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Prevalence of selected congenital anomalies in Saudi children: a community-based study

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BACKGROUND AND OBJECTIVES: Limited data are available on the prevalence of congenital anomalies based on a community survey in Middle East countries. The prevalence of congenital anomalies is expected to be high in these countries because of the high consanguinity rate and high maternal age. The aim of this cross-sectional study was to establish the prevalence of congenital anomalies in Saudi Arab children.

DESIGN AND SETTINGS: This is a prospective, cross-sectional, community-based study conducted over 2 years among the Saudi population.

SUBJECTS AND METHODS: The study sample was determined by a multi-stage probability random sampling of household representatives of the Saudi Arab population. The health status of children was obtained during household visits by primary care physicians who performed a history and physical examination of all children and adolescents younger than 19 years. All cases of congenital anomalies were recorded.

RESULTS: During the 2-year study period (2004-2005), a total of 45 682 children were screened. The commonest congenital anomalies found in this survey were Down syndrome, congenital deafness, and congenital blindness with prevalence rates of 6.6 per 10 000, 4.8 per 10 000, and 1.3 per 10 000 children, respectively. The prevalence of cleft lip with or without cleft palate was 0.9 per 10 000 children, achondroplasia was 0.7 per 10 000, and Dandy-Walker syndrome was 0.4 per 10 000. Crouzon syndrome, Treacher-Collins syndrome, Angelman syndrome, and Turner syndrome had equal prevalence of 0.2 per 10 000 children.

CONCLUSION: The data suggest a significant decline in the prevalence of Down syndrome; however, the prevalence of other anomalies like congenital deafness is still high.

Worldwide surveys have shown that the frequency of congenital anomalies varies greatly from country to country. The frequency depends on the time of observations after birth, the types of malformations included, and the differences in reporting, and statistical procedure.[1] Prevalence studies of congenital anomalies are useful to establish baseline rates, document changes over time, and identify clues to etiology. They are also important for health services planning and evaluating antenatal screening for congenital anomalies, particularly in high-risk populations.[2]

Despite the fact that the prevalence of some types of congenital anomalies has been declining during the past decade in some countries, they are still a major cause of perinatal mortality and childhood disability.[2,3] With the control of infectious diseases and malnutrition, particularly in developed countries, congenital anomalies are now making a proportionally greater contribution to ill health in childhood.[4]

Available evidence suggests that congenital and genetic disorders are responsible for a major proportion of infant mortality, morbidity, and handicap in countries in the Middle East.[5]

In Saudi Arabia (SA), the population is characterized by large family size, high maternal or paternal age, and high consanguinity rate of 56%, with the first cousin type the most common.[6] All of these factors have relatively high risk of producing offspring with congenital anomalies.[7]

Apart from isolated reports,[8-12] population-based data on the prevalence of major congenital malforma-

tions in Saudi children are lacking. Therefore, the objective of this study was to provide community-based prevalence of selected congenital anomalies. We believe that this study will be supportive to the future national registry of these anomalies.

SUBJECTS AND METHODS

This study was conducted in the field over 2 years (2004 and 2005). It was part of the Health Profile Project for Saudi Children and Adolescents. The study sample was selected by a multi-stage probability sampling procedure from a stratified listing based in the year 2000–2001 population census available at the time of study design. Accordingly, the sample, which includes all children below the age of 19 years, was the representative of all the socio-economic strata with a weighted urban-rural representation from each of the 13 administrative regions of Saudi Arabia. Data collection was made by house-to-house visits where a medical history and clinical examination of all children below 19 years of age were completed by field teams consisting of primary care physicians and nurses.

The collected data were recorded in the survey questionnaire designed to provide basic information about the subject, including birth date, perinatal history, nutrition, childhood illnesses, drug history, previous and current diagnosis, and socio-economic status of the family. The children, including those with a negative history of the disease, were subjected to complete physical examination to ascertain their general health. The families of children with identified chronic illnesses were requested to provide any available medical reports indicating diagnosis and therapy; children with suspected congenital anomalies, but no previous medical assessment, were referred to the district hospital for evaluation. The congenital anomalies were classified based on diagnosis documented by a medical report or assessment by the field team and reconfirmation of the diagnosis by the district hospital. More details of the data-collecting methods were published earlier.[6,7]

For the purpose of this study, congenital anomalies were defined as structural defects, chromosomal abnormalities, inborn errors of metabolism, and hereditary disease diagnosed before, at, or after birth.[2]

To facilitate communication, all documents including instructions and questionnaire were provided to the field teams in both Arabic and English languages. A workshop training of all field team members was conducted well before the start of data collection in each region of the country. These workshops included oral presentations and small group training on procedures for locating households in the study sample, explanation of the questionnaire, family interviews including history of illness and clinical examination of all children in the household, in single or repeated visits, and field visit demonstration. The data were analyzed using descriptive statistics (SPSS, Chicago, IL USA).

A total of 45 682 Saudi children below the age of 19 years from all the parts of SA were screened over 2 years (2004 and 2005) during house-to-house visits.

Table 1. Prevalence of major congenital anomalies in 45 682 children.

	Male	Female	Total	Prevalence per 10 000
Down syndrome	14	16	30	6.6
Congenital deafness	11	11	22	4.8
Congenital blindness	5	1	6	1.3
Cleft lip/palate	4	0	4	0.9
Achondroplasia	2	1	3	0.7
Multiple congenital anomalies	3	0	3	0.7
Dandy-Walker syndrome	2	0	2	0.4
Crouzon syndrome[a]	1	0	1	0.2
Treacher-Collins syndrome	1	0	1	0.2
Angelman syndrome[b]	1	0	1	0.2
Turner syndrome	0	1	1	0.2
Absent uvula	1	0	1	0.2
Congenital diaphragmatic hernia	1	0	1	0.2
Imperforated anus	1	0	1	0.2

[a]Crouzon syndrome; AD, branchial arch syndrome.

[b]Angelman syndrome; neurogenetic disorder characterized by severe intellectual and developmental delay.

RESULTS

The prevalence of the common congenital anomalies identified in Saudi children is shown in **Table 1**.

All the cases of congenital anomalies were diagnosed by specialists and were known to the families, most of whom had medical reports. In only a few cases, the diagnosis was made by the field team physicians by means of history and physical examination and referral to the district hospitals for confirmation of the diagnosis.

The commonest chromosomal anomaly found in this survey was Down syndrome (Trisomy 21), occurring in 30 children, 14 males and 16 females, with a prevalence of 6.6 per 10 000 children. Congenital deafness either syndromic or non-syndromic accounted for 22 children with equal numbers in both sexs; the prevalence was 4.8 per 10 000 children. One of the children

with congenital deafness reported twice in this study, as he had Treacher-Collins syndrome (mandibulofacial dysostosis), and deafness was part of his syndrome.

Congenital blindness represents third most common congenital anomalies in Saudi children with a prevalence of 1.3 per 10 000. Cleft lip with or without cleft palate was seen in 4 children of the study population; all of them were boys with a prevalence of 0.9 per 10 000 children. Achondroplasia is a common cause of dwarfism inherited as an autosomal dominant genetic disorder; the prevalence was 0.7 per 10 000 children. The rest of the syndromes and malformations listed in the table show less prevalence in the community; the prevalence rate of congenital anomalies were higher in males, except Down syndrome.

DISCUSSION

Demographic and environmental factors may influence the prevalence of anomalies.[2] Available evidence suggest that congenital and genetic disorders are responsible for a major proportion of infant mortality, morbidity, and handicap in Arab children.[5] Several factors may contribute to the high prevalence of genetically determined disorders. The most important contributing factor is the high consanguinity rate.[2,5,7] In a more recent survey of a representative sample of Saudi families defined by a multi-stage random sampling procedure representing both urban and rural settlements, the prevalence of consanguinity was 56%, with the first cousin type was the most common.[6] The high consanguinity rate is known to increase the incidence of recessive disorders[13] which may explain the high prevalence of congenital deafness (4.8 per 10 000) reported in this study, compared with (2 per 10 000) reported in the United Kingdom.[2] The spectrum of hereditary deafness is broad and ranges from simple deafness without other clinical abnormalities to genetically determined syndromes in which deafness is one of a number of clinically recognized signs, together comprising the syndrome as we observed in one of the children who presents Treacher-Collins syndrome with complete deafness as a result of complete atresia of the external auditory canal, a feature of the syndrome.[14] However, only approximately 30% of genetically determined deafness is said to occur in a syndromic form and 70% in a non-syndromic form.[15] Most surveys agree that the most common form of genetic deafness is autosomal recessive accounting for more than 75% of cases.[5,16]

These observations can explain the high prevalence of congenital deafness in any community with a high consanguinity rate, as in our study. Childhood blindness is one of the priorities in vision 2020: the right to sight.[17] Population-based data on the prevalence of childhood blindness, which are needed to set priorities and plan strategies to reduce the childhood blindness, are limited worldwide.[18] No previous data are available on the community prevalence of congenital blindness in Saudi children. In this study, we noticed that congenital blindness affecting mainly males, a ratio of 5:1 with a total prevalence of 1.3 per 10 000. Tabbara KF[19] studied 187 patients attending special educational institutions in Saudi Arabia and found that 84% of childhood blindness were genetically determined diseases, and 56% of this group was the product of consanguineous marriage.

Maternal age is strongly associated with the prevalence of chromosomal anomalies especially Down syndrome.[2] The rate of children with Down syndrome in some Arab countries exceeds from 12 to 17 per 10 000 typical for industrialized countries.[5] In Saudi Arabia, the prevalence of Down syndrome has been reported to be 18 per 10 000 live births.[8] The rising proportion of older mothers is likely to have contributed to the high trend in the prevalence of this anomalies in Saudi children.[5,8] Down syndrome was the commonest chromosomal disease found in our study. The prevalence was 6.6 per 10 000 children compared to the previously reported prevalence of 18 per 10 000.[8] Abortion in the case of pre-natal diagnosis of Down's syndrome is not permitted in Saudi Arabia. Therefore, this reduction in the prevalence of Down syndrome in Saudi children over 15 years (the time difference of the 2 studies) can be explained by the increased awareness of the risk to a bear fetus affected with Down's syndrome in mothers over 40 years.

In contrast, the prevalence of Down syndrome in developed countries increased over the past 20 years from 13 to 25 per 10 000 total birth, while infant mortality decreased; so, there was an increase in long-term survivors.[20]

In some industrialized countries, the parents have the option of selective termination of pregnancies of affected fetuses.[5] It seems likely that the influence of increasing maternal age outweighed the effect of antenatal diagnosis and termination of pregnancy in Europe.[20]

In most Arab countries, except Tunisia, selective termination of pregnancies is not legally available. A better method of prevention would be the general availability of preconception information and family planning for older mothers, which would be expected to lead to a reduction of around 50% in the frequency of new cases of Down syndrome.[5]

Considering the high mortality rate of congenital anomalies during the infantile life, survivors who have been visited at home cannot be the representative of all children with congenital anomalies.

Apart from Down syndrome and congenital deafness, we found no difference in the prevalence of congenital anomalies compared with regional or global studies.[1,2,21-25]

In conclusion, this descriptive epidemiological study of congenital anomalies in SA suggest that despite a considerable decline in the prevalence of some types of congenital anomalies like Down syndrome, the prevalence of other anomalies like congenital deafness is still high; efforts should be made to improve this situation.

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Conflict of interest

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