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PEDIATRICTHEME

Prevalence of congenital malformations recorded on the birth certificate and fetal death certificate, Mexico, 2009 to 2010

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ABSTRACT

Background. Congenital malformations are a main cause of infant death, chronic illness and disability in several countries. The expected frequency is ~2-3% in live newborns and ~15-20% in stillbirths. In 2010 in Mexico, infant mortality ranked in second place with a rate of 336.3/100,000 births. In order to estimate prevalence and main causes of congenital malformations in live births and stillbirths, national base registries of newborns and stillbirths were evaluated for 2009-2010.
Methods. Databases of neonatal live births and fetal deaths were combined.
Results. From a total population of 4,123,531 certificates, 99.3% were live born and there were 0.7% fetal deaths. Congenital malformations were registered in 30,491 cases, 91.7% of live newborns and 8.3% of fetal deaths with a prevalence rate of congenital malformations of 73.9/10,000.
Conclusions. The reported prevalence was lower than expected. It is necessary to enforce registry systems through system validation and training of personnel.

Key words: congenital malformations, prevalence, causes, population-based registries, national registries.

INTRODUCTION

According to the World Health Organization (WHO), congenital malformations, congenital anomalies or birth defects are important causes of infant mortality, chronic disease and disability in many countries and a major cause of health care for surviving children. These affect 1/33 infants and cause 3.2 million disabilities annually, with great impact on those affected, their families, health systems and society. An estimated 270,000 newborns died in 2010 within the first 28 days of life due to congenital malformations. Therefore, recommendations have been made for the international community to work with the creation and strengthening of national programs.[1] Congenital malformation is defined as an abnormality in some structure and is present at birth, producing a physical or mental disability and, in some cases, death. The most severe malformations occur during the first 8 weeks of gestation during organogenesis.

Congenital malformations are classified according to certain characteristics. Its magnitude is classified into greater or lesser malformations. A greater congenital malformation is understood to be one that causes limitations in biological, psychological and social spheres of an individual; lesser malformations are those that do not meet these conditions. When the diagnosis is made clinically, these are classified as external malformations. When the diagnosis requires alternative measures they are classified as internal. According to its etiology they may be genetic, environmental or undefined. There may only be one malformation or there may be multiple malformations. Depending on their location, they are classified according to their position in certain organs or systems. The dysmorphic classification will depend on the stage of development of prenatal life during which the alteration occurs and its mechanism, labelling malformations as deformities or disruptions. Depending on the histology, they are called

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aplasias, hypoplasias or dysplasias. Finally, there are sequences, associations and well-defined syndromes.[2] The influence of congenital malformations in certain diseases has been estimated in different population groups. The numbers referred to are variables due to the methodology used in the manner of collecting the information, the characteristics themselves of the population studied and the diagnostic criteria. The information can be obtained from national databases, national and regional medical records and records of birth defects. The frequency of major malformations in live births reported at the international level varies between 2 and 3%, and in fetal deaths between 15 and 20%.[2] In Mexico, studies have been carried out with that objective: in Guadalajara, Jalisco, the incidence of congenital malformations in a population of 7791 consecutive live births was 12.8/1000.[3] In the Federal District of Mexico (DF) an incidence of 1.2% major congenital malformations was found and 2.1% lesser congenital malformations in 3283 live births.[4] Also, in the DF an incidence of 2.6% in 12,659 newborns was detected and 8.7% in 208 fetal deaths.[5] In Monterrey, Nuevo Leon, the incidence was 2.31% in 9675 newborns.[6] Due to the epidemiological significance and transcendence of these alterations, organizations have gathered at the international level to carry out a surveillance of congenital malformations. The *European Surveillance of Congenital Anomalies* (EUROCAT) began in 1979 with a registry of >1.7 million births per year. It includes 21 countries and covers 29% of the European population. One of its principal objectives is to evaluate the effectiveness of primary prevention in health systems.[7]

In a collaborative project of the International Clearinghouse for Birth Defects Surveillance and Research (ICBDSR), there were 29 countries with different registries involved. Of these, eight contribute with population baselines at the national level: CCASS Canada, Czech Republic, Costa Rica, Finland, Hungary, New Zealand, Norway and Switzerland.[8]

The trend of infant mortality in <1 year in Mexico has shown an important decline in the last 30 years, with rates on the order of 3889.9/100,000 recorded births in 1980 to 1498.6 in 2010. This means a decrease of 61.5% and in absolute terms of 94,116 deaths to 28,865, representing 69.3%. In contrast, there have been rates for congenital malformations of 224.4/100,000 births in 1980 with 5,031 deaths, and 336.3/100,000 births in 2010 with 6,477 deaths, representing an increase of 28.7% in the rate and in absolute numbers, 49.9%. In 1980, congenital malformations occupied fourth place, accounting for 5.2%, relative to the total infant deaths. In 1990, congenital malformations occupied fourth place with 7127 deaths (10.9%). In 2000, congenital malformations occupied second place with 7212 deaths (18.7%) and in 2010, second place with 22.4%. This change to second place came about in the mid-1990s.[9,10]

In 1978 the Registry and Surveillance of Congenital External Malformations (RYVEMCE) was begun in Mexico and has been a member of ICBDSR since 1980. The information is collected from 21 hospitals in 11 cities throughout Mexico and covers ~3.5% of annual births in the country.[10] The studies already mentioned in the Mexican population have low representation because their estimates correspond to small populations. The RYVEMCE manages information of congenital malformation prevalence by specific causes and does not mention an indicator of prevalence of the total congenital malformations.[11,12]

In May 2007, the Department of Health created a subsystem of information at the national level that gathered data of live births in a format denominated "birth certificate," which contains information regarding the mother, data of the newborn, data of the person certifying, other methods of information, and general and specific instructions on filling out the form and certification. In a similar manner, the format called "fetal death certificate" was generated, which contains information relative to the data of the product, the pregnancy and the occurrence, causes of the fetal death, data of the mother, data of the informant, data of the person certifying and instructions on how to fill out the document. Both records are coded and captured in the National Institute of Geography and Statistics (INEGI). Once the review, coding, capture process and closure of the corresponding year are completed, data are sent to the Department of Health for its exploitation and dissemination of the information. Accordingly, the objective of this study was to estimate the prevalence of congenital malformations at birth and of the principal causes recorded in the birth certificates and fetal death certificates during the period from 2009-2010.

MATERIALS AND METHODS

Databases containing the information from the birth certificate of live births and fetal death certificates were merged

with respect to common variables using the Statistical Package for the Social Sciences (SPSS) v.19. Comparative tables were obtained with distributions of frequencies and percentages and also prevalence rates. From the live births database, the variable referred to as "congenital anomalies, diseases or lesions of the newborn" were selected, and for the fetal death the variable "basic cause of death." The overall study included newborns and fetal deaths from 22 to 42 weeks gestation and with weight ≥500 g as well as the usual residence was in Mexico.

The reasons were analyzed taking as a base the groupings suggested by the International Classification of Diseases 10th revision (ICD-10) by chapters. Specifically selected was Chapter XVII "Congenital Malformations, Deformities and Chromosomal Anomalies" from which the causes by group and categories were studied, and according to order of frequency from highest to lowest was selected.

A table was constructed with some of the indicators selected between Mexico and Costa Rica for purposes of international comparisons. The latter was chosen because it is a program based on populations and its system of registry of congenital malformations represents ~98% of its births, whereas Mexico includes 100%.[13]

A map with prevalence rates of congenital malformations according to each state was created. It was grouped into three strata: high, intermediate and low, based on the natural break technique of the program System of Geographical Information in Epidemiology (SIGEPI) (v.4.1).

RESULTS

A database with 4,123,531 births, of which 99.3% corresponded to live births and 0.7% for fetal deaths, was created. The resulting number of cases of congenital malformations was 30,491, of these, 91.7% were live births and 8.3% fetal deaths. The proportion of congenital malformations in newborns was 0.7% and 8.4% in fetal deaths (Table 1). Depending on the causes by chapter according to the ICD-10, 92.3% corresponded to healthy children, problems of the live births and deaths related to maternal factors and complications of pregnancy, labor and delivery (2.1%), congenital malformations, deformities and chromosomal abnormalities (0.7%) and those poorly coded or missing data (4.9%).

The national prevalence rate for the period from 2009-2010 was 73.9/10,000 births (Table 2); 83.9% of all cases were concentrated in the first six groups. In first place were musculoskeletal system malformations because in this group almost 2/5 of the cases were concentrated. Following, in order of frequency but with a smaller percentage, were congenital diseases of the eyes, face and neck, malformation of the genital organs, congenital disorders of the nervous system, other congenital dysmorphias and, finally, malformations of the lips, mouth and palate (Table 2). With regard to the category, the 20 principal causes were concentrated in 73.9% of all congenital malformations (Table 3). Specifically, the ten main categories accounted for 60.5%. The causes according to the group of malformations of the musculoskeletal system, in order of frequency, were deformities of the feet, hip, polydactyly, unclassified disorders of the osteomuscular system, other deformities of the bones of the skull and face and syndactyly. All were practically in the group of live births, representing 32.2% of the overall congenital malformations. Testicular malformations were mainly due to lack of descent or hypospadias, representing 8.1% of the total malformations; 9.3% corresponded to anomalies of the lip and palate. With respect to malformations of the nervous system, anencephaly and spina bifida, subject to epidemiological monitoring in Mexico, these barely represented 4.5%. With respect to the distribution of prevalence rates of congenital malformations according to state during the study period, the fact that rates of greater prevalence were mainly concentrated in the states in the center and two of the south of the Mexican Republic was notable: Aguascalientes, Colima, Queretaro, DF, Tlaxcala, Yucatan, Hidalgo, Puebla, Guanajuato, Michoacan and Tabasco (Figure 1).

DISCUSSION

Prevalence of congenital malformations observed in the present study was 73.9/10,000. It is lower compared to other studies that have been reported at the national and international level 2-3% in live births and 15-20% fetal deaths. The percentages obtained in this study were 0.7% and 8.4%, respectively. This may be due to differences in the methodology for detecting congenital malformations. This coupled with the fact that the registry subsystem of live births began in 2007, so it may require more training in the detection of congenital malformations and the appropriate recording of data on the certificate.

This study estimated congenital malformations with a population base derived from a program recently implemented at the national level and has heterogeneous representation as the staff who request the certificates of births and fetal deaths are physicians of various specialties including pediatricians, obstetricians/gynecologists and others as well as personnel authorized by the Department of Health.

On the international scale, countries that use national population databases to estimate the prevalence of congenital malformation and that participate in CLEARINGHOUSE are Canada, Costa Rica, Czech Republic, Finland, Hungary and Switzerland. For the comparison of the rates by specific causes reported in this study, we selected congenital malformations that are identified by examination and only Costa Rica was chosen as a reference because it is a population-based program and covers 98% of all births. Other countries with population-based registries were excluded because prenatal diagnosis and termination of pregnancy for this reason is legally allowed.

Congenital malformations such as anencephaly and cleft lip and cleft palate with cleft lip are higher in Mexico than in Costa Rica (Table 4). It is higher in 31% (2.1/1.6) for the first cause and higher in 11% (4.9/4.4) for the second. Percentages for other causes are lower in Mexico than

Table 1. Percentage distribution of births (Mexico, 2009-2010)

Causes according to classifications of the ICD-10[1]	Liveborn	Fetal deaths	Total	
			No.	%
Healthy	3,806,201	0	3,806,201	92.3
(P00-P99) Fetus and newborn affected by maternal factors and by complications of pregnancy, labor and delivery	58,889	27,443	86,332	2.1
(Q00-Q99) Congenital malformations, deformities and chromosomal anomalies	27,968	2,523	30,491	0.7
Absence of data, incongruent information and coding errors	200,507	0	200,507	4.9
Total*	4,093,565	29,966	4,123,531	100.0

[1]ICD-10, International Classification of Diseases, 10th revision.
*Includes products 22-42 weeks of gestation with weight ≥ 500 g
Source: Databases of newborns and fetal deaths, Dirección General de Información en Salud, Secretaría de Salud.

Table 2. Prevalence of births according to groups of congenital malformations (Mexico, 2009-2010)

Causes of chapter "Q00-Q99 Congenital malformations, deformities and chromosomal anomalies"[1]	Liveborns	Fetal deaths	Total		
			No.	%	Prevalence rate[2]
(Q65-Q79) Malformations of the musculoskeletal system	11,152	148	11,300	37.1	27.4
(Q10-Q18) Congenital diseases of eyes, face and neck	3,503	6	3,509	11.5	8.5
(Q50-Q56) Malformations of genital organs	2,967	1	2,968	9.7	7.2
(Q00-Q09) Congenital diseases of the central nervous system	1,915	820	2,735	9.0	6.6
(Q89-Q89) Other congenital dysmorphic conditions	1,506	1,142	2,648	8.7	6.4
(Q35-Q38) Malformations of cleft lip and palate	2,397	6	2,403	7.9	5.8
(Q90-Q99) Chromosome abnormalities not otherwise classified	1,871	101	1,972	6.5	4.8
(Q39-Q45) Other malformations of the gastrointestinal tract	1,313	25	1,338	4.4	3.2
(Q20-Q28) Congenital diseases of the circulatory system	670	162	832	2.7	2.0
(Q60-Q64) Malformations of the urinary organs	367	64	431	1.4	1.0
(Q30-Q34) Congenital diseases of the respiratory tract	307	48	355	1.2	0.9
Total*	27,968	2,523	30,491	100.0	73.9

[1]ICD-10, International Classification of Diseases, 10th revision.
[2]Per 10, 000 births (live births + fetal deaths = 4,123,531).
*Includes products of 22-42 weeks of gestation and weighing ≥ 500 g
Source: Database of live births and fetal deaths, Dirección General de Información en Salud, Secretaría de Salud.

Table 3. Prevalence of births according to 20 principal categories of congenital malformations (Mexico, 2009-2010)

Categoríes of causes of chapter "Q00-Q99 Congenital malformations, deformities and chromosomal anomalies"[1]	Live births	Fetal deaths	Total		
			No.	%	Prevalence rate[2]
(Q66) Congenital deformity of the feet	3,295	0	3,295	10.8	8.0
(Q65) Congenital deformity of the hip	2,771	0	2,771	9.1	6.7
(Q53) Undescended testicle	1,978	0	1,978	6.5	4.8
(Q89) Other congenital malformations not classified elsewhere	730	1,134	1,864	6.1	4.5
(Q17) Other congenital malformations of hearing	1,819	0	1,819	6.0	4.4
(Q69) Polydactyly	1,608	1	1,609	5.3	3.9
(Q90) Down syndrome	1,473	41	1,514	5.0	3.7
(Q79) Congenital malformations of the bone/muscular system not classified elsewhere	1,210	98	1,308	4.3	3.2
(Q18) Other congenital malformations of the face and neck	1,211	4	1,215	4.0	2.9
(Q37) Cleft lip/palate	1,033	5	1,038	3.4	2.5
(Q36)) Cleft lip	1,002	1	1,003	3.3	2.4
(Q00) Anencephaly and similar malformations	300	558	858	2.8	2.1
(Q03) Congenital hydrocephalus	575	130	705	2.3	1.7
(Q05) Spina bifida	473	35	508	1.7	1.2
(Q54) Hypospadia	498	0	498	1.6	1.2
(Q75) Other congenital malformations of the skull bones and face	424	24	448	1.5	1.1
(Q38) Other congenital malformations of the tongue, mouth and pharynx	447	0	447	1.5	1.1
(Q24) Other congenital malformations of the heart	310	134	444	1.5	1.1
(Q70) Syndactyly	380	0	380	1.2	0.9
(Q35) Cleft palate	362	0	362	1.2	0.9
Other causes	6,069	358	6,427	21.1	15.6
Total*	27,968	2,523	30,491	100.0	73.9

[1]ICD-10, International Classification of Diseases-10th revision.
[2]Per 10,000 births (live births + fetal deaths = 4,123,531).
*Includes products of 22-42 weeks of gestation and weighing ≥ 500 g
Source: Databases of live births and fetal deaths, Dirección General de Información en Salud, Secretaría de Salud.

in Costa Rica, probably because greater emphasis has been given to the identification of the first two aforementioned defects, together with defects of neural tube closure.

Nervous system malformations, anencephaly and spina bifida are predominant in the database of fetal deaths as it is known that they are causes of infant morbidity and mortality. According to the figures reported in this study, the focus of prevention and control of congenital malformations in neural tube defects (specifically spina bifida) with folic acid intake represents in our country only 1.7% of all congenital malformations. However, other causes continue to significantly contribute to child morbidity and mortality and, therefore, should also be subject to medical and epidemiological surveillance so that with specific programs these can contribute to a better quality of life for those children affected.

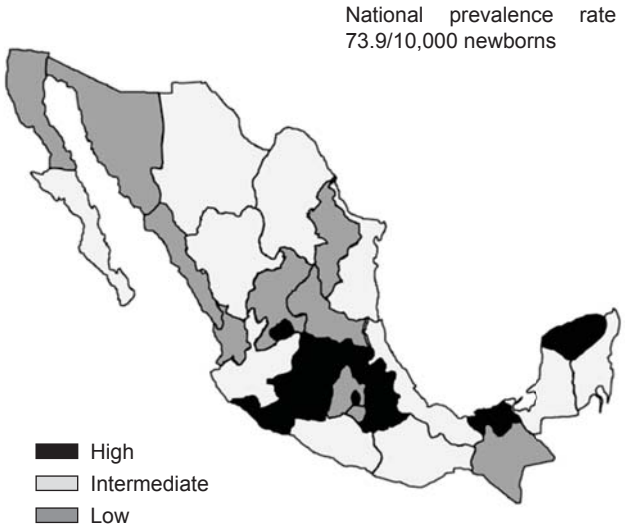
With regard to other unclassified congenital malformations, these represent 6.1% with 1864 cases. This number is significant and is an essential task for those at the managerial and policy levels to assist in the training of staff responsible for certification in order to specify a better diagnosis.

An additional health problem in cases of congenital malformations is the possibility that these are not isolated cases and may have risks of recurrence at the family level. An accurate diagnosis as well as establishing the etiology will allow possible prevention using appropriate genetic counseling.

Distribution of prevalence rates of congenital malformations according to state showed a higher concentration in the central and south of the country, which is related in a very similar way to the behavior of infant mortality due to congenital malformations during the same period of analysis. This suggests conducting a study to help us understand the possible causes of this phenomenon.

The analysis performed allows for the identification of positive and negative aspects. Among the positive aspects

Federal Entity	Rate*
Nuevo León	26.8
Chiapas	34.7
Sonora	37.8
Nayarit	42.7
Sinaloa	46.9
Zacatecas	51.7
Baja California Norte	56.5
Morelos	57.0
México Edo de	58.3
San Luis Potosí	60.2
Oaxaca	63.6
Tamaulipas	66.1
Veracruz	66.1
Campeche	66.8
Chihuahua	67.7
Durango	69.0
Jalisco	69.9
Quintana Roo	70.0
Guerrero	71.8
Baja California Sur	76.1
Coahuila	77.3
Tabasco	88.9
Michoacán	90.6
Guanajuato	93.0
Puebla	94.5
Hidalgo	95.4
Yucatán	102.8
Tlaxcala	105.5
Distrito Federal	118.4
Querétaro	134.0
Colima	134.5
Aguascalientes	187.4
National	73.9



Source: Databases of live births and fetal deaths, Dirección General de Información en Salud, Secretaría de Salud.

Figure 1.

Prevalence rate of congenital malformations according to strata (live births + fetal deaths by federal entity. Mexico, 2009-2010. *Rate per 10,000 births.

are coverage at the national level, identification of external congenital malformations and possibility of being able to carry out an epidemiological surveillance of congenital malformations that are identified in these information systems. Among the negatives are the underreporting, which may be caused by the lack of a precise diagnosis and verifying that the information on live births is current.

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Table 4. Comparison of prevalence rates according to cause of congenital malformation selected (Mexico-Costa Rica)

Cause according to ICD-10[1]	Mexico, 2009-2010	Costa Rica, 2008
(Q00) Anencephaly and similar malformations	2.1	1.6
(Q01) Encephalocele	0.4	0.8
(Q02) Microcephaly	0.4	2.3
(Q03) Congenital hydrocephalus	1.7	3.9
(Q05) Spina bifida	1.2	1.2
(Q35) Cleft palate	0.9	0.7
(Q36-Q37) Cleft lip/palate	4.9	4.4
(Q69) Polydactyly	3.9	10.3
(Q90) Down síndrome	3.7	9.1

[1]ICD-10, International Classification of Diseases-10th revision.
[2]Per 10,000 births (live births + fetal deaths = 4,123,531).
Source: Databases of live births and fetal deaths, Dirección General de Información en Salud, Secretaría de Salud and Annual Report 2010 with data for 2008, Costa Rican Registry of Congenital Malformations (Reference 13).

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