Prenatal Diagnosis of Congenital Anterior Abdominal Wall Defects in Villa Clara, Cuba, 1994 - 2019: A Retrospective Analysis

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Abstract

Objectives: To describe trends in prevalence of the abdominal wall defects, maternal age-specific prevalence and accuracy of prenatal diagnosis methods.


Results: The prevalence of abdominal wall defects was 8.47 per 10,000 live births. There was an increase of 34.6% of the overall prevalence. Gastroscisis had the highest increase (36.51%) from 4.30 to 5.87 per 10,000 live births. Gastroscisis was associated with an increased risk for youngest mothers (< 20 years of age) (OR = 3.78; CI = 1.64 - 3.37), whereas for omphalocele it was for women aged ≥ 35 years (OR = 4.22; CI = 2.3 - 7.78). The sensitivity of antenatal diagnosis of anterior abdominal wall defects by ultrasonography was 42.7% and by serum alpha-fetoprotein was 74.2%, with major accuracy of alpha-fetoprotein for gastroscisis (88.9%) and ultrasonography for omphalocele (65.9%) (p < 0,01 and p = 0, 01, respectively).

Conclusion: A rising prevalence trend has been noted in recent decades in Villa Clara. The present study revealed a pattern for young mothers of cases with gastroscisis and older maternal age for omphalocele. The use of fetal ultrasound and alpha-fetoprotein screening allowed the prenatal diagnosis of the majority of cases in Villa Clara.

Keywords: Prenatal Diagnosis; Abdominal Wall Defects; Gastroscisis; Omphalocele; Cuba

Introduction

Abdominal wall defects (AWD) are one of the most frequent congenital defects (CDs). They range from the mild umbilical cord hernia to the highly complex limb-body wall, pentalogy of Cantrell (CDs of the heart, pericardium, diaphragm, sternum, besides AWD), cloacal or bladder exstrophy, body stalk anomaly, ectopia cordis and OEIS complex (acronym by: Omphalocele, Exstrophy of cloaca, Imperforate anus and Spinal defects). The most common AWD are gastroschisis (GS) and omphalocele (OM) [1-4].

Nowadays, the diagnosis is usually made antenatally by ultrasound (US), which also detects associated major CDs. Elevated alpha fetoprotein (AFP) level has been correlated with AWD besides neural tube defects [5].

The services of medical genetics in Cuba are integrated under the National Program for Diagnosis, Management and Prevention of genetic diseases and CDs, started in the 1980s. The screening of maternal serum AFP was implemented since 1982. The determination of AFP is made during gestational age 15 - 19 weeks. Also in the early 1980s, the practice of US for prenatal diagnosis of CDs was introduced in Cuba. Currently, an US was performed to all pregnant women in each trimester of pregnancy [5,6].

Villa Clara province is in the central region of the Cuban archipelago and at 8 411.81 km² is the fourth-largest Cuban province, representing 7.6% of the country’s total landmass. Administratively it is divided into 13 municipalities and has a population of 780,749. Between January 2013 and December 2018, a total of 46,007 births occurred in the province [7].

Aim of the Study

The aim of this study was to describe trends in prevalence at birth of AWD, frequency by fetal sex, maternal age-specific prevalence, accuracy of prenatal diagnosis (PND) and the sensitivity of prenatal diagnosis of congenital AWD by serum AFP and by US.

Patients and Methods

This retrospective, observational study was performed from January 1994 to December 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 CDs.

These registries cover livebirths, fetal deaths from 20 weeks' gestational age and all terminations of pregnancy for any CDs and they include a textual description of all the observed CDs performed either by US or by physical examination. The different malformations were coded by trained staff of the Villa Clara provincial department of medical genetics according to the 10th revision of the International Statistical Classification of Diseases and Related Health Problems for Diagnoses (ICD-10), accompanied with a written description for each CDs.

The original records of RECUMAC and RECUPREMAC in the Villa Clara province are stored at the Provincial Department of Medical Genetics (PDMG). We manually scrutinized all of them for the study time period. Data of the provincial statistical department, the Villa Clara Provincial Health Direction was consulted for population statistics, as the live births number per years.

High levels of AFP were considered in cases with AFP values greater than 2 multiple of the normal median at 15 - 19 weeks of pregnancy.

The prevalence rate at birth of AWD was calculated by dividing the numerator (registered cases of LB, stillbirth or elective pregnancy terminations with any AWD) 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. The overall birth prevalence of abdominal wall defects was determined for the whole period of twenty-six years under study. Besides, the prevalence was estimated for each specific type of AWD in two different periods (from 1994 to 2006 and from 2007 to 2019).

**Statistical analysis**

Descriptive statistics are presented as frequencies and percentages or median and ratio for categorical and continuous variables, respectively. We conduct a statistical hypothesis test on the basis of the results of the Z test to compare proportions. Categorical data were analyzed using Pearson’s Chi-square test ($X^2$) and Fisher’s exact test (FET). The odds ratio (OR) and 95% confidence interval were calculated to evaluate the risk of maternal age. $P$ value < 0.05 was considered statistically significant. Statistical analysis was done using SPSS Version 22 software.

**Ethics**

This study is based on data analysis from records from which all identifying information had been removed, guaranteeing complete patient anonymity. The study was approved by the Ethics Committee of Villa Clara Medical University’s Biomedical Research Unit.

**Results**

A total of 143 cases of AWD were recorded, of these, 87 (60.84%) were GS, 44 (30.77%) were OM, and 12 (8.39%) were others AWD. Among the latest group, four cases with limb-body wall complex, one case with body stalk anomaly, another with Ectopia cordis and three cases with OEIS complex and pentalogy of Cantrell, respectively were included.

Elective pregnancy termination occurred in 124/143 cases (86.71%), in 3/143 intrauterine fetal death was described (2.09%), 16/143 were LB (11.19%) and no cases of affected stillbirths was reported. Six of these LB cases, (37.5%) had PND and parents decided to continue pregnancies.

The cases were ascertained from 168 827 LB. The overall prevalence of AWD (per 10,000 LB) was 8.5. The prevalence of GS was 5.2 cases, for OM was 2.6 cases and 0.7 for others congenital AWD. That is, one case of GS in 1941 LB, whereas OM was found in 1: 3837 LB.

There was an increase of 34.57% of the birth prevalence (per 10,000 LB) for all AWD during the study timeframe, with rates of 7.03 in the first thirteen years of study and 9.46 in the second period. GS had the highest increase (36.5%) from 4.30 to 5.87, although there were not statistically significant differences between these prevalence rates (Table 1).

<table>
<thead>
<tr>
<th>Type of AWD</th>
<th>First Period</th>
<th>Rate</th>
<th>Second Period</th>
<th>Rate</th>
<th>%</th>
<th>p</th>
</tr>
</thead>
<tbody>
<tr>
<td>Gastroschisis</td>
<td>33</td>
<td>4.30</td>
<td>54</td>
<td>5.87</td>
<td>36.5</td>
<td>0.37</td>
</tr>
<tr>
<td>Omphalocele</td>
<td>18</td>
<td>2.34</td>
<td>26</td>
<td>2.83</td>
<td>17.3</td>
<td>0.46</td>
</tr>
<tr>
<td>Others AWD</td>
<td>5</td>
<td>0.65</td>
<td>7</td>
<td>0.76</td>
<td>14.5</td>
<td>0.49</td>
</tr>
<tr>
<td>Total</td>
<td>54</td>
<td>7.03</td>
<td>87</td>
<td>9.46</td>
<td>34.6</td>
<td>0.30</td>
</tr>
<tr>
<td>Total live births</td>
<td>76831</td>
<td></td>
<td>91996</td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

According, the distribution by sex of cases with AWD, 84/143 (58.74%) were males, 55/143 (38.46%) were females, whereas in 4/143 (2.80%) was no possible to determine their sex. Gender was documented in all but three cases, (two GS cases and one patient with OM and with other AWD, respectively).

Mothers below 20 years of age represented 36.8% of all cases of GS and the prevalence for this group was 14.2 per 10,000 LB, whereas in OM and other congenital AWD the highest prevalence rates were in mother aged 35 years and more (8.7 and 1.5 per 10,000 LB, respectively). Moreover, 74/87 (85.06%) and 13/87 (14.94%) cases of GS were detected in mothers aged 24 years or less and older mothers, respectively (Table 2).

<table>
<thead>
<tr>
<th>Maternal ages</th>
<th>Anterior Wall Defects</th>
<th>Total</th>
<th>Births by maternal ages</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>GS No. Rate</td>
<td>OM No. Rate</td>
<td>Others AWD No. Rate</td>
</tr>
<tr>
<td>&lt; 20</td>
<td>32 14,21</td>
<td>3 1,33</td>
<td>3 1,33</td>
</tr>
<tr>
<td>20-24</td>
<td>42 10,09</td>
<td>11 2,64</td>
<td>4 0,96</td>
</tr>
<tr>
<td>25-29</td>
<td>9 1,62</td>
<td>13 2,34</td>
<td>2 0,4</td>
</tr>
<tr>
<td>30-34</td>
<td>4 1,14</td>
<td>5 1,43</td>
<td>1 0,36</td>
</tr>
<tr>
<td>≥ 35</td>
<td>0 0,0</td>
<td>12 8,71</td>
<td>2 1,45</td>
</tr>
<tr>
<td>Total</td>
<td>87 5,15</td>
<td>44 2,61</td>
<td>12 0,71</td>
</tr>
</tbody>
</table>

Table 2: Birth prevalence of congenital abdominal wall defects per 10,000 live births related to maternal ages, Villa Clara, Cuba. 1994 - 2019.

AWD: Abdominal Wall Defects; GS: Gastroschisis; OM: Omphalocele. Others AWD: Others Abdominal Wall Defects; No.: Number of cases by each age group and number of births by maternal ages; Rate: Prevalence rate per 10,000 live births; %. Percent.

The incidence of all AWD as group was significantly associated with decreasing maternal age: less than 20 years: \(X^2 = 21.71\); age 20 - 24: \(X^2 = 20.74\); age 25 - 29: \(X^2 = 8.73\); age 30 - 34: \(X^2 = 12.85\) and 35 years and over: \(X^2 = 0.51\).

Compared to women age less than 20 years (OR = 2.35, IC: 1.64 - 3.37), odds of all AWD as group decreased with advancing maternal age (age 20 - 24: OR 1.96; age 25 - 29: OR 0.54; age 30 - 34: OR 0.34 and age 35 and more: OR 0.27.

GS was significantly associated with maternal age less than 24 years old (\(X^2 = 21.71, p = 0.000, \text{FET}= 1.82, \text{IC}: 1.27 - 2.42\)) but the risk is even higher in women less than 20 years old (\(X^2 = 41.39, p = 0.000, \text{FET}= 3.22, \text{IC}: 2.52 - 5.67\)); whereas OM was significantly associated with maternal age over 35 years old (\(X^2 = 21.48, p = 0.000, \text{FET}= 4.22, \text{IC}: 2.3 - 7.78\) as its shown in table 3, where to facilitate the visualization of the results of statistical analysis, we reduce the maternal ages groups (Table 3).

<table>
<thead>
<tr>
<th>Congenital defect</th>
<th>Age groups</th>
<th>Less than 20 years</th>
<th>20-34 years</th>
<th>35 years and more</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>(X^2(p))</td>
<td>FET (p, OR, CI)</td>
<td>(X^2(p))</td>
<td>FET (p, OR, CI)</td>
</tr>
<tr>
<td>All AWD</td>
<td>21,71 (0,000)</td>
<td>1,8-05 (2,35) [1,64-3,37]</td>
<td>1,82 (0,17)</td>
<td>0,09 (0,86 [0,69-1,07]</td>
</tr>
<tr>
<td>Gastroschisis</td>
<td>41,39 (0,000)</td>
<td>3,2-08 (3,78) [2,52-5,67]</td>
<td>12,06 (0,000)</td>
<td>4,0-05 (1,89 [1,42-2,52]</td>
</tr>
<tr>
<td>Omphalocele</td>
<td>7,36 (0,006)</td>
<td>0,001 (0,23 [0,08-0,66]</td>
<td>8,63 (0,003)</td>
<td>0,001 (0,57 [0,39-0,83]</td>
</tr>
</tbody>
</table>

Table 3: Results of the statistical analysis of congenital abdominal wall defects related to mother's age groups, Villa Clara, Cuba. 1994 - 2019.

AWD: Abdominal Wall Defects; \(X^2(p)\): Chi Square Value and (p Value); OR: Odds Ratio; FET: Fisher’s Exact Test; CI: Confidence Interval.

Maternal age for GS cases had a range of 14 to 31 years, (mean age 21.1 years). Meanwhile, the mean maternal age for OM and other AWD was 28.2 (range of 17 to 42 years old) and 25.7 (range of 19 to 38 years old), respectively.

PND was performed in 133/143 (93.0%) cases of congenital AWD. It was more accurate in GS (82/87, 94.3%), followed by others congenital AWD (11/12, 91.7%) and OM (40/44, 90.9%).

PND of congenital AWD improved significantly from 87.0% during 1994-2006 period to 98.9% between years 2007 - 2019, (p = 2.9E-04) with a significant improving for GS and others congenital AWD from 84.9% and 80.0% respectively, in period 1994-2006 to 100% of cases prenatally diagnoses for the period 2007-2019. (p = 9.4 E-12 and p = 9.7 E-03, respectively) (Table 4).

<table>
<thead>
<tr>
<th>Type of AWD</th>
<th>Period from 1994 to 2006</th>
<th>Period from 2007 to 2019</th>
<th>Z</th>
<th>p</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>No.</td>
<td>%</td>
<td>No.</td>
<td>%</td>
</tr>
<tr>
<td>Gastroschisis (n = 33 and n = 54)</td>
<td>28</td>
<td>84.9</td>
<td>54</td>
<td>100.0</td>
</tr>
<tr>
<td>Omphalocele (n = 18 and n = 26)</td>
<td>15</td>
<td>83.3</td>
<td>25</td>
<td>96.2</td>
</tr>
<tr>
<td>Others AWD (n = 5 and n = 7)</td>
<td>4</td>
<td>80.0</td>
<td>7</td>
<td>100.0</td>
</tr>
<tr>
<td>Total (n = 54 and n = 87)</td>
<td>47</td>
<td>87.0</td>
<td>86</td>
<td>98.9</td>
</tr>
</tbody>
</table>

Table 4: Prenatal diagnosis of congenital abdominal wall defects in two different periods in Villa Clara, Cuba. 1994 - 2019.

High levels of AFP allowed the diagnosis of 72 cases with AWD; 77.8% of them (56/72) were GS cases, whereas the 42.6% of cases detected by means of fetal US (26/61) were GS cases. On the other hand, fetal US enabled diagnosis of 47.5% (29/61) of OM cases in contrast with only 15.3% diagnosed by means of maternal AFP determination. These differences were statistically significant (Table 5).

<table>
<thead>
<tr>
<th>Type of AWD</th>
<th>No.</th>
<th>%</th>
<th>No.</th>
<th>%</th>
<th>X²</th>
<th>P</th>
</tr>
</thead>
<tbody>
<tr>
<td>Gastroschisis</td>
<td>56</td>
<td>77.8</td>
<td>26</td>
<td>42.6</td>
<td>17.26</td>
<td>0.000</td>
</tr>
<tr>
<td>Omphalocele</td>
<td>11</td>
<td>15.3</td>
<td>29</td>
<td>47.5</td>
<td>16.35</td>
<td>0.000</td>
</tr>
<tr>
<td>Others AWD</td>
<td>5</td>
<td>6.9</td>
<td>6</td>
<td>9.8</td>
<td>0.36</td>
<td>0.546</td>
</tr>
<tr>
<td>Total</td>
<td>72</td>
<td>100</td>
<td>61</td>
<td>100</td>
<td>18.38</td>
<td>0.000</td>
</tr>
</tbody>
</table>


High levels of AFP were more frequently detected in GS cases compared to OM cases, with a statistically significant difference (Table 5).

In 68.3% of GS prenatally diagnosed cases (56/82), the diagnosis was performed by means of AFP screening, but only in 27.5% of cases with OM (11/40). Meanwhile, the sensitivity of US in AWD cases was 42.7% (61/143), with major accuracy in OM (29/44, 65.9%) in comparison with 29.9% (26/87) in GS.
Discussion

The prevalence rate of OM (per 10,000 live births) during the study timeframe was one-half those of GS (2.6 and 5.2 respectively). Our study showed slight higher results than other reports which stated that the prevalence of OM was 1.92 per 10,000 LB in United States of America (USA) and 2.17 per 10,000 LB in Singapore [8,9].

It is consistent with trends described in the literature where is claimed that the incidence of OM has plateaued since the 1970s and is now less common than GS at around 1 to 2.5 per 10,000 LB or even low: 0.6-2.2 per 10,000 LB [10,11].

Nevertheless, other report stated that OM occurs around 4 per 10,000 pregnancies, with a possible increase from 2.9 to 4.3 per 10,000 births from 1997 to 2007 [12].

We found one case of GS in 1941 LB, which it’s slightly higher than findings of different authors whom stated that GS is a fairly common CDs with incidence around 1 per 2000 - 3000 LB [13-16]. But it’s in accordance with the current incidence found in United Kingdom, where it varies from 1 per 3000 to 1:8000 LB [11].

According to the two periods of 13 years each, the prevalence rate for GS in Villa Clara was increased around 37%. This increasing prevalence is concordant with ongoing concerns of a worldwide increase in the prevalence of GS [10,14,15,17-22].

In a series of 18 years with data pooled from 15 USA states by Jones, et al. [20] they reported an increasing of 30% in GS prevalence when compared two time periods (1995 - 2005 and 2006 - 2012).

The prevalence of the AWD varies worldwide and tends to be lower in Asian countries [23]. For instance, the prevalence of GS during 2006 to 2015 in the Chinese province of Liaoning, was 2.30 per 10,000 LB with a significant decreasing of 12.63% per year for the 10-year period of study [24].

Even smaller prevalence’s were described in Taiwan and Japan. A study conducted in Taiwan concluded that the prevalence of GS and AWD from 1998 to 2013 was 0.50 and 1.65 per 10,000 LB, respectively and decreased over time [23]. Meanwhile, in Japan even when the prevalence of GS increased from 0.1 per 10,000 LB in 1975-1980 to 0.5 per 10,000 LB in 1996-1997, it still remains very low [25].

In contrast, according to the population-based CDs data in the USA, 2012-2016, the overall prevalence estimates (per 10,000 LB) were 4.3 for GS and 2.1 for OM [8]. In accordance with European registries, where the data of the EUROCAT working group revealed that the total prevalence of GS was 3.09 per 10,000 births and LB prevalence was 2.63 per 10,000 in 2011 [15]. Nevertheless, in the largest population-based study summarizing current epidemiology of GS in California (USA) from 1995 to 2012, the prevalence of GS was 2.7 cases per 10,000 LB [18].

A higher OM prevalence was found over 22 years of data collection in the Wessex region of England and the Channel Isles, where 335 cases of OM from 614 321 births were reported from 1994 to 2015, for a prevalence of 5.45 per 10,000 pregnancies [12].

However, to the best of our knowledge, the highest worldwide prevalence rates were reported in a study conducted in the world’s largest island (Greenland) from 1989 to 2015, where the annual point prevalence for GS increased from 8.0 to 35.1 (average 10.7) per 10,000 LB and stillborn infants. For OM, the prevalence varied from 8 to 11 per 10,000 LB and stillborn infants [26].

Our data show an increase in the prevalence rate of AWD from 7.03 to 9.46 per 10,000 LB over the twenty-six years of the study, with rising in both, GS and OM prevalences; in contrast with other authors whom claimed that the worldwide incidence of GS has risen inexo-
rably over the past 30 years, whereas the incidence of OM, at least at the time of birth, has been either relatively static [10,26] or with modest increases over time [19].

In all cases of AWD, including GS and OM, we found a male predominance, but it was lower in GS than in OM (55.2% and 65.9%) respectively. This finding is consistent with that reported in the literature [8,13,16,27]. Although in other studies GS prevalence has not been shown to differ significantly by infant sex [10,22] or unlike GS, the prevalence of OM has been observed to differ by infant sex, with a higher prevalence in males compared with females [8,27].

In contrast, in a study conducted from 2008 to 2019 in Malaysia, where 73 cases of OM were identified, there was female sex preponderance (53.4%) versus 39.7% of male sex [11]. Whereas Gábor, et al. [28] described in both cases of GS and OM, similar female dominance.

We found the highest GS prevalence among younger mothers aged 24 or less years and, especially in teenager’s mothers aged less than 20 years, in line with several previous studies; where the association between GS and low maternal age has consistently been documented [2,15,18-21,26,29,37].

Whereas the highest OM prevalence rate in our series was observed among mothers aged 35 years or more, with a range of ages from 17 to 42 years. Our results support the findings of Marshall., et al. [8] who reported an increase in OM in offspring of older mothers.

The range of ages found in Villa Clara province in mothers of GS and OM cases is, by far, wider than the observed in a previous study in Greenland, where a range of 16 to 31 years for GS and 21 to 34 years for mothers of infants with OM was described [26].

Young maternal age has been recognized as one of the strongest risk factors for GS [18,20,26,27]. Younger mothers are significantly more likely than women older than this age to have a child with this congenital defect [15].

However, in a study of the increasing prevalence, time trend and seasonality of GS in Sao Paulo state, Brazil, from 2005 to 2016; a significantly increased of GS prevalence per year was observed and this trend was higher in mothers aged 30 - 34 years than in mothers of other age groups [30].

In the literature, studies examining the prevalence for OM by maternal age, reported a higher prevalence among women of advanced maternal age [8,31]. Other studies have reported inconsistent results regarding maternal age [19,32] and Thakur, et al. [2] stated that, as compared to GS, the mothers age did not produce a significant risk factor for OM.

PND programmes are designed to detect CDs during early pregnancy, thus allowing couples to decide whether to terminate or continue the pregnancy. If a couple chooses to continue the pregnancy, the early diagnosis enables them and the healthcare provider to set a strategy for the child’s treatment and follow-up. Routine prenatal screening and diagnosis of the AWD and concurrent anomalies is important as it allows for effective prenatal counseling and optimal perinatal management [32,33].

Maternal serum biomarkers have been used worldwide in prenatal screening for decades. AFP is a 69 kDa fetal-derived glycoprotein which is mainly produced by yolk sac, gastrointestinal tract and fetal liver in early pregnancy; which is encoded by a gene with locus at 4q11-q13 [5].

Screening the levels of AFP in maternal serum is a valuable tool in the diagnosis of a number of CDs by means of the US. In the present study this biomarker allows prenatally diagnosing the double of cases of GS (89%) compared with of OM cases (41%) and is thought to be due to free diffusion of AFP from the fetal circulation into maternal circulation [1].
AFP has proven to be more sensitive in detecting GS than omphalocele, likely due to the intact, overlying membrane in OM. An elevated maternal AFP level on routine obstetrics blood screening should prompt a comprehensive US evaluation [33]. In our series when AFP was combined with the US evaluation allowed detecting prenatally over 90% of all AWD cases (94% and 91% in GS and OM, respectively).

Our data revealed that prenatal sonographic diagnosis was more accurate in OM than in GS (66% vs 30%) in line with previous studies that claimed that US remains the mainstay for the PND of OM, with high sensitivity for these anomalies from about 14 weeks’ gestation, but more typically at 20 - 22 weeks of gestation [15,33].

PND has a high level of accuracy in the Villa Clara Medical Genetics Service, in the current study 93% of cases with AWD, 133 of 143 cases was prenatally diagnosed, this allows antenatal following by clinical geneticists at the PDMG and planning deliveries and outcomes at the provincial Maternity and Pediatric hospitals, located at the Santa Clara city, centers with experience in neonatology and perinatal care, pediatric surgery and specialized obstetrics services for all pregnant women.

These results are encouraging when viewed as a reflection of medical genetics care provision to all pregnant women in Cuba, integrated under the National Program for diagnosis, management and prevention of genetic diseases and CDs.

Accuracy of prenatal diagnoses of the AWD varies widely worldwide. In a series of 335 OM cases in a region of United Kingdom 92% of cases (307/335) were detected prenatally [12]. In South America, in a study conducted to assess the accuracy of PND of CDs in 18 South American hospitals on the basis of prenatal sonographic records, 33 of 38 AWD were prenatally diagnosed, that is a detection rate of 86.8% [34].

In a retrospective study of 19 newborns undergoing surgery for AWD (8 patients with OM and 11 cases of GS) conducted by Watanabe, et al. [29] over 16 years of experience from a single clinic in Japan, PND was made in 17 of the 19 cases (89.47%) with detection rates of 87.5% and 90.9% for OM and GS, respectively.

In a population-based case-malformed control study conducted by Given, et al. [21] using data from 18 European congenital anomaly registries, 1577 GS cases were identified, of whom 69% (1088/1577) were prenatally diagnosed, including cases with termination of pregnancy for fetal anomaly.

Other series reported lower prenatal detection rates, for instance, in a prospective study of 42 neonates with GS in Kampala, Uganda; 24% (10/42) of mothers had undergone an antenatal US scan, but only one (10%) had been given the correct diagnosis. In this country, antenatal US scans that do happen, are often performed in the private sector with varying levels of reliability [35].

In a series of 73 OM cases, identified at the neonatal unit of a tertiary hospital in Yogyakarta, Indonesia; from 2008 to 2019, only 15.1% (11/73) of OM cases were diagnosed prenatally by US [11].

Moreover, Hasan, et al. [36] in a study done in the largest tertiary care pediatric surgery center, from 2014 to 2017, in Bangladesh; found only 4% of GS patients (3/75) with PND. Authors stated that this is due to less public awareness about prenatal care and screening in a developing country like Bangladesh and the limited experience of radiologists and primary caregivers at rural area might also be contributing factors.

Despite studies on the prevalence of AWD and the increasing prevalence of GS, the etiology of these defects remains poorly understood. Moreover, given the prevalence and clinical impact of these defects, further research is needed to gain insight into the etiology and differences reported in the birth prevalence by various maternal and infant characteristics for these birth defects [22].

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Conclusion

In conclusion, a rise in AWD cases has been noted in recent decades in Villa Clara. The present study revealed a pattern for young mothers of cases with GS and older maternal age for OM. The use of fetal US and AFP screening in medical genetics care services in Villa Clara allowed the PND of the majority of AWD, with subsequent opportunities for parental genetics counseling, fetal intervention, and optimal perinatal management.

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Authors’ Contributions

All authors have contributed either obtaining or analyzing data as well as in the article preparation. They have read and approved the final manuscript.

Competing Interests

The authors have no competing interests to declare.

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