ORIGINAL RESEARCH PAPER

Volume-7 | Issue-2 | February-2018 | PRINT ISSN No 2277 - 8179

INTERNATIONAL JOURNAL OF SCIENTIFIC RESEARCH

INCIDENCE OF CONGENITAL ANOMALIES DURING PREGNANCY AND ASSOCIATED RISK FACTORS AT UNIVERSITY HOSPITAL IN JEDDAH

Radiology

Awad Elkhadir	Diagnostic Radiology Department, Faculty of Applied Medical Sciences (FAMS), King Abdulaziz University (KAU), Jeddah, Saudi Arabia
Laila Ashkar	Department of Diagnostic Radiology, King Abdulaziz University Hospital (KAUH), Jeddah, Saudi Arabia
Bander S Almutairy	Department of Diagnostic Radiology, King Abdulaziz University Hospital (KAUH), Jeddah, Saudi Arabia
Bashair Alhidri	Diagnostic Radiology Department, Faculty of Applied Medical Sciences (FAMS), King Abdulaziz University (KAU), Jeddah, Saudi Arabia
Bayan Alsaud	Diagnostic Radiology Department, Faculty of Applied Medical Sciences (FAMS), King Abdulaziz University (KAU), Jeddah, Saudi Arabia
Esraa Jadallah	Diagnostic Radiology Department, Faculty of Applied Medical Sciences (FAMS), King Abdulaziz University (KAU), Jeddah, Saudi Arabia
Samaher Binmahfouz	Diagnostic Radiology Department, Faculty of Applied Medical Sciences (FAMS), King Abdulaziz University (KAU), Jeddah, Saudi Arabia

ABSTRACT

Background: Several types of congenital fetal malformations (CMs) can be detected in pregnant ladies by ultrasound (U/S). Socioeconomic, demographic and genetic factors are to be considered the most influential risk factors.

Objective: To assess the frequency and nature of CMs, as well as the associated risk factors during conception, among pregnant ladies at King Abdulaziz University Hospital (KAUH) during the study period.

Material and Methods: From 2012-2015 a total of 9352 U/S were performed at KAUH in pregnant ladies. Data was collected retrospectively from the patient's records in the radiology information system regarding the risk factors and analyzed for the type and frequency of CMs.

Results: 9352 U/S reports were surveyed. Of 70 foetuses with CMs sonographically detected, the incidences of head/neck was 61.42%, urinary system 20%, limbs 7.14%, pelvic/spine 4.29%, abdominal wall 4.29% and cardio respiratory system was 2.86%. Nationality, consanguinity marriage, family history and maternal age constituted the most common risk factors. Consanguineous marriage was more common among Saudi citizens; 74.3% compared to non-Saudi 25.7%.

Conclusion: Several types of fetal CMs were detected in pregnant ladies at KAUH -Jeddah, in particular among Saudi citizens during the study period. Socioeconomic, demographic and genetic factors are influential risk factors that need to be considered during the antenatal visits and history taking.

KEYWORDS

Congenital malformations, pregnant, U/S, consanguineous marriage.

Introduction:

According to the World Health Organization (WHO), congenital anomalies are also known as birth defects, congenital disorders or congenital malformations (CMs) and can be defined as structural or functional anomalies (e.g. metabolic disorders) that occur during intrauterine life as well as can be identified prenatally, at birth or later in life [1]. Stuart Campbell produced the first sonogram of a congenital anomaly in the human foetus in 1972 [2]. It was found that CMs appear in 3% of all infants [3,4]. Almost more than quarter will result in perinatal death [5,6]. When ultrasound (U/S) techniques became sufficiently powerful, many countries introduced U/S screening to detect these anomalies during the second trimester [7,8]. In this study we will concentrate on all types of CMs that were detected at King Abdulaziz University Hospital (KAUH) in Jeddah, Saudi Arabia during the past four years.

The prevalence and different kinds of CMs vary from one nation to another, even in the same nation from one area to another. This depends on the definition of CMs utilized; procedure of their detection, trimester and the characteristics of the population under exam including their different ethnics and socio-economic backgrounds [9,10]. Based on WHO reports, about 3 million foetus and infants are born each year with major malformations [10,11]. The influence of these CMs on the newborn and foetus is great as they are responsible for 495,000 deaths worldwide [11] and thus contribute mostly to infant mortality rate. Unfortunately, there is also a lack of awareness by both families and the health authorities of the importance of genetic counselling in the prevention of CMs [12]. At this moment, no nationwide birth defect monitoring record exists in the Kingdom of Saudi Arabia (KSA).

Only a small number of published information is available from online peer review medical journals about the prevalence of the CMs among live births and still births. We were cited of these to support this research of ours.

Material and Methods:

A retrospective study and cross sectional study design was conducted in the Jeddah region of KSA at KAUH. Data was collected from the files of all pregnant female patients scanned by U/S for four consecutive years. Collection of data started on August 2015 and finished on February 2016 after we got the ethical approval from the hospital. *In spite of KAUH* having a unique hospital information system, we couldn't obtain *some data with regards to* consanguineous marriages, so we contacted the scanned pregnant ladies to get some important information such as consanguinity and family history. We were able to reach 59% (48/70) of them.

Nine thousand three hundred fifty two obstetric U/S reports, during the period from January 2012 and December 2015 were reviewed, to identify sonographic findings of CMs using patient's information recording system (Sectra). Out of 9352 reports, a total of 70 (0.7485%) maternal data was collected, because only those subjects have fulfilled the criteria of our needed outcome measures recorded for our study. Gathered data was analyzed by SPSS to measure the relationship between the incidence of CMs and related variables such as consanguinity marriage, mother's age, family history, nationality and if there is any other influential risk factors over the past four years**.** Analysis included frequency, percentages and the chi-squared test was used to compare variation between the data. Statistical significance was acceptable at P value <0.05.

Results

Among the total of nine thousands and three hundred fifty two foetuses studied during the period (2012–2016) there, were seventy foetuses (0.7485%) that had CMs. Our study showed that the incidence of CMs in consanguinity marriages was confirmed in 28.6% of the cases while 37.1% didn't shown any consanguineous marriage and in 34.3% of the cases the data was missing (Table 1). According to the risk factor of family history, our study revealed 12.8% affected by this risk, 52.9% were not and 34.3% of the cases couldn't be contacted (Table 1). Distribution of maternal age group reported ages between 30-39 years 48.6%, less than thirty years 41.4% and more than forty years 10% (Table 1). Regarding the nationality 74.3% were Saudi and 25.7% were non-Saudi (Table 1).

It's worth noting that our study showed that the significant risk factors associated with our samples were consanguinity marriage, family history, age and nationality ($p < 0.05$) (Table 1). The incidences of CMs were categorised into head/neck 61.42% (43/70), urinary system 20% (14/70), limbs 7.14% (5/70), pelvic/spine 4.29% (3/70), abdominal wall 4.29% (3/70) and cardio respiratory system 2.86% (2/70) (Table 2). Generally, the most frequent CM diagnosed during this study was hydrocephalus (Figure 1). Figure 2 demonstrated CMs in the cardio respiratory system.

Table 1. Risk factors of CMs through 70 foetuses.

Risk factor	Yes		No		Inaccessible		Total		P-value
Consanguinity marriage	n	%	N	%	n	%	N	%	
	20	28.6%	26	37.1%	24	34.3%	70	100.0%	.000
Family history	9	12.8%	37	52.9%	24	34.3%	70	100.0%	.000
Age groups of mother	Less than 30 years		30-39 years		More than 40 years		70	100.0%	.000
	N 29	41.4%	N 34	48.6%	N 7	10.0%			
Nationality	Saudi		non-Saudi		-		70	100%	.000
	N 52	74.3	N 18	25.7%	-				

Table 2. Site of Cms.

Area	N	%
Limbs	5	7.14%
Pelvis / spine	3	4.29%
Head / neck	43	61.42%
UT	14	20%
Respiratory system / CVD	2	2.86%
Abdominal wall	3	4.29%
Total	70	100%

Discussion:

Deformities, abnormalities and the demise of embryos before birth are caused by many intrinsic and extrinsic factors. Abnormalities that occur as a result of genetic mutations, aneuploidies and translocation are known as malformations [13]. There is no doubt that differences in the reported prevalence rates of CMs over time and among countries, or even within the same country, may depend on several factors, such as study design, classification and inclusion criteria and accuracy of diagnosis [14,15].The overall incidence of CMs in this study was(0.7485%) foetuses. The study reviewed the incidence of CMs that occurred in pregnant ladies over the last four years and the factors that contribute in it occurrence. In this study, CMs were mostly diagnosed in central nervous system (CNS)/oro-facial system (61.42%) followed by urinary system (20%), limbs (7.14%), pelvic/spine (4.29%), abdominal wall (4.29%) and finally cardio respiratory system (2.86%). These findings agreed with previous studies that showed a significant variation in incidences of CNS anomalies in different regions of the world including Europe (Barkovich, 2005). A previous study at KAUH by Fida etal, showed that the rate of birth defects in the musculoskeletal system was less than in the current study, which indicates that skeletal anomalies have risen [16]. Also, there was another research on birth defects in Canada that showed that the most prevalent CMs were in the musculoskeletal defects while in our research the most common anomalies were brain defects [17].

Regarding the most common defects of the cardiovascular system (CVD), our study detected 2.86% compared to Becker et al, [18] where they found a relatively higher proportion of atrioventricular septal defects (ASD's) and Tetralogy of Fallot (TOF's) than reported in other large epidemiological studies of more heterogeneous populations [19]. A high proportion of ASD cases had been reported at another center in KSA [20]. Our present study supports the effected prevalence rate of CMs and maternal age which was also showed in a study done in Egypt by R.M. Shawky etl [21,22]. It is widely acknowledged that consanguineous marriages are playing an important role in the occurrence of CMs in the Middle East [23]. In the present study, which included all nationalities, consanguineous marriage was significantly increased by 28.6% similar to Fida etal, 's study which demonstrated 27.2% in Saudi [16] and others report 38.9% [24]. Similarity was also reported in other Arab countries and in Iran [23,25,26,27]. Zlotogora [28], Pinto Escalante [29] and a study done in Egypt [30] also reported same results. Family history of CMs was reported to occur in 12.8 % in this study and was considered statistically significant ($p < 0.05$) and this also was more common among relatives of consanguineous marriages [27]. The reason of high incidence of CMs in Saudis compared to non-Saudis (74.3 % versus 25.7%), might be related to the high rate of consanguineous marriages among Saudis in KSA. Therefore further research needs to be conducted in this aspect.

Conclusion

In summary, CMs still remain a serious cause of mortality and morbidity in foetuses and even in children.It is important during our clinical practice to consider and document the mentioned socioeconomic, demographic and genetic factors during the history taking at the antenatal visits. As well, the frequency of the different types of fetal congenital anomalies need to be well known to the technologists and physicians performing the antenatal U/S studies, for accurate diagnosis.

Further researches need to be conducted to study these different types of malformations and their related risk factors to reduce or prevent the incidence of CMs. U/S is the first medical imaging tools to detect CMs, because it is affordable, speedy, safe and available everywhere [31,32].

Ethical statements

Ethical Research Committee (REC) approved to conduct this study with (reference No 316-15) - Retrospective study.

Conflict of Interest

The author(s) declare(s) that there is no conflict of interest.

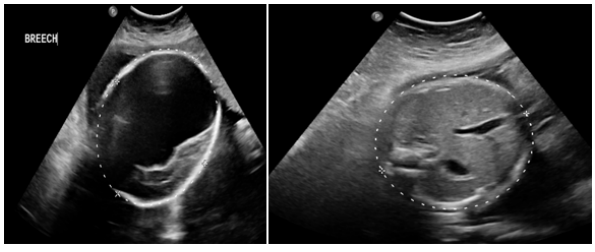

Figure 1: Single viable foetus in breech presentation with severe hydrocephalus.

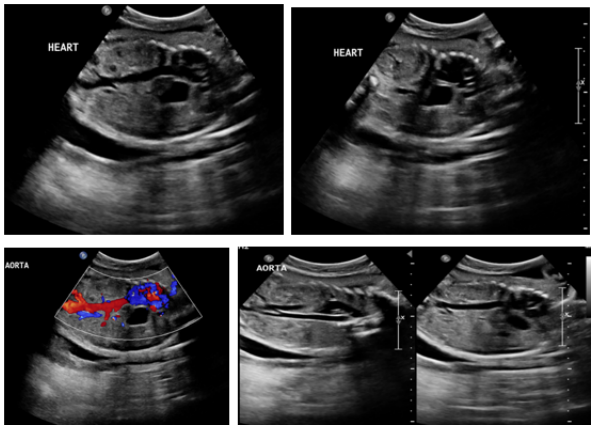

Figure 2: Cephalic presentation of single viable foetus with anomaly in the heart and dilated aorta and inferior vena cava.

References

[1] http://www.who.int/mediacentre/factsheets/fs370/en/.
[2] Campbell S, Johnstone FD, Holt EM, May P. Anencephaly: early ultrasonic diagnosis and active management. Lancet 1972; 2: 1226-1227.
[3] Reerink JD, Herngreen WP, Verkerk PH, Ruys JH, Verloove-Vanhorick SP. Congenital disorders in the first year of life. Ned. Tijdschr. Geneeskd. 1993; 137: 504-509.

[4] De Galan-Roosen AE, Kuijpers JC, Meershoek AP, van VD. Contribution of congenital malformations to perinatal mortality. A 10 years prospective regional study in the Netherlands. Eur. J. Obstet. Gynecol. Reprod. Biol. 1998; 80: 55-61.
[5] Chung CS, Myrianthopoulos NC. Congenital anomalies: mortality and morbidity, burden and classification. Am. J. Med. Genet. 1987; 27: 505-523.
[6] Prevalence of congenital malformations in the Northern Netherlands 1981-2006. 2007; http://www. rug. nl/umcg/faculteit/disciplinegroepen/MedischeGenetica/Eurocat/professionals/tabellen.
[7] Stoll C, Tenconi R, Clementi M. Detection of Congenital Anomalies by Fetal Ultrasonographic Examination across Europe. Community Genet. 2001; 4: 225-232.
[8] Smith-Bindman R, Hosmer W, Feldstein VA, Deeks JJ, Goldberg JD. Second-trimester ultrasound to detect fetuses with Down syndrome: a meta-analysis. jama 2001; 285: 1044-1055.
[9] Christianoon RE, Van Den Berg BJ, Milkovich WF, Oechsli L. Incidence of congenital anomalies among white, black live births with long-term follow-up. J Public Health 1981;71:1333–41.
[10] Aihw NP, Birch MR, Grayson N, Sullivan EA. Recommendations for development of a new Australian birth anomalies system: a review of the congenital malformations & birth defects data collection. AIHW Cat. No. PER 23, Sydney: AIHW National Perinatal statistics Unit (Birth Anomalies Series no.1); 2004.
[11] Eurocat working group. Eurocat Report 7, Scientific institute of Public Health Louis Pasteur, Brussels, 1997;
[12] A prospective study of congenital malformations among live born neonates at a University Hospital in Western Saudi Arabia By:Fida, NM (Fida, Nadia M.); Ai¬Aama, J (Ai¬Aama, Jumana); Nichols, W (Nichols, Waaa); Alqahtani, M (Alqahtani, Mohamed) SAUDI MEDICAL JOURNAL Volume: 28 Issue: 9 Pages: 1367¬1373 Published: SEP 2007 View Journal Information.
[13] Gilbert SF. Environmental regulation of animal development. In Developmental biology 5th edition. Sinauer Associate, Inc. Sunderland, Massachusetts, USA; 1997. p 805-841.
[14] Lechat MF, Dolk H. Registries of congenital anomalies.EUROCAT. Environ Health Perspect 1993; 101 (Suppl 2):153-157.
[15] International Clearinghouse for Birth Defects Monitoring Systems. Annual Report 2003. Rome, Italy: International Centre for Birth Defects; 2003.
[16] A prospective study of congenital malformations among live born neonates at University Hospital in Western Saudi Arabia.Fida et al. Saudi Med J 2007; Vol. 28 (9).Electronic ISSN 1658-3175. Print ISSN 0379-5284.Avilable on: http://smj.psmmc.med.sa/index.php/smj/article/view/5960.
[17] Theregionaldisparityofcongenitalanomaliesinsaskatechewananditsimpacto ntheutilizationofhealthservices.amylynnmetcalfe,august2007.
[18] Becker K, Seller MJ, Pal K, Davies AF. A 46, XX fetus with external female and internal male genitalia, facial dysmorphic features and mildly dilated lateral ventricles of the brain: a new syndrome? Clin Dysmorphol 2001; 10: 215-217.
[19] Fixler D, Talner N. Compilation of seven international studies in epidemiology of congenital heart disease. In: Oski FB et al, editor. Principles and practice of pediatrics. Philadelphia: J.B. Lippincott; 1994. pp 393.
[20] Jaiyesimi F. Pattern of congenital heart disease in King Fahd Specialist Hospital. Ann Saudi Med 1993; 13:407-411.
[21] Nazer HJ, Cifuentes OL, Aguila RA, Ureta LP, Bello PMP,Correa CF, Melibosky RF. The association between maternal age and congenital malformations. Rev Med Chil 2007;135(11):1463–9.
[22] The Egyptian Journal of Medical Human Genetics (2011) 12, 69–78 by Elsevier B.V. Peer review under responsibility of Ain Shams University. doi:10.1016/j.ejmhg.2011.02.016
[23] Hudgin L, Cassidy SB. Congenital anomalies. In: Marten RJ, Fanaroff AA, Walsh MC, editors. Neonatal – perinatal medicine. Philadelphia: Mosby – Elsevier; 2006. p. 561–86.
[24] Khayat RG, Saxena PC. Consanguinity and its effect on infant and child mortality in Egypt. http://ussp2005princeton.edu.
[25] Ali A, Zahad S, Masoumeh A, Azar A. Congenital malformations among live births at Arvand Hospital, Ahwaz, Iran – a prospective study. Pak J Med Sci 2008;24(1):33–7.
[26] Al-Ghazali LI, Dawodu AH, Sabarinathan K, Varghese. The profile of major congenital abnormalities in the United Arab Emirates (UAE) population. J Med Genet 1995;32:7–13.
[27] Mosayebi Z, Movahedian AH. Pattern of congenital malformations in consanguineous versus nonconsanguineous marriages in Kashan, Islamic Republic of Iran. East Mediterr Health J 2007;13(4):868–75.
[28] Zlotogora J. Genetic disorders among Palestinian Arabs: effects of consanguinity. Am J Med Genet 1997;68:427–35.
[29] Pinto Escalante D, Castillo Zapata I, Ruiz Allec D, Ceballos Quintal JM. Spectrum of congenital malformations observed in neonates of consanguineous parents. An Pediatr (Barc) 2006;64(1):5–10.
[30] Shawky RM, El-Baz FM, Elsobky ES, Osman A, Elsayed SM. High resolution cytogenetic study of patients with multiple congenital anomalies, M.D. Thesis, Ainshams University, 2005.
[31] Rocha SMS, Oliveira IRS, Widman A, Chisman BSK, Fukushima JT, Oliveira LAN, et al. Hepatometria ultrasonografica em criancas: proposta de novo metodo. Radiol Bras 2003; 36: 63¬70.
[32] Vazozzo DCP, Rocha DC, Cerri GG. Ultra¬sonografia abdominal. la ed. Sao Paulo:Sarvier;1993.