Gene therapy may cure 'bubble boy disease'

Treatment seems to have cured 8 of 10 kids who had rare, deadly condition

The Associated Press
updated 4:59 p.m. ET, Wed., Jan. 28, 2009

NEW YORK - Gene therapy seems to have cured eight of 10 children who had potentially fatal "bubble boy disease," according to a study that followed their progress for about four years after treatment.

The eight patients were no longer on medication for the rare disease, which cripples the body's defenses against infection. The successful treatment is reported in Thursday's issue of the New England Journal of Medicine and offers hope for treating other diseases with a gene therapy approach.

Bubble boy disease is formally called severe combined immunodeficiency, or SCID. This genetic disorder is diagnosed in about 40 to 100 babies each year in the United States. The nickname comes from the experience of a Houston boy, David Vetter, who became famous for living behind plastic barriers to protect him from germs. He died in 1984 at age 12.

He had the most common form of SCID. Recent studies found that gene therapy produced impressive results for that form of the disease, but also carried a risk of leukemia.

The new study involved a different, less common form of SCID — and one that holds a key position in medical history. In 1990 it became the first illness to be treated by gene therapy, according to the U.S. government. Two Ohio girls improved but continued to take medication.

This form of SCID arises in babies with a genetic defect that leaves them deficient of an enzyme called adenosine deaminase. Patients can be treated with twice-weekly shots of the enzyme or a bone marrow transplant, but the medicine is expensive and marrow transplants don't always work.

No ill effects appearing
Gene therapy for the new study was performed in Italy and Israel. Researchers removed marrow cells from the patients, equipped the cells with working copies of the gene for the enzyme, and injected the cells back into the patients. In most cases, that was done before age 2.

The journal article reports the outcome two to eight years later, with an average of four years. All 10 patients were still alive, but two needed further treatment. None showed signs of leukemia or other health problems from the therapy, the researchers said.

Dr. Donald Kohn, a SCID expert at Childrens Hospital Los Angeles and the University of Southern California, said scientists are trying to understand why gene therapy produces a leukemia risk with the most common form of SCID but not the enzyme-related form.

The new findings are good news for the idea of using gene therapy to treat some other blood cell disorders, including sickle cell disease, said Kohn, who didn't participate in the new study.

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