Chorionic villus sampling

What is chorionic villus sampling (CVS)?[1]

Chorionic villus sampling remains the only diagnostic test available in the first trimester and allows for diagnostic analyses, including fluorescence in situ hybridisation (FISH), karyotype, microarray, molecular testing, and gene sequencing.

Chorionic villus sampling has been performed before 9 weeks in the past, though this has shown to increase the risk of limb deformities and, therefore, is no longer recommended.

Chorionic villus sampling may be performed via either transcervical or transabdominal approach. Via either approach, chorionic villi are collected for genetic evaluation under ultrasound guidance without entering the amniotic sac.

Chorionic villus sampling allows for earlier prenatal diagnosis, subsequently decreasing time of uncertainty and allowing for earlier (and therefore, safer) pregnancy termination if desired.

A disadvantage of chorionic villus sampling is that approximately 1% to 2% of chorionic villus sampling results may reflect confined placental mosaicism rather than true fetal chromosomal abnormalities.

Pregnancy loss attributed to chorionic villus sampling is approximately 1 in 455.

Chorionic villus sampling has decreased in frequency with the recent increased uptake of cell-free DNA screening.

In the UK, the Royal College of Obstetricians and Gynaecologists recommends:[2]

- Chorionic villus sampling should not be performed prior to 10+0 weeks' gestation. Where possible, to reduce the risk of technical challenges, CVS should be performed from 11+0 weeks' gestation onwards.
- Chorionic villus sampling, carried out to obtain placental villi for analysis, is usually performed between 11+0 and 13+6 weeks of gestation.
- If required, chorionic villus sampling can be performed between 14+0 and 14+6 weeks' gestation. Individualised counselling of the merits of chorionic villus sampling versus amniocentesis should be provided for women considering chorionic villus sampling during this time period.
- Women with multiple pregnancies should be informed that the additional risk of miscarriage for twin pregnancy following chorionic villus sampling or amniocentesis performed by an appropriately trained operator is around 1%.

Genetic counselling

Genetic counselling should ideally be offered prior to any pregnancy, when there is a family history of a condition which might be diagnosed either by amniocentesis or chorionic villus sampling.

It is clearly important to avoid unnecessary invasive testing in pregnancy where possible.

Diagnostic testing should be provided within the context of informed consent and autonomy, both about the conditions being tested and about the implications for the continuation of the pregnancy.[3] Pre- and post-test genetic counselling are both indicated.

Indications

It is important to remember that women choose whether or not to undergo chorionic villus sampling. The aim of the procedure is to provide chorionic material for prenatal diagnosis of chromosomal or single-gene abnormalities.

CVS is most likely to be used in the following situations:

- Positive antenatal screening test - eg, the combined test for trisomy.
- Past history of a genetic or chromosomal abnormality.
- Familial chromosomal rearrangement.
- Biochemical or molecular diagnosis of a familial genetic disorder.

Chorionic villus sampling cannot be used to screen for structural problems such as neural tube defects, which have no known metabolic or molecular basis (unlike amniocentesis). Suspected neural tube defects should be investigated by biochemical and ultrasound markers.

See also separate Amniocentesis article.

Contra-indications

To chorionic villus sampling:

- Active vaginal bleeding.
- Infection.

Transcervical route of sampling is also contra-indicated by:

- Cervical polyps.
- Fibroids.
- Fundal placenta.
- Retroverted uterus with posterior placement of placenta.

Procedure

- Chorionic villus sampling is usually performed between 11 and 13 weeks (11^{+0} and 13^{+6}).[2]
- Informed written consent should be obtained.

- Rhesus immunoprophylaxis should be given where appropriate (fetomaternal transfusion is a risk in amniocentesis and chorionic villus sampling).
- The placental sample is obtained either by ultrasound-guided transabdominal needle, or ultrasound-guided transcervical cannula aspiration or biopsy forceps
- Results are usually obtained within 7-14 days, although newer tests can reduce reporting time to 24-48 hours.[4]
- Use of biopsy forceps rather than cannula aspiration for the transcervical route may be more effective and less painful for the woman but there is no difference in the risk of miscarriage.[5]
- Chorionic villus sampling allows diagnosis earlier in the pregnancy than with amniocentesis (which may only be safely conducted in the second trimester) and an earlier opportunity to consider termination of pregnancy in the event of fetal abnormality.

Risks and complications of chorionic villus sampling

A Cochrane review concluded that second trimester amniocentesis increases the risk of pregnancy loss, and early amniocentesis is not as safe as second trimester amniocentesis (increased pregnancy loss and talipes). Transcervical chorionic villus sampling compared with second trimester amniocentesis may be associated with a higher risk of pregnancy loss.[6]

- Sampling failure may occur due to laboratory failure, mosaicism, ambiguous results, insufficient sample or maternal cell contamination.[2]

- Miscarriage risk:[7] [8]
    - There is evidence to suggest the risk is reducing, probably due to improvements in technique.[9]
    - In a low-risk population, background pregnancy loss is about 2%. Overall risk of miscarriage following chorionic villus sampling is about 3% but CVS is often performed in the presence of conditions associated with a higher rate of pregnancy loss.[2]
    - A systematic review and meta-analysis, which included only studies published since 2000 therefore reflecting current practice, suggests the procedure-related risks are much lower at 0.2%.[7]
- Transcervical chorionic villus sampling is more technically demanding than the transabdominal route and more likely to lead to sample failures and multiple insertions, and to cause vaginal bleeding.
- Very experienced surgeons (more than 100 procedures per year) may have higher success rates and lower procedure-related miscarriages.[2]
- Potential complications of chorionic villus sampling also include amniotic fluid leakage, vaginal (higher for transcervical than transabdominal route) and sepsis (rare).
- Chorionic villus sampling at 8-9 weeks has, in case reports, has been linked with an increased incidence of fetal limb deficiencies (oromandibular limb hypoplasia and isolated limb disruption). The association between these abnormalities and early chorionic villus sampling is uncertain, as subsequent analyses haven't been able to confirm a link. Chorionic villus sampling is no longer performed prior to 10 weeks, in part due to the greater technical difficulties involved.[2]

One systematic review found that the procedure-related risks of miscarriage following amniocentesis or chorionic villus sampling are lower than currently quoted to women. The risks appeared to be negligible when these interventions were compared to control groups of the same risk profile.[10]

Dr Mary Lowth is an author or the original author of this leaflet.

Further reading

- Ogilvie C, Akolekar R; Pregnancy Loss Following Amniocentesis or CVS Sampling-Time for a Reassessment of Risk. J Clin Med. 2014 Jul 8;3(3):741-6. doi: 10.3390/jcm3030741.
- Jones TM, Montero FJ; Chorionic Villus Sampling. StatPearls, December 2022.

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