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Funding needed to cure a disease that makes infants waste away

By Loren A. Eng

Fifty years ago, Francis Crick and James Watson ignited a revolution in modern science. Toying with a collection of cardboard cutouts and X-ray photographs, the two scientists discovered the structure of deoxyribonucleic acid, known as DNA.

Their spontaneous observation of the "double helix" sparked a torrent of research into DNA that today has us on the verge of finding a cure for certain genetic diseases. One of these close to a cure is spinal muscular atrophy, an often fatal and prevalent genetic children's disease.

Though virtually unknown, spinal muscular atrophy is the leading genetic killer of infants and toddlers. Up to 1,000 babies, one in every 6,000 live births, are born with this condition annually. More than 25,000 people have the disease, and 1 in every 40 Americans (7 million people) carry the gene.

Like amyotrophic lateral sclerosis, or Lou Gehrig's disease, spinal muscular atrophy is a neuromuscular illness marked by the wasting of skeletal muscles. It is caused by a missing or defective gene that maintains the health of spinal nerves called motor neurons, which control voluntary muscle function. Within a few months of birth, infants with this condition begin losing physical strength. Without respiratory support, 95 percent of babies with the most severe form die before age 2. Patients require extensive physical therapy, lengthy hospitalization and frequent surgery. There is no treatment.

Researchers have isolated the gene involved, SMN, as well as the protein produced by the gene. Even more remarkable, scientists have located a backup gene capable of taking over some protein-producing functions. Drugs to improve the action of the backup gene are approaching the testing stage. The National Institute of Neurological Disease and Stroke recently designated spinal muscular atrophy as the model for its new approach to funding "translational research." Translational research develops findings in the lab by scientists into drugs and treatments. Funding of \$20 million to \$30 million per year could help develop a treatment within five years.

Unfortunately, the National Institutes of Health has been slow to fund research. Spinal muscular atrophy funding peaked this year at \$5 million, after remaining under \$3 million

for several years. This is remarkably low compared to research funding dedicated to diseases of similar prevalence and severity. Translational research funding will not, due to purely administrative reasons, be available until January 2004. The NIH's inaction on a disease so close to a cure is troubling to many.

Along with U.S. Sen. Tom Harkin, D-Iowa, U.S. Sen. Arlen Specter of Philadelphia has strongly endorsed the translational research concept, urging sufficient resources to maximize chances of success and evaluate whether a similar approach will work for other diseases.

Fifty years of biological research has brought us to the verge of one of the highest achievements attainable by humanity: the eradication of a dreaded disease. Only sufficient federal dollars now stand between spinal muscular atrophy and a cure.

Loren A. Eng is president of the Spinal Muscular Atrophy Foundation, based in New York. You can find out more about the condition at www.smafoundation.org.