Is prenatal screening for rare diseases like spinal muscular atrophy too costly?

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Spinal Muscular Atrophy (SMA) is one of many serious disorders for which prenatal testing is available. SMA affects approximately 1 in 10,000 live births and is the leading genetic cause of infant mortality and the second most common autosomal recessive disorder, after cystic fibrosis. Although the American College of Medical Genetics recommends carrier testing for all couples, the American College of Obstetricians and Gynecologists has issued a recommendation to the contrary, citing lack of information about the costs and benefits of screening for SMA. An article which may shed light on this controversy appears in the March 2010 issue of the *American Journal of Obstetrics & Gynecology*.

Using a decision analytic model, the authors found that 12,500 women need to be screened to prevent one case of SMA, at a cost of $5 million per case averted. They also determined that at $4.9 million per Quality-Adjusted Life Year (QALY), such screening was not cost-effective. Maternal QALY was used to measure the combined effects of caring for a child who suffers from the disease with resultant premature death and disability.

The cost of the procedures involved, maternal and paternal genetic testing, fetal amniocentesis and genetic testing of fetal cells, were all estimated from data published in the literature. The model assumed that couples with a positive fetal test would elect pregnancy termination. Lifetime costs of caring for an affected child were estimated from costs for similar diseases documented in the medical literature. All costs were

adjusted to 2009 dollars using the Consumer Price Index.

The authors found that the cost per case averted was the most influenced by the baseline prevalence of disease in the population screened. For those couples with a higher prevalence, such as those with a family history of disease, screening may be a cost-effective strategy.

Writing in the article, Sarah E. Little, MD, Department of Obstetrics and Gynecology, Massachusetts General Hospital, Boston, and co-authors state that "our estimated cost per added QALY falls far beyond what is usually considered to be cost effective. SMA screening does not approach the cost-effective range ($50-100,000/QALY) until the cost of severe disease is over $7 million or the cost of the mild disease is over $17 million, both of which are more than 20 times the baseline estimates. As such, we feel there is little chance that the basic finding that universal SMA testing is not cost effective would change appreciably with different model inputs."

More information: The article is "The cost effectiveness of prenatal screening for spinal muscular atrophy" by Sarah E. Little, MD; Vanitha Janakiraman, MD; Anjali Kaimal, MD, MAS; Thomas Musci, MD; Jeffrey Ecker, MD; and Aaron B. Caughey, MD, PhD. It appears in the American Journal of Obstetrics & Gynecology, Volume 202, Issue 3 (March 2010).

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