Frimley Health NHS Foundation Trust

Prenatal Diagnosis, CVS and Amniocentesis Guideline

Key Points

- Amniocentesis
- Chorionic villus Sampling
- Fetal abnormality
- Array CGH

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Abbreviations

Amnio	Amniocentesis
CGH	Comparative genomic hybridisation
CPM	Confined placental mosaicism
CVS	Chorionic villus sampling
DNA	Did not attend
GLH	Genomic laboratory hubs
LocSSIP	Local Safety Standards for Invasive Procedures
NIPT	Non invasive prenatal testing
PND	Pre-natal diagnosis
QF-PCR	Quantitative fluorescent-polymerase chain reaction
T21	Trisomy 21 (Down's syndrome)
T18	Trisomy 18 (Edwards Syndrome)
T13	Trisomy 13 (Patau's syndrome)

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1. Introduction

Prenatal diagnosis (PND) is a diagnostic procedure carried out on pregnant women to detect the presence of genetic or other abnormalities in the developing fetus.

Ultrasound scanning remains the cornerstone of prenatal diagnosis. Other procedures include chromosome and enzyme analysis of fetal cells obtained by amniocentesis or, at an earlier stage of pregnancy, by chorionic villus sampling (CVS).

2. Indications for prenatal diagnosis

Pregnant women are offered amniocentesis or chorionic villus sampling (CVS) for prenatal diagnosis for a variety of reasons including:

- A high chance combined or quadruple screening result.
- A higher chance NIPT result
- Fetal structural anomaly identified or suspected on the 12 week or 18-20+6-week anomaly scan.
- A known risk of inherited genetic disease

3. Chorionic Villus sampling (CVS)

CVS is a transabdominal or sometimes transcervical procedure performed under continuous ultrasound guidance. It is usually performed from 11 to 14 weeks of pregnancy. A sample of placental tissue is obtained for chromosomal or genetic analysis. If required, CVS can be performed between 14^{+0}- and 14+6-weeks' gestation. Individualised counselling of the merits of CVS versus amniocentesis should be provided for women considering CVS during this time period.

4. Amniocentesis

Amniocentesis is a procedure performed under continuous ultrasound guidance and can be performed from 15+0. A sample of amniotic fluid is obtained for chromosomal or genetic analysis. Careful counselling is required if Amniocentesis is supported at or close to a viable gestation. In very unusual circumstances a third trimester amniocentesis may be supported but care must be taken to consider chance of preterm delivery and maternal steroids should be considered prior to the procedure.

5. Twin pregnancies

Prenatal diagnosis (CVS or Amniocentesis) for multiple pregnancy is more complicated and all women considering PND will be referred to a tertiary centre, it will not be performed locally. Additional counselling is required to support decision making in the uncommon event of one baby presenting with a confirmed diagnosis and the other baby not. Selective termination of pregnancy for example would require specialist discussion and only be supported at a tertiary centre. Other technical considerations include there may be a need to insert the needle twice to get samples of placenta or fluid from each baby or with CVS there is a small chance of getting 2 samples from the same baby, which could give misleading results.

6. Miscarriage risk

- Women should be informed that miscarriage risk following amniocentesis or CVS, performed by a skilled operator is approximately 0.5%.
- Women with multiple pregnancies should be informed that miscarriage risk following amniocentesis or CVS performed by a skilled operator is approximately 1.0%.

7. Pre procedure counselling

- Frimley Park and Wexham Park have limited availability to offer CVS and Amniocentesis on site and availability is managed by the antenatal screening team. If there is no local availability the screening team are responsible for referring women to the tertiary referral centres. Frimley Park refer to the fetal medicine team at St Georges Hospital in Tooting and Wexham Park refer to the fetal medicine team at the John Radcliffe Hospital in Oxford.
- All women should be counselled, ideally face to face, by one of the specialist Midwives in the antenatal screening team for high chance Combined , Quadruple or NIPT screening results .If there is a known risk of inheriting a genetic condition then specialist written information must be available from the genetics team or women will need to be referred to the team to confirm if PND is available for the specific conditional and ensure the parents are fully informed of the reason for testing , the possible results , timing of results and choices the woman may need to consider. If a structural anomaly is suspected on scan the women should be referred to fetal medicine for a further scan and discussion to ensure PND is an appropriate consideration.
- Screening Midwife must clearly explain and document the reason for prenatal diagnosis, the expected results and limitations and risks associated with the procedure. The screening teams will use the national fetal anomaly screening programme information for parents to support discussions.
- It is not always possible to get a result from the first CVS or amniocentesis. Up to 6 in 100 women (6%) will be offered a second procedure.
- Women should be directed to the appropriate Fetal anomaly screening programme information.
  https://www.gov.uk/government/publications/downs-syndrome-edwards-syndrome-and-pataus-syndrome-options-after-a-higher-chance-screening-result/your-choices-after-a-higher-chance-screening-result
  https://www.gov.uk/government/publications/cvs-and-amniocentesis-diagnostic-tests-description-in-brief/nhs-fetal-anomaly-screening-programme-chorionic-villus-sampling-cvs-and-amniocentesis-information-for-parents

8. Scheduling of procedures

The screening teams schedule the CVS or Amnio on the fetal medicine scan lists. In some cases, decision to go ahead with the procedure is not made until the consultant has counselled the women and completed a scan. Following confirmation that the women wishes to proceed the decision to complete an invasive procedure also has many practical considerations such as placental position, uterine views, fulness of bladder etc. It is safe practise to reschedule and delay procedure if the optimum situation does not present.

9. Review of screening and booking bloods

- Infectious disease screening
  Must be completed prior to completing a procedure. If the woman has a positive infectious disease screening result this needs to be discussed with the fetal medicine consultant prior to scheduling a procedure. All women with a positive HIV result or a positive Hepatitis B result require targeted counselling and should understand the additional risk of passing on these viruses to their unborn baby. Viral load levels may support decision making and may need to be repeated prior to scheduling an appointment.

- Sickle and thalassaemia screening
  Must be completed prior to completing a procedure. If indicated biological father screening must be completed prior to scheduling PND in the unlikely event that a mother may present with a high chance from trisomy screening but also have a risk of inheriting a genetic condition.

- Blood group.
  If the woman is rhesus negative then Anti D is required, unless the fetal cell free DNA result is available, and the fetus is also predicted to be rhesus negative. Anti D can be ordered by the screening team on the day of the procedure. A group and save must be sent prior to collecting the anti D. If over 20 weeks gestation a Keilhauer must also be ordered to ensure the standard dose of anti D is sufficient.

NB The screening team should take the opportunity to complete the booking blood flow sheet prior to the scheduled appointment with fetal medicine.

10. Documentation. Procedure and consent

- It is the fetal medicine consultant's responsibility to complete the EPIC documentation.
- Fetal medicine consultant is required to place an EPIC order for the relevant procedure. Consultant must choose either ***Diagnostic amniocentesis*** or ***Chorionic villus sampling***
- The clinician selects the order from the intake tab and completes procedure consent form 2 and LocSSIP questions with the patient.

11. Preparation

- It is the responsibility of the screening team to have all stock available. This is stored in the scan room cupboards and includes dressing pack, gauze, non-latex sterile gloves and skin preparation.
- Needles: Specific Amniocentesis and CVS procedure needles are ordered by the screening team and stored in the procedure scan room cupboards.
- CVS transport medium is sent to the laboratory teams by the Viapath laboratory and collected by the screening teams and stored on the Frimley site in the ANC fridge and on the Wexham site in the fridge on Ward 21.

12. Testing of CVS or Amniocentesis Samples

All NHS providers are linked to a genomic hub to support a national testing network. Frimley Health genetic testing is supported by the Southeast Genomic Laboratory Hub (GLH) based at the Viapath genetics laboratory, Guy's and St Thomas' NHS Foundation Trust. All samples taken on the Frimley or Wexham sites are couriered directly to the Viapath laboratory. The lab will arrange the courier and the screening teams are responsible for coordinating with the laboratory team when a courier is required.

Request forms for all genetic testing including pre-natal diagnosis, parental chromosome or carrier bloods can be obtained from the screening team or directly from the Via path website.

Guy's and St Thomas' NHS Foundation Trust (2016) *Genetics specimen form.* Available at: http://www.viapath.co.uk/sites/default/files/upload/MF-G-RequestForm_2.pdf
Please note Oxford NHS Foundation Trust are mapped to the West Midlands regional genomic hub based at Birmingham's Women and Childrens NHS Foundation Trust. Women completing PND at Oxford following a tertiary referral will have their samples sent there and the Oxford team will ensure results are shared with the women and the local screening team.

13. Post procedure

- Women should be given a copy of the Screening in pregnancy: CVS and amniocentesis information for parents, the screening team and triage contact details should be clearly available.
- One in 200 women who have a CVS or amniocentesis will miscarry. We do not know why some women miscarry after these procedures. Most miscarriages happen within 3 days of the procedure, but they can happen up to 2 weeks afterwards.
- All women will be advised to contact triage (FPH) or MAC (WPH) regardless of gestation with any post procedure complications or concerns. It is important to note that some women may have completed PND following presenting with a very sick baby on scan therefore risk of miscarriage maybe higher and it is important women are cared for in a sensitive and dignified manner within an appropriate environment It is not appropriate for women to attend A&E with complications following a procedure.
- A serious infection following a procedure is rare, women should be asked to contact triage at Frimley Park or MAC at Wexham Park if they experience constant or severe abdominal pain, bleeding, signs of rupture of membranes, a high temperature, chills or shivering or contractions following a procedure regardless of gestation. Obstetric doctors should treat with a broad-spectrum antibiotic if symptoms of infection are present.

14. Possible results

- **PND after a higher chance result from combined screening, Quadruple screening or NIPT.**
  An initial QF-PCR result should be received within 3 days. In some rare cases the laboratory may advise if the sample is a possible confined placental mosaicism (CPM) as placental DNA may not be the same as the baby's DNA and that parents should wait for the full karyotype result which should be available within 2 weeks. In some cases, the laboratory in conjunction with the genetic team may recommend an amniocentesis test as this will analyse the fetal cells present in the amnio fluid and would eliminate the chance of a CPM finding.

In most cases the result will confirm one way or another if the baby does or does not have the condition the test was looking for. When indication for testing is due to a high chance combined, quadruple or NIPT screening test women need to understand that the test may detect a different condition i.e Turners syndrome or a tripody pregnancy. The screening teams are supported by specialist genetic services based at either St Georges NHS Foundation Trust or Oxford NHS Foundation Trust and can refer women who may require more specialists advise following the receipt of an unexpected result or if following a confirmed diagnosis and more targeted counselling is needed.

- **Pre-natal diagnosis for risk of inheriting a specific genetic condition including high risk couples identified through the sickle cell and thalassaemia screening programme.**
  All testing will be arranged following direction from the specialist genetic counselling teams. Not all GLH's test for all conditions but in most cases the Via path (Synnovis) laboratory will culture the sample and send required material to the specific laboratory for specialist testing. In most cases a QF-PCR result for the 3 main trisomy's (T21, T18 7 T13) will be reported by Synnovis. This should be taken into consideration when counselling and taking consent as women must be aware what is and is not being tested for and what and when to expect results. Timing of specialist genetic test results may take between 2- 3 weeks but QF-PCR results for T21, T18 & T13 should still be received within 3 days. In this situation consideration can be taken for women to decline combined or quadruple screening as a dating scan may be supported earlier than 12 weeks to allow for scheduling of the CVS procedure.

- **Fetal structural anomaly identified or suspected on the 12 week or 18-20+6-week anomaly scan.**
  Testing will be supported following in person consultation with the fetal medicine consultant. In some situations, a referral to the tertiary centres or the genetic services is required for parents to have more targeted counselling and then the women can return for the procedure to be performed locally. Advise from the tertiary and / or genetics team will inform the screening teams what additional information should accompany the sample such as what specific test is required and if maternal or paternal blood samples should also be sent. Specialist testing and giving of results is often coordinated by the genetics team.
  Women may attend the tertiary centre for specialist care and complete the pre-natal diagnosis there. In this situation the tertiary centre may send samples to an alternative GLH and is responsible for informing the women of the results and liaising with the local screening team to support ongoing care and decision making.

- **When PND results show the baby does not have the condition tested for**
  The woman should be informed of the result and continue with antenatal care. The outcome of the pregnancy will be reported by the screening team to National Congenital And Rare Diseases Registration Service (NCARDRS).

- **When PND results show the baby has a condition tested for**
  A discussion of the results with the screening Midwives and /or fetal medicine consultant will be arranged. A specific plan for results giving can be discussed with the women following the procedure and her wishes should be documented on her EPIC record. Women should be supported to make personal informed choices about their ongoing care and pregnancy options.

NB Fetal sexing will not be reported unless clinically relevant such as in cases of X linked genetic conditions. If, following confirmation of an abnormal result, the parents decide to discontinue the pregnancy and are requesting to know the fetal sex the laboratory can be contacted and in most cases are able to provide this information.

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Full version control record

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Version Control Sheet

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1.0	November 23	K. Franks	Final	First cross site version

Related Documents

None.