Trisomy 21 in prenatal screening: How to talk with parents

FEB 7, 2019

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While cell-free fetal DNA screening is useful in identifying potential chromosomal abnormalities, results from it can easily be misunderstood by parents as diagnoses. In the case of an abnormal Down syndrome result, parents might also make erroneous assumptions about their child’s prognosis and its potential impact on family members.

Overcoming misinformation and emotional high stakes and can be a sizable task for physicians and their care teams. The following are offered as tips for helping expecting parents understand prenatal screening results and make informed decisions about pregnancy outcomes that align with their goals and values.

The AMA Code of Medical Ethics provides guidance on informed consent, such as opinion 4.1.2, “Genetic Testing for Reproductive Decision-Making,” which includes steps to take to when providing genetic testing to patients who request it, as well as when offering screening and diagnosis to individuals and couples considering having a child.

The following advice is summarized from an article published in the AMA Journal of Ethics® (@JournalofEthics) by pediatricians Eva Schwartz, MD, and Kishore Vellody, MD. This advice responds to a hypothetical case involving a pregnant woman and her partner who have decided to have an abortion after receiving prenatal screening results indicating their fetus has a high probability of having Down syndrome.

Start the conversation right away. Evidence cited in the commentary clarifies that mothers prefer a diagnosis to be given as soon as possible and in the presence of the father or partner. This gives the expectant parents time to seek support and make decisions.
Provision up-to-date information and resources. This includes Down syndrome’s causes and expectations for parenting people with Down syndrome today. Physicians might also provide a bibliography of resources about the condition.

In addition, mothers like to be offered opportunities to make social connections with parents of children with Down syndrome, who might be able to provide social, cultural, emotional and practical support and education beyond what a physician can offer during a clinical encounter.

Be sensitive, nonjudgmental and unbiased. In each conversation, the authors noted, physicians should set aside personal opinions and avoid value judgments and value-laden language, such as, “I have bad news to share,” or terms like “mongolism” and “retarded.”

Also, avoid attempting to convince a patient to make a particular decision, including by “speaking up on behalf” of people with Down syndrome. Likewise, counseling discussions should focus on a patient’s and family’s goals—not the physician’s values.

Tap specialists for help. In early conversations, select a health care professional knowledgeable about Down syndrome to communicate the test results, and note that this might not always be an obstetrician. In addition, offer follow-up appointments not only with an obstetrician but also with a specialist—such as a genetic counselor, Down syndrome specialist or cardiologist—who can help answer questions.

Even if a patient refuses offers of counseling, she retains the right to decide which pregnancy outcomes best align with her family’s goals and values.

The AMA Code of Medical Ethics is AMA policy. The AMA Journal of Ethics, however, is an editorially independent scholarly publication of the AMA.