

# Indications and Complications of Amniocentesis in 16–20 Weeks in a Tertiary Center in Northern Kerala: A Descriptive Study

Anoop Venkatapura Bylaswamy<sup>1</sup>, Juvaina Puthiyakam<sup>2</sup>, Jyoti Ramesh Chandran<sup>3</sup>, Ellezhuthil Devarajan<sup>4</sup>

Received on: 14 June 2023; Accepted on: 04 December 2023; Published on: xxxx

## ABSTRACT

**Aim and background:** Chromosomal anomalies frequently occur. Around the world, at least 7.6 million kids are born every year with severe genetic or congenital deformities. One of the biggest problems in contemporary perinatology is the diagnosis of chromosomal abnormalities in fetuses. Trisomies 21, 18, 13, monosomy X, and other sex chromosome aneuploidies are the most prevalent chromosomal abnormalities found in infants. Prenatal diagnostics use a number of approaches to assess a fetus's health and condition. Invasive and non-invasive procedures can be used to diagnose pregnancies. Our objective was to identify indications and complications of amniocentesis.

**Materials and Methods:** A total of 111 pregnant women of gestational age between 16th and 20th weeks who either had abnormal first-trimester scan at 11–14 weeks with positive aneuploidy markers or abnormal mid-trimester anomaly scan with findings suggestive of underlying chromosomal disorders or intrauterine infections were referred from Department of Obstetrics and Gynecology to Department of Radiodiagnosis in Government Medical College, Kozhikode, to undergo amniocentesis to determine the fetal chromosomal structure, numerical aberrations, or translocations.

**Results:** The most common indication for amniocentesis was absent/hypoplastic nasal bone (78%), followed by the combination of absent/hypoplastic nasal bone with increased nuchal fold thickness (7%), smaller percentages including fetal ventriculomegaly (2%), combination of absent/hypoplastic nasal bone with short femur (1%). The complication rate was very minimal; only one patient had a bloody tap. None of the patients had any other complications, such as miscarriages/bleeding per vagina. Mild pain in the abdomen was experienced by a few patients.

**Conclusion:** The most common indication for amniocentesis was absent/hypoplastic nasal bone, followed by a combination of absent/hypoplastic nasal bone with increased nuchal fold thickness. The complications were very minimal.

**Clinical significance:** Amniocentesis is a safe procedure with minimal to no complications. Hence, it can be advised to patients with a positive screening test.

**Keywords:** Absent/hypoplastic nasal bone, Amniocentesis, Positive soft markers in second-trimester ultrasound, Prenatal genetic diagnostic testing.

*International Journal of Infertility and Fetal Medicine* (2024); 10.5005/jp-journals-10016-1335

## INTRODUCTION

Chromosomal abnormalities in fetuses can have minor to severe repercussions, such as severe physical and mental impairment and a short lifetime. Despite significant advancements in medical research, it is still difficult to diagnose chromosomal abnormalities in fetuses. Trisomies 21, 18, 13, monosomy X, and other sex chromosome aneuploidies are the most frequent chromosomal abnormalities found in infants.<sup>1</sup>

The most frequent reason for invasive prenatal testing is the prenatal detection of fetal chromosomal disorders. More than 50% of clinically confirmed early pregnancy losses involve chromosomal abnormalities. Aneuploid fetuses are responsible for 6–11% of stillbirths and neonatal mortality. Around 0.65% of newborns have chromosomal abnormalities that are compatible with life but cause significant morbidity, and 0.2% of newborns have structural chromosomal rearrangements that will eventually impact reproduction.<sup>2</sup>

Prenatal genetic diagnostic testing aims to ascertain with as much accuracy as possible if the fetus has a certain genetic illness or condition. Prenatal genetic testing, in contrast, aims to determine if a patient is more likely than not to have a fetus affected by a genetic condition.<sup>3</sup>

<sup>1,2,4</sup>Department of Radiodiagnosis, Government Medical College, Kozhikode, Kerala, India

<sup>3</sup>Department of Obstetrics & Gynecology, Government Medical College, Kozhikode, Kerala, India

**Corresponding Author:** Anoop Venkatapura Bylaswamy, Department of Radiodiagnosis, Government Medical College, Kozhikode, Kerala, India, Phone: +91 7259108046, e-mails: anoop\_1094@live.com; anoopvbusmle@gmail.com

**How to cite this article:** Bylaswamy AV, Puthiyakam J, Chandran JR, *et al.* Indications and Complications of Amniocentesis in 16–20 Weeks in a Tertiary Center in Northern Kerala: A Descriptive Study. *Int J Infertil Fetal Med* 2024; <https://doi.org/10.5005/jp-journals-10016-1335>.

**Source of support:** Nil

**Conflict of interest:** None

**Patient consent statement:** The author(s) have obtained written informed consent from the patient(s) for publication of the research details and related images.

As a result, there are well-established screening and diagnostic programs to identify the most prevalent autosomal trisomies in liveborn newborns, including Down syndrome.<sup>2</sup>

The term "amniocentesis" comes from the Greek words "pricking" and "amnon," which refer to the inner membrane that surrounds the fetus. This method is mostly utilized in the prenatal identification of chromosomal abnormalities and fetal infections. Amniocentesis was first applied in Germany in the early 1880s to treat hydramnios. Amniocentesis was previously used to help locate the placenta during fetal evaluations and even as a technique of pregnancy termination by injecting hypertonic saline into the uterus. In order to evaluate the health of the fetus, Alvarez performed an amniocentesis in Uruguay in 1950.<sup>4</sup>

Our objective was to study the indications and complications of amniocentesis.

## MATERIALS AND METHODS

A total of 111 pregnant women of gestational age between 16th and 20th weeks who either had abnormal first-trimester scan at 11–14 weeks with positive aneuploidy markers or abnormal mid-trimester anomaly scan with findings suggestive of underlying chromosomal disorders or intrauterine infections or high-risk for fetal aneuploidy detected by biochemistry—double marker/quadruple marker, previous child with aneuploidy/single gene disorder/inborn metabolic disorders, or positive family history for chromosomal disorders (maternal/paternal) were referred from Department of Obstetrics and Gynecology to Department of Radiodiagnosis in Government Medical College, Kozhikode

to undergo amniocentesis to determine the fetal chromosomal structure, numerical aberrations, or translocations.

## RESULTS

In this study, 111 patients underwent amniocentesis. The most common indication for amniocentesis was absent/hypoplastic nasal bone (78%), followed by the combination of absent/hypoplastic nasal bone with increased nuchal fold thickness (7%), smaller percentages include fetal ventriculomegaly (2%), a combination of absent/hypoplastic nasal bone with short femur (1%), combination of short humerus and short femur (1%), increased nuchal fold thickness and choroid plexus cyst (2%), fetal pyelectasis and single umbilical artery (1%), and absent nasal bone and echogenic cardiac focus (1%).

The most common indication for amniocentesis was positive soft markers or in combination with positive soft markers and biochemical abnormalities (Table 1).

The most common positive soft marker in ultrasound for which amniocentesis was done is absent/hypoplastic nasal bone (Table 2) (Fig. 1).

The complication rate was very minimal; only one patient had a bloody tap. None of the patients had any other complications, such as miscarriages/bleeding per vagina. Mild pain in the abdomen was experienced by a few patients.

## DISCUSSION

The indications of amniocentesis were biochemical abnormalities (double marker positivity or quadruple marker test positivity), positive soft markers in second-trimester ultrasound (absent/hypoplastic nasal bone is the most common), positive family history, genetic abnormality in previous child, and one of these or in combination.

The soft markers for which amniocentesis is required include absent/hypoplastic nasal bone, increased nuchal fold thickness, fetal ventriculomegaly, short femur, short humerus, choroid plexus

**Table 1:** Indications of amniocentesis

<i>Indications of amniocentesis</i>	<i>Frequency</i>
Biochemical abnormalities	78
Positive soft markers in ultrasound	88
Positive family history	1
Genetic abnormality in the previous child	13

**Table 2:** Positive soft markers in second-trimester ultrasound

<i>Positive soft markers in second-trimester ultrasound</i>	<i>Frequency</i>	<i>Percentage</i>
Absent/hypoplastic nasal bone	69	78
Absent/hypoplastic nasal bone and increased nuchal fold thickness	6	7
Increased nuchal fold thickness	4	5
Fetal ventriculomegaly	2	2
Absent/hypoplastic nasal bone and short femur	1	1
Short femur and short humerus	1	1
Increased nuchal fold thickness and choroid plexus cyst	2	2
Fetal pyelectasis and single umbilical artery	1	1
Absent nasal bone and echogenic cardiac focus	1	1
Increased nuchal fold thickness, absent/hypoplastic nasal bone, and echogenic bowel	1	1
Echogenic cardiac focus	1	1
Total	88	

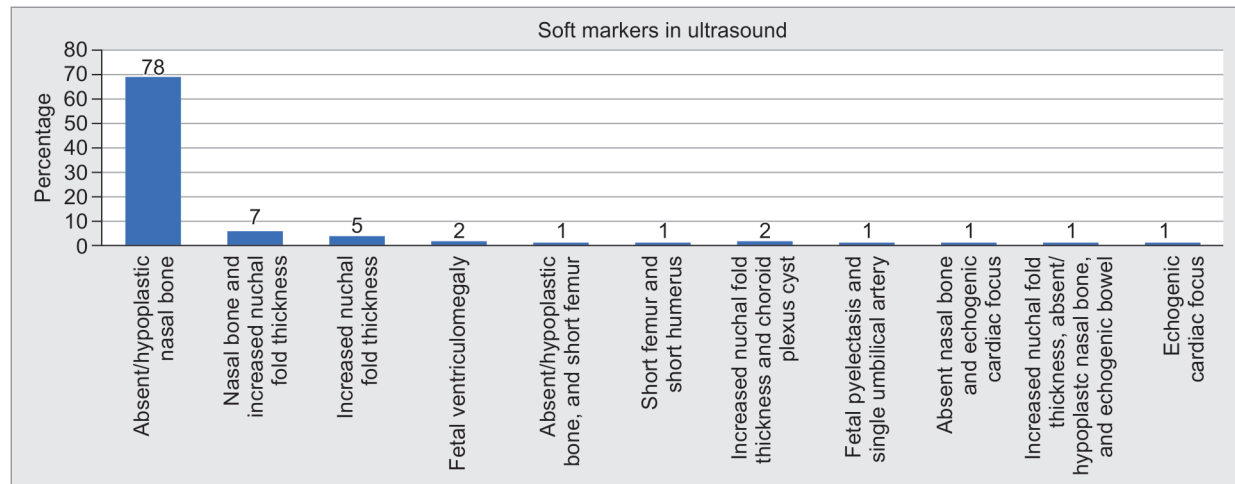


Fig. 1: Bar graph of positive soft markers in second-trimester ultrasound

cysts, echogenic cardiac focus, and echogenic bowel. The most common indication was absent/hypoplastic nasal bone (78%), followed by a combination of absent/hypoplastic nasal bone and increased nuchal fold thickness (7%).

In none of the patients, miscarriage was noticed following the procedure. Only mild pain and minimal leak per vagina were complained by a few patients. Hence, amniocentesis is a safe procedure.

## CONCLUSION

The most common indication for amniocentesis was absent/hypoplastic nasal bone, followed by a combination of absent/hypoplastic nasal bone with increased nuchal fold thickness. The complications were very minimal, which could be used to counsel the patients to undergo amniocentesis.

## Clinical Significance

Amniocentesis is a safe procedure with none to very minimal complications. Hence, it can be advised to patients with a positive screening test.

## ORCID

Anoop Venkatapura Bylaswamy <https://orcid.org/0009-0005-4823-5575>

Juvaina Puthiyakam <https://orcid.org/0000-0001-5832-2086>

## REFERENCES

- Sharma A, Kaul A. Late amniocentesis: better late than never? A single referral centre experience. *Arch Gynecol Obstet* 2023;308(2):463–470. DOI: 10.1007/s00404-022-06662-6
- American College of Obstetricians and Gynecologists. ACOG Practice Bulletin No. 88, December 2007. Invasive prenatal testing for aneuploidy. *Obstet Gynecol* 2007;110(6):1459–1467. DOI: 10.1097/01.AOG.0000291570.63450.44
- Practice bulletin no. 162: prenatal diagnostic testing for genetic disorders. *Obstet Gynecol* 2016;127(5):e108–e122. DOI: 10.1097/AOG.0000000000001405
- Mukherjee K, Chaudhury K. Amniocentesis is a safe and effective prenatal diagnostic tool: a clinical study in Eastern India. *Int J Reprod Contracept Obstet Gynecol* 2015;4(5):1330–1335. DOI: 10.18203/2320-1770.ijrcog20150705