

Prenatal diagnosis: Beyond decisions about termination

To the Editor:

An editorial in the January issue of *The Journal*¹ commented on 2 articles that studied the impact of prenatal diagnosis on the prevalence of Down syndrome² and cystic fibrosis.³ The author characterized prenatal diagnosis as a process designed to detect and prevent adverse outcomes.¹ But prenatal identification of selected conditions has value far beyond that limited use, and can inform a variety of decisions during or after pregnancy. Ultrasound identification of cardiac abnormalities in a fetus with Down syndrome, for example, may direct the choice of delivery to a hospital in which pediatric cardiac resources are readily available for a family not interested in termination. As the number of diagnoses that can be identified during pregnancy increases, uses for this information will continue to expand.

Counseling about a particular diagnosis may need to be updated as therapeutic advances affect outcomes for persons with that condition. Another article in the January issue of *The Journal*⁴ noted that prevalence of Down syndrome in the Netherlands was higher than expected, with increasing maternal age at conception and limited uptake of prenatal screening by older women. That study, which was not mentioned in the editorial, aimed to assist efforts to improve quality of life in persons with Down syndrome by identifying and defining baseline measures of features affecting morbidity and mortality. We wonder whether this attention to quality of life is reflected in the descriptions of Down syndrome given to parents during counseling about screening.

Van den Berg et al⁵ offered prenatal screening for Down syndrome to younger women in the Netherlands, most of whom rejected the option. The authors concluded that routine offers of screening without careful counseling may hinder informed decision making. This could have a real impact in the United States, given the recent proposal for universal screening for Down syndrome.⁶ More discussion is needed about the counseling process as prenatal screening is being offered to an increasingly wider audience.

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Reply

To the Editor:

We read with interest the letter from Mattheis et al responding to an editorial published in the January issue of *The Journal*.¹ The editorial discussed our article on the prevalence of Down syndrome² and another on the prevalence of cystic fibrosis³ and referred to "selective pregnancy terminations and reduced birth prevalence" as "a desirable and attainable goal." We agree with Mattheis et al that prenatal screening has uses beyond the prevention of adverse outcomes. At present, a fetal diagnosis of Down syndrome is most likely to result in a termination of pregnancy in our population, but certainly some families choose to continue the pregnancy or to not undergo prenatal testing at all.

Our article highlighted the fact that despite the widespread use of prenatal testing, the live birth rates of Down syndrome underscore the need for continuing attention to providing high-quality services to families with a child with Down syndrome. Although this lies outside the scope of the epidemiologic data presented in our article, we welcome and support the call for discussion and research on the counseling process accompanying prenatal screening programs. All prenatal testing should be based on informed and free choice, both in the decision to undergo testing and when making a decision after receiving a prenatal screening or diagnostic test result. Contributing to this is the development and evaluation of a decision aid for prenatal testing for fetal abnormalities, aimed at improving women's informed decision making.⁴ A cluster randomized trial conducted by our group demonstrated that this tailored prenatal testing decision aid played an important role in improving women's knowledge of their screening options and helping them make decisions according to their values.

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