Comparing Three Screening Strategies for Combining First- and Second-Trimester Down Syndrome Markers

To the Editor:

I read with interest the report by Palomaki et al1 on the hypothetical detection rates for integrated, sequential, and contingent prenatal screenings for Down syndrome. An integrated screen is a statistical merger of the results from a first-trimester screen (nuchal translucency, pregnancy-associated plasma protein-A, human chorionic gonadotropin [hCG]) and a second-trimester quadruple screen (alpha-fetoprotein, unconjugated estriol, hCG, inhibin-A). In this integrated approach, the results of the first-trimester screen are not disclosed to women; only a composite value is given after the second-trimester screen has been performed. With a sequential screen, by contrast, women receive the results of the first-trimester screen at the time when it is given and are offered immediate diagnostic testing if they fall above “an initial risk cutoff level.” Contingent screening differs from sequential screening by also having a “low-risk cutoff level.” With both sequential and contingent screens, women are informed that their first-trimester screens are either “positive” or “negative,” and the authors conclude that “it is possible to select risk cutoffs for both sequential and contingent strategies that minimize losses in efficiency while maintaining early detection and early completion” (p. 367).

Mothers have told us that this practice is unacceptable.2 They have asked obstetricians to report all prenatal screenings as risk assessments and not as “positive” or “negative” results. Instead of the medical community assigning value to arbitrary cutoff levels, mothers have asked that they be given the numerical results (eg, 1:270) so that they can make their own personal decision on how to proceed. Obstetricians and genetic counselors can aid in explaining how to interpret probabilities, but mothers should be able to make the final call. Women have varying degrees of comfort, and what might be deemed a “risky” result to one person might fall within the satisfactory range for another.

The authors point out that “neither contingent nor sequential screenings have yet been formally tested in real world settings” (p. 373). If the medical community listens to mothers, the screens never should.

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REFERENCES

Editors Note:
The authors declined to respond.