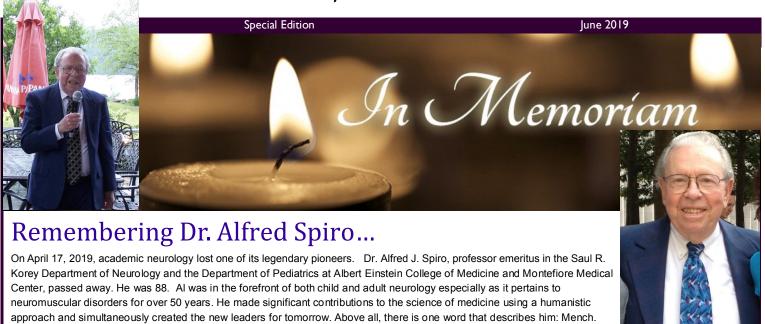


## Child Neurology News

The Children's Hospital at Montefiore

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Al graduated with his MD degree in 1955 from the University of Bern, Faculty of Medicine in Bern, Switzerland and then did his pediatric residency in Babies Hospital in Newark New Jersey followed by a neurology residency at the Hospital of University of Pennsylvania and child neurology residency at the Children's Hospital in Philadelphia. In between his residencies he was in active duty in the United States Public Health Service. He was appointed at Albert Einstein in July of 1966 and he remained with us for the rest of his career where he developed an outstanding child neurology division and neuromuscular program beyond compare. Al was the director of child neurology at Einstein from 1980 to 1995. He trained scores of pediatric neurologists that became leaders in the field, in all aspects of pediatric neurology, some of whom are writing letters supporting this nomination. Although very interested in neuromuscular disorders, he made sure that all aspects of neurology were covered, providing an encyclopedic knowledge to the residents he trained

From very early on, AI was interested in neuromuscular diseases and he was the first to describe a variety of neuromuscular diseases using histochemistry. Indeed, he created one of the first histochemistry labs in the US, which was absolutely necessary before neurogenetic testing became available many decades later. He was and still is a master at muscle biopsy interpretation. He described a series of mitochondrial myopathies and these diseases can perhaps be best classified as "Spiropathies". He was able to describe clinical features that made the diagnosis possible and the confirmation by detailed immunological studies. He made foundational contributions in the areas of congenital myopathies (myotubular, nemaline), Duchenne muscular dystrophy, myotonic dystrophy, sarcoglycanopathies, limb girdle muscular dystrophy and spinal muscular atrophy. He described unique features that aid in the diagnosis of neuromuscular diseases such as minipolymyoclonus in children with spinal muscular atrophy. Here is a brief reflection of the breadth and depth of Al's clinical work is his publication history, rich with case reports and case series that span the field of neuromuscular disorders. In 1965 (JAMA Neurology), he described a hereditary occurrence of nemaline myopathy, a disorder that had been identified by Shy et al. only 2 years before and that had appeared not to be hereditary in all other reports up to that time. In 1966 (Arch Neurol), Al reported his examination of an adolescent boy whose muscle biopsy revealed fetal-type myotubes instead of mature muscle cells, and thus described myotubular myopathy for the first time; now, the field recognizes the genetic abnormalities and complex relationships among various forms of myotubular and centronuclear myopathies. He reported in 1970 both a new mitochondrial myopathy (JAMA Neurology) and familial cases of hereditary spastic paraparesis with sensory neuropathy (Dev Med Child Neurol). As early as 1977, Al was offering diagnostic algorithms for the child with muscle weakness, a common and puzzling situation for clinicians. Limb-girdle muscular dystrophy, nemaline myopathy, adrenoleukodystrophy, myotonic dystrophy, amyotrophic lateral sclerosis, polymyositis, and of course Duchenne and Becker muscular dystrophies: all of these disorders came under Al's scrutiny, and he shared his findings with us.

In his neuromuscular program, he included patients not only with muscular dystrophy or mitochondrial myopathies but all aspects of the neuromuscular disorders. He developed the first such clinic in the New York metropolitan area which grew over time and attracted the interest of many doctors keen to better understand neuromuscular diseases. He molded his neuromuscular clinic into a comprehensive, multi-disciplinary program that was a regional magnet for patients of all ages, newborn to elder, suffering from all types of neuromuscular disorders. The genetic maps he drew required multiple family members and multiple generations, more than might be recruited through a purely pediatric practice. He taught generations of trainees on the approach to the child with muscle weakness.

Al excelled in all aspects of Child Neurology. He was the director of the Einstein/Montefiore Child Neurology program from 1980-1995, where he established a world class training program addressing all aspects of child neurology. Many of his students have advanced the field based on his original contributions. As a clinician, as a writer, and as a researcher, he always kept in the forefront the needs of his patients. He did not let the rest of us forget for a moment that, what we do, has a major impact on the lives of individuals. He kept medicine humane even when few or no treatment options were available to offer to our patients. Yet, Al was a very unassuming guy, downplaying his accomplishments. But not the Child Neurology Society which recognized his achievements by bestowing to him the 2018 Child Neurology Society Roger and Mary Brumback Lifetime Achievement award.