

No US commercial laboratories fully meet guidance for noninvasive prenatal screening

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An analysis of the reports and materials provided by commercial laboratories offering noninvasive prenatal screening (NIPS) for genetic disorders finds that none of them fully meet the recommendations published by the American College of Medical Genetics and Genomics (ACMG). The report from a team of specialists in medical genetics is being published in the journal *Genetics in Medicine*.

"It's been more than two years since the ACMG published its recommendations about NIPS, and we could not find a single commercial lab in the U.S. that adhered to all of the recommendations," says Brian Skotko, MD, MPP, of the Division of Medical Genetics at MassGeneral Hospital for Children, corresponding author of the paper. The ACMG position statement includes guidance to laboratories on the test reports used by clinicians and expectant parents.

"Incomplete or inaccurate reporting can lead to confusion and improper counseling," Skotko says. "The ACMG was clear that laboratories should not offer screening when some of their recommendations were not met, so I think our findings will be concerning to clinicians and expectant parents trying to identify quality NIPS reporting in today's marketplace."

NIPS testing analyzes a blood sample from an expectant mother for DNA fragments derived from the placenta, which can reveal genetic or chromosomal conditions in a fetus. The most commonly screened-for disorders include Down syndrome, caused by an extra copy of chromosome 21; other "trisomies" involving chromosomes 13 or 18; and extra or missing copies of the X or Y sex chromosomes. NIPS has the highest detection rate and lowest false positive rate of any screening test used for these conditions. However, NIPS is a screening test, and false positive and negative results are known to occur at rates that can vary depending on the

specific condition being screened.

Among its recommendations, the ACMG specified five statistics that should be reported for each screened-for condition:

- detection rate—the percentage of fetuses with a condition that will accurately be diagnosed by the test,
- specificity—the percentage of fetuses without a condition that will receive a negative result,
- [positive predictive value](#) (PPV) - the percentage of fetuses with positive test results that will actually have the condition,
- negative predictive value (NPV) - the percentage of fetuses for which a negative test result will be accurate,
- fetal fraction—the percentage of cell-free DNA in the mother's bloodstream that originated from the placenta.

"Parents deserve to know how likely their positive screening result is to be a true positive," Skotko explains. "Parents' decision to have the NIPS test—and how they respond after a positive result—may hinge on their understanding of PPV and NPV."

The ACMG recommendations also limited the number of conditions for which screening should be offered, specified that test results be provided in ways that help parents and providers understand the findings and make decisions, and called upon labs to direct parents and providers to resources providing additional education and support when test results are positive.

"By electing NIPS, expectant couples can receive an early indication of the chance their fetus might have a condition such as Down syndrome," says Skotko, who is director of the Massachusetts General Hospital (MGH) [Down Syndrome Program](#). "For highly suggestive results, they might elect to

pursue more definitive diagnostic testing, such as amniocentesis. When a diagnosis becomes definitive, couples have the option of terminating the pregnancy, making arrangements for adoption, or beginning to prepare for raising their child. Many couples choose to learn more through reading and meeting other families during the prenatal period so they feel well prepared for the birth."

The team's consensus-based analysis of test results and patient education materials provided by 10 commercial laboratories offering NIPS found considerable inconsistencies and inadequacies in reporting the full range of ACMG-reported statistics. Overall, while all of them met some of the ACMG recommendations, none met them all.

"Our study can't tell us why labs are choosing not to meet all the ACMG recommendations," says Skotko, the Emma Campbell Endowed Chair on Down Syndrome at MGH and an associate professor of Pediatrics at Harvard Medical School. "Laboratories have been very quick to capitalize on the ACMG position that all pregnant women be informed about the availability of NIPS testing, but they have been selective about the other recommendations. It's true that some recommendations may need updating—such as the fact that reporting a PPV for a negative test result doesn't make sense and that PPV might not be easily calculated for some chromosomal [conditions](#)—but we hope our findings will soon become outdated as companies begin to adhere to more of the ACMG recommendations."

He adds, "We hope our paper encourages laboratories to revise their reports to include patient resources and [test](#) metric data that can help physicians and obstetric care providers help their patients make informed decisions based on [accurate information](#)."

As a resource for providers and parents, Skotko and his collaborators in the Prenatal Research Information Consortium have published and will regularly update a chart showing individual laboratories' compliance with the ACMG recommendations, freely accessible here: <https://prenatalinformation.org/table>.

Skotko adds, "The ACMG recommendations state that labs should support the educational and access needs of parents and providers, and while half the labs made good faith efforts to meet our criteria, around half did not recommend any specific resources. Expectant families deserve to make informed choices, but they will struggle to do so without access to quality resources and information such as those recommended by ACMG."

More information: *Genetics in Medicine* (2019). [DOI: 10.1038/s41436-019-0485-2](https://doi.org/10.1038/s41436-019-0485-2)

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