RESEARCH ARTICLE

DIAGNÓSTICO PRENATAL MEDIANTE AMNIOCENTESIS EN GESTANTES DE CÚCUTA, NORTE DE SANTANDER (ENERO DE 2018 A DICIEMBRE DE 2023)

PRENATAL DIAGNOSIS THROUGH AMNIOCENTESIS IN PREGNANT WOMEN FROM CÚCUTA, NORTH SANTANDER (JANUARY 2018 TO DECEMBER 2023)

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RESUMEN

El diagnóstico prenatal es el pilar fundamental para disminuir las complicaciones fetales, lo que ha promovido el avance en métodos diagnósticos y disminuir el riesgo fetal. En Colombia es reducida la disponibilidad de técnicas no invasivas lo que hace que la amniocentesis sea el método principal para la determinación de trastornos genéticos, que según la técnica y el factor operador son mínimos los riesgos durante el procedimiento. El objetivo de este estudio fue determinar la prevalencia de la Amniocentesis diagnóstica en la ciudad de Cúcuta, Norte de Santander, entre los años 2018 y 2023, por medio de un enfoque cuantitativo, con metodología descriptiva de corte transversal. Durante este periodo de tiempo se realizaron 124 amniocentesis en gestantes cuyas edades oscilaron entre 16 y 46 años, de las cuales el 12.1% presentó alteraciones cromosómicas en el reporte, siendo el síndrome de Patau y el síndrome de Edwards los más frecuentes. A partir de este estudio se evidenció la importancia de los hallazgos fetales por medio de la ecografía como principal indicación para la amniocentesis y el diagnóstico temprano conlleva a la reducción de complicaciones materno-fetales y así mismo permite la preparación para la llegada de un hijo con alteración cromosómica.

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Palabras clave: Amniocentesis, Diagnóstico Prenatal, Anomalías cromosómicas, Aneuploidía, Trisomía.

ABSTRACT

Prenatal diagnosis is the fundamental pillar to reduce fetal complications, which has promoted advancements in diagnostic methods and decreased fetal risk. In Colombia, the availability of non-invasive techniques is limited, making amniocentesis the primary method for determining genetic disorders, where the risks during the procedure are minimal due to the technique and operator factor. The objective of this study was to determine the prevalence of diagnostic amniocentesis in the city of Cúcuta, Norte de Santander, between 2018 and 2023, using a quantitative approach with a descriptive, cross-sectional methodology. During this period, 124 amniocenteses were performed on pregnant women aged between 16 and 46 years, of which 12.1% presented chromosomal abnormalities in the report, with Patau syndrome and Edwards syndrome being the most frequent. This study highlighted the importance of fetal findings through ultrasound as the main indication for amniocentesis, and early diagnosis leads to a reduction in fetal and maternal complications and also allows for preparation for the arrival of a child with a chromosomal alteration

Keywords: Amniocentesis, Prenatal Diagnosis, Chromosomal Anomalies, Aneuploidy, Trisomy.

Introducción.

Amniocentesis is an invasive prenatal diagnostic test performed in the second trimester of pregnancy to detect chromosomal abnormalities, single-gene disorders, fetal infection and intra-amniotic inflammation, as well as the degree of hemolytic anemia, blood type, or platelets. This procedure utilizes a technique that involves the extraction of amniotic fluid from the uterine cavity by introducing a needle through a transabdominal approach under continuous ultrasound guidance, which allows for obtaining a sample of exfoliated fetal cells, transudates, urine, or secretions. (1-2).

The development of non-invasive tests for effective first-trimester screening of chromosomal abnormalities, such as combined risk assessment using ultrasound and biochemical markers, as well as cell-free fetal DNA analysis in maternal blood, has significantly reduced the current use of amniocentesis (3).

For prenatal genetic studies, amniocentesis can technically be performed at any time after 12-13 weeks of gestation ; however, the optimal period for performing this

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procedure is between 15-16 and 18-19 weeks of gestation. (4).

It was first performed for the diagnosis of fetal genetic disorders by Fuchs and Riis in 1956. The most common indications for prenatal genetic diagnosis include advanced maternal age (>35 years), abnormal biochemical markers in first and second-trimester screening, ultrasound detection of fetal anomalies, a personal or family history of chromosomal abnormalities in previous pregnancies, abnormal prenatal karyotype, and balanced parental translocation. (5)

Amniocentesis is associated with complications mainly such as vaginal bleeding in 2-3%, feto-maternal hemorrhage 2.6%, premature rupture of membranes 1.7%, infection 0.1%, and fetal loss in 0.11% (5,6,7).

Prenatal screening for the most common fetal aneuploidies includes Down syndrome (trisomy 21), Edwards syndrome (trisomy 18), and Patau syndrome (trisomy 13). (8,9)

Globally, congenital anomalies account for 2.1% of individuals with disabilities, with approximately 434,000 deaths occurring each year, 97% of which take place in developing countries, contributing to 3.3 million deaths among children under five years of age. In Latin America, the prevalence of disability due to congenital defects is approximately 4.5%, prompting the adoption of policies aimed at raising awareness of infant morbidity and mortality, as well as promoting access to the prevention and treatment of congenital anomalies. Given the global impact of congenital anomalies, the study of amniotic fluid plays a critical role in providing genetic counseling for pregnant women (10,11).

In our region of Norte de Santander, access to genetic diagnosis through non-invasive prenatal testing is limited. As a result, amniocentesis has become the cornerstone of prenatal diagnosis for congenital anomalies. The majority of these procedures are performed at the Maternal-Fetal Medicine Unit (NORFETUS), as the analysis of amniotic fluid is not conducted in the city of Cúcuta and must be sent to Bogotá. This administrative limitation prevents the procedure from being carried out in local clinics and at the Erasmo Meoz University Hospital. Therefore, given the significance of this procedure, understanding its prevalence, the characteristics of the patients who have undergone amniocentesis, and the perinatal outcomes will support the continued integration of screening for these conditions and the early

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detection of maternal-fetal risk.

Métodos.

This is a descriptive cross-sectional study, in which reports of amniocentesis procedures performed on pregnant women who attended the Maternal-Fetal Medicine Unit NORFETUS, in the city of Cúcuta, Norte de Santander, Colombia, between 2018 and 2023 were reviewed. The objective was to determine the most frequent indications for this procedure, the characteristics of the pregnant women who underwent it, abnormal findings in the extracted amniotic fluid, the prevalence of the resulting diagnoses, and perinatal outcomes.

Each amniocentesis report was recorded, collecting information on maternal age, gestational age, indication for the procedure, number of fetuses evaluated, karyotype results, and complications. In addition, telephone surveys were conducted with mothers whose amniocentesis results were pathological, in order to obtain information on perinatal outcomes. All collected data were compiled into a database for descriptive statistical analysis, and the relevant population variables were evaluated.

Inclusion criteria consisted of pregnant women with a medical indication for amniocentesis during the established time period, regardless of nationality or age. Exclusion criteria included cases lacking complete information on the required variables. The final sample included 124 diagnostic amniocentesis procedures (an average of 1.7 cases per month), of which 39 were excluded due to incomplete data; none of these excluded cases had pathological findings in the amniotic fluid. Ultimately, a total of 85 amniocentesis procedures were analyzed, allowing for the characterization of the studied pregnant population.

Resultados.

A total of 85 amniocentesis procedures were included, performed over a six-year period at the Maternal-Fetal Medicine Unit NORFETUS, in the city of Cúcuta, Colombia. These procedures were carried out in pregnancies ranging from 14 to 36 weeks of gestation, with the majority

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occurring between 23 and 27.6 weeks (35.3%) (see Figure 1). The most common gestational age at the time of puncture was 18 weeks. All patients who underwent the procedure had singleton pregnancies; no intra-amniotic punctures were performed in multiple pregnancies. Maternal ages ranged from 16 to 45 years, with a mean age of 24 years. Pregnant women aged 35 years or older represented 27% of the analyzed population (see Figure 2).

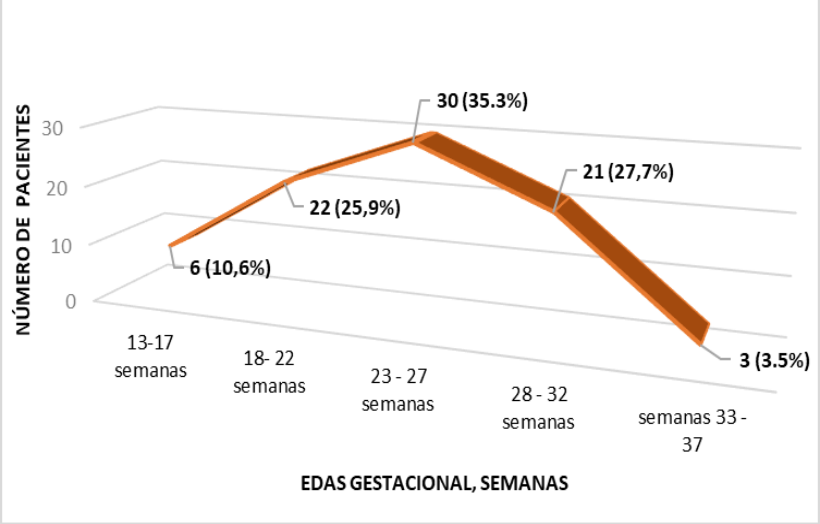

Figure 1. Gestational Age of Pregnant Women at the Time of Amniocentesis

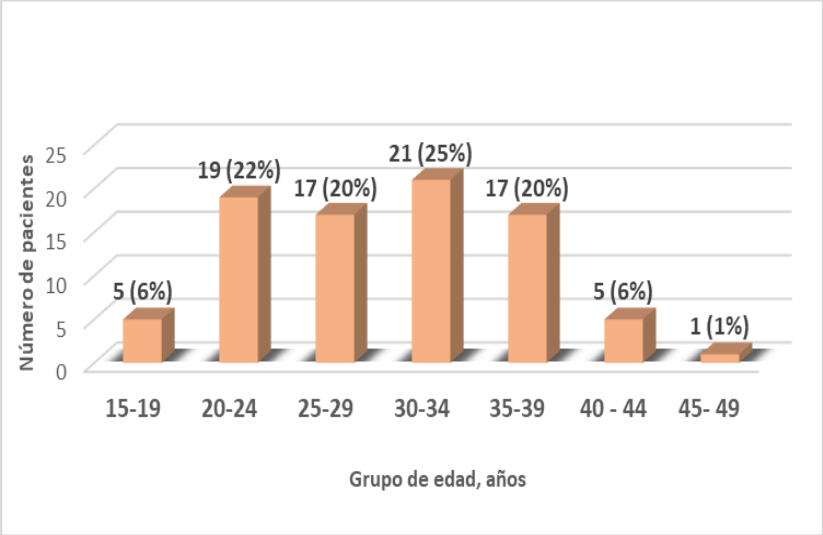

Figure 2. Patient's Age at Diagnostic Amniocentesis.

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The vast majority of women underwent amniocentesis due to suspected fetal aneuploidy, based on the presence of fetal anomalies identified during prenatal ultrasound evaluation (73%). The second most common indication was suspected toxoplasmosis infection (27%) (see Figure 3). Among the amniotic fluid results in patients who underwent the procedure due to suspected active toxoplasmosis infection, it is noteworthy that 100% of the results were negative.

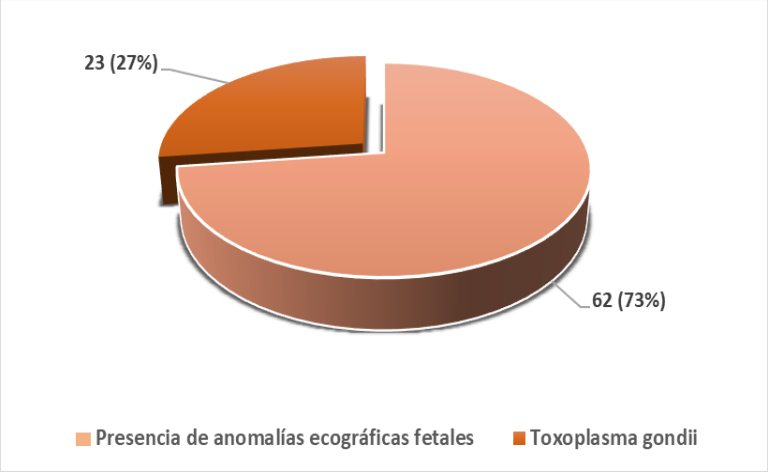

Figure 3. Diagnostic Study Indication.

Of the total population studied (124 patients), 12.1% of the women who underwent amniocentesis were found to have abnormal chromosomal results (see Figure 4)

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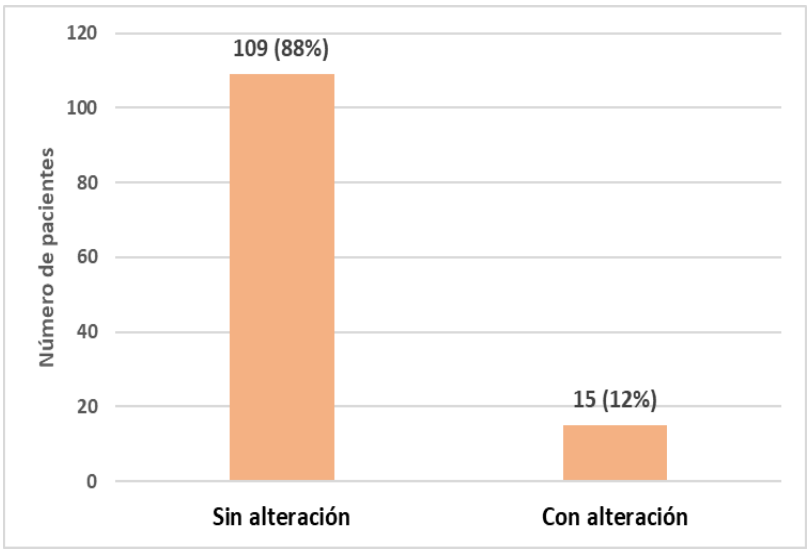

Figura 4. Resultados de la Amniocentesis

Among the 15 amniocentesis procedures with pathological findings, the most frequently diagnosed chromosomal abnormalities were Patau syndrome (4 cases, 26.7%) and Edwards syndrome (4 cases, 26.7%), followed by Down syndrome and Turner syndrome, each with 3 cases (20%). There was only one case of Rethoré syndrome (trisomy 9p), accounting for 6.6% of the pathological findings (see Figure 5).

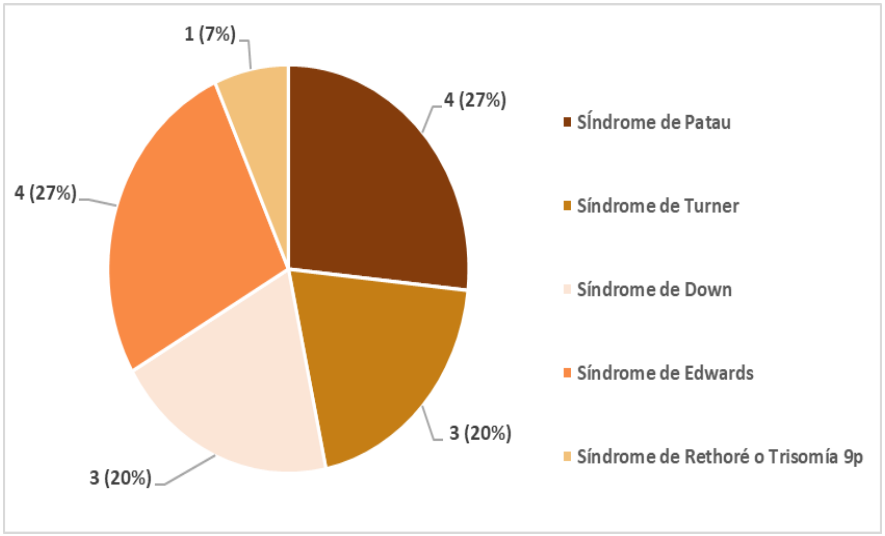

Figure 5. Chromosomal abnormalities diagnosed by Amniocentesis in Cúcuta – Colombia.

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A survey on perinatal outcomes was successfully conducted with 33.4% of the women who had pathological amniocentesis results, despite the time elapsed since the earliest diagnoses and the emotional impact on the patients. Based on the information provided by the respondents, the following outcomes were identified: two cases of intrauterine fetal death (at 28 and 37 weeks of gestation), associated with Down syndrome and Edwards syndrome, respectively; one case of pregnancy termination at 29 weeks due to Patau syndrome; and two cases of live births. One of these was a full-term infant with Turner syndrome who is currently alive, while the other was a preterm newborn with Edwards syndrome who was admitted to the Neonatal Intensive Care Unit and died at two months of age.

During the performance of diagnostic amniocentesis in the total study population, no complications or adverse events were observed. There were no reported cases of procedure-related complications.

It is worth noting that in the excluded population, two cases were identified in which amniocentesis results showed absence of cellular growth in culture, thereby preventing the generation of karyotype reports.

Discusión.

This research study reports a 12.1% prevalence of chromosomal abnormalities among the 124 amniocentesis procedures performed over a six-year period at the Maternal-Fetal Medicine Unit – NORFETUS. This figure is comparable to international data and slightly lower than that reported in another Colombian study conducted in Bogotá, which found a 17.5% prevalence of chromosomal abnormalities among pregnant women (3). At the Latin American level, a study in Costa Rica involving 625 pregnant women reported fetal chromosomal abnormalities in 9% of cases (12), while a study in Cuba with 803 participants found a prevalence of 2.7% (9). Similarly, a Mexican study reported chromosomal alterations in 4.5% of amniotic fluid samples (13). In Europe, a Turkish study with 6,124 cases identified chromosomal abnormalities in 3.6% of pregnancies (14), and in Asia, a Korean study found a prevalence of 3.1% (15).

Trisomies were the most common chromosomal abnormalities identified in this and other studies (9,13,14,16). Within the trisomies observed in our cohort, trisomy 13 (Patau syndrome)

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and trisomy 18 (Edwards syndrome) were the most frequent, each accounting for 26.7% of pathological cases, followed by Down syndrome (trisomy 21), which represented 20% of the cases. This percentage for Down syndrome is notably lower compared to other studies: 75% in the Cuban study, 47.1% in the Mexican study, and 46% in the Colombian study (3,9,13).

In contrast, the frequencies of trisomies 13 and 18 in our data were higher than those reported in other studies: 8.8% for trisomy 13 and 14.7% for trisomy 18 in the Mexican study; 4.3% and 11.8%, respectively, in a Turkish study; and 1.2% and 10.2%, respectively, in the Korean study. Another Turkish study from a different center reported 3.1% and 10.6% for trisomy 13 and 18, respectively (13,14,15,17). All referenced studies show a consistent trend of higher prevalence for trisomy 18 (Edwards syndrome) compared to trisomy 13 (Patau syndrome).

One case of Rethoré syndrome (trisomy 9p) was identified in the pregnant population of Cúcuta. This condition involves a duplication of the short arm of chromosome 9 and is extremely rare, with limited survival rates. It is characterized by psychomotor delay, malformations affecting various organs, and occasionally epilepsy. Rethoré syndrome is considered the fourth most frequent chromosomal disorder, following trisomies 21, 18, and 13. First described in a neonate in 1973, just over 150 cases have been reported worldwide to date (18,19,20).

In this study, 27% of the pregnant women who underwent amniocentesis were over 35 years of age. Although advanced maternal age is traditionally associated with a higher risk of chromosomal abnormalities, some studies do not support this conclusion (13,16). This emphasizes the importance of considering amniocentesis for pregnant women of all ages, particularly when fetal anatomical abnormalities are detected on ultrasound, which is considered a sufficient indication for invasive diagnostic testing (21). In our study, this indication accounted for 73% of cases, similar to the 47.1% reported in a study from Cali, Colombia (13,14,22,23). Amniocentesis plays an integral role in the basic prenatal care framework, enabling comprehensive and early evaluation of potential chromosomal anomalies (24). Abnormal ultrasound findings, especially during the second trimester, showed a significant correlation with the likelihood of genetic abnormalities during pregnancy (25). Therefore, diagnostic amniocentesis based on ultrasound findings emerges as a critical and consistent strategy to mitigate congenital defects in the context of prenatal diagnosis.

Conclusion

Amniocentesis is a prenatal diagnostic

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procedure that involves the analysis of amniotic fluid, primarily for the detection of genetic disorders. When performed by experienced professionals, it carries a low risk of complications and effectively identifies chromosomal abnormalities in fetuses with anatomical anomalies detected via ultrasonography. In the city of Cúcuta, Colombia, this diagnostic test is available, enabling improved management of fetuses and/or neonates with congenital anomalies.

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