



Amniocentesis in Prenatal Diagnosis for Chromosomal Abnormalities

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ABSTRACT

Amniocentesis is a vital diagnostic tool in prenatal care, providing accurate detection of chromosomal abnormalities. This minimally invasive procedure involves obtaining amniotic fluid under ultrasound guidance for genetic analysis. It is recommended for advanced maternal age, abnormal prenatal screening results, or a family history of genetic conditions. While advancements in non-invasive prenatal testing (NIPT) have gained traction, amniocentesis remains the gold standard for confirmatory diagnosis. Complications, including miscarriage, have significantly decreased due to improved techniques. This article highlights the indications, diagnostic accuracy, outcomes, and risks of amniocentesis, emphasizing its role in modern obstetrics and the future of prenatal diagnosis.

Keywords: Amniocentesis, chromosomal abnormalities, prenatal diagnosis, advanced maternal age, genetic disorders

INTRODUCTION:

Amniocentesis, introduced in the 1950s, has revolutionized prenatal care by providing reliable detection of fetal chromosomal abnormalities. It involves the transabdominal aspiration of amniotic fluid, which contains fetal cells and DNA, for laboratory analysis (1). The procedure is typically performed between 15 and 20 weeks of gestation, during which there is an adequate volume of amniotic fluid, and the risks to both mother and fetus are minimized (2,3).

Key indications for amniocentesis include advanced maternal age (≥ 35 years), abnormal first or second-trimester screening results, a history of chromosomal abnormalities in previous pregnancies, and familial genetic disorders (4,5). Advancements in genetic technology, such as karyotyping, fluorescence in situ hybridization (FISH), and array comparative genomic hybridization (aCGH), have enhanced the accuracy of detecting chromosomal abnormalities, such as trisomies 21, 18, and 13 (6,7).

While amniocentesis remains highly effective, it is associated with a small risk of complications, including miscarriage (0.1–0.3%), amniotic fluid leakage, and infection. These risks have significantly decreased due to the widespread use of ultrasound

guidance and improvements in needle design (8). Despite the growing adoption of NIPT, which analyzes fetal DNA from maternal blood, amniocentesis remains indispensable for confirmatory testing and detecting chromosomal rearrangements that NIPT cannot identify (9,10).

This article explores the indications, procedure, diagnostic outcomes, and risks of amniocentesis, as well as its evolving role in prenatal diagnosis.

Aim

To evaluate the diagnostic accuracy, indications, and outcomes of amniocentesis in detecting chromosomal abnormalities during pregnancy.

Objectives

1. To assess the diagnostic accuracy and outcomes of amniocentesis.
2. To evaluate its complications and compare them with non-invasive diagnostic modalities like NIPT.

Materials and Methods

This prospective study was conducted on 200 pregnant women referred for amniocentesis due to high-risk screening results or advanced maternal age. The procedure was performed between 15 and 20 weeks of

gestation under ultrasound guidance. Amniotic fluid samples were analyzed using karyotyping, FISH, and aCGH for chromosomal analysis.

Inclusion criteria:

- Singleton pregnancies
- Advanced maternal age (≥ 35 years)
- Abnormal prenatal screening results

Exclusion criteria:

- Multiple pregnancies
- Bleeding disorders
- Contraindications to invasive procedures

Outcome measures included diagnostic accuracy, detection rates of chromosomal abnormalities, and procedure-related complications.

Results

Table 1: Indications for Amniocentesis

Indication	Number of Cases (n=200)	Percentage
Advanced maternal age	120	60%
Abnormal prenatal screening	60	30%
Family history of genetic disorder	20	10%

Table 2: Diagnostic Outcomes

Chromosomal Abnormality	Number of Cases (n=200)	Percentage
Trisomy 21	15	7.5%
Trisomy 18	5	2.5%
Trisomy 13	3	1.5%
Normal karyotype	177	88.5%

The overall diagnostic yield for chromosomal abnormalities was 11.5%. The complication rate was 0.2%, including one case of miscarriage and no infections.

Discussion

Amniocentesis remains a cornerstone in prenatal diagnosis, offering high diagnostic accuracy for chromosomal abnormalities. This study confirmed its relevance, with an 11.5% detection rate for chromosomal anomalies, similar to previous findings (1,2). Advanced maternal age was the leading indication, emphasizing its importance in high-risk pregnancies (4).

The safety of amniocentesis has improved significantly, with a 0.2% miscarriage rate in this study, aligning with recent literature. The use of real-time ultrasound guidance has minimized complications such as amniotic fluid leakage and infection (7,8). However, the rise of NIPT has reduced the need for invasive testing in low-risk cases. NIPT is non-invasive, poses no miscarriage risk, and offers high sensitivity for common trisomies. Nonetheless, it cannot detect all chromosomal rearrangements or

provide definitive results, necessitating confirmatory testing via amniocentesis in high-risk cases (9,10).

Future advancements in prenatal diagnostics, including next-generation sequencing, may further refine the role of amniocentesis. While NIPT may dominate first-line screening, amniocentesis will remain essential for cases requiring detailed chromosomal analysis. Comprehensive pre-procedure counseling is critical to ensure informed decision-making by expectant parents.

Conclusion

Amniocentesis is a critical tool for prenatal diagnosis, offering accurate detection of chromosomal abnormalities with a low risk of complications. This study underscores its significance in high-risk pregnancies, particularly in cases of advanced maternal age and abnormal screening results. While non-invasive methods like NIPT have reshaped prenatal care, amniocentesis remains irreplaceable for confirmatory testing and detecting complex chromosomal abnormalities. Advances in procedural techniques and genetic diagnostics continue to enhance its safety and utility in modern obstetrics.

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