

# Prevalence and outcomes of birth defects in newborns of South Wollo and Oromia zones of Amhara regional state: A retrospective study

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## Abstract

**Background:** Congenital malformations (CMs) are structural and functional anomalies that have a significant but under-recognized cause of mortality and morbidity among infants and children under 5 years of age. CMs are not only life threatening, but also result in long-term disabilities that negatively affect individuals, families, health care systems and societies.

**Objectives:** The purpose of this study was to describe the prevalence and outcomes of birth defects in newborns of South Wollo and Oromia zones of Amhara regional state.

**Methods:** A retrospective cross-sectional study was done in three hospitals of South Wollo and Oromia zones, where 22,624 infants were born between January 2015 and December 2017. The presence and type of birth defects in the infants, as well as their outcomes, were identified by reviewing the medical records of their mothers. Medical records with incomplete information were excluded from the study. The data were collected using a pre-tested checklist for data collection.

**Results:** A total of 22,624 infants were born during the study period. Three hundred and twenty-four (1.43%) newborns were delivered with birth defects. Anomalies of the central nervous system (CNS) were the most frequent type of birth defect (43.2%), followed by defects of the musculoskeletal system (19.7%), orofacial malformations (11.0%) and gastrointestinal system anomalies (6.1%). Most of the malformed infants were born from mothers with a history of alcohol intake (55.8%), lack of iron and folic acid supplementation (58.4%), family history of birth defects (58.7%) and history of diseases during pregnancy (53.5%). The outcomes of newborns with birth defects were significantly affected by the type of birth defect, gestational age, weight of the newborn and presence of twin pregnancy ( $p$ -value  $\leq 0.05$ ).

**Conclusions:** The prevalence of CNS anomalies was the highest, followed by musculoskeletal system anomalies. The type of birth defect, gestational age, weight of newborn and twin pregnancy significantly affected outcomes of the newborns with congenital anomalies. Therefore, in order to reduce the prevalence of CMs, the identification and management of risk factors should be the focus of stakeholders. *Ethiop. J. Health Dev.* 2019; 33(3):00-00]

**Key words:** Birth defect, Prevalence, Newborns, Outcomes, South Wollo.

## Introduction

A congenital anomaly can originate before birth (during intrauterine life) and can be identified antenatally, at birth or later in life (1). A congenital anomaly interferes with normal body function, with the cosmetic appearance of the individual, and may lead to infant morbidity or mortality (2-4).

The World Health Organization (WHO) estimates that 270,000 deaths worldwide (about 7% of all neonatal deaths) were caused by birth defects in 2010. Birth defects are among the leading causes of childhood death, chronic illness, and disability in many countries (5). The impact of congenital anomalies is severe in middle- and low-income countries – approximately 95% of children who die from birth defects are from such countries (6).

For 50% to 60% of congenital anomalies, the etiology is unknown (7). However, risk factors for congenital anomalies are multifactorial. They include advanced maternal age, prior pregnancy or family history of birth defect, consanguinity, maternal disease, alcohol drinking, drug intake, viral infection, and exposure to ionizing radiation during pregnancy (8). Congenital anomalies can be also caused by single gene defects,

chromosomal disorders, multifactorial inheritance, environmental teratogens or micronutrient deficiencies (6).

Contrary to the commonly held view that congenital disorders are not a public health issue in developing countries, in recent years, a number of developing countries have experienced an epidemiological transition, with significant declines in infant mortality rates, reduction of infections and malnutrition, and a relative increase in morbidity and mortality due to CMs (9).

Congenital anomalies are not only leading causes of fetal loss, but also contribute significantly to pre-term birth, childhood and adult morbidity, along with considerable repercussions on mothers and their families (10).

A prevalence study conducted in central and north west Ethiopia in Dessie Referral Hospital (DRH), one of the present study sites, reported a relatively high prevalence of CMs at 1.9% (11).

## Objectives

Congenital anomalies pose a significant impact at individual and national level in any given country.

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Knowing the prevalence and outcomes of congenital anomalies is important for planning and implementing programs to reduce morbidity and mortality resulting from birth defects. The main purpose of this study was to estimate the prevalence of birth defects, identify their outcomes, and assess the presence of any risk factors. In addition, the data generated in this study can be used as baseline for further prevalence studies, and for planning and implementing appropriate clinical management strategies by considering the outcomes of birth defects.

### Methods and subjects

**Study area and study period:** The study was conducted in South Wollo and Oromia zones, Amhara regional state, from January 2015 to December 2017. South Wollo and Oromia zones are found 300-470km north east of Addis Ababa. They cover a large area in the eastern Amhara Region.

**Study design:** A retrospective, institution-based, cross-sectional study design was used. Data were collected by reviewing the medical records of mothers to describe the prevalence and outcomes of newborns with CMs.

**Source population:** The source population was all newborn babies with or without birth defects delivered in the health institutions of South Wollo and Oromia zones of Amhara regional state.

**Study population:** The study population was infants born with birth defects, during the study period, in selected health institutions: Dessie Referral Hospital (DRH), Boru Meda Hospital (BMH) and Kemissie General Hospital (KGH).

**Inclusion and exclusion criteria:** Records of mothers with malformed infants were included in the study. Records of mothers with incomplete information were excluded from the study.

**Sample size determination:** The sample size was all the medical records of newborns with CMs in the selected hospitals. A total of 324 records of mothers with malformed infants were identified and reviewed. The records selected were: 252 from DRH, 47 from KGH and 25 from BMH. From these 324 medical records 14 (11 from DRH and three from KGH) were incomplete and were excluded from the study.

**Sampling method:** Registration books of all births attended in the selected study hospitals during the study period were screened for CMs and all documents of mothers with malformed infants were reviewed and used for this study. The study hospitals were selected by way of purposive sampling.

**Data collection method:** Permission to collect data was obtained from hospital managers and medical directors. The data were collected by trained midwives and nurses from medical records using checklists. The

period of data collection was from January 2018 to March 2018.

**Dependent variables:** Congenital malformations and outcomes of newborns.

**Independent variables:** Socio-demographic factors; family history; behavioural factors such as alcohol intake, drug usage during pregnancy; exposure to teratogenic factors; chronic diseases; iron and folate supplementation; and types of CMs.

**Data quality control:** To maintain the quality of data obtained from the documents, checklists were used that were pre-tested before actual data collection. The documentation of information in the checklists was carried out by midwives and nurses who were working at the delivery wards.

**Data analysis:** The data obtained were checked, cleaned and entered into SPSS version 23. Descriptive analysis (frequency) for types and proportions of birth defects was conducted, and risk factors of birth defects seen in the mothers were calculated, with a 95% confidence interval for outcomes of birth defects. P-values of  $\leq 0.05$  were considered as statistically significant. The results were presented in the form of text, figures and tables.

**Ethical considerations:** A support letter was obtained from Wollo University College of Medicine and Health Sciences and submitted to the authorized personnel of the respective health institutions. After obtaining permission from medical directors, the health care providers were contacted and the aims of the study were presented and explained. The patients' anonymity and confidentiality were maintained throughout the study period. Patients' data obtained from each document were kept in a locked cabinet.

### Results

A total of 22,624 infants were delivered in DRH, BRH and KGH from January 2015 to December 2017. Of these newborns, 12,047 (53.25%) were male and 10,577 (46.75%) were female. Of these, 324 (1.43%) were born with congenital malformations (CMs), of which 198 (61.1%) were male and 126 (38.9%) were female. A total of 14 records (11 from DRH and three from KGH) out of 324 were incomplete and were excluded from the study; data from 310 records/documents were then used for analysis. A total of 14 infants out of 1,000 births were born with congenital anomalies.

As shown in Table 1, the socio-demographics of the study subjects showed that there were more male cases of birth defects (63.9%) than female (36.1%). The majority of infants were born from mothers of rural residence (63.2%). Of the infants with CMs, the majority (57.7%) of the cases were born from mothers whose age was greater than or equal to 35 years and whose family's occupation was farming (41.6%).

**Table 1: Socio-demographic characteristics of study subjects in the study hospitals, January 2015 to December 2017**

Characteristics	Variables	Frequency (n=310)	%
Sex of newborn	Male	198	63.9
	Female	112	36.1
Age of mother	< 35 years	131	42.3
	≥35 years	179	57.7
Residence	Urban	114	36.8
	Rural	196	63.2
Occupation	Farmer	129	41.6
	Private employee	79	25.5
	Government employee	90	29.0
	Other	12	3.9

Congenital anomalies pertaining to the central nervous system (CNS) accounted for the majority of the cases (43.2%) followed by musculoskeletal system defects (19.7%), orofacial malformations (11.0%), gastrointestinal system anomalies (6.1%), genitourinary system defects (4.2%), head and neck malformations (3.5%), cardiovascular system defects (2.9%) and Down syndrome (2.6%). Twenty-one of the cases (6.8%) were not specified in the documents.

Among the infants with CNS anomalies, spina bifida was the most common, accounting for 121 (39.0%) of

the total cases of CMs, followed by hydrocephalus (4.2%). Of the musculoskeletal system malformations, club foot was the most common, which accounted for 42 (13.6%) cases followed by 19 omphalocele (6.1%) cases. Combined cleft lip and palate (4.9%) was the most prevalent orofacial malformation, followed by cleft lip (3.2%) and cleft palate (2.9%). Of the gastrointestinal anomalies, imperforated anus accounted for 11 (3.5%) cases, followed by seven cases (2.3%) of infantile hypertrophic pyloric stenosis (IHPS) (Table 2).

**Table 2: Frequency distribution of congenital malformations in the study hospitals, January 2015 to December 2017**

Body systems	Type of CM	Frequency (310)	%
Central nervous system		134	43.2
	Spina bifida	121	39.0
	Hydrocephalus	13	4.2
Musculoskeletal system		61	19.7
	Club foot	42	13.6
	Omphalocele	19	6.1
Orofacial anomalies		34	11.0
	Cleft lip	10	3.2
	Cleft palate	9	2.9
	Cleft lip and cleft palate	15	4.9
Gastrointestinal system		19	6.1
	Imperforated anus	11	3.5
	IHPS	7	2.3
	Other	1	0.3
Genitourinary system		13	4.2
Head and neck CMs		11	3.5
Cardiovascular system		9	2.9
Down syndrome		8	2.6
Unspecified		21	6.8
<b>Total</b>		<b>310</b>	<b>100</b>

IHPS: infantile hypertrophic pyloric stenosis; CMs: congenital malformations

Among the mothers with malformed infants, 129 (41.4%) used iron and folic acid supplementation during pregnancy, whereas 181 (58.4%) did not. The majority of mothers (55.8%) had a history of alcohol

intake. One hundred and eighty-two (58.7%) of the mothers had a family history of birth defects, and about 166 (53.5%) had diseases during the period of pregnancy (Table 3).

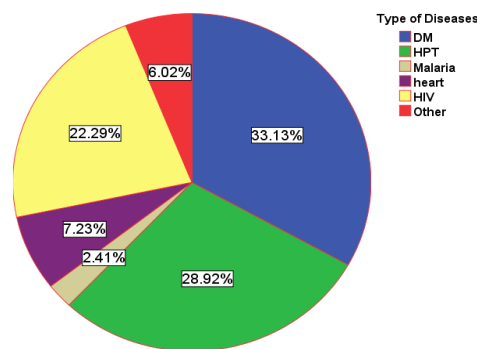
**Table 3: Frequency of risk factors of birth defects observed in mothers of malformed infants in the study hospitals, January 2015 to December 2017**

Variables	Response	Frequency (n=310)	%
Iron and folic acid supplementation	Yes	129	41.6
	No	181	58.4
History of alcohol intake	Yes	173	55.8
	No	137	44.2
Family history of BDs	Yes	182	58.7
	No	128	41.3
Disease during the pregnancy	Yes	166	53.5
	No	144	46.5
Medication history during the pregnancy	Yes	163	52.6
	No	147	47.4
<b>Total</b>		<b>310</b>	<b>100</b>

BDs: Birth defects

Among mothers with a history of disease during pregnancy, about 53 (33.13%) had diabetes mellitus (DM), followed by hypertension (HPT) (28.92%),

HIV/AIDS (22.23%), heart problems (7.23%), other diseases (6.02%) and malaria (1.41%) (Figure 1).

**Figure 1: Types of diseases on mothers of malformed infants during pregnancy in the study hospitals, January 2015 to December 2017**

Of all the mothers, about 163 (52.6%) used medication during the period of pregnancy, while 147 (47.4%) did not. Of those mothers with a history of taking medication, the majority used antidiabetic (34.4%),

antihypertension (25.8%), and antiretroviral drugs (22.7%), drugs for heart problems (7.3%), and other drugs (7.3%) (Table 4).

**Table 4: Types of drugs used during pregnancy by mothers of malformed infants in the study hospitals, January 2015 to December 2017**

Type of drug	Frequency (n=163)	%
Antidiabetic	56	34.4
Antihypertensive	42	25.8
Antimalarial	4	2.5
Heart disease	12	7.3
Antiretroviral	37	22.7
Others	12	7.3
<b>Total</b>	<b>163</b>	<b>100</b>

In this study, of the total 310 infants with CMs, 104 (33.5%) infants died. Outcomes of the infants were significantly affected by the type of CM, with a linear association of 7.92 and p-value of 0.005. Infants with malformations of the central nervous system (CNS),

gastrointestinal system (GIS), cardiovascular system (CVS), genitourinary system (GUS), Down syndrome (DS) and head and neck died more frequently than infants with musculoskeletal and orofacial malformations (Table 5).

Table 5: Outcomes of infants and types of birth defect in the study hospitals, January 2015 to December 2017

Variable	Outcome of malformed infants			Chi square	Df	p-value
	Died	Survived	Total			
Type of CMs	CNS	47 (35.1%)	87 (64.9%)	7.92	1	0.005
	MS	11 (18%)	50 (82%)			
	Orofacial	4 (11.8%)	30 (88.2%)			
	CVS	5 (55.6%)	4 (44.4%)			
	GIS	9 (47.4%)	10 (52.6%)			
	DS	4 (50%)	4 (50%)			
	GUS	9 (69.2%)	4 (30.8%)			
	Head and neck	5 (45.5%)	6 (54.5%)			
	Unspecified	10 (47.6%)	11 (52.4%)			
<b>Total</b>	<b>104 (33.5%)</b>	<b>206 (66.5%)</b>	<b>310 (100%)</b>			

CNS: central nervous system; GIS: gastrointestinal system; DS: Down syndrome  
MS: musculoskeletal system; CVS: cardiovascular system; GUS: genitourinary system

Gestational age, weight of the newborn and presence of twin pregnancy also significantly affected the outcome of newborns, with Pearson chi square values of 3.744, 21.647 and 12.741, and p-values of 0.036, 0.001 and

0.001, respectively. Malformed infants born under 37 weeks of gestational age, below 2.5kg body weight and with twin pregnancy died more often than expected (Table 6).

Table 6: Outcomes of infants with congenital malformations in relation to gestational age, weight and twin pregnancy in the study hospitals, January 2015 to December 2017

Variables	Outcome of malformed infants			Chi square	Df	p-value
	Died	Survived	Total			
GA (weeks)	<37	40 (41.2%)	57 (58.8%)	3.744	1	0.036
	≥37	64 (30%)	149 (70%)			
Birth weight (kg)	<2.5	31 (62%)	19 (38%)	21.647	1	0.001
	≥2.5	73 (28.1%)	187 (71.9%)			
Twin pregnancy	Yes	15 (68.2%)	7 (31.8%)	12.741	1	0.001
	No	89 (30.9%)	199 (69.1%)			
<b>Total</b>	<b>104 (33.5%)</b>	<b>206 (66.5%)</b>	<b>310 (100%)</b>			

## Discussion

The prevalence and types of CMs differ from one country to another and even in the same country from one region to another (12).

In the present study, attempts have been made to find the prevalence of CMs, maternal risk factors observed and their outcomes in hospital deliveries.

The prevalence of CMs in this study was found to be higher than that reported in a study conducted in India (1.25%) (1). However, the prevalence of CMs in the current investigation was slightly lower than the prevalence reported by studies conducted in central and north west Ethiopia (1.9%), India (1.53%), Egypt (2%), and Brazil (2.2%) (11-14). Cohort studies with follow-up programs indicate a higher incidence of CMs than document-based retrospective studies, because they allow the collection of valid information on CMs which are manifested later in life (15).

The relatively lower prevalence of CMs in this study may have been because of the relatively small sample

size used as well as missing data on CMs that could be manifested later in life.

In the present investigation, male infants appeared to be more affected by CMs than female infants. This observation agrees with the findings of a study conducted in western Ethiopia that showed the majority of CMs cases were male children (58.5%) (11).

Congenital anomalies affecting the CNS were the most common BDs observed in this study. This finding is similar to studies conducted in Tanzania, Brazil, other areas of Ethiopia, and Egypt (2,13,16,17). A relatively high prevalence of congenital CNS anomalies was reported by studies in Tanzania (29.8%) (2), Brazil (32.6%) (13), and Egypt (32.1%) (17). In the present study, the prevalence of congenital anomalies affecting the CNS was relatively high. This may be due to the fact that DRH and KGH are among the hospitals in South Wollo and Oromia zones which have many referral linkages with other hospitals and health centers, and receive most of the pregnancies with

suspected difficulties and abnormalities. A much higher prevalence of anomalies of the CNS was reported in a prospective cohort study conducted in northern Ethiopia (68.8%) (16).

In the present study, following CNS anomalies, musculoskeletal system, orofacial, gastrointestinal system and genitourinary system anomalies had the highest frequencies, respectively. Similar observations have been reported by other studies, where CNS anomalies (48%) were the most prevalent, followed by anomalies of the musculoskeletal system (18,19). However, there are other investigations that contradict the findings of our study. In one such study, it was reported that the most prevalent congenital anomalies were observed in the digestive system, which accounted for the maximum number of anomalies of all cases (20). The discrepancy of results may be explained by differences in methodology used. In the present study, a retrospective, document review-based approach was used, whereas in other studies, a prospective descriptive study design was employed. A prospective study design allows collection of information of CMs which is manifested later. Moreover, most CMs (such as CMs of the heart, GIS, respiratory and GUS structures) may not be visible externally on the bodies of newborn infants.

The most prevalent CM of the CNS, in this study, was spina bifida. This finding agrees with the findings of studies conducted in central and western Ethiopia (11), as well as in Mekelle and Ayider hospitals (16), which reported that the most prevalent congenital anomaly of the CNS was spina bifida (51.5% and 70%, respectively). The prevalence of spina bifida is much higher in these two studies compared to that of our study, especially in the study conducted in Mekelle and Ayider hospitals. This may be due to the limitations of a retrospective study compared to a prospective study. The present study was a retrospective study, whereas the study in Mekelle and Ayider hospitals was a prospective study. In the present study, there may be instances of misdiagnosis or failure of health care professionals to carefully document the spina bifida cases.

In this study, club foot had the highest prevalence of cases with musculoskeletal anomalies, while cleft lip and palate were highly prevalent in the cases with orofacial malformations. These findings are consistent with the level of prevalence reported by another similar study (17). However, compared to our finding, a much higher prevalence of orofacial malformations has been reported in a study that was carried out in central and western Ethiopia (11). This discrepancy in the observed prevalence of orofacial malformations may have been due to the difference in the sample size used and difference in the duration of the study period. In the study conducted in central and western Ethiopia (11), the sample size was much larger compared to that of our study. Moreover, in the study conducted in central and western Ethiopia, data were collected from referral hospitals for surgical corrections of malformations and the duration of the study period was

five years compared to that of ours, which was three years.

In this study, the frequency of risk factors associated with the prevalence of CM were analyzed using descriptive statistics. The factors identified as risk factors were maternal age above 35 years, lack of iron and folic acid supplementation, a history of alcohol intake, and a family history of birth defects. In this study, it was observed that the majority of mothers were exposed to risk factors of birth defects. In the present study, the level of correlations between risk factors and prevalence of CMs was not assessed, as the focus of the study was the prevalence of CMs in newborns, relative distribution of BDs in relation to the body systems, and identification of outcomes and presence or absence of risk factors related to CMs.

The mortality rate of the newborns with birth defects was relatively high in the present study compared to that reported by a study conducted in Egypt (20). The outcomes of infants with birth defects were significantly affected by the types of congenital anomalies. Infants with CNS, GIS, CVS, GUS, Down syndrome and head and neck malformations died more than expected, whereas infants with musculoskeletal and orofacial malformations died less than expected. This finding is in line with published data of studies conducted in England and Wales, and in Brazil (21,22).

### Conclusions

In this study, the prevalence of birth defects in newborn infants has been observed to be relatively high, and female infants were less likely to have birth defects compared to male infants. Of all systems of the body, the CNS was the most affected, followed by the musculoskeletal system. Maternal age above or equal to 35 years, lack of iron and folic acid supplementation, history of alcohol intake and history of diseases and drug usage during pregnancy were the risk factors observed in the mothers of malformed infants. The outcomes of the newborns with congenital anomalies were affected by type of birth defect and presence of twin pregnancy. This information gives insight to health care providers to help them plan and implement appropriate preventive, curative and rehabilitative management strategies, depending on the type of CM.

### Limitations of the study

The strength of the present study was the concern given to investigate outcomes of birth defects that provide information in the study settings that have limited research possibilities and coverage in the country. In the present study, because we used a retrospective study approach, getting complete information and avoiding biases were challenging. In addition, there was no control group to evaluate the association of risk factors with birth defects.

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