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Consanguinity and hearing impairment in developing countries: a custom to be discouraged

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Abstract

Consanguineous marriage is a tradition which is commonly practised among Asian, African, and Latin American communities whether they are living in their own countries or settled in Europe or the USA.

These communities, in addition to their custom of interrelated marriage, have large families and are a rapidly growing population. The siblings of consanguineous marriages have a significantly higher incidence of autosomal recessive diseases including hearing impairment.

Two epidemiological surveys were carried out 10 years apart. There were 6 421 subjects from Riyadh City and 9 540 from all other parts of the Kingdom of Saudi Arabia. A random sample was examined otologically and a questionnaire was filled in that included age, sex, family relation, number of siblings, etc. ENT examination and audiological assessment were performed.

Consanguinity was found among 22 per cent as first cousins and 23 per cent as second cousins in the first survey. In the second survey 19 per cent were first cousins and 28 per cent second cousins. The rate of consanguinity was 45 per cent in the first survey and 47 per cent in the second. The prevalence of hereditary sensorineural hearing loss (SNHL) was 66.07 per cent and 36.6 per cent in the first and second survey respectively.

The incidence of hereditary hearing impairment is very high in developing countries compared to developed countries. Prevention is essential to reduce the incidence of genetic hearing loss. Consanguinity should be discouraged through health education of the public about the adverse effect of interrelated marriage. Genetic counselling, premarital and antenatal screening are to be applied whenever possible, at least for those at risk of developing genetic diseases including hearing impairment.

Key words: Consanguinity; Hearing Disorders; Health Surveys; Preventive Medicine

Introduction

Consanguinity (family intermarriage) is commonly practised in many Asian, African, and Latin American communities.[1–3] In Saudi Arabia, consanguineous marriage as in the mating of first cousins is encouraged as part of social customs especially among tribes, although certain degrees of kinship, for example uncle-niece mating are prohibited by religion. The siblings of consanguineous marriages have a significantly higher incidence of hereditary diseases including hearing impairment.[4]

The great majority of hereditary deafness is caused by single gene autosomal recessive inheritance.[5] Consanguinity, which refers to marriage of parents with a recent common ancestor greatly increases the chance that the two parents may each give the child the same detrimental recessive gene inherited from that common ancestor. In fact, the genetic significance of consanguinity was reported as early as 1902 by Garrard, who ranks with Galen as a founder of medical genetics.[6] The effect of consanguinity on hereditary deafness has been well studied and documented. Using statistical principles of population genetics, many authors calculate that all cases of familial deafness where there is no family history, but with consanguineous parents, could be accounted for by an autosomal recessive gene.[4,7]

The purpose of this paper is to study the frequency of consanguineous marriage in the Kingdom of Saudi Arabia, its effect on the frequency of hereditary hearing loss especially SNHL and to review the literature and the possibilities of preventing or minimizing this custom.

Materials and methods

Two surveys 10 years apart were carried out in the country to study the prevalence of hearing impairment in general and SNHL (the hereditary type) in particular. The frequency of consanguineous marriage was reported together with the prevalence of hearing impairment among the siblings of consanguineous and non-related pairings.

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The first survey was completed in 1990 in the capital city of Riyadh with its cosmopolitan population. Several families emigrated to Riyadh from various parts of the kingdom because of better schooling facilities and job prospects. A random sample of 6421 Saudi infants and children below the age of 12 years was accomplished. The sampling design was essentially a three-stage stratified random sampling using age and sex as stratifying factors in the final stage. The city was divided into 93 administrative areas and these areas were distributed into six strata according to socioeconomic homogeneity. One fifth of the areas in each stratum were chosen by a simple random method. Each area was further subdivided into roads and the latter were subsequently divided into smaller blocks of approximately equal size; and a sample of each block was randomly selected. Within each block selected, a systematic process was used whereby a random starting point was chosen and a predetermined zigzag route followed, so that every other household was studied. The survey team, which included an otolaryngologist, audiologist, a social worker and a nurse, visited houses randomly to find out if there were any children. The visits of the team were carried out in the afternoon and early evening to ensure that children had returned from school or kindergarten. A questionnaire was prepared which included the particulars of the family, including among other variables the age, sex, parents' relationship, and family history of hearing loss. All children were physically examined and subjected to an audiological assessment using a screening Madsen 28 audiometer and tympanometry. As part of the testing done, pure tone audiometry or auditory-evoked response was carried out in the hospital to determine the extent and type of any hearing impairment. The auditory brain stem evoked response was used only in children who were unable to cooperate. These tests were performed by a qualified audiologist in the audiology unit. A pure tone audiologic evaluation for young children used the methods of limits by air conduction of 250 to 8000 Hz and bone conduction of 500 to 4000 Hz. The standard technique was employed for masking air and bone conductions.

In the second survey, 9540 Saudi infants, preschool and school age children, below 15 years were assessed during September 1997 to May 2000. The sample selection was randomly chosen to represent children covering all socio-economic and demographic groups from the different provinces of Saudi Arabia. Based on the information obtained from the ministry of health (MOH), there were 1750 health centres scattered all over the kingdom in the various provinces. Each centre has a certain number of inhabitants registered and they represent a fair and approximate census of population of that area. According to socioeconomic homogeneity the areas with health centres were divided into four strata, from each stratum, a number of centres were randomly selected and 70 centres were selected to represent the actual socioeconomic distribution of the population. Five centres each from the Southern and Eastern and 30 centres each from the Western and Central provinces were selected. The population registered in each selected centre was called to bring their children for otological examination and hearing assessment, most of the children being accompanied by one or both parents. The overall response was 69 per cent. Each survey team consisted of an ENT specialist, a nurse and a social worker. Each child was carefully examined for hearing status and a questionnaire was completed with the help of the parents. The questionnaire was based on the WHO/PDH ear examination form with some modifications. It consisted of information regarding age, sex, consanguinity of parents, family history of deafness, hearing and speech deficits and exposure to various known risk factors for hearing impairment.

Hearing impairment in the children was tested by free field speech testing and tuning fork tests. Pure tone audiometry (0.5–2 kHs2) and tympanometry were used for further confirmation and the assessment of the severity of hearing loss. The type of hearing impairment was determined by pure tone average based on the air conduction threshold at 500 Hz, 1000 H and 2000 Hz. The degree of hearing loss is generally expressed as an average in decibels hearing loss (dBHL): slight 16–25 dBHL, mild 26–45 dBHL, moderate 46–75 dBHL, severe 76–100 dBHL and profound 100+ dBHL. Evoked response audiometry was performed in the case of very young children below four years of age whose hearing was suspect. The data were analysed by χ^2 test using EPI-INFO computer software. A value of $p<0.05$ was considered as statistically significant.

A child was considered to have hereditary hearing impairment if there was a positive family history. A sibling was considered as having a positive family history if a parental sibling of direct parental ancestors was deaf without environmental factors, regardless of the parental hearing status. Children with positive environmental factors (prematurity, meningitis, rubella, mumps, etc.) were considered to be deaf as a result of these factors. Children with neither family history nor environmental history were considered to have SNHL of unknown cause.

Results

In the first survey a total sample of 6421 Saudi children, 55 per cent males and 45 per cent females, were recruited. Table I shows the age and sex distribution of the children.

TABLE I
THE AGE AND SEX DISTRIBUTION OF THE 6421 CHILDREN OF THE FIRST STUDY (1990)

Age	Number
Up to 4 years	3214
>4–8 years	2010
>8–12 years	1178
No data	19
Sex	
Male	3531
Female	2890
Total	6421

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TABLE II
CAUSES OF SENSORINEURAL HEARING IMPAIRMENT AMONG 6421 SAUDI CHILDREN

Cause	Number	Overall prevalence	Proportional prevalence
Hereditary	111	1.73	66.07
Prematurity	17	0.26	10.12
Meningitis	15	0.23	8.93
Rubella	4	0.06	2.38
Mumps	4	0.06	2.38
Down's syndrome	2	0.03	1.19
Unknown	15	0.23	8.93
Total	168	2.62	100

The relationships of the parents were as follows: 1 419 (22.1 per cent) were first cousins; 1 477 (23 per cent) were second cousins; 3 088 (48.1 per cent) were not relatives while 437 (6.8 per cent) gave no definite answers. Parents' relationships more distant than second cousins were labelled as being unrelated. The number of infants and children with sensorineural hearing impairment was found to be 168 (2.6 per cent with standard error of 0.002). Hereditary factors were thought to be the cause of 111 cases of sensorineural deafness. In other words, the prevalence of hereditary hearing loss was 1.7 per cent among the population studied. The distribution of the other contributing factors of hearing impairment is shown in Table II.

In the second survey a total of 9 549 Saudi children were included in the study. There were 4 189 (43.91 per cent) male and 5 351 (56.09 per cent) female children. The age distribution of the children was as follows: up to four years (2 054), >four to eight years (3 431), >eight to 12 years (3 615) and >12 to 15 years (440) Table III.

Consanguinity of parents of the second study

The parents of 1 809 (19 per cent) children were first cousins, whereas the parents of 2 672 (28 per cent) children were either second cousins or other relatives. In 4 439 (47 per cent) children, the parents had no earlier family relationship, while 620 (six per cent) subjects failed to give a definite answer (Table I and IV).

TABLE III
AGE AND SEX DISTRIBUTION OF 9540 CHILDREN OF THE SECOND STUDY (2000)

Age category	Male	Female	Total
Up to 4 years	1108	946	2054
>4 to 8 years	1749	1682	3431
>8 to 12 years	1251	2364	3615
>12 years	81	359	440
Total	4189	5351	9540

Number of siblings of all children

Twenty-eight had three to four siblings and 46.6 per cent had five or more siblings. The average number of siblings was 5.39 with a range from one to thirteen.

Hearing impairment

In the first study 494 out of 6 421 (7.7 per cent) were found with hearing impairment. There were thirteen cases (0.2 per cent) with unilateral SNHL, 155 (2.4 per cent) bilateral SNHL and mixed HL and 326 (5.1 per cent) with the conductive type of HL. In the second survey out of 9 540 children surveyed, 1 241 (13 per cent) were found to be hearing impaired. A total of 150 (1.57 per cent) of the children had hearing impairment (HI) in the left ear, and 216 (2.26 per cent) in the right ear whereas 875 (9.17 per cent) had HI in both ears. The prevalence of hearing impairment was found to be significantly higher in the children whose parents were either first cousins (16.14 per cent, $p<0.001$) or relatives (12.42 per cent, $p<0.01$) as compared to the children whose parents were not related (10.38 per cent, Figure 1). The prevalence of SNHL was found to be 1.5 per cent, 10.4 per cent of those tested had conductive hearing loss and 1.1 per cent had mixed hearing loss. The various causes of SNHL are shown in Table IV and the extent of bilateral SNHL in Table V.

The two samples were combined to examine consanguinity effect.

TABLE IV
CAUSES OF SNHL AMONG 9540 SAUDI CHILDREN

Cause	Number	Proportional prevalence %	Overall prevalence %
Hereditary	52	36.6	0.55
Renal tubular acidosis (RTA)	2	1.4	0.02
Retinitis pigmentosa	2	1.4	0.08
Prematurity + Low birth weight (LBW)	8	5.6	0.08
Jaundice	4	2.8	0.04
Meningitis	5	3.5	0.05
Mumps	7	4.9	0.07
Measles	3	2.1	0.03
Rubella	2	1.4	0.02
Toxoplasmosis	2	1.4	0.02
Sickle cell disease	1	0.7	0.01
Fever of unknown aetiology	4	2.8	0.04
Head injury	4	2.8	0.04
Unknown cause	46	32.4	0.48
Total	142	100%	1.5%

Note: because of rounding up effect, the percentage may not add up to exactly 100 per cent.

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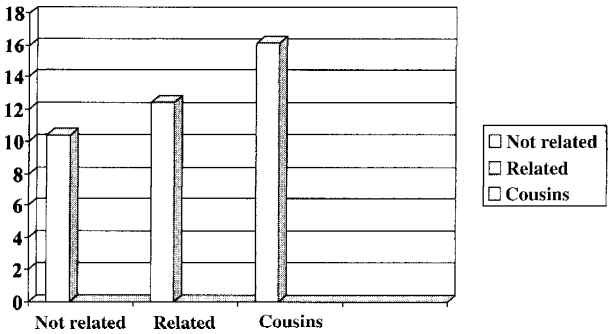

FIG. 1

Prevalence of hearing impairment in the children and consanguinity of parents.

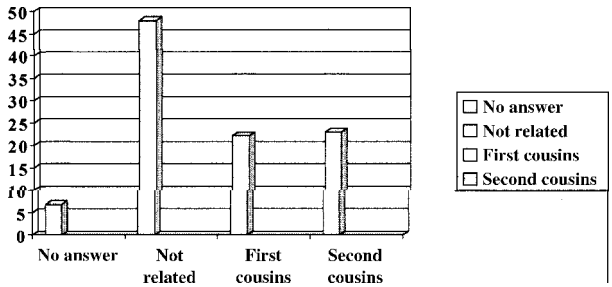

FIG. 2

Percent frequency distribution of total subjects according to consanguinity of parents.

Discussion

The prevalence of consanguineous mating is dictated to a large extent by culture, tradition, religion and civil law. It is extremely common in some parts of the world such as South India[8] where it is a tradition; while it is not common in other areas such as North America, because the low birth rate reduces the number of relatives available and also because some consanguineous marriages are prohibited by law and religion. This study showed that consanguinity is widely practised in the Saudi community: marriage between first cousins was observed in 22.1 per cent of the parents of the children and more distant consanguinity (second cousins) in 23 per cent (Figure 2). This high frequency of consanguinity is due to social customs, the practice of arranged marriage within the families, and public unawareness of the adverse genetic effect of such practices.

The second study showed that consanguineous marriages are still common (Figure 3). The parents of 19 per cent children were first cousins and 28 per cent were either second cousins or other relatives. Earlier investigations have also reported a high frequency of consanguineous marriages in Saudi Arabia. Al Mazrou *et al.*[9,10] in a study of 8 482 married urban and rural females showed that 34 per cent of all marriages were to first cousins in urban areas and 41 per cent in rural areas. Among literate groups of 30 per cent of marriages were consanguineous and 39 per cent in illiterate groups were consanguineous. We observed the overall prevalence of hearing impairment in Saudi children to be 13 per cent, which is comparatively higher than that reported in earlier studies from Saudi Arabia.[11] Sensorineural hearing loss was found among 1.5 per cent. There was a significantly higher prevalence of hearing impairment in the children of consanguineous parents.

The Arabs in general are genetically diverse. Their demographic features include high rate of consanguinity, which is a tradition, large family size and rapid population growth. The consequences are the high frequency of autosomal recessive disorders. In every ethnic, demographic or racial group, there are certain inherited diseases that occur more frequently than in the general population.

Many authors have suggested that approximately one half of sensorineural hearing loss in children can be attributed to hereditary causes.[12] In this study, hereditary deafness accounted for 66.1 per cent of 168 children with sensorineural hearing loss in the first survey Table VI and 36.6 per cent of 142 in the second one. There is a big difference in the prevalence of hereditary deafness between the two samples due to inclusion of other provinces of the kingdom in the second survey, where familial deafness and consanguinity are minimal. It could be also explained by the increased awareness of the families in big cities like Riyadh and to the improved health services provided during the last decade. This high relative incidence of hereditary deafness may be explained by the actual increase of hereditary deafness cases due to consanguinity and also because of the increased efficiency of detecting genetic deafness. Taylor *et al.* (1975) reported that most, if not all, cases of deafness previously classified as of 'unknown cause' are cases of autosomal recessive inheritance.[13]

The other significant result to emerge from this study is the high absolute prevalence of hereditary hearing impairment (1.7 per cent) in comparison with less than 0.6 per cent incidence reported by many authors.[5,13] This high prevalence may be

TABLE V

DEGREE OF BILATERAL SNHL (SECOND STUDY)

Degree	Number	Proportional prevalence %	Overall prevalence %
Mild (20–40 dBHL)	45	33.3	0.47
Moderate (40–70 dBHL)	22	16.3	0.23
Severe (71–90 dBHL)	36	26.7	0.38
Profound (>91 dBHL)	32	23.7	0.34
Total	135	100%	1.42%

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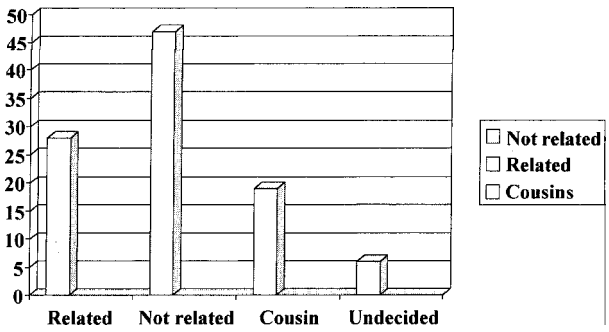

FIG. 3

Percent frequency distribution of total subjects according to consanguinity of parents.

explained, in part at least, by the widespread practice of consanguineous marriages. Table VI shows the effect of consanguineous mating by comparing the prevalence of hereditary deafness among children of parents whose degree of relationship varies.

In the majority of cases of hereditary deafness (75 to 88 per cent) inheritance is by a recessive single gene.[5] In this mode of inheritance, consanguineous marriage, although it does not increase the proportion of abnormal genes in the next generation, does affect their assortment into genotype by increasing the proportion of homozygotes at the expense of heterozygotes. Consanguineous marriage also increases the risk of transmission of polygenic (multifactorial) inheritance. This uncommon type of inheritance is not fully understood, but it is postulated that multiple genes contribute to the disease and that each individual has a threshold above which the abnormality will be manifest.[14] A further point is that for multifactorial inheritance, the risk to subsequent siblings is higher when the parents are consanguineous than when they are unrelated, in contrast to autosomal recessive inheritance, where the risk is the same whether or not the parents are consanguineous.[3,4]

Bergstrom *et al.*[15] have observed that the children of deaf parents without consanguinity, have only slightly increased risk of hearing impairment due to the rare possibility that both father and mother would be affected by the same genetic deafness. On the other hand the children of consanguineous parents are at significantly higher risk of hearing impairment, because their parents are more likely to be homozygous and capable of passing the trait to their offspring. The effect of consanguinity on the development of childhood hearing impairment also depends on the closeness of the relationship of parents. A marriage between first cousins poses a greater risk, whereas a distant consanguinity has comparatively low risk of producing defective offspring,[4] which is also supported by our findings.

TABLE VI

DISTRIBUTION OF HEREDITARY SENSORINEURAL HEARING IMPAIRMENT TO THE PARENTAL RELATIONSHIP (FIRST STUDY)

Parents' relationship	No. and (%) of children	No. of hereditary deaf
First cousins	1419 (22.1)	39
Second cousins	1477 (23)	23
Not relative	3088 (48.1)	43
Undetermined	437 (6.8)	6
Total	6421	111

A preventive programme is necessary to limit the number of children affected through public health education regarding the possible outcome risks of consanguineous marriage. Screening to identify carriers of genetic disorders is an essential aspect of prevention. School screening, premarital, prenatal as well as antenatal screening should be part of the programme. Genetic counselling is the process by which a family is given information about a genetic condition that might affect them, so that they can take the appropriate decisions about marriage, reproduction and health management. Islamic teaching encourages counselling (M Albar, unpublished data).

This counselling may be provided by a variety of professional (the health providers) which should include a clinical psychologist, social workers and public health nurse. The counselling must be based on a correct diagnosis which is the responsibility of the otologist.

The target groups for screening (at-risk population) should be identified, laboratory services should be available and people should accept prevention. There are considerable social, ethical, religious and legal aspects which should be understood.

Conclusion

In conclusion these studies illustrate the high prevalence of hereditary deafness and consanguineous marriage among the population studied. It clearly demonstrates a definite role of consanguinity in the prevalence of childhood hearing impairment. Prevention is the only means to reduce the incidence of genetic hearing loss. This prevention can be accomplished by genetic counselling of high-risk individuals and families. Adequate understanding by the general public and medical profession is necessary. A well planned counselling programme to create awareness of the adverse effects of consanguineous marriages will be helpful in saving our population from the disability of hereditary deafness.

Early intervention for the children who are affected, in the form of hearing aid fitting, speech training and support for their families during the preschool period and at school are of great value. Those who need cochlear implants should be referred to the relevant authority. Hearing and speech centres should be available, at least in the big cities, to look after these children.

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