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Frequency of Birth Defects and its Relationship to Parents Having Interfamily Marriages at a Tertiary Care Hospital

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Abstract

Background: Inter family cousin marriages carries a risk of increased birth defects. The exact contribution of interfamily to birth defects risk is controversial. The aims of this study were to determine the frequency of birth defects in in relation to interfamily or outside family marriages.

Methods: This cross sectional study was conducted in the Civil hospital Sukkur from 9th November 2013 to 13th December 2015. Mothers giving birth to babies married in interfamily or outside family with their consent obtained were included in the study. Mothers not giving consent for study and having any known major illnesses were excluded from this study. Data was analyzed using SPSS version 21.0.

Results: The mean age of enrolled participants is 26.72 ± 7.07 years, the mean years of marriage was 5.95 ± 6 years and 49.4% were residents of urban area. Overall 89.9% were Muslims, 8.4% Christians and 2.8% Hindus, 60.7% infant had gestational age of <37 weeks. Overall 11.4% of newborns have congenital malformations. Frequency of congenital malformations among interfamily marriages was 15.6% compared to outside family marriages accounted for 3.7% cases (p=0.021).

Conclusion: It was concluded from this study that congenital malformations are common among participants of interfamily marriages.

Keywords: Congenital malformations; Interfamily marriages; Outside family marriages

Introduction

Birth defects are presenting as gross physical, hidden inside body as well as defects at cellular level like enzymes that present at birth resulted in physical and /or mental disabilities [1]. The underlying etiology of birth defects is multifactorial which include genetic factors, maternal age, and intake of drugs, exposure to teratogenic agents or radiation, maternal illness/infection, smoking, interfamily and alcohol consumption. A birth defect can be single or multiple, live threatening or non-life threatening [2]. In developed countries, the proportion of birth defects is rising due to good infection control and low prevalence of under nutrition [3,4]. The reported prevalence of birth defects varies in different geographic areas, ethnicities and populations [5]. In Pakistan, about 6% to 9% perinatal deaths are attributed to birth defects [6]. Common birth defects are neural tube defects, congenital heart diseases, gastrointestinal defects, genitourinary defects, face defects, musculoskeletal defects and ear anomalies [7] Long-term survival of children with birth defects is significantly lower than normal children [8].

Inter-family cousin marriages carry a risk of increased birth defects. Most of the published literature has reported the effect of interfamily marriages on birth defects as causative. However, risk factors like gestational diabetes mellitus (GDM), parity, socioeconomic strata, maternal age and literacy, intake of the teratogenic drug in the first trimester of pregnancy, and peri-conception chemical exposure. Scientist around the world has different schools of thoughts one school of thought believes that consanguinity produces specific types of birth defects [9-11] and others believe that the contribution of consanguinity to the etiology of major birth defects has been overestimated [12].

Interfamily marriages between first or second cousins are very common in Sindh, Pakistan [10], where interfamilial unions account for 20% to 50% of all marriages. First and second cousins marriages are favored by most of the families. Another study reported that rate of birth defects by interfamily marriages was 7% whereas in outside family marriages it was recorded to be 2% [13].

The exact contribution of interfamily to birth defects risk is controversial because we lack reliable local data and many studies did not take account of confounding factors and interactions with other exposures at the time of analysis. Accordingly, we would like to conduct this study because it addresses the issues related to birth defects and shows practical/public health relevance in societies with interfamily marriages vs. outside family marriages.

Interfamily first and second cousins marriages are prevalent in most of the families of Sindh because of social, cultural and other reasons. The exact contribution of interfamily marriages to birth defects risk is controversial because we lack reliable data. The present study addressed the frequency of birth defects in interfamily vs. outside family marriages.

Material and Methods

This was a cross sectional study conducted in Civil Hospital Sukkur from 9th November 2013 to 13th December 2015. Newborn delivered in the Civil Hospital Sukkur were screened for gross congenital malformations. Infants were enrolled after taking informed consent from mother. Mothers refused to participate were excluded from the study. Detailed history was taken with emphasis on age of mother, gestational age, duration of marriage, inter-family marriage and antenatal detection of congenital defect. Detailed physical examination of newborn was carried out. Anthropometric measurements (weight, length and fronto-occipital circumference) were also taken. Two readings were taken and average was taken for the analysis. Examination of eye, mouth, ear, facial features, cardiovascular system, gastrointestinal system, musculoskeletal system, abdomen, chest and genitourinary system was done. The findings were recorded in a proforma.

Statistical analysis

Sample size was collected through non-probability purposive sampling. A total of 450 mothers were included in this study. Data was analyzed by using SPSS 21.0 (IBM, incorporation, USA). Frequencies and percentages were presented for categorical variables like gender, interfamily marriage and congenital malformations. Mean ± SD was reported for continuous variables. Chi square test or Fisher exact test was applied for comparison of categorical and student`s t-test was applied for comparison of continuous variables. P-value of <0.05 was considered significant.

Results

A total of 534 patients were enrolled in this study during study period (Figure 1).
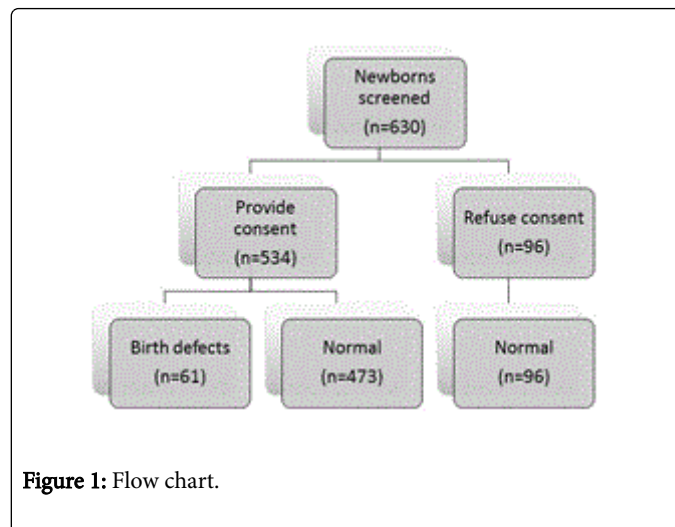

Figure 1: Flow chart.

The mean age of enrolled participants is 26.72 ± 7.07 years, and 55.2% of mothers were >30 years of age, the mean years of marriage was 5.95 ± 6 years. Overall 49.4% women were residing in urban area and 50.6% in rural areas. Majority of women were Sindhi speaking as mother tongue accounting for 45.5% cases.

Approximately, 89% women were Muslims, 8% Christians and 3% Hindus, 60% women had gestational age of <37 weeks, 68.4% had interfamily marriages. Overall 11.5% of newborns have congenital malformations.

Comparison of baseline characteristics is summarized in Table 1 and type of malformation is summarized in Figure 2.

Variables		Birth defects		Total (n=534)	P-value
		Yes (n=61)	No (n=473)		
Age of mother	<30 years	24 (39%)	271 (57%)	295 (55%)	0.006
	≥ 30 years	37 (61%)	202 (43%)	239 (45%)	
Years of marriage	<5 years	22 (36%)	214 (45%)	236 (44%)	0.11
	≥ 5 years	39 (64%)	259 (55%)	298 (56%)	
Residency	Urban	32 (53%)	232 (49%)	264 (49%)	0.357
	Rural	29 (46%)	241 (51%)	270 (51%)	
Ethnicity	Sindhi	27 (44%)	216 (46%)	243 (46%)	0.367
	Mohajir	6 (10%)	46 (10%)	52 (10%)	
	Pathan	28 (46%)	190 (40%)	218 (41%)	
	Punjabi	0 (0.0%)	21 (4%)	21 (4%)	
Religion	Islam	49 (80%)	425 (90%)	474 (89%)	0.016
	Christians	8 (13%)	37 (8%)	45 (8%)	
	Hinduism	4 (7%)	11 (2%)	15 (3%)	

Gestational age	<37 weeks	50 (82%)	274 (58%)	324 (61%)	<0.001
	≥ 37 weeks	11 (18%)	199 (42%)	210 (39%)	
Relation	Inter-family marriage	44 (72%)	321 (68%)	365 (68%)	0.02
	Outside family marriage	17 (28%)	152 (32%)	169 (32%)	
Type	One anomaly	23 (38%)	44 (100.0%)	68 (64%)	<0.001
	Multiple anomaly	37 (61%)	0 (0.0%)	37 (35%)	

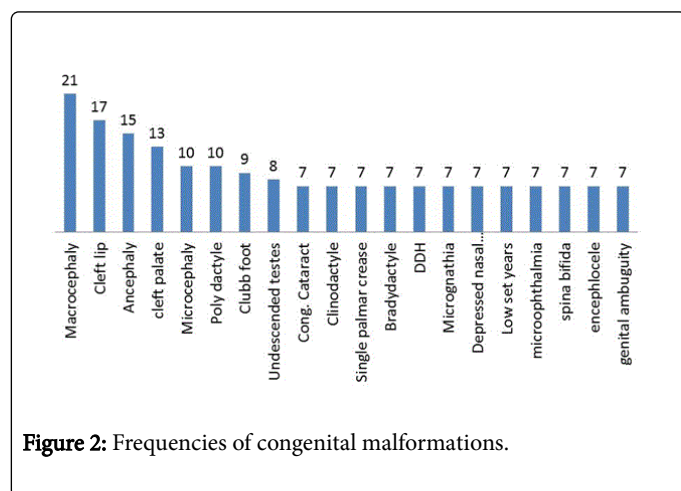**Table 1:** Baseline characteristics of enrolled participants by outcome.

Figure 2: Frequencies of congenital malformations.

Discussion

Birth of abnormal child is a stressful situation for mother as well as for society. About 8 million children are born each year with congenital abnormalities, out of which 3.3 million die before the age of five while 3.2 million survivors suffers from severe mental or physical disability [14]. Although it is a major global concern but advancing technology and early antenatal diagnosis of birth defects has altered the trends in birth prevalence of the congenital abnormalities [15].

Worldwide, the prevalence rates of all genetic birth defects combined range from a high of 82/1,000 live births in low income regions to a low of 39.7/1,000 live births in high income regions [16]. These malformations have multi-factorial etiologies and 40% of cases are idiopathic but there is an impression that they are more prevalent in populations with consanguineous marriages [17].

Despite amazing advances in science and technology, detection of causes of congenital malformation still remain obscure in most of the cases and only few identifiable factors led the scientist to rethink the situation which is global burden. The epidemiological survey of pattern and prevalence of congenital anomalies not only helps in understanding the frequency of malformations in specific area but also contribute to the general knowledge about the predisposing factors and different patterns of birth defects.

In the tropics, malnutrition and infections are main causes of infant morbidity and mortality while in the temperate zones, cancer, accidents and congenital abnormalities are the key causes of infant morbidity and mortality [18]. Prevalent studies of congenital anomalies are useful to establish baseline rates, to document changes over time and to identify clues to etiology. They are also important for health services planning and evaluating antenatal screening in populations with high risk. The study is also important as it may help to raise the awareness of surgical Pediatric intervention and to emphasize the loss of babies with congenital abnormalities. In this study the frequency of congenital malformation was 11.5% of them 50% had CNS malformations followed by 20% had cleft lip and palate, 10% each GI, musculoskeletal and ear, nose & throat malformations.

The commonest anomaly detected in the study was the involvement of central nervous system. In a study it was revealed that alimentary tract, nervous system and cardiovascular system are the most commonly affected parts in descending order of frequency [19]. In accordance with other studies [20,21]. Neural tube defects (NTD) were the commonest anomaly found in this study accounting for 46.67% of cases. Consanguinity is considered a controversial association with congenital malformations (CMs). Increased incidence of CM in consanguineous couples is due to homozygous expression of recessive genes inherited from common ancestors [22]. The current study found CMs to be more common in consanguineous parents than non-consanguineous which was in accordance with an earlier study (61.3%) [23]. Another study also reported that CMs were 3.5 times more common in consanguineous than non-consanguineous marriages. This association is also supported by another study [23,24].

In this study we found that 64% had interfamily marriages and the frequency of congenital malformations is common among interfamily marriages compared to marriages outside families (p=0.021). In a study, it was reported that cousin marriages with 1st degree cousin accounted for 83.33%. Among interfamily marriage people, 15.6% had congenital malformations. In a study, consanguinity was accounted for 22% cases of congenital malformations.

There are some limitations to this study. Chromosomal analysis and Viral infections markers (TORCH) were not conducted in this study due to high cost of these tests which further add burden to the patient so only symptomatology of viral infections were asked to determine the risk in particular patient. In this study we also have not compared risk factors in controls so in future we will conduct study to determine the relative risk of each risk factor with congenital anomalies. Non probability sampling may lead to sampling bias and since sample is not impartially chosen it may not representative of population. Hence the external validity of this study is low. One more limitation, this hospital mostly serves the rural population, where most of the parents left against medical advice in case of anomalies due to culture reason. Parent are thinking that infants with congenital malformation can be treated by spiritual healers and hence often not come into medical attention especially if infants born at night.

Conclusion

In conclusion, Neural tube defect is identified as major congenital abnormality in this community and consangious relationship being the biggest risk factor associated with it. Knowledge of incidence and pattern of CMs are important to plan preventive strategies at different levels by healthcare providers. Consanguineous marriages with first degree cousin and maternal relationship identified as major contributor and risk for congenital anomalies so discouraging intermarriages, and early termination of pregnancies in fatal cases will definitely reduce birth defects in community. In future large study will be required for genetic association to see family trends in this population and viral marker till that time consanguineous marriages will be considered as major risk factor in this population for increased risk of congenital malformation especially neural tube defect. It is therefore recommended that general awareness should be created regarding peri-conceptional use of folic acid use. Since most of the women presented during late pregnancy, it is important that antenatal care should be emphasized and it should be improved to detect and manage congenital anomalies in time.

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