Data from the program may also inform future attempts to change abortion laws. Building evidence on the true prevalence of specific disorders in other countries has in the past given leverage to those attempting to change abortion laws. In Iran, thorough analysis of the prevalence of beta-thalassemia led the country to amend its abortion laws, largely to reduce the financial and social burden of the condition on the health system and families [33]. Alternatively, if data indicate that the conditions screening and diagnostic services aim to detect do not contribute substantially to the overall public health burden, funds and human resources can be better allocated to the conditions that are substantial burdens.

Fourth, many KIs expressed views that antenatal screening and diagnostic services should be made more widely available but identified significant contextual factors limiting their availability and uptake, in addition to the aforementioned human resource challenges. The most prominent factor is the restrictive abortion law, but we find the legal issues difficult to disentangle from financial and sociocultural factors. All have come together to create what one key informant called a "problematic ambivalence" among providers and health administrators about the current situation of antenatal screening and diagnosis of congenital disorders. With laws prohibiting except when a mother's life is threatened, the free health system has little reason to devote its

limited financial resources to a screen or diagnostic test that provides results about fetal well-being. Government officials and health administrators accept that the services are available in the private sector for those who desire them. Consultant obstetricians must act under financial and logistical constraints in government hospitals to determine which women need additional services, but they are free to discuss non-routine services and refer patients on for services they feel necessary. Religious opposition has stalled legal change, and the Sri Lanka patient population does not demonstrate strong initiative to demand policy change or request additional services from the free health system. Interestingly, though our participants attribute much of the resistance to genetics services and termination of pregnancy to religious beliefs, they also suggest that a person's religious views may not be reflected in his or her individual actions.

Finally, though non-invasive prenatal testing plays only a minor role in this report, the details of its availability in Sri Lanka were not known prior to this study. Our participants confirmed that NIPT is available but not widely known or utilized. Other studies have suggested that NIPT might be better suited to the contextual factors working against more traditional methods of screening for chromosomal abnormalities: performing the simple blood draw required for NIPT may be more feasible than training health workers to perform invasive screening and diagnostic procedures [23]. We found if we were to take into account only the material and human resource scarcities in Sri Lanka, this might be the case. However, we find that with respect to the most significant

contextual factors, NIPT would fare no better. First and foremost, NIPT implementation would face the same legal opposition as all other methods of antenatal screening and diagnosis. Furthermore, if the conditions most burdensome in Sri Lanka are not among the few that can be reliably detected by NIPT, there is less incentive to build capacity to deliver it. And while eventually the cost of NIPT may drop, the price of the test is currently prohibitive for much of Sri Lanka's population.

4.1 Implications for policy

In answering our questions about antenatal care and congenital disorders, KIs and PHMs provided valuable insights into issues that have public health and policy implications. They suggest a need for policy-makers and health providers to assess current patterns of healthcare delivery and understand how they contribute to health disparity.

One issue that demands attention is the shift of a growing proportion of medical care from the public to the private sector in recent decades. According to Thresia et al., this shift means that larger number of services are financed through out-of-pocket payments, exacerbating economic inequality and contributing to medical impoverishment [13]. Another notable issue is that with respect to antenatal care personnel, there is significant overlap between the public and private sectors. The participants in our study highlighted the dual role of VOGs (consultant obstetricians) in public hospitals and private practices. Their view is backed by other reports that

government health workers are becoming more and more attracted to private sector consultation services [48]. Providers' desire to have a private-sector appointment in addition to a public role may change the ways providers interact with patients in public versus private settings; PHMs reported that mothers feel they receive more attention in the public sector during delivery if they have seen a consultant in his or her private clinic earlier in pregnancy. Russell et al. found similar views in their 2005 study of treatment-seeking behaviors in two urban poor communities in Colombo: people reported that they not only saved time but also built better relationships with doctors in the private sector [21]. If the treatment discrepancies perceived by patients perceive are real, individuals who lack the means to see doctors in their private sector appointments are disadvantaged in their satisfaction with, and possibly in the quality of, the care they receive.

Another issue illuminated by our participants is that the current abortion policy has different implications for different population subsets. Abortion is widely practiced across socioeconomic groups, but individuals of lower socioeconomic status cannot pay for safe abortion services and suffer disproportionately from complications and sepsis. What has developed is a stratification of reproductive practices. "Stratified reproduction" is a concept labeled by bioethicists Shellee Colen, Rayna Rapp, and Faye Ginsburg that refers to a hierarchical organization of reproductive health rights and child rearing capabilities that ultimately gives some women more reproductive

autonomy than others [49]. Dorothy Roberts recently applied the concept to describe the current situation in the United States, where affluent women have access to advanced reproductive technologies that allow them rule out the possibility for their children to have certain genetic features [50]. In Sri Lanka, too, affluent women are the ones who have access to antenatal genetic services and specialized reproductive technologies. They are also the ones who have access tp safe abortion services they learn of a fetal abnormality. With the provision of testing and the provision of abortion stratified, the likelihood of bearing a child with a congenital disorder is also stratified. Those under the most economic strain are those most likely to face the additional strain of caring for a child who requires additional heath resources and developmental support.

Finally, our participants highlighted shortcomings in policy and healthcare provision, shortcomings primarily related to the recognition of and support for individuals affected by congenital disorders. Though our research does not answer the question of whether scaling up antenatal screening and diagnostic services is currently feasible or appropriate for Sri Lanka to address the burden of congenital disorders, it does shed light on actions and services that are currently feasible and appropriate. There was nearly universal agreement among participants that the government should increase its support for individuals with congenital disorders. There was also strong agreement that consultant obstetricians should be trained to better recognize potential abnormalities during routine ultrasound scans. Such training might inform their referral

practices and increase the detection of conditions for which antenatal interventions could improve health outcomes. Despite finding baseline ultrasound skills lacking among obstetric service providers, Dias et al. found that relatively short training significantly improved machine-setting and fetal biometry skills [19]. For Down syndrome in particular, improving knowledge among healthcare providers of proper management practices might improve health outcomes; a recent study by Senanayake et al. found that screening for complications of Down syndrome is often delayed and the use of appropriate growth charts is low, both insufficiencies in management that may negatively impact a child's survival and quality of life [51].

4.2 Implications for further research

Prior to this study it was not generally known that NIPT was offered in Sri Lanka, and the perspectives of key informants suggest that NIPT will only be useful if its detection capabilities improved and if its cost decreased. In countries with institutionalized free health systems where expense is a major determinant of service availability, cost-benefit analyses have proven useful. In the United Kingdom, Morris et al. took a model-based approach to estimate the costs and outcomes associated with offering NIPT within the national Down syndrome screening program [52]. Australia undertook a similar study using decision-tree methodology to compare the costs of the screening system currently in place to one that included NIPT within first-trimester screening [44]. Moving from models into practice, another project is ongoing in the UK

to evaluate the limited implementation of NIPT as a screen in state-funded hospitals; study implementers hope that their findings will inform policy decisions and guide the development of protocols for appropriate reporting and counseling [53]. Analyses like these are not feasible in Sri Lanka given its resource limitations and the limited role of the antenatal genetic screens that NIPT would replace or supplement. However, as the aforementioned studies continue and report their results, policy-makers in countries like Sri Lanka, where NIPT is on the horizon, may look to them for guidance on situating NIPT within their own health systems.

While healthcare providers, administrators, and genetics experts provided valuable insights into the knowledge and attitudes toward antenatal screening and diagnosis, an important next step is to gain the perspectives patients themselves: current and future parents. What do they want to know about their pregnancies? Do they feel they have adequate resources during pregnancy, and are their most important questions answered? How do they view children with congenital disorders and what are the primary considerations of raising a child with a disorder?

Some research has explored the attitudes of Sri Lankan health professionals and of parents of children with Down syndrome and found that both stakeholder groups are open to screening for Down syndrome, not for the chance to consider termination but for the chance to prepare for life with a special needs child [28, 29, 54, 55]. However, there has been little research on these topics conducted recently, and additional work is

necessary to understand how views might have changed over the past decade in light of marked socioeconomic development, the end of a decades-long civil war, and ever-increasing access to technology.

Further inquiry into reproductive trends is also necessary to understand how the impact of congenital disorders may change as Sri Lanka continues to rise in its level of development. Our participants spoke little of how reproductive practices are changing, but evidence suggests that in countries moving through an epidemiological transition, as maternal age increases and babies who would have died in the past now survive, congenital disorders rise in prevalence [56]. Demographic analyses that aim to characterize trends in reproductive patterns and forecast changes in the prevalence of congenital disorders may provide useful information to those creating health policy and guiding service provision.

4.3 Study strengths and limitations

This study has several limitations. First, its geographic specificity limits the ability to generalize the views of informants and PHMs to the nation as a whole. While there is some variability among MOH areas in the Galle District (in population average income, religious and ethnic composition, and distance to tertiary health facilities), it is not representative of the variability found nationwide, with other districts facing different health burdens and healthcare access constraints. For instance, among residents of districts near the center of the island, access to care is limited by extremely

mountainous terrain. Galle is comprised overwhelmingly of Sinhala people, the ethnic majority, but districts in Northern and Central Sri Lanka are comprised almost completely of Tamil people, an ethnic minority. Adding to our results' geographic specificity is the fact that a pilot project for birth defects surveillance was operating in the Southern province at the time of the study. Procedures for collecting data on congenital disorders are likely to differ between the Southern Province and other provinces where the program has not been implemented. However, the fact that PHMs were unaware of the practicalities of reporting even in the pilot district suggests that there is limited awareness of the program among PHMs nationwide.

Our participant recruitment and data collection strategies may also limit our findings. Focus groups were conducted in Sinhala, transcribed, and translated into English by one of two research assistants. Though the RAs were fluent in English and their translations were checked for accuracy, we cannot rule out that nuances or alternative meanings were lost in translation. Through convenience and minimal snowball sampling we obtained small sample sizes for certain stakeholder groups of key informants. We are confident that we achieved data saturation in the health administrator group, but we may have been able to tease out richer data from the health provider group if we had been able to recruit additional providers. That said, some of the difficulty in recruiting specialized maternal health providers reflects the scarcity of these providers in Sri Lanka overall and supports one of our key findings: a limited

health workforce is a significant factor limiting greater implementation and uptake of genetic screening and diagnostic technologies.

5. Conclusion

This study provides information on both the feasibility and utility of increasing the scope and spread of services for the antenatal detection of congenital disorders in Sri Lanka. Key stakeholders in healthcare policy, antenatal care delivery, and genetic services suggest that making antenatal genetics services part of routine care in the public health system is not feasible within the current financial, legal, and social context. Yet they also suggest that current patterns of availability and utilization contribute to regional and economic disparities. They restrict access to wealthy individuals in proximity to major health centers in the Southern Province. Encouragingly, the National Birth Defects Surveillance Program will soon provide higher quality information on the impact of specific disorders. This information may provide the evidence needed to improve equitable access to antenatal genetic services within the public health system.

It is clear that even before the program returns information on specific disorders in the next few years, key constituents in Sri Lanka can take action now to bring continued gains and reduced disparities in maternal and child health. The improvements won't be achieved by stasis. Our participants reported that understanding of congenital disorders and means to detect those disorders is low among patients and healthcare providers. They also reported that support for individuals with congenital disorders is inadequate and difficult to access. Public and private health policy-makers can examine their guidelines for patient referral to ensure

every woman receives the care appropriate for her risk level regardless of her socioeconomic and geographic position. Lobbyists calling for change in abortion policy can strengthen their arguments by examining the policy's disparate consequences for the rich and the poor and its contribution to maternal mortality and morbidity. The government can augment the services it provides to support individuals and families affected by congenital disorders and clarify the processes through which services can be accessed.

Appendix A: Map of MOH Areas, Galle District

Source: co-PI Dr. Amila Chandrasiri, own work.

Appendix B: Questions asked during interviews and focus group discussions

Core Questions for Key Informant Interviews
For Health Administrators
1. Please describe your role in maternal healthcare.
2. What is included in the routine system of antenatal care in Sri Lanka?
3. What are your policies/programs related to antenatal care?
4. What role does antenatal screening and testing for congenital abnormalities play in the routine system of care?
5. Do your patients request prenatal genetic screening and testing services?
6. Is data collected on the incidence of congenital disorders, fetal abnormalities, and neonatal mortality?
7. Are you familiar with non-invasive prenatal testing?
8. Do you think that NIPT would be useful in Sri Lanka?
9. What do you see as the major barriers to scaling up antenatal screening and testing for congenital abnormalities in Sri Lanka?
For Healthcare Providers
1. Please describe your role in maternal healthcare delivery.
2. Tell me about prenatal care in Sri Lanka.
3.Do you currently offer prenatal genetic screening or testing to your patients?
4.Do you inform your patients about different options for prenatal genetic screening and testing?
5. Do your patients request prenatal genetic services?
6. Who is responsible for explaining the results of screens and tests to patients?
7. Are you familiar with non-invasive prenatal testing?
8. Would you say your patients want to know as much as possible about their pregnancies?
9. If NIPT were to become available, would you offer it to your patients?
10. Is data collected on the incidence of congenital disorders, fetal abnormalities, and neonatal mortality?
11. What do you see as the major barriers to scaling up antenatal screening and testing for congenital abnormalities in Sri Lanka?
For Genetics/Genomics Experts
1. Please describe your background in genetic technology.
2. What genetic testing services do you perform/does your facility offer?
3. Do you see a need for additional screening/testing services?
4. Are any of the screens/tests you offer occur during the antenatal period?
5. Tell me about the people who utilize your services.
6. Do people understand the screens and tests you offer?

7. Does your facility incorporate counseling services into the delivery of test results?
8. Are you familiar with non-invasive prenatal testing?
9. Do you think that NIPT would be useful in Sri Lanka?
10. What are/were the barriers to introducing new genetic and genomic technologies in Sri Lanka?
Note: While interviews were partially guided by these questions, additional probes and questions were utilized to promote the flow of conversation during interviews and to extract additional information as deemed pertinent.

Core Questions for Focus Group Discussions
1. How many years you have been practicing, and in what aspects of antenatal care are you involved?
2. How far into a woman's pregnancy does your first visit take place?
3. How many times do you see a woman during her pregnancy?
4. What conditions are you most concerned about during a woman's pregnancy?
5. What conditions are women most concerned about?
6. Do you see any women who you think are at higher risk for genetic conditions?
7. What genetic conditions are you aware of?
8. What genetic conditions have you most frequently seen in your experience?
9. What genetic conditions, if any, have women asked you about?
10. If a child is born with an abnormality (Down syndrome, a birth defect, etc.), do you report this outcome to someone?
11. When I say the words "antenatal testing ", what is the first thing that comes into your mind?
12. Where do you get information about antenatal genetic testing?
13. Generally, how much do you think women know about antenatal genetic testing?
14. In your experience, have any women asked you about specific genetic tests? If so, which ones have they asked you about?
15. Tell me about your recent experiences discussing prenatal genetic testing with your patients.
16. If you discuss it, what do you find difficult about discussing/explaining antenatal genetic testing, if anything?
17. What additional resources would better equip you to discuss prenatal genetic testing?
18. Are there any other issues that you would like to raise regarding prenatal genetic testing?
Note: While FGDs were guided by these questions, additional probes and questions were utilized to promote the flow of conversation and to extract additional information as deemed pertinent.

Appendix C: High-risk conditions in pregnancy

(from "Maternal Care Package: A Guide to Field Healthcare Workers", pages 41-45)

High-risk conditions related to past obstetric history:

- Previous stillbirth
- History of 2 or more consecutive 1st trimester miscarriages
- History of preterm delivery before 37 weeks, or 2nd trimester miscarriage
- History of PIH (pregnancy-induced hypertension), eclampsia/pre-eclampsia
- History of antepartum hemorrhage (APH) or postpartum hemorrhage (PPH)
- History of retained placenta
- History of cesarean section/Myomectomy
- History of surgery on reproductive tract, including removal of septum, cone biopsy, and large loop excision of transformational zones (LLETZ)
- History of a birth with birth weight < 2500 g or > 3500 g
- History of Intrauterine growth restriction (IUGR)
- History of feeding difficulties/breast surgery

High-risk conditions related to present pregnancy:

- Maternal age: < 20 years or > 35 years
- Height < 145 cm
- BMI: < 18.5 kg/$m^{2,}$, 25-29.9 kg/m^2, or > 30 kg/m^2
- Primigravida
- Parity (5 or more)
- Multiple pregnancy
- Diastolic blood pressure (DBP) > 90 mmHg
- DBP > 110 mmHg or systolic blood pressure (SBP) > 160 mmHG
- Vaginal bleeding (any time)
- Maternal jaundice
- Severe maternal anemia (Hb% < 7 g/dL)
- Maternal malaria
- Maternal syphilis or maternal HIV
- Uncertain dates/irregular periods
- Intrauterine growth restriction (IUGR)
- Malpresentations and abnormal lie after 34 weeks

Other risk conditions:

- Diabetes mellitus/Liver disease and renal failures
- Rheumatic and congenital heart disease
- Epilepsy
- Mental disorders
- Asthma
- Rh negative mother
- Subfertility

High-risk conditions related to social problems:

- Unmarried mothers/widows
- Geographically marginalized and other social factors

References

1. The World Bank. World Development Indicators - Table 4.1: Growth of output. Wasington, DC: World Bank, 2015. Retrieved from http://wdi.worldbank.org/table/4.1#. Accessed October 14, 2015.

2. Wijesinha A. Sri Lanka's Middle-Income Transition: Thinking Beyond the "Optics", to the "Mechanics". Talking Economics website. http://www.ips.lk/talkingeconomics/2013/10/19/sri-lankas-middle-income-transition-thinking-beyond-the-optics-to-the-mechanics/. Published October 19, 2013. Accessed October 30, 2015.

3. United Nations Sri Lanka. Sri Lanka Millennium Development Goals Country Report - 2014. http://www.lk.undp.org/content/dam/srilanka/docs/mdg/MDG-Country-Report-2014.pdf. Accessed November 15, 2015.

4. Family Health Bureau (FHB). National Strategic Plan: Maternal and Newborn Health (2012-2016). Colombo: Ministry of Health Sri Lanka, 2011. http://fhb.health.gov.lk/web/index.php?option=com_content&view=article&id=175:maternal-and-child-health-policy-framework-directives&catid=10&Itemid=120&lang=en#national-policy-on-maternal-and-child-health. Accessed October 3, 2014.

5. World Health Organization (WHO). Community Genetics Services, Report of a WHO Consultation on community genetics in low- and middle-income countries. Geneva, Switzerland: World Health Organization, 2011. http://www.who.int/genomics/publications/reports/en/. Accessed October 3, 2016.

6. WHO. Provisional agenda item 11.7. In: Birth defects: report by the Secretariat, in Sixty-third World Health Assembly, Geneva, 17-21 May 2010. 2010, World Health Organization: Geneva. http://apps.who.int/gb/ebwha/pdf_files/wha63/a63_10-en.pdf. Accessed January 4, 2016.

7. Christianson A, Modell B. Medical genetics in developing countries. Annu Rev Genomics Hum Genet, 2004; 5:219-65. doi: 10.1146/annurev.genom.5.061903.175935

8. de Silva DC. Genetic Variation and Genetic Epidemiology of Sri Lanka. In: Genomics and health in the developing world, D. Kumar, Editor. Oxford, UK; Oxford University Press, 2012:984-998.

9. Institute for Health Metrics and Evaluation (IHME). The Global Burden of Disease: Generating Evidence, Guiding Policy. Seattle, WA: Institute for Health Metrics and Evaluation, Human Development Network, The World Bank, 2013. http://www.healthdata.org/sites/default/files/files/data_for_download/2013/WorldBank_SouthAsia/IHME_GBD_WorldBank_South%20Asia_FullReport.pdf. Accessed September

30, 2014.

10. Haththotuwa, R. et al. Models of care that have reduced maternal mortality and morbidity in Sri Lanka. Int J Gynaecol Obstet, 2012. 119 Suppl 1: p. S45-9. http://www.ijgo.org/article/S0020-7292(12)00157-9/abstract. Accessed September 23, 2014.

11. Mudiyanse RM, Perera A, Bandara D. Thalassemia in Sri Lanka. In: 1st Pan-Asian Conference on Haemoglobinopathies, A. Maggio, Editor. Bangkok, Thailand; PAGEPress Publications, 2012: 4.

12. Department of Census and Statistics (DCS), Household Income and Expenditure Survey 2012/13 Final Report. Colombo; Ministry of Policy Planning, Economic Affairs, Child, Youth, and Cultural Affairs, 2015: 89. http://www.statistics.gov.lk/HIES/HIES2012_13FinalReport.pdf. Accessed January 15, 2016.

13. Thresia CU. Rising private sector and falling 'good health at low cost': health challenges in China, Sri Lanka, and Indian state of Kerala. Int J Health Serv, 2013 43(1):31-48. doi: http://dx.doi.org/10.2190/HS.43.1.c

14. WHO. Addressing noncommunicable diseases in a lower-middle-income country: Sri Lanka's approach. Colombo; World Health Organization, Country Office for Sri Lanka, 2013. http://www.searo.who.int/srilanka/documents/WHO_Booklet.pdf. Accessed Sepctember 20, 2014.

15. Weeratunga L. Factors Affecting Demand for Health Care Services in Sri Lanka. Asian Profile, 2010; 38(5):467-481.

16. Goonewardene M, Dias, T. Antenatal care: paradigm changes over the years. Ceylon Med J, 2013; 58(2):47-50. doi: 10.4038/cmj.v58i2.5678

17. Office of Director General of Health Services. Antenatal Care Circular No. DGHS/D/FHB/2010. Colombo; Ministry of Health Sri Lanka, 2014. http://fhb.health.gov.lk/web/index.php?option=com_phocadownload&view=category&download=579:antenatal-care-circular-english&id=2:antenatal-postanatal-care&Itemid=148&lang=en. Accessed January 4, 2016.

18. Benn P, Borell A, Chiu R, et al. Position statement from the Aneuploidy Screening Committee on behalf of the Board of the International Society for Prenatal Diagnosis. Prenat Diagn, 2013; 33(7):622-9. doi: 10.1002/pd.4139

19. Dias T, Ruwanpura L. Systematic introduction of obstetric ultrasound skills into practice. 2012. Sri Lanka J Obstet Gynaecol, 2011; (33):154-157. doi: 10.4038/sljog.v33i4.4803

20. Dias T, Cooper D, Fernando A, et al. Knowledge of Down's syndrome Screening amongst Patients and Health Care Professionals in Sri Lanka. Gynecol Obstet (Sunnyvale), 2014; 4(7):234. doi:10.4172/2161-0932.1000234

21. Russell S. Treatment-seeking behaviour in urban Sri Lanka: Trusting the state, trusting private providers. Soc Sci Med, 2004; 61(7):1396-1407. doi:10.1016/j.socscimed.2004.11.077

22. Dassanayake RS, Mahadevan K, Gunawardene YI. Development of single-strand conformational polymorphism to screen for mutations in hotspot regions of beta-globin gene of beta-thalassemia patients of Sri Lanka. Southeast Asian J Trop Med Public Health, 2013; 44(1): p. 114-23. http://www.tm.mahidol.ac.th/seameo/2013-44-1-full/16-5574-12.pdf. Accessed September 24, 2014.

23. Chandrasekharan S, Minear MA, Hung A, Allyse M. Noninvasive prenatal testing goes global. Sci Transl Med, 2014; 6(231): 231fs15. doi: 10.1126/scitranslmed.3008704

24. Daley R, Hill M, Chitty LS. Non-invasive prenatal diagnosis: progress and potential. Arch Dis Child Fetal Neonatal Ed, 2014; 99(5): F426-f430. doi: 10.1136/archdischild-2013-304828

25. Papasavva T., et al. Next generation sequencing of SNPs for non-invasive prenatal diagnosis: challenges and feasibility as illustrated by an application to beta-thalassaemia. Eur J Hum Genet, 2013; 21(12): 1403-10. doi: 10.1038/ejhg.2013.47

26. Ohno M, Caughey, A. The role of noninvasive prenatal testing as a diagnostic versus a screening tool - a cost-effectiveness analysis. Prenat Diagn, 2013; 33(7): 630-635. doi: 10.1002/pd.4156

27. Simpson, B. Negotiating the Therapeutic Gap: Prenatal Diagnostics and Termination of Pregnancy in Sri Lanka. J Bioeth Inq, 2007; 4(3):207-215. doi: 10.1007/s11673-007-9070-5

28. Simpson, B, Dissanayake VH, Jayasekara RW. Contemplating choice: attitudes towards intervening in human reproduction in Sri Lanka. New Genet Soc, 2005; 24(1):99-117. doi: 10.1080/14636770500037859

29. Simpson B, Dissanayake VH, Wickramasinghe D, Jayasekara RW. Prenatal testing and pregnancy termination in Sri Lanka: views of medical students and doctors. Ceylon Med J, 2003; 48(4):129-32.

30. de Silva D. Emerging issues in genetic testing in Sri Lanka. Indian J Med Ethics, 2007; 4(3):133-4. http://www.ncbi.nlm.nih.gov/pubmed/18624146. Accessed July 7, 2015.

31. Kumar RMSM. Abortion in Sri Lanka: The Double Standard. Am J Public Health, 2013; 103(3):400-404. doi: 10.2105/AJPH.2012.301154

32. Dissanayake VHW. Medical Genetics and Genomics in Sri Lanka. In: Genomics and health in the developing world, D. Kumar, Editor. Oxford, UK; Oxford University Press, 2012: 953-963.

33. Ballantyne, A. Prenatal diagnosis and abortion for congenital abnormalities: is it ethical to provide one without the other? Am J Bioeth, 2009; 9(8): 48-56. doi: 10.1080/15265160902984996.

34. Atapattu H. Sequel of Unsafe Abortion. In: Legislation of termination of pregnancy - Are we ready? 45th Annual Scientific Sessions of the Sri Lanka College of Obstetricians and Gynaecologists, Abstracts. Colombo; SLCOG, 2012:31. http://www.slcog.lk/sljog/Abstracts%202012/SLCOG%20ABSTRACTS%202012.pdf. Accessed July 8, 2015.

35. Arambepola C, Rajapaksa LC. Decision making on unsafe abortions in Sri Lanka: a case-control study. Reprod Health, 2014; 11:91. doi: 10.1186/1742-4755-11-91

36. DCS. Births Deaths, Marriages and Infant and Maternal Death Rate - 1999 to 2013. Colombo: Department of Census and Statistics, 2013. http://www.statistics.gov.lk/PopHouSat/VitalStatistics/Indicators/Table%201.pdf. Accessed September 21, 2016.

37. Perera H. Proposed changes to the law of abortion. 45th Annual Scientific Sessions of the Sri Lanka College of Obstetricians and Gynaecologists, Abstracts. Colombo; SLCOG, 2012:32. http://www.slcog.lk/sljog/Abstracts%202012/SLCOG%20ABSTRACTS%202012.pdf. Accessed July 8, 2015.

38. Central Intelligence Agency. The World Factbook: Sri Lanka. CIA website, 2014. https://http://www.cia.gov/library/publications/the-world-factbook/geos/ce.html. Accessed October 3, 2015.

39. DCS. Census of Population and Housing Final Report - Southern Province. 2012, Colombo; Ministry of Finance and Planning, 2012. http://www.statistics.gov.lk/PopHouSat/CPH2011/index.php?fileName=Southern&gp=Activities&tpl=3. Accessed October 2, 2015.

40. Corbin J, Strauss A. Chapter 4: Strategies for Qualitative Data Analysis. In: Basics of Qualitative Research: Techniques and Procedures for Developing Grounded Theory. Thousand Oaks, CA: SAGE Publications, Inc.; 2008:65-87.

41. Mayring P. Einführung in die qualitative Sozialforschung [Introduction to Qualitative Research]. 5th ed. Weinheim, Germany: Beltz-UTB; 2002.

42. WHO. Genomics and World Health: Report of the Advisory Committee on Health Research. Geneva; World Health Organization, 2002. www.who.int/rpc/genomics_report.pdf. Accessed September 30, 2014.

43. Singer PA, Daar AS. Harnessing genomics and biotechnology to improve global health equity. Science, 2001; 294(5540): 87-9. doi:10.1126/science.1062633

44. Ventura W, Nazario-Redondo C, Sekizawa A. Non-invasive prenatal diagnosis from the perspective of a low-resource country. Int J Gynecol Obstet, 2013; 122(3): 270-273. doi: 10.1016/j.ijgo.2013.03.031

45. Smith DK, Shaw RW, Slack J, Marteau TM. Training obstetricians and midwives to present screening tests: Evaluation of two brief interventions. Prenat Diagn, 1995; 15(4): 317-324. doi: 10.1002/pd.1970150404

46. Vithana PV, Ariyaratne MA, Jayawardana PL. Effectiveness of an educational intervention among public health midwives on breast cancer early detection in the district of Gampaha, Sri Lanka. Asian Pac J Cancer Prev, 2015; 16(1): 227-32.

47. FHB. Guidelines on Birth Defects Surveillance Pilot Implementation – Southern province. Colombo: Family Health Bureau, Ministry of Health; 2014. http://fhb.health.gov.lk/web/index.php?option=com_phocadownload&view=category&id=18:maternal-child-morbidity-mortality-surveillance&Itemid=149&lang=en#. Published August 11, 2015. Accessed January 5, 2016.

48. Silva KT. Social determinants of health: Lessons from Sri Lanka.. In: Social Determinants of Health: Assessing Theory, Policy and Practice, Bhattacharya, Editor. London: Orient BlackSwan, 2010.

49. Rapp R. Gender, body, biomedicine: How some feminist concerns dragged reproduction to the center of social theory. Medical Anthropology Quarterly, 200; 15(4): 466-77. doi: 10.1525/maq.2001.15.4.466

50. Roberts, D. Privatization and Punishment in the New Era of Reprogenetics". Faculty Scholarship, 2005; Paper 579. http://scholarship.law.upenn.edu/faculty_scholarship/579. Accessed January 17, 2016.

51. Senanayake MP, Jayasinghe JA. Adherence to preventive health care in children and young adults with Down syndrome (DS) in Sri Lanka. Ceylon Med J, 2014; 59(1): 13-5. doi: 10.4038/cmj.v59i1.6733

52. Morris KS, Chung N, Hill M, Chitty LS. Model-based analysis of costs and outcomes of non-invasive prenatal testing for Down's syndrome using cell free fetal DNA in the UK National Health Service. PLoS ONE, 2014; 9(4): e93559-e93559. doi: 10.1371/journal.pone.0093559

53. Hill M, Wright D, Daley R, et al. Evaluation of non-invasive prenatal testing (NIPT) for aneuploidy in an NHS setting: a reliable accurate prenatal non-invasive diagnosis (RAPID) protocol. BMC Pregnancy Childbirth, 2014. 14: 229. doi: 10.1186/1471-2393-14-229

54. Simpson B. On Parrots and Thorns: Sri Lankan Perspective on Genetics, Science and Personhood. Health Care Anal, 2007; 15(1): 41-9. doi: 10.1007/s10728-006-0036-2

55. de Silva DC, Jayawardana P, Hapangama A, et al. Attitudes toward prenatal diagnosis and termination of pregnancy for genetic disorders among healthcare workers in a selected setting in Sri Lanka. Pren Diagn, 2008; 28(8): 715-721. doi: 10.1002/pd.2021.

56. Weatherall D. Foreword II. In: Genomics and Health in the Developing World, D. Kumar, Editor. Oxford, United Kingdom: Oxford University Press; 2012.