

March of Dimes Foundation

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Media Relations

## 2009 MARCH OF DIMES PRIZE IN DEVELOPMENTAL BIOLOGY

### BIOGRAPHICAL SKETCH

**LOUIS M. KUNKEL, PH.D.**

**Principal Investigator, Howard Hughes Medical Institute  
Professor of Pediatrics and Genetics, Harvard Medical School  
Director, Program in Genomics, The Children's Hospital  
Boston, Massachusetts**

Dr. Louis Kunkel once gambled his entire career on finding a gene underlying Duchenne muscular dystrophy a genetic disease that attacks a person's muscles.

That was more than 20 years ago, and fortunately, Dr. Kunkel's gamble paid off. Today, he is an internationally-renowned geneticist and a third-generation member of the National Academy of Sciences. He serves as chairman of the Muscular Dystrophy Association's Scientific Advisory Committee, has authored hundreds of journal articles, received many honors and awards, and is the 2009 co-recipient of the March of Dimes Prize in Developmental Biology.

More than 250,000 Americans are affected by one of the 40 known neuromuscular disorders, which include nine types of muscular dystrophy, facioscapulohumeral muscular dystrophy, amyotrophic lateral sclerosis (ALS, or Lou Gehrig's disease), spinal muscular atrophy and others. Muscular dystrophy, is a genetic disorder in which the muscles progressively degenerate. Among this group are late-onset conditions that appear in adulthood, such as amyotrophic lateral sclerosis (ALS, or Lou Gehrig's disease), as well as childhood conditions such as Duchenne and Becker muscular dystrophies and spinal muscular atrophy.

Living in Boston in the early 1980s, fresh from a postdoctoral stint at the University of California San Francisco, Dr. Kunkel found work in the famed laboratory of Dr. Samuel Latt at Children's Hospital Boston, sorting X chromosomes. Dr. Kunkel was given one year to find another postdoctoral fellow-ship

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that would support him and his research. Under that pressure, he wrote an ambitious grant application proposing to identify and map the gene that causes Duchenne muscular dystrophy, and submitted it to the Muscular Dystrophy Association (MDA). He won the grant.

“My whole career depended on this. I put my whole career on identifying that gene, and MDA really took a chance on me,” said Dr. Kunkel. By 1986 he had succeeded in identifying the DMD gene and its protein product, dystrophin. Mutations in the gene produces defective or absent dystrophin, leading to Duchenne and Becker forms of muscular dystrophy.

To study the DMD gene and dystrophin, Dr. Kunkel made one of the first successful attempts at a then-novel approach called positional cloning, now a staple of DNA research. It involved identifying missing segments from the DNA of patients with the disease in order to determine the location of the gene on the X chromosome.

As a child, Dr. Kunkel bred flowers, specifically irises, with his father and planned to become a botanist after graduating from Gettysburg College in Pennsylvania. Then he had an internship with the late Dr. Victor McKusick, the founder of the field of medical genetics and an early proponent of mapping the human genome. It was Dr. McKusick who suggested that the young Kunkel apply to the human genetics program at Johns Hopkins University, where he went on to earn his doctorate in biology in 1978.

Dr. Kunkel has now dedicated his career to the study of muscular dystrophy. His lab has identified 15 dystrophin-related or associated genes and their protein products in the last three decades. Mutations in three of those genes cause limb-girdle muscular dystrophy.

Duchenne and Becker muscular dystrophies are allelic disorders – that is, they are caused by different mutations in the same DMD gene. The diseases occur when the DMD gene doesn't produce enough of the protein dystrophin, or produces poor quality dystrophin. Dystrophin is a key element in a group of proteins that provide muscle cells with the structural stability they need to work properly.

### March of Dimes Prize/Kunkel-3-

This finding led Dr. Kunkel to work in 1987 and 1988 on dystrophin with Dr. Kevin Campbell, now his co-recipient of the March of Dimes Prize

Because of Dr. Kunkel's findings, health professionals can now diagnose and help manage muscular dystrophy earlier and with much greater precision, and provide better genetic counseling to families. Dr. Kunkel's work has led to the development of potential new treatments that are offering hope to those affected by various forms of muscular dystrophy, for which there are no known treatments.

One potential new treatment is gene therapy, which involves implanting normal dystrophin genes into those patients who have Duchenne muscular dystrophy. However, studies have shown that the muscles do not produce enough dystrophin to repair themselves. Dr. Kunkel also is investigating stem-cell based treatment for muscular dystrophy.

He also has started a database of children who have received treatment at Children's Hospital Boston that includes not only their medical/genetic backgrounds but also environmental factors – the interaction of which may explain a variety of diseases, including autism.

Dr. Kunkel and his wife, Susan, have three daughters, Sarah, Johanna, and Ellen and live outside of Boston in Westwood, Massachusetts.

The March of Dimes Prize in Developmental Biology has been awarded annually since 1996 to investigators whose research has profoundly advanced the science that underlies the understanding of birth defects. The March of Dimes Foundation created the Prize as a tribute to Dr. Jonas Salk, who received Foundation support for his work to create a polio vaccine.

The March of Dimes is the leading nonprofit organization for pregnancy and baby health. With chapters nationwide and its premier event, March for Babies®, the March of Dimes works to improve the health of babies by preventing birth defects, premature birth and infant mortality. For the latest resources and information, visit [marchofdimes.com](http://marchofdimes.com) or [nacersano.org](http://nacersano.org).