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Research Article

Prevalence of Congenital Anomalies Detected by Antenatal Ultrasound

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Abstract

Objective: This study aims to provide statistical distribution of different types of congenital anomalies prevalent in hospitals of Al-Baha city.

Materials and Methods: This study followed a retrospective design for reviewing 267 pregnant women files who underwent an ultrasound scan in both King Fahad Hospital and Prince Meshari Hospital during the time period from January 1, 2005 to December 31, 2018.

Results: It showed that 19.9% participants had a first-degree family history of congenital anomalies, 70.8% were consanguineous, and 29.2% were non-consanguineous. Anomalies of urinary system (41.6%) were most common, followed by anomalies of the CNS (34.1%) and anomalies of circulatory system (19.9%). Most common reason for terminations was congenital anomalies related to digestive system. Therefore, prenatal counselling and antenatal screening should be encouraged to increase the survival rate of infants.

Conclusion: The knowledge of the involvement of both consanguinity and first-degree family history of anomalies in cases of congenital anomalies can also be useful in designing future health awareness endeavors.

Keywords: *Congenital Anomalies; Consanguinity; Family History; High Maternal Age; NICU Admission*

Introduction

A congenital anomaly (CA) is defined as a metabolic disorder that occurs within the uterus and is often detected at the time of birth. Such anomalies result from the defects in the parental genomes and are considered harmful as they negatively impact the developmental process of a child. According to the information provided by Mahela and Talukdar [1], 276,000 infants, out of 3 million reported births, die within a month on account of congenital anomalies. As per the report of World Health Organization [2] each year, 303,000 babies are born with frequently lethal congenital anomalies. Down syndrome [2], heart defects [3] and the abnormalities of the central nervous system (CNS) [4] are some of the most common congenital anomalies. The ratio of the prevalence of the congenital anomalies in Saudis was 412:10,000 [5].

In many regions, these anomalies are detected in the second trimester [6]. However, the prevalence of these anomalies is mostly related to family history, paternal consanguinity and low birth weight in Saudi Arabia [7]. The high prevalence of genetically determined disorders

is attributed to many factors; for instance, the high consanguinity rate in Arab societies may be the most important factor [8]. The study of Al Salloum., *et al*. [9] revealed that the prevalence of congenital abnormalities in infants often results in Down syndrome.

Congenital anomalies are further classified into two types: single system malfunction and multiple system malformations [10]. Vyas., *et al*. [11] investigated the prevalence of different types of congenital anomalies and the findings of the study revealed that the overall percentage of the occurrence of congenital malformations is 1.23% (12.3/1000 births), and that the congenital anomalies of central nervous systems were the most common. Another report released by the Global Burden of Disease Study indicated that congenital anomalies claim the lives of nearly 510,400 every year [12].

Based on the above-mentioned facts and figures, the study aims to provide the statistical distribution of different types of congenital anomalies prevalent in the two largest hospitals of Al-Baha city during the time period from January 2005 to December 2018. The study involved review of the records of antenatal ultrasounds carried out during the aforementioned time period. It is strongly hoped that the findings of this study will be beneficial for medical experts as the data collected and analyzed in this study focus on the occurrence of congenital anomalies and diagnosis of congenital anomalies which is an important factor in the timely provision of remedial treatment. Remedial treatment of mothers is very important as it possibly protect infants from the adverse effects of the problem. Besides, the findings of the current may also help women in general and pregnant women of Al-Baha in particular to realize the importance of timely check-ups during pregnancy.

Materials and Methods

This study followed a retrospective design which reviewed files of pregnant women who underwent an ultrasound scan in both King Fahad Hospital and Prince Meshari Hospital during the time period from January 1, 2005, to December 31, 2018. This chart review was carried out in the medical record department of each hospital. Informed consent was duly obtained from the participants before they took part in the study. The study was conducted in accordance with the Helsinki Declaration, and the study protocol was also approved by the Ethics Committees of Ministry of Health and Al-Baha University (2378/r/19). To enhance ethical aspect of the study, the collected data was stored in a password-protected computer in order to limit its accessibility to the researcher only to ensure confidentiality of the data.

The study sample consisted of 267 women but 8.9% of them missed the follow-up; however, their data was included in the analysis where available. The mean maternal age was 25.8 ± 7.6 years and ranged between 17 to 45 years. The median score for parity was 2 ± 3, ranging from 0 to 12 and 11% (29) women were primigravida. Patients suffering from congenital anomalies were included in the study sample; however, any patient with a documented non-viable fetus at the time of diagnosis was excluded from the study sample. Likewise, scans that showed a viable fetus with an isolated soft marker were also excluded.

The retrieved data included the following variables: maternal parity, maternal age, gestational age at the time of diagnosis and delivery, diabetic status, family history of anomalies and consanguinity. Additionally, data describing the type and number of anomalies such as (CNS, Eye, Ear, Face, Neck, Circulatory, Respiratory, Cleft Lip and Palate, Digestive, Genital, Urinary, Musculoskeletal) were collected according to the (ICD-10) criteria and the results of karyotyping when available. Documented neonatal outcomes included survival that was defined as signs of life at the time of birth: newborn was declared alive and had an APGAR score after birth. The rate of NICU admissions was also documented to determine whether a definitive diagnosis or anomaly was clear after birth or not.

Simple descriptive statistics via SPSS software, version 24 was used to analyze the collected data. Quantitative variables were summarized as either Mean and Standard Deviation for normally distributed data or Median and 25th and 75th Percentile for skewed data; the qualitative variables were calculated in terms of frequencies and percentages.

Results

Table 1 shows the demographic characteristics of the participants. The main maternal age of consanguineous and non-consanguineous couples was 24.8 and 26.1 years respectively. 70.8% (189) participants were consanguineous, while 29.2% (78) participants were non-consanguineous. 19.9% (53) participants had a first-degree family history of congenital anomalies, while the percentages of consanguineous and non-consanguineous couples with first-degree family history of congenital anomalies were 19.6% and 20.5% respectively.

	Participants 267 (100%)	Consanguineous couples 189 (70.8%)	Non-Consanguineous couples 78 (29.2%)	P
Maternal age Mean (range)	25.8 (17 - 45)	24.8 (17 - 42)	26.1 (18 - 45)	0.063
17-25	115 (43.1%)	79 (41.8%)	36 (46.2%)	
> 25 - 35	62 (23.2%)	43 (22.8%)	19 (24.3%)	
> 35 - 45	90 (33.7%)	67 (35.4%)	23 (29.5%)	
Parity	2 (0 - 12)	2 (0 - 10)	2 (0 - 12)	0.09
Primigravida	29 (10.9%)	18 (9.5%)	11 (14.1%)	
Para 1-3	172 (64.4%)	123 (65.1%)	49 (62.9%)	
Para 4-7	49 (18.3%)	40 (21.2%)	9 (11.5%)	
Para 8-12	17 (6.4%)	8 (4.2%)	9 (11.5%)	
Family history of congenital anomalies	53 (19.9%)	37 (19.6%)	16 (20.5%)	-

***Table 1**: Demographic characteristics of participants.*

Table 2 shows the proportions of the prevalence of each anomaly from ICD 10 anomalies with comparison between consanguineous and non-consanguineous couples. Among the most common congenital anomalies, anomalies of the urinary system were the most frequent (41.6%). The second highest frequency was of the congenital anomalies of the CNS (34.1%), followed by the anomalies of the circulatory system (19.9%). The results also showed that non-consanguineous couples did not suffer from anomalies of eyes, ears, face, neck, respiratory system, cleft lip and palate, digestive system, genital organs, musculoskeletal system and chromosomes (p-value < 0.05).

Type of Anomalies (ICD 10)	Frequency for all participants 267	Consanguineous couples 189	Non- Consanguineous couples 78	P
CNS	91 (34.1%)	64 (33.9%)	27 (34.6%)	0.09
Eye, ear, face, neck	1 (0.4%)	1 (0.5%)	0	0.05
Circulatory	53 (19.9%)	37 (19.7%)	16 (20.5%)	0.074
Respiratory	1 (0.4%)	1 (0.5%)	0	0.02
Cleft lip and palate	1 (0.4%)	1 (0.5%)	0	0.01
Digestive	3 (1.1%)	3 (1.6%)	0	0.03
Genital	1 (0.4%)	1 (0.5%)	0	0.03
Urinary	111 (41.6%)	77 (40.7%)	34 (43.6%)	0.89
Musculoskeletal	3 (1.1%)	3 (1.6%)	0	0.01
Chromosomal	1 (0.4%)	1 (0.5%)	0	0.05
Other	1 (0.4%)	0	1 (1.3%)	0.08

***Table 2**: Prevalence of each anomaly of the 10th ICD categories.*

Table 3 shows the data related to the fetal outcomes for each of the 10th ICD categories. Among them, 23.1% babies survived and 68.1% were admitted in NICU, who were delivered at 37+1.6 weeks with CNS related anomaly, and the termination rate of 31.9%. No case of termination was reported for the congenital anomalies of eyes, ears, face, and neck, respiratory system, and cleft lip and palate (p-value > 0.05). However, termination was reported in case of chromosomal anomaly. Majority of the terminations (66.7%) were reported in the cases of congenital anomalies of the digestive system.

Type of anomaly	GA at Delivery (median) weeks				Baby Survived				NICU admission				Termination			
	T	C	NC	P	T	C	NC	P	T	C	NC	P	T	C	NC	P
1- CNS	37 + 1.6	37	37 + 1.5	0.06	23.1% (21/91)	25% (16/64)	18.5% (5/27)	0.89	68.1% (62/91)	75% (48/64)	51.9% (14/27)	0.98	31.9% (29/91)	31.3% (20/64)	33.3% (9/27)	0.60
2- Eye, ear, face, neck	38	38	-	0.98	100% (1/1)	100%	-	0.72	100% (1/1)	100%	-	0.05	0 %	0%	-	0.07
3- Circulatory	35 + 3.1	34 + 2.2	38	0.05	50.9% (27/53)	51.4% (19/37)	50% (8/16)	0.70	50.9% (27/53)	51.4% (19/37)	50% (8/16)	0.06	1.9% (1/53)	2.7% (1/37)	0%	0.07
4- Respiratory	31	31	-	0.06	100% (1/1)	100%	-	0.60	100% (1/1)	100%	-	0.07	0 %	0%	-	0.09
5- Cleft lip and palate	39	39	-	0.07	100% (1/1)	100%	-	0.07	100% (1/1)	100%	-	0.09	0%	0%	-	0.09
6- Digestive	38 + 3.7	38 + 3.7	-	0.09	0%	0%	-	0.07	33.3% (1/3)	33.3% (1/3)	-	0.09	66.7% (2/3)	66.7% (2/3)	-	0.50
7- Genital	26	26	-	0.09	0%	0%	-	0.09	0%	0%	-	0.89	100% (1/1)	100%	-	0.07
8- Urinary	39 + 2.1	38 + 1.6	38 + 1.5	0.06	98.2% (109/111)	97.4% (75/77)	100% (34/34)	0.09	11.7% (13/111)	14.3% (11/77)	5.9% (2/34)	0.72	1.8% (2/111)	2.6% (2/77)	0%	0.40
9- Musculoskeletal	37 + 2.1	37 + 2.1	-	0.07	66.7% (2/3)	66.7% (2/3)	-	0.50	66.7% (2/3)	66.7% (2/3)	-	0.70	33.3% (1/3)	33.3% (1/3)	-	0.06
10- Chromosomal	30	30	-	0.09	0%	0%	-	0.07	0%	0%	-	0.60	100% (1/1)	100%	-	0.98
11- Other	25	-	25	0.93	0%	-	0%	0.40	0%	-	0%	0.50	100% (1/1)	-	100%	0.05

***Table 3**: Total participants fetal outcomes for each of the 10th ICD categories.*

Discussion

According to the findings of the current study, congenital anomalies of the urinary system were the most frequently diagnosed anomalies, followed by congenital anomalies of the CNS and the circulatory system respectively. These findings are relatively in line with the similar study conducted in Riyadh [8]. Table 2 provide details of the associated obstetric and fetal outcomes for each of the eleven ICD 10 categories. The anomalies of the urinary system include conditions such as renal agenesis, renal pylectasia, renal cysts, hydronephrosis, etc. However, individuals suffering from these anomalies were the least likely to suffer from major adverse outcomes, and, therefore, the highest fetal survival rate and the highest median GA at delivery was reported in cases of such anomalies, despite the fact that these anomalies occurred most frequently (41.6%). Besides, these anomalies were also associated with the lowest rate of termination of pregnancy and need for NICU admission. These findings are in line with the findings of the previous studies [9] and imply that urinary anomalies detected in the ultrasound scan can be expected to have a relatively benign course of development despite their high frequency of

occurrence. Notably, the second most frequently recorded type of congenital anomalies were associated with the CNS (34.1%). However, they predicted a largely negative course of development and fetuses with such anomaly were the most susceptible to the need of NICU admission. Moreover, these anomalies were also associated the highest total number of terminated pregnancies and deaths following birth. These findings are also in line with the findings of the previous studies [8,13].

Congenital anomalies of the circulatory system were the third most frequently recorded (19.9%) congenital anomalies. These anomalies showed intermediate spectrum of severity in comparison with the milder urinary and more severe CNS groups which may be due to the fact that such anomalies are less likely to be lethal, possibly due to a higher amenability to surgical and medical therapy, even in cases in which major structural heart defects are detected. The study of Anyanwu, Danborno, Hamman [14]; however, provided contradicting results; their findings revealed that the prevalence of congenital anomalies mostly causes abnormalities in the central nervous system and other affected organs include genitourinary system, gastrointestinal system, mouth and musculoskeletal system. Similar results were also obtained in other studies [11,12], which revealed that out of 13,614 births, almost 167 infants were diagnosed with congenital malformations. However, the most adverse effects of congenital anomalies were detected in the central nervous system (CNS). The results are in contrast with those obtained in the current study.

The American College of Obstetricians and Gynecologists (ACOG) and The International Society of Ultrasound in Obstetrics and Gynecology (ISUOG), suggest at least one routine mid-trimester fetal ultrasound scan for the general population [15,16]. Therefore, it may be inferred that when a physician encounters an abnormal antenatal CNS scan, he must inform the patient about the high likelihood of complications associated with this diagnosis. Thus, this strategy may serve as an opportunity to educate and alert the involved families and caregivers to follow the pregnancy more closely. Mothers often voluntarily miss out the early antenatal visits either out of complacency or poor health awareness. Other reasons behind this trend are over-crowded clinics and long traveling time required to reach medical centers from peripheral towns and villages. However, the findings of this study help us affirm with surety that delay in the early diagnosis surely increases the risks of morbidity and mortality of both mother and fetus. According to the previous studies, the reported median gestational age at diagnosis was 31 weeks, while this study revealed that it was 22 weeks, and this could be considered as an encouraging sign indicated by this study [8].

Advanced maternal age is a recognized risk factor for the occurrence of a multitude of congenital anomalies. In a study conducted by WHO which included 65,000 pregnancies in the US, the proportion of mothers with over 34 years of age was 20% in the major anomalies arm. The mean maternal age in this study was 27 years and the proportion of mothers with over 34 years of age was only 14%. Therefore, it can be inferred that maternal age cannot be recommended as a dependable marker to rule out fetal anomalies. Estimates of the prevalence of consanguinity in the Middle East consistently show an elevated level. The high consanguinity rate (70.8%), documented in this study could be a by-product of the selection of a population with positive findings. On the other hand, first-degree family history of anomalies was extremely prevalent (19.9%). This leads to a presumption that public awareness about the risk of congenital anomalies and adverse fetal outcomes associated with consanguinity and family history is still severely low even in adversely affected families [8,9].

Study Limitations

The results of study are limited since it was a retrospective study and did not include a control group to make a baseline comparison. This limitation makes it difficult to ascertain the extent to which the recorded risk factors contributed to the likelihood of increased morbidity or mortality. Therefore, future studies should carry out a cross-sectional analysis and should include a control group. Future studies should also focus on carrying out a comparison between different types of congenital anomalies in couples with and without consanguinity. The study also lacks a set of documented variables to cover the complete spectrum of possibly implicated etiologies: tobacco exposure, nutritional deficiencies, or environmental pollution. Future studies should also focus on clearing this contentious area.

Conclusion

Congenital anomalies occur worldwide and cause perinatal and infant deaths and postnatal physical disabilities. The study concluded that anomalies related with the urinary system were the most frequently diagnosed congenital anomalies, followed by anomalies of CNS and circulatory anomalies. Based on these findings, this study recommends that prenatal counselling and antenatal screening should be conducted especially in cases of consanguinity and the cases with family history of anomalies. This strategy may reduce the prevalence of congenital anomalies and facilitate more timely interventions. Areas of strength in this study include the documentation of hundreds of cases of congenital anomalies detected by ultrasound screening and description of their nature and ensuing complications. Furthermore, the study findings provided a summary of the baseline characteristics of the studied population which can be used by practitioners to familiarize themselves with the common patterns they may encounter during their clinical practice. In addition, the study findings may aid in enlightening the families about the outcomes that may arise from the diagnosis of congenital anomalies. As revealed in the study finding, the knowledge of the involvement of both consanguinity and first-degree family history of anomalies in cases of congenital anomalies can also be useful in designing future health awareness endeavors.

Competing Interest

The author declares no competing interest.

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