

Epidemiology of congenital abnormalities in Bahrain

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وبائيات الشذوذات الخلقية في دولة البحرين شيخة سالم العريض

أجريت هذه الدراسة لمعرفة معدل وقوع الشذوذات الخلقية في البحرين . وتمت دراسة إحصائيات وزارة الصحة خلال السنوات العشر من ١٩٨٠ إلى ١٩٩٠ ووجد أن معدل وقوع هذه الشذوذات في البحرين يبلغ ٢,٧٪ من المواليد الأحياء . وتم بحث كل شذوذ بمفرده وقورن بمعدلات حدوثه في البلدان الأخرى كلما أمكن . وتبين أن معدل شذوذات الجهاز الحركي كان أعلى المعدلات (٢,٢٨ في الألف) وجاءت من بعدها شذوذات الجهاز البولي (١,٣ في الألف) بينما بلغ معدل حدوث الاضطرابات في الصبغيات ٠,٩ في الألف .

This study was carried out in order to find out the incidence of congenital anomalies in Bahrain. Statistics of the Bahraini Ministry of Health for 11 years from 1980 to 1990 were studied. The overall incidence rate of congenital anomalies in Bahrain was found to be 2.7% of live births. Each anomaly was studied separately and compared with the incidence in other countries, whenever possible. It was found that anomalies of the musculoskeletal system have the highest incidence (2.28 per 1000), followed by the genitourinary system (2.13 per 1000), while the incidence of chromosomal disorders was 0.9 per 1000.

Epidémiologie des anomalies congénitales à Bahreïn

Cette étude a été réalisée afin de déterminer l'incidence des anomalies congénitales à Bahreïn. Les statistiques produites par le Ministère de la Santé de Bahreïn sur une période de 11 ans de 1980 à 1990 ont été examinées. On a trouvé que le taux global de l'incidence des anomalies congénitales à Bahreïn s'élevait à 2,7% des naissances vivantes. Chaque anomalie a été étudiée séparément et son incidence a été comparée avec celle d'autres pays, chaque fois que possible. Cette étude a montré que l'incidence la plus élevée était celle concernant les anomalies du système ostéo-articulaire et musculaire (2,28 pour 1000) suivie par l'incidence des anomalies des organes génitaux et de l'appareil urinaire (2,13 pour 1000), tandis que l'incidence des anomalies chromosomiques s'élevait à 0,9 pour 1000.

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Introduction

Because of the decline in fatal infectious diseases, in the near future congenital abnormalities will become one of the major causes of infant mortality in Bahrain, as is the case in developed countries.

Methods

Deliveries in state hospitals constitute more than 80% of the deliveries in the country; the rest of the deliveries are conducted at home or in private hospitals. The statistics of the Ministry of Health for 11 years (1980–1990) were classified and analysed to find out the incidence of each anomaly separately. The classification of congenital malformations presents certain difficulties, as other investigators in this field have found. The following classification has been adopted to maximize the information available [7].

Every malformation was classified, according to the system, as follows:

1. central nervous system
2. cardiovascular system
3. musculoskeletal system
4. genitourinary system
5. gastrointestinal system
6. respiratory system
7. chromosomal disorder.

This covers all the abnormalities that were diagnosed in the delivery suites immediately after birth, together with cases that were diagnosed by a paediatrician during the first year of life. However, the statistics may not represent the actual number of cases [7], because of underdiagnosis and underreporting of some types of malformations on account of the following:

- Lethality of these disorders, causing death before birth or before diagnosis is made.
- Impossibility of diagnosis of certain disorders that manifest themselves with the functional development of the infant, e.g. mental retardation, eye and ear abnormalities.
- Difficulty of diagnosing internal organ abnormalities as compared with external organs.

Results and discussion

Table 1 shows the number of deliveries and the incidence of congenital anomalies each year, together with the overall incidence.

Table 2 shows the incidence of these diseases every year, as well as the average incidence.

Table 1 Incidence of congenital anomalies in Bahrain, 1980–1990

Year	No. of births	No. of abnormal cases	Percentage
1980	10 097	86	0.9
1981	11 248	159	1.4
1982	11 248	148	1.3
1983	11 633	161	1.4
1984	12 254	189	1.5
1985	12 394	230	1.9
1986	8 544	355	4.2
1987	9 809	463	4.7
1988	9 978	489	4.9
1989	10 063	348	3.5
1990	10 230	248	2.4

Overall incidence 2.5%

Table 2 The annual incidence (per thousand births) of each anomaly

Anomaly	1980	1981	1982	1983	1984	1985	1986	1987	1988	1989	1990	Average incidence
NTD	2.18	1.01	1.24	1.46	1.14	1.21	0.54	0.49	0.42	0.44	0.35	0.95
CHD	0.49	2.03	2.22	1.63	2.53	2.5	0.53	0.75	0.90	0.58	0.38	1.32
Respiratory	—	—	0.26	0.34	0.16	0.24	0.07	0.08	0.04	0.03	0.05	0.12
Cleft palate	0.39	0.55	0.53	0.52	0.73	0.81	—	—	0.25	0.07	0.05	0.35
Gastrointestinal	0.59	1.02	0.98	1.03	0.98	2.02	0.50	0.66	0.66	0.24	1.27	0.90
Genitourinary	0.89	3.69	3.02	2.41	2.36	3.87	0.93	1.27	1.33	0.90	0.65	1.93
Undescended testicle	0.37	1.29	0.89	0.69	0.73	1.53	0.35	0.46	0.37	0.18	0.27	0.65
Hypospadias	0.39	2.03	1.24	1.03	0.82	1.13	0.16	0.49	0.50	0.45	0.13	0.76
Chromosomal	0.79	1.01	0.89	0.77	1.14	1.13	0.42	0.45	0.79	1.06	0.20	0.79
Musculoskeletal	1.78	3.50	2.49	3.00	3.75	4.03	0.90	0.89	0.90	1.06	0.47	2.07
TEV	0.59	2.03	—	1.20	1.22	1.05	0.39	0.42	0.32	0.39	0.18	0.71

NTD = Neural tube defects

CHD = Congenital heart defects

TEV = Talipes equinovarus

Central nervous system

The incidence of neural tube defects (NTD), which include anencephaly, spina bifida and encephalocele, was found to be 0.95 per 1000, which is considered low incidence. The incidence of NTD in the United Kingdom (in certain parts of Wales, Ireland and Scotland) is as high as 4–8 per 1000. In general, most NTD are of multifactorial inheritance [4].

Cardiovascular system

Table 2 shows the overall incidence of congenital heart defects to be 1.32 per 1000 in Bahrain. In the United Kingdom the incidence is 8.14 per 1000. The cause of the low incidence here is underdiagnosis and not merely rare occurrence. More of these malformations are diagnosed at a later stage in the development of the infant. The malformation of great vessels is underreported because usually it can be diagnosed only at

autopsy. The majority of cardiovascular system anomalies have a multifactorial mode of inheritance [5].

Musculoskeletal system

The anomalies involving this system are the most common of all anomalies. The overall incidence was found to be 2.07 per 1000. The most common category of these is talipes equinovarus (TEV), which had an incidence of 0.87 per 1000.

Genitourinary system

The incidence was found to be 1.93 per 1000, which ranks second in frequency after musculoskeletal disorders. The most frequent types found were undescended testes (0.65 per 1000) and hypospadias (0.76 per 1000). The reasons for this may be the easy diagnosis of these disorders in comparison with the diagnosis of kidney abnormalities.

Gastrointestinal system

The average incidence of anomalies involving this system was 0.90 per 1000. In 1985 the incidence was 2.02 per 1000. The incidence of cleft palate and lip was found to be 0.35 per 1000. In Europe it is 0.6 per 1000 [8].

Respiratory system

We found that the incidence of anomalies involving this system was 0.12 per 1000. The same was noticed in other studies (0.2 per 1000) [7]. Due to the lethality and difficulty of diagnosis of this anomaly, this is usually an underestimation.

Chromosomal disorders

Chromosomal disorders had an incidence of 0.79 per 1000. The most common category was Down syndrome. In 1984 the incidence was 1.14 per 1000, compared to the international incidence of 1.4 per 1000.

Conclusion

We found from our study that the incidence of congenital malformations in Bahrain falls within the world range of 2.5–6%, even for individual anomalies [4]. It is thought that the majority of such anomalies have a multi-

factorial origin, caused by the joint action of a genetic liability (polygenic inheritance) and environmental factors. The recurrence risk depends on the number of affected individuals in the family, the severity of the disorder and the sex of the index case. For an isolated case the recurrence risk varies between 1% and 7% depending on the type of malformation. The malformation that occurs in more than one member in the same family can have at least four causes: teratogens, an inherited chromosome abnormality, multifactorial inheritance and Mendelian inheritance [8]. The first of these may be established by taking a careful history of the pregnancy, and the second by chromosomal analysis. However, to ascertain whether two affected siblings or an affected parent and affected child reflect multifactorial or Mendelian inheritance we need to collect data from a large number of the members of the affected family [9].

The above frequencies are an underestimation since not all congenital malformations can be detected at birth or shortly thereafter. Some may not be diagnosed in the first year. In other studies, researchers have found that they diagnosed 43% of malformations at birth and 82% during the first 6 months [9].

References

1. Worcester J, Stevenson SS, Rice RG. 677 congenitally malformed infants and associate digestational characteristics. *Pediatrics*, 1950, 6(27):208–22.
2. Gustavson KH, Jorulf H. Recurrence risks in a consecutive series of congenitally malformed children dying in the perinatal period. *Clin Genet*, 1976, 9:307–14.
3. Marden PM, Smith DW, McDonald MJ. Congenital anomalies in the newborn infant, including minor variations. *J Pediatrics*, 1964, 6:357–61.
4. *Community approaches to the control of hereditary diseases*. Report of a WHO advisory group. Geneva; World Health Organization, October 1985.

5. Van Regemorter N et al. Major congenital malformations in 5448 newborns: comments on genetic counselling and prenatal diagnosis. *Acta Paediatr Belg*, 1976, 34:73-81.
6. Van Regemorter N et al. Congenital malformations in 10 000 consecutive births in a university hospital: needs for genetic counselling and prenatal diagnosis. *J Paediatr*, 1984, 3:386-90.
7. McIntosh R et al. The incidence of congenital malformations: A study of 5964 pregnancies. *Pediatrics*, 1958, 14:505-22.
8. Neel JV. A study of major congenital defects in Japanese infants. *Am J Hum Genet*, 1958, 10:399-43.
9. Holmes LB. Inborn errors of morphogenesis. *N Engl J Med*, October 1974, 763-73.
10. Skinner R. Genetic counselling. In: Emery AEH, Rimoin DL, eds. *Principles and practice of medical genetics*, vol. 2. Edinburgh, Churchill Livingstone, 1983:1429-36.
11. Stoll C et al. Usefulness of a registry of congenital malformations for genetic counselling and prenatal diagnosis. *Clin Genet*, 1986, 29:204-10.