Victor Almon McKusick, MD, renowned worldwide as the father of modern medical genetics, was awarded with the 2008 Japan Prize for his work in medical genomics and genetics. The prize, created in 1985, is chosen annually by a committee of Japanese scholars based on nominations from an international pool of academics.

Dr. McKusick, University Professor of Medical Genetics at Johns Hopkins University School of Medicine in Baltimore, will be given a medal and 50 million yen (about $470,000) by the Science and Technology Foundation of Japan on April 23.

Dr. McKusick graduated from the Johns Hopkins University School of Medicine in 1946, joined the medical faculty in 1947 as a cardiologist, and is the longest-serving faculty member in the school’s history. He has served as director of the Division of Medical Genetics (1957-1975) and physician in chief of the Johns Hopkins Hospital (1973-1985).

His foray into genetics began when he developed an interest in Marfan syndrome, a genetic connective tissue disease marked by unusually tall height, heart defects, and other abnormalities.

He has since been a leader in mapping the location of genes on chromosomes and relating gene location to diseases, including Marfan syndrome and dwarfism. Thomas Bird, MD, professor of medicine, neurology, and medical genetics at the University of Washington in Seattle, WA, praised Dr. McKusick as a founding father of medical genetics. “His contributions to this field have been staggering in terms of research, training, and education,” he told Neurology Today. “He represents a world class model of how to build bridges between molecular genetics and the practice of medicine.”

Dr. McKusick established the first training program in medical genetics in the U.S. at Johns Hopkins in 1957, devoted to the study and management of inherited diseases and predispositions.

In the 1960s, he performed studies on the Old Order Amish — an inbred community that keeps diligent genealogical records — which enabled him to trace rare recessive disorders and classify previously unrecognized, genetic diseases. These findings are incorporated into his most famous work, Mendelian Inheritance in Man, a vast compilation of human genes and genetic disorders (1966). The catalogue, now known as OMIM (Online Mendelian Inheritance in Man) and maintained online, contains descriptions of all known genes.

Dr. McKusick is known for his early recognition of the significance of mapping the entire human genome. He helped found and served as the first president (1988-1990) of the Human Genome Organization, an international group established to encourage mapping and sequencing the human genome. The phenomenal Human Genome Project, which has aided in the understanding and eventual prevention or cure of human diseases, was completed in 2003.

Dr. McKusick is a member of numerous scientific societies, has been awarded with over twenty honorary degrees, and has also won the highest scientific honor in the U.S., the National Medal of Science (2001).