Birth Defects Report

Jan 2008-Sept 2014

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ABBREVIATIONS

CVS	Cardiovascular system
CNS	Central Nervous System
CRS	Congenital Rubella Syndrome
EPI	Extended programme for immunization
ICD10-CM	Tenth Revision of the International Classification of Diseases clinical modification
IGMH	Indira Gandhi Memorial Hospital, Male
LMIC	Low and middle income countries
LBW	Low Birth Weight
MDHS	Maldives Demographic and Health Survey
MDG	Millennium Development Goal
MOD	March of Dimes
MPHD	Ministry of Planning and Housing Development, Maldives
NGO	Non-Governmental organizations
NICU	Neonatal intensive care unit
NTC	National Thalassemia Centre
NTP	National Thalassemia Programme
NTD	Neural tube defects
SEAR	WHO South-East Asia Region
SHE	Society for Health Education
UNICEF	United Nations Children's Fund
WHO	World Health Organization

EXECUTIVE SUMMARY

Congenital anomalies are defined as abnormalities of body structure or function that are present at birth and are of prenatal origin [1]. Synonymous terms that are often used are “birth defects”, “congenital anomalies” and “congenital malformations [2]. The increased awareness of mortality by birth defects on infant and under-five mortality rates have led to a global movement in identifying, raising awareness and instituting preventive measures.

This short term consultancy for reporting and analysing the birth defect register will provide information on the prevalence of sentinel defects in a hospital based setting and identify the maternal risk factors, associations and deficient areas. The birth defect register maintained in Neonatal intensive care unit(NICU) in Indira Gandhi Memorial Hospital (IGMH) since 2008 has accumulated data of birth defects cases admitted to the NICU; this includes both intramural and extramural cases.

This study identifies birth defects requiring hospital care and trained neonatal support. Data from the register was extracted and categorized with 316 cases identified from Jan 2008-Sept 2014. Comparative analysis and prevalence rates are presented, however due to deficient data for maternal risk factors no statistical relations could be established from this present study.

Annual rates	The birth prevalence of birth defects in IGMH is between 11.9-20.5 per 1000 live births, a noticeable rise was seen in 2010 and 2011.
Birth Defects	The most prevalent defects are Cardiac and circulatory system anomalies (26%). The most common condition reported is Atrial Septal defect. Single body system involved in 73% of cases. Second commonest is central nervous system defects
Gender	Male predominance with 56%
Demography	Increased number of cases seen in Haa Alif, Dhaalu, Noonu and Aliff Aliff atoll when compared to the population numbers.

Maternal age	Birth defects highest in mothers of advanced maternal age with 13.4 per 1000 live births for >40years.
Gestational age	74% seen in Term infants (37-40 weeks).
Birth weight	27.7% of babies with birth defects have low birth weight. Higher than the national average of 10.8% (2009-2011).
Method of delivery	Number of Caesarean sections done on mothers of infants with birth defects increased from 47.3% in 2008 to 71.1% in 2014.
Mortality	Mortality (per 1000 live births) due to birth defects is varied with 4.1 in 2008 to 0.6 in 2013. Highest mortality is seen associated with Cardiac and Central nervous system disorders.

INTRODUCTION

The importance of birth defects and genetic inherited disorders has come to light as Infant mortality rates (IMR) decline with improving primary health care and decreased mortality due to communicable diseases. The March of Dimes (MOD) Global Report on Birth Defects, [2] reports worldwide 7.9 million births occur annually with serious birth defects and 94% of these births occur in the middle and low income countries.

In an effort to decrease the number of congenital anomalies worldwide, the Sixty-third World Health Assembly adopted a Birth defects resolution. As one of its objectives, this resolution encourages countries to build in-country capacity related to the prevention of congenital anomalies and to raise awareness about their effects [5].

Maldives, within the region have done well with various mortality indicators with under-five mortality11; infant mortality rate 9; neonatal mortality rate 7[2].The WHO - Birth defects in South East Asia- Situation analysis reports 10.7% of neonatal deaths in Maldives are attributed to birth defects[4].

The health care delivery system of Maldives is organized into a four tier referral system with the island level health facilities (primary health care) referring patients to higher level facilities in the atolls regions and to central level(tertiary care)[3]. Perinatal care is provided at two main hospitals in Male' the capital, Indira Gandhi Memorial Hospital (IGMH) is a government hospital; 36-42% (2008-2011) of births in the country takes place in IGMH [6]. The neonatal unit in IGMH was upgraded in 2006 from a 5 bedded nursery care to a level 2 NICU. It is 20 bedded with trained neonatal nurses providing care around the clock. Facilities are in place for ventilation, CPAP, full monitoring and minor paediatric surgical procedures. ADK- a private hospital; had basic nursery care and upgraded their NICU in 2013 (4 beds and ability to ventilate). Their new NICU inaugurated in august 2014 is 8 bedded with trained neonatal staff, full monitoring and ventilation facilities.

No previous study of birth defects has been done before in Maldives, and this report will be instrumental in the development of a nationwide birth defects surveillance programme. It will provide a basic understanding of the burden and impact birth defects have on the local health system and population.

1 BIRTH DEFECTS REGISTER.

The birth defect register under study is based in the Neonatal intensive care unit (NICU) at IGMH and is a hospital based study. It was initiated in January 2008 and continues till date. No reporting or analysis of this data has been undertaken before. This register captures some maternal details, birth and outcomes of infants born with birth defects. Intramural (born in IGMH) and extramural (born outside the hospital/referred) who were admitted in the NICU of IGMH are included.

Diagnosis of birth defects was based on clinical evaluation, radiographic examination, ultrasonography and echocardiography. Chromosomal analysis of the newborn whenever recommended was sent to a speciality lab in Mumbai,India for confirmation.

2 OBJECTIVES

- Analyse and report data from Birth Defects register based in the Neonatal Care unit in Indira Gandhi Memorial Hospital from January 2008- Sept 2014.
- Identify prevalence and outcomes of infants born with birth defects
- Identify high risk groups.
- Provide data to develop public health programs and help health care professionals for use in planning and implementing surveillance and awareness programmes

3 DATA COLLECTION, METHOD OF ANALYSIS AND CASE DEFINITION

Data collection was prospective and was manually abstracted from maternal and baby records when a case was identified. Data was entered by nursing Staff into the register when a case was diagnosed and the outcomes at the end.

To improve the quality and accuracy of data- births and deaths for the hospital were taken from hospital, Vital Registration system and the delivery register maintained in the birthing unit at IGMH. All infants with reportable and confirmed birth defects are included; however data for terminations and still birth due to birth defects was not collected in this register and hence not included in this study. From a total of 346 cases 30 were excluded as many were birth traumas and others had inconclusive diagnoses. Data for 316 cases from the register was classified annually and entered into an electronic data base on Microsoft Excel 2010 manually. Factors like maternal age, parity, gestational age and birth weight were divided into strata and data allocated into these strata. Microsoft Excel functions were used to filter and compare these results

Case:	A case is an individual infant with one or more birth defects. A case with more than one birth defect condition may be counted in two or more categories of birth defects.
Rate:	A rate is used to adjust for differences in population sizes.
Birth Prevalence:	Birth defects are a prevalence measure as the at-risk population foetuses with defects/spontaneous abortions (denominator) cannot be identified and hence an incidence cannot be calculated. Prevalence is useful since it allows comparison between populations of different sizes. It is usually presented as per 1000 or 100000 live births.
Proportions:	In a proportion, the cases in the numerator must be included in the denominator. Percentage is a proportion multiplied by 100.
Counts:	Counts present the simple enumeration of cases.

4 LIMITATIONS

As a hospital based study; only those requiring immediate care have been captured in this group, the birth defect register does not include a significant number who have non-life threatening birth defects, within the hospital or in the atolls and islands. The lack of details on data about terminations and still births deters the analysis as a true prevalence and mortality rates cannot be calculated based on this. Due to deficient maternal information especially maternal risk factors no causative relations could be analysed in this study.

Quality and reliability of data is questionable in some cases as there is failure to update changes or confirmation to a diagnosis after initial entry into the register at admission or probable diagnosis, e.g. karyotype confirmation of infants with Trisomy 21,

The missing data for neonatal admissions (2008, 2009 and 2013) and deaths (2008, 2009) is a serious setback as the true burden of neonates with birth defects on the neonatal population within the health facility cannot be generated.

5 BIRTH DEFECTS ANALYSIS (Jan 2008- Sept 2014)

5.1 Birth Prevalence

The birth prevalence of intramural birth defects in IGMH from 2008-2014 shows birth prevalence ranges between 11.9 - 20 per 1000 live births. An increase in 2010 and 2011 was noted with no cause for this increase yet identifiable. Extramural cases (16) have not been included in the birth prevalence. The birth prevalence in IGMH is expected to be high as this is the main tertiary centre and mothers with that of antenatal diagnosis of birth defects who are referred for obstetric care and delivery to the centre. This only gives a prevalence of sentinel birth defects born in IGMH.

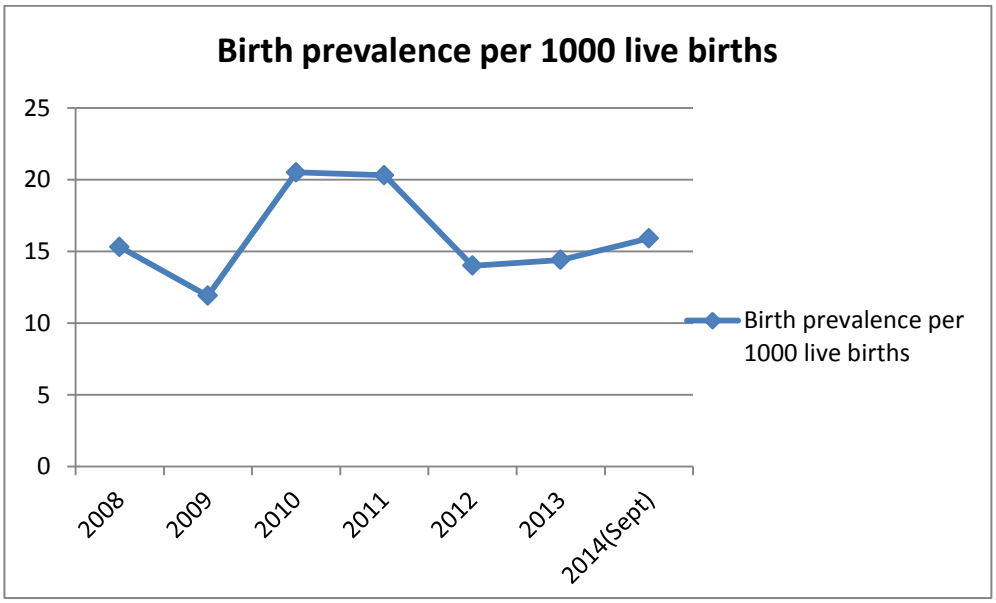
Table 5-1-Birth defects- Birth Prevalence per 1000 live births for Jan 2008-Sept 2014

Year	Live Births IGMH*	Birth Defects Intramural**	Birth prevalence per 1000 live births
2008	2418	37	15.3
2009	3011	36	11.9
2010	2679	55	20.5
2011	2605	53	20.3
2012	2838	40	14.0
2013	3035	44	14.4
2014(Sept)	2136	34	15.9

*Data from delivery register in IGMH

** Data from NICU based Birth defect register Jan 2008- Sept 2014

Figure 5-1 Birth defects- Birth prevalence per 1000 live births for Jan 2008- Sept 2014



5.2 Birth defects – Prevalence in NICU admissions.

The annual rate of NICU admissions receiving care with birth defects shows an average of 7.1% for 2010, 2011 and 2012. Both intramural and extramural cases have been included. The proportion of birth defects in the first 9 months of 2014 is 7.7%, however as this is only partial data it has not been included to calculate an average. Data for NICU admissions for 2008, 2009 and 2013 was not available.

Table 5-2 Percentages of Birth defects in infants for NICU admissions

Year	NICU admissions	Intramural	Extramural	Total cases N	Percentage %
2008	na	30	**7**	**37**	na
2009	na	36	**2**	**38**	na
2010	885	55	**7**	**62**	7.0
2011	734	53	**3**	**56**	7.6
2012	621	40	**3**	**43**	6.9
2013	na	44	**1**	**45**	na
2014(sept)	451	34	**1**	**35**	7.7

na- not available. N=316

5.3 Birth defects- Systemic classification

Both intramural and extramural cases have been included and conditions broadly classified into body systems. Standard coding is not done while entering information in the Birth Defects Register at NICU, and therefore not available for this analysis. The diagnosis is made by the treating medical team/doctor and entered in the birth defect register by NICU staff. A total of 408 anomalies were detected in 316 cases from Jan 2008- Sept 2014.

The graph and table below summarises the birth defects categorised into the different systems identified in the ICD 10-CM.

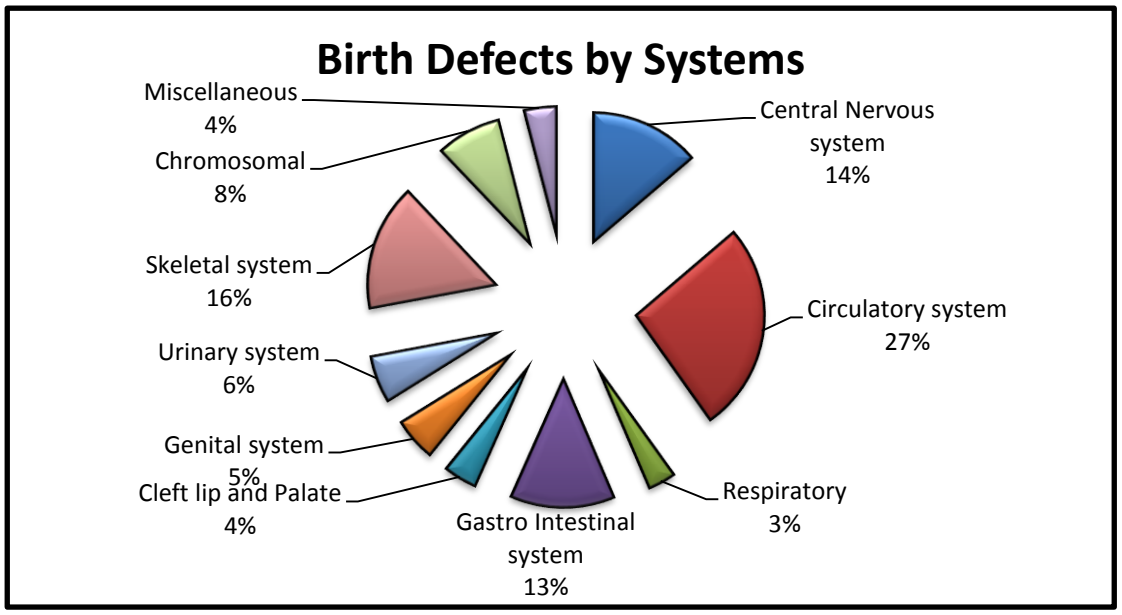


Figure 5-2 Birth defects by body systems

Table 5-3 Birth defects and Prevalence by Body systems Jan 2008-Sept 2014

Congenital Malformations	2008	2009	2010	2011	2012	2013	2014	Total	
								N	%
Central Nervous system	8	7	6	11	8	9	7	56	13.7
Circulatory system	15	14	26	14	17	15	7	108	26.4
Respiratory	1	0	2	5	2	1	3	14	3.4
Gastro Intestinal system	7	6	7	9	12	5	7	53	12.9
Cleft lip and Palate	2	1	4	2	2	4	2	17	4.1
Genital system	3	2	6	1	5	3	2	22	5.3
Urinary system	3	3	5	4	4	2	3	24	5.8
Skeletal system	3	5	8	16	9	15	9	65	15.9
Chromosomal	9	5	4	6	5	2	2	33	8
Miscellaneous	2	2	6	0	2	3	1	16	3.9

N= number of defects (408)

Cardiac and Circulatory disorders has the highest proportion (26.4%). Among these acyanotic heart conditions ASD-9%, VSD-5.2% are the most common. However there are a significant number of congenital heart defects which do not have a definitive diagnosis or details unrecorded during their stay in hospital. Cardiac defects includes Patent ductus arteriosis (7%) which can be a common physiological condition in preterm infants[7].

Intestinal obstruction involves both proximal and distal conditions including atresia's (excluding trachea-oesophageal fistula and pyloric stenosis). These accounted for 7.8% of the defects diagnosed.

Hydrocephalus births represent 5.9% of defects; However a full classification was not possible i.e. Obstructed/unobstructed or with or without Spina bifida as this was not clear from the register.

5.0% of births were reported as trisomy 21, however there is no record if these infants were confirmed by karyotyping.

Inherited metabolic and haemoglobinopathies like Thalassemia and G6PD are not recorded in the register and hence not included as part of this study.

Table 5.4 shows all the cases divided into ICD 10-CM based classification. Unclassified and multiple anomalies have been included into unspecified birth defects.

Birth defects Systemic Division Jan 2008-Sept 2014				
Congenital Anomalies of the Central Nervous System			**Congenital Anomalies of Urinary System**	
Anencephaly	3		Renal Agenesis/Hypoplasia	5
Spina Bifida	12		Polycystic kidney disease	2
Hydrocephalus	25		Obstructive Genitourinary Defects	11
Encephalocele	3		Not identified- hydronephrosis	8
Microcephalus	5			
			Congenital Anomalies of Genital system	
Congenital Anomalies of the Circulatory System			Hypospadiasis	4
Double oulet right ventricle	2		Ambiguous Genitalia	6
Transposition of Great Vessels	4		Congenital Hydrocele	5
Tetralogy of Fallot	0		Undescended Testes	6
Ventricular Septal Defect	22			
Atrial Septal Defect	37		**Congenital Anomalies of the Musculoskeletal System**	
Pulmonary Valve Atresia and Stenosis	5		Congenital Talipes equinovarus(CTEV)	11
Hypoplastic Left Heart Syndrome	1		Limb Deformities	14
Patent Ductus Arteriosis	29		Gastroschisis/Omphalocele	1
Coarctation of Aorta	4		Congenital Hip Dislocation	5
Pulmonary Artery Anomalies	1		Diaphragmatic Hernia	8
Other- Dextrocardia, Cardiomyopathy	12		Polydactyl/Syndactyl	5
Not Classified- Lesion not recorded	33		Achondroplasia	3
			Others -	16
Congenital Anomalies of the Respiratory System			**Chromosomal Anomalies**	
Lung Agenesis/Hypoplasia	7		Trisomy 13	1
Choanal Atresia	1		Trisomy 21-Down Syndrome	21
Congenital cyctic adenomatoid malformation	1		Trisomy 18	1
Others	5		Others- Pierre Robin, Turners, Multiple defects	3
Cleft Palate and Cleft Lip			Other and Unspecified Congenital Anomalies	16
Cleft Palate	1			
Cleft Lip w and w/o Cleft Palate	16			
Congenital Anomalies of the Alimentary Canal				
Esophageal Atresia/ Tracheoesophageal Fistula	14			
Intestinal Obstruction and atresia	33			
Pyloric Stenosis	1			
Hirshsprung's Disease (congenital megacolon)	1			
Others- mesentric cyst, glossal cyst, hernia	3			

Table 5-4 Birth defect case classification into body systems Jan 2008-Sept 2014

5.4 Birth defects by Gender Jan 2008-Sept 2014.

Sex differences in the prevalence of specific human birth defects are common, and male infants are at greater risk for birth defects than female infants [8]

Our analysis portrays the above finding with a predominance of male infants born with birth defects. 56 % were male and 42% were female. Ambiguous genitalia were noted in 6 babies(<1%), There were also differences among broad categories and specific birth defects by gender as some conditions are exclusive to males eg- congenital hydrocele, hypospadiasis.

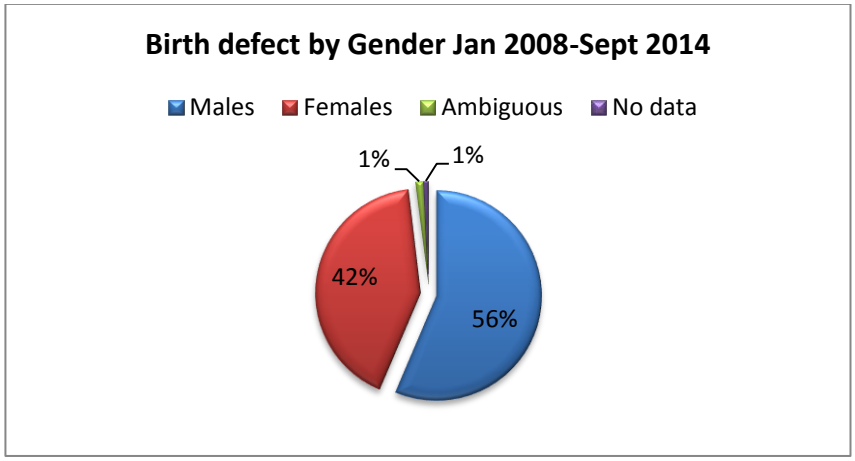


Figure 5-3 Birth Defects by Gender

5.5 Birth defects by affected systems- Jan 2008-Sept 2014

Each case may have one or more defects and each defect is assigned to its respective body system, Figure 5-4 below shows number of systems involved. 75% have a single system affected while 25 % have 2 or more anomalous systems/anomalies identified.

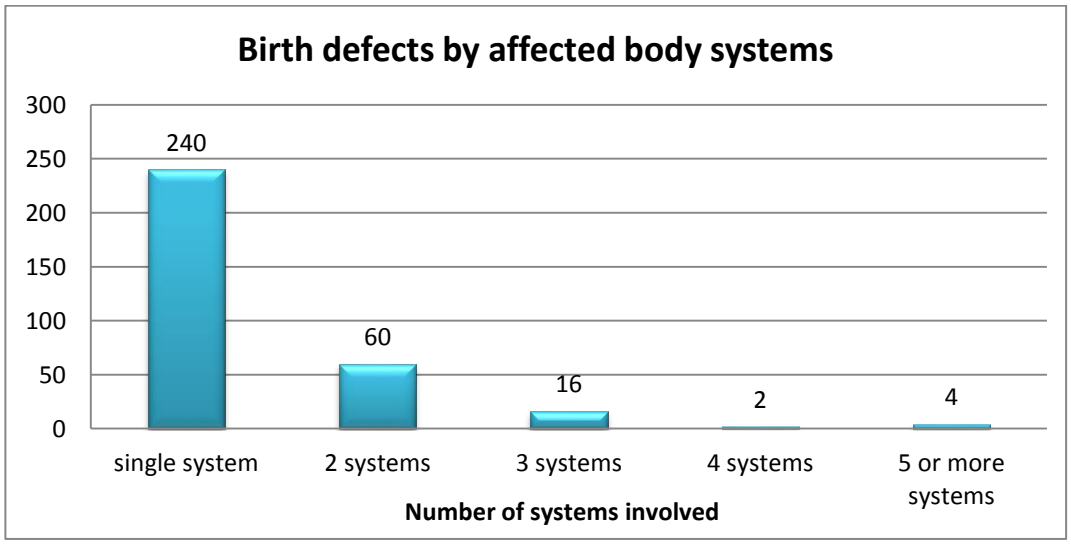


Figure 5-4 Birth Defects by affected body systems

5.6 Birth defects by Demography Jan2008-Sept 2014

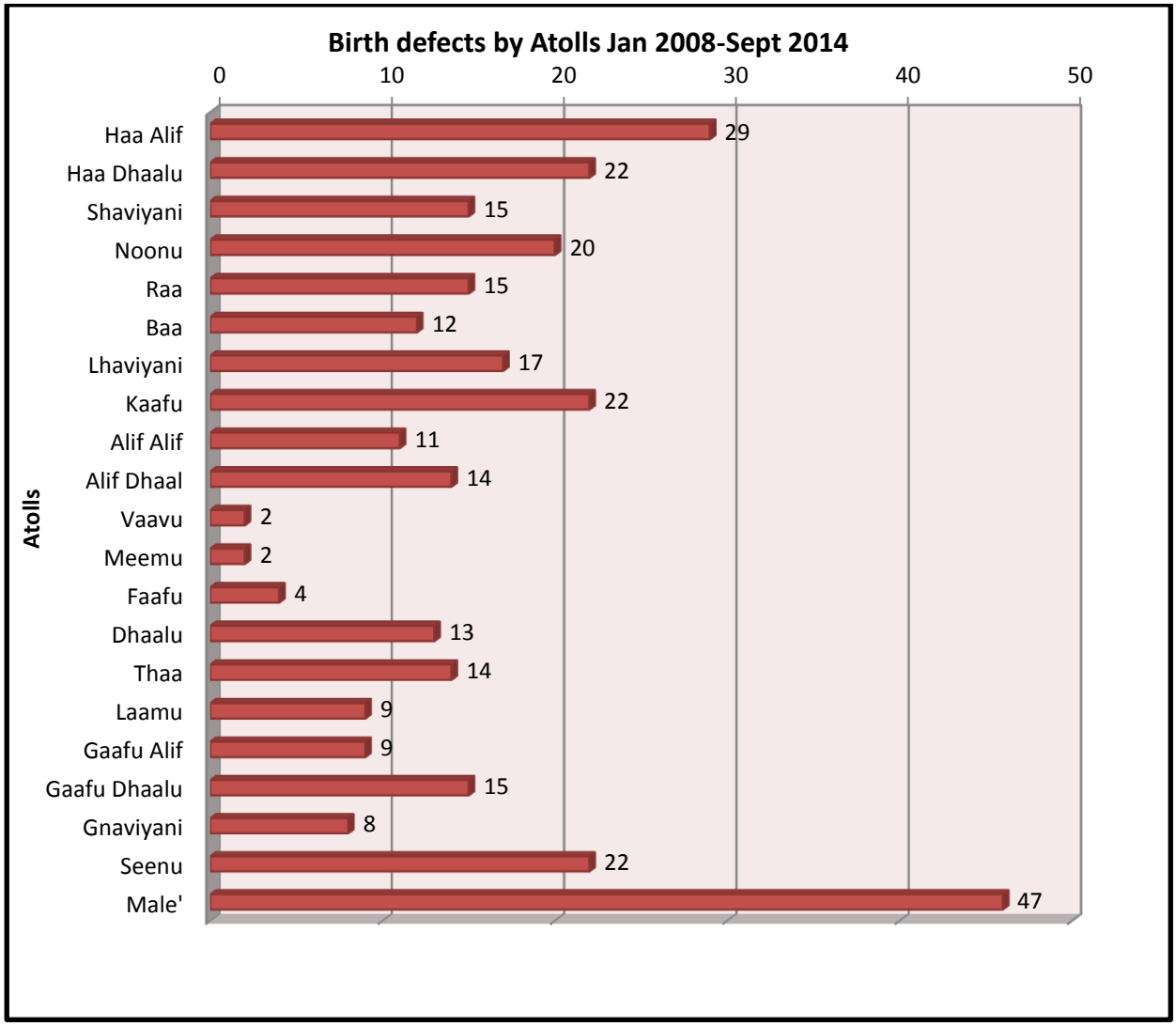
There are 20 atolls in total in the Maldives. The capital city Malé and islands Villingili and Hulhumale are considered a separate region and contain one-third of the total population of the country [9]. Demography details were obtained from the address provided in maternal notes for place of permanent residence. Atoll-based analysis (table 5-5) was done as sample size limitations prevent analysis by smaller geographical units.

As transport services have improved with regular ferries and air transfers available, the number of mothers going to deliver out of their catchment area to the regional hospitals and especially to Male' is high and no mechanism in place to identify this moving population. Male' has a large emigrant population from atolls and islands who have taken up residence there and nearby inhabited islands.

Table 5-5 Proportion of Birth Defect cases- Atoll distribution- Jan 2008- Sept 2014

Atoll	Birth Defects	Proportion %	Live births 2008-2013	Population
Haa Alifu	29	9.0	1106	13,495
Haa Daalu	22	6.8	3934	16,237
Shaviyani	15	4.6	508	11,940
Noonu	20	6.2	548	10,015
Raa	15	4.6	1905	14,756
Baa	12	3.7	724	9,578
Laviyani	17	5.2	1036	9,190
Kaafu	22	6.8	220	15,441
Aliff Aliff	11	3.4	240	5,776
Aliff Dhaalu	14	4.3	626	8,379
Vaavu	2	0.6	40	1,606
Meemu	2	0.6	238	4,710
Faafu	4	1.2	274	3,765
Dhaalu	13	4.0	397	4,967
Thaa	14	4.3	350	8,493
Laamu	9	2.8	1455	11,990
Gaafu Aliff	9	2.8	267	8,262
Gaafu Daalu	15	4.6	1598	11,013
Gnaviyani	8	2.4	1159	7,636
Seenu	22	6.8	1985	18,026
Male	47	14.6	23560	103,693

Figure 5-5 Total number of Birth Defects by atolls -Jan 2008- September 2014



Higher case numbers are seen in highly populated atolls/islands like Male', Haa Alif, Haadhaalu, Gaafu Daalu and Seenu atolls. 9 out of 29 cases (31%) in Haa Alif atoll prevail from Ihavandhoo. In Haadhaalu- 9 out of 22 (40%) cases are reported from Kulhudhoofushi which bases the regional hospital for the northern atolls. In the southern atolls, Hithdhoo in Seenu atoll has 9(40%) out of 22 cases and Maradhoo 6 (27%)cases. Hithadhoo island houses the regional hospital for the southern atolls. Notable atolls when compared to the population and livebirths with higher numbers of birth defects are Aliff Aliff, Noonu, Dhaalu

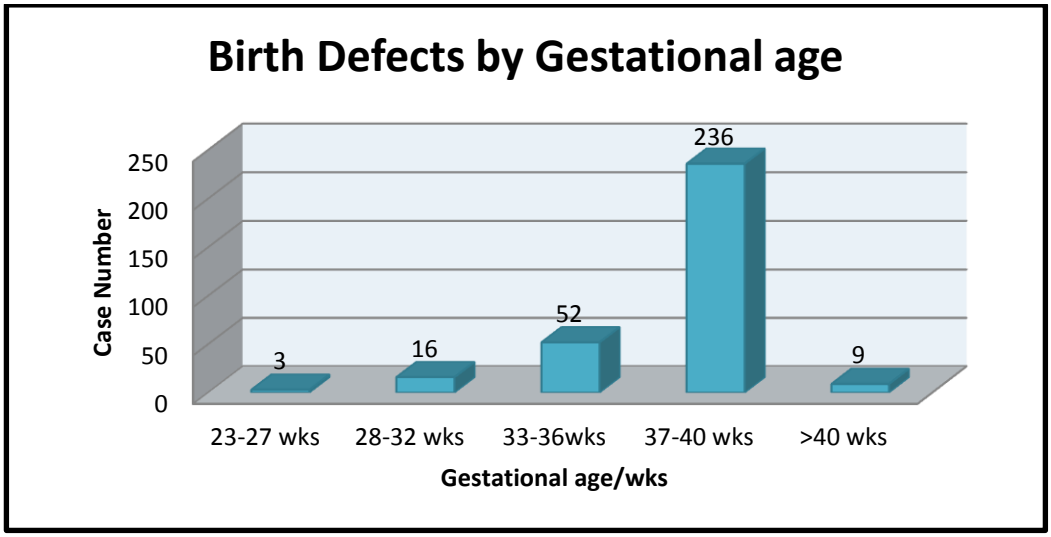
Complete prevalence data can be obtained only from a population based Birth Defects Surveillance system or a population based research or study. Accurate details of the place of residence and live birth numbers of the atolls and island populations taking into consideration the moving population is essential to create a demographic prevalence.

5.7 Birth defects by Gestational age Jan 2008-Sept 2014.

73.5% (236) of defects are seen in term babies between 37-40 weeks. 22% (71) are seen in preterm infants.

As term gestations have the highest number of births in the population, this data needs conversion into relative data to identify the true relation of birth defects to gestational age. This data for gestations for all livebirths is not available.

Figure 5-6 Birth defects by Gestational age Jan 2008- Sept 2014



Data not available in 5 cases

5.8 Birth defects by Birth weight Jan 2008-Sept 2014

Infants with birth defects have a higher incidence of having low birth weight (<2500gm). This association of LBW and malformations has been well documented in other studies[11,12].Our study shows 27.7% have a low birth weight and 3.7 % have higher than >4.0kg weight. 69% had a normal birth weight. The average percentage of low birth weight infants for from 2009-2011 in Male' Population is 10.6% and in atolls- 9% (Maldives Health statistics 2012)[6].

Table 5-6 Birh weight distribution in infants with birth defects Jan 2008-Sept 2014

Birth weight		
	Numbers	Percentage%
Low birth weight <2500gm	89	27.7
Normal Birth weight 2500-3999 gm	222	69
High birth weight >4000gm	12	3.7

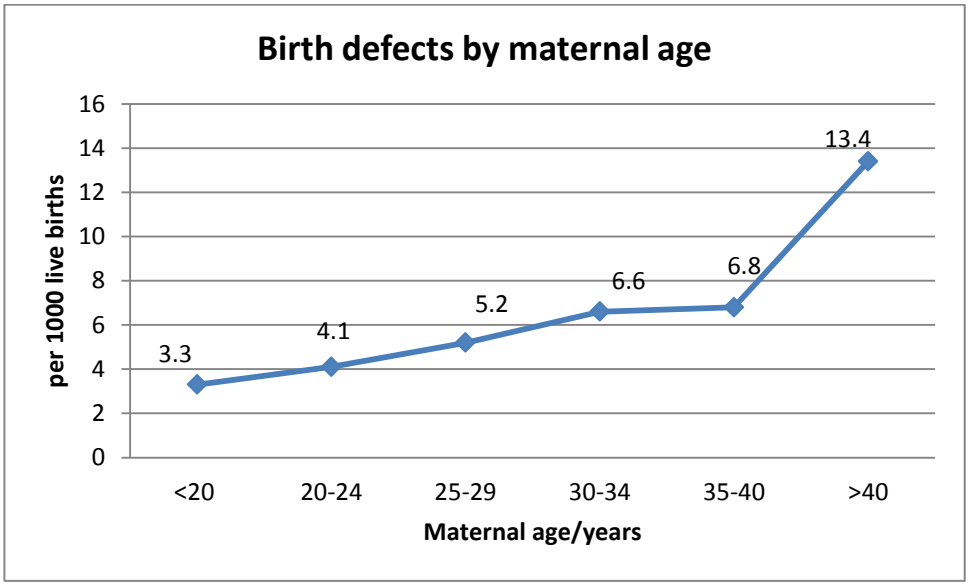
5.9 Birth defects by maternal age

Studies have reported that babies born to those at an advanced age are at increased risk of non-chromosomal abnormality, chromosomal abnormality and congenital anomaly [14] and others include those <20 years with increasing numbers of defects[11] .

In this study maternal age was stratified into sub groups and allocated. Maternal age for live births was available only from 2008-2011. The findings from the study were similar to the above studies with prevalence of birth defects increasing with maternal age.

Individual conditions and their relation to maternal age have not been done as numbers are small and 46 cases do not have maternal age recorded

Figure 5-7 Birth defects by maternal age (Jan 2008- Dec 2011)



Source –Maldives Health Statistics 2009, 2012- Ministry of Health.
No data was available for 46 cases

5.10 Birth defects by method of delivery

The register has limited maternal data and it is not possible to know which of the infants had an antenatal diagnosis and if the method of delivery especially caesarean sections were planned in advance. The indication for caesarean does not have to be due to the birth defect and could be due to other maternal of foetal complications.

However this data is presented due to the change in trend noted over the study time period, a true relation can be obtained only after considering maternal risk factors and indications. In 2008, 52.6 % had normal deliveries and 47.3% mothers whose infants had birth defects had caesarean sections. In 2014 28.2% had normal deliveries and 71.7% of mothers had caesarean sections. The percentage of caesarean sections nationally in 2011 was 41.1% [6].

Figure 5-8 Birth defects by method of delivery Jan 2008- Sept 2014

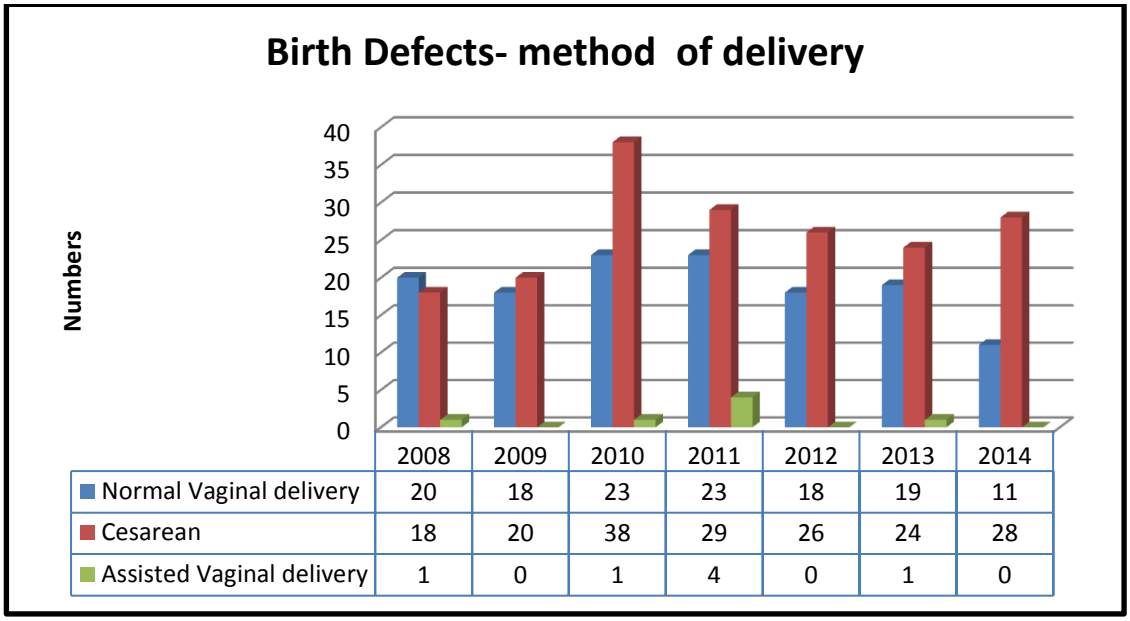


Table 5-7 Prevalence of caesarean sections in mothers of infants born with birth defects

Year	Live Births IGMH	Birth defects with Caesarean Section	Prevalence/ 1000 live births
2008	2418	18	7.4
2009	3011	20	6.6
2010	2679	38	14.1
2011	2605	29	11.1
2012	2838	26	9.1
2013	3035	24	7.9
2014	2136	28	13.1

5.11 Birth defect cases- Outcomes.

Globally an estimated 270 000 newborns die during the first 28 days of life every year from congenital anomalies [1].

The presence of a birth defect does not necessarily indicate that the birth defect is the direct cause of death. Average mortality for infants with birth defects is 2.6 per 1000 live births for 2008- 2014. This only includes live births as still births with birth defects cannot be accounted for.

The table 5-7 shows intramural, extramural and infant deaths per 1000 live births. The proportion of deaths looks at the number of infants who died with birth defects to the number of neonatal deaths due to all causes in IGMH. The mortality due to birth defects on neonatal population is as high as 58.3% in 2011. The proportion and prevalence of deaths are expected to be higher as this is the main referral centre.

Table 5-8 Mortality due to birth defects Jan 2008- Sept 2014

Year	Live Births IGMH	NICU admissions	NICU Deaths	Deaths due to Birth defects	Percentage* %	Mortality due to birth defects **
2008	2418	na	na	10	--	4.1
2009	3011	na	na	6	--	1.9
2010	2679	885	35	8	21	2.9
2011	2605	734	12	7	58.3	2.6
2012	2838	621	20	7	35	2.4
2013	3035	na	12	2	-	0.6
2014(Sept)	2136	451	16	**6**	37.5	2.8

* Percentage = (number of infant deaths with birth defects)/total number of infant deaths in NICU) x100
** Number of deaths per 1000 live births = (infant deaths)/(live births)x1000

49% of infants were transferred to mothers care or discharged directly home(fig 5-10). 36% required speciality care, as the health system could not facilitate specialized investigations or treatment for these conditions, and were referred to speciality centres outside the country. Specific referral details were not recorded in the register and hence not included in this study. Infants who may have died later or at referral centres cannot be accounted for as follow up data of these infants is not available after discharge or referral.

Figure 5-9 Neonatal outcome of cases with Birth defects

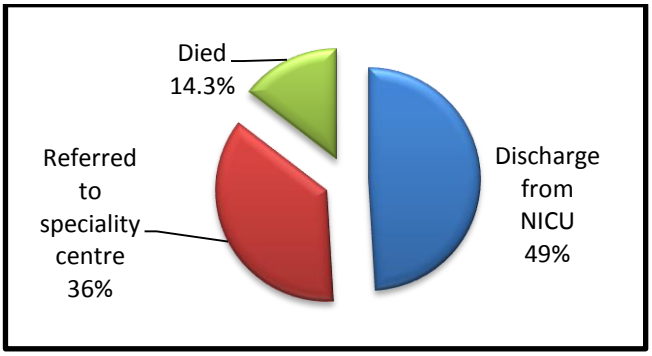


Table 5-8 shows mortality related to the body system involved. As no data on primary cause of death is available for these mortality cases, there is replication in conditions where two or more systems are involved. There are few conditions with multiple anomalies and a definitive diagnosis not reached and classed as a miscellaneous group. The highest mortality was associated with CVS and CNS defects.

Table 5-9 Mortality in relation to body system affected Jan 2008-Sept 2014

Body System	Total defects (N)	No of deaths	Percentage %	Total N=408
Central Nervous system	56	12	21.2	2.9
Circulatory system	108	14	13	3.4
Respiratory system	14	9	64	2.2
Gastro Intestinal system	53	-	-	-
Cleft lip and Palate	17	-	-	-
Genital system	22	-	-	-
Urinary system	24	8	33	1.9
Skeletal system	65	6	9	1.4
Chromosomal	33	4	12	0.9
Miscellaneous	16	6		1.4

6 DISCUSSION

As infant mortality rates decline due to reduction in deaths by infections and other preventable causes, a growing recognition arises of the impact non-communicable diseases, genetic disorders and birth defects have on child mortality, morbidity and its effect on local health economics and the burden to the health system. The diverse demography of Maldives makes deliverance of quality and equal health care a difficult challenge. A significant moving population is created due to unavailability of resources and reduced confidence in available medical services in atolls, which very often prompt travel to regional hospitals and tertiary centres for antenatal, obstetric care and birth.

Maternal factors such as social, education, nutrition, residence and environmental factors have various influences over the aetiology of birth defects. Though consanguineous marriages are uncommon in the Maldives, due to the small populations in some islands (600-1000s), close inter related marriages are seen within small remote and isolated island populations. High case numbers in relation to the atolls population are seen in certain atolls (Haa Alif, Dhaalu, Noonu, and Aliff Aliff). Variations in prevalence of birth defects might be explained by social and racial influences that are commonly known in genetic disorders. The higher number of CVS and CNS disorders in our study is supported by two studies done in Iran[14,15].

NTD are the second most common birth defect identified from this study and a preventable defect in majority of cases. Folic acid supplementation is initiated in antenatal mothers and during breast feeding, however food fortification with folic acid or periconceptional use has still not been established in the Maldives[16]. Folic acid intake among pregnant women increased from 50% in 1999 to 87% in 2004- (RH Survey 2004). No cases of hypothyroidism were reported in this study due to possible diagnosis at later stages or infants not requiring NICU care and hence not entered in the register. The Micronutrient survey in 2007 identified >99% use of iodized salt in households in the Maldives and 1.4% women of reproductive age were found with severe iodine deficiency with the majority of severe and moderately deficient women belonged to Malé region. Among children 6 months to 5 years of age, only 0.7% children were found to be severely deficient with majority belonging to the South central region (Meemu, Faafu, Dhaalu, Thaa and Laamu).

Antenatal care nation-wide is good with 85% having 4 or more antenatal visits [9]. Universal screening for HIV, Hepatitis B and Syphilis is required as part of antenatal work up although reporting gaps have been identified.

TORCH screening has been done for women with history of multiple abortions since 1995 and later expanded in 1998 to include women with only one history of miscarriage or stillbirth. Maldives Rubella/Congenital Rubella syndrome (CRS) project in 2003 by WHO SEARO reported an outbreak of rubella in 2000. However due to failure in notification as rubella was not then included as a notifiable disease and poor CRS surveillance; no cases were identified but suspicion of cases was reported. MMR (Mumps,Measles and Rubella) vaccine at 18months of age was introduced into the EPI programme for immunization in 2007.Measles, Rubella and VDRL are now under the list of notifiable diseases to Health Protection Agency.

Ultrasonogram(USG) facilities are available in all the atoll hospitals and ultrasound scanning are routinely done by the obstetricians, however their expertise in identification of congenital anomalies maybe limited. Foetal anomaly scanning and availability of foetal echocardiography would be most valuable in screening for serious congenital malformations; this would provide a chance of referral to speciality care early and opportunity for consideration of termination if required. An “Abortion Fatwa” issued in 1999 legalised prenatal diagnosis and terminations; it was revised in late 2013 and allows terminations of pregnancy for certain conditions including terminations for thalassemia and serious malformations within 120 days of conception.

The high incidence of Thalassemia- a genetic inherited disorder led to the development of a Thalassemia programme in 1994. In the year 2000, the National Thalassemia programme (NTP) was incorporated into the Health Master Plan and health budget. In November 2012 the thalassemia control act (Act4/2012) was passed which addressed screening, preventive and curative measures. This included mandatory premarital screening, improved diagnostic support and services, finance for medical care to be borne by the government for these individuals and to provide equal opportunities.

The role of non-governmental organizations (NGOs) is highlighted, by Society of Health education (SHE) which took a lead role in identifying the presence of thalassemia in the population and initiating an island-wide Thalassemia screening programme. Thalassemia screening by SHE identified an average beta thalassemia carrier rate of 18% (1 in 5) with significant inter island variations (9 islands with prevalence of 30%, 28 islands with prevalence of more than 25% and 25 islands with less than 5%)[17]. A preliminary cord blood study also indicated an Alpha + thalassemia incidence of 28%[18]. Other haemoglobinopathies identified included HB E Carrier 0.8%, Hb D carrier 0.29%, and Hb S 0.14%[17]. Three types of thalassemia mutations accounted for more than 95% of the cases paving the way to establish a cost effective prenatal diagnostic service [18].

Sickle hotspots identified include AA. Thoddoo: Beta Thalassaemia 17% and Hb S: 8%; GA. Nilandhoo: Beta Thalassaemia 13% and Hb S: 6%; GA. Dhiyadhoo: Beta Thalassaemia 30% and HbS 10%; GDh. Fares: Beta Thalassaemia 11% and Hb S 3% (Source- SHE 1992). Associated with these haemoglobinopathies , a high prevalence of Glucose -6- phosphate dehydrogenase deficiency (G6PD) was also noted.

The National Thalassemia centre (NTC) was established by the government in December 1994 and functions as a day care centre catering for the thalassemia patients and screening services for the public. NTC was renamed as the Centre for Thalassemia and Haemoglobinopathies following the thalassemia act in Nov 2012. The centre functions along with the National blood transfusion services under the Maldives Blood Services (MBS). A DNA lab has been established in SHE in Dec 2005, and two Obstetricians trained in Chorionic villus sampling at CMC, Vellore. The DNA lab services; although a once free service under the NTP now requires out of pocket payment by clients due to funding cuts.

NGO's play a big role in advocating and raising awareness about birth defects and disabilities. Maldivies Thalassaemia Society (MTS) established in 1994, is a parent led support group to improve and enhance quality of lives of Thalassemia patients in Maldives. Tiny Hearts of Maldives, established in 2009 works together with families with children who have congenital heart defects. They provide counselling, parental support groups, organizing fund raisers, Cardiac health camps, assistance for families travelling abroad for further care and advocacy. Beautiful Eyes - Down Syndrome Association founded in 2012 works with families with Down

syndrome. Their work is towards achieving better care and education for these individuals, and public health awareness. They run a training centre to help individuals with developmental skills. Other NGO's like Hemophilia society, Blind and Visually Impaired Association of Maldives, Care Society, Maldives Autism Association, Maldives Deaf Association (MDA) and Maldives Association of Physical Disables (MAPD) are all involved in working with individual disabilities and empowering individuals and creating awareness.

High mortality numbers are noted as the population analysed are sick infants and often have serious and life threatening defects incompatible with life. Follow up information to identify long term outcomes is not available for infants discharged from the unit or following referral to a speciality care centre.

Improvements in care with increased availability of services and resources have contributed to the decline in certain types of referral made to speciality centres. In Jan 2010 the employment of a trained local cardiologist and ability to do echocardiography has helped significantly in diagnosing and monitoring infants with cardiac disorders. NICU staff has been trained in feeding practices to help infants with cleft lip and palate till they are ready for corrective surgery. A number of infants still require travelling abroad for paediatric surgical procedures, certain diagnostic investigations and treatment.

The national health insurance scheme (Aasandha) covers all medical bills of the infant which can extend over a prolonged period or an entire lifetime depending on the birth defect and associated disability. The financial burden on families are high as accommodation and other expenses are borne by the clients, including possible loss of pay for working parents/carers requiring to be away for a prolonged time period. Social difficulties arise when there are other siblings and family issues involved.

This report highlights some of the deficient areas that need to be addressed and the logistical difficulties that are faced by the system and families. It identifies the need for a robust and effective birth defect surveillance programme that detects prevalent birth defects, clusters within islands/atolls and highlight maternal risk factors that could predispose to some of these birth defects identified in the Maldives infant population.

7 RECOMMENDATIONS

1. Development of systems to capture and secure reliable data and maintain this data.
2. Planning and development of a birth defect surveillance programme to detect, establish trends in prevalence of various defects, demographic details and risk factors.
3. Identify high prevalence areas and conduct epidemiological survey to identify possible predisposing agent/event.
4. Utilising the above data to guide in health policy decision making and establishing prevention and public health programmes.
5. Monitoring and strengthening of nutrient supplementation especially folic acid and initiation of food fortification programmes.
6. Maternal information to include social and environmental factors including residence during pregnancy, education, antenatal history, smoking and substance abuse.
7. Plan and establish a newborn screening programme and laboratory support for diagnostics.
8. Establish follow up pathways for infants discharged or referred abroad for diagnosis and care, and information to be maintained in the register.
9. Establish a monitoring system for notifications and reporting of eminently preventable birth defects including NTD, CRS, congenital syphilis, Haemoglobinopathies and hypothyroidism.
10. Health professional training and in identification of common birth defects and establish pathways for notification and referral be easily accessible.
11. Partnerships among policy stakeholders including Governmental and NGO's in taking the programmes to the community

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