Prevalence and patterns of birth defects among newborns in Southwestern Ethiopia: Retrospective study

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Abstract

Background : Intrauterine development can be considered as normal development as well as abnormal development. Abnormal development commonly called birth defects occur because of interference of normal development from genetic disorders, environmental factors and the combination of both genetic and environment factors during the critical period of embryogenesis. The present study aimed to evaluate the prevalence and patterns of birth defects at birth in Southwestern Ethiopia.

Methods : Institutions based cross-sectional study design was used. Six hospitals were selected from the existing hospitals in southwestern Ethiopia purposively based on case load. The study was conducted from September 2011 to December 2015 during which 45,951 deliveries were attained. All records of births that were occurred in the selected hospitals during the study period were identified from medical records, to locate the birth defect records.

Results : Twenty-one different birth defects were recorded. Of these, 49.6 % of the birth defects recorded were Anencephaly and Hydrocephalus. Five types of birth defects namely: Anencephaly (25.0%), Hydrocephalus (24.6%), Spinal bifida (13.1%), Meningomyelocele (7.1%), and Umbilical hernia (4.8 %) accounted about three-fourth (75%) of all recorded birth defects. The present study revealed that the prevalence of the birth defects at birth was found to be 5.5 per 1000 births. Among twenty one birth defects identified in this study, the neural tube defects were the most frequent with prevalence rate of 4.1 per 1000 birth followed by gastrointestinal defects (0.7 per 1000 births)

Conclusions: As compared to other study, the prevalence of birth defect identified in the present study was found to be higher. The neural tube defects were the most prevalent with the frequency of 4.05 cases per 1000. The present study also identified that nearly equal proportions of birth defect occurred among males and females newborns. The majority of the mother who gave birth to birth defects was younger than 35 years old. As the birth defect was found to be a main cases of infant mortality and morbidity, there need to an urgent interventions to control the cases.

Background

Intrauterine development can be considered as normal development as well as abnormal development. Abnormal development occurs because of interference of normal development from genetic disorders, environmental factors and the combination of both genetic and environment factors during the critical period of embryogenesis. These lead to abnormal cytogenesis, histogenesis and morphogenesis with which the neonate born with defect [1, 2]

Congenital anomalies (CA) commonly called birth defect (BD) is structural as well as functional abnormality present at birth and can be described in terms of four clinically significant types based upon the causes, timing, and extent of the developmental interference during prenatal life by teratologic agents [3]. These are malformation, disruption, deformation, and dysplasia [4, 5]. The World Health Organization (WHO) estimates that approximately 260,000 deaths (7% of all neonatal deaths) globally were caused by

BDs in 2004 [6]. It is estimated that the prevalence rates of birth defect is 4.7% in the developed countries, 5.6% in the middle-income countries, and 6.4% in the low-income country [7].

The incidence of CA varies among different ethnic groups [8]. Extensive literature is available on the incidence of CA in First-World countries and studies have also been done in several Third-World countries [8]. However, in Africa south of the Sahara, limited information is available on the incidence of CA, especially in the black population. Previous studies all had limitations in that they were either retrospective, performed on small sample numbers, or reported the frequency of a single abnormality or a few specific abnormalities [1].

BD is emerging as important prenatal problem contributing to the prenatal mortality and morbidity [9]. The difference between the frequency of types of congenital malformation (CM) in different parts of this country and reports from other countries may be due to genetic background and geographic nutritional and socioeconomic differences [9]. CM begins to emerge as one of the major childhood health problems as well as a cause of infant mortality and morbidity throughout the world especially in developing countries [10]. In most cases, infants with malformation do not survive, more than 70% die in the first month of life [11, 12 - 15]

The frequency of BD in the developing countries including Ethiopia is underestimated because of the deficiencies in diagnostic capabilities and lack of reliability of medical records and health statistics. Instead, the available recorded diagnoses in vital health statistics relay on obvious illnesses, rather than on preexisting congenital defects present at birth that increase infant mortality and morbidity leading to the significant community problem [1, 16, 17].

CMs are the fifth leading cause of years of potential life lost and a major cause of morbidity and mortality throughout children of the world [18]. Regardless of their clinical importance there are little studies conducted directly related the prevalence and predisposing risk factors [11, 19 - 22].

Methods

Study setting and period

The study was carried out in six hospitals of three Zones in southwestern Ethiopianamely:Jimma University Specialized, Shanan Gibe, Limmu Genet, Mattu Karl, Agaro and Nekemte hospitals; located in Jimma, Ilu Abba bori and East Wollega zones. The three zones are among the seventeen Zones in the west Oromia Regional State- the largest state in Ethiopia. The study was conducted retrospectively from September 2011 to December 2015. During this period a total of 45,951 deliveries were attended in the hospitals.

Study and Sample Population

The Sampling frame included all deliveries attended in selected hospitals in southwestern region of Ethiopia during the period of September 2011 to December 2015.The sampling population involved all

deliveries with birth defects that included both alive and still birth.

Study Design

Institutions based cross-sectional study design was used. Six hospitals were selected from the available hospitals in southwestern Ethiopia purposively based up on the doses of the cases. All records of births that were occurred in the selected hospitals during the study period were identified from log books and card numbers were used to locate the records. Both client cards and log books were used to retrieve for potential CAs to determine the burden of BD in the hospitals.

For a child born with defects, type of birth defect, socio-demographic variable (age, educational status, income, residence, religion of the mother) and obstetric history (parity, gravida, history of abortion and stillbirth, Gestational age, birth weight) were retrieved just to characterize the child.

Data Analysis

The collected data were cleaned, entered to EpiData Manager Version 2.0.8.56 computer software and analyzes were made using SPSS software program version 22.0. Descriptive statistics like frequency was used to describe the study participants. BDs were classified using the International Classification of Diseases. The overall prevalence of birth defect was computed from total hospital births during the same period of time. An infant/fetus with more than one anomaly was counted once only based on the primary diagnosis. P-value less than 0.05 were considered as statistically significant between common types of BD and child of mothers' characteristics using Fisher exact test.

Ethical Considerations

Ethical approval was obtained from Institutional Review Board of College of Health Sciences (IRB-CHS), Addis Ababa University research and postgraduate study office in collaboration with Jimma University institutional review board to conduct the research. The ethical and supportive letters were submitted to medical directors of all study hospitals to commence with data collection. Data collection began after permission was obtained from medical director of each study hospital. The data collected from medical record were kept confidentially. The study participants were not present during data collection as the study was retrospective study.

Results

From September 2011 to December 2015, 45,951 total deliveries were recorded. Out of these, 253 births were with a BD that was detected at birth. This makes the overall prevalence of BD 5.5 per 1000 births. The majority (92%) of the mothers were younger than 35 years. About half of birth defect (49.6%) occurred among mothers whose ages were between 25 - 35 years (table 1). 51.8% and 48.2% of children born with birth defect were males and females respectively by their sex. About 63.1% of neonates with a BD were born as stillbirth. The majority of the mothers had no previous history of abortion and stillbirth, 92.5% and 93, 3% respectively (table 1).

Table 1: Socio-demographic and obstetric of mothers and child born with defect, southwest Ethiopia.

Characteristics		Frequency (%)	Percentage
Religion of the mother	Muslim	115	45.6
	Orthodox	33	13.1
	Protestant	26	10.3
	Not mentioned	78	31.1
Age of the mothers	15-24	105	41.7
	25-35	125	49.6
	36-43	12	4.8
	Missing	10	4.0
Onset of Labor	Spontaneous	202	80.2
	Induced	50	19.9
Mode of delivery	Vaginal	213	84.5
	Caesarean section	39	15.5
Sex of the newborn	Male	126	50.0
	Female	126	50.0
Status at birth	Alive birth	93	36.9
	Stillbirth	159	63.1
History of abortion	Yes	19	7.5
	No	233	92.5
History of stillbirth	Yes	17	6.7
	No	235	93.3
Gestational age	Term	139	55.2
	Preterm	99	39.3
	Post term	3	1.2
	Missing	11	4.4
Birth order	1-4	116	46.0
	5-9	41	16.3
	Missing	95	37.7

Twenty-one different BDs were recorded. These can be grouped as neural tube defects (NTD) (73.9%), gastrointestinal defects (13.4%), musculoskeletal defects (11.1%) and Genitourinary defects (2%) (Table 2). The major BDs identified during the study period, NTDs were the most prevalent with frequency of 73.9% followed by gastrointestinal defects (13.4%). Genitourinary defects were the least prevalent with the frequency of 2% followed by musculoskeletal (11.1%) (table 2).

Birth defects	Male (n = 131)		Female (n = 122)		Total (n= 253)	
	Frequency	%	Frequency	%	Frequency	%
Neural tube defects	89	67.9	97	79.51	186	73.52
Gastrointestinal defects	20	15.3	14	11.48	34	13.44
Genitourinary defects	5	3.8	0	0	5	1.98
Musculoskeletal defects	17	13.0	11	9.02	28	11.07
Total	131	100	122	100	253	100

Table 2: Frequency of birth defects by sex 2011 – 2015 in Southwestern Ethiopia.

Of the NTDs recorded, Anencephaly (33.9%) and Hydrocephalus (33.3%) were found to be the most frequent followed by spinal bifida (17.7%). While, Microcephaly (1.6%) and Craniorachischisis (1.6%) were the least frequent (table 3).

Table 3: Frequency of neural defects 2011 – 2015 in Southwestern Ethiopia.

Neural tube defects	Male (n=89)		Female (n = 97)		Total (n = 186)	
	Frequency	%	Frequency	%	Frequency	%
Anencephaly	25	28.1	38	39.2	63	33.87
Hydrocephalus	32	17.2	30	30.9	62	33.33
Spinal bifida	15	16.9	18	18.6	33	17.74
Meningomyelocele	10	11.2	8	8.3	18	9.67
Encenphocele	2	2.3	2	2.1	4	2.15
Microcephaly	3	3.3	0	0	3	1.61
Craniorachischisis	2	2.3	1	1.03	3	1.61

Among gastrointestinal defects, the proportion of umbilical hernia, imperforate anus, Gastroschisis, Duodenal Atresia, congenital inguinal hernia and omphelocell was 35.3%, 26.5%, 17.7%, 8.8%, 8.8% and 2.9% respectively. Among gastrointestinal defects, the umbilical hernia was the most prevalent, 35.3% and omphelocell was the least frequent, 2.9%, (table 4).

Among musculoskeletal defects, the proportion of Clubfoot, Cleft, cleft palate, both cleft lip and palate and Chest deformity was 36%, 24%, 20%, 16% and 4% respectively in decreasing order of their frequencies (table 5).

Table 4: Frequency of gastrointestinal defects 2011 – 2015 in Southwestern Ethiopia.

Gastrointestinal defects	Male (n=21)		Female (n = 13)		Total (n = 34)	
	Frequency	%	Frequency	%	Frequency	%
Umbilical hernia	3	14.3	9	69.2	12	35.3
Imperforate anus	7	33.3	2	15.4	9	26.5
Gastroschisis	5	23.8	1	7.7	6	17.7
Duodenal atresia	2	9.5	1	7.7	3	8.8
Congenital inguinal hernia	3	14.3	0	0	3	8.8
Omphelocell	1	4.8	0	0	1	2.9

Table 5. Frequency of musculoskeletal defects 2011 – 2015 in Southwestern Ethiopia

Musculoskeletal defects	Male (n=15)		Female (n = 10)		Total (n = 25)	
	Frequency	%	Frequency	%	Frequency	%
Clubfoot-bilateral	3	20.0	6	60.0	9	36.0
Cleft Lip	4	26.7	2	20.0	6	24.0
Both cleftlip and palate	3	20.0	2	20.0	5	20.0
Cleft palate	4	26.7	0	0.0	4	16.0
Chest deformity	1	6.7	0	0.0	1	4.0

Genitourinary defects were the least frequent constituting 2% of the total major BDs identified in this study. Among genitourinary defects, the proportion of hypospadias, meatal stenosis, ambiguous genitalia was 50%, 37.5% and 12.5% respectively (table 6). The result showed that hypospadias was the most frequent and ambiguous genitalia were the least frequent among identified genitourinary defects.

Table 6: Frequency of genitourinary defects 2011 – 2015 in Southwestern Ethiopia

Genitourinary defects	Male (n=7)		Female (n = 1)		Total (n = 8)	
	Frequency	%	Frequency	%	Frequency	%
Hypospadias	4	57.1	0	0	4	50
Meatal stenosis	2	28.6	1	100	3	37.5
Ambiguous genitalia	1	14.3	0	0	1	12.5

Of the twenty – one BDs identified in the present study, five types of BDs namely: Anencephaly (25.0%), Hydrocephalus (24.6%), Spinal bifida (13.1%), Meningomyelocele (7.1%), and Umbilical hernia (4.8 %) accounted about three-fourth (75%) of all recorded birth defects (table 7).

Table 7: Over all types of birth defects recorded from September 2011 to December 2015 in Southwest Ethiopia in order of their frequency.

Types of Birth defects	Frequency (%)	Percentage
Anencephaly	63	25.0
Hydrocephalus	62	24.6
Spinal bifida	33	13.1
Meningomyelocele	18	7.1
Umbilical hernia	12	4.8
Imperforate anus	9	3.6
Clubfoot-bilateral	9	3.6
Cleft Lip	6	2.4
Gastroschisis	6	2.4
Both lip and palate cleft	5	2.0
Hypospadias	4	1.6
Cleft palate	4	1.6
Encenphocele	4	1.6
Duodenal atresia	3	1.2
Microcephaly	3	1.2
Meatal stenosis	3	1.2
Craniorachischisis	3	1.2
Congenital inguinal hernia	3	1.2
Chest deformity	1	0.4
Ambiguous genitalia	1	0.4
Omphelocell	1	0.4
Total	253	

The association between the common types of defects and characteristics of the mothers and children were computed using Fisher's Exact Test. Only status of the child at birth (P-value =0.0001), birth weight (P-value =0.0001) and gestational age (P-value =0.0001) were found to be associated with types of birth defects.

Discussion

Birth defects, commonly called CA, occur at a rate of 1 in every 33 babies born. However, some are rather minor and others are major defects. In most cases, the major defects are the leading cause of high perinatal mortality and morbidity in developing countries as well as in developed countries [23].

According to World Health statistics, about 260,000 neonatal deaths worldwide are caused by congenital anomalies [10]. Accordingly, about 7% of all neonatal deaths, but varying from 5% in the South-East Asia Region to more than 25% in the European Region. It was also indicated that between-country variation, from 4% (Bangladesh, Equatorial Guinea, Ethiopia, Liberia, Mali and Sierra Leone) and an estimated 8% in China to 38% [10]

In the present study, a total of 253 births were identified with a BD at birth from 45951 total deliveries during the study period. The overall prevalence rate of BDs in the present study was found to be 5.5 per 1000 total births where the NTDs were the most prevalent constituting about 73.5% of all the defects identified in the current study with prevalence rate of 4.05/1000 total births.

The study done in referral hospital of Northwestern Ethiopian supported the present study that the NTDs were the most prevalent types of birth defect with 32.5% [24]. According to study done in china, the prevalence rate of NTD (20.1 per 10000 births) including anencephaly (6.9 per 10000), spinal bifida (10.6 per 10000), and encephalocele with 2.7 per 10000 was the highest, followed by congenital heart disease 17.1 per 10000 [3]. In the present study, the neural tube defects with higher prevalence rate of (40.5 per 10000 child birth) with predominant types of NTDs namely: Anencephaly (13.7 per 10000), Hydrocephalus (13.2 per,10000), Spinal bifida (7.2 per 10000)and Meningomyelocele (3.9 per 10000).The prevalence rate of NTD was twice (40.5 per 10000 total births, when compared with prevalence rate (20.1 per 10000) of NTDs of the study done in China. While the prevalence rate of spinal bifida (7.2 per 10000) slightly less than that of the prevalent rate of Spinal bifida (10.6 per 10000).

As shown in the present study, Anencephaly and Hydrocephalus were the most common associated BDs with the frequency of 25% and 24.6% respectively. The other 1.43per 1000 child births were belongs to the rest the BDs identified in the present study indicating that the NTDs were the most prevalent. In line with the our study, Abbey et al., 2017 [25] demonstrated that the prevalence of major CA at the UPTH in the

Niger Delta during the period 2011–2014 was 20.7 per 1,000 live births, with those of the central nervous system predominating at 27% of the total.

The study done in china identified that the prevalence rate of BD was 15.6 per 1000 [3].On the other study, the overall prevalence of a major BD was 446.3 per 10,000 births [26]. In both studies, the BDs were seems to be higher as compared to the present study. This may be because of differences in sociodemographic, registration and record keeping, racial or environmental factors. Unlike the present study where the NTDs were found to be the most prevalent defects, a study done in china indicated that septal defects (138.2 per 10,000) were the most prevalent followed by congenital hip dislocation (652 per 10,000) [26]. Unlike the presents, the study done in Addis Ababa and Amhara region by Mola et al., 2018 [19] revealed that were the most frequent the Oro-facial defects (34.2%) followed by Neural tube defects (30.8%).

The European Surveillance of CA (EUROCAT) reported that the prevalence of major BD in 2003–2007 was 239 per 10,000 births, of which 80% were delivered, 17.6% were terminated by induced abortion, 2.5% died after birth, and 2% were stillbirths [26, 27].The male to female ratio in the present study was nearly 1:1. There was a Preponderance of females, with a female to male ratio of 1.4:1 as indicated in the study done in north – west Nigeria [27, 28] which is nearly in line with the present study.

Conclusions

The present study revealed that the prevalence of the BD at birth was found to be 5.48 cases per 1000 births. As compared to other study, the prevalence of BD identified in the present study was found to be high. Among twenty one BDs identified in this study, the NTDs were the most prevalent with the frequency of 4.05 cases per 1000 birth. Whereas all the rest constitute only one fourth of the total BDs identified with prevalence rate of 1.43 cases per 1000 birth. The present also identified that nearly equal proportions of BD occurred among males and females newborns. The majority of the mother who gave birth to BDs was younger than 35 years old. As the birth defects were founded to be leading causing of neonatal morbidity and mortality, planning, designing and implementation of prevention and management schemes for BDs are crucial.

Abbreviations

BD	Birth defect
CA	Congenital anomaly
CM	Congenital malformation
NTD	Neural tube defect
WHO	World Health Organization

Declarations

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Author's Contributions

Conceived the idea: SAG.

Designed the study: SAG, LD.

Conducted the study: SAG, GG, DA, LD.

Analyzed the data: SAG, GG, DA, LD.

Interpreted the results: SAG, GG, DA, LD.

Wrote the draft Manuscript: SAG.

Revised and edited the final manuscript: SAG, GG, DA, LD.

Approved the manuscript for submission: SAG, GG, DA, LD.

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Availability of data and materials

The data set supporting this study are available in the manuscript.

Ethical Approval and consent participants

Ethical approval was obtained from Institutional Review Board of College of Health Sciences (IRB-CHS), Addis Ababa University research and postgraduate study office in collaboration with Jimma University institutional review board to conduct the research. The data collection began after permission was obtained from medical director of each study hospitals upon submission of ethical and supportive letters each medical director. As the data collected from medical record, the study participants were not present during data collection.

Consent for publication

Not applicable.

Competing interests

The authors declare that they have no competing interests.

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