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American College of Medical Genetics and Genomics
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ACMG Responds to *The New York Times* article regarding Noninvasive Prenatal Screening, "Tests Predicting Rare Disorders in Fetuses are Usually Wrong"

January 2022

The American College of Medical Genetics and Genomics (ACMG) took swift action in response to *The New York Times (NYT)* investigative article about noninvasive prenatal screening published on January 2, 2022, "Tests Predicting Rare Disorders in Fetuses are Usually Wrong." The ACMG is concerned that the article may be misleading to readers as it fails to clearly distinguish between screening and diagnosis, omits information on the accuracy of professionally recommended aneuploidy screening versus screening including microdeletions, and overlooks the problems caused by the lack of appropriate pre-test genetic counseling prior to ordering of the test. ACMG sent a Letter to the Editor authored by President-elect Dr. Susan Klugman and approved by the Board within hours of the *NYT* article being published.

The ACMG has been a longstanding advocate for the proper use of noninvasive prenatal screening and the importance of both pre- and post-test genetic counseling. ACMG has stressed the importance of medical professionals who are knowledgeable about prenatal genetics being involved in the counseling of patients about such screening tests, the ordering and interpretation of such tests, and the need for follow-up diagnostic testing. ACMG has also made clear recommendations about the type of conditions these screening tests should and should not include and has stressed that NIPS is a *screening* rather than a diagnostic test and recommends the use of the term noninvasive prenatal *screening.*

Detailed recommendations can be found in ACMG's 2016 statement: "Noninvasive prenatal screening for fetal aneuploidy, 2016 update: a position statement of the American College of Medical Genetics and Genomics" at https://www.gimjournal.org/article/S1098-3600(21)04448-8/fulltext.

Following is ACMG's Letter to the Editor (please note that *The New York Times* has a word limit of 200 words for Letters to the Editor, hence the ACMG statement is brief.)

Noninvasive Prenatal Tests Are for Screening, Not Diagnosis
In "Tests Predicting Rare Disorders in Fetuses Are Usually Wrong" (front page, Jan. 2), the authors describe the conundrum facing pregnant patients undergoing elective prenatal genetic screening tests. However, their use of the word "wrong" is misleading.

These prenatal tests, like routine mammography, are for screening, *not* diagnosis. A screening test is designed to reliably identify persons with a condition, recognizing that some individuals will have false positive results. Follow-up diagnostic tests are needed to determine true positive results.

In practice, these screening tests are not offered directly to consumers, but through healthcare professionals who have knowledge of prenatal genetics in keeping with national organizations' guidelines, not commercial marketing materials. Pretest counseling is crucial when undergoing any medical test. Patients need to understand the testing or screening they choose, and what follow-up tests are indicated before taking any action related to their pregnancy.

Current guidelines including those from ACMG and ACOG recommend noninvasive prenatal screening for common disorders, but not for less common syndromes caused by microdeletions, or missing pieces of chromosomes.