

Study finds screening for spinal muscular atrophy not cost effective

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In a study to be presented today at the Society for Maternal-Fetal Medicine's (SMFM) annual meeting, The Pregnancy Meeting, in Chicago, researchers will unveil findings that show that it is not cost effective to screen for spinal muscular atrophy.

Spinal muscular atrophy (SMA) is the most common genetic cause of infant mortality and the second most common inherited autosomal recessive disorder. There is controversy about whether prenatal carrier detection should be routinely offered to couples. In the November 2008 issue of the *Genetics in Medicine* Journal, the American College of [Medical Genetics](#) recommended that carrier screening for SMA should be made available to all families. However, the American College of Obstetricians and Gynecologists, in a committee opinion from May 2009, recommended against preconception and [prenatal screening](#) in the general population.

In the study presented at the Society for Maternal-Fetal Medicine's meeting, researchers describe the analytic model that was created to compare a policy of universal prenatal SMA screening to that of no screening. Baseline assumptions included a disease prevalence of 1 in 10,000 for a carrier rate of 1 in 50 with 70% of affected children having severe disease. They assumed a 90% sensitivity rate for carrier screening and that 2% of SMA cases arise from de novo mutations. Baseline cost estimates (2009 dollars) included \$400 for a carrier screen and \$260,000 for the lifetime cost of a child with severe disease. Maternal quality-adjusted life-years (QALYs) were calculated assuming a 22% reduction

in quality of life for having a child with severe disease and 8% for fetal loss.

The results showed that 11,000 women would have to be screened to prevent one case of SMA, at a cost of 4.7 million dollars per case averted.

"Our findings show that screening everyone for SMA is not cost effective," said one of the study's authors, Dr. Sarah Little of Massachusetts General Hospital. "Our results, however, were most sensitive to changes in the baseline prevalence of disease, suggesting that prenatal SMA screening may be cost effective in high risk populations, such as those with a family history of disease."

Provided by Society for Maternal-Fetal Medicine

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