

# Louis Kunkel Biography

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# Biography

Louis Kunkel is renowned for his research on the **genetics** of pediatric neuromuscular disorders. Most notably, he is responsible for discovering the **gene** that causes Duchenne muscular dystrophy.

Kunkel received his B.A. degree from Gettysburg College in 1971, and completed his Ph.D. at Johns Hopkins University, with a specialty in human genetics. He was appointed chief of the division of genetics at Children's Hospital in Boston in 1988, and an investigator of the Howard Hughes Medical Institute in 1990, while holding a post as professor of genetics and pediatrics at Harvard University.

In 1986, Kunkel and colleague Tony Monaco discovered the gene that causes Duchenne muscular dystrophy. The genetic disease, which is found on the **X chromosome**, affects approximately one out of every 3,000 to 4,000 newborns, primarily males. Duchenne muscular dystrophy attacks the muscles, wearing them away until those afflicted are unable to walk. Most patients with Duchenne muscular dystrophy die by the time they reach their mid-20s.

By comparing the **DNA** from healthy cells to those of diseased cells, the scientists were able to isolate the Duchenne gene. The gene produces the protein dystrophin which, when missing or malfunctioning, causes the muscular degeneration seen in both Duchenne and Becker muscular dystrophies. This discovery, as well as the identification of the gene behind **cystic fibrosis** three years later, raised hopes that **gene therapy** could be used to disrupt the progress of inherited diseases.

After identifying the Duchenne gene and describing the dystrophin protein, Kunkel and his team discovered a method for **cloning** the gene. Subsequent work focused on identifying dystrophin-associated proteins in normal muscle, as well as in the brain, in order to understand how an absence of these proteins leads to the development of muscular dystrophy. Kunkel's research was the first step towards halting the progress of the disease, as well as improving diagnosis and treatment of other neuromuscular diseases.

In 1999, a team of scientists at Children's Hospital and Harvard University, led by Kunkel and Richard Mulligan, announced a breakthrough in the treatment of muscular dystrophy. They were able to inject healthy bone-marrow **stem cells** into mice with muscular dystrophy. These donor cells traveled through the bloodstream, infusing and regenerating weakened muscles with the dystrophin protein. The new procedure was hoped to eventually be used in the treatment of other degenerative muscle diseases.