

The [ESHG/ASHG joint policy statement on cfDNA](#) provides an excellent review of the ethical issues inherent in this new technology. Here are the main points from this publication:

- cfDNA offers improved accuracy when testing for trisomy 21, trisomy 18, and trisomy 13 as compared to compared other screening tests. However, cfDNA is still a SCREENING test. False positives occur for a variety of reasons, and women should be advised to have a positive result confirmed through diagnostic testing (they recommend amniocentesis specifically) if they are considering a possible termination of pregnancy.
- Although this test may be a more accurate screen for some conditions in pregnancy, the importance of patient education, counseling, and informed consent is imperative. ESHG/ASHG guidelines identify a goal of prenatal screening programs to provide women and couples with the opportunity for informed reproductive choices, and counseling and education to allow for informed choices about cfDNA is an essential component of that.
- The ESHG/ASHG recognizes that it may be impossible to avoid unexpected, incidental findings through cfDNA. Pre-test counseling should involve discussion of this possibility, and the wishes of the patient/couple should be respected regarding whether to receive or not receive information regarding incidental findings.
- The ESHG/ASHG recommends AGAINST using cfDNA to screen for sex chromosomal abnormalities and microdeletions given the associated ethical concerns, counseling challenges, and potential for the expansion into rare conditions with a higher likelihood of false positive to reverse the reduction in invasive testing achieved with implementation of cfDNA for aneuploidy.
- Currently, the ESHG and ASHG do not recommend that cfDNA be used for any conditions outside of what may be considered 'serious congenital and childhood onset disorders'.
- In countries where cfDNA is offered as a public health program, governments and public health authorities should adopt an active role to ensure the responsible use of this technology that ensures not only laboratory quality control, but also to include oversight of the quality of counseling and education provided to patients, as well as equity of access.
- Different scenarios for cfDNA-based screening for common autosomal aneuploidies are possible, including cfDNA as an alternative first-tier option. All of the possible benefits, risks, and limitations of each pathway should be carefully evaluated when developing a screening program.
- More tools to enable informed choices regarding prenatal screening and measures to evaluate these tools are needed.



American Society of Human Genetics (ASHG) and European Society of Human Genetics (ESHG) Summary

- A proactive professional and societal debate about what the future scope of prenatal screening for fetal abnormalities is necessary.