

Title: **Real incidence of gastroschisis and omphalocele. An eighteen-year study in Cuba.**

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Abstract: Background/Purpose: Omphalocele and gastroschisis are frequent affections whose incidence seems to have decreased with prenatal diagnosis. A study is carried out to know the real incidence of these defects not clearly defined in the literature. Methods: It is a descriptive study of all the cases that presented gastroschisis or omphalocele in twelve years, including the born alive and those interrupted pregnancy or withspontaneous abortion for abdominal wall defects. In the last group all the diagnosis was corroborated by pathological studies. Results: In the revised literature not work at all makes reference to the real incidence of the affection was found. The born alive with these affections were 24, 15 gastroschisis and 9 omphaloceles, there were 67 abortions: 22 gastroschisis and 45 omphaloceles, which rises the figure to a total of 91 cases, for which the real incidence was 1.76/10 000 for gastroschisis and 2.92/10 000 for omphalocele. Conclusions: The identification of the real incidence allows to have a more exact knowledge of the quantity of omphaloceles and gastroschisis. Although their incidence appears to be low, there seems to be an increase of these affections nowadays.

Keywords: Omphalocele; Gastroschisis

Introduction

Described respectively by Paré in 1634 and Calder in 1733, omphalocele and gastroschisis are among the most frequent defects of the abdominal wall. They are classic and at the same time a challenging pediatric surgical problem. With the appearance and development of the prenatal diagnostic ultrasound these affections are easily identifiable in such early stages as the tenth intra-uterine week of life ¹. In many places the interruption of pregnancy is done due to the possibility or reality of other associated serious congenital anomalies. This brings about that the real number of cases with these malformations is not accurately known. The consulted works report the incidence of the born alive patient, but the real incidence has not been studied. It would be given at one time by all the alive born patients that present with these affections plus the interruptions carried out by this cause with a positive pathological diagnosis in specific place and period of time.

Many studies have been carried out aimed at avoiding or prevent these defects with different results, since knowing the real incidence is an element of great importance not only for the epidemiologists ^{1,2}.

Harrison ³ drew the attention in relation to the hidden mortality in diaphragmatic hernia in a report in 1979, undoubtedly he noticed the real incidence and magnitude of the problem this affection meant. The prenatal diagnosis has defined the hidden mortality in pathologies such as diaphragmatic hernia, bilateral hydronephrosis, sacroccocygeal teratoma, cystic hygroma, intestinal volvulus with necrosis and gastroschisis-omphalocele. Most of these affections are interrupted once diagnosed keeping in mind its poor prognosis of general survival ⁴.

The frequency in the literature in relation to gastroschisis and omphalocele is not the real one, it only refers to the ones born alive, but with the course of time bigger frequency of spontaneous abortions take place or are induced before the diagnosis of these affections. Stoll, of the French service of clinical genetics mentions 2.18 per 10,000 of incidence for omphalocele and 1.76 per 10,000 for gastroschisis in a study carried out among the years 1979 and 1998, where they followed 265,858 serial births, but they do not include the abortions ⁵.

The incidence of gastroschisis in an area of South Africa has increased in relation to the omphalocele from 1981 through 2001 but there are not final conclusions ⁶.

The opposite is reported in China, with changes between rural and urban areas. Overall incidence of omphalocele in China was 1.52 per 10 000 live births, with an increasing trend during 1996 to 2000 ⁷. Incidence of omphalocele was 1.40 per 10 000 and 1.83 per 10 000 in the rural and urban areas respectively, but the study was made in born alive only ⁷.

The prevalence of gastroschisis in Norway, as reported for the Medical Birth Registry of Norway, increased regularly and sixfold from 0.5 to 2.9 per 10,000 births during 1967-1998⁸.

In the United States of America important works reported a general coincidence with the above mentioned reports. It appeared that the prevalence of gastroschisis was rising from 1992 to 1999, while prevalence of omphalocele was decreasing⁹.

Between January 1989 and November 1996 a total of 44 cases of anterior abdominal wall defects were detected in a study carried out in the University of Alabama. There were 29 cases of omphalocele and 15 of gastroschisis but their incidence is not analyzed¹⁰.

In Utah, gastroschisis prevalence increased from 0.36 to 3.92 cases per 10,000 live births over 31 years¹¹. In Atlanta from 1968 through 1975, the rate of gastroschisis was stable at 0.8 per 10,000 births. After 1975, the rate of gastroschisis was 2.3 per 10,000 births with no significant increase observed from 1976 through 2000. An increase in the rate of gastroschisis was observed in the mid-1970s, but no temporal trend has been observed since that time¹².

In Georgia from 1994 to 2002, the birth prevalence of gastroschisis was 1:3600 and omphalocele 1:3400, but from 2000 to 2002, gastroschisis increased to 1:1667, while omphalocele increased to only 1:2709. The birth prevalence of abdominal wall defects in general is increasing, but more notably for gastroschisis¹³.

In the University of North Carolina the birth prevalence of gastroschisis increased from 1.96 per 10,000 births in 1997 to 4.49 per 10,000 births in 2000¹⁴.

In Tampa the number of cases of omphalocele and gastroschisis were similar, compared to the expected 3:2 ratio, suggesting an increase in the occurrence of gastroschisis. Cases with omphalocele had more syndromic and nonsyndromic anomalies, more chromosomal anomalies, a higher mortality rate, and older mothers¹⁵.

In Taiwan, a retrospective review was conducted of 115 cases seen between January 1990 and June 2000 at two tertiary medical centres. Data included perinatal events and associated anomalies. Of 115 patients, 65 were classified as having gastroschisis and 50 as having omphalocele. Other anomalies were found in 24 omphalocele cases, compared with 23 gastroschisis cases¹⁶. In Denmark the average point prevalence of gastroschisis at birth was 1.33 per 10 000 live and stillbirths. During the first decade, an increase in prevalence occurred culminating in 1976, followed by a decrease reaching its initial value in 1983 and then a new increase. Overall, no significant linear trend could be demonstrated for the entire period. The average point prevalence at birth for omphalocele was 2.07 and for gross abdominal wall defect 0.12 per 10 000 live and stillbirths with no significant change in the period¹⁷.

In Australia there has been a sustained increase in the birth incidence of gastroschisis over the past decade, particularly in teenage women. A significant fetal death rate in the third trimester is observed ¹⁸.

In the Czech Republic during the period of 1961-2000 a total of 2293 cases of abdominal wall defects were registered. From this total number of notified defects 1915 cases were diagnosed after delivery, prenatal diagnosis was made in 378 cases and pregnancy was therefore terminated prematurely. From the total number of abdominal wall defects there were 1450 cases of omphalocele (including 136 prenatally diagnosed cases) and 843 cases of gastroschisis (including 242 cases diagnosed prenatally). The authors found a significant decrease in the incidence in the neonatal population of the Czech Republic due to the advances of prenatal diagnosis in the recent decade ¹⁹.

In the International Center for Birth Defects, 36 programs from Europe, the Americas, Asia, Australia, and South Africa participate in the clearinghouse and cumulatively monitor 3.3 million births each year. The head office of the clearinghouse, the International Center for Birth Defects, registers and evaluates these data. The 19 registries recorded 3073 cases of gastroschisis. The overall prevalence at birth was 0.29 (95% confidence interval 0.21 to 0.40) per 10 000 births in 1974 and 1.66 (1.51 to 1.85) per 10 000 births in 1998. Prevalences varied among programs. Nine areas had significant increases in the prevalence of gastroschisis at birth from Europe (five registries), Australia, Japan, and the Americas (two registries) ²⁰.

Material and Method

This is a retrospective study of all diagnosed cases of abdominal wall defects in the pediatric population of Holguín province in Cuba, assisted between January 1990 and December 2008. It includes the ones born alive such as those cases in which an pregnancy was interrupted prematurely practiced because of positive diagnosis of abdominal wall defects made by the genetics department of the province.

In the last group all the diagnoses were corroborated by the Department of Pathological anatomy.

All the ones born alive were assisted in the service of neonatal surgery of the Pediatric Hospital and in case the parents decided the interruption of the pregnancy or a spontaneous abortion took place because of the diagnosis, it was always corroborated by anatomopathological studies.

The official statistic registrations of the county were consulted to obtain the data of the children born alive born per years and a a revision of the literature was carried on in this topic.

Results:

Demographic data

In the period under study 184 413 births were registered in the county. A number of 24 children were diagnosed with these affections, 15 gastroschisis and 9 omphaloceles that represent an incidence of 0.59 per 10 000 born alive for gastroschisis and 0.32 per 10 000 births for the omphalocele.

In the same period 67 interruptions were made by these causes, 22 for gastroschisis and 45 for omphalocele, for a real incidence of 1,76 per 10 000 births for gastroschisis and of 2,92 per 10 000 possible born alive for omphlocele. There were not articles in the literature revised that included in their analysis the incidence of the interruptions were they already spontaneous or induced.

DISCUSSION

The fact that they are not in the revised literature similar reports to the present work, forces to propose a new term to refers to the alive born patients with these affections and include the interrupted pregnancies by these causes. It would be the real incidence of gastroschisis and omphalocele.

Big epidemic series have found that the omphalocele incidence is of 1 for each 4 000 births and that of gastroschisis of 1 for each 6 000 at 10 000 with same frequency in females that in males ^{20,21,22}. In our casuistry, the initial study shows that in studied time gastroschisis were more frequent than omphalocele, but when determining the real incidence of both affections the omphaloceles number it is bigger than that of gastroschisis agreeing this way with that referred in the classic literature. On the other hand it is also shown a frank tendency to the increment of the incidence of the gastroschisis, observed during the last decades by several authors ^{1,5, 7, 10,23,24}. In our opinion the same thing can be happening to the omphalocele although the fact has not been reflected when not being kept in mind the gestations that have been interrupte by this cause so that the current results would not reflect the problem exactly.

The number of cases operated by these causes in our service could not be significant regarding the incidence of other malformative congenital pathologies, but if the real incidence is analyzed, these malformations have a tendency to the increase and they continue constituting a challenge for surgeons and geneticists for the sake of to decipher the etiology and to achieve its prevention.

The literature also picks up as surprising discovery having found a wide series in wich the incidence of both defects was a lot but discharge keeping in mind the cases with prenatal ultrasonografic diagnosis reaching until one of each 2 500 fetuses ^{21,25,26}.

However we don't find any series that showed the incidence no longer contemplating the cases diagnosed prenatally by ultrasound with it which it is difficult to differ among a defect and other, but checked by the morphological study of the autopsy like it is the case of our report.

CONCLUSIONS

1 - The identification of the real incidence allows to have a more exact knowledge of the amount of this affections.

2 -Although there appear to be a decreased what is really happening is an increase in their frequency.

RECOMMENDATIONS

1 – Follow up the control of these affections with the methodology presented in this work.

2 – Applies the results of this experience to other congenital malformations.

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