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Cure for genetic disease requires money

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Fifty years ago today, James Watson's and Francis Crick's scientific paper ignited a revolution in modern science. Fidgeting with cardboard cutouts and X-ray photographs, Watson and Crick discovered the spiral staircase structure of DNA. Their description of the double-helix sparked a torrent of research into DNA that today has us on the verge of finding a cure for genetic diseases.

One such disease is spinal muscular atrophy, or SMA, the leading genetic killer of infants and toddlers.

More than 7 million adults carry the gene that can give their newborns this crippling disease ---- and most do not know they have it. An estimated 25,000 Americans suffer from SMA and up to 1,000 babies each year are born with this deadly genetic malady that dooms them to suffering and premature death, usually before age 2.

Like Lou Gehrig's disease, SMA is a neuromuscular illness that attacks nerve cells of the spinal cord and causes skeletal muscles to waste away. Victims of SMA lose muscle control and strength, leading to a progressive inability to walk, stand, sit up, move and --- - eventually ---- breathe. In less severe forms, patients require physical therapy, frequent hospitalization and repeated surgery. No effective treatment is known.

The disease has been difficult to diagnose and some physicians fail to recognize it. However, thanks to scientific innovations and the persistence of a select group of lawmakers from California, there is hope.

Researchers at the National Institute of Neurological Disorders and Stroke and the National Institutes of Health recently designated SMA as a model for translational research funding, which uses basic scientific research to devise actual drugs and treatments. Translational research is critical to the development of a treatment for SMA and similar diseases.

In a recent letter to the director of the NIH, leading scientists and clinicians worldwide urged the NIH to implement translational research for SMA. They wrote that with NIH

funding of \$20 million to \$30 million a year, an effective therapy for SMA could be achieved in five years.

Unfortunately, the NIH has been slow to fund this research. One reason is that SMA is an "orphan" disease that has attracted little public attention. Two noteworthy exceptions are Reps. Randy "Duke" Cunningham, R-Escondido, and Susan Davis, D-San Diego. They have been at the forefront of the struggle, leading a coalition of congressional supporters from California, fighting for SMA families and urging the NIH to accelerate research funding.

In a recent letter to the NIH, nearly one-third of the U.S. Senate and more than 75 members of the House urged the NIH and NINDS to fund translational research for SMA. Cunningham and Davis were the House leaders in this effort, with help from Democratic California U.S. Sens. Barbara Boxer and Dianne Feinstein and 13 other members of California's congressional delegation.

Fifty years of biological research has brought us to the verge of eradicating a dreaded disease. SMA research has come far with limited funding and attention. Full federal funding could help us complete the cure.

George Eng lives in San Diego and is a co-founder of the SMA Foundation.
www.smafoundation.org.