Scientific Research Publishing

Open Journal of Radiology, 2023, 13, 67-76
https://www.scirp.org/journal/ojrad
ISSN Online: 2164-3032
ISSN Print: 2164-3024

Burden of Congenital Defects Diagnosed through Ultrasonography in Soba Fetomaternal Unit, Khartoum, Sudan

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How to cite this paper: Osman, M.A.M.A., Elhassan, I.A.A., Khan, F.N., Alimam, A., Noma, M. and Fazari, A. (2023) Burden of Congenital Defects Diagnosed through Ultrasonography in Soba Fetomaternal Unit, Khartoum, Sudan. *Open Journal of Radiology*, **13**, 67-76.
https://doi.org/10.4236/ojrad.2023.131007

Received: January 20, 2023
Accepted: March 24, 2023
Published: March 27, 2023

Abstract

Background: Congenital anomalies are among the leading causes of fetal loss, despite it can be identified prior to birth through advanced technology in expert hands. Our research aimed at estimating the prevalence of congenital anomalies in Sudan. **Methods:** A facility-based retrospective cross-sectional study combined with a community-based survey through a telephone interview was implemented on a purposive convenient sample of 138 participants. The data were computerized in Epi Info 7. Google Earth Pro enabled to collect the geographical coordinates for the residence of the participants. Descriptive statistics were performed through SPSS 23 and ArcGIS 10.3 was used to generate the geographical distribution map of congenital defects to visualize the catchment areas of Soba Ultrasonography Unit. **Results:** Of the 138 participants, the estimated prevalence of congenital defects was 2.2/10,000 live births. The ultrasonography screening revealed that neural tube defects were the most prevalent anomalies with 13.0% (18/138), which represented 47.4% (18/38) of all defects. Concerning children, a mortality rate of 23.2% (32/138) was reported. **Conclusions:** The child mortality rate post ultrasound screening of 23.2%, and the neural tube defects being the most common anomalies appealed to Sudan health authorities for focusing on more preventive antenatal practices to strengthen and promote maternal and child health.

Keywords

Prevalence, Congenital Defects, Ultrasound, Spatial Distribution

1. Introduction

Congenital anomalies are conditions of prenatal origin that can be identified prenatally, at birth, or may only be detected later in infancy. They include structural and functional abnormalities that impact fetal or infant health, development and/or survival. Congenital anomalies have a significant impact on individuals, families and healthcare systems as they contribute to perinatal mortality and morbidity. These anomalies can occur in isolation (single defect) or as a group of defects (multiple defects), and have different names such as congenital abnormalities, malformations, disorders or defects. An estimated 240,000 newborns die worldwide within 28 days of birth every year due to birth defects. Birth defects cause a further 170,000 deaths of children between the ages of 1 month and 5 years. Moreover, low- and middle-income nations are disproportionately impacted by congenital abnormalities, which are one of the primary causes of the global disease burden [1] [2].

Ultrasound Scanning (USS) is an ideal imaging procedure for a primary diagnostic and screening method during pregnancy. The detection of anomalies could be hampered by factors that intervene with visualization like maternal obesity, oligo/anhydramnios, fetal position and reverberation caused by bone. This would indicate another screening modality like magnetic resonance imaging. The types of fetal anomalies, which can be detected by ultrasonic diagnosis in different gestational ages, include: central nervous system, genitourinary, cardiovascular, respiratory, gastrointestinal, musculoskeletal, facial deformity, ascites and pleural effusion, cystic hygroma, teratoma and multiple malformations [1] [3].

Second-trimester scan, between 18 and 22 weeks, remains the standard for fetal anatomical assessment worldwide. However, significant improvement in detecting fetal abnormalities in the first trimester of pregnancy is also recognized [4].

The European Surveillance of Congenital Anomalies (EUROCAT) was set up for detecting any epidemic of congenital anomalies. The prevalence and trend of 61 congenital anomaly subgroups (excluding chromosomal) in 25 population-based EUROCAT registries (1980-2012) indicated a significant increase in Congenital Heart Disease (CHD) which was attributed to the increase in the number of diabetics as well as overweight mothers; while the decrease of the prevalence of limb reduction could not be explained. The increase in renal anomalies was due to rigorous screening; the reported birth prevalence of congenital heart disease had reached an estimate of 9/1000 live births in the last 15 years; the birth prevalence of congenital heart disease varies according to the geographical location of the patient and the severity of the heart defect [5].

The Netherlands National Screening Program on prenatal detection of severe congenital heart anomalies was evaluated. It was found that the detection rate of all CHD increased significantly from 35.8% before to 59.7% after the introduction of the National Screening Program ($p < 0.001$). It was concluded that pre-

natal detection of CHD remains challenging, especially for ultrasonographers who were minimally exposed to these anomalies [6]. It cannot be denied that computerized birth registries and new software applications play a significant role in analyzing and identifying trends; consequently, the current study attempted to apply similar technologies in spatially distributing the types of defects for visualization and easy capture of regions that require more attention in terms of antenatal health services. The identification of multiple congenital defects was made more accurate by combining population-based birth defect data such as EUROCAT with epidemiological data in a computer-based algorithm [7].

A household survey was conducted in Nepal villages on a sample of 21,111 women and 27,201 children with congenital defects. The prevalence of congenital defects was 52.0/10,000 children (95% CI: 44.0 - 61.0), and the majority were born to mothers with poor health. One of the most severe forms of congenital defects was Neural Tube Defects (NTDs) which can be prevented through proper nutrition and folic acid supplements [8]. The province of Shanxi in China had the highest reported worldwide incidence of congenital heart defects which was partially attributed to the presence of coal mines and many other minerals in the soil. Various strategies were applied in order to reduce the incidence. A spatial and temporal analysis of a live and stillbirths was conducted in two Chinese localities between the years 1998-2012. The findings indicated that the interventions implemented by the government, such as food fortification by adding five micro-nutrients, might have a positive impact on reducing the overall incidence of NTDs. The results also revealed the existence of significant spatial heterogeneity. NTD clusters were identified in areas close to coal sites and main roads even after intervention [9].

Food fortification with folic acid is a proven strategy to reduce neonatal and under-five mortality in general and those associated with spina bifida in particular, and it is recommended that countries implement mandatory folic acid fortification of staple foods without further delay [10].

This study aimed to estimate the prevalence of congenital defects, their types and geographical distribution in pregnant women who had ultrasonography examinations at the Fetomaternal Unit of Soba University Hospital, Sudan.

2. Methodology

A facility-based retrospective record-based study combined with a community-based survey was implemented. The research was conducted in the Fetomaternal Unit of Soba University Hospital in Al Khartoum (Sudan), where a purposive convenient sample of 138 ultrasonography records was extracted, in March-May 2018, from the electronic database of a total of 2500 patients examined during the period of January 2016 to December 2017. Hence, the sample examined represented 5.5% of the women who had ultrasonography screening during the period of January 2016 to December 2017. A standardized data tool

was used to extract the data needed to address the research objective. The research tool had two parts; Part 1 for recording maternal characteristics and part 2 for fetal characteristics. A community survey was conducted through a telephone interview to collect the missing data on the residence of participants, mother and child's current health status (at the time of interview) and the outcome of the pregnancy.

The data collected were computerized using a template elaborated in Epi Info™ 7.1.5.2, free software developed by the Center for Disease Control, Atlanta, USA. The Statistical Package for Social Sciences (SPSS version 23) was used to summarize the data numerically (mean, standard deviation and median) and graphically (frequency tables for estimating prevalence and graphics). Google Earth Pro 7.1.8.3036 (32bit) was used to obtain the geographical coordinates (latitudes and longitudes) of the residence of the participants. The Geographical Information System (ArcGIS 10.3 for Desktop version 10.3.043322) was used to elaborate the spatial distribution map of congenital defects.

Ethical Approval and Consent to Participate

The research was reviewed by the Institutional Review Committee of the University of Medical Sciences and Technology (UMST) and was authorized by the General Director of Soba University Hospital. The community-survey obtained a verbal well informed consent from all the participants.

3. Results

3.1. Characteristics of Participants

The age of the 138 females, who went through ultrasonography screening, ranged from 17 to 40 years with an average age (median) of 29 years. 40.6% (56/138) were highly educated whereas 5.8% (8/138) had never attended a formal schooling. Their gynecological and obstetrical backgrounds revealed that their gravidity ranged from 1 to 8 pregnancies with an average of 3 pregnancies; their average parity of 2 varied from 0 to 8. They had between 0 to 8 miscarriages as revealed in Table 1. Figure 1 displayed the geographical distribution of the participants according to their respective state of residence.

3.2. Types and Prevalence of Congenital Defects

Types of Congenital Defects

Congenital defects were present in 38 of 138 pregnant women who went through ultrasonography screening. This represented a proportion of 27.5% (38/138) congenital defects of all types. Table 2 revealed the details of the ultrasonography screening results and Figure 2 displayed the distribution of congenital anomalies in the involved states. The ultrasonography screening revealed that neural tube defects were the most prevalent anomalies with 13.0% (18/138), which represented 47.3% (18/38) of all defects. The ultrasonography examination also revealed cardiac (10.5%, 4/38), renal (7.9%, 3/38), musculoskeletal (5.3%, 2/38) and abdominal wall (2.6%, 1/38) anomalies. Multiple defects (more than one system

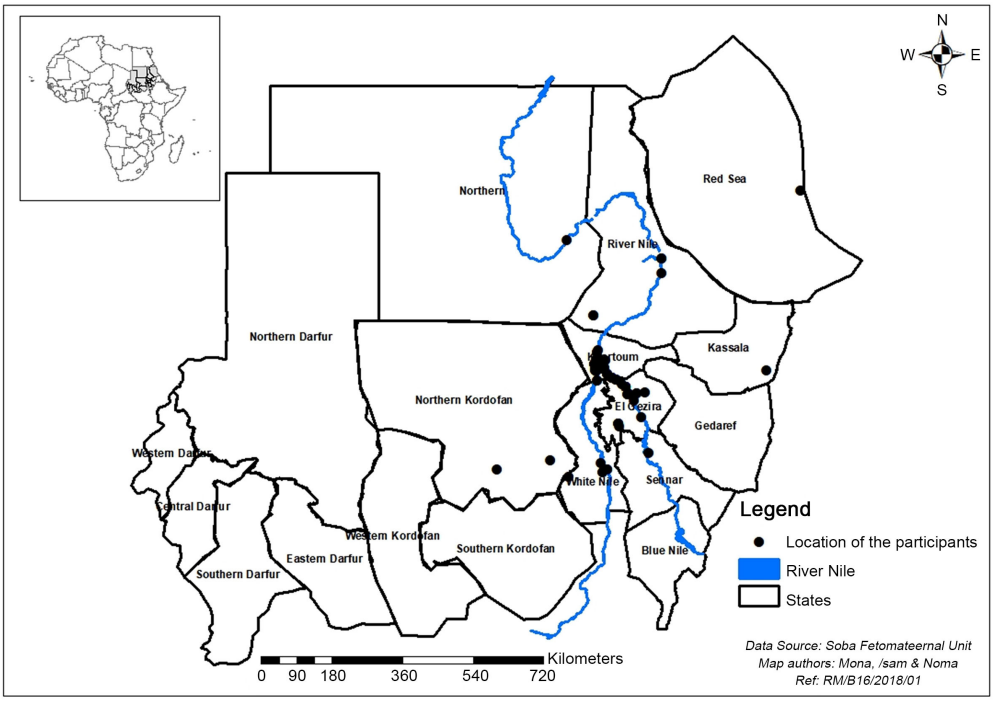

Figure 1. Geographical distribution of the study participants based on their state of origin (n = 138).

Table 1. Characteristics of the study participants (n = 138).

Variable	Number	%
Study setting (n = 138)		
Soba University Hospital (SUH)	138	100
Education levels (n = 138)		
University or higher	56	40.6
Secondary	46	33.3
Primary	28	20.3
Never been to school	8	5.8
Age in years (n = 138)		
Median	29	
Min-Max	17 - 40	
Gravidity (n = 138)		
Median	3	
Min-Max	1 - 11	
Parity (n = 138)		
Median	2	
Min-Max	0 - 8	
Miscarriage (n = 137)		
Median	0	
Min-Max	0 - 8	

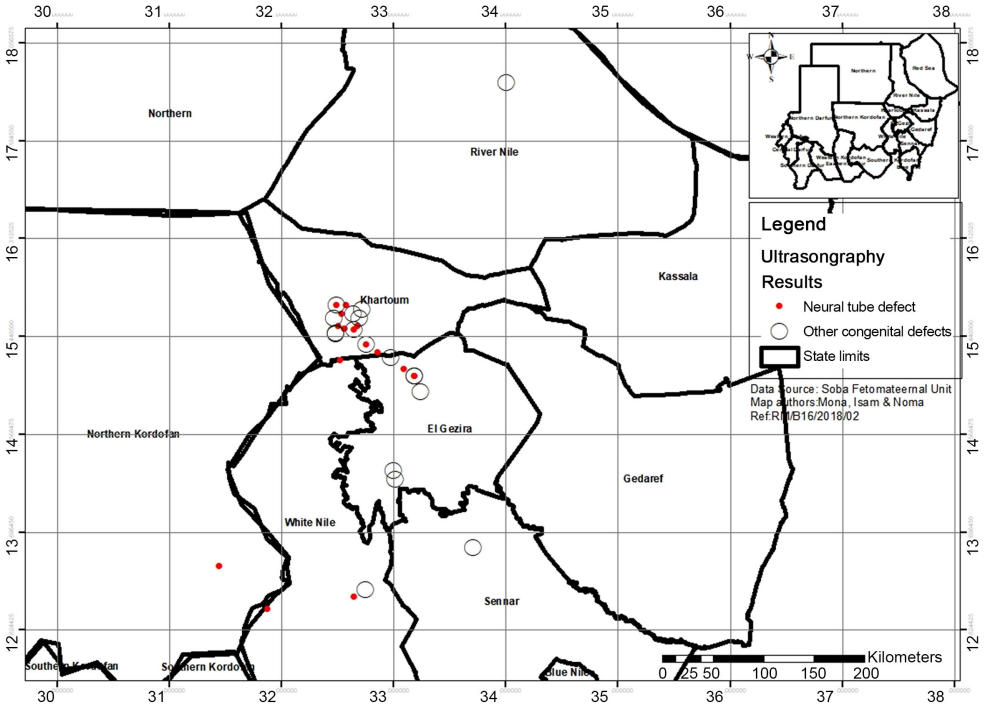

Figure 2. Geographical distribution of the congenital defects (n = 38) diagnosed through ultrasonography in Soba Fetomaternal Unit.

Table 2. Results of 138 women who went through ultrasonography screening in Soba Fetomaternal Unit.

Type of congenital defect	Number	%
No structural abnormality	100	72.5
Neural tube defect	18	13.0
Multiple defects	5	3.6
Others	5	3.6
Cardiac defect	4	2.9
Renal defect	3	2.2
Musculoskeletal defect	2	1.4
Abdominal wall defect	1	0.8

affected) represented 13.2% (5/38) of the total anomalies detected (n = 38) whereas hydrops, cystic hygroma and down syndrome categorized as "others" were 13.2% (5/38).

The *neural tube defect* was diagnosed in young adult women aged on the average (median) 29 years (range: 21 - 39 years). These women had a history of 4 pregnancies (range: 1 - 11) which lasted on an average delivery of 2.5 ranging from 0 to 8. Unfortunately, they presented late for ultrasound screening with an average (median) gestational age of 31 weeks.

The *other types of congenital defects* (n = 20), except neural tube anomaly, were diagnosed in women aged 28 years ranging from 17 to 40 years, they had

an average of 3 pregnancies (range: 1 - 6) and delivered between 0 and 5 newborns with an average of 2 newborns. They presented to ultrasound examination earlier than those with neural tube defects with an average of 29 weeks ranging from 14 to 36 weeks; 85.0% (17/20) who presented at a gestational age ≥ 25 weeks, one participant (1/20) was examined at a gestational age of 14 weeks and 10.0% (2/20) at the period between 18 and 24 weeks.

Maternal and Child Health Status

Telephone interviews enabled to collect the data which were missed from the ultrasound reports. These data were the pregnancy outcome, the mothers and children health status at the time of the interview. Except one participant whose husband verbally consented to provide the above information, all the participants freely provided the data related to their pregnancy outcome, their health status and the child condition.

Regarding the outcome of the pregnancy, 79.7% (110/138) of the participants delivered live newborns without disability and 1.4% (2/138) of the newborns presented anomalies at the delivery. Stillbirth was recorded in 4.4% (6/138) of the participants. A perinatal mortality rate of 14.5% (20/138) was reported with respectively 6.5% fetal and 8.0% neonatal deaths (Table 3).

Table 3. Pregnancy outcomes, mother and children health status at time of interview and ultrasound screening results (n = 138).

	Ultrasonography results					
Variable	**Neural tube**	**Other congenital**	**No congenital**	**Total defects**	**Total**	**%**
Pregnancy outcome						
Alive without apparent disability	12	7	91	19	110	79.7
Alive with a disability	0	0	2	0	2	1.4
Fetal death	3	5	1	8	9	6.5
Neonatal death	3	4	4	7	11	8.0
Stillbirth	0	4	2	4	6	4.3
Total	**18**	**20**	**100**	**38**	**138**	
Maternal health status						
Unwell	0	1	2	1	3	2.2
Well	18	19	98	37	135	97.8
Total	**18**	**20**	**100**	**38**	**138**	
Child health status						
Alive and well	5	5	86	10	96	69.6
Alive with complication	4	2	4	6	10	7.2
Deceased	9	13	10	22	32	23.2
Total	**18**	**20**	**100**	**38**	**138**	

At the time of the interview, 97.8% (135/138) of the participants were in good health; of the three participants who reported not being healthy, their ultrasonography screening had not detected any congenital defect in two and one was diagnosed with a congenital defect other than NTDs. Concerning the children, a mortality rate of 23.2% (32/138) was reported; the ultrasonography screening was normal for 31.3% (10/32) and congenital defects were diagnosed in 68.8% (22/32). 7.2% (10/138) of the children lived with complications. Overall, only 69.6% (96/138) of the children were living healthily at the time of interview.

4. Discussion

Of the 138 cases that underwent ultrasound examination in our study, the prevalence of congenital structural defects was estimated at 2.2/10,000 live births (range: 0.3 - 7.4/10,000). This prevalence was comparable with the 7.2% and 2.5% reported in the literature [3] [11]. Neural tube defects were the most prevalent congenital defects (13.0%); this was in line with published data [10] and contradicted those publications [8] [9] where the most prevalent anomalies were congenital heart defects. As it is known that folic acid treatment in the first trimester can prevent neural tube defects, our findings indicated that the antenatal care provided in primary health care settings should be enhanced by a health promotion message emphasizing the importance of taking folic acid supplements.

It has been suggested that congenital malformations may emerge in the first trimester of pregnancy as a result of genetic aberrations or gene-environment interaction. The etiology is predominantly multifactorial, arising from complex gene-environment interactions that impair normal embryo-fetal development. Environmental factors (such as chemical toxins, infectious agents, maternal sickness, and exogenous factors) can have pre-conceptional mutagenic, post-conceptional teratogenic, peri-conceptional endocrine disruption and epigenetic effects [11]. Unfortunately, our research did not investigate the impact of environmental factors in our country where irrigated farming using pesticides is widely practiced. Another limitation of our research was the convenient sampling technique used due to the inaccessibility of the entire database of ultrasound records available. However, as an explorative study, the findings revealed that neural tube defects, preventable through supplementation, were the predominant anomaly. The use of spatial distribution software for mapping health conditions facilitated the visualization of locations with the highest concentration of affected population, hence enhancing relevant stake-holders ability to respond promptly. Healthcare providers are urged to promote maternal health, incorporate ultrasonography screening into routine antenatal care and support pregnant women with abnormal fetal outcomes.

Availability of Data and Materials

The data are available upon request at any time needed from the corresponding

author.

Funding

The research was fully funded by the researcher Osman, M.A.M.A. and Elhassan, I.A.A. in the course of their field research related to their thesis for the diploma in Research Methodology and Biostatistics.

Authors' Contribution

1) Osman, M.A.M.A.: Designed and implemented the research as well as the data management and drafting of the manuscript.

2) Elhassan, I.A.A.: Contributed to the implementation of the research and the data management, and reviewed the draft manuscript.

3) Alimmam A.: Facilitated access to the ultrasound reports.

4) Noma, M.: Supervised the data management, orientated the GIS analysis and edited the final manuscript.

5) Fazari, A.: Designed and contributed to drafting, and edited the final manuscript.

6) Khan, F.N.: Designed and contributed to drafting, and edited the final manuscript.

All the authors read and approved the final manuscript prior to submission.

Acknowledgements

We acknowledge Dr. Durreya Alrayah, and her staff of the Habib Fetomaternal Unit of Soba University Hospital, with special thanks to the nurses for their support. The University of Medical Sciences and Technology for guidance in the framework of the Diploma in Research Methodology and Biostatistics, in particular Prof. Humeida and Dr. Hanan Tahir. We are also greatly in debt to our Batch Diploma colleagues in particular Dr. Al Nazeer and Ms. Maria for their assistance.

Conflicts of Interest

The authors declare no conflicts of interest regarding the publication of this paper.

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