



Anomalies Associated with Gastroschisis and Omphalocele in Villa Clara, Cuba: a 30-year series from 1990 to 2019

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Abstract

Background: Gastroschisis and omphalocele are the most common anterior abdominal wall defects. The aims of our study were to describe the prevalence trends of congenital anterior abdominal wall defects and to determine the frequency and type of associated congenital anomalies in gastroschisis and omphalocele cases.

Methods: A retrospective, observational study, with data obtained by the Cuban Registries of Congenital Malformations; was conducted in the Cuban province of Villa Clara from 1990 to 2019.

Results: A total of 158 cases with anterior abdominal wall defect were identified, 93 of them (59%) with gastroschisis and 53 with omphalocele. There was an increasing trend in the prevalence rate of the anterior abdominal wall defects during the study timeframe, mainly in gastroschisis prevalence. The prevalence rate of gastroschisis (per 10,000 live births) during the study timeframe was two fold those of omphalocele (4.8 and 2.7 respectively). The majority of GS cases did not have additional anomalies (80/93 [86%]). There was a statistically significant difference between the proportions of isolated and associated cases. ($p= 2, 2E-08$). Omphalocele cases were more likely to be diagnosed with at least one other CD than those with GS (25/53 [47%] vs. 12/93 [13%]).

Conclusions: An increase in anterior abdominal wall defects prevalence has been noted in recent decades in Villa Clara province. The present study revealed that omphalocele cases were more likely to have other congenital anomalies, predominantly in musculoskeletal and cardiovascular systems.

Keywords: Abdominal wall defects; Gastroschisis; Omphalocele; Anomalies; Cuba

Introduction

Anterior abdominal wall defects are a common cause of morbidity and even mortality. These include the most common congenital anterior abdominal wall defects (AWD): gastroschisis (GS) and omphalocele (OM) or exomphalos. GS is a congenital defect (CD) where the small intestine, part of the large intestine and occasionally other abdominal organs protrude through a lateral defect in the ventral abdomen without any protective membrane,

whereas OM is an amnion covered sac enclosing a range of viscera from a few loops of midgut to the entire midgut, liver and spleen. GS usually is an isolated defect, whereas OM is often seen in association with chromosomal and other systemic congenital defects (CD) (1-3). Villa Clara province is located in the central region of the Cuban archipelago, with a superficial extension of 8 411, 81 square kilometres, occupying the fourth place among the fifteen Cuban

provinces and representing the 7.6% of the total area of the country (4). With this study we aimed to describe the prevalence trends of AWD and to determine the associated anomalies that have been identified in GS and OM cases and to provide an insight into the more affected system of organs.

Patients and Methods

This retrospective, observational study was performed from January 1, 1990, to December 31, 2019 with data obtained by the Cuban Registry of Congenital Malformations and by the Cuban Prenatal Congenital Malformations Registry (RECUMAC and RECUPREMAC respectively, for their names in Spanish), a multicenter, hospital and community-based registers, which recorded all pre- and postnatally detected congenital defects. Data about associated congenital defects were obtained besides the US and post-mortem reports, from the Pathology Department of the Provincial Maternity Hospital "Mariana Grajales", where all anatomopathological macroscopic and microscopic studies were performed. We evaluated data from all cases diagnosed with either GS or OM. All cases were classified in isolated when no other congenital defects were observed neither by US, physical examination nor autopsies. The associated congenital defects were counted in each affected system. All cases of other AWD rather than GS or OM were not included. The prevalence rate at birth of AWD was calculated by dividing the numerator (registered cases of live birth (LB), stillbirth or elective pregnancy terminations) by the denominator (total number of live births in the studied period). The prevalence rate was expressed as the number of cases per 10,000 LB. We conduct a statistical hypothesis test on the basis of the results of the Z test to compare proportions. P value <0.05 was considered statistically significant. Statistical analysis was done using SPSS Version 22 software. This study is based on an analysis of existing registered data, which were anonymously collected, according the ethical issues on human investigations. It was approved by the Ethical Committee of the Biomedical Research Unit of the Villa Clara Medical University, following the World Medical

Association Declaration of Helsinki.

Results

A total of 158 cases of AWD were recorded, of these, 93 (58.9%) were GS, 53 (33.5%) were OM, whereas 12/158 (7.6%) were other AWD not included in the present study. The cases were ascertained from 193 916 LB. The overall prevalence of AWD (per 10,000 LB) was 8.15, meanwhile the prevalence of GS and OM was 4.80 and 2.73, respectively. That is, one case of GS in 2085 LB, whereas OM was found in 1: 3658 LB.

(Figure 1) shows an increasing trend in the prevalence rate (per 10,000 LB) of AWD over the thirty studied years in Villa Clara, from 3.83 in 1990 to 7.47 in 2019. The GS prevalence was over the OM prevalence most years during the study timeframe. A temporal cluster of GS was observed in year 2006 (8 GS cases, and the highest prevalence rate: 17.92 per 10,000 LB). The majority of GS cases did not have additional anomalies (80/93 [86%]). There was a statistically significant difference between the proportions of isolated and associated cases. ($p= 2, 2E-08$). Meanwhile, (30/53 [57%]) of OM cases had associated CD, although, there were not statistically significant differences between these proportions ($p=0, 18$). We identified 71 associated CD in 43/146 [49%] cases of GS and OM. In 13/93 [14%] of GS cases and in 30/53 [57%] of OM cases at least one associated CD was observed. (Tables 1). Of the 43 cases with associated CD, 13 (30%) had more than one CD. OM cases were more likely to be diagnosed with at least one other CD than those with GS (25/53 [47%] vs. 12/93 [13%]). This was true for most categories of involved system of organs, which all shown statistically significant differences, but respiratory and gastrointestinal (Table 2). One-half of cases with associated CD affecting Central Nervous System had Neural tube defects (5/10) and most of three quarters of them (4/5 [80%]) were identified among OM cases and interestingly, all cases with chromosomal abnormalities were observed among OM cases (4/53 [8%], including three cases with Trisomy 13 and another with Triploid.

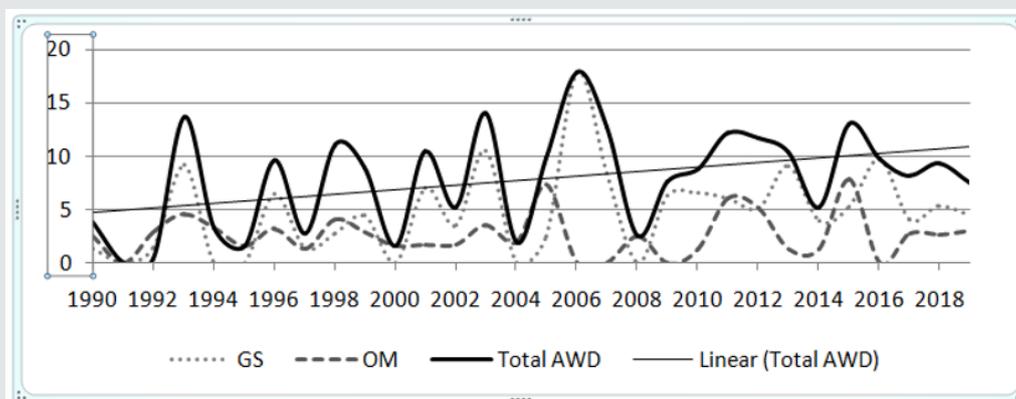


Figure 1: Prevalence trends of gastroschisis, omphalocele and the total of congenital abdominal wall defects in Villa Clara, Cuba. 1990- 2019.

Table 1: Distribution of gastroschisis and omphalocele cases according the presence or not of associated congenital defects. Villa Clara, Cuba.1990- 2019.

| | Type of AWD | | Isolated cases | | Associated cases | |
|---------------|-------------|-------|----------------|-------|------------------|---------|
| | No. | % | No. | % | Z | p |
| Gastroschisis | 80 | 86.02 | 13 | 13.97 | 5,47 | 2,2E-08 |
| Omphalocele | 23 | 43.39 | 30 | 56.6 | 0,89 | 0,18 |

Table 2: Proportion of associated congenital defects observed per system of organs in gastroschisis vs. omphalocele cases. Villa Clara, Cuba.1990- 2019.

| System of organs | Gastroschisis | Omphalocele n=53 | Z | p |
|---------------------------|---------------|------------------|------|---------|
| | n= 93 | | | |
| Cardiovascular | 1.15 | 22.73 | 4,20 | 1,3E-05 |
| Respiratory | 0 | 2.27 | 1,41 | 0,07 |
| Central Nervous | 2.3 | 11.36 | 2,17 | 0,01 |
| Musculoskeletal | 1.15 | 31.82 | 5,20 | 9,6E-08 |
| Gastrointestinal | 4.6 | 2.27 | 0,65 | 0,25 |
| Genitourinary | 2.3 | 9.09 | 1,75 | 0,03 |
| Chromosomal abnormalities | 0 | 7.55 | 3,20 | 6,7E-04 |

Discussion

The prevalence rate of GS (per 10,000 live births) during the study time frame was two fold those of OM (4.8 and 2.7 respectively). It is consistent with trends widely described in the literature, where is stated that prevalence of GS in recent decades shows worldwide remarkable and worrying increasing trend, suggesting role of environmental factors (1, 5- 7). From an incidence of 0.06–0.8 per 10,000 in the 1960s, GS has become more prevalent over the last few decades to its current rates of 4.5–5.13 per 10,000 pregnancies (5). The prevalence rates (per 10,000 LB) of GS and OM in our series are consistent with Schmieding (8) who reported 4–5 for GS and 2 for OM, and almost the same prevalence as Stalling (9) found for a larger series in the USA (4.3 for GS and 2.1 for OM). Otherwise, the lowest prevalence rates (per 10,000 LB) of GS worldwide had been described in Asian countries. In the Liaoning province in China, the prevalence of GS was 2.30 in a series of ten years of study and 0.50 in Taiwan and Japan, respectively (10-12). In our series, 86% of GS were isolated and the remaining 14% had associated CD; this is also consistent with other series, where the incidence of associated anomalies in GS varies from 5 to over 20% (13- 15). Reported associations include cardiac abnormalities and increased prevalence of central nervous system anomalies (amyoplasia), and limb and kidney anomalies and may influence the prognosis of the child with GS significantly (13). In a study conducted by Given, et al. (16) using data from 18 European congenital anomaly registries, they identified 1577 GS cases, 83.0% of which were isolated. The higher frequency of isolated GS cases than of those with associated CD has been shown in other series as well. (17) However, in a study conducted in a Mexican hospital, 52/108 (48%) of infants with GS had one or more associated anomalies, for 1.4 associated defects per case (18). We found the most frequent associated

CD in GS cases affecting the gastrointestinal system. Intestinal atresia has been reported in up to 25% of GS cases in some series, and especially when the bowel was thickened and coated (15). Similar to prior reports, we observed that OM cases were more frequently diagnosed with associated CD compared with those with GS. Musculoskeletal and cardiac anomalies were the most prevalent associated CD found in OM cases in our study, which is consistent with the literature, where it is claimed that neonates with OM have higher rates of death compared to neonates with GS, this difference in mortality is thought to be related to the high incidence of associated CD and specifically, cardiac abnormalities among neonates with OM (19). In our series, 57% of OM cases had associated CD. These findings are consistent with other reports of prenatal diagnosis and management of OM where it is stated that about 40–80% of all OM cases would have at least one concurrent anomaly. Associated anomalies include cardiac (7–47%), gastrointestinal (3–20%), genitourinary (6–20%), chromosomal (3–20%), musculoskeletal (4–25%) and central nervous system (4–30%) (2). There is a known association between genetic disease and OM. We found 8% of chromosomal abnormalities among OM cases. This frequency is lower than the reported in a single-center study of AWD prenatally diagnosed, where chromosomal abnormalities were found by karyotype in 8/29 (28%) fetuses with OM (20). To the best of our knowledge this is the first study conducted in Cuba including a large series of years to determine the frequency and type of associated congenital anomalies in GS and OM cases. Limitations of our study include lack of data regarding number of stillbirths during the study timeframe, for that reason when prevalence rate was estimated; in the denominator only live births were included, and the limited number of chromosomal studies performed to the AWD cases. In conclusion, a rise in AWD prevalence has been noted in recent decades in Villa Clara province.

The present study revealed that OM cases were more likely to have other congenital anomalies, predominantly in musculoskeletal and cardiovascular systems.

Conflict of Interest Statement

The authors declare no potential conflict of interest.

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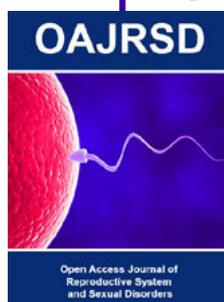
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