J. Fetal Med. (September 2016) 3:131–135
DOI 10.1007/s40556-016-0084-0

REVIEW ARTICLE

Amniocentesis

Nandita Dimri[1] · **Ashok Baijal**[1]

Received: 16 December 2015 / Accepted: 4 April 2016 / Published online: 27 May 2016

Abstract Amniocentesis is the most common invasive prenatal diagnostic procedure worldwide. It is a technique of withdrawing amniotic fluid from the uterine cavity using a needle, via a transabdominal approach, under continuous ultrasound guidance, in order to obtain a sample of fetal exfoliated cells, transudates, urine, or secretions. The amniotic fluid contains amniocytes and fetal epithelial cells. Amniotic fluid can be tested directly or grown in culture for various chromosomal, bio-chemical, molecular, and microbial studies. Amniocentesis for genetic testing is generally performed between the gestational age of 16 and 20 weeks. Diagnostic amniocentesis is commonly used for prenatal diagnosis of chromosomal abnormalities, single gene disorders, fetal infection, and intra-amniotic inflammation. Common indications cited are advanced maternal age, positive maternal screening results for aneuploidy, structural abnormality on ultrasound, and inconclusive or positive noninvasive prenatal testing. Pre-procedure counseling and screening ultrasound should always be done before subjecting a woman to amniocentesis. Complete procedure is performed under ultrasound guidance with continuous visualization of the needle under proper aseptic conditions. Post-procedure follow-up includes documentation of fetal viability immediately after the procedure by ultrasound studies, anti-D immunoglobulin (300 g) to be administered to RhD negative women, and house rest for 24 h.

Keywords Amniocentesis · Ultrasound · Chromosomal abnormality

✉ Nandita Dimri
dimnan@yahoo.com

[1] Department of Fetal Medicine, Sir Ganga Ram Hospital, Rajinder Nagar, New Delhi 110060, India

Introduction

Amniocentesis is the most common invasive prenatal diagnostic procedure worldwide. Amniotic fluid tapping had been practised for over a 100 years, although few cases have been recorded. Amniocentesis in late trimester was reported by Prochownick et al. in 1877, and Schatz in the 1890s. Nadler and Gerbie published "Role of amniocentesis in the intra-uterine diagnosis of genetic defects" in 1970 [1]. This was probably the first initiative in genetic amniocentesis and diagnosis. Amniocentesis is a technique of withdrawing amniotic fluid from the uterine cavity using a needle, via a transabdominal approach under continuous ultrasound guidance, in order to obtain a sample of fetal exfoliated cells, transudates, urine, or secretions. The amniotic fluid contains amniocytes and fetal epithelial cells. Amniotic fluid can be tested directly or grown in culture for various chromosomal, biochemical, molecular, and microbial studies. Amniocentesis for genetic testing is generally performed between the gestational age of 16 and 20 weeks.

Indications

Diagnostic amniocentesis is commonly used for prenatal diagnosis of chromosomal abnormalities, single gene disorders, fetal infection, and intra-amniotic inflammation. Common indications cited are advanced maternal age, positive maternal screening results for aneuploidy, structural abnormality on ultrasound, and inconclusive or positive noninvasive prenatal testing.

Performing amniocentesis for evaluation of neural tube defects, determination of fetal lung indices, or determination of degree of fetal anemia are almost obsolete in this era of high resolution real time ultrasonography and better

noninvasive techniques. Therapeutic use of amniocentesis is seen for polyhydramnios to relieve maternal distress, infusion of fluids (amnio infusion) in pregnancies complicated by oligohydramnios to improve visualization before placement of shunts in obstructive uropathy, or intrapartum to prevent fetal heart rate decelerations.

No absolute contraindication to amniocentesis has been put forward till now. Relative contraindications include maternal retroviral or hepatitis B infection and early gestation as these have been associated previously with increased chances of fetal loss. History of vaginal spotting and stained amniotic fluid are the other relative risk factors associated with increased fetal loss after amniocentesis [2].

Pre-procedure Counseling

Prenatal invasive procedure carries a lot of apprehension and anxiety in a pregnant woman. Pre-procedure counseling forms an important aspect before subjecting a woman to amniocentesis. Counseling includes obtaining informed consent after explaining the proper indications, technique, and risks involved in performing the procedure, and post-test counseling.

The decision to undergo an invasive procedure may be difficult for a woman and an occasion for worry. Knowledge of women's concerns and worries, understanding of the reactions, and the process of decision-making about prenatal diagnosis are of importance to the caregivers providing counseling. Major concerns of women before amniocentesis are abnormal test result, possible termination, injury to the fetus, risk of miscarriage, and procedure-related discomfort or pain [3, 4].

Pre-amniocentesis counseling should emphasize the fact that the actual pain and anxiety experienced during the procedure is significantly lower than expected. In fact, on a scale of 0–10, the mean level of pain is only 2.1, with a slightly higher mean level of anxiety [4].

Procedure

Maternal history, clinical data documentation, and relevant investigations are essential prerequisites before the test.

Screening Ultrasound Ultrasound evaluation before the procedure is based on a sweep of the uterine cavity in serial transverse views of the maternal abdomen for:

- Placental location
- Position of fetuses and determination of chorionicity in multiple gestation
- Fetal position and fetal movements
- Estimation of gestational age and viability
- Estimation of liquor amnii, (maximum vertical pocket–MVP)
- Determination of cervical length
- Absence of chorioamniotic separation

Position Supine

Technique

The following points should be kept in mind

1. Probe should be kept perpendicular to maternal abdominal surface.
2. Correct length of the needle should be chosen according to maternal habitus. Preferred length of the needle ranges from 9 to 15 cm with 20–22G thickness. (Fig. 1).
3. Maximum vertical pool of liquor should be taken in transverse view of abdomen avoiding oblique views and lateral entry.
4. Proper image magnification with complete view of maternal skin up to amniotic fluid pool should be recorded. Transplacental puncture should only be attempted in cases of risk of failure of procedure, extreme lateral entry, and nonconducive fetal position. Transplacental approach is contraindicated in alloimmunization, HIV, HBsAg, and HCV infections in mother.
5. In cases of multiple gestations, two separate punctures should be attempted with two separate needles to withdraw amniotic fluid and proper mapping of fetuses is documented to avoid errors in identification of samples.
6. The abdomen is prepared with antiseptic solution [povidone-iodine or chlorhexidine] (Fig. 2). The transducer is draped in a sterile manner to facilitate real-time imaging of needle trajectory.

Fig. 1 22G/9 cm long needle for amniocentesis

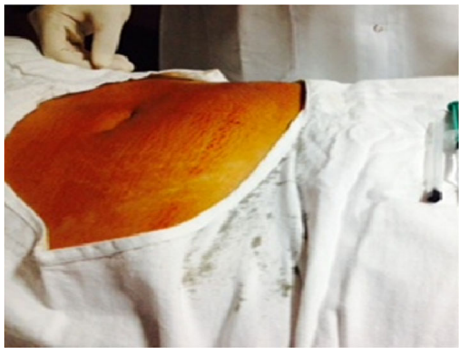

Fig. 2 Sterile preparation of maternal abdomen

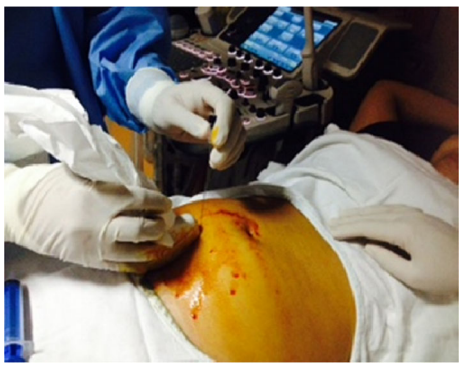

Fig. 3 Needle being introduced at 45° to maternal midsagittal plane

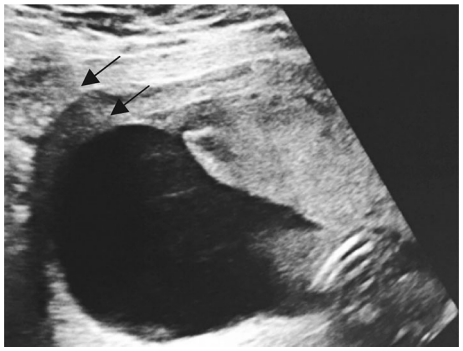

Fig. 4 Complete visualization of needle under ultrasound guidance

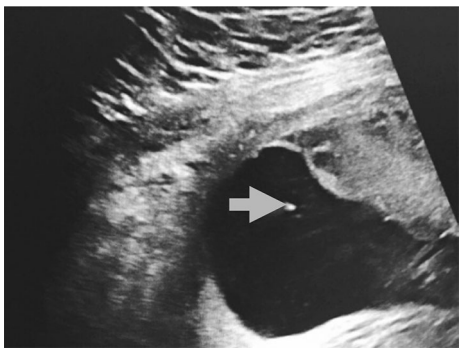

Fig. 5 Correct placement of needle in amniotic cavity

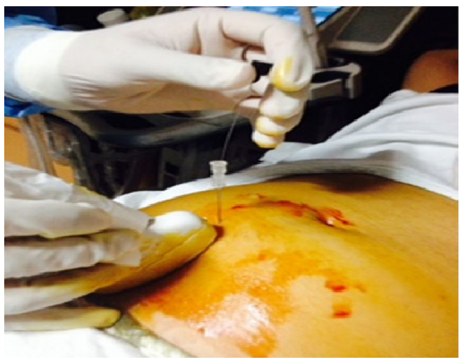

Fig. 6 Stylet removal after correct placement for aspiration of amniotic fluid

Ultrasound-guided amniocentesis is performed using the 'freehand' technique, in which the specialist performing the procedure freely manipulates the ultrasound transducer with one hand and needle with the other. Complete procedure is performed under ultrasound guidance with continuous visualization of the needle. Needle insertion has four stages: abdominal skin puncture, uterine puncture, entry into amniotic cavity, and advancement of the needle.

Introduction of the needle through abdomen is at 45° to the maternal midsagittal plane and 90° contralateral to the probe (Fig. 3). Care should be taken to visualize the entire trajectory to avoid injury to adjacent bowel loops (Fig. 4) [5].

Uterine puncture may evoke a startle due to pain or focal myometrial contraction for which, orientation of the needle needs to be readjusted.

Amniotic cavity insertion needs a "jab" to avoid tenting of membranes or failure of aspiration of amniotic fluid. Thrusting or twisting of needle in case of tenting of membranes may be tried for successful aspiration.

Needle advancement into amniotic cavity under complete visualization is advised. After correct placement in the cavity (Fig. 5), stylet is withdrawn (Fig. 6) and 20 mL amniotic fluid is aspirated after discarding first 2 mL (Fig. 7). The amniotic fluid should be collected in two or three sterile tubes (Fig. 8) if initially there is blood straining of the amniotic fluid, the first 2–3 mm of the fluid should be discarded.

Needle can be kept inside the amniotic cavity for around 1 min, after which it should be extracted if the attempt remains unsuccessful. A new puncture site should be chosen and the needle changed to avoid contamination. Maximum of two attempts are recommended in case of failure to obtain a satisfactory sample and a repeat procedure is suggested a week later.

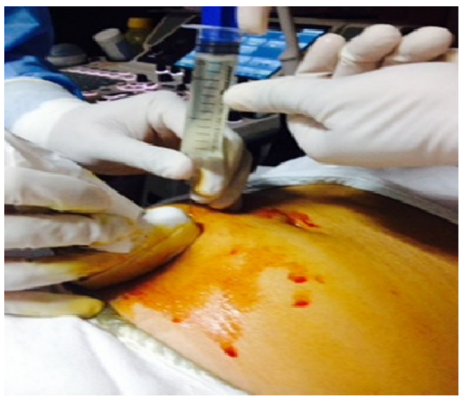

Fig. 7 Aspiration of amniotic fluid

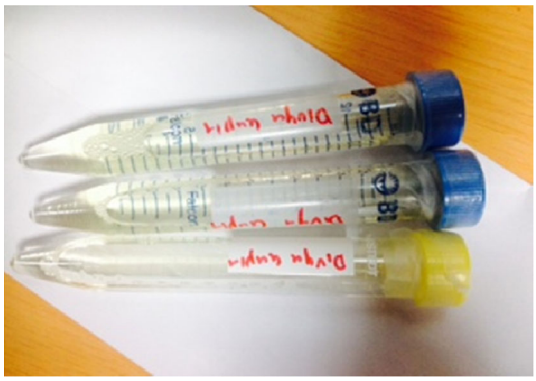

Fig. 8 Collection and transport of amniotic fluid in sterile tubes

Post-procedure Follow-up

1. Document fetal viability immediately after the procedure by ultrasound studies.
2. Anti-D immunoglobulin (300 μg) should be administered to RhD negative women.
3. House rest for 24 h. Normal activities for personal hygiene and in-house activities are allowed. In spite of great social pressure to increase the recommended house-rest period, there is no scientific evidence to justify this.
4. Alarming symptoms that require the woman to contact the emergency room include bleeding or amniotic fluid leakage, intense abdominal pain, and fever equal to or above 38 °C.
5. Ultrasonography should be performed 1 week after the procedure to confirm fetal viability and evaluate the puncture area.

Usually, results of QF-PCR/FISH are available within 48–72 h, PCR for fetal infection in one week, array-CGH in two weeks, and karyotyping in three weeks. This may vary from center to center.

The rate of miscarriage after amniocentesis is reported as approximately 0.5 %. However, these data were based on earlier studies with less modern techniques. Recent studies (randomized controlled trials and cohort studies with or without a control group; the control group may have no procedure or an alternative procedure) show a range of post-midtrimester amniocentesis loss of 0.75 to 3.3 % (9 studies; mean 1.41 %, median 1.1 %) [6, 7]. The risk of miscarriage prior to 24 weeks who underwent amniocentesis and CVS was 0.8 and 2.1 %, respectively. The background rates for miscarriage in women from the control group that did not undergo any procedures were 0.6 % for amniocentesis and 1.6 % for CVS [8]. According to a recent study in China, procedure-related fetal loss is reported lower than 0.59 % and increased association is seen with advanced maternal age, bad obstetric history, and abnormal vaginal bleeding in present pregnancy, fibromyomas, and stained amniotic fluid [9]. Transplacental passage of the needle initially, seemed to increase the rate of pregnancy loss, but subsequent work has shown no increase in loss rates with this approach. Leakage of amniotic fluid may occur after a genetic amniocentesis, although favorable pregnancy outcomes are seen in more than 90 % of these women with expectant management. Amniotic fluid culture fails in approximately 0.1 % of cases. Outcome studies of infants after amniocentesis show no long-term sequelae.

Compliance with Ethical Standards

Conflict of interest None.

References

1. Nadler HL, Gerbie AB. Role of amniocentesis in the intrauterine detection of genetic disorders. N Engl J Med. 1970;282:596–9.
2. Theodora M, Antsaklis A, Antsaklis P, Blanas K, Daskalakis G, Sindos M, et al. Fetal loss following second trimester amniocentesis. Who is at greater risk? How to counsel pregnant women. J Matern Fetal Neonatal Med. 2016;29(4):590–5.
3. Cederholm M, Axelsson O, Sjödén PO. Women's knowledge, concerns and psychological reactions before undergoing an invasive procedure for prenatal karyotyping. Ultrasound Obstet Gynecol. 1999;14:267–72.
4. Ferber A, Onyeije CI, Zelop CM, O' Reilly-Green C, Zelop CM, Divon MY. Maternal pain and anxiety in genetic amniocentesis: expectation versus reality. Ultrasound Obstet Gynecol. 2002;19:13–7.
5. Cruz-Lemini M, Parra-Saavedra M, Borobio V, Bennasar M, Gonce A, Martinez JM, et al. How to perform an amniocentesis. Ultrasound Obstet Gynecol. 2014;44:727–31.
6. Towner D, Currier RJ, Lorey FW, Cunningham GC, Greve LC. Miscarriage risk from amniocentesis performed for abnormal maternal serum screening. Am J Obstet Gynecol. 2007;196(608):e1–5.
7. Wilson RD, Langlois S, Johnson JA. Society of obstetricians and gynaecologists of Canada. Mid-trimester amniocentesis fetal loss rate. J Obstet Gynaecol Can. 2007;29(7):586–95.

8. Akolekar R, Beta J, Picciarelli G, Ogilvie C, D'Antonio F. Re: risk of miscarriage following amniocentesis and chorionic villus sampling. Ultrasound Obstet Gynecol. 2015;45(6):755–7.

9. Huang L, Jiang T, Liu C. Fetal loss after amniocentesis: analysis of a single center 7957 cases in China. Clinc Exp Obstet Gynecol. 2016;29(4):590–5.