

EXTERNAL STRUCTURAL CONGENITAL ANOMALIES DIAGNOSED AT BIRTH IN TAMALE TEACHING HOSPITAL

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Abstract

Background: In the year 2013, about 276,000 of the 2.71 million neonatal deaths were directly attributable to congenital anomalies of which an estimated 95%, occurred in low and middle-income countries.

Materials and method: This study was part of surveillance on external structural congenital anomalies conducted in the Tamale teaching hospital from 1st January 2011 to 31st December 2015. Aggregate data on maternal characteristics and outcome of delivery were collected from the records in the antenatal clinics, labour and delivery room, and midwifery monthly returns.

Results: A total of 35,383 live births were recorded in the study. Overall, 161 external congenital anomalies were recorded over the period. The overall incidence of external congenital anomalies over the five-year period was 455 per 100,000 live births. The gastrointestinal

(GIT) system accounted for (77) 47.8%, Central nervous system (65) 40.4%, Major musculoskeletal (9) 5.6%, Genitourinary system about (2) 1.2%, Multi-systemic and syndromes accounted for (8) 5.0% of all external structural congenital anomalies. The incidence of specific congenital anomalies found during the study period were as follows; Spina bifida 104.6 per 100,000 live births, Exomphalus/omphalocele 84.8 per 100,000 live births and Anencephaly 5.7 per 100,000 live births. The incidence of Oro-facial cleft, Hydrocephalus and Imperforate anus were 73.5, 48.0 and 39.6 per 100,000 live births respectively.

Conclusion: External structural congenital anomalies rate in the northern sector of Ghana is unacceptably high. Trend analysis has demonstrated the steady decline in neural tube defects

Key Words: Congenital anomaly, Spina bifida, Exomphalus, Anencephaly, Oro-facial cleft

Introduction

Congenital anomalies are associated with significant morbidity and mortality. In the year 2013, about 276,000 of the 2.71 million neonatal deaths were directly attributable to congenital anomalies¹. Also, congenital anomalies are associated with reduced quality of life, social stigma and the potential of endangering family resources². Globally, congenital anomalies accounts for 49.1- 69.5 million disability adjusted Life-years (DALY) of which sub-Saharan Africa contributes over 80%^{3,4}. It is estimated that, 94% of all congenital anomalies occurs in low and middle-income countries².

Congenital anomalies are critical global health priorities requiring urgent measures to address the unacceptably high incidence in developing countries⁵. It is estimated that the incidence of congenital anomalies is about 10 times higher in Africa compared to developed countries. Fortunately, most of congenital

anomalies are preventable^{6,7}.

Congenital anomalies has been defined to include morphological, biochemical or physiological defects present at birth, which could be diagnosed prenatally, at birth or later in life⁸. It is broadly classified into structural and functional anomalies. Structural anomalies are further described as external or internal. It is believed that the incidence of birth defects were being underestimated in the low and middle income countries due to lack of equipment for the diagnosis of some congenital anomalies particularly the internal and functional anomalies^{9,10}. Most studies about congenital anomalies in developing countries focused on external birth defects because of ease in diagnosis as opposed to the functional and internal birth defects that may require equipment lacking in developing countries.

It is absolutely essential to ensure that couples have healthy anomaly-free children so as to prevent the consequence of birth defects on the child, family and the community. To this end, many developed countries have programs, support and consensus statement aimed at addressing the issue of congenital anomalies¹¹. However in many developing countries little is done beyond folic acid and other multivitamin supplementations. Data on the incidence and trends in congenital anomalies necessary to inform program

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implementation is significantly lacking. The aim of this study was to obtain the baseline incidence of congenital anomalies and also provide the needed information for surveillance.

Methods

Study settings and recruitment

This study was conducted in the Tamale teaching hospital. It used data from 1st January 2011 to 31st December 2015. The Tamale teaching hospital is the only teaching hospital located in the northern sector of Ghana. It has a walk-in delivery service for expectant women in the Tamale Metropolis but also accepts referrals from the district hospitals within its catchment area. All pregnant women presenting for delivery services were eligible for enrolment in the study. There were about 6400 to 7000 deliveries per annum during the study period. Aggregate data on maternal characteristics and outcome of delivery were collected from the records in the antenatal clinics, labour and delivery room, and midwifery monthly returns.

Physical examinations and diagnosis

All babies born in the facility were physically examined first by trained midwives or medical doctor, an obstetrician or a pediatrician whenever necessary to diagnose any congenital anomaly present. However, the obstetrician made the final ultimate diagnosis. Congenital anomalies found during the study period included Spina bifida, Exomphalus/ omphalocele, Anencephaly, Oro-facial cleft, Hydrocephalus, Imperforate anus and Talipes Equinovarus. Others included; Meningocele, Meningoencephalocele, Down syndrome, Gastroschisis, Siamese twins and Encephalocele. External Structural anomalies were studied because they are relatively easy to diagnose without the requirement of sophisticated investigative tools to attain a fairly accurate diagnosis. This provides a fairly accurate measure of incidence of the anomalies of interest in this baseline study. A congenital anomaly case is one diagnosed as having any of the above anomalies. A baby having multiple congenital defects that fits a syndrome is diagnosed as such. Where as those not fitting any particular defect/syndrome but with multiple major external structural anomalies were classified as others/ multiple defects. The study did not include congenital anomalies found in aborted fetus and maternal deaths with undelivered fetuses.

Data collection and analysis

Data were collected and stored by trained public health nurses and midwives. Data were first captured into registers and analyzed monthly to generate monthly statistics. This study puts together all the monthly data for this analysis.

Data were entered and managed using EpiData 3.1 (Atlanta, US). Data analysis was carried out using STATA version 11 (College Station, Texas) and Microsoft Excel. Maternal age was categorized a priori

using a five year interval, 10 – 14, 15 -19, 20 – 24, 25 – 29, 30 -34 and 35 and above. Tabulations were carried out to obtain the characteristics of mother and baby. The Incidence per 100,000 live births was calculated by dividing the number of anomalies by total live births multiplied by 100,000. Anomalies were later grouped under their respective systems to generate the incidence by systems. Line graphs were generated for all the anomalies individually and then as total anomalies. However, only graphs showing significant trends were reported. The study used the proportion of women completing second dose of intermittent prophylactic therapy (IPT) for malaria as a proxy measure of the regular intake of folic acid. Folic acid and IPT were part of routine medications given at antenatal clinic in Ghana. First dose of IPT was served at quickening and the second dose after a month of quickening by directly observed therapy. It remains a challenge to know which of the women actually took the dispensed folic acids. Hence our decision to use the second dose of directly observed therapy for IPT as proxy measure of folic acid intake during pregnancy. It is assumed that a client that took at least two doses of the IPT is more likely to have taken the routine antenatal clinic medications including folic acid in previous pregnancies and the index pregnancy.

Results

Maternal and baby characteristics at delivery

A total of 37,303 births (live births plus still births) were recorded in the study. Total live births amounted to 35,383. The peak age group of mothers at time of delivery was within 25 to 29 years accounting for 31.1% of all deliveries. Vaginal delivery was the preferred mode of delivery accounting for 73.1% of all deliveries over the five-year period. Caesarean section was the method of delivery in 25.8% of the pregnant women while vacuum accounted for 1%. With regards to the sex of the baby, females accounted for 53.7% while males accounted for 45.7%. About 95.1% of all the deliveries were singleton deliveries with twin delivery accounting for 4.9%. Stillbirths constituted 5.2% of all births. Complete details of maternal and baby characteristics at delivery is shown table 1 below.

Incidence of selected congenital anomalies

Overall, 161 external congenital anomalies were recorded over the period. This overall incidence of external congenital anomalies over the five-year period was 455 per 100,000 live births. Spina bifida was the commonest congenital anomaly accounting for 23.0 % of all external structural anomalies. The incidence of spina bifida varied over the study period, however, the five-year incidence of spina bifida was 104.6 per 100,000 live births. Exomphalus was second commonest with a cumulative incidence of 84.8 per 100,000 live births. The incidence of Oro-facial cleft, hydrocephalus and imperforate anus were 73.5, 48.0 and 39.6 per 100,000 live births respectively. The

relatively rare congenital anomalies in our environment include siamin twin and Encephalocele, which recorded a cumulative incidence of 2.8 per 100,000 live births each. Incidences of other major structural anomalies at birth are included in table 2 below.

The gastrointestinal (GIT) system accounted for most of the anomalies representing (77) 47.8% of all external structural congenital anomalies, an incidence of 206 per 100,000 live births.

Table 1: Characteristics at delivery

Maternal and Baby Characteristics	Frequency	Proportion (%)
Mother characteristics		
Age of mother at Delivery (years)		
10 – 14	3	0.0(≈0.01)
15 – 19	1,837	5.1
20 – 24	8,094	22.6
25 – 29	11,134	31.1
30 – 34	9,617	26.8
35 and above	5,148	14.4
Mother completing two doses of Intermittent Preventive Therapy (IPT) for malaria		
Completed two IPT doses	31,731	89.4
Did not complete two IPT doses	3,761	10.6
Mode of Delivery		
Vaginal	25,958	73.1
Caesarean Section	9,168	25.8
Vacuum	366	1.0
Mother’s Anti-Retroviral (ARV) medications history		
Taking ARV	102	0.3
Not taking ARV	35,390	99.7
Baby characteristics		
Sex of Baby		
Male	16,193	45.7
Female	19,015	53.7
Outcome of Delivery		
Singleton	33,414	94.9
Twins	1,739	4.9
Triplets	40	0.1
Quadruplets	1	0.0(≈0.003)
Births		
Live births	35383	94.9
Still births	1920	5.1

Central nervous system followed with (65) 40.4% representing 174 per 100,000 live births.

Major musculoskeletal system anomalies accounted for (9) 5.6%, 25 per 100,000 live births. Genitourinary system recorded the least anomaly rate with a five-year incidence of about (2) 1.2%, 6 anomalies per 100,000 live births. Multi-systemic and syndromes accounted for (8) 5.0% of all anomalies and recorded a cumulative incidence of 21.5 per 100,000 live births.

Table 2: Incidence of major structural anomalies at birth

	Congenital anomaly	No.	%	Incidence per 100,000 live Births
1	Spina bifida	37	23.0	104.6
2	Exomphalus/ omphalocele	30	18.6	84.8
3	Oro-facial cleft	26	16.1	73.5
4	Hydrocephalus	17	10.6	48.0
5	Imperforate anus	14	8.7	39.6
6	TalipesEquinov arus	9	5.6	25.4
7	Meningocele	6	3.7	17.0
8	Down syndrome	4	2.5	11.3
9	Gastrochisis	3	1.9	8.5
10	Anencephaly	2	1.2	5.7
11	Meningoenceph alocele	2	1.2	5.7
12	Achondroplasia	2	1.2	5.7
13	Imperforate urethra	2	1.2	5.7
14	Siamin twins	1	0.6	2.8
15	Encephalocele	1	0.6	2.8
16	Others	5	3.1	14.1
	Total	161	100.0	472.0

*Total births – live births plus stillbirths

Trend analysis

Total structural anomaly rate has been sinusoidal over the study period as found in Fig 1.

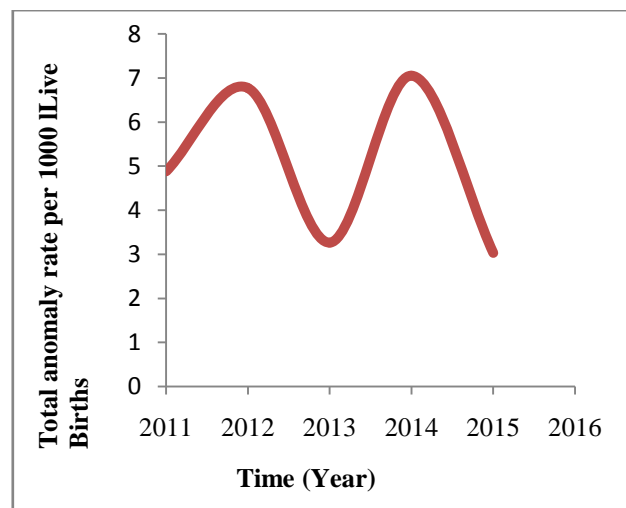


Fig 1: Trend in total anomalies at birth rate per 1000 live births

However, neural tube rate per 1000 live births continue to reduce as shown in Fig 2.

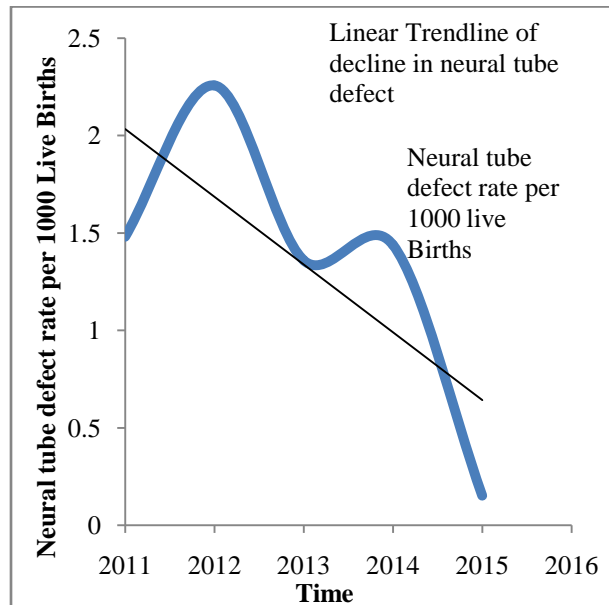


Fig 2: Trend in Neural Tube Defect Rate Per 1000 Live Births

A trend line showed a steady decline. This study used the proportion of women completing second dose of intermittent preventive therapy for malaria (IPT2) as a proxy measure for routine antenatal clinic (ANC) attendance and regular intake of the routine antenatal medications served at ANC including folic acid. Fig 3, demonstrated that with increase in folic acid supplementation there was a decrease in neural tube defects in our environment.

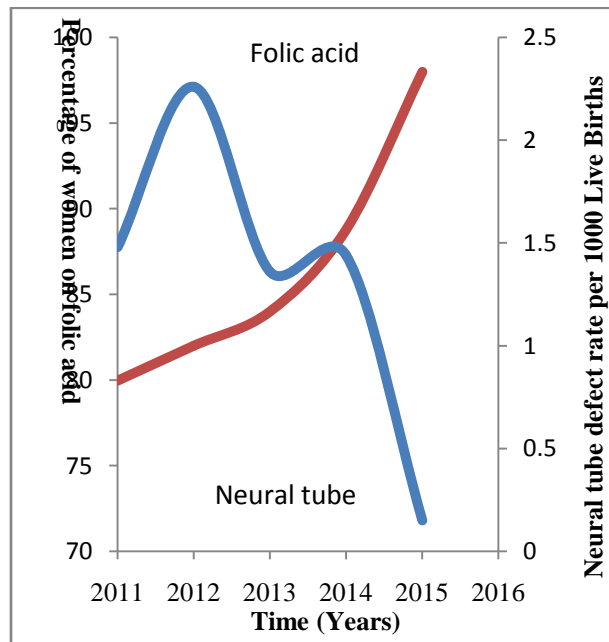


Fig 3: Trend in neural tube defect rate per 1000 live births versus percentage of women completing second dose of intermittent preventive therapy for

malaria used as a proxy measure of folic acid intake during pregnancy

Exomphalus/ omphalocele rate has been sporadic over the period of study. However, between March 2013 and May 2015, there was a surge in the rate of exomphalus/omphalocele. Fig 4 demonstrates this trend.

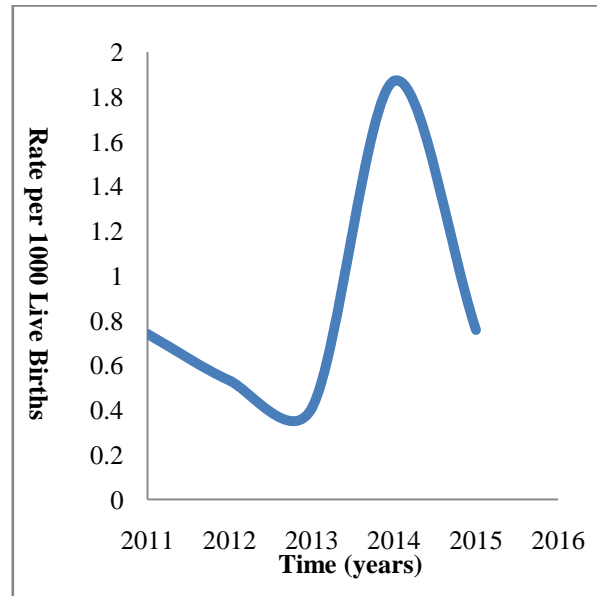


Fig 4: Exomphalus rate per 1000 live births versus time in years

Discussion

Main findings

Spina bifida was the commonest congenital anomaly found in this study. The annual rate decreased steadily over the five-year period. Conclusive evidence from several randomized controlled trials emphasized the beneficial role of preconception folic acid supplementation in the prevention of neural tube defects¹². There is a demonstrated risk reduction in spina bifida through folic acid supplementation^{13,14,15}. The general rule in prevention of neural tube defects is; two or three month of preconception folic acid supplementation and up to three month of antenatal folic acid supplementation¹⁶. Most pregnancies in Africa are unplanned. As such, some authors rank food fortification as the best approach to prevent neural tube defects^{17,18}. In Ghana however, data on pre-conception folic acid supplementation as well as food fortification is scarce. Available data on folic acid supplementation is that of antenatal clinic folic acid supplementation program. Antenatal Folic acid supplementation program in Ghana is near universal for pregnant women attending antenatal clinic. Antenatal clinic attendance has progressively risen over the years with over 97% of women attending antenatal clinic¹⁹. This may contribute to the reduction in spina bifida over the study period. Also, the national health insurance

operates a free maternal health services for all pregnant women. This we believe had increased the access to folic acid and other supplementation for pregnant women who otherwise could not buy the supplements. There is the need for further studies in our environment to ascertain the prevalence of preconception folic acid supplementation among women in the reproductive age group.

Gastrointestinal system (GIT) anomalies were most common in this study. Exomphalus/ omphalocele were the most common GIT anomalies recorded. The literature on omphalocele suggests a generally stable pattern²⁰. However, this study found, contrary to a generally stable pattern, a surge in the incidence of Exomphalus with a pattern suggestive of a common source exposure outbreak. Exomphalus also known as omphalocele is a defect in muscle development in-utero and has been strongly linked to chromosomal abnormality^{21,22}. The curve obtained in this study suggests some environmental exposure, which might have precipitated the chromosomal changes. Some studies have found the association between congenital anomalies and ambient air pollution²³. Another gastrointestinal defect of high incidence was cleft lip/palate. Oro-facial clefts remain a relatively common birth defect worldwide²⁴. In Ghana, there are programs aimed at repair of clefts. However, little is done in its prevention. A study conducted in the United States found smoking to be associated with Oro-facial cleft. It was estimated that cessation of smoking in early pregnancy would prevent 430 Oro-facial cleft in the United States²⁵.

Application of the findings

The findings from the study with regards to the incidence of structural congenital anomalies have demonstrated a high incidence of some congenital anomalies in our environment. However, the success associated with the folic acid supplementation program should encourage practitioners to encourage women to strictly adhere to the folic acid supplementation guidelines. This study provides the baseline data for the establishment of surveillance on congenital anomalies in Tamale. This study also demonstrated a possible environmental exposure for the development of some congenital anomalies, which would require further studies into environmental exposures in our environment, which may be linked to congenital anomalies. The high incidence of congenital anomalies requires a more comprehensive approach at reducing the incidence that would include preconception prevention strategies, prenatal diagnosis and counseling, and post-natal interventions to reduce the incidence, morbidity and mortality associated with congenital anomalies.

Strengths and limitation of the study

Delivery information on large number of women and babies were included in this study. It also spanned

a five-year period. The above reasons were likely to have estimated the true incidence of external structural congenital anomalies in our catchment area. However, congenital anomaly from pregnancies in which maternal mortality was recorded with fetus undelivered was not included in the study, which could underestimate the true incidence in our environment. Also the study included only women delivering in the hospital. This could underestimate the true incidence due to proportion of women in the northern region who prefer to deliver at home, with traditional birth attendants and private maternity homes. Nevertheless, health facility delivery in Ghana has been described as persistently high(26).

Conclusion

External structural congenital anomalies rate in the northern sector of Ghana is unacceptably high. Trend analysis has demonstrated the steady decline in neural tube defects over the five-year period, which is due to the success of folic acid supplementation program hereby strongly encouraged.

Acknowledgements

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