Gene replacement tx beneficial in spinal muscular atrophy

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(HealthDay)—Gene replacement therapy is beneficial in spinal muscular

atrophy type 1 (SMA1), and nusinersen is beneficial for infants with spinal muscular atrophy, according to two studies published online Nov. 1 in the *New England Journal of Medicine*.

Jerry R. Mendell, M.D., from the Center for Gene Therapy at the Research Institute at Nationwide Children's Hospital in Columbus, Ohio, and colleagues examined functional replacement of the mutated gene encoding survival motor neuron 1 (*SMN1*) in SMA1. Fifteen patients with SMA1 received a single dose of intravenous adeno-associated virus serotype 9 carrying SMN complementary DNA encoding the missing protein (three received a low dose; 12 received a high dose). The researchers found that at 20 months of age, all 15 patients were alive and event-free compared with 8 percent survival in a historical cohort.

Richard S. Finkel, M.D., from Nemours Children's Hospital in Orlando, Florida, and colleagues conducted a randomized, double-blind, sham-controlled trial of nusinersen in 78 infants with spinal muscular atrophy. In a prespecified interim analysis, the first primary end point (motor-milestone response) was assessed. The researchers found that the percentage of infants with a motor-milestone response was significantly higher in the nusinersen versus the control group (41 versus 0 percent), prompting termination of the trial. The percentage with motor-milestone response at the final analysis was 51 versus 0 percent, respectively.

"Among infants with spinal muscular atrophy, those who received nusinersen were more likely to be alive and have improvements in motor function than those in the control group," Finkel and colleagues write.

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